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Pediatric Rehabilitation

Principles and Practice

Fifth Edition

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DEDICATION

To Dr Gabriella Molnar, who passed away since the last edition. She has left a void that will not be filled. She is a recognized founder of our field of pediatric rehabilitation medicine. Dr Molnar created our first textbook, edited the subsequent two editions, and wrote numerous state-of-the-art reviews for the Child with Physical Disability. After escaping from Hungary in 1956 from the Russian occupation and communist regime, Dr Molnar displayed much foresight and courage throughout her professional career. Her guiding principle has always been that children are not miniature adults, but individuals with changing physical, intellectual, and emotional abilities and needs. At every age, therefore, the principles of rehabilitation medicine have to be adapted to these changing aptitudes. Beginning as a resident at Albert Einstein College of Medicine, Dr Molnar quickly rose through the ranks from faculty instructor to full tenured professor, while developing and running the Pediatric Rehabilitation Medicine Service. Concluding her career at Children's Hospital and Research Center in Oakland, California, where she created a new Department of Pediatric Rehabilitation Medicine, she finished training her last of over 50 domestic and international fellows. Her speaking career has included invitations from all over the world, including Australia, Europe, Asia, and England. She has served on the editorial boards for the *Archives of Physical Medicine and Rehabilitation* from 1976 to 1994 and *Developmental Medicine and Child Neurology* from 1992 to 1997. She is a recipient of the Krusen Award from the American Academy of Physical Medicine and Rehabilitation (AAPMR), the highest honor obtainable for proven performance in clinical expertise, contributions to the literature, and administration in the field of rehabilitation medicine. Simply stated, Dr Molnar defines the standard for the rest of us to follow. Her husband's generous contribution to the PMR Foundation created an award for lifetime achievement in pediatric rehabilitation and funds research stipends to increase the body of knowledge in pediatric rehabilitation. Her life is an inspiration to all of us.

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PREFACE

This is the second edition of *Pediatric Rehabilitation* that has not been under the leadership of Dr Gabriella Molnar, but her influence and wisdom are still apparent in the text.

Our field has changed again since the last edition. Every chapter in this new fifth edition has been thoroughly revised and expanded. We have covered many of the more common genetic disorders throughout the textbook and added new chapters on sports injuries, concussions, and rehabilitation of pain and conversion disorders that will be of great interest to physiatrists. We have also added a chapter on ultrasound, as many of us are actively involved in this exciting adjunct to our practice of medicine.

You will notice that some chapter authors have returned and we have asked them to incorporate new pediatric rehabilitation specialists, as it is our hope that these new coauthors will become the senior authors of future chapters and perhaps editors of future editions.

We are happy to present to you this compiled wisdom of the brightest and most enthusiastic clinicians in our tightly knit group of pediatric rehabilitation specialists.

On a somber note, the editors are saddened to announce the recent passing of Dr Ellen Kaitz. Her help in the previous and the current edition is greatly appreciated.

Michael A. Alexander
Dennis J. Matthews

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Pediatric Rehabilitation: Principles and Practice, Fifth Edition

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HISTORY AND EXAMINATION

Maureen R. Nelson, Michael A. Alexander, and Gabriella E. Molnar[†]

The physiatric history and examination of a child require a blend of medical diagnostic skills to establish or confirm the diagnosis as well as knowledge of child development and behavior to evaluate functional assets and challenges for the intervention phase of rehabilitation.

SETTING THE TONE

To ensure the best cooperation, especially in the preschool age, the environment should be child-friendly. Exposure to crying and upset children should be avoided in the waiting room or other areas. Families will do better if someone else can take care of the child's siblings rather than bring them to the appointment so that parents can focus on the interview without distraction. The examination room may have a small table and chair with an assortment of toys for children of different ages to make them comfortable and relaxed. The examiner's attire also influences the child. Many times a good rule is to "lose the white coat," unless it has a pocket full of toys. The child is not impressed by the coat and may be intimidated by past medical visits. A child-oriented décor such as pictures of cartoon characters or animals on the wall, small toys, and decals on instruments helps to create a playful atmosphere and alleviate the child's fears.

Start the visit by introducing yourself, including telling the patient and parents something about yourself, and what will happen during the visit, and how long it will last. Ask the parents to tell you why they came and what specific questions they have for you.

Concerns stated by the referral source should be shared with the parents. Many parents are unsure about what information the visit can provide. This is the opportunity to explain what pediatric rehabilitation is, its focus on function, and what it can offer the child and family. The examiner should also explain that it is part of the examination to watch the child so that the parents will not feel offended by the examiner's wandering gaze. Because observation of spontaneous behavior is one of the most informative aspects of evaluating youngsters with a disability, examination begins from the moment the child is in the physician's view. Questions about history and illnesses should be asked in simple terms so that the family can understand them and provide proper information. It is also important to have someone clarify insurance coverage and whether additional tests or treatment can be performed on the same day or must await approval.

HISTORY

PRENATAL AND PERINATAL HISTORY

The prenatal and perinatal history includes the preconceptual period and the parents' ages and health before and since the birth of the child. Maternal factors during gestation may lead to fetal malformations. Examples of these associations include febrile illnesses (1) anticonvulsants (2) with spina bifida; maternal diabetes with caudal regression syndrome and sacral agenesis; and rubella, thalidomide, or fetal alcohol syndromes. Weak or eventually lost fetal movements may be the earliest sign of a motor disability of prenatal origin. Prenatal care, unusual weight gain or loss, hypertension, or any other gestational problems should be explored. Mode and duration of delivery, use of anesthesia, induction, intrapartum complications, and expected and actual date of birth should be noted. Any history of previous pregnancies, deliveries, and fetal loss is significant. Prenatal cerebral damage is increased in infants of mothers with previous spontaneous abortions (3). A detailed neonatal history is essential, including birth weight, Apgar scores, onset and success of breastfeeding, as well as the infant's age at discharge. Weak lip seal and sucking force and inadequate feeding may be preliminary signs of oral motor dysfunction. If care in the neonatal intensive care unit (NICU) was required, what were the specifics of that, including medications and ventilator support? Neonatal seizures may signal pre- or perinatal brain damage.

[†] Deceased.

Prematurity, particularly very low birth weight, is a frequent cause of cerebral palsy (2). Large birth weight may lead to intrapartum trauma, brachial plexus palsy, or, on extremely rare occasions, spinal cord injury, particularly with breech or other fetal malposition. When extended hospitalization was required, one should note the infant's age, weight, and condition on discharge, including means of feeding and need for ventilatory or other supportive measures at home, since these may predict subsequent, persistent, or recurrent problems.

DEVELOPMENTAL HISTORY

The developmental history should cover all major aspects of function and behavior. For details of developmental milestones and testing, refer to Chapter 2. This discussion presents only general guidelines for the purpose of diagnostic interpretations. Discrepancies between different areas of functioning provide clues about the nature of medical diagnosis and developmental disability.

Delayed accomplishments, primarily in motor function, suggest a neuromuscular deficit. One of the earliest signs that parents report is a lack of spontaneous movements when the infant is held or placed in the crib. They may add that the baby feels limp or stiff, suggesting either hypotonia or spasticity. In all cases of motor dysfunction, it is important to clarify whether the dysfunction was a steady, continuing delay from an early age, suggesting a static disease, or an arrest or regression noted at a particular point. However, slow deterioration due to progressive neurologic disease may be masked for a time by the relatively fast pace of early motor development. Developmental history and subsequent assessment must take into consideration the interactive effect of coexistent deficits. Slow development in personal and adaptive tasks that require both motor and cognitive abilities may be related to impairment in either area. A significant cognitive dysfunction by itself may delay gross and fine motor development (4). This also frequently exacerbates the functional consequences of a neuromuscular disability and may create the impression that the motor deficit is more severe than it actually is.

A history of delay in communication development raises several differential diagnostic possibilities: (a) true language dysfunction affecting receptive or expressive domains or both, (b) oral motor dysfunction interfering with speech production, and (c) significant hearing loss. In a child with motor disability, language dysfunction may result from diffuse or focal cerebral lesions, such as head injury or cerebral palsy, particularly when cognitive function is also affected.

The ability to follow simple and, later, complex commands indicates preserved receptive language even in the absence of verbalization. Parents report a variety of responses, such as smiling, cooing, crying, pointing, or vocalization with inflection as a substitute for speech. Oral motor dysfunction is also associated with cerebral palsy, most often with spastic quadriparesis or dyskinetic disorders due to suprabulbar or pseudobulbar palsy. Bulbar palsy in medullary involvement affects speech production, for example, in spinal muscular atrophy or spina bifida with syringobulbia. There is a close association between anatomic structures and neurologic control for speech and oral feeding. Concurrent oral motor dysfunction with feeding difficulties is an additional sign of bulbar or pseudobulbar pathology and confirms the suspicion of speech production deficit. In such cases, history of early feeding is most relevant. For example, was there a good lip seal and strong suction on breastfeeding? When bottle-fed, the infant can handle 4 ounces in about 10 minutes, and feedings every 3 to 4 hours are generally adequate. The need for longer and more frequent feeding to maintain weight gain, especially during the first few months; coughing; nasal regurgitation of liquids; and then later difficulty with drinking from a cup; and difficulty with introduction of solid food due to chewing problems are early symptoms of oral motor dysfunction and a possible subsequent deficit in speech production. Augmentative communication training should be initiated early in such cases.

Hearing is an essential factor for speech development. Early cooing and babbling are innate characteristics of infants and involve the same vocal components, regardless of the language spoken in their environment. Infants with hearing loss start to fall behind after 6 to 8 months of age when learning of auditory-dependent vocalization begins. Parents may notice a decrease even in spontaneous babbling at that age. All neonates and infants at high risk for developmental disability or recurrent ear infections should have an initial and, if warranted, repeat hearing evaluations. Correction of a hearing deficit should be initiated as soon as possible after it is detected (5).

For infants and young children, the history is obtained from parents or caretakers. While gathering information from one person about another, the examiner gains an understanding of both and establishes rapport with parents and child. Early-school-age children can provide some information about themselves and should be encouraged to do so. Preadolescents and particularly adolescents generally prefer to give an account of their problems and achievements. Adolescents may wish to have privacy without the parent present, at least for part of the visit.

GENERAL HEALTH HISTORY

The examiner should determine whether the patient is an essentially well child with impairment or a sick child who has been hospitalized several times. In the latter case, one should explore in detail the frequency, reasons, tests, and treatments. Even if one has access to records,

the parents should be asked to tell the child's history in their own words. Their account provides an insight into their knowledge and participation in the child's care. One should ask how many visits they make to medical centers and therapists and how much time is spent in transit for the child's care.

History of allergies to medications or other substances should be noted. An early history of allergies to different and often inconsistent formulas may indicate that the child in fact had feeding difficulties that were attributed to allergy. Multiple exposures to latex and any signs of allergy should be determined, particularly in spina bifida or after repeated surgeries. Any medications that the child takes regularly, including dietary supplements and homeopathic or alternative medications or aerosols, should be recorded with dosage and schedule.

The risk and incidence of seizures are higher in static and progressive diseases of the central nervous system (CNS). Overt or suspicious signs, type and frequency of seizures, anticonvulsants, and their effectiveness and possible side effects should be recorded.

Nutrition, with special consideration for the child's disability, should be reviewed. Feeding difficulties or behavior problems may lead to inadequate consumption of calories and essential nutrients. Dietary intake may be lower than required for the increased energy expenditure on physical activities in children with motor disability. In contrast, caloric intake may be excessive when physical activity level is restricted and lead to obesity, commonly in wheelchair users with spina bifida (6) or muscular dystrophy, and after transition from walking with assistive devices to mobility via wheelchair. In children with caloric restriction, there is often a need for supplemental vitamins, protein, and calcium (7). Dietary information and guidance are fundamental for regulation of neurogenic bowel incontinence. Cultural and family eating patterns should be taken into consideration. Injuries, burns, fractures, and spinal cord and head trauma are followed by a period of a catabolic state. Monitoring of weight, nutrition, and fluid intake is essential during inpatient rehabilitation for major injuries, and after return to home. Caloric requirements for children are calculated from age-appropriate standards, which take into consideration growth. In children with motor disability, upward or downward adjustment in height and weight may be needed, depending on their level of physical activity and individual growth trend. Specific recommendations are available for children with spina bifida to avoid obesity (8,9). Obesity is a risk factor for secondary issues from the primary diagnosis, including pain and fatigue, as well as a cardiovascular risk and social challenge (10).

History of respiratory complications, past or present, should be explored in children with pertinent diagnoses. Central ventilatory dysfunction (CVD) is a potential severe complication of Arnold–Chiari malformation in spina bifida (11). Syringobulbia may cause similar symptoms. Nightmares, insomnia, and night sweating are complaints associated with hypercapnia, and may be reported in advanced stages of muscular dystrophy. Hypercapnia and sleep apnea may occur in diseases of the CNS. Intercostal muscle paralysis in high thoracic paraplegia with spinal cord injury or spina bifida, spinal muscular atrophy, or advanced muscle diseases leads to inefficient pulmonary ventilation and handling of secretions. With severe spastic or dyskinetic cerebral palsy, the respiratory musculature may lack coordination. Such children are prone to recurrent bouts of pulmonary infections. Coexistent feeding difficulties with minor aspirations or restrictive pulmonary disease due to spinal deformities may add to pulmonary dysfunction.

Restricted mobility of the spine and thoracic cage may be present in ankylosing spondylitis or severe systemic-onset juvenile rheumatoid arthritis/juvenile idiopathic arthritis (JIA). Detailed information about home management and use and frequency of equipment must be included in the history. Exercise dyspnea may be a sign of pulmonary compromise or deconditioning with the high energy cost of physical activities in children with some motor disabilities. Scoliosis may exacerbate respiratory disease in some of these children. Cardiac decompensation with right-sided failure, a potential complication of pulmonary dysfunction, is more likely to occur in older children or young adults with the previously mentioned disabilities. Myopathic conduction defects and arrhythmias are often symptom-free in the absence of heart failure. Consultation with pulmonary and/or cardiology specialists should be arranged when history reveals suspicious symptoms.

Visual and hearing impairments are more frequent in those with childhood disabilities. Inquiry about these aspects of function is a part of the history. Regular hearing and visual assessments are required. Prenatal infections, anoxic or infectious encephalopathy, metabolic diseases, meningitis, hydrocephalus, and head injury warrant exploration of visual and auditory function. Like all children, youngsters with disabilities are prone to a variety of childhood illnesses. In some cases, however, acute symptoms and febrile illnesses may be directly related to complications of a specific disability. Vomiting, headache, irritability, or lethargy may be prodromal signs of decompensating hydrocephalus in spina bifida, cerebral palsy (2), or an intercurrent unrelated illness. Recurrent headaches are also a manifestation of autonomic dysreflexia in spinal cord injury. Fever may represent central hyperpyrexia in severe head injury or hyperthermia due to pseudomotor paralysis in high thoracic spinal cord injury. However, these diagnoses can be made only after other causes of fever have been excluded. In those with a neurogenic bladder, urinary tract infection should always be investigated as a possible cause of febrile illness. A history of the usual pattern of the amount and frequency of voiding is essential in neurogenic bladder dysfunction. Systematic daily recording is a guide for bladder training. Fluid intake, in accordance with pediatric norms, needs

to be monitored at home, and records of both bladder and bowel schedules should be available on the medical visit.

Immunization history is part of all pediatric visits. The child in good health may not have received the recommended vaccinations because of excessive concern on the part of the family or pediatrician or the child may have been ill when scheduled for immunization.

HISTORY OF BEHAVIOR

The examiner should ask about the child's behavior in terms of temperament and personality. The parents may state that the child was always a good baby, but this report may mean that the youngster never cried and slept more than expected for his or her age. In other cases, parents may report excessive crying and restlessness both while the child is awake and asleep. Some children may show excessive mood swings from lethargy to hyperactivity, whereas others are even-tempered and react appropriately. One should ask the parents whether the child is friendly, outgoing, and sociable, or shy and withdrawn, particularly in group situations. Parental guidance may be needed to encourage interactive behavior by the child. Compliance or problems with obedience, daily activity level, attention span, sleeping and eating habits, and special interests and dislikes are revealing information. Separation from the parents may be a problem for some children with disability. The parents may be uncomfortable to leave the child with relatives or other caretakers. In this context, it is important to point out the need and possible approaches to foster the child's independence.

EDUCATIONAL AND SOCIAL HISTORY

Very young children may be enrolled in an early intervention program, either home- or center-based. Frequency, length of sessions, components of training, the child's tolerance and cooperation in the program, and its effectiveness, as perceived by the parents, should be clarified. The same applies when the slightly older child attends a preschool program. In school-age children, information about the type of class—mainstream, integrated, or special education—is important. Academic expectations are different in each of these educational pathways and should be taken into consideration when report card grades are interpreted. Individualized Education Program (IEP) meetings and environmental accommodations are other pertinent details. The child may have special interests and strengths that should be further developed or difficulties in certain subjects, which may require additional help and adjustment of the IEP. Review of educational status is a consistent part of follow-up visits, and assistance should be offered when problems arise, including cognitive and neuropsychological testing.

Opportunities to meet and play with other children in addition to school or home contacts, visits and sleepovers with friends, and participation in various recreational activities are formative experiences that prepare all youngsters for social functioning and adulthood. Asking the parents to describe the child's daily schedule, including regular and occasional activities on weekdays and weekends, yields a valuable insight into these aspects of the entire family's lifestyle. Time spent in school, therapy, homework, play, and leisure activities with family members, friends, or alone should be noted. Housing, employment of the parents, siblings and their ages, and social support of the family provide further understanding of the physical and social environment. Some families with a disabled child experience social isolation. Information about or referral to community resources may be helpful for many families. This may include recreational and sports training and teams, theater, music, and social outings.

FAMILY HISTORY

In motor or other developmental disabilities, a detailed family history must be obtained to explore the possibility of a genetic disease. Health and function of the parents, siblings, and other family members on the maternal and paternal sides should be explored through several preceding generations. One should ask specifically whether there are other children in the family with developmental delay or adults with known motor disability, limb deficiency, or other malformations. Historical information is at times incomplete until further questioning brings to light additional facts. Family albums and pictures of relatives may be helpful to detect dysmorphic facial or other features. Consanguinity is an increased risk for genetic disease, especially diseases with an autosomal recessive inheritance pattern. In some autosomal-dominant conditions, mild variants of a disease may be missed until a thorough investigation of suspected family members is carried out, including congenital myotonic dystrophy and facioscapulohumeral dystrophy. In X-linked conditions, affected males typically have a maternal familial history. Diseases with multifactorial inheritance, such as spina bifida, are complex, and may or may not have a known familial history (2,8). Referral for genetic workup is necessary whenever there is the possibility of a genetic condition. Pregnant mothers of affected children should be referred for genetic counseling. Prenatal diagnostic testing is available.

STUDIES

Radiographic imaging is useful for a variety of children. For those with bone abnormalities, plain films will be

helpful. For those with brain involvement, computed tomography (CT) and MRI may add to the diagnostic specificity. Electrodiagnostic testing is helpful in those with brachial plexus palsy or other nerve or muscle lesions. Electrodiagnosis is used less as genetic evaluations become much more specific for muscle and nerve diseases. Chromosomal and DNA testing are available for many diagnoses. Prenatal chromosomal testing, fetal DNA from the maternal bloodstream, newborn screening, and carrier screening are all available, with advances in techniques continuing.

EXAMINATION

This chapter provides only general guidelines for the format and structure of the pediatric rehabilitation examination at different ages. Specific details of diagnostic signs and interpretation of findings are discussed in subsequent chapters about different disabilities.

OBSERVATION

The examination begins as soon as the family and child enter the examination room, before the child is actually touched or asked to perform. Sometimes, it may be the most informative phase of the examination. Specific behaviors to observe and note include reaction to separation from the parents (especially in young children); apparent visual and auditory awareness; temperament (calm or hyperactive, compliant, or challenging); spontaneous exploration and interest in toys, games, or books in the room; style, concentration, attention span, or distractibility during play; level and manner of motor activities; attempts to engage the parents and the examiner in conversation, vocabulary, complexity of language, and quality of speech; and interaction with parents or examiner (appropriate, shy, or demanding). Observations of the parents' response and their way of handling the child's behavior are also revealing.

EXAMINATION BY AGE

For infants and young children, the examiner must create an atmosphere of trust. Friendly advances during history taking or while the child is at play allay initial fears and anxiety. At this age, most, if not all, of the examination can be accomplished with the child in the parent's lap if the child remains fearful. Interactive play in this phase of the examination can incorporate developmental testing by offering toys for grasping or raisins to test pincer grasp. Blowing bubbles makes most youngsters comfortable and happy, and one can assess their visual motor coordination as they reach to pop them. Balls are also extremely versatile and nonthreatening items to engage a child in his or her exam in a variety of ways. Hearing, vision, cranial nerves, and postural abnormalities also can be observed.

As the parent gradually undresses the child, gentle touch and tickling or funny sounds with a smile help to maintain relaxation and to facilitate hands-on examination. Inspection and palpation of body parts and gentle movements to examine tone are performed at this point. The examiner should be prepared to improvise if the child shows increasing anxiety.

The actual hands-on examination, consisting of bodily handling and manipulation, is the last stage; anxiety-provoking or painful tests are deferred to the end. If the examination requires placement of the child on a table, the parent can sit at the end and let the child's head rest on his or her lap. With anxious children, performance of gross motor activities, such as sitting, crawling, standing, or walking, also can be conducted with the family's help. One should note the quality of movements, postures, weakness, incoordination, asymmetry, or reflex abnormalities that reflect a motor deficit. Range of motion, deep tendon reflexes, or primitive reflexes that need physical manipulation should be examined after evaluation of active mobility. Tests that require instrumentation, such as sensation, fundoscopy, otoscopy, and oral function, conclude the examination.

Giving choices involves the preschool child in the examination. For example, the examiner may ask, "Should we look at your arm or leg first?" On the other hand, questions such as "Can I look at your arm?" should be avoided because if the child says "no," confrontation results. Parents can often bring out many capabilities of their children without the examiner touching them.

SCHOOL-AGE AND ADOLESCENT PATIENTS

The customary method of systematic medical examination is applicable. Children with cognitive deficit need to be approached according to their cognitive rather than chronological age. Children in this age group, particularly adolescents, are usually embarrassed about walking in underwear in front of their parents. Shorts or a bathing suit is more acceptable. Adolescents may be seen with and without their parents. Their concerns may be different from those of the family and should be addressed with respect for their privacy.

The scope of the examination is expanded to reflect the growing child's increasing functional needs in activities of daily living (ADLs) and other areas of competence. A comprehensive examination includes screening in educational achievements, reading, writing, and arithmetic. Formal psychological or psychoeducational testing follows in case of deficits.

GROWTH

Parameters of physical growth should be routinely measured on each visit and plotted on the standard growth chart. Height and weight are obtained at all ages, and head circumference is measured in children under 3 years and thereafter in children with deviations. Serial monitoring is necessary in hydrocephalus, regardless of etiology, and microcephaly, which reflects defective brain growth. In spina bifida and other disabilities that require full-time wheelchair use, arm span measurement is recommended instead of height (8). Extremity length and girth are recorded in children with localized growth disturbance due to neurogenic weakness, epiphyseal fracture, or arthritis. In growth disturbances that involve one side of the body, one must determine whether the condition represents hemihypertrophy or hemiatrophy. Hemihypertrophy unrelated to neurologic causes requires investigation for renal tumor.

INSPECTION

General appearance and special features may help to establish a diagnostic entity. Dysmorphic facial features, epicanthal folds, increased intercanthal distance, external ear anomalies, and malformations of the toes or fingers suggest a prenatal disorder, possibly teratogenic or genetic, and at times, an identifiable syndrome (12). Blue sclerae are a sign of osteogenesis imperfecta. Asymmetric facial and palpebral fissures and pupils may indicate facial palsy or Horner's syndrome, whereas craniofacial asymmetry and vertical strabismus may be present in torticollis. Dolichocephaly is typical in premature infants and children. A bald spot or area of short, thinning hair over the posterior skull is commonly a sign of weak neck muscles, most likely associated with generalized weakness, and may also be seen with torticollis. Extraocular, facial, and tongue muscle weakness may represent cranial nerve dysfunction, myopathy, or other neurologic disease. Involuntary eye movements and nystagmus are noted in cerebellar or other CNS disorders.

The skin should be inspected for telangiectasias, nevi, or other lesions. Café au lait spots or pigmented skin areas are seen in neurofibromatosis. In children with ataxia, telangiectasias may be seen over the flexor surface of the knees and elbows. Malar rash suggests a rheumatic disease. The combination of adenomatous rash, seizures, and hemiplegia is seen in tuberous sclerosis. Hairy patches, dimples, or other skin lesions over the spine are frequent signs of spina bifida occulta (8). A small sinus, dermal tract, or pilonidal cyst in the gluteal crease also may accompany occult spina bifida. Sudden weakness in such cases may indicate an infection penetrating into the spinal canal or a neurologic complication related to underlying malformation in or around the spinal cord. In children with sensory deficit, the involved area must be routinely examined for skin lesions, pressure abrasions, ulcerations, and infections. Foot deformities, varus or valgus deformity, or claw toes lead to abnormal weight distribution and callus formation consistent with the pathologic posture. Calluses over the dorsum of the feet and knees, the so-called "housemaid's knee," develop in older children whose preferred mode of locomotion is crawling. Multiple scars, bruises, and abrasions in various stages of healing may indicate frequent falls or child abuse/nonaccidental trauma (NAT).

Asymmetry in the size of skeletal muscles should be noted in terms of location and distribution. Anterior axillary and upper chest muscle atrophy may represent absent pectoralis muscle or atrophy due to a brachial plexus injury. Congenital clubfeet or multiple joint deformities are manifestations of prenatal muscle weakness due to spina bifida, arthrogryposis, or myotonic dystrophy, other muscle diseases, or may be idiopathic. A hypertrophic, "muscle-bound" appearance is a sign of myotonic dystrophy. Deformed, fusiform, dimpled joints may be seen in arthrogryposis. Lower extremity joint positions reflect the distribution of muscle weakness in newborns with spina bifida. Pseudohypertrophy of the calf muscles is an early sign of Duchenne muscular dystrophy. Symmetrical, well-developed musculature of the shoulder girdles and upper extremities is a convincing indication of functional crutch walking or effective wheelchair locomotion. An enlarged limb with bruit detectable by palpation or auscultation may signal an arteriovenous shunt and increased blood flow in the extremity.

Flaring of the ribs, or the so-called bell-shaped chest, suggests ineffective intercostal muscle function in children with motor unit disease or high spinal cord dysfunction. In scoliosis, the thoracic cage may develop asymmetry.

PALPATION

In infants and young children, the fontanelles and cranial sutures should be palpated for patency, tension, and size with the child in sitting position and while the child is quiet and not crying. A tense fontanelle in a vigorously crying child does not necessarily mean increased intracranial pressure. In the case of ventriculoperitoneal shunt, the reservoir may be checked for ease of emptying and speed of refill. Skin should be felt for texture, temperature, and absent or excessive perspiration. Pseudomotor paralysis in spinal cord injury eliminates sweating below the level of the lesion, and compensatory excessive perspiration occurs above the level of the lesion with high environmental temperature. Vasomotor dysfunction with coldness to touch and paleness or slight cyanosis of the skin may be present in severe upper motor neuron impairment. It is seen in the lower extremities of some children with cerebral palsy. Subcutaneous abnormalities

may be palpable, such as hard calcific deposits in dermatomyositis or neurofibromatous nodules along the course of peripheral nerves. When arthritis is suspected, each joint should be felt for the cardinal signs of inflammation, warmth, discomfort, and swelling due to synovial thickening and effusion.

Much can be learned from palpation of muscles. Tone and bulk are reduced in lower motor neuron paralysis; in long-standing denervation, the muscle tissue feels less resilient and fibrotic. The pseudohypertrophic calf muscles in Duchenne muscular dystrophy have a typical rubbery, doughy, hard consistency. A fibrotic nodule may be palpable in the sternocleidomastoid muscle in congenital torticollis. In an infant who has an isolated knee extension contracture, a palpable nodule in the quadriceps indicates fibrotic muscle changes at the site of previous repeated intramuscular injections. Localized pain and swelling accompany injuries to soft tissue or bone. Osteoporotic fractures in lower motor neuron lesions with sensory deficit show swelling but are painless. Tenderness in multiple muscle groups with weakness, fatigue, or skin rash is suspicious for myositis due to collagen disease or parasitic or viral infections.

ORGAN SYSTEMS

Although the primary health care of children with disabilities remains the responsibility of the pediatrician, the pediatric physiatrist should perform a selective general physical examination. The emphasis is placed on organ systems that are at increased risk in certain handicaps and may affect both overall health and successful rehabilitation.

Vital signs, including blood pressure and heart rate, are obtained in all patients. In myopathies and collagen diseases, cardiac auscultation should be performed because of the possibility of associated heart disease. In a child with developmental delay, the presence of a heart murmur may suggest an undiagnosed syndrome. Blood pressure monitoring is particularly important in spinal cord injury, neurogenic bladder, Guillain–Barré syndrome, and residual poliomyelitis, as well as in children receiving stimulant medications.

In disabilities that cause ineffective ventilation and involve the risk of minor aspirations, auscultation of the lungs may be revealing. Myopathies, thoracic spinal cord dysfunction due to injury or malformation, severe spastic quadriparetic cerebral palsy, and any disability with oral motor dysfunction are such indications.

Abdominal and rectal examinations are essential in children with neurogenic bladder and bowel dysfunction to evaluate bladder distention, bowel or rectal impaction, and anal sphincter tone. Stool consistency, intermittent or continuous bladder incontinence, and gross appearance and microscopic examination of the urine should be noted. Umbilical movements in response to eliciting superficial abdominal reflexes help to delineate the spinal cord level in thoracic lesions. Absent abdominal muscles result in loose skin folds resembling a prune; seen in prune-belly syndrome.

NEUROMUSCULAR SYSTEM

Examination of neuromuscular function consists of testing reflexes, tone, active motion, strength, and coordination. Limited understanding and cooperation in infants and young children requires adaptation of traditional methods of testing. After 4 to 5 years of age, the standard examination is generally applicable.

In infancy, reflex testing includes age-appropriate responses that reflect early immaturity and subsequent maturation of the CNS. In newborns and young infants, state of alertness, activity, and comfort influence muscle tone (13–16). If the baby is anxious, upset, restless, or crying, this part of the examination should be postponed. Valid assessment may require several attempts. In the first few months of life, flexor tone predominates. Hypotonia or hypertonicity signals neurologic abnormalities. Increased tone is the symptom of corticospinal or basal ganglion damage. Myopathy, cerebellar dysfunction, and lower motor neuron lesions due to anterior horn disease, neuropathy, or spina bifida all can result in hypotonia. However, a hypotonic stage usually precedes the appearance of increased tone in anoxic brain damage (17). This stage of hypotonicity tends to last longer in dyskinetic cerebral palsy than in spastic types. On passive motion of hypotonic muscles or extremities, no resistance is felt. The infant with generalized hypotonia is limp and floppy with handling and, in severe cases, may feel like a "rag doll"—a descriptive term for this finding. In hypotonia related to motor unit disease or lower motor neuron lesion, deep tendon reflexes are diminished or absent. In contrast, they are present or increased in floppy infants during the transient hypotonic phase of CNS damage (17).

Spastic hypertonicity and related postures are influenced by position in space and the effect of gravity. The child should be examined in supine, prone, and vertical positions to elicit typical postures. Perhaps the most dramatic yet common example of this is the increased hip adduction into scissoring, extension of the legs, with plantar flexion of the feet when a child with spastic cerebral palsy is suddenly lifted into vertical suspension. Resistance to both slow and fast stretching of muscle should be tested to differentiate rigidity from spasticity (18). In infants and young children, one may use a number of developmental reflexes to examine active movements and strength (19). The Moro reflex includes shoulder abduction followed by forward flexion of the arm. Eliciting palmar or plantar grasp reflexes demonstrates finger or toe flexor function. Asymmetric responses in the upper extremities may suggest Erb's paralysis or hemiplegia. Unilateral or bilateral absence of protective extension

response is likewise suggestive of weakness in the respective extremity. A 4-month-old infant elevates the head and trunk on extended arms in the prone position. Scapular winging during this activity is a sign of a weak serratus anterior muscle (20). In older children, the wheelbarrow maneuver demonstrates the same finding (20). Lifting up under the axilla elicits spontaneous active shoulder depression. When these muscles are weak, the shoulders slide upward, virtually touching the ears. These signs suggest proximal weakness, possibly due to myopathy.

Young children often adopt ingenious substitutions or compensatory movements to cope with weakness of particular muscles. With weakness of the deltoid, they may fling the arm forward by momentum or substitute the long head of the biceps for shoulder flexion. In advanced shoulder and elbow weakness, they may "walk up" the arm on the torso, using their fingers to get the hand to the mouth. Combat crawl is a usual way of crawling in lower extremity paralysis. Deformities around a joint commonly reflect an imbalance of strength in muscles acting on the joint. The deformity is in the direction of overpull. Such imbalance may be spastic or paralytic.

Visual observation during performance of functional activities to detect muscle weakness should consider the child's age and the achievements expected for the child's developmental stage. Walking on tiptoes, squatting and rising without using the arms for assistance, and straight sitting up from the supine position without rolling to the prone position or to the side are mastered by children around 3 years of age (21). Thus, the inability of younger children to perform these activities in a mature pattern should not be interpreted as weakness of the plantar flexors, hip and knee extensors, or abdominal muscles. Testing for Trendelenburg's sign and grading the triceps surae by having the child rise on the toes of one leg must be deferred until 4 years of age, when children develop adequate balance.

The standard technique of manual muscle testing can be used after school age, except in children who have serious behavioral problems or cognitive impairment (22–25). The customary grading system of scores from 0 to 5 or 0 to normal is used. Above fair grade, the wide range of normal variations in growth patterns should be considered in judging good versus normal strength. Because children are adept in using substitution for movement, the examiner must pay special attention and adhere to precise technical conduct of testing individual muscles. Side-by-side comparison may detect even mild neurologic weakness, although disuse atrophy or mild bilateral neurologic weakness may escape detection. Quantitative strength determination with comparison of both sides is helpful to demonstrate unilateral disuse atrophy in such strong muscles as the quadriceps. This determination is particularly advisable in teenage athletes after knee injury. Resumption of training for competition before virtually equal bilateral quadriceps strength is regained predisposes them to recurrent injuries. Testing of strength in upper motor neuron lesions requires the well-known considerations for position in space and orientation of head and major joints, which may affect recruitment of motor units and produce synergistic movement patterns.

A common sign of central movement disorders is impaired coordination. Proprioceptive sensory loss or parietal lobe syndrome may contribute to incoordination. Movement abnormalities associated with cerebellar dysfunction, basal ganglion disease, dyskinetic disorders, or spastic incoordination present with specific distinguishing signs. The detection of a coordination deficit is based mostly on observation of gross and fine motor function in children less than 2 to 3 years of age. Concurrent mild delay of motor development is not unusual. After 3 years of age, the examination becomes more specific for testing the quality of performance in complex and more advanced developmental skills. Around 3 years of age, the child can walk along a straight line, unsteadily placing one foot in front of the other. In comparison, facility at tandem walking at 5 years of age is a good illustration of continuing refinement of motor skills with age. The pediatric physiatrist may be asked to evaluate the appropriateness of coordination in children without an overt physical disability (26). Clumsiness of handwriting and drawing, difficulties in physical education or sports, and other subtle signs may be present. Such children may have a motor incompetence of apraxic nature, sometimes related to visuomotor perceptual deficit (27). It also may be associated with learning and behavioral dysfunction. A number of tests are available for examining motor proficiency and dexterity in children without physical disability (28,29). Tasks to evaluate youngsters with minor neurologic dysfunction include imitation of gestures (30), hopping (31), handclapping (32), and pegboard performance (33,34).

MUSCULOSKELETAL SYSTEM

Examination of the musculoskeletal system includes inspection and palpation of bones and soft tissues, measurement of active and passive joint range of motion, and assessment of stance and gait (35–38). It is complementary to neuromuscular assessment. As in previous parts of this chapter, only developmental variations are discussed.

Bone configuration and joint mobility change during the growing years (39,40). Full-term infants may lack as much as 25 degrees of elbow extension because of predominant flexor tone. In contrast, joint hyperextensibility and hypotonia allow increased passive motion in preterm infants. The scarf sign is a good illustration of excessive joint mobility in premature babies. Holding the infant's hand, the examiner draws one arm across the chest, like a scarf, toward the contralateral shoulder. In premature infants, the elbow crosses the midline, indicating hypotonic laxity of the shoulder and elbow joints. Full-term

neonates have incomplete hip extension with an average limitation of 30 degrees as a result of early flexor tone predominance (39,40). The limitation decreases to less than 10 degrees by 3 to 6 months. At birth and during early infancy, hip external rotation exceeds internal rotation (39,41). With the resolution of early hip flexion attitude, internal rotation gradually increases. Differences between bilateral hip abduction, apparent shortening of one leg, and asymmetric gluteal and upper thigh skin folds are highly suggestive of congenital or acquired hip dysplasia or dislocation (40). Alignment of the femoral neck in neonates is consistent with prenatal coxa valga and increased anteversion. Femoral inclination is 160 degrees, and the angle of anteversion is 60 degrees. Respective adult measurements of 125 and 10 to 20 degrees develop postnatally and are accelerated by weight-bearing.

Persistent fetal configuration in nonambulatory children with physical disabilities enhances the effect of neurogenic muscle imbalance on the hip joint and contributes to acquired hip dislocation in spina bifida and cerebral palsy. The popliteal angle is 180 degrees in the hypotonic preterm infant, compared with 90 degrees in full-term neonates. A combination of increased flexor tone and retroversion of the proximal tibia causes this limitation of knee extension in mature newborns. By 10 years, tibial retroversion resolves spontaneously. An early varus configuration of the tibia contributes to the physiologic bowleg appearance in infancy and corrects itself by 2 to 3 years of age. A systematic review of skeletal development, with examination of the spine and extremities, is presented in Chapter 10.

Normal variations of stance and gait should not be mistaken for pathology in the growing child (37,42,43). Gait abnormalities evident on clinical observation include asymmetric stride length and stance phase in hemiparesis; toe walking and scissoring with lower extremity spasticity; crouch posture and gait in diplegic cerebral palsy; Trendelenburg's gait in motor unit diseases and hip dislocation; gastrocnemius limp with lack of pushoff in L4 to L5 weakness due to spina bifida; and various types of gait deviations associated with involuntary movements, such as ataxia, tremor, or dyskinesias, in dysfunction of the CNS.

SENSORY EXAMINATION

A complete examination of all peripheral sensory modalities is possible only in older children (44). Nevertheless, some modalities can be tested in infants and young children, and provide significant information. An infant who cries and squirms to move away from pinprick obviously perceives pain (45). A sleepy infant may be slow to respond and requires repeated stimuli. Withdrawal of the leg from painful stimuli may represent the triple flexion spinal withdrawal reflex in thoracic spinal cord lesion and should not be mistaken for active movement and presence of sensation. Comparing the infant's reaction to pinprick on the arms or face differentiates actual sensory perception in such cases. Older infants respond to touch and vibration by turning toward or moving away from the stimulus. The presence of superficial reflexes signals an intact afferent and efferent reflex arc. The neurosegmental levels are T8 to T12 for abdominal reflexes, L1 to L2 for the cremasteric reflex, and S4 to S5 for the anocutaneous reflex. In spina bifida, absence of these reflexes generally coincides with sensory deficit in the respective dermatomes. In young children who cannot be tested for proprioceptive function, ataxia and incoordination may suggest absence of this sensation. Testing of position sense is usually reliable by school age.

Cortical sensory function is impaired in parietal lobe damage (44,46). The most frequent childhood example is hemiparetic cerebral palsy. Disproportionately poor spontaneous function, neglect, and visual monitoring during use of the arm and hand are suspicious signs. Objective evaluation is generally feasible after 5 to 6 years of age, using the same technique as in adults for stereognosis, two-point discrimination (47), and topognosia with single or double sensory stimulation. Testing for graphesthesia may be attempted by using a circle or square. Around 8 years of age, the traditional number identification gives more accurate information. Cutaneous sensation and proprioception must be intact, and adequate cognitive ability is a prerequisite for testing cortical sensory function.

The child's age and ability to cooperate need to be considered in the examination of special senses. Moving a bright light or attractive object across the visual field is used to test vision in infants. At 1 month, the infant will follow to midline and at 3 months, from side to side through a 180-degree arc. The Stycar test and the illiterate E chart are used for screening preschool children at risk for visual deficit (48,49). At an early age, unilateral impairment or loss of vision and visual field defects, such as hemianopsia, are more likely to remain undetected than bilateral deficits. A child with strabismus or suspicion of diminished vision should see an ophthalmologist as soon as the problems are discovered. Early treatment with eye patching or corrective lenses is necessary to prevent amblyopia ex anopsia, or suppression amblyopia, partial loss of vision caused by cortical suppression to prevent diplopia (50,51). Central dysfunction of visual attentiveness, discrimination, and information processing may be misinterpreted as diminished vision and require both ophthalmologic and neuropsychological investigation. Cortical visual impairment usually shows improvement over time.

Screening of auditory function is a routine procedure in the neonatal nursery, pediatric office, and school. The examination of handicapped infants and children also should include a simple screening of hearing, eliciting the blink or startle reflex. Responses to handclapping; to speech of conversational loudness or whisper; perception

of finger rubbing near the ear; and reaction to tuning fork, bell, or cricket toy are methods of testing. Absent, lost, or delayed speech, articulation deficits, inattentiveness to sound, a history of recurrent otitis media, head injury, or failure to pass the screening test indicates a need for complete evaluation of auditory function (45,50,52,53).

FUNCTIONAL EVALUATION

The pediatric rehabilitation examination is meaningless if the physiatrist does not construct from it a coherent picture of the child's functional achievements. This evaluation both complements and integrates the variety of information derived from all phases of the examination.

The developmental diagnostic evaluation is a convenient, functionally oriented assessment tool for infants and preschool children (21,54). Language, fine motor and adaptive skills, gross motor abilities, and personal–social behavior are the four major areas of function in the organizational framework of developmental testing. The same functional domains are considered in the evaluation of older children and adolescents. However, in these age groups, the examination includes a wider range of developmental expectations and abilities to function in school and society. ADLs and gross mobility skills need to be assessed in this context. In addition to speech, testing of language function includes other modes of communication: reading, writing, spelling, and, if indicated, augmentative communication. Drawing, design construction, arithmetic problems, and questions about handling hypothetical situations in daily life offer a brief, preliminary insight into cognitive and learning abilities. A number of specific assessment instruments were designed for various childhood disabilities (55–58). These instruments are useful functional assessment tools for their designated conditions and appropriately complement the customary developmental evaluation.

INFORMING INTERVIEW

Informing the family about the findings of the examination and their implications is an important responsibility of the physician. Factual information must be imparted with a caring attitude. Informing the parents about a newly established diagnosis should be considered as crisis intervention. A diagnostic label is insufficient without explanation of its meaning. The parents need to know the estimated prognosis, including the uncertainties of early prognostication, particularly in CNS dysfunction, with the possibility of multiple handicaps. Future needs in care and functional rehabilitation should be outlined. One should emphasize the need to avoid focusing on the physical disability alone and to consider the child's developmental and social needs. Effective counseling and communication skills are essential for establishing a partnership between the physician and family to ensure the successful outcome of a comprehensive rehabilitation program. Information for ongoing support is crucial.

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2

MEDICAL CARE OF CHILDREN WITH DISABILITIES

Susan D. Apkon and Emily Myers

Children with special health care needs (CSHCN) are a population who have been or are at risk for a chronic physical, developmental, behavioral, or emotional condition, and require health and related services of a type or amount beyond that required by children generally (1). An estimated 15.1% of children in the United States had a special health care need in a 2009 to 2010 survey (2). Due to this high prevalence, primary care providers (PCPs) and pediatric subspecialists are commonly called to care for this group of children. Routine health maintenance visits are frequently omitted in favor of visits for acute illnesses, which can result in a failure to discuss routine health care issues, such as growth and development, immunizations, vision, hearing, and dental care. Evaluation for acute illnesses in this population poses unique challenges to care providers due to the oftentimes extensive past medical and surgical histories, lengthy lists of medications, and the atypical presentation of typical childhood illnesses. This chapter will focus on a discussion of the provision of medical care to CSHCN using a medical home model, the routine health maintenance issues for children with disabilities, and the common acute medical issues that a PCP may be asked to evaluate in this group of children. Finally, this chapter will discuss the issue of palliative care for CSHCN.

MEDICAL HOME

The concept of a medical home has long been endorsed by the American Academy of Pediatrics (AAP) as the optimal model for the provision of primary care for all children. The definition of medical home includes six attributes: (a) a usual place for both acute and preventative care; (b) a personal nurse or doctor; (c) no difficulty receiving referrals to subspecialists; (d) appropriate care coordination; (e) family-centered care; (f) services to provide for necessary transition to adulthood that improve treatment adherence, decrease health care disparities, and access preventive health services (3). These six attributes are used to measure the success of state Title V programs at achieving Healthy People 2020 goals.

Beyond the provision of acute and routine medical care, the medical home can provide both "vertical links" within the medical system and "horizontal links" to the wider community. Within such a network, families should feel that they have a supportive, effective, informed, and caring network to rely on to help them meet the acute, chronic, and often unanticipated problems faced by their CSHCN.

Families rely on the primary care physician to make appropriate referrals to and communicate with the multiple subspecialists who also provide care to many of these children. It can be of enormous benefit to have a designated individual in the office or clinic who is able to coordinate multiple appointments on the same day, thus lessening the burden of travel for these families. Having translator services available, as well as written materials in the family's primary language, is an additional benefit.

CSHCN often require therapeutic as well as supportive services. Examples of therapeutic services include home nursing; physical, occupational, or speech therapy; and in some cases, mental health services. Supportive services may include the provision of letters of medical necessity, assistance with transportation, acquisition of durable medical equipment, provision of information regarding financial entitlements and respite care, connections to community support groups, and communication with schools. Knowledgeable individuals within the medical home, typically experienced registered nurses and social workers, should facilitate care coordination. These services play a pivotal role in decreasing the care burden on the family, promoting maximal independence of the child and enabling full participation in community life.

Unfortunately, approximately one-half of CSHCN do not have access to a medical home (4). Less than 25% of children who require supportive and therapeutic services have a high-quality medical home (5). Ongoing advocacy for a medical home is critical to improve family and patient health outcomes.

DISABILITY CHARACTERIZATION

The International Classification of Functioning, Disability and Health, known more commonly as ICF, is a classification of health and health-related domains. The components focus on body function and structure, activity and participation, and additional information including severity and environmental factors. Care providers should be familiar with standardized tools that capture the ICF components with a focus on cognitive and learning, motor, speech–language, and social and adaptive skills. These assessments are used by medical, educational, psychological, and therapeutic care providers to determine and advocate for particular educational and state supports. The focus on the following dialogue will be on assessments of motor and speech function.

Speech and language impairment can dramatically affect an individual's ability to communicate his or her wants and needs. Characterization of speech and language impairment provides means to tailor therapeutic programs and supports to meet an individual's needs. Table 2.1 lists some commonly used tools utilized by speech–language pathologists to measure the different components of these skills.

Motor skills have a significant impact on early childhood development, as early social and cognitive development partially depends on an individual's ability to explore his or her environment. Later, motor development can impact adaptive and social function. Assessing these functions in all individuals with disabilities and a therapeutic program focusing on the limitations optimize developmental and psychosocial outcomes. Table 2.2 lists common pediatric assessment and categorization tools for individuals at risk for disability.

ROUTINE HEALTH MAINTENANCE

CSHCN need to see physicians more frequently, and have seven times the number of nonphysician visits than typically developing children (6). For this diverse group of individuals, acute care concerns often distract PCPs and caregivers during these health surveillance visits. The PCP must ensure that routine health care needs are being addressed. The following are examples of health care topics that should be addressed during routine visits.

Care Notebooks are very helpful in the ongoing management of children with multiple and complex medical needs, and should be reviewed and updated at routine visits. The AAP provides specialized forms for families and medical personnel to build a Care Notebook. Care Notebooks include a child's medical history, current medications, past medical complications and their typical presentations, and a treatment plan based on presenting signs and symptoms (7). An up-to-date Care Notebook provides medical providers important information, allowing for safe and efficient care. Table 2.3 details a list of commonly used medications that are regularly utilized among individuals with CSHCN and are frequently listed in Care Notebooks.

GROWTH AND NUTRITION

Assessment of growth is a fundamental component of the routine physical examination of children. Length, weight, and head circumference should be obtained at each health maintenance visit. Obtaining a weight on a child who is unable to stand on the office scale is often accomplished by having parents hold the child while stepping on the scale themselves. This is more effective in a smaller child, but is more difficult in a larger child or one with severe spasticity or hypotonia. It is recommended that an office that cares for large numbers of children who are nonambulatory obtain a wheelchair scale, which allows the child to be weighed easily in his or her own wheelchair. Assessment of the length of a child is also problematic when the ability to stand is limited. Use of arm span as a substitution for height may be an acceptable option. Alternatives to standing height also include individual measurements of lower extremity segments when significant joint contractures are present. To obtain segmental measurements, the child is placed in the supine position

TABLE 2.1 COMMON MEASURES OF SPEECH AND LANGUAGE

Clinical Evaluation of Language Fundamentals, Fifth Edition (CELF-5) (63)	Expressive and Receptive Syntax, Semantics, Morphology	Used for age range 5 to 21 years
Comprehensive Assessment of Spoken Language (CASL) (64)	Expressive and Receptive Syntax, Semantics, Morphology, Pragmatics	Used for age range 3 to 21 years
Peabody Picture Vocabulary Test, Fourth Edition (PPVT-4) (65)	Receptive Vocabulary	Used from 2 years 6 months to adulthood
Preschool Language Scale (PLS-5) (66)	Receptive and Expressive Language	For use from birth to 7 years 11 months

TABLE 2.2 COMMON MEASURES OF MOTOR FUNCTION

INSTRUMENT	DESCRIPTION	COMMENTS
Gross Motor Function Classification System (GMFCS) (67)	Measures self-initiated movement status of individuals with cerebral palsy	Most useful for individuals between 2 years and 18 years of age Focus on functional limitations
Alberta Infant Motor Scale (AIMS) (68)	Designed to monitor and identify infants with gross motor delay	Useful for infants from birth to 18 months
Manual Ability Classification System (MACS) (69)	Describes how children with cerebral palsy use their hands to handle objects in their daily activities	Used for individuals 4 to 18 years
Beery–Buktenica Developmental Test of Visual–Motor Integration, Fifth Edition (BEERY VMI) (70)	Screen for visual–motor deficits	Used for individuals 2 years through adulthood
Bayley Scales of Infant and Toddler Development, Third Edition (71)	Cognitive, language, motor, social–emotional, and adaptive development measured	Used for children 1 to 42 months
Bruininks–Oseretsky Test of Motor Proficiency (BOT-2) (72)	Norm reference test of motor abilities	For children 4 to 21 years
Peabody Developmental Motor Scales (PDMS) (73)	Measures both gross and fine motor skills	Used for children from birth to 83 months of age
Motor Assessment of Infants (MAI) (74)	Measures muscle tone, primitive reflexes, automatic reactions, and volitional movements of infants who had been treated in the neonatal care unit	Utilized in the first year of life

on the examination table and the assessment is done by adding all of the measurements obtained from the head to pelvis, the pelvis to knees, and knees to feet. Use of knee height has also been used as another means of monitoring a child's growth (8–10).

Plotting a child's anthropometric data on a growth chart allows the PCP to track his or her nutritional status. A weight-to-length ratio below the fifth percentile may represent failure to thrive. However, growth velocity is the more important piece of information. Many children with disabilities will be below the fifth percentile for their age, but as long as their weight and length increase in parallel to a normal curve, growth may be appropriate. A child's age should be corrected for prematurity until 2 years of age. One must remember that some CSHCN have short stature as part of their disease process or syndrome. Special growth charts are available for children with Down syndrome (11), Turner syndrome (12), achondroplasia, arthrogryposis–amyoplasia, Marfan syndrome, Noonan syndrome, Prader–Willi syndrome, and Williams syndrome.

A nutritional assessment should be completed during routine health maintenance visits. When there is concern about a child's growth, a more careful investigation into the food intake is necessary. The amount, variety, and consistency of food eaten may provide the examiner with information regarding the caloric intake of the child. Assessment of possible nutritional deficiencies may provide useful information about general health. Following serum ferritin, vitamin D, and essential elements can be very important in individuals with developmental disabilities. The amount of food eaten is important, but the amount of time it takes a child to complete a meal is also essential. It is not unusual for a child with severe cerebral palsy (CP) or an infant with spinal muscular atrophy (SMA) to eat a meal over a prolonged period. The amount of energy to consume a meal may be significant. An assessment of oral motor function during eating should be performed at health supervision visits if there is a concern for feeding problems. Prevalence estimates suggest that 80% to 90% of individuals with developmental disabilities will experience dysphagia during their lifetime (13). Signs and symptoms of feeding problems include coughing or choking while eating, a wet vocal quality during or after the meal, poor sucking, gagging easily, and vomiting after a meal. A referral to a comprehensive feeding clinic should be considered if there is a concern about the weight of the child, or his or her safety while eating. An interdisciplinary clinic may include an occupational and speech therapist, nutritionist, gastroenterologist, and/or developmental or rehabilitation physician. Assessments used to evaluate feeding in this population include behavioral feeding examinations, oral motor feeding observations, and video fluoroscopic swallow studies.

TABLE 2.3 COMMON PHARMACOLOGIC THERAPIES FOR CHILDREN WITH SPECIAL HEALTH CARE NEEDS

MEDICATION (BRAND NAME)	INDICATION (MECHANISM OF ACTION)	CONSIDERATIONS
Polyethylene glycol (Miralax)	Constipation (osmotic laxative)	Can cause diarrhea, bloating, cramping
Docusate (Colace)	Constipation (surfactant laxative; stool softener)	Can cause diarrhea, bloating, cramping
Sodium phosphate enema (Fleets Enema)	Constipation (saline enema)	Can cause dizziness, rectal trauma, electrolyte disturbances
Odansetron	Nausea ($5HT_3$ receptor antagonist)	Can be sedating
Omeprazole (Prilosec)	Gastroesophageal reflux (proton pump inhibitor)	Give prior to meal on an empty stomach
Lansoprazole (Prevacid)	Gastroesophageal reflux (proton pump inhibitor)	Give prior to meal on an empty stomach
Ranitidine (Zantac)	Gastroesophageal reflux (histamine type 2 receptor antagonist)	Food does not interfere with absorption
Famotidine (Pepcid)	Gastroesophageal reflux (histamine type 2 receptor antagonist)	Food may increase bioavailability of medication
Metoclopramide (Reglan)	Gastroparesis (prokinetic agent)	Extrapyramidal symptoms more common in children
Erythromycin	Macrolide antibiotic (prokinetic agent	Limited data available
Levetiracetam (Keppra)	Antiepileptic	Metabolized in blood, metabolites renally excreted, lots of drug interactions, behavioral problems reported
Valproic Acid (Depakote)	Antiepileptic	Hepatotoxicity is the major side effect (is a substrate, inhibitor, and inducer of many liver enzyme pathways), also should be avoided in pregnancy
Oxcarbazepine (Trileptal)	Antiepileptic	Induces CYP3A4 liver enzyme, many drug interactions, metabolized by liver, hyponatremia reported and should be monitored
Phenobarbital	Antiepileptic, barbiturate	Interacts with many liver enzymes, is hepatotoxic, can get tolerant over time, very sedating
Topiramate (Topamax)	Anticonvulsant	Used for infantile spasms, seizures, and migraines, metabolic acidosis and nephrolithiasis reported
Gabapentin (Neurontin)	Antiepileptic, therapy for neuropathic pain	Metabolism unknown, many drug interactions
Melatonin	Sleep initiation agent	Occasionally can see paradoxically activating side effects
Trazodone	Antidepressant, serotonin reuptake inhibitor/antagonist, sleep adjunctive agent	Limited data available but used frequently in children with developmental disabilities, important side effect is priapism

IMMUNIZATIONS

Routine immunization against childhood diseases is recommended for all children with disabilities. The most current schedule can be obtained through the Centers for Disease Control and Prevention (CDC) and is approved by the AAP and American Academy of Family Physicians (14). Although children with disabilities are not necessarily at higher risk for contracting childhood infections, they may have greater morbidity and mortality when ill. Administration of diphtheria, tetanus, and pertussis (DTaP) and measles, mumps, and rubella (MMR) vaccines to children with a personal or family history of seizures is controversial as these vaccines can increase the risk of seizures (15). The seizures are typically short in duration, generalized, self-limited, and associated with a fever. Because the pertussis immunization is given during infancy, the onset of a seizure after the vaccine can be confusing. It is recommended that the DTaP vaccine be delayed until a complete neurologic evaluation is completed and the cause of the seizure determined. Conversely, the MMR vaccine should not be withheld because it is typically first given after the onset of infantile seizures and the etiology of the seizure is generally already known.

Special attention should be given to children who are immunocompromised. Children with physical disabilities, such as those with rheumatologic diseases and Duchenne muscular dystrophy (DMD) who are on chronic corticosteroids, are included in this special population. In general, it is not recommended that children who are immunocompromised from corticosteroid use receive live bacterial or viral vaccines. Although definitive guidelines do not exist, the current Red Book recommendation is that children receiving high doses of systemic corticosteroids given daily or on alternative days for more than 14 days not receive live-virus vaccines until 1 month after the discontinuation of the medications. High-dose corticosteroids are defined as dosages greater than 2 mg/kg per day or greater than 20 mg/day if the child weighs more than 10 kg. In the case of DMD, it is recommended that children receive all of their immunizations prior to the initiation of corticosteroids (16).

Immunization against influenza of CSHCN, families, and medical providers on a yearly basis decreases the potential devastating morbidity and mortality associated with this virus. Influenza immunization of all high-risk children older than 6 months of age and their close contacts should be strongly encouraged each fall (17). Children who are high risk include those with recurrent pneumonias or upper respiratory infections, and those with CP and neuromuscular diseases such as SMA, congenital myopathies, and muscular dystrophies. Chemoprophylaxis during an influenza outbreak is also recommended to decrease the duration the child is infectious. Children who may have increased risk from complications due to pneumococcal disease should receive the pneumococcal conjugate and/or polysaccharide vaccine (15). Children born prematurely or having certain cardiopulmonary diseases often qualify for respiratory syncytial virus (RSV) prophylaxis (15).

Among the general community, concern remains regarding risk of vaccines causing other developmental difficulties including autism spectrum disorder (ASD). Though there is no evidence to support these concerns, families continue to decline immunizations for their children. It is very important for providers to document families' concerns and attempt to sensitively counsel them regarding the state of the medical evidence and the risks posed to children and communities that do not immunize.

DENTAL CARE

Tooth decay is one of the most common diseases of childhood (18). Almost 80% of CSHCN in the United States are reported to need dental care (19), and as few as 10% of dentists report that they serve this population (20). Tooth decay and poor dental hygiene in children with disabilities are related to swallowing problems, drooling, and gastroesophageal (GE) reflux. Many medications are given with sweeteners to increase palatability, and increase risk of tooth decay; others cause gingival hyperplasia (eg, phenytoin) or decrease saliva production. Routine dental care of a child or adolescent with severe developmental disabilities may be challenging for parents and caregivers due to an oral aversion, a tonic bite reflex, or the inability of the child to follow instructions to open his or her mouth. Other daily care activities, such as administration of multiple medications or respiratory treatments, may make dental hygiene less of a priority. Once a child takes over the care of his or her own teeth, the quality of cleaning may not be optimal because of cognitive and physical limitations.

Dental health of children with CP compared to children with other disabilities is most frequently described in the literature. The incidence of dental caries in children with CP is similar to the general population, although the quality of the caries is different. The size of the carious lesions is greater than what is seen in typical children (21–23). Periodontal disease is more prevalent in children with CP, likely due to the presence of gingival hyperplasia from those receiving phenytoin (24). Malocclusion and developmental enamel defects were also more common in children with CP (25–29). Erosion of primary and permanent teeth has been attributed to chronic GE reflux. The severity of erosion has been correlated with the duration of the GE reflux disease, frequency of vomiting, pH of the acid, and the quality and quantity of saliva (30–33). Despite the fact that children with CP do not participate in high-risk activities as frequently as their able-bodied peers, dental trauma is more common (34,35). These

injuries, most commonly to the maxillary incisors, are related to trauma during transfers or falls.

There is little information about dental problems for children with spina bifida. An important issue that must be addressed at each visit is to ensure that the dental office or operating room provide a latex-free environment (36). Families may need to remind the dentist and hygienist of the child's risk for an allergic reaction to latex. Latex-free gloves must be available to reduce the risk of an allergic reaction. Boys with DMD can have malocclusion with anterior and posterior open bites, which are associated with lip incompetence, mouth breathing, and macroglossia. Deteriorating oral muscle function as the child gets older is associated with increased plaque and calculus formation and gingival inflammation, but not necessarily with the presence of dental caries (37,38). Boys with DMD have a greater risk of malignant hyperthermia when anesthesia is used for dental care (39,40).

Routine examinations and cleaning to maintain optimal dental hygiene should be performed by a dentist comfortable in the care of children with special needs. Some of the dental care may need to be accomplished under anesthesia in order to obtain the maximum benefit. Combining dental procedures with other necessary procedures, such as a brainstem auditory evoked response (BAER), local injections with phenol or botulinum toxin, or certain orthopedic procedures, may limit the exposure to anesthetic agents. The AAP Policy Statement on oral health care states that CSHCN be referred to a dentist as early as 6 months of age and no later than 6 months after the eruption of their first tooth, or 12 months of age (whichever comes first) (41). Visits will provide the dentist with the opportunity to provide specific education to the family to allow for optimal dental care.

VISION

Vision screening and eye examination should be a component of all routine health care visits. The AAP recommends that the evaluation begin in the newborn period and then at all subsequent visits, with the goal of identifying conditions that might result in visual impairments or represent serious systemic diseases (42). In the child with a disability, this is especially important, given the frequent association of visual disorders with neurologic diseases. The eye evaluation from birth to 3 years should include a vision assessment, which is accomplished by having the infant or young child fix on an object. The examiner assesses the child's ability to maintain the fixation and follow the object into different gaze positions, a skill that by 3 months of age is developmentally appropriate. Further evaluations of infants and young children should also include external inspection of the eye and lids, pupillary and red reflex examination, and ocular alignment. Assessment of the child older than 3 years should also include age-appropriate visual acuity measurements and an attempt at ophthalmoscopy.

Ophthalmologic disorders frequently seen in children with CP require very close follow-up with an ophthalmologist (43). Annual evaluation for cataracts should be completed in children with myotonic dystrophy or those on chronic corticosteroids, such as boys with DMD or a child with a juvenile rheumatoid arthritis (16,44). Detailed and accurate documentation of the ophthalmologic examination of a child with spina bifida can be helpful when assessing possible ventriculoperitoneal (VP) shunt malfunctions. For example, a malfunctioning VP shunt may cause papilledema or changes in extraocular movements. These are early indications that may manifest prior to more obvious signs, such as headaches, lethargy, or vomiting.

The eye examination of a child with a disability is best performed by a pediatric ophthalmologist due to the child's high risk for ophthalmologic problems. The ophthalmologists have the skill needed to obtain a thorough assessment. A referral to a pediatric ophthalmologist for specialized tests, such as an electroretinogram (ERG), may be useful in assisting with the diagnosis of rare neurologic conditions, such as mitochondrial diseases.

HEARING

Newborn hearing screening is the standard of care in the United States. In 1999, the AAP endorsed the implementation of a universal newborn hearing screening program (45). Two technologies are used for newborn hearing screening: BAER and otoacoustic emissions (OAEs). Periodic reassessments of children with disabilities are important, since these children are particularly at risk for hearing impairment, and hearing loss will affect their developmental skills.

PCPs should pay special attention to children with specific disabilities, as they are at greater risk for developing hearing loss. For example, children with Down syndrome are at increased risk of otitis media and concomitant transient conductive hearing loss (46). Children with congenital cytomegalovirus (CMV), both symptomatic and asymptomatic at birth, are at risk for progressive and late-onset hearing loss (47). Children with athetoid CP due to kernicterus have a high incidence of hearing loss, as do children who have been treated with ototoxic antibiotics for systemic infections (48,49). Children who have been placed on extracorporeal membrane oxygenation (ECMO) have ongoing risk for hearing loss and need regular hearing screening. Finally, all children showing signs of speech or communication difficulties should have their hearing screened.

ILLNESSES IN THE PRIMARY CARE OFFICE

Children with disabilities present to PCPs with the same childhood illnesses as their typically developing peers, but the presenting signs and symptoms may be quite different. Medical personnel should be aware of these differences in order to provide accurate and timely management. Referring to previous medical records is helpful in determining the unique risk factors for a particular child, and a patient's Care Notebook is a useful diagnostic tool as well.

The following section reviews specific acute and chronic medical concerns of CSHCN and strategies for the PCP to facilitate an appropriate diagnosis and treatment plan. For a more detailed list of commonly used medications in the primary care setting for treatment of a multitude of comorbid conditions for children with developmental disabilities, see Table 2.3.

RESPIRATORY COMPLICATIONS

Drooling

Difficulty in managing oral secretions in children with disabilities results from poor oral motor control. Parents may express concern over their child's drooling, frequent cough, or increased upper airway congestion. Management of drooling can be pharmacologic or surgical. Treatment is recommended when drooling causes significant skin irritation, social problems, or the child is having recurrent respiratory infections secondary to poor secretion management (50).

When oral secretions are copious, use of a suction catheter by caretakers can keep the oral cavity and upper airway clear. Families should be instructed in the appropriate technique of oral cavity suctioning and have a portable suction machine that can be used in and out of the home setting. Use of medications such as glycopyrrolate or the scopolamine patch can decrease the volume of secretions. The use of botulinum toxin injections into the submandibular and parotid glands is being recommended more frequently for children with CP (51). Surgical ligation of the glands is typically reserved for cases that are unresponsive to medications. As saliva is very important in the prevention of dental caries, care must be taken to maintain sufficient production of saliva to protect tooth health.

Drooling may indicate that a child is having difficulty with eating, drinking, or swallowing. In a child with a degenerative neuromuscular disease, such as SMA, the development of increased drooling or difficulty managing oral secretions should prompt a further investigation into his or her feeding status. A referral to a feeding team, and consideration of a swallow study should be initiated. Alternative feeding modalities, such as a nasogastric tube or a gastrostomy tube, may be necessary. Use of medications to dry secretions in a child with muscle weakness may be counterproductive, as thicker secretions may be more difficult to clear.

Respiratory Distress

Children with upper respiratory infections commonly present to their PCP with fever, increased work of breathing, and tachypnea. The evaluation and treatment of a child with a disability who presents with these symptoms should be similar to a typically developing child. However, the deterioration may be accelerated, requiring a rapid diagnosis and initiation of treatment. The assessment should begin with a review of vital signs, including pulse oximetry. The physical examination focuses on assessing the child's level of alertness, his or her work of breathing, and a chest examination. Children with neuromuscular diseases will frequently increase their respiratory rates in order to maintain oxygen saturation. Unfortunately, a child can decompensate quickly in this situation as a result of significant fatigue. Oxygen saturations can be falsely reassuring in the face of hypoventilation.

Diagnostic testing may include pulse oximetry, chest x-ray, venous or arterial blood gas, sputum culture looking for a bacterial etiology, and viral studies for identification of common viruses such as influenza and RSV. Viral etiologies are the most common causes of upper respiratory infections in both typically developing children and children with disabilities.

Use of antiviral medications should be considered in children with disabilities because of their high risk for significant morbidity. Enteral or parenteral antibiotics should be reserved for suspected bacterial etiologies. Coverage for anaerobic bacteria should be initiated when aspiration pneumonia is suspected.

A child with a neuromuscular disease, such as SMA or DMD, may need assistance with secretion mobilization and airway clearance. Secretion mobilization can be addressed with chest percussion or a vibratory vest, skills that a family should be comfortable performing. Airway mobilization can be accomplished with the use of a cough-assist machine. The in–exsufflator, a commercially available device that provides a positive pressure breath followed by a large exhalation, improves peak cough expiratory flow rates (52,53). Children and caregivers should be familiar with the different techniques and initiating their use at the first signs of a respiratory illness. When symptoms increase and evidence of hypoventilation is present, use of noninvasive and invasive respiratory support may be necessary. Noninvasive support may include negative pressure ventilation or positive pressure ventilation with bilevel positive airway pressure (BiPaP) (54). It is important for PCPs to have knowledge of the various options for respiratory support and to understand

the family's wishes on the extent of treatment the family wants in the case of acute decompensation. Acute events are less stressful when families and their PCPs have discussed their wishes while the child is well and prior to the event.

NEUROLOGIC COMPLICATIONS

Seizure Activity

Children with CP are at increased risk of having seizures (55). The PCP is frequently asked to evaluate a child who is having increased seizure activity. Identification of an intercurrent illness, which may lower the seizure threshold, and a review of adherence to the current medication regimen are critical questions that must be asked. Obtaining levels of the antiepileptic medication is useful in determining whether suboptimal levels are the etiology of the increased seizure activity and there is a subsequent need to increase the medication dose if levels are low. A recent increase in weight might trigger the need to adjust the current dose. A referral for an EEG and consultation with a pediatric neurologist may be generated if the pattern of seizures is determined to be changing. Empowering a family to treat seizure activity with fast acting benzodiazepine in the home setting is an important way to decrease the need for emergency room visits.

New-onset seizures in a child with a disability should be evaluated thoroughly. For example, a new seizure in a child with spina bifida and shunted hydrocephalus may represent a shunt malfunction with subsequent worsening hydrocephalus.

Spasticity

The majority of children with CP have spasticity as a component of their upper motor neuron disorder. PCPs are frequently asked to evaluate a child with increasing tone. The acute onset of increased tone may represent an intercurrent illness, such as an otitis media or a urinary tract infection, causing pain in a child, which is manifested as spasticity. The sole presenting signs of an acute fracture of an extremity in a child who is nonverbal may be increased tone. A careful assessment of all extremities is necessary when the diagnosis is unclear. Treatment of the increased spasticity should focus on treatment of the underlying illness. Use of antispasticity agents such as diazepam and baclofen may be a necessary adjunct when tone is markedly increased. Acute withdrawal from a malfunctioning intrathecal baclofen pump (see the chapter on cerebral palsy, Chapter 14) may present with increased tone, diaphoresis, tachycardia, hypertension, and irritability. The irritability may be related to the pruritus that is an idiosyncratic reaction not associated with a rash. PCPs who care for children with intrathecal baclofen pumps should be familiar with these common presenting symptoms and management of the withdrawal from baclofen. Immediate administration of oral baclofen or intravenous (IV) diazepam will help decrease the symptoms, but referral to a center that can evaluate and treat the malfunction is needed to resolve the problem (56).

Pain

Though there is relatively little literature addressing pain in children with disabilities, the data reports that individuals with a wide range of disabilities have increased risk for pain (57). The etiology of the pain experienced by individuals with disabilities is as varied as the types of disabilities themselves. For example, individuals with CP may have significant pain from muscle spasms or in the presence of fractures or pressure wounds. Individuals with CP also have increased risk of renal and ureteral stones when taking certain antiepileptic medications such as topiramate. Individuals with disabilities more frequently suffer from constipation and GE reflux, both of which when chronic can lead to significant discomfort. Individuals with Down syndrome have increased risk of middle ear dysfunction and ear infections. Individuals with disabilities also have increased numbers of necessary medical painful medical procedures.

It is the clinician's responsibility to assess for pain at every visit, and to address concerns raised by both the patient and the caregiver. Evaluation includes a thorough review of systems pertinent to the complaint, which can be more difficult in patients who are nonverbal. Consider thorough otolaryngologic, oral, neurologic, gastrointestinal, musculoskeletal, and urologic causes. Utilize an individual's Care Notebook for prior experiences as a guide for evaluation and management. People with disabilities are regularly undertreated for pain (57). Treatment should include management of the underlying problem as well as appropriate analgesia.

ORTHOPEDIC COMPLICATIONS

Fractures

The incidence of fractures associated with minimal trauma is increased in children with CP, spina bifida, and DMD (58). This is related to reduced bone mineral density secondary to immobilization or limited mobility. Children with CP or DMD with an acute fracture typically present with pain and/or irritability. However, children with spina bifida or a spinal cord injury may only present with swelling of the limb due to their lack of sensation. Radiographs should be utilized when swelling of a limb is present, even when no trauma history is elicited.

Treatment of fractures in children with disabilities varies, depending on the diagnosis, type, and location of

the fracture. Casting of a limb in a child may depend on his or her degree of mobility. A child who is wheelchair-dependent may not need a surgical procedure but a bulky splint may be applied to maintain alignment and allow healing. This is especially true for a child who is insensate, since a plaster or fiberglass cast may lead to pressure sores. Prophylactic treatment of reduced bone mineral density in CSHCN is controversial and should be addressed by a specialist in disorders of bone metabolism (59).

Reproductive/Sexual Health

Most individuals with developmental disabilities are able to have children. Thus, caregivers and individuals with disabilities need appropriate guidance and management about sexual health, reproductive health, and pregnancy (60). The benefits of this type of guidance and education include decreased risk of sexual abuse as well as improved feelings of autonomy and self-esteem. Education in this realm can also reduce the fear and anxiety that oftentimes accompanies these topics for both individuals and caregivers. Individuals should have appropriate education provided to them regarding sexual and reproductive health, and it should be described in the Individualized Education Program (IEP).

For adolescent females with disabilities, menarche and menstruation can be particularly challenging. Periods can be very painful and add another level of self-care skills for the adolescent. In some circumstances, patients and caregivers may seek elimination of periods. Menstruation suppression can be achieved by utilizing intrauterine devices, progesterone-only injections, and oral contraceptives if appropriate. Permanent sterilization is now considered unethical, and should not be pursued. Gynecologic examinations should be performed in the most sensitive manner, allowing for the individuals to be as comfortable as possible. Assent from the adolescent should be obtained if possible, and in severe circumstances pharmacologic analgesia and anxiolysis may be used.

Sleep Health

Individuals with disabilities often present with a wide variety of concerns regarding sleep, and it is a common complaint when coming to a clinician's office. Sleep disturbances not only impact the child but can significantly disrupt an entire family's sleep. For example, individuals with CP and intellectual disability have increased prevalence of bruxism, commonly occurring during sleep, which can lead to family sleep disruption, tooth erosion, headache, and tooth sensitivity (61). Obstructive sleep apnea is common among children with developmental disabilities, particularly those who have abnormal muscle tone or neuromuscular diagnoses. Individuals can present with behavior problems and irritability. Snoring, difficulty breathing in sleep, and breath pausing are commonly present. Assessment of snoring and breath pausing includes a complete otolaryngologic examination to assess for adenotonsillar hypertrophy and, commonly, a sleep study. Individuals may need an adenotonsillectomy, continuous positive airway pressure (CPAP), or BiPAP.

Individuals with visual impairments and other developmental disabilities can have circadian sleep disturbances resulting in irregular sleeping patterns. Treatment of sleep disturbances such as these includes strict adherence to the principles of healthy sleep hygiene, which include a regular and set bedtime routine, use of appropriate transitional activities before sleep (reading, quiet play, bath), and positive reinforcement for appropriate bedtime behavior. The best studied pharmacologic therapy used in children for sleep is melatonin, and is commonly used for individuals who have difficulty with sleep initiation.

PALLIATIVE CARE

Primary care for CSHCN may include consideration for palliative care services. Children who should be referred for palliative care are those with potentially life-limiting diseases. This diverse group includes children with diagnoses of advanced or progressive cancer, neuromuscular diseases, severe CP, acquired brain injuries, severe central nervous system (CNS) malformations, complex and severe cardiac abnormalities, and chromosomal or metabolic abnormalities. Children with HIV infection, severe immunodeficiency, cystic fibrosis, and severe epidermolysis bullosa also meet the criteria for palliative care. Palliative care for children can and often does include life-prolonging treatments, such as a tracheostomy placement for a boy with DMD, as well as potentially curative treatments, such as chemotherapy for a child with advanced cancer.

In a policy statement on palliative care, the AAP states, "Palliative treatments focus on the relief of symptoms (eg, pain, dyspnea) and conditions (eg, loneliness) that cause distress and detract from the child's enjoyment of life. It also seeks to ensure that bereaved families are able to remain functionally intact" (62). Palliative care focuses on the quality of the life remaining to the child. Palliative care addresses not only the physical symptoms, but also the psychological, social, and spiritual issues of a child who lives with life-threatening or terminal conditions.

CONCLUSION

In summary, CSHCN account for a large percentage of children in a primary care practice. It is imperative that the PCP be familiar with the associated medical conditions of

each child and the need to provide routine health care. Furthermore, it is critical for the PCP to be familiar with the frequently occurring acute medical illnesses in children with disabilities and the common manner in which they may present.

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3

PSYCHOLOGICAL ASSESSMENT IN PEDIATRIC REHABILITATION

Jamie L. Spohn and Jane A. Crowley

Children's needs within a rehabilitative setting are different from adults in that recovery of skills and return to baseline are not the end point. Rather, it is an evolution toward the continued development of ever-changing abilities in emotional, behavioral, and cognitive structures. The goal of any pediatric rehabilitation process is to foster the continuing work of childhood. This additional distinguishing dimension of pediatric rehabilitation relates to the central requisite of the pediatric population—development. There is a dual goal: rehabilitation to prior levels and capacity for the remaining development in that child's or teen's life. An important tool for establishing current levels, setting future goals, and tracking progress and/or changes over time is psychological assessment.

The rehabilitation physician and the team will treat a wide array of medical conditions among their patients. Rehabilitation medicine departments will encounter requests for treatment for those with congenital disability, acquired disability from illness or injury, and chronic medical conditions. A recent estimate of the incidence of severe chronic illness seen in rehabilitation is more than 1 million children in the United States (1). These children, and those who survive catastrophic illness or injury, are a growing population due to medical advances that reduce mortality, covering the full age range from infancy to young adulthood.

In acknowledging that normal development assumes an intact sensory, motor, and overall neurologic system for interaction with the environments of family and the larger world, the children and teens we work with do not have the standard equipment or interrelationships among the aforementioned skills. For example, a child's motor disability can easily alter the basic emotional developmental tasks. The protraction of physical dependence that is a reality for a child with a congenital disability like spina bifida, at the very least, risks altering the psychological and developmental milestones of separation/individuation. Cognitive sequelae of that central nervous system (CNS) disorder can also result in academic, social, and adaptive behavior deficits. In these cases, standard developmental schema often does not apply (2). These developmental scripts do not apply not only because of deficits, but because there are unique tasks to be mastered with a disability. Functional use of a wheelchair, doing activities of daily living (ADLs) with one arm, self-catheterization, and visual competence with a field cut are but a few specific "milestones" our patients face. In the case of a traumatic injury, the disruption of a normal life as well as typical developmental progress and engagement in the world, is an emotional maelstrom for the patient and his or her family (3).

It is possible that many problematic distortions in many aspects of the nurturing and individuating demands of competent development abound in children with disabling conditions (4). The barrage of medical technology and interventions is vast in variety and effectiveness. Yet, the psychological cost of these necessities can be astronomical. The challenges of hospitalization, a disruption of familiar routine, the therapy demands of the rehabilitation structure, absence of parents, and intrusive or painful medical procedures are additional tasks against which to hinder the patient's progress (5). In a broader context, there is prejudice against those with disability, and children must face the extra demands of bridging ignorance and misconceptions among their peers and anyone else they may encounter.

In line with the centrality of development, the objects of assessment constitute a "moving target." Environmental demands change, as does the child's or teen's abilities to meet them. At school age, the child must now function competently in the ever-increasing demands for independence reflected in the school setting. Furthermore, the medical condition can itself change over a child's development, causing a need for constant adaption. A disease process can progress (eg, juvenile rheumatoid arthritis), or increasing body size can change the nature of mobility (eg, spina bifida), and prior function can be lost, requiring significant adjustment on the part of the child or teen, as well as their parents or guardians. The task is to have these experiences remain *challenges* to development and

not become *barriers.* This argues for continued monitoring throughout a child's development as a vital factor and the importance of psychological assessment as a vital part of that monitoring to be utilized throughout the pediatric course of a patient's life. This allows for continuous modifications as support throughout the individual's developmental course.

The relationship of family functioning to outcome in pediatric disability has been widely demonstrated (6,7). The challenge to a family is to walk an unfamiliar path, as few families have direct experience with childhood disability. The effects may be bidirectional (8), with the deficits from the medical condition interacting with parental features or the child's status resulting in disrupted and possibly ineffective parenting approaches. Parents often must assume an additional role as case manager and advocate in the medical and educational systems. In addition, they have to "translate" their child's issues to other family members at the nuclear and extended family levels. The family becomes a vital arena of intervention. The family is the first-order site of development and stimulation as well as a filter for the world at large.

ADJUSTMENT VERSUS PSYCHIATRIC DIAGNOSIS

It is important to recognize the distinction between *psychiatric disturbance* and *adjustment problems* as informed concepts in assessment for a pediatric rehabilitation population. Indeed, psychiatric disturbance is not common in children with chronic conditions, as some studies show that their functioning is better than children in the mental health clinic population. Taken together, however, children and teens seen in rehabilitation medicine settings do have a greater risk for adjustment problems (9). Their medical conditions act as life stressors not encountered by their healthy peers. To use psychiatric diagnoses in this population belies the reality of behavioral symptomatology that is indeed adaptive to the conditions and situations of a child's medical condition. Though some behaviors may be unusual in the healthy child, they may be adaptive to this population (9). The concept of *adjustment* encompasses the variability that these patients encounter. It can express the unique trajectory that these children's lives will take, and recognizes it as adaptive in that it is age-appropriate for those conditions and oriented ultimately toward healthy adult functioning.

A large body of literature focuses on the adjustment of children with chronic physical conditions. This group was twice as likely to have adjustment problems as healthy children in a meta-analysis of 87 articles by Lavigne and Faier-Routman (9). Though the specific prevalence rates were higher among these children, only a minority showed maladjustment. Such children, then, are more vulnerable than those who are healthy. Newer assessment instruments have been developed that utilize the concept of *quality of life* (10) and will be discussed here in the section "Population-Specific Assessments." With this approach, the nature of a child's or teen's adjustment and the reflection of the uncontrollable factors in his or her situation are captured for a wider rubric than the inadequate dichotomy of normal versus abnormal.

However, the use of psychiatric diagnoses can be appropriate in both this population and those with neurodevelopmental disabilities. The latter group showed a rate six times that of the general population for significant emotional and behavioral problems (drawn from an outpatient clinic population) (11). The identified problems run the gamut, encompassing a breadth of both emotional and behavioral disorders, and are more likely to persist into adulthood. There is the primary impairment of the neurologic disorder and a secondary impairment of psychosocial support problems (4). With the disturbance in the brain being primary, these findings are not surprising. Other factors exist as well to either exacerbate or ameliorate the brain's abnormality, but the overall picture is one of significant neurologic and psychological morbidity, with the source of disability either congenital or traumatic.

The most common disabling injury of childhood, in fact, is traumatic brain injury (TBI; 12), which carries a substantial risk for long-term physical, cognitive, emotional, and behavioral difficulties (13). Expressed differently, incidence figures are such that by the tenth grade, 1 in 30 students would have had a TBI (across severity ranges of mild, moderate, or severe). This population will be a substantial part of a pediatric rehabilitation medicine practice. The causative link between cognitive and behavioral functioning represents the juncture of thinking and adaptive behavior that can be devastating to the ongoing development of a child survivor of an acquired injury (14). There is a particular danger in the misattribution that easily occurs. Behavioral deficits will likely be attributed to more common etiologies, as opposed to the organic brain disorder from the injury. With misattribution comes the inevitable inappropriate treatment. Cognitive limitations are not accounted for in treatment efforts, or the wrong premise (eg, antecedent versus contingent programming) used, and failure occurs or even exacerbation of the original problems. Awareness and consideration of the brain damage from an injury means assessment must encompass a wide focus, looking at emotional, behavioral, cognitive, personality, and adaptive domains. Neuropsychological testing is the centerpiece in these children and teens, representing a subtype of general psychological assessment that will be an important aspect of many rehabilitation cases.

NATURE OF MEASUREMENT

The essence of psychological assessment lies in the construction of the instruments used to explore various domains of adjustment, personality functioning, behavior,

emotion, and cognition. This construction always has at its core the notion of standardization through its reference to a normative group, whose performance is characterized by a transformation of the raw score earned by an individual. Even the most skilled observer could not provide the richness of the information gleaned from a psychometrically sound instrument. Such a test allows for the comparison of that subject to the typical performance of his or her peers in a fair and objective way. The value of standardized assessment depends on some core concepts, elucidated in the following sections. It is standard for clinicians to rely on not only standardized instruments, but also behavioral observation and input from the child's exterior world, including parental information as well as information from the child's school. Conclusions are drawn when all sources of information have contributed.

NORM-REFERENCED MEASUREMENT

Norm-referenced tests are standardized on a clearly defined group, referred to as the normative group, and scaled so that each individual score reflects a rank within that group. The examinee's performance is compared to the group, generally a sample that represents the child population of the United States. The comparison is carried out by converting the raw score into some relative measure. These are derived scores and indicate the standing of a patient relative to the normative sample by which the test was standardized. These scores also allow for comparison of the child's performance on different tests. Stanines, standard scores, age- and grade-equivalent scores, and percentile ranks are the most common tests.

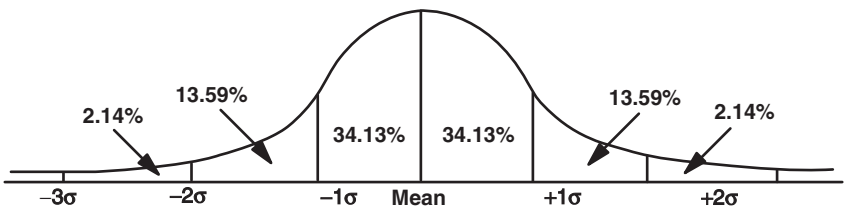
A central concept in the expression of individual performance as compared to a norm group is the normal curve. The normal curve (Figure 3.1) is a bell-shaped curve. It represents the distribution of many psychological traits, with the greatest proportion at the "middle" of the curve, where it is the largest, and the abnormal levels—both below and above average—at the two "tails." All derived scores have a distinct placement on the normal curve and are varying expressions of the location of an individual's performance on that curve.

Stanines are expressed as whole numbers from 1 to 9. The mean is 5, with a standard deviation of 2. Substandard performance would be judged with stanines in the range of 1 to 3 and above average at 7 to 9. In this transformation, the shape of the original distribution of raw scores is changed into the normal curve.

Standard scores are generally the preferred derived scores (15). Their transformation of raw scores yields a mean for the normative group and a standard deviation. This places a given score across the normal curve, and the scores express the distance from the mean of that patient's performance.

T scores, *z* scores, and the well-known *IQ* of the Wechsler scales are all standard scores. Like all standard scores, the *z* score derives a constant mean and standard deviation across all age ranges The *z* score has a mean of 0 and a standard deviation of 1. It expresses below-normal performances with the minus sign and above average with the plus sign, with scores in a range of -3 to +3. These scores are often transformed into other standard scores to eliminate the positive and negative signs (see Figure 3.1). *T* scores and the IQ scores are drawn from the *z* score, with different numerical rubrics that eliminate the plus or minus sign associated with the *z* score.

Multiplying by 10 and adding a constant of 50 yield a *T* score ranging from 20 to 80, with an average of 50. Another transformation occurs by multiplying the standard score by 15 and adding 100. This provides a range from 55 to 145, with a mean of 100 and a standard deviation of 15 or 16, depending on the test used. This is the method that produces the *Deviation IQ,* the form of derived score used on the Wechsler intelligence batteries. The alternative to the Deviation IQ is the *Ratio IQ,* which is the ratio of mental age to chronological age multiplied by 100, used in the Stanford-Binet tests.



Z scores	–3.0	–2.0	–1.0	0.0	1.0	2.0	3.0
Percentile	0.13	2.5	16	50	84	97.5	99.87
T - score	20	30	40	50	60	70	80
IQ (SD = 15)	55	70	85	100	115	130	145
SAT score	200	300	400	500	600	700	800
Stanine	1	2	3	4 5 6	7	8	9

FIGURE 3.1 The normal curve.

What appear more understandable, but are not as psychometrically sound as standard scores, are percentile ranks and age- and grade-equivalent scores. Percentile ranks offer easy interpretation, with the rank reflecting the point in a distribution at or below which the scores of a given percentage of individuals fall. To a lay audience, this is often confused with percentages, which are *not referenced to a normative population*—only to the number correct compared to the total number of items. For example, function at the 50th percentile is average performance, whereas a grade of 50% on a test would be considered failing.

Even more straightforward appeal exists for age- and grade-equivalent scores. These scores are obtained by discerning the average raw score performance on a test for children of a given age or grade level. These scores are often used when the child is given a test of academic ability. The individual patient's score on that test is compared to that value. Grade equivalencies are expressed as tenths of a grade (eg, a grade equivalency of 4.1 represents the beginning of fourth grade). Despite their appeal, there are limitations with these forms of derived scores. First, a grade-equivalency value does not mean that a child is performing at that particular level within his or her own school, as the curricular expectations of the school might be different from the mean score established by the normative sample. Some actual age- or grade-equivalency values might not have been earned by any specific member of a normative sample, but instead are extrapolated or interpolated from other points of data. Furthermore, age or grade equivalencies may not be comparable across different tests. The meaning of a first grader who obtains a raw score similar to a third grader is not that the child is functioning as a third grader in that subject. He or she shares that score, but the assumption that the child in first grade has all the skills of a third grader is inappropriate. Similarly, a 12-year-old patient who achieves an age equivalency score of 8 years, 4 months seldom actually functioned on the test the way a typical 8-year-old child would, and certainly should not be treated like an 8-year-old for most issues in rehabilitation or academic programming.

Finally, as is the case with percentiles, age- and grade-equivalents cannot be used in statistical tests, as there is an unequal distribution of scores. Both require conversion to another scale before they can be used in data analysis.

RELIABILITY

This concept of *reliability* refers to the ability of a test to yield stable (ie, reliable) results if readministered at different points in time. There needs to be a consistency and stability of test scores, and the nonsystematic variation reduced as much as possible. Psychometric theory holds that any score is composed of the measurement of the actual trait that a child possesses as well as an error score, which represents the variation or standard error of measurement. The reliability coefficient is the familiar statistic to express this property. It can vary from 0.00, indicating no reliability, to 1.00, indicating perfect reliability. High-reliability coefficients are considered particularly important for tests used for individual assessment. In the case of cognitive and special ability tests, a reliability coefficient of 0.80 or higher is required for sufficient stability to be a useful test. Reliability coefficients are calculated for a test across three conditions of reliability. One is *test–retest reliability,* meaning the capacity of the test to yield a similar score if given a second time to a child. Another is *alternate-form* reliability, where the child is tested with an alternate form of the test, measuring the same trait and in the same way as the initial testing. A third kind refers to *internal stability* in a test, where in the ideal test, item responses are compared to another item on the test to demonstrate the equivalence of items in measuring the construct in a replicable manner. Active judgments must be made in the choice of tests, with reliability coefficients reviewed in the process of test selection.

VALIDITY

Validity is another crucial component to consider in the construction and use of standardized measures. Validity is the extent to which a test actually measures what it intends to measure and affects the appropriateness with which inferences can be made based on the test results. Validity of a given test is expressed as the degree of correlation, with external criteria generally accepted as an indication of the trait or characteristic.

Validity is discussed primarily in terms of *content*—whether test items represent the domain being measured as claimed—or *criterion*—the relationship between test scores and a particular criterion or outcome. The criterion may be concurrent, such as comparison of performance on neuropsychological test measures with neurophysiologic measures (eg, computed tomography, electroencephalography).

Alternatively, the criterion may be *predictive*—the extent to which test measures relate in a predictive fashion to a future criterion (eg, school achievement). In the rehabilitation context, various events and contingencies may affect predictive validity. An appropriate determinant of predictive validity is the likelihood that the individual's test performance reasonably reflects performance for a considerable period of time after the test administration. Acute disruption in physical or emotional functioning could certainly interfere with intellectual efficiency, leading to nonrepresentative, or failure to measure what it is intended to measure, results. In contrast, chronic conditions would be less likely to invalidate the child's performance from a predictive standpoint because

significant change in performance as a function of illness or impairment would not be expected over time. With therapeutic interventions, a patient's performance could improve, so test results from prior to that would not be valid. The more time that passes between test administrations, the more likely extraneous factors can intervene and dilute prior predictive validity, making updated testing imperative for continued ability. Anxiety, motivation, rapport, physical and sensory handicaps, bilingualism, and educational deficiencies can all effect validity (15). For an inpatient population, the effects of acute medical conditions (eg, pain, the stress of hospitalization, medical interventions themselves, fatigue) can also affect validity. Wendlend and colleagues (16) noted that in a study of cognitive status postpoliomyelitis, the deficit seen could well have been due to the effect of hospitalization as opposed to the disease. The understanding of acute effects of hospitalization on psychological testing makes it imperative to retest the child in order to have results to compare to as the child is released from the inpatient setting.

Construct validity refers to the extent to which the test relates to relevant factors. Another important component of validity is *ecological validity,* which refers to the extent to which test scores predict actual functionality in real-world settings. Test scores are typically obtained under highly structured clinical testing situations, which include quiet conditions, few distractions, one-on-one guidance, explicit instructions, praise, redirection, and so on. These conditions do not represent typical everyday tasks or settings (17). This disconnect between the test setting and real life is especially relevant in children with brain-related illness or injury. These children, who have high rates of disordered executive functioning (eg, distraction control, organization, planning, self-monitoring, etc.) benefit disproportionately from the highly directive nature of clinical testing, and test scores may overestimate true functional capacity for everyday tasks (18).

A test's reliability affects validity in that a test must yield reproducible results to be valid. However, as detailed previously, validity requires additional elements.

In the rehabilitation population, all of these issues have particular importance. Most tests are developed on a physically healthy population. Motor and sensory handicaps and neurologic impairment are not within the normative samples. Issues of validity predominate here, though with transitory factors as noted previously, reliability can be affected as well. Standardized procedures may have to be modified to ensure that a patient is engaged in the testing in a meaningful way.

USES OF ASSESSMENT

The use of psychological assessment aims to further the functioning and adjustment of children with a wide range of disabilities or chronic illnesses. These purposes encompass issues directly related to the medical and rehabilitation setting, but often have equal utility in educational planning. Unique to the field of pediatric rehabilitation is this necessity for interaction between what are arguably the two biggest public systems for children: medicine and education. Both have their productive and counterproductive forces and hold a vital place in the individual child's or teen's life. Furthermore, both can act to hinder or potentiate the salutary effect of the other. The needs and parameters of engagement with both is at the crux of the navigation of development for our patients, and psychological assessment contributes significantly to this process.

Psychological testing is often associated solely with IQ testing. The IQ concept of intellectual development is too narrow for many of the applications in a pediatric rehabilitation setting. Instead, the broader notion of *cognitive* abilities, not merely *intellectual* abilities, is considered more important. Cognitive assessment covers testing the wide array of known components of the brain's thinking, reasoning, and problem solving. Assessing these intake, processing, and output modalities of thinking, their individual elements or the combination of these skills is a vital factor in school or in medical rehabilitation. School is children's work, and the interface with this system is critical, as it is the arena where many key adjustment and developmental issues are played out. Psychological adjustment—indeed, overall functioning—is closely tied to cognitive status. Coping with frustration, functioning within a group, and inhibiting and planning for long-term goals, are examples of processes vital to school that have cognitive capacity at their center.

Within the schools, the psychological assessment performed has typically included only intellectual and academic achievement testing as prime components. Though that is changing in some settings, it is not yet common that *cognitive* processes are assessed within the school setting. For the populations common to a rehabilitation medicine practice, many conditions have brain involvement (eg, TBI). Their needs are clearly beyond the limitations of typical school testing capabilities. Eligibility for services within the special education system under the qualifying conditions of TBI (mandated by the federal government in 1998) cannot be done without consideration beyond IQ and achievement testing. Indeed, TBI as its own inclusion category was done to reflect the serious misunderstanding of the disorder when only evaluated by IQ and achievement testing alone. TBI evaluation lies in stark contrast to individuals with learning disabilities, which is typical what the school is able to piece together through the use of just intellectual and academic achievement testing. The intellectual assessment of children with spina bifida needs explication beyond IQ testing as well. Often, the component parts of the Full-Scale IQ score are so divergent in children with spina bifida and other brain conditions that it does not represent a true summary score, and thus must be interpreted on an individual

subtest level, rather than merely looking at composite scores. To understand a child's condition fully, further assessment of cognitive processes needs to be done. Pertinent abilities are attention, concentration, memory, and executive functions. In the wide array of conditions known to affect brain functioning there are primary and secondary effects. Primary effects are seen from brain tumors, seizure disorders, or cancer processes. Secondary, or "late" effects on cognitive processes are seen in the process of infectious disease or cancer treatment. It is necessary to evaluate a broader array of abilities rather than relying solely on IQ to understand the full spectrum of required cognitive skills for competent development.

In order to promote the fuller understanding of medical conditions and their effects on cognitive functioning, the rehabilitation practitioner will often be consulted for more specialized assessment to capture the full nature of functioning within his or her patients. Input into the Individualized Educational Plan (IEP), which is the centerpiece of planning in the special education system, is essential in brain-based disorders to ensure full consideration of the medical condition, its own process, and its unique effect on brain functioning. The dynamic nature of recovery is notably absent from most students receiving special education services, but is often a primary part of the course in TBI, brain infectious processes, cancer, or strokes. Frequent reassessment, specific remediation or rehabilitative-focused services, or specialized support in school reentry, and transition are several of the unique concepts that are vital to sound educational planning in our population but are largely unknown to the traditional process of special education. This is the most critical juncture of school and medical factors in a pediatric rehabilitation process.

As per Section 504 of the Rehabilitation Act, *accommodations* are often sought on either a long-term or transitory basis in rehabilitation medicine patient groups. These are efforts to "level the playing field" within the school setting in acknowledgement of disability that skews a student's ability to benefit from the standard educational setting. These students do not require the breadth or type of actual intervention or service gained through special education classification and do not require special classification. Instead, these students need modifications in the system in order to demonstrate their capacities or adequately access the learning environment and mainstream curriculum. Results of psychological/neuropsychological evaluations can be useful in demonstrating such needs related to cognitive issues. For example, deficits in information processing speed can have a global effect on functioning within the group instructional environment of school. Accommodations such as reduction in homework, extended time for tests, quizzes, or assignments, or lecture notes, among others, can all be sought with the documentation provided by evaluation results. The issue of how long the accommodations are required can be answered by repeated testing. An example is in the case of a brain injury where recovery occurs and accommodations may no longer be necessary.

It is of the utmost importance for the clinician to recognize and have a solid understanding of the role he or she can play in securing vital (but not necessarily typical) medical treatment for a patient. This includes speech and language or occupational therapy, cognitive remediation, or adjustment-focused cognitive behavioral work. The documentation of that need, based on the medical diagnosis or history, can be obtained much quicker and with the proper focus through the medical system in terms of both insurance coverage and proper treatment frequency and formulation. Obtaining assessment from a public school system can be a lengthy process. For rehabilitation patients, this can waste valuable time and, therefore, cannot meet the time frame needed for an acute recovery. A typical school psychological assessment could miss acute issues and be even less likely to detect weaknesses that could hamper development or skill acquisition distant from the injury or illness. Such evaluation needs the medical framework of rehabilitation psychology to be timely and pertinent. Furthermore, with a rehabilitation psychology perspective and knowledge, appropriate documentation emerges to secure services covered by medical insurance or from legal settlement funds, if such exists. Further, a broader range of knowledge exists within a rehabilitation setting related to access to other services, resources within the patient's community, and specific school and classroom placement planning. Keeping the intervention within the medical perspective can make it more integrated with disease or injury sequelae and, therefore, more targeted and appropriate in terms of goals and treatment techniques.

It can be seen that the assessment of a child's or teen's learning process is essential to both the school and medical setting. Memory processes, language abilities, planning, and capacity to inhibit are essential functional elements in either system. The preference of one modality over another, or the explication of memory functioning, can be of great use in school issues and in rehabilitation. The need to master specialized tasks, such as wheelchair skills or self-catheterization, can be enhanced when general learning styles of an individual patient can be discerned.

This understanding of a patient's cognition can inform educating the patient about his or her medical disorder, or the rationale about a medical procedure. The feelings of victimization that can evolve around a painful surgery and the subsequent effect on adjustment or even personality formation are secondary sources of potential morbidity in a child's development. The child or teen senses that he or she was regarded enough in the consideration of procedures to be included in the decision and planning process. The experience of this and the skill to be a meaningful participant are vital long-term skills and are promulgated by knowing the proper way to present material in a way to ensure understanding. Decisions

about a child's ability to benefit from a specific treatment such as biofeedback, relaxation training, or the varieties of behavioral programming available are part of diagnostics that guide treatment.

Change as the result of intervention can be quantified by assessment. However, change without overt intervention, but to chronicle the long-term outplay of a medical condition, is arguably the most common use of assessment in rehabilitation. The risk for long-term sequelae in TBI or from cancer processes and treatment is well known (13,19). The serial assessment of a patient, particularly through known critical developmental periods or illness interventions, is at the core of sound pediatric rehabilitation practice. A developmental lag becomes the object of treatment, whether to spur development or to teach compensatory strategies. As the physical process of a disease is monitored through traditional outpatient clinic visits, so is the status of cognitive/behavioral functioning in relation to the demand of one's medical condition or to changing developmental expectations is equally important to monitor.

Understanding the individual experience of a child or teen in relation to his or her body experience is another use of assessment. Understanding the experience, whether through a questionnaire about pain, assessment of specific mood states like depression or anxiety, or a general personality assessment of that patient, can be quite useful. Differential diagnosis can be important, as in the case of posttraumatic stress disorder, where cognitive symptoms of that disorder can be mistaken for the effects of a mild brain injury or concussion. In that circumstance, the deficits are due to the effects of the stress and not to the mechanical disruption of trauma.

TYPES OF ASSESSMENTS

The purpose of psychological assessment is to discern the status of an individual in relation to an appropriate peer group. Jerome Sattler discerns four pillars of child assessment as norm-referenced tests, interviews, observations, and informal assessment (15). This is a broader list than many referral sources would recognize, as typically "tests" are all that might be considered as psychological assessment or evaluation. However, a central tenet in psychology is that test scores or results cannot be interpreted in isolation. Information from naturalistic settings must be sought through the methods of interview, observations, and informal assessments, as enumerated by Sattler.

In a discussion of cognitive testing, the issues of single tests versus batteries are an important consideration. Single tests are designed to tap a specific dimension of cognition, like verbal learning or visual–motor abilities. As useful as they are for more in-depth examination of a single construct, this strength is a source of limitation as well. Seldom is the question at hand to be answered by examining a single ability. Abilities are not the unitary concepts that evolve from theoretic models. The influence of other overarching cognitive abilities, such as attention or processing speed, is not addressed directly and is discernible only through observation. Normative samples for single tests can be restricted and not large enough or representative enough to draw firm conclusions as to standing within one's peer group. Therefore, the use of a *test battery* is preferred.

APPROACHES TO ASSESSMENT

Historically, there are three approaches to assessment, especially neuropsychological assessment, that have been widely used. The first is called a *fixed battery of tests.* The best-known example of a test battery is the Wechsler batteries for intelligence assessment, comprising a number of subtests. These collections cover an array of abilities. The fixed battery is a predetermined set of subtests that are administered in a standard format to every patient. This same set of tests is administered regardless of the referral questions or set of symptoms that a patient presents with. The advantages of a fixed battery include a comprehensive view of the patient's cognitive domains. They also provide the strongest basis for comparison of a patient's performance across subtests, as norms are based on this arrangement of tests, given in the established order or both strengths (what a patient is able to do) and weaknesses. The use of a standardized format in fixed batteries is also highly useful for research purposes. Another advantage is when the evaluation results are questioned, such as in a court proceeding, where the examiner will be required to defend his or her evaluation findings. This approach is also beneficial for green practitioners who are not yet comfortable with choosing their own batteries.

There are several disadvantages for the use of a fixed battery approach. One is that it is exceedingly time-consuming. Since all tests within the battery must be administered, the time it takes to do such is often a barrier to choosing this type of approach. The fixed battery approach requires the clinician to examine all areas, even those that appear intact. This results in an excessive collection of data that may have little or no use to the clinician. Another disadvantage is the lack of flexibility in different clinical situations. This makes it sensitive to detecting focal deficits, as opposed to diffuse deficits, which is often what a child presents with.

A *flexible battery* is composed of a number of single tests, assembled with the patient's referral question or known medical condition and/or symptoms in mind, with an eye to tapping tests most likely to explicate suspected deficits. Thus, the battery is individually tailored to each patient based upon a diagnostic question. Flexibility can allow for modification and possibly individualization of directions during the assessment (20).

Advantages of the flexible battery include possible shorter administration time, lower economic costs, and the ability to adapt to individual needs and situations. Disadvantages include the potential for examiner bias or omission of deficits due to a lack of comprehensiveness, which is problematic in situations where an examiner is required to defend his or her findings (eg, court proceedings). Another disadvantage is a lack of standardized administration rules for some of the tests, and a limited capability of developing a research base, thereby making retest reliability difficult. A common approach to alleviate these disadvantages would be to utilize a core set of tests that assess major neuropsychological domains, and to supplement the battery with additional tests as they are needed.

Lezak and colleagues (21) noted a survey of neuropsychologists where 70% responded that they use a flexible battery approach. They note the position that fixed batteries involve more testing than some patients need. Further, the flexible approach continues to increase in popularity, as health insurance corporations are continuing to restrict reimbursement for longer evaluations (22).

Automated or computer use in testing has increased substantially since the 1980s. Prior to that, automated and later computerized administration and scoring of tests was quite limited. Initially, computerized testing of attention was developed (Connors Continuous Performance Test). More recently, computerized tests have been developed for concussion diagnosis and monitoring, in addition to research purposes. Such techniques offer repeatability, sensitivity to subtle cognitive changes, and ease of administration. Reliability, validity, and other considerations pertinent to general issues in more traditional so-called pen-and-paper tests are pertinent to this type of assessment as well. Computerized testing for concussion will be discussed in the Concussion chapter of this book (Chapter 12).

Evaluation, then, is a robust and multifactorial process, not to be confined to a set of test scores or descriptions of test performance, but also to include natural setting, or qualitative observation, data. The norm-referenced placement of a patient has a role, but the assessment setting in and of itself imposes a high degree of structure. While this one-to-one administration is not replicated in real life, it is necessary for the standardization of administration and the reference to a normative sample, as described previously. This offers a "best case scenario" for the patient, which has implication when compared to "real-life" settings. Therefore, the addition of perspectives from natural settings of the home, school, and community is necessary, as is the consideration of the aspects of the medical condition.

The interpretation of standardized tests must take these factors into account: issues about the course of a disease or injury recovery, the unique interface that the course of an illness or recovery has on the timetable of childhood development, and the actual length of the struggle with the medical condition.

Some of these elements are captured in a good history taking and/or record review. Reserve factors concerning coping and response are also gathered in history but can additionally be tapped by standardized questionnaires, whose responses are sought from a variety of sources. These encompass figures from the major settings in a child's life (ie, parents and teachers). The value of such instruments is that they can reference responses to those of a normative population such that the degree of divergence from standard development can be expressed. Some include consistency scales that add information about the nature of the responses given.

CULTURE-SENSITIVE ASSESSMENT

Culturally sensitive psychological assessment is often a challenging aspect of the testing process. Most measures have a culture bias in terms of content and validity, and normative data are seldom adequately representative of diverse groups. Not all examiners are sufficiently sensitive to the impact of cultural issues on children's performance, and when interpreted without caution, results can be misleading. The assessment of English language learners, children who have reduced mastery of the English language because their parents' primary language is not English, is particularly challenging. Use of interpreters or test translations carries limitations, such as lack of equivalent concepts in the two languages, minimal provision for dialectical variations, performance anxiety to having an "audience" present, and possible changes in the level of difficulty or meaning of translated words (15).

Several "culture-fair" tests have been developed to reduce culture bias by limiting the amount of verbal exchange, using more abstract content that is less grounded in culture and language, and using more diverse groups during the norming process. This represents an important step in culturally sensitive assessments, and some of these tests are discussed in the following sections. However, there is no way to truly eliminate cultural bias from tests, and demographic data on normative groups must be carefully examined before assuming that it is any more representative of the specific patient than traditional tests. For example, many of the "culture-fair" tests are normed only on children in the United States. Their use for students with different backgrounds, such as children from refugee camps in Africa with little to no formal schooling, is clearly limited.

Culture-sensitive assessments in pediatric populations are made even more complicated by the frequency of mild to severe motor impairment. Examiners assessing individuals with motoric impairment rely heavily on tests of verbal cognitive skills and try to reduce the number of tasks that require speeded or complex motor responses. Examiners assessing individuals from linguistically or culturally diverse backgrounds rely heavily on tests of nonverbal cognitive skills and try to reduce the

verbal component. Examiners assessing individuals from linguistically diverse backgrounds with motor impairments are limited indeed in terms of valid options. Even in pediatric groups that do not have motor impairment, the higher frequency of *discrepancies* in functioning (significant strengths and weaknesses in a single individual, such as may be caused by damage to right versus left hemisphere or cortical versus subcortical areas) makes the traditional practice of assessing nonverbal skills and considering the results representative of *general* functioning highly questionable. School and community-based clinicians may not be aware of the complexity of issues involved and may provide scores without adequate caution regarding limitations.

SPECIFIC INSTRUMENTS

NEUROPSYCHOLOGICAL EVALUATION

Originally, the neuropsychological assessment was directed at diagnosing the presence, nature, and site of brain dysfunction. The focus has shifted from diagnosis to assessment of a child's function to identify and implement effective management, rehabilitation, or remediation services.

NEUROPSYCHOLOGICAL BATTERIES

As mentioned earlier, neuropsychological batteries have been developed to provide a comprehensive evaluation of cognitive abilities. The two most common in practice today are the downward extensions of the Halstead–Reitan Neuropsychological Battery and the NEPSY-II, developed specifically for children.

The Halstead–Reitan Battery has been refined and redefined over the years since Ward Halstead's original conceptualization in the 1940s to a larger series of tests to diagnose so-called brain damage for ages 14 and above (23), and subsequently the downward extension for ages 9 to 14, called the Halstead-Reitan Neuropsychological Test Battery for Older Children (HRNB-C). It takes approximately 4 to 6 hours to administer, and it uses subtests from the adult Halstead–Reitan Battery, with some modifications. The battery for children aged 5 to 8 is called the Reitan Neuropsychological Test Battery and requires a similar time interval for administration. These batteries, in wide use earlier, are criticized for a number of pivotal problems. The first is on conceptual grounds, in that the battery was not developed for children, but for adults, and is perhaps reflected in the minimal assessment of memory, academics, and language, with no direct measure of attention. The psychometric properties are widely acknowledged to be quite poor, such that reliance on those alone for interpretation is inappropriate. Considerable clinical acumen is required to interpret findings. Dean concludes a review of the batteries saying, "The HRNB cannot be recommended for general clinical use without considerable training and familiarity with research on the battery (24)." Considering norms published in the interim, Lezak et al. (21) is more favorable to the HRNB in saying that what statistics it yields are misappropriated by "naïve clinicians," implying the same point as Dean.

The only neuropsychological battery ever developed specifically for children is the NEPSY-A Developmental Neuropsychological Assessment (25), with the newest version, the NEPSY-II (26), published in 2007. Both batteries are based on the diagnostic principles of the Russian neuropsychologist Alexander Luria. The original NEPSY had two forms and covered ages 3 to 4 and 5 to 12, with a core battery of 11 to 14 subtests represented to tap five functional domains: attention and executive functions, sensorimotor functions, language, visuospatial processing, and memory and learning functions. This original version was criticized for its content and psychometric properties (27). It is well standardized, and though some instability is noted in some subtests, this may indeed reflect the reality of the developmental status of the brain.

The most recent version expands the age range to 16 years, extending one ostensible benefit of a battery that covers the childhood range, allowing for the ideal serial assessment. The content has also changed, with targeted groupings of subtests for various diagnoses, nonverbal elements, and new measures of executive functioning, memory, and learning, which reportedly solves some of its statistical problems. A functional domain in social perception has been added as well.

ATTENTION, CONCENTRATION, AND INFORMATION PROCESSING

The processes of attention, concentration, and information processing are often central concerns for any patient with a medical condition involving the brain (28). In many ways, they form the basis on which the other component processes occur. Overall performance on other tests looking at other domains of cognitive functioning is significantly impacted by attention and information processing.

Attention has been conceptualized in a number of ways, generally relating to an organism's receptivity to incoming stimuli. Most do regard the issues of automatic attention processes versus deliberate/voluntary as central dimensions. Other characteristics include sustained, purposeful focus—often referred to as concentration—and the ability to shift attention as required by a stimulus. Being able to ward off distractions is usually seen as part of concentration (28). Vigilance is conceptualized as maintaining attention on an activity for a period of time. There are the needs to respond to more than one aspect of a stimulus or competing stimulus—the capacity to divide attention—alternating with shifts in focus.

The multitude of processes subsumed in the concept of attention is necessary because of the overall effect. Most notable is the developmental nature of attention in childhood and adolescence. Increasing demands in school participation are seen in the shifting requirements throughout the academic process. In the early grades, a child is more directly engaged by the teacher, but as the years progress, the capacity for independent (ie, voluntary/deliberate) processes grows. This capacity is matched with growing expectation and demands for independent work as a child progresses through grades. Attentional processes are a central aspect of the changing capacity of typical development. Attention's vulnerability to normal variation, as with fatigue or anxiety, is a part of typical functioning. Attentional processes require a certain "tone" to the brain's functioning; attention and its concomitants are often affected in brain disorders. Furthermore, with acquired deficits in the disordered brain, the demands are higher, as an individual struggles with recognizing the need to attend along with implementing a specific compensatory task.

Lezak and colleagues (21) note that underlying many attention problems is slowed cognitive processing. This can be misinterpreted as a memory disorder (29), as competing stimuli in normal activity interrupt the processing of the immediately preceding stimuli and something is "forgotten," in common parlance. The discernment of this specific problem is important, as strategies alleviating the effects of slowed processing would be different from those for memory per se. All of these aspects warrant examination, notably in those with a brain disorder, due to the overall effect on functioning and the demand for acquisition of academic and adaptive behaviors throughout childhood. The effects of anxiety about an illness process, its treatment, and demands for coping can all affect attention, and in a competent diagnosis are differentiated from primary brain disruption.

Due to the issue of time in competent attention processes, computerized testing has real utility to control for calibration of presentation and response. In the absence of a fully computerized administration, the use of taped auditory stimulus in attention testing allows for standardized presentation increments. Typically, the computerized tasks involve visual stimulus and the taped presentations involve auditory ones. This differentiation between verbal and nonverbal, or auditory versus visual, is necessary to capture these two central aspects of stimulus processing; there may be a distinct difference in one's ability to maintain visual attention versus auditory attention. This has significant implication with one's ability to learn and absorb information.

The recent development of a battery of attention tasks for children, the Test of Everyday Attention-Children, will be described next. It attempts to cover a number of aspects of attention processes and for the comparison of subtest scores to allow for relative differentiation of components.

Inattention, slowness of cognitive processing, and poor concentration have a wide-ranging effect on competent cognitive and adaptive functioning. Other processes may be quite competent, but attention and its aspects can be a primary "rate limiting" factor. These should be addressed in even a screening of functioning, whether at the bedside or in the clinic, both as an overall indicator of current cognitive activity and as a harbinger for developmental problems to come, signaling the need for more stringent monitoring. Commonly used tests are described in Table 3.1.

TABLE 3.1 TESTS OF ATTENTION AND SPEED OF PROCESSING

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Test of Everyday Attention Test of Everyday Attention-Children (TEA-Ch) (30)	Batteries of eight or nine tasks for ages 17 and above; TEA-Ch ages 6 to 16	Taps visual/auditory attention including dual tasks; selective, sustained and executive control
Paced Auditory Serial Addition Test (PASAT) Children's Paced Auditory Serial Addition Test (CHIPASAT) (31)	Adding pairs of digits presented at four rates of speed, controlled by the audiotape presentations; adult and child forms; ages 8 and above	Highly sensitive to deficits in processing speed; sensitive to mild disruption, but can be a stressful test to take, as many items can be missed at normal ranges
Continuous Performance Tests (32)	Covers a category of tests; visual or auditory stimulus where the individual must respond to a target stimulus in the presence of distractors; various versions for ages 4 and up	Many versions exist; sustained, vigilance and inhibition tapped; Connors Continuous Performance Test II and Test of Attention are well known.
Symbol Digit Modalities (SDMT) (33)	Oral or written; requires visual scanning and tracking to match preset symbol and number pairs	Taps information processing; Spanish version with norms; seen as selectively useful.
Trail-Making Test (TMT) (34)	Subject draws lines to connect consecutively numbered (Part A) and alternating numbers and letters in order (Part B). Ages 9 and up	Part of Halstead–Reitan battery; test of speed, visual search, attention, mental flexibility, and fine motor; needs interpretation with other tests; Part B is most sensitive

PROBLEM-SOLVING AND EXECUTIVE FUNCTIONING TESTS

Executive functioning is a cognitive domain that relates to how other cognitive skills (such as attention, memory, language, and nonverbal reasoning) are executed in different situations. Executive functioning requires self-control over impulses and emotions, the ability to flexibly shift, and skills related to manipulating information in mind, planning, and organization. Deficits in problem solving and executive functioning can have a devastating effect on functioning, just as difficulties with attention. Executive functions have both metacognitive and behavioral components. Cognitive process can be intact, but with executive functioning impairments, the output can be substantially derailed. The basic tasks of life can suffer, along with the ever-present demand in childhood to acquire new skills. These deficits can be more obscured in children than in adults, as there is a natural support of activity by parents or other family members, thus creating a situation where many of these deficits will not be identified until early adulthood. Return to school can be the point at which executive functioning problems can clearly be seen for the first time since an acquired illness or injury. TBI presents a particular vulnerability to deficit in these skills. Executive functions are associated with the frontal and prefrontal areas of the brain, where, due to the mechanisms of closed head injury and the shape of the brain and skull convexities, damage can be focused across the full range of severity. Rehabilitation efforts suffer, both in commitment to the process and in learning strategies to compensate for deficits (23).

The competent measurement of executive skills requires a multidimensional approach and can be quite complex (35), given the variety of skills encompassed under the umbrella term of executive function. Testing of these functions imposes a degree of structure required by standardization such that vital elements can be obscured. Attempts at quantification in real-life situations becomes particularly important, as that is where executive skills are often played out. Questionnaires for parents and teachers elicit descriptions of behavior that can be compared to normative expectations. Particularly for parents, this can be useful in understanding the need for treatment. Teachers have a normal sample of age-appropriate peers for comparison in the classroom and can be more aware of such problems. In that circumstance, the questionnaire process can illuminate the component elements to be addressed, as a deficit in classroom performance can be composed of many factors, with differing contributions to the overall presentation. The Behavior Rating Inventory of Executive Function (BRIEF), described in the section on "Psychosocial Evaluation," is an instrument focused on these behaviors. It covers the preschool period through adolescence, as well as a self-report questionnaire for older children, with basic forms for teachers and parents to complete.

There are many models, as in attention, as to what comprises these skills and how to measure the components, since it is far from a unitary concept. Again, as in attention, the developmental progress of these skills is a central aspect of the developing child. In the teenage patient, assessment of these skills is vital, as adult-like capabilities for work, driving, and independence can be severely affected and, in the particular case of driving, can have disastrous results. The enactment of graduated driver license requirements for teen driving in some states implies the centrality of these skills and their necessity for that activity. Stepwise exposure and supervision of driving for teens allows for a graduated experience before full driving privileges are granted. Specialized assessment through rehabilitation-based driving evaluations using computer simulation should be considered by a rehabilitation team in any teen with a history of brain disorder.

Executive skills include the capacity for planning and flexible use of strategies, and the ability to generate, maintain, and shift cognitive sets; to use organized search strategies; and to use self-monitoring and self-correction, as well as the capacity to utilize working memory. It is distinct from general intelligence, though it does correlate at lower levels of intelligence. Again, as in attention skills, these skills are vulnerable and easily disrupted in many circumstances, as they are largely acquired throughout childhood as an essential central process of competent development. Therefore, deficits acquired can be unknown or hidden until they are called on for future development. The range of tasks is wide, from inhibiting behavior in the absence of visible authority to planning how to accomplish several assignments due at the same time.

Though the cognitive aspects are difficult to quantify, the literature on these is substantial. However, the emotional and behavioral aspects of executive skills has not been studied as much (36). Executive skills act to regulate behavior (37,38), inhibit and manage emotions, tolerate frustration, and provide persistence. Notable is the result of limited empathy (ie, taking the position of the other). They are observed collaterally in any sound testing process, but are captured better, to the extent possible, in the "real-world" questionnaire approach discussed previously. The effect of impairment in these skills can be widespread and debilitating, especially as expectations for empathy and self-awareness increase in adolescence.

One of the questionnaires does differentiate these two factors. In the BRIEF (131), questions about such skills yield feedback for the behavioral regulation composite, as differentiated from another composite reflecting the cognitive aspect, metacognition composite. A list of tests that cover this wide-reaching domain is listed in Table 3.2.

TABLE 3.2 TESTS OF PROBLEM SOLVING AND EXECUTIVE FUNCTION

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Halstead Category (HCT) (39)	Versions exist for ages 5–9 and 9–14, as well as through adulthood; part of Halstead–Reitan battery	Machine and booklet forms; measures conceptualization and abstraction abilities
Wisconsin Sorting Test (WCST) (40)	Revised manual offers norms for ages 6.5 and above. Computerized and standard administration	Requires inference of correct sorting strategies and flexible use
Tower of Hanoi (TOH) (41)	Computer and standard administration; ages 4 and up	Taps working memory, planning, rule use, and behavioral inhibition
Tower of London—Drexel, 2nd Edition (TOLDX) (42)	Arrange balls on pegs to match picture; norms for 7 and up	Taps inhibition, working memory, anticipatory planning
Stroop Color-Word Test (43)	Well-known test, quick administration in paper form; several versions	Test of inhibition, selective attention, and switching sets
Matching Familiar Figures (MFFT) (44)	Must find identical match for stimulus picture; ages 6 and up	Measures impulsivity
Fluency Tasks Verbal and Design	Speeded tasks of response generation to verbal and nonverbal stimuli	Taps self-monitoring, initiating, and shifting; included in many batteries
Delis–Kaplan Executive Function System (D–KEFS) (45)	Battery of six subtests; ages 8 and above	Battery aids comparison of subtest scores

NONVERBAL/VISUAL-PERCEPTUAL FUNCTION TESTS

It is essential that before a clinician makes any judgments regarding one's deficits in sensory-perceptual abilities, the clinician concludes that primary visual systems are not damaged. This type of cognitive task is seen as one of the two major classes of cognitive input/output. From earliest infancy, humans already have sufficient visual perception to mimic another's facial expression (27). Related to the developmental aspects of childhood, by age 9, visual processes are integrated with tactile and proprioceptive functions. Tests of visuoperceptual, visuospatial, and visuomotor function are all within this domain and include the ability to discriminate between objects, distinguish between left and right, judge spatial orientation and the relationship among objects in space, copy a model, understand symbolic representations of maps and routes, and solve nonverbal problems. Concrete functional outputs include being able to navigate the environment and depth perception. Testing of these functions can illuminate visual field cuts, visual neglect, agnosia, and apraxia.

As with most domains of cognitive functioning, these tasks often involve other aspects of cognition, such as attention, memory, speed of thinking, and motor impairment. At the base is the requisite of normal visual acuity, which is screened for in the pediatric clinic setting or in school admission testing. However, after a brain injury or illness, the intactness of the basic perceptual components of nonverbal functioning should not be assumed. Issues like cortical visual defects or loss of binocular vision require examination by vision specialists; in more severe TBIs, such injuries are not uncommon. Consideration of the myriad factors involved in this domain's assessment requires examining other test performances to discern patterns, as well as factors that may affect visual function output but not be a deficit in these processes per se. The importance of these functions as a basic component of input and output of cognitive function is seen in their presence in all intellectual and neuropsychological batteries. Table 3.3 provides a representative listing of these tests.

LANGUAGE FUNCTIONING TESTS

The examination of language is imperative, as it is felt to be a central aspect of brain functioning and the foundation of human thought. Language development is well delineated. It proceeds from spoken to written competency in typically developing childhood. Receptive and expressive abilities require separate assessment, as do the modalities of written language. There is a distinct difference between speech and language. Speech as the mechanical aspect of oral communication is assessed as distinct from language per se. One element can be intact, while the other has significant impairment, and evaluation distinguishes between the two.

TABLE 3.3 TESTS OF NONVERBAL/VISUAL-PERCEPTUAL FUNCTION

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Rey–Osterrieth Complex Figure Test (ROCF) (46)	Copying of complex figure and a delayed recall condition; alternate forms exist for repeat administration; ages 6 and up	Taps planning, visual organization, and memory for complex visual information
Beery Developmental Test of Visual-Motor Integration. 6th Edition (VMI) (47)	Design-copying test of 24 forms of progressive difficulty; supplemental tests of visual perception and motor coordination; Ages 3+; procedures for younger children	Long-standing test with new additions; visuoperceptual, visuomotor integration is assessed
Facial Recognition Tests (48)	Requires direct matching and side profiles of photos of human faces; norms for ages 6+	Present in standalone tests, but also in batteries, including memory aspect; implications of right hemisphere function
Wide Range Assessment Battery of Visual Motor Ability (WRAVMA) (49)	Battery of three tests: drawing, matching, and pegboard. Can be administered individually or as a battery yielding a composite; ages 3–16	Sensitive to right cerebral dysfunction; nonverbal test of visual spatial ability
Judgment of Line Orientation Test (JLOT) (50)	Pictures of line segments whose position can be matched to 2 of 11 full-length lines; ages 7–74	Sound psychometrics yield standard scores and percentile for each subtest
Hooper Visual Organization Test (51)	Series of 30 line drawings of familiar objects that are divided into fragments; child must mentally reassemble fragments to determine what the object is; ages 5+	Measures visual analysis, synthesis, conceptual reorganization, and mental rotation

An acute injury most notably, even an illness, may produce a blatant aphasia, but this often resolves from its most dramatic state fairly quickly. Deficits remain that are more subtle, but important to address. Common deficits after injury differ between an adult and a child. In childhood and adolescence, deficits in word finding, dyscalculia, and problems with formulating written language are common (27). Functional problems, like difficulty with instructions or following commands, require analysis to determine the degree of linguistic difficulty versus other factors, such as attention or memory. As is always the case, the determination of concurrent difficulty in other tasks provides the diagnostic information to hone in on the core problem(s).

Components like phonological processing, confrontation naming, language comprehension (oral and written), and understanding the syntactic structure of language, as well as the productive aspects of language, should be covered. The profound impact of language deficits on academics makes it a particularly important aspect for assessment. Problems with reading comprehension are often seen, and because reading is one of the primary tools for learning once a child enters elementary school, the impact can be widespread on competent school functioning. Deficits in the understanding of phonemes are often an early sign of reading disability.

There are many well-accepted tests to measure all these aspects. For receptive language, the Peabody Picture Vocabulary Test-IV (52), Token Test—Second Edition (53), and the Bracken Basic Concept Scale—Third Edition (54) are widely used. These vary from single-word comprehension to grammatical/syntactical structure to linguistic concepts. Similar tests exist to investigate expressive oral language. Batteries like the Clinical Evaluation of Language Functions (55) or the Test of Written Language-2 (56) offer the advantages of batteries, while covering various aspects of language so that differential levels can be discerned.

A long tradition in neuropsychological evaluation is the evaluation of aphasia, the disturbance in the basic language capacity of the brain. This capacity begins at the level of auditory discrimination and phonological awareness, proceeding to words, then meaningful word combinations. The Boston Diagnostic Aphasia Examination (57), though its full utility with children has been questioned, has long been in use. Issues such as fluency skills, where the ability to generate words within a parameter, such as the beginning sound, or rapid naming are basic language skills that can be lacking due to developmental or acquired problems. Though they are not everyday language skills, they represent an automaticity of language that can affect more complex skills, such as reading.

MEMORY AND LEARNING TESTS

Memory is often a presenting problem in children with brain dysfunction. Memory involves cognitive mechanisms used to register, retain, and retrieve previous events, experiences, or information (58). All aspects of this activity need to be assessed to provide sound diagnostic information in addition to developing remediation or compensatory strategies. Questions of ecological validity are particularly cogent in memory evaluation, as necessary types of memory cannot be assessed in the testing situation. Adaptive behavior questionnaires and devices that attempt to incorporate real-life situations, like the Rivermead Behavioral Memory Tests, are useful to round out more traditional assessment tools, which are largely based on theoretical laboratory models. The nature of material to be remembered in everyday life is different, but so is a naturalistic setting, and the attendant natural distractions are part of many instances where memory is needed.

A full examination of memory covers a number of distinctions, including declarative/explicit versus implicit/procedural memory, recognition versus recall, encoding versus retrieval issues, prospective and remote memory, and short-term/working memory versus long-term memory. An important distinction is verbal versus nonverbal memory, and it should be included as a referral question in most situations.

Findings need to be viewed in the context of the recognized developmental changes in memory functioning through childhood (59). Developmental changes that mark the progression toward mnemonic competence are attributable to the child's growing proficiency in the use of compensatory strategies to aid encoding and retrieval of information.

In situations of TBI, there is a specific role for monitoring the time span where brain functioning was insufficient to record ongoing environmental input, referred to as posttraumatic amnesia (PTA). This is done through tracking orientation and return of continuous recall. The latter refers to the brain's resumption of the capacity to register everyday occurrences on an automatic basis. For pediatric rehabilitation, the Children's Orientation and Amnesia Test (COAT) was developed for this purpose by Ewing-Cobbs and colleagues (60), based on the Galveston Orientation and Amnesia Test for adults. The duration of PTA has been shown to more reliably predict recovery than the Glasgow Coma Score (GCS), the rubric used in general medicine to judge severity and, by implication, prognosis. Retrograde amnesia should also be assessed, representing the time span for which formation of long-term memory was disrupted, so that minutes, hours, and sometimes days prior to the injury are not recalled. This also requires serial monitoring, as restoration of retrieval processes results in more information being recalled as the brain recovers. For retrograde amnesia, the monitoring is essentially just patient responses to questioning of events leading up to the injury (Table 3.4).

SENSORY-PERCEPTUAL AND MOTOR TESTS

Although at times minimized or omitted entirely, tests of these functions can be illuminative for laterality issues as well in determining the extent of impairment in the corresponding cerebral hemisphere. Peripheral disorders must be ruled out as the cause of discrepancies or abnormal scores. There are well-established norms from age 3 and up pertinent to motor sequencing, various hand movements,

TABLE 3.4 TESTS OF MEMORY AND LEARNING

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Rivermead Behavioral Memory Test, 3rd Edition. Children's Version (61)	Tasks are analogues of everyday memory; has immediate and delayed tasks; two versions: adult (age 11+) and children (ages 5–10); four parallel forms	Novel approach with everyday tasks increases utility in case planning and remediation. May miss moderate to mild deficits; alternative forms very useful, though enough statistics are not given for full utility; shows general disruption
Wide Range Assessment of Memory and Learning 2 (WRAML 2) (62)	Traditional memory battery covers nonverbal and verbal, immediate, recognition, and delayed; ages 5–90	Excellent psychometrics; widely used; has a screening form
Test of Memory and Learning-2 (TOMAL-2) (63)	Ages 5–60; traditional battery; covers nonverbal, verbal, immediate, delayed, and cued recall	Good psychometrics; easy to administer
Child Memory Scale (CMS) (64)	Ages 5–16; battery; parallel structure of adult Wechsler Memory Scale	Widely used; enables comparison with IQ and achievement as part of Wechsler series
California Verbal Learning Test-C (CVLT-C) (65)	Ages 5–16; verbal memory assessed; short and long delay (20 min) procedures	Hard to score by hand; good psychometrics

and reciprocal coordination. This area includes tests of tactile discrimination and fine motor or hand–arm movements. Rates of competence between the sides in simple items and in items with gradually increasing complexity are done for both tactile and fine motor functioning. The techniques of A. R. Luria (66) are often used for fine motor examination, with elements of executive function abilities intrinsic to completion of the more complex movements. Specific tests would include the Grooved Pegboard (67) for skill motor movements—a timed task involving peg placement in holes at various orientations to the shape of the pegs. Though interpretation must be done in the context of other data, such tests can provide information about the course of a disorder. An example is in chronic hydrocephalus, where monitoring with tactile proprioception as in finger recognition and number-writing perception can signal progression of the cerebral pathology.

Brief Smell Identification (68) allows for standardized, forced-choice odor identification, with 12 microencapsulated odorants as a screening test for olfactory function. Many studies have documented a high incidence of olfactory dysfunction post brain injury in adults, correlated with higher-order cognitive skills that can be elusive to discern in a direct fashion. The role in the developing brain is less delineated. Norms have been developed from age 5 and up.

COMPUTERIZED ASSESSMENT

Within this area, a number of devices have already been listed under other sections, notably in the section titled Attention, Concentration, and Information Processing. The discussion here is relative to concussion. See "concussion" chapter (Chapter 12) regarding the use of computerized assessment of cognitive functions specific to abilities typically disrupted by concussion.

To characterize concussions presenting to an emergency room (69), a version is being developed for children aged 5 to 10.

COGNITIVE AND INTELLECTUAL MEASURES

A central component of all psychological assessment, usually included in nearly all testing, has been a measurement of intellectual or cognitive ability. There is a large body of literature on general intellectual testing as well as many theories, all beyond the scope of what can be included here. As this pertains to children, the purpose is typically to predict and plan for academic capacity and appropriate educational programming. Tests of this nature have also allowed clinicians and educators to detect students who may be at risk for learning problems and benefit from special services.

Of the major general cognitive tests, each is based on different theoretical models, as mentioned previously; however, all share a fundamental similarity: separate assessment of verbal and nonverbal skills, with scores combined to yield a general composite score. In the rehabilitation population, children whose illness or disability differentially affects verbal or visual-spatial skills require a more sophisticated selection, analysis, and understanding of tests. These children are more likely than the typical population to show significant differences and scatter on different types of skill sets, and composite scores may not provide much useful information. For example, a child who scores in the average range on visual-spatial tasks and in the impaired range on verbal tasks may be given an overall composite score in the low-average range—which does little to describe the child's actual abilities and even less in terms of guiding programming. Therefore, individual indexes and subscales must be examined in order to gain a clearer picture of the child's ability.

In cases of significant physical or sensory impairment, such as hemiparesis, clinicians are simply not able to fully and adequately assess the full range of intellectual functioning. Tests that require rapid bilateral fine motor skills have to be modified, thus negating valid interpretation, and replaced with less involved tests that require pointing. These tests cannot be assumed to measure precisely the same skills—and may even be skipped altogether in favor of using scores on verbal-response tests as the primary index and then assuming that the score reflects general capacity across domains. This practice is ill-advised even in normal populations, much less in children where there is evidence of neurologic impact that may differentially affect various skill sets. In general, with children like these, scores on cognitive tests should be carefully interpreted, with cognizance of limitations, and used *as part* of a larger body of neuropsychological assessment that applies more sophisticated and specified measures to best assess the full span of skills that are commonly affected by illness or disability.

The Wechsler scales include the Wechsler Intelligence Scale for Children, 4th Edition (WISC-IV) (70), the Wechsler Adult Intelligence Scale, 4th Edition (WAIS-III) (71), the Wechsler Preschool and Primary Scale of Intelligence, 3rd Edition (WPPSI-III) (69), as well as an abbreviated battery, the Wechsler Abbreviated Scale of Intelligence (72). The factor structure of the WISC-IV was significantly changed from the previous editions. The WISC-IV includes a full-scale score made up of four separate composites, each of which is made up of several different subtests. The four composites are verbal comprehension, perceptual reasoning, working memory, and processing speed. The core working memory subtests are primarily verbal in nature, and the core processing speed subtests are primarily nonverbal in nature. The WISC-IV is designed for use with children aged 6 to 16 years. The WAIS-IV is used with individuals aged 16 to 90 years. The factor structure of the WAIS-IV

was also updated. Similar to the WISC-IV, it yields a full-scale score comprising verbal, performance (nonverbal), working memory, and processing speed scaled scores. Each index is made up of several different subtests. The WPPSI-III has two different score structures, depending on age. For children aged 2.5 to 4 years, there is a full-scale score comprising verbal, performance, and general language composites. For children aged 4 to 7 years, 3 months, there is one additional composite score: processing speed. Important considerations in the assessment of preschool-age children are addressed in the following section, "Instruments for Use With Young Children." The WASI-II is an abbreviated measure of intellectual functioning that measures both verbal and performance abilities, leaving out working memory and processing speed. This will often be used when other measures tapping these skills are administered.

The Stanford-Binet Intelligence Scales, 5th Edition (73) is designed for use with individuals aged 2 to 89 and up. The full-scale score is made up of five factor indexes: fluid reasoning, knowledge, quantitative reasoning, visual-spatial processing, and working memory. Each factor index includes separate assessments of nonverbal and verbal skills. It should be noted that some of the "nonverbal" tasks require significant receptive language skills, which may complicate interpretation in a child with a basic discrepancy in verbal and nonverbal skills.

The Kaufman Assessment Battery for Children, 2nd Edition (K-ABC-II) (74) was designed for use with children aged 3 to 18. It is unusual in that guidelines are provided for interpreting results within two different theoretical models: the Luria neuropsychological model and the Cattell–Horn–Carroll psychometric model. Using the Luria model can provide some coherence within a broad neuropsychological assessment. Under this model, there are five scales (sequential processing, simultaneous processing, planning ability, knowledge, and learning ability), each comprising multiple subtests. There is also a distinct nonverbal index that can be administered entirely through nonverbal gestures and responses, which can be useful for children with certain disabilities.

INSTRUMENTS FOR USE WITH YOUNG CHILDREN

Tests of infant ability have been developed in an attempt to measure the developmental status of infants and young children. Such tests are primarily useful in describing current developmental status, with minimal relationship of these early childhood competencies to skills considered crucial during later developmental phases (75). Predictive validity is considered viable only with infants who are significantly developmentally delayed in the first year of life (76,77). Furthermore, tests of infant abilities heavily emphasize assessment of motor skills and cooperative behaviors, which are areas compromised in a child with chronic or acquired disability, causing additional complications for achieving test validity in this population.

Research generally indicates that the younger the child, the less predictive intelligence tests are of later test scores and academic performance as the child ages (78,79). Although this was a common belief and well proven in the 1960s and 1970s, this is currently being examined in the research to discern if there is moderate predictability of some of these tests (80). The assessment of young children typically requires adaptation and expansion of existing tests to obtain reluctant and valid information. Factors to be considered are that the young child cannot be expected to perform on request and exceptional efforts may be necessary to elicit the degree of responsiveness and cooperation necessary to obtain sufficient and meaningful information. According to Stevenson and Lamb (81), an infant's response to a strange-adult-influenced test performance and "sociably friendly" infants scored higher on measures of cognitive competence. Ulrey and Schnell (75) noted that preschool children have had minimal experience with test situations, show minimal concern for responding correctly, and have limited experience with the feedback process that is contingent on being right. Usually, the process of merely asking young children to complete a task may not yield an accurate indication of their capabilities. It is, therefore, incumbent on the examiner to make a judgment about the extent to which the child's performance represents optimal functioning. The likelihood of obtaining ecologically valid information can be enhanced by incorporating observations and analyses of infants' or young children's interactions with the environment (eg, parents, siblings, or caregivers) during spontaneous play.

The Bayley Scales of Infant and Toddler Development, 3rd Edition (Bayley-III) (82) can be used to measure cognitive and motor ability in children aged 1 to 42 months. The cognitive scale measures memory, visual preference, visual acuity, problem solving, number concepts, language, and social development. The language scale measures social communication, semantics, morphology and syntax, prelanguage vocalizations, and comprehension. (Separate receptive and expressive language subtests are included.) The motor scale measures functional grasp and hand skills, object manipulation, visual–motor integration, head control, trunk control and locomotion, motor planning, and quality of movement. (Separate fine and gross motor subtests are included.) There is also a social-emotional scale (covered in the section titled Psychosocial Evaluation) and an adaptive behavior scale that is the same as the early childhood version of the Adaptive Behavior Assessment System-II (83), which is covered in the section titled Adaptive Behavior. The Bayley-III is considered the best available instrument for infant assessment (15).

The Brazelton Neonatal Assessment Scale (BNAS) (84) is administered to infants between 3 days and

4 weeks of age to generate an index of a newborn's competence. This scale includes 27 behavioral items and 20 elicited responses to assess. Test scores may be most useful when the test is repeated over the first several weeks of life, so that *changes in scores* can be examined to assess the infant's ability to respond to parenting and recover from the stress of birth. It is this recovery pattern that predicts later functioning in childhood (85). Scores have also been used to teach parents how to provide sensitive and confident care to their infants, with small to moderate effects (86).

ALTERNATIVE TESTS OF COGNITIVE FUNCTION

Alternative tests of cognitive ability are of particular utility with rehabilitation populations, where patients often have specific impairments (eg, motor impairments, sensory impairments) that preclude the valid use of more common measures. Some of the alternative measures rely less on verbal responding, or reduce requirements for motor output or speed of responding. In a pediatric rehabilitation population, it is often necessary to use alternative assessment measures to accommodate a range of conditions that may interfere with the child's ability to meet requirements of standardized test administration on traditional measures.

Given that many of these alternative measures were designed for particular populations, scores generated are not interchangeable with scores of the major intelligence scales. Furthermore, the special formatting of these tests limits the applicability of results to "real-world" environments, where such intensive accommodations are not always made, and scores may not be as predictive of actual functioning in major settings such as school, home, or community. These instruments may be most useful as screening or supplemental tools in the assessment or interpretation processes.

The Universal Nonverbal Intelligence Test (UNIT) (87) is a test of intelligence that is designed to be completely nonverbal. It can be used with children aged 5 to 17 years. Administration is done through eight specified pantomime gestures. Responses are also entirely nonverbal, and consist of pointing, paper–pencil, and manipulating items. Multiple standardized teaching items are provided to help ensure that the examinee understands the purpose of gestures. The UNIT is most useful for children who have significant hearing or oromotor limitations, or who do not speak English. Relatively normal fine motor functioning is required for valid use of the test. There are four overlapping scales (memory, reasoning, symbolic, and nonsymbolic), and a full-scale score. The nonsymbolic scale is designed to measure abstract symbolic functioning, which is typically measured through *verbal* scales on cognitive tests. Some children who *can* hear seem to find the examiner's complete reliance on nonverbal pantomime to be somewhat off-putting at first.

The Leiter International Performance Scale-3rd Edition (Leiter-3) (88) is a nonverbal test of intelligence for use with individuals aged 3 to 75 years. It retains the best of the widely used Leiter-R subtests and includes a number of new measures. It also uses a refined block-and-frame format plus foam manipulatives for easier manipulation by all examinees, which was a disadvantage of the previous edition. Leiter-R subtests were combined and items with similar difficulty levels were removed, reducing the number of subtests from 20 to 10. The Leiter-3 measures nonverbal cognitive, attentional, and neuropsychological capabilities.

This test is useful with individuals with hearing or oromotor limitations, or who do not speak English.

The Comprehensive Test of Nonverbal Intelligence, 2nd Edition (C-TONI-2) (89) is designed to assess intelligence in individuals aged 6 to 89 years. It includes an overall composite and two subscales: pictorial and geometric, which include two subtests each. The test can be administered orally or in pantomime (and is standardized using both). The option of oral administration is for use with children who are not hearing impaired, as these children can be confused when a test is administered completely nonverbally. The C-TONI has the additional advantage of requiring no more complex motor response than pointing to the correct answer. Tests requiring only pointing are sometimes further modified by clinicians to accommodate severely impaired children for whom even pointing is too difficult (eg, the examiner points to each option and the examinee provides indication through predetermined head or trunk movements when the correct choice is reached).

Raven's Progressive Matrices include three separate forms: Coloured Progressive Matrices (90) designed for children aged 5 to 11, Standard Progressive Matrices (91) for children aged 6 to 17, and Advanced Progressive Matrices (92) for older adolescents and adults, including individuals suspected of above-average intellectual ability. The tests are brief measures made up of abstract visual arrangements, with the examinee required to select one of multiple choices to complete the arrangement. Instructions can be administered orally or through pantomime. These tests can be used with children with oromotor or hearing impairments, or who do not speak English. The examinee responds by pointing, so it is useful for children with motoric impairment. They are limited as a measure of general cognitive functioning because they assess only one specific type of skill, which may be particularly problematic in a neurologic population where highly specific strengths and weaknesses are often seen.

The Peabody Picture Vocabulary Test-III (PPVT-III) (93) is a receptive vocabulary test, where the respondent is given a vocabulary word and points to the best match from a series of pictures. It is sometimes used as a screening device to estimate verbal cognitive abilities for students with expressive speech and/or motor difficulties, though, of course, great caution is warranted, as the

PPVT-III assesses only a single skill set. Visual-perception and native English skills are required. The PPVT-III can be used with individuals aged 2.6 to 90+ years.

As noted previously, the K-ABC-II (74) includes a distinct nonverbal index that can be administered entirely through nonverbal gestures and responses, which can be useful for children with certain disabilities. This test requires relatively complex and rapid motor responding, and would not be appropriate for use with individuals with even mild motoric impairment. Table 3.5 provides a complete listing.

ACHIEVEMENT TESTS

The assessment of academic achievement is an imperative component of the evaluation of children and adolescents, as school is the "work" of childhood. An important task of assessment is separating academic knowledge from the rate of production (referred to as academic fluency) in children with response speed deficits due to motoric impairment or brain injury. Many tests of achievement include a speeded component. Overall scores may be less helpful than specific scores that separate out fluency and basic skills. In addition, academic testing in youth with recent-onset illness or injury may overestimate long-term academic capacity. Academic testing generally measures previously learned knowledge, which may be intact in children whose illness or disability has not yet affected schooling. Whether a child can continue to make progress is a critical question. This is particularly true in brain-injured youth whose deficits in attention, executive functions, and anterograde memory have a strong impact on mastery of new academic skills, and applies to other types of recent-onset conditions that place higher coping demands on the child, leaving fewer resources available for basic academic learning. Often, frequent retest is necessary to track progress.

TABLE 3.5 ALTERNATE TESTS OF COGNITIVE ABILITY

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Universal Nonverbal Intelligence Test (UNIT) (87)	Nonverbal test that measures both symbolic and nonsymbolic cognitive skills in the nonverbal domain. Age range: 5–17.	Requires some fine motor functioning; designed to reduce cultural bias; easy to administer; useful with individuals with auditory or oromotor limitations, or who do not speak English.
Leiter International Performance Scale-Revised (Leiter-R) (88)	Nonverbal test developed for use with hearing- or language-impaired subjects; measures visual-spatial reasoning and nonverbal attention and memory. Age range: 3–75.	Motor responses are relatively simple, but some items are scored for speed, so motor impairments may affect results. Useful with individuals with auditory or oromotor limitations, or who do not speak English.
Comprehensive Test of Nonverbal Intelligence (C-TONI) (89)	Nonverbal test with pictorial and geometric subscales to measure concrete and abstract nonverbal skills. Only motor skill required is pointing, and this can be further adapted for severely motor-impaired individuals. No time limits.	Nonverbal test with option for oral administration in English-hearing individuals. Useful for individuals with combined limited motor functioning and auditory or oromotor limitations or who do not speak English.
Raven's Progressive Matrices Tests (90,91)	Measures nonverbal reasoning; three different forms for different age ranges; limited motor skills required; advanced version is useful for individuals considered to have above-average intelligence; no time limits.	Limited in that it uses a single type of task; useful for individuals with auditory, oromotor, or physical disabilities, or who do not speak English.
Peabody Picture Vocabulary Test-III (PPVT-III) (93)	Multiple-choice test of receptive vocabulary; for individuals aged 2.6–90+; pointing is the only response required, and further adaptations can be made for severely motor-impaired; no time limits.	Useful as a screening device for measuring verbal functioning in children with significant expressive verbal or motor impairments; sometimes used to estimate general cognitive functioning in individuals who cannot participate in other types of assessment, but should be interpreted with great caution.
Kaufman Assessment Battery for Children-II (KABC-II) (74)	General intelligence battery that includes a nonverbal index that can be administered entirely without spoken language. Relatively complex and rapid motor responses are required.	Suitable for individuals with auditory or oromotor impairments, or non-English speakers; not for use with individuals with even mild motor impairment.

Some of the more frequently used, individually administered, norm-referenced, and wide-range screening instruments for measuring academic achievement spanning kindergarten through twelfth grade include the Kaufman Test of Educational Achievement, 2nd Edition (KTEA-II) (93), the Wechsler Individual Achievement Test, 3rd Edition (WIAT-II) (94), and the Woodcock–Johnson Psychoeducational Battery, Third Edition (WJ-III) (95). The Wide Range Achievement Test, 4th Edition (WRAT-IV) (96), is frequently used, but is a brief measure that yields limited information. The Peabody Individual Achievement Test-Revised (97) addresses generally similar content areas as the other major assessment tools, but minimizes the verbal response requirement by using a recognition format (eg, point to correct response based on four choices). Although this format may allow assessment of children presenting with certain impairments, language or motor, the results may not provide the best indication of expectations for student performance in the classroom, where recall and more integrated answers are required.

New assessment guidelines under the Individuals with Disabilities Education Act (IDEA, 2004) for diagnosing learning disabilities in public education settings include options for using response to intervention (RTI), which is a process of assessing progress in skill acquisition in response to scientifically supported interventions, using frequent brief assessments rather than a single cluster of standardized testing. While RTI is not specified for use in qualifying children under other special education diagnostic categories, such as health impairment, orthopedic impairment, sensory/physical impairment, or brain injury, the RTI model provides a potential structure for assessing *progress* in the school setting.

Table 3.6 provides a listing of achievement measures.

ADAPTIVE BEHAVIOR

Adaptive behavior includes behaviors and skills that reflect competence to meet the demands of everyday living in order to achieve a developmentally appropriate level of independence. The American Association on Mental Retardation (AAMR) distinguishes three major categories of adaptive functioning. Conceptual skills include language, functional academics, and self-direction. *Social* skills include establishing friendships, social interaction,

TABLE 3.6 MEASURES OF ACHIEVEMENT

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Kaufman Test of Educational Achievement-II (KTEA-II) (93)	Reading (decoding and comprehension), math (computation and applications), and written language composites (spelling and composition), as well as additional subtests measuring reading-related skills and oral language. Ages 4.6–25.	Age- and grade-based norms provided; norms broken down by fall, winter, spring; reading-related subtests help identify specific deficits in phonological awareness or rapid naming.
Wechsler Individual Achievement Test-III (WIAT-III) (94)	Subtests measure pseudoword decoding, word reading, comprehension, numerical operations, math reasoning, written expression, spelling, oral language, and listening comprehension. Ages 4–50.	Age- and grade-based norms provided; norms broken down by fall, winter, spring; conormed with the Wechsler Intelligence Scale for Children-IV to promote statistically sound comparisons between IQ and achievement scores.
Woodcock–Johnson III Tests of Achievement (WJ-III) (95)	Scales assess reading, oral language, mathematics, written language, and knowledge. Separate scales assess basic skills, applications, and fluency for reading, math, and written language. Multiple additional scales of highly specified skills are included. Ages 2–90+.	Age- and grade-based norms provided; scoring provided through use of computer software only; lack of hand-scoring option limits the clinician in interpretation in some cases; specific fluency scores useful in populations with processing speed deficits; written expression subtest relatively simplistic.
Wide Range Achievement Test-IV (WRAT-IV) (96)	Subtests include sentence comprehension, word reading, spelling, and math computation. Ages 5–94.	Brief measure that does not assess some critical aspects of academic functioning.
Peabody Individual Achievement Test-Revised (PIAT-R) (97)	Includes subtests for general information, reading recognition, reading comprehension, mathematics, spelling, and written expression. Ages 5–18.	Uses a recognition format that accommodates individuals with language and motor impairments; measures relatively limited set of skills compared to other tests.

and social comprehension. *Practical* skills include basic self-care skills and navigation of home, school, and community tasks and environments. In later adolescence, vocational functioning is also assessed as part of the practical domain.

Deficits in adaptive behavior are one of the core criteria in determining a diagnosis of mental retardation, along with significantly impaired intellectual functioning. Adaptive functioning is assessed primarily through structured interviews and rating scales completed by persons familiar with the child in natural settings, such as parents and teachers. These scales are open to the response bias inherent in this type of assessment, but are also directly linked to programming assistance. There is great utility in using responses to adaptive skills to identify target skills for rehabilitation. Several issues are especially noteworthy in using these assessments with rehabilitation populations. First, adaptive scores may be disparate with intellectual testing scores in a TBI population, because they represent more procedural learning and are often less affected directly after the injury. The failure to gain subsequent abilities can be a source of substantial disability as time goes on, due to impairments in sensory or cognitive abilities. Second, in contrast to individuals with developmental mental retardation, who may be expected to show a general pattern of mastery of easier skills and nonmastery of more difficult skills on each scale, the rehabilitation population is more likely to show uneven peaks and valleys across skills even within the same domain. For example, a person with motoric impairment may struggle with some "easier" self-care skills, but have the cognitive and adaptive ability to handle more "difficult" skills in the same domain. In these individuals, standardized scores may not provide a meaningful picture, but analysis of specific items can provide direction for rehabilitation programming.

The Vineland Adaptive Behavior Scales-II (98) is a widely used set of scales that has four forms: Survey Interview, Parent/Caregiver Rating, Expanded Interview, and Teacher Rating. Each assesses four broad domains. The communication domain assesses expressive, receptive, and written communication. The daily living skills domain assesses personal, community, and domestic skills. The socialization domain assesses interpersonal relationships, play and leisure time, and coping skills; the motor skills domain assesses fine and gross motor skills for young children. The domain scores are combined to yield a composite index. A maladaptive behavior domain surveys inappropriate social or behavioral displays. The survey interview and rating scales take 20 to 60 minutes to complete, while the expanded interview is lengthier. The second edition includes updated content and increased coverage of early childhood adaptive behavior for use down to early infancy.

The Adaptive Behavior Assessment System-II (ABAS-II) (83) includes five forms, each taking 15 to 20 minutes to complete: Parent/Primary Caregiver Form for birth to 5 years, Teacher/Daycare Provider Form for children aged 2 to 5 years, the Teacher Form for ages 5 to 21 years, the Parent Form for ages 5 to 21 years, and the Adult Form for ages 16 to 89. In the second edition of the system, the domains are closely aligned with the AAMR definition of adaptive behavior. The conceptual domain assesses communication, functional academics (or pre-academics), and self-direction. The social domain assesses leisure and social skills. The practical domain assesses self-care, home/school living, community use, health and safety, and, for older adolescents and adults, work skills. The scales are well validated. Table 3.7 provides a complete listing of these tests.

PSYCHOSOCIAL EVALUATION

The assessment of psychosocial status has different conceptual bases, depending largely on the age of the child. A multimethod, multisource assessment is critical, as

TABLE 3.7 MEASURES OF ADAPTIVE FUNCTIONING

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Vineland Adaptive Behavior Scales-II (99)	Age: Birth to 90 years. Measures four domains: communication, daily living skills, socialization, and motor. Also includes a maladaptive behavior scale.	Assessment of adaptive motor skills relevant for a rehabilitation population. Rating scale and interview formats available.
Adaptive Behavior Assessment System-II (ABAS-II) (83)	Age: Multiple scales covering birth to 89 years. Measures three domains: conceptual, social, and practical.	Composite areas specifically match AAMR guidelines.

Abbreviation: AAMR, American Association on Mental Retardation.

different sources are sensitive to different areas of functioning (100). Structured interview, observational methods, performance evaluation, and careful analysis of both medical data and psychosocial variables should be combined, and, where possible, multiple sources of information should be included, such as parents, teachers, and child self-report.

CAVEATS

One of the trickiest issues in psychosocial assessment in rehabilitation populations is the need to account for the biologic factors on assessment results. Most psychosocial assessment tools are not specifically designed for use with children with disabilities or chronic illness, making their interpretation of results questionable. There is to be expected a level of adjustment that exists in children with disabilities or chronic illness. While children with chronic physical conditions appear to be at increased risk for psychological adjustment problems, the majority of children in this population do not show evidence of maladjustment (100). Furthermore, assumptions based on group membership by disability or medical condition can be inaccurate. For example, intuitive reasoning would indicate that individuals with disfigurements, such as amputations or burns, would be particularly affected. Such is not the case, however, as demonstrated in research of these groups (99).

It is important to be aware that some items on psychosocial assessment scales can elicit medical as opposed to psychological distress. Particularly in children, "somatization"—or the tendency to express high levels of physical symptoms—is often assessed in scales measuring emotional functioning. A high level of somatization is considered indicative of internalizing problems such as depression and anxiety in general child populations, and high somatization scores can lead to high scores on composite scales meant to measure general internalizing problems. Obviously, in youth with chronic illness, the extreme physical symptoms relating to the medical condition may, *even in the absence of other areas of significant symptomology*, yield a score on the somatization subscale that is high enough to lead to elevated "total" emotional symptoms scores. It is incumbent on the professional to analyze the general profile and individual items in these cases. If there are low rates of *other* indicators of emotional distress besides those symptoms specific to the medical condition, it is important not to overinterpret the elevated scores. At the same time, high total scores should not be disregarded just because they are in part due to medical symptoms, as this population does frequently show elevated symptoms of distress, even when somatic items are not included in scoring (101). An intimate familiarity with the items making up the measure and the specific variables associated with the individual child's medical condition is required for psychosocial assessment in this population. Physicians should be wary of scores provided by school and community clinicians who are not specifically familiar with the challenges in the assessment for this population. *Referral to clinicians who specialize in pediatric rehabilitation should be strongly considered when psychosocial concerns are an issue*.

Unique to the arena of personality of psychosocial functioning is the empirically based or criterion-group strategy of assessment. This approach grew in response to the serious liabilities presented by self-report tests, which used items that had face validity. For example, an item that asks about arguing with others was a direct question, just as could be asked in a live interview. There are great liabilities to that approach; it assumes that subjects can evaluate their own behavior objectively, that they understand the item in the way it was intended, and that they chose to respond candidly. In a radical departure, the developers of what came to be known as the Minnesota Multiphasic Personality Inventory (MMPI; now in the second edition and restructured form—MMPI-2-RF) formulated the test with the main premise that nothing can be assumed about the meaning of a subject's response to a test item—the meaning can be discerned only through empirical research. Items are presented to criterion groups, such as depressed, schizophrenic, or passive–aggressive personality disorders, and control groups. By their answers as a diagnostic group, the items *become* indicative of a given disorder or personality outplay, regardless of what the content of the items was or an intuitive judgment of what it *should* indicate. This approach also allows for the determination of respondent's bias—whether an adolescent self-reporting, as in the case of the Minnesota Multiphasic Personality Inventory-Adolescent (MMPI-A), or parents filling out a behavioral checklist such as the Personality Inventory for Children-2.

In young children, temperament is a more cogent concept than that of personality. The dynamics of psychological functioning are the effect of innate temperament in interaction with parents and other caregivers within the basic sensorimotor exploratory nature of infancy and early childhood. If school is children's work, play is the work of this youngest group. What an interview or a self-report measure yields in older children, the observation of play provides in the preschooler. To quote Knoff (102), "This information reflects the preschooler's unique perceptions of his or her world, perceptions that are important in any comprehensive assessment of a referred child's problems." Projective techniques such as the Rorschach are not recommended in this population because of the need to interpret ambiguous visual stimuli. The active developmental maturation of visual-perceptual systems and the attendant normative variability mitigate against the appropriateness in preschoolers.

INDIVIDUAL ASSESSMENT TOOLS

Functional behavior assessment (FBA) is highly appropriate when young children, as well as older youth, with disability or illness are displaying significant behavior problems (103). When the ability to effectively communicate or independently access one's wants and needs is inhibited by cognitive or physical disability, rates of inappropriate behaviors can increase as the child learns (sometimes subconsciously) that these behaviors can effectively serve a function. FBA is a structured assessment method for determining the underlying function (ie, purpose) of inappropriate behavior. This assessment method has the advantage of being directly linked to intervention strategies—when a function is identified, environmental interventions can be developed to teach the child to use more appropriate behaviors to meet his or her purpose. There is an adaptive emphasis for children who cannot use developmentally appropriate language or mobility, and children with even severe impairments in cognitive, language, sensory, or motor functioning can be assessed through this method. Functional behavior assessment includes structured interviews examining the antecedents and consequences of behavior, structured observations of behavior in naturalistic settings to identify environmental mediators, and experimental manipulation of environmental conditions (functional analysis) to determine whether behaviors serve to meet children's need for attention, tangible items or activities, to escape from nonpreferred situations, or to meet internal needs, such as the release of endorphins through self-injury.

Transdisciplinary play-based assessment (TPBA) (104) is a standardized observation of play. It provides an exhaustive listing of developmentally cogent play behaviors under four domains: cognitive, language and communications, sensorimotor, and social-emotional development. It allows the child to engage in the most natural of activities, but is limited in that there may not be an expression of a specific behavior of interest but rather a global picture of the child in interaction with the environment. Because of the limitations of individually administered tests in the young child, this acts as cross-validation of parental report and is less influenced by the demanding characteristics of traditional testing. The advantage of hearing spontaneous language production is particularly useful, for this is often the primary *shutdown* of younger children in an evaluation setting (105). There are other systems for play observation. Some are designed for the more evocative structure of play designed to tap certain themes (eg, abuse) used in children. In the rehabilitation population, nonpathologic issues such as adjustment and developmental integrity predominate, so the TPBA offers an excellent choice.

The Bayley Scales of Infant and Toddler Development, 3rd Edition (82) provides a normative framework for this domain by providing scaled scores for the popular Greenspan Social-Emotional Growth Chart (106), which is a parent-report instrument to assess early indicators of social-emotional functioning in children aged 0 to 42 months.

The MMPI-A (107) is based on the criterion-group strategy described in the introductory comments to this section. It is the first revision of the original MMPI specifically for use with adolescents. For the original test (MMPI), adolescent norms were developed in the 1970s, but it was only a downward extension at best. Now, new items tap specific adolescent developmental or psychopathologic issues. There are new supplemental scales that give feedback relative to alcohol and drug problems and immaturity. There are 15 new content scales in addition to the original 10 clinical scales. Development of the validity and response bias of the subject was expanded by devising response-inconsistency scales.

The original MMPI interpreted with adolescent norms had been used extensively with adolescent medical populations, including those with physical disability (108). For the development of the MMPI-A, extensive rewriting and some revision of test items were done. A national representative adolescent sample was used for normative data (not the case in the original MMPI). The new length is 478 test items presented in a booklet form, with true/false response. The reading level required is best considered to be seventh grade, although it had been designed with the goal of fifth-grade comprehension. In actuality, the range is from fifth to eighth grades. The test is available in an audiotape format as well, which takes about 90 minutes. Each item is read twice. This aspect was designed for access by the visually impaired, but doubles for individuals who have reading comprehension problems. The language comprehension level required for the audiotape format is fifth grade. A computer-administered form is also available that presents items singly and with a response entered on the keyboard.

The effective use of the MMPI-A with pediatric rehabilitation patients is contingent upon cautious interpretation. For example, elevated scores on scales such as "hypochondriasis" or "lassitude-malaise" will be interpreted differently in a patient with chronic illness than in general populations. A correction factor is recommended for use with spinal cord injury to obviate responses to items that reflected the reality of the medical condition, as opposed to the criterion value assigned to the item (109). Recommended uses for the MMPI-2, which would also appear appropriate for the MMPI-A, in medical assessments include assessment of response bias, as the validity scales allow for assessment of the accuracy of the patient's self-report, identification of emotional distress factors relating to the medical condition that may influence recovery, and comorbid psychiatric conditions that would be expected to affect

recovery and participation in rehabilitation. Attempts to use the MMPI-2 (and likely the MMPI-A) to differentiate between organic and functional conditions are discouraged, as research suggests that elevated scores on scales suggestive of somatic preoccupation can reflect the *effect*s of the medical condition (110). The updated version of the MMPI-2 is now called the MMPI-2-Restructured Form (RF). This new form has about 200 fewer questions, although the same normative sample was used to create it.

The Personality Inventory for Children, 2nd Edition (111) is a behavior rating scale for children aged 5 to 19. It comprises 275 items to be completed by a parent. There is a brief form that takes about 15 minutes to complete. Composite scales include cognitive impairment, impulsivity and distractibility, delinquency, family dysfunction, reality distortion, somatic concern, psychological discomfort, social withdrawal, and social skill deficits. Three validity scales are designed to assess response biases, including inconsistency, dissimulation, and defensiveness, that may invalidate responses. Sattler (112) finds that additional research is needed on the reliability and validity of this new version of the scale, and there have been some concerns noted about the use of previous versions with specific rehabilitation populations—notably those with brain injury.

The Achenbach System of Empirically Based Assessment (113,114), including the Child Behavior Checklist for Ages 6–18 (CBCL/6–18), the Child Behavior Checklist for Ages 1.5–5 (CBCL/1.5–5), the Youth Self-Report (YSR), and Caregiver-Teacher Report Forms (TRF), are commonly used measures of psychosocial adjustment. They were each developed through factor analysis (or the *statistical* grouping of items into clusters/scales, as opposed to using clinical judgment to group items), but also include *DSM*-oriented scales developed through clinical judgment. Broad domains include internalizing symptoms and externalizing symptoms. The CBCL/6–18, TRF, and YSR each include 112 items in eight scales. The CBCL and TRF are designed for completion by parents or teachers, respectively, of children aged 6 to 18 years. The YSR is designed for self-report of adolescents aged 11 to 18, and requires a fifth-grade reading level. The CBCL/F/2-5 and Caregiver–Teacher Form, for use with younger children, each consist of 100 items, separated into seven and six scales, respectively. The scales are commonly used in children with chronic physical conditions (99). Limitations of its use with children in this population include limited sensitivity to milder adjustment problems, a possible confound by medical symptoms, incomplete assessment of social functioning, and methodological concerns (115).

The Behavior Assessment System for Children-2 (BASC-2) (116) includes three parent rating scales (Preschool, aged 2–5 years; Child, aged 6–11; and Adolescent, aged 12–21); three teacher rating scales, following the same age ranges; and three self-report of personality scales (Child, aged 8–11 years; Adolescent, aged 12–21 years; and Young Adult, aged 18–25 years, attending a postsecondary school). Each scale takes 20 to 30 minutes to complete and requires a third grade reading level. Parent rating scales include composite scores for adaptive skills, behavioral symptoms, externalizing problems, and internalizing problems. Teacher rating scales measure these four areas and add a school problems scale. The self-report scales include composite measures of emotional symptoms, inattention/hyperactivity, internalizing problems, personal adjustment, and school problems. The BASC-2 scales also include several indexes to measure response sets that would indicate invalid scores, such as high rates of negative answers, high rates of positive statements, endorsement of nonsensical or implausible items, or inconsistent responses. The BASC-2 system is well validated and provides an integrated multisource system of assessment (114).

The Rorschach Inkblot Technique (117) remains a widely used test in children and adolescents. It is the classic technique of 10 inkblots presented with the instruction to say what it looks like to the examinee. An alteration in administration with younger people is to follow up each card with the inquiry, asking why it looked like whatever the response was, whereas with adults, this is done only after all blots are viewed. Normative data on this technique for children and adolescents began appearing in the 1970s; however, these are not representative of the general population, being over-representative of children with above-average intelligence, with incomplete attention to race and socioeconomic status (117). Despite the fact that some norms exist down to age 2 years, most authors agree that the Rorschach should not be used with children below the age of 5 years. There is little experience with this type of test in assessing the type of adjustment issues common to the rehabilitation population. Therefore, it should be used guardedly.

Children's Apperception Test (CAT) and Thematic Apperception Test (TAT) (116) represent another type of projective test, but this time, the stimuli are ambiguous pictures and the subject is asked to make up a story concerning what is happening, what led to the scene in the picture, and what will happen next. It requires considerable skill on the part of the examiner, and should be given only by a professional, as is the case with all projective techniques. There is usually follow-up questioning about the story given, and the recording is verbatim. There are no real normative data on the CAT, but some authors believe that it remains a powerful technique in discerning children's personalities (118). Some believe it taps themes of confusion and conflict, with the child's resolution being a central focus of interpretation. It is based on the author's personality theory as opposed to a pathologic model. The entire set contains 20 cards, although a standard administration uses only selected pictures. Over the years, individual cards have been identified as being

particularly useful with certain age groups. There are concerns regarding lack of adequate reliability and validity of data (112).

In these days of cost-efficiency considerations, more specific, shorter measures are of great utility. The choice of a specific construct is often suggested by the results of other examinations or by knowledge of the presenting problem. Anxiety is a common correlate of chronic physical conditions (119). The Revised Children's Manifest Anxiety Scale for Children (RCMAS) (120) is a single-construct measure of anxiety. The RCMAS has 37 short statements to which the child responds yes or no. There is a total anxiety score, as well as a lie subscale that examines the candidness and honesty of the response set. The brevity of the instrument results in the three anxiety subscales that can be generated but are of limited use. The standardization sample was large and representative of socioeconomic status, demographics, race, and gender. Validity and reliability are extensively reported in the manual and are helpful in informed interpretation. The reading level is third grade, so a wide variety of children and adolescents can use this device. Because of its brevity and specificity, it should be only one part of a battery.

The Children's Depression Inventory-2nd edition (CDI-2) (121) is a well-recognized self-report measure of depressive symptoms in children aged 7 to 17 years. There are five subscales: negative mood, interpersonal problems, ineffectiveness, anhedonia, and negative self-esteem. Reliability for the total score is stronger than for subscales. Though a popular measure, questions have been raised about the psychometric properties (112) of the first edition. Although research on the newest edition is sparse at this point, this was to be one of the issues addressed.

The Behavior Rating Inventory of Executive Function (BRIEF) system (122) includes a parent version (ages 5–18) a teacher version (5–18), a preschool version of Parent and Teacher Rating Scales that can be completed by parents or teachers/daycare providers (ages 2–5), and a Self-Report (ages 11–18). The behavioral rating of executive functioning is an important addition to the assessment of psychological functioning in any child with neurologic impairment. Soliciting the observation of executive functioning in natural environments is especially important in light of previously mentioned concerns regarding ecological validity of clinical tests of executive functioning due to the highly structured, directive nature of clinical assessment. The preschool version of the BRIEF includes three broad indexes—inhibitory self-control, flexibility, and emergent metacognition—and a global composite, as well as two validity scales to identify excessive negativity or inconsistency in responding. The other versions have two broad indexes—metacognition and behavioral regulation—and a global composite, as well as the two validity scales. Table 3.8 provides a complete listing.

FAMILY ENVIRONMENT

The instruments noted here are part of the ever-growing recognition of the pivotal importance of family functioning in the face of a child's disability and adjustment. The most dramatic impetus has been the requirement of a family service plan in all early-intervention services for children up to 3 years of age. Beyond the case to be made in the youngest age group, many studies show a strong relationship between family functioning and a child's psychological adjustment across a number of different medical conditions (1). Further, the maximization of skills from treatment to the community requires the dedication of family members to be involved in the recovery process. The importance of such considerations is clear. The following are synopses of two widely used instruments for populations often within the scope of a rehabilitation practice.

The Home Observation for Measurement of the Environment Sale (HOME) (124) is a checklist designed to assess the quality of a child's home environment. It is an involved process including observation of the home setting and an interview with parents. Six areas are assessed: responsiveness of parent, parental acceptance of child, organization of physical environment, provision of appropriate play materials, parental involvement with child, and opportunities for variety in stimulation. In young children, the home setting is a strong predictor of later functioning.

The Family Environment Scale (FES) (125) rates parental perception of the social climate of the family, and is rooted in family systems theory. It contains 90 true–false items that break down into 10 subscales: cohesion, expressiveness, conflict, independence, achievement orientation, intellectual-cultural orientation, active-recreational orientation, moral-religious orientation, family organization, and family rules. Scores are plotted on a profile, with two forms available—the actual state of the family as perceived by individual members and the ideal state. Profiles derived from each parent can be compared, from which the family incongruence score is calculated.

There has been controversy about the psychometric properties of the FES relative to the stability of its factor structure. It was suggested that the factor structure varies, depending on which family member's perceptions were used. There is some caution expressed about its use as a clinical diagnostic tool in a rehabilitation setting with adults (126). Others have used it successfully in studies of children with chronic medical conditions. In one such study by Wallander and colleagues (127), family cohesion made a significant contribution to social functioning in children with spina bifida. A measure of family functioning specific to children with disabilities (Parents of Children with Disabilities Inventory [PCDI]) is presented in the following section on population-specific assessments. Table 3.9 provides a full listing of these tests.

TABLE 3.8 MEASURES OF GENERAL PSYCHOSOCIAL FUNCTIONING

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Functional Behavior Assessment (FBA)	A style of observation-based behavioral assessment geared toward identifying the underlying purpose of problem behavior.	Results are directly linked to interventions for behavior change. Can be successfully used with individuals with severe disabilities in any domain.
Transdisciplinary Play-Based Assessment (TPBA) (106)	Normed for 6 months to 6 years. Administered in home or clinic. Structured play observation.	Designed with intervention development as the primary goal. Taps a naturalistic activity; more engaging for young children.
The Bayley Scales of Infant and Toddler Development-III (82)	Ages 0–42 months. Provides normative framework for major social–emotional milestones.	Conormed with the cognitive measures on the Bayley Scales.
Minnesota Multiphasic Personality Inventory-Adolescent (MMPI-A) (107)	Objective self-report for adolescents aged 14–18. Revision of most widely used personality test for this age. Detailed assessment of response bias.	Excellent standardization and psychometric properties. Audiotape administration available. Likelihood of continued widespread use facilitates comparison across different groups. Length can be problematic in terms of engagement by subjects. Some subscales specifically measuring physical complaints must be interpreted carefully.
Personality Inventory for Children-2 (PIC-2) (111)	Two versions cover ages 3–16 years. Parent-report rating scale. Separate norms for mother and father as respondents. Assesses response bias.	Well normed for clinical population, but less research in rehabilitation population. Some concerns noted in use with brain injury.
Achenbach System of Empirically Based Assessment (123)	Includes parent report (CBCL), and teacher report (TRF), scales ranging from ages 1.5–18 years, and a self-report scale (YSR) for ages 11–18. Empirically driven and *DSM*-oriented scales provided.	Parent and teacher forms are widely used instruments in rehabilitation and nonrehabilitation populations. Does not assess response bias. Subscales measuring physical complaints must be interpreted carefully in a rehabilitation population.
Behavior Assessment System for Children-2 (BASC-2) (114)	Age: Parent and teacher scales range from 2–21 years. Self-report scales range from 8–25 years. Several scales measuring response bias.	Computer-scoring program provides easy comparison of information from multiple sources. Subscales measuring physical complaints must be interpreted carefully in a rehabilitation population.
Rorschach Inkblot Technique (117)	Projective personality test using inkblots as ambiguous stimuli. Standardized scoring norms provided for ages 5–16.	Psychometrically unsound. Concerns regarding impact of visual-perceptual impairments in rehabilitation population.
Children's Apperception Test (CAT) and Thematic Apperception Test (TAT) (116)	Projective personality test using ambiguous pictures. Some structured scoring.	Assesses themes of confusion and conflict, but requires careful interpretation. Absence of psychometric/normative data.

Abbreviation: *DSM, Diagnostic and Statistical Manual of Mental Disorders*.

POPULATION-SPECIFIC ASSESSMENTS

While most of the measures listed previously are designed for general use in the assessment of psychosocial functioning in children and adolescents, an increasing number of measures are being developed specifically for use with pediatric rehabilitation populations. Population-specific measures are more sensitive to the unique adjustment challenges that these youth face.

The PCDI (128) was designed to assess not only the frequency of disability-related stressors, but also parent perceptions of the stressors, which are an important factor in family adjustment. Four areas of concern are measured: medical and legal, concerns for the child, concerns for the family, and concerns for the self. Limited psychometric data is available, though initial estimates of reliability and concurrent and construct validity appear adequate. Further validation and normative studies are needed.

TABLE 3.9 MEASURES OF SINGLE DIMENSIONS

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Revised Children's Manifest Anxiety Scale for Children (RCMAS) (120)	Self-report of anxiety. Includes a lie scale to assess response bias.	Items assessing physiologic symptoms must be interpreted with caution in rehabilitation population.
Children's Depression Inventory (CDI) (121)	Self-report measure of depression. Five subscales: negative mood, interpersonal problems, ineffectiveness, anhedonia, and negative self-esteem.	Well-recognized scale. Some questions have been raised about the psychometric properties.
Behavior Rating Inventory of Executive Function (BRIEF) System (122)	Parent, teacher, and self-report rating scales. Measures behavior regulation and metacognition. Two response-bias scales included.	Allows for assessment of executive skills in naturalistic environment, which is important, as this can be hard to validly assess in clinical settings.

The Pediatric Inventory of Neurobehavioral Symptoms (PINS) (129) has the advantage of having been specifically designed for the assessment of personality, emotional, and behavioral issues associated with TBI. It has the disadvantage of having less research support, though there is some evidence of construct validity. It comprises 54 items, and can be completed by the parent or the teacher. Five general scales are obtained: mental inertia, social inappropriateness, dissociation of affect and behavior, episodic symptoms, and biologic symptoms.

The Pediatric Pain Questionnaire (PPQ) (130) is a structured interview completed with patients and parents. It measures both pain intensity and location, using body outline and visual analogue, as well as the emotional and perceptual experience. There are separate forms for children, adolescents, and parents. The adolescent form also covers the social and environmental influences on the experience.

History taking is an integral part of the process, including extensive history of treatments, child and family pain history, and environmental aspects. The analogue scale provides no numbers or markings, but instead elicits present and worst pain intensity of the past week. Different semantic anchors are used for children (not *hurting* versus *hurting a lot*), along with happy and sad faces. The adolescent and parent versions are anchored by *no pain* and *severe pain* and pain descriptors of hurting and discomfort. The body outlines are age-appropriate on the children and adolescent forms. The child can indicate four levels of pain intensity by coloring in the body outline with a choice of eight crayons. The child chooses colors to demonstrate the intensity gauged by four categories of pain descriptors. In this way, the child can show multiple sites and register the appropriate range of intensity in each. A separate list of pain descriptors is provided that assesses the evaluative, emotional, and sensory quality of the child's own experience. Words are provided for younger children or anyone who may have trouble generating labels.

The multidimensional aspect of the PPQ is appealing for anyone who has struggled to understand the experience of pain in children. It allows for engaging visual representations as well as standard language expression. Expecting parent reports to match the child's is erroneous. As in the adult literature, the subjectivity of the pain experience mitigates against this being the case. Comparison of child and parent reports is useful more as a gauge of convergence in the relationship between parent and child, not as a validating measure. Despite the unusual structure of some of its components, reliability and validity have been shown for the PPQ, and it holds considerable promise.

Measurement of health-related quality of life (HRQOL) represents an important component in the assessment of psychosocial functioning in pediatric populations. The PedsQL (131) is designed to measure HRQOL through brief child and/or parent ratings, with separate scales designed for different age groups within the 2- to 18-year range. Physical, emotional, social, and school functioning scales are included in the generic core scale, and supplemental condition-specific modules are available for asthma, rheumatology, diabetes, cancer, and cardiac conditions. Additional disease-specific measures of HRQOL are available for use with other populations such as epilepsy (132) and cystic fibrosis (133). A listing of population-specific measures is shown in Table 3.10.

The assessment of disease-related knowledge should not be overlooked. Most children with chronic illness or disability face the dual challenge of needing to cope with higher demands (as compared to normal populations) in terms of medical treatment regimens, using lower general coping resources due to primary symptoms and secondary deficits. Treatment adherence is of critical concern. Assessment of general developmental

TABLE 3.10 POPULATION-SPECIFIC MEASURES

INSTRUMENT (REF.)	DESCRIPTION	COMMENTS
Parents of Children with Disabilities Inventory (PCDI) (128)	Assesses frequency and perceptions of family stressors in the areas of medical/legal, concerns for child, concerns for family, concerns for self.	Limited psychometric data available. Assessment of perceptions of stressors is important, as this construct is related to adjustment.
Pediatric Inventory of Neurobehavioral Symptoms (PINS) (129)	Designed to assess sequelae associated with traumatic brain injury. Five domains assessed: mental inertia, social inappropriateness, dissociation of affect and behavior, episodic symptoms, and biologic symptoms.	Limited research on scale, though some construct-validity data is available.
Pediatric Pain Questionnaire (PPQ) (130)	Assesses pain intensity and location, as well as emotional and perceptual experience. Different scales for children, adolescents, and parents.	In-depth assessment of highly subjective experience.
PedsQL (131)	Measures health-related quality of life through child and parent ratings. Generic core scale measures physical, emotional, social, and school functioning. Condition-specific modules available for asthma, rheumatology, diabetes, cancer, and cardiac conditions.	Measures important aspect of functioning in pediatric populations. Disease-specific measures tap unique issues within separate illnesses.

maturity and psychosocial adjustment is a key indicator for addressing this issue. Informal assessment of patient understanding may help identify barriers to treatment adherence.

CONCLUSION

This chapter seeks to be a point of reference primarily to physicians working within rehabilitation settings, but it also has utility for other members of a pediatric rehabilitation team. It details the uses of psychological and neuropsychological assessment and hopefully acts as a primer of sorts on how to be a "good consumer" of such services. Since the first edition of this book an important development in the *Current Procedural Terminology* (CPT) codes has occurred. In 2002, the addition of the health and behavior assessment and intervention codes established diagnostic interview and interventions for psychosocial adjustment and psychoeducational purposes as a legitimate and billable activity. Prior to this, treatment or even a referral for evaluation would require assigning a psychiatric diagnosis for CPT coding, which was inappropriate and most times did not match what was actually happening with the patient. With this change, there is now a complete framework for the supportive and intervention role of mental health staff to assist patients and families in the significant coping challenges in the medical setting. This recognition removes all barriers to the inclusion of valuable psychosocial interventions to enable our patients and their families to have the services vital to the optimum outcome of the rehabilitation process.

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4

LANGUAGE DEVELOPMENT AND DISORDERS OF COMMUNICATION AND ORAL MOTOR FUNCTION

Lynn Driver, Rita Ayyangar, and Marie Van Tubbergen

Communication, as defined by the National Joint Committee for the Communicative Needs of Persons with Severe Disabilities (1), refers to "any act by which one person gives to or receives from another person information about that person's needs, desires, perceptions, knowledge, or affective states. Communication may be intentional or unintentional, may involve conventional or unconventional signals, may take linguistic or nonlinguistic forms, and may occur through spoken or other modes." Communication is clearly a dynamic process used to exchange ideas, relate experiences, and share desires.

Communication takes a variety of forms, including speaking, writing, gesturing, and sign language. As we know, interference with the physical ability to perform any of these acts has a significant impact on communication. Oral motor and neurologic impairments that affect communication may also significantly affect swallowing.

The purpose of this chapter is to provide a basic understanding of the acts of communication and swallowing, as well as an understanding of the primary disorders resulting from abnormal development or acquired injury of structures or systems related to these acts. The chapter is divided into two sections. The first section describes speech and language development and disorders through the exploration of four main topics:

- The primary anatomic systems of speech along with brief examples of deficits that result from disruption in these systems
- The primary components of speech and language, including milestones for acquisition
- Common speech and language disorders, both developmental and acquired, as well as common associated disorders
- A brief description of speech and language assessment and intervention

The second section describes feeding and swallowing processes and disorders and includes the following:

- Development of feeding skills, including expected milestones
- Anatomy and physiology of the swallowing mechanism
- Common disorders of deglutition, both congenital and acquired
- Feeding and swallowing assessment and intervention

SPEECH AND LANGUAGE DEVELOPMENT AND DISORDERS

Within the field of communication sciences and disorders, we think of communication as broadly comprising speech and language. Speech generally refers to aspects of communication that involve motor output for production of speech sounds. Production of speech sounds requires functional input from respiratory, phonatory, and articulatory systems (Table 4.1). Language generally refers to the process by which we both encode and process meaning within messages, and is divided into three primary components: form, content, and use. These components can be further subdivided based on five key aspects of language—specifically, phonology, morphology, syntax, semantics, and pragmatics.

ANATOMY OF SPEECH SYSTEMS

As noted previously, production of speech requires input from *respiratory, phonatory,* and *articulatory* systems. An airstream is generated by the lungs, passes through the vocal cords, and is then shaped by the articulators to form speech sounds. Impairments in any of these systems most likely will have a significant impact on speech production.

TABLE 4.1 COMPONENTS OF SPEECH

RESPIRATORY	PHONATORY	ARTICULATORY
Upper Airway	*Larynx*	Lips
Nose	Vocal Cords	Tongue
Mouth	Cartilage	Palate
Pharynx	Muscle	
Nasopharynx	Mucous	
Oropharynx	Membrane	
Hypopharynx	Ligaments	
Lower Airway		
Trachea		
Lungs		

Respiration

The *respiratory* system is composed of the upper and the lower airways. The upper airway consists of the nose, mouth, pharynx, and larynx, and the lower airway consists of the tracheobronchial tree and the lungs (2).

UPPER AIRWAY. The upper airway has many functions. The mucous membranes covering much of the upper airway structures are softer, looser, and more fragile in infants and young children than in older children and adults, and more susceptible to edema and injury from trauma.

NOSE. All children are obligate nasal breathers during the first 6 months of life, during which time the soft palate is in close anatomic approximation with the epiglottis. This factor, combined with the relatively large size of the tongue relative to the oral cavity at this age, renders nasal patency essential for maintaining an airway. Those children with nasal obstructions from conditions such as choanal atresia are at risk for respiratory compromise (cyanosis) during feeding.

MOUTH. The lips, mandible, maxilla, cheeks, teeth, tongue, and palate are the most important components of the oral cavity with regard to manipulation of airflow for respiration and speech production. The infant tongue takes up a larger area in the mouth and rests more anteriorly in the oral cavity than that of the adult. There are numerous congenital craniofacial anomalies, often associated with syndromes, that have an adverse impact on airflow. Some anomalies, such as cleft palate, prevent sufficient valving of the airstream, resulting in inaccurate production of speech sounds. Other anomalies, such as glossoptosis (oropharyngeal or hypopharyngeal obstruction during feeding caused by tongue retraction; common in disorders such as Pierre Robin sequence), can result in blockage of the airstream and subsequent respiratory distress.

PHARYNX. The pharynx, a muscular tube shared by the respiratory and digestive tracts, is sometimes referred to as the aerodigestive tract, and serves vital functions for both respiration and swallowing. It is divided into three portions: the nasopharynx, oropharynx, and the hypopharynx. The pharynx in an infant is gently curved, and as the child grows and develops, the angle increases to approximately 90 degrees.

The *nasopharynx* is the portion of the pharynx directly behind the nasal cavity, extending from the roof of the nasal cavity to the roof of the mouth. In addition to conducting air, the nasopharynx acts as a resonator for voice. The eustachian tubes from the middle ear open into the nasopharynx.

The *oropharynx* is that portion of the pharynx directly behind the oral cavity, extending from the roof of the mouth (pharyngeal aspect of the soft palate) down to the base of the tongue, at the level of the tip of the epiglottis. Movement of the pharyngeal walls in this portion, together with elevation of the soft palate and the posterior portion of the tongue, is crucial for velopharyngeal closure. Inadequate closure, or velopharyngeal incompetence, can result in disordered speech production.

The *hypopharynx* extends from the base of the tongue at the level of the hyoid bone and tip of epiglottis down to the entrance of the larynx and esophagus.

Lower Airway

The lower airway consists of the tracheobronchial tree and the lungs. The tracheobronchial tree consists of a system of connecting tubes that conduct airflow in and out of the lungs and allow for gas exchange.

TRACHEA. The trachea is situated anterior to the esophagus, beginning at the cricoid cartilage and extending inferiorly to the carina, where it bifurcates into the right and left main-stem bronchi. It is composed of C-shaped cartilage rings joined by connective tissue. These cartilage rings assist in keeping the trachea open during breathing. As noted previously, the mucous membranes of the trachea are softer, looser, and more fragile than those of the adult and more susceptible to damage, increasing the risk of obstruction from edema or inflammation.

LUNGS. The lungs are situated in the thoracic cavity, enclosed by the rib cage and diaphragm, the major muscle of ventilation, which separates the thoracic cavity from the abdominal cavity. The diaphragm in an infant is flatter than that of an adult, resulting in less efficient functioning for respiration. The air passages in infants and small children are much smaller, increasing their

susceptibility to obstruction. The respiratory bronchioles, alveolar ducts, and alveoli grow in number until about 8 years of age, after which they continue to grow in size. Impairments in lung function can occur as a result of birth-related conditions such as bronchopulmonary dysplasia (BPD) and diaphragmatic hernia, or due to acquired disorders such as spinal cord injury (SCI). These impairments often require tracheostomy and/or mechanical ventilation, which in turn have an impact on speech production.

CONTRIBUTION OF RESPIRATORY DYSFUNCTION TO SPEECH DISORDERS

Speech disorders related to respiratory dysfunction are often secondary to the presence of tracheostomy and/or ventilator dependence. The primary diagnoses of children requiring chronic tracheostomy and/or ventilator dependence include conditions due to trauma such as brain injury, SCI, and direct injury to the trachea; congenital conditions; progressive neurologic disorders; and acquired nontraumatic conditions such as Guillain–Barré syndrome and anoxic encephalopathy (3). It is important to note that the causes of respiratory failure and subsequent need for mechanical ventilation are not always respiratory disease or disorder. The lungs themselves may be healthy, but access to them or the systems that contribute to their function may be impaired.

A primary means of airway management in the presence of chronic respiratory insufficiency is a tracheostomy. A tracheostomy is an artificial opening created between the outer surface of the neck and the trachea between the second and third tracheal rings. The opening itself is referred to as the stoma, and the tracheostomy tube inserted into the trachea through the stoma serves to maintain the opening, as well as provide means for connecting mechanical ventilatory devices. Tracheostomy provides a secure airway, long-term airway access, and a means for interface with mechanical ventilatory devices, and as such, is the most frequently used method of airway management. Placement of the tracheostomy tube diverts airflow away from the trachea through the tube and out the neck, bypassing the upper airway, including the vocal cords. Depending on the size and type of tracheostomy tube, a portion of the airflow will still pass around the tube and through the vocal cords; this may or may not be sufficient to produce sound. In the event that it is not sufficient, options to facilitate sound include downsizing of the tracheostomy tube to a smaller diameter and use of a unidirectional flow valve such as the Passy–Muir valve (4), which directs greater airflow through the upper airway and out the nose and mouth. Table 4.2 shows a decision flow chart for manipulating factors related to the tracheostomy tube in order to facilitate voicing.

TABLE 4.2 TRACHEOSTOMY TUBE DECISION FLOW CHART

TTS—CUFF ↓ → ⇓	LW PRESS AIRCUFF↓ → ⇓	LW PRESS AIRCUFF↑ → ⇓	TALKING TRACH →	AAC
CUFFLESS	CUFF ↓	PRTCUFF ↓ ADJ VENT →		
SAME SZ → ⇓	ADJ VENT ⇓	⇓		
PHON W/LEAK → ⇓	PHON W/LEAK → ⇓	PHON W/LEAK → ⇓	ENT- VF EXAM	
		CUFF↓DAY		
UFV IN LINE	UFV IN LINE	CUFF↑NT ⇓		
		UFV IN LINE		

Note: A double arrow pointing down indicates progression if successful with that step; arrow pointing to the right indicates progression if that step was not successful. Single arrow pointing down indicates deflate/decrease pressure. Arrow pointing up indicates inflate/increase pressure.

Abbreviations: AAC, augmentative and alternative communication; ADJ, adjust; ENT, otolaryngologist; LW PRESS, low pressure; PHON, phonation; PRT cuff, partial cuff; Same SZ, same size; TTS, tight-to-shaft cuff; UFV, unidirectional flow valve; VF, vocal folds.

Source: Adapted from Ref. (5). Driver LE, Nelson VS, Warschausky, SA, eds. *The Ventilator Assisted Child: A Practical Resource Guide.* San Antonio, TX: Communication Skill Builders; 1997:73.

Phonation

The *phonatory* system comprises the larynx, and provides the sound source for speech. When this sound source is disrupted, it may result in alterations in voice quality, thus affecting communication.

The larynx is made up of cartilage, ligaments, muscles, and the mucous membrane. It protects the entrance to the lower airway and houses the vocal cords (Figure 4.1) (6).

Sound is *generated* in the larynx, and that is where pitch and volume are manipulated. The strength of expiration of air from the lungs also contributes to loudness, and is necessary for the vocal folds to produce speech (Figure 4.2) (6).

Most of the muscles of the larynx receive their innervation via the recurrent laryngeal branch of the vagus nerve. This branch descends downward and wraps around the aorta, and for this reason, children who undergo cardiac surgery can sometimes experience voice disorders. If the recurrent laryngeal nerve is stretched or damaged during surgery, innervation to the vocal cords can be disrupted, and vocal hoarseness can occur.

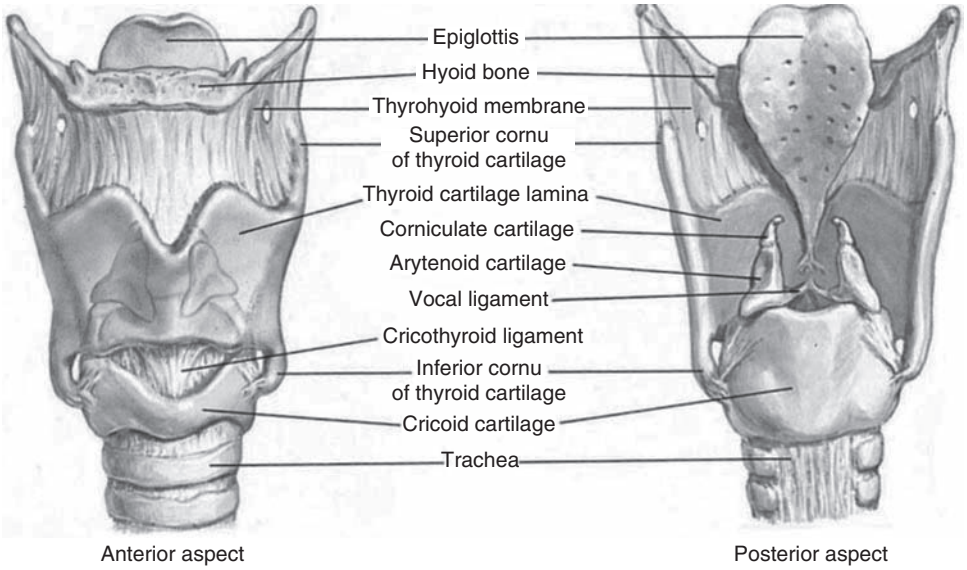


FIGURE 4.1 The larynx.

CONTRIBUTION OF PHONATORY DYSFUNCTION TO SPEECH DISORDERS

Speech disorders related to *phonatory* dysfunction are generally classified as voice disorders, and include dysphonia (abnormal voice quality) and aphonia (loss of voice). Dysphonia is an impairment of voice secondary to cranial nerve involvement, laryngeal pathology, or tracheostomy, and is characterized by varying degrees of breathiness, harshness, and vocal strain. Dysphonia may be a prominent feature of dysarthria related to cranial nerve involvement. Laryngeal pathologies resulting in dysphonia may include polyps, granulomas, nodules, or other lesions affecting the vocal fold mucosa. A common vocal fold trauma resulting in dysphonia is traumatic intubation following serious injury requiring assisted ventilation.

Articulation/Resonance

The *articulatory/resonatory* system is composed of the structures of the oral and nasal cavities, which modulate the airstream into the acoustic waveforms perceived as speech. Articulators responsible for production of speech sounds include the lips, tongue, and palate.

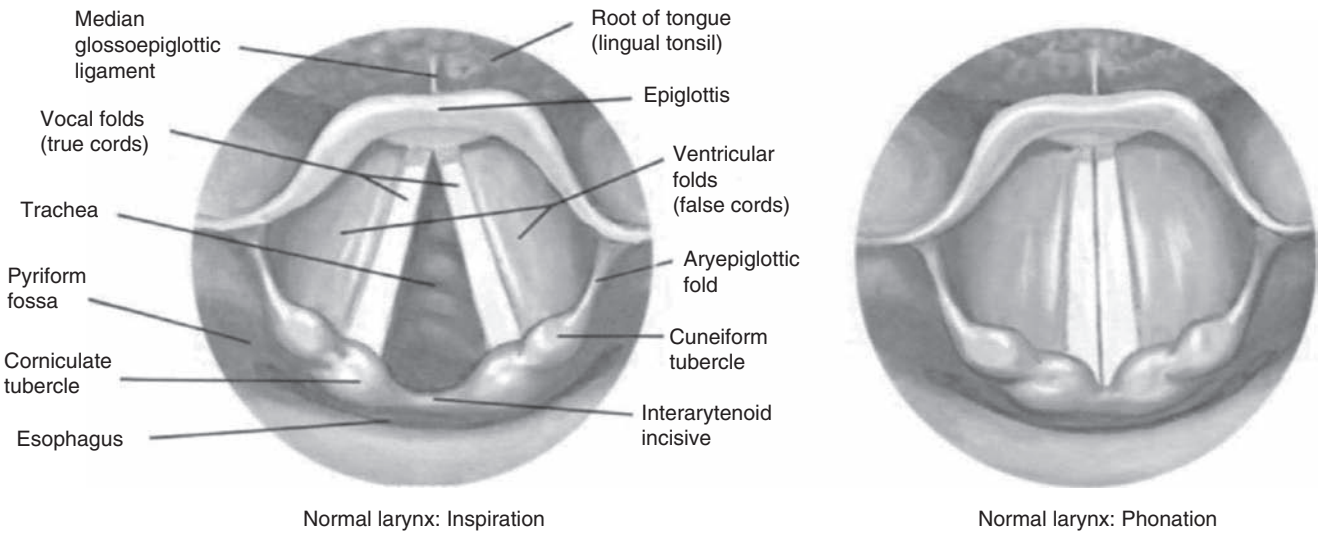


FIGURE 4.2 The vocal cords.

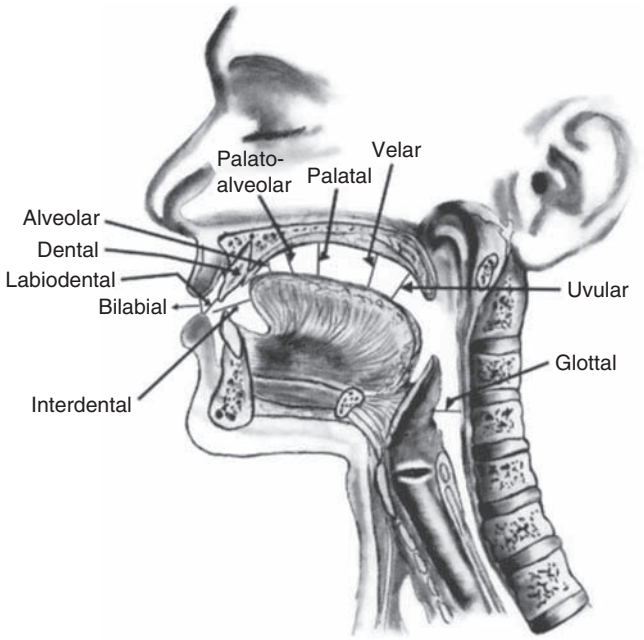


FIGURE 4.3 Places of articulation.

In addition to the placement of articulators, successful production of accurate speech sounds requires adequate functioning of the oral and nasal cavities as resonating chambers (resonance). Modulation of the airstream by these structures is a complex process that relies on intact structures as well as precise neuromuscular coordination. Figure 4.3 illustrates where various articulators are located, as well as places of articulation for various speech sounds. Impairment in one or more of these components is likely to result in a disorder of articulation/resonance.

CONTRIBUTION OF ARTICULATORY/RESONATORY DYSFUNCTION TO SPEECH DISORDERS

Speech disorders related to *articulatory/resonatory* dysfunction include disorders that result from impairment in any component of the *articulatory/resonatory* system, and as such are quite comprehensive. They include all motor speech disorders, including dysarthria and apraxia, as well as disorders resulting from congenital conditions such as cleft palate.

Although the three components of speech described previously are considered separately as individual components, they function as a single coordinated and interactive unit for production of speech, and as such, are subsystems of a complex motor act requiring precise coordination of muscle groups. It is easy to understand how impairments in any of these components can have an impact on communication, as the extent and complexity of the speech system make it susceptible to the influence of a myriad of factors.

MOTOR SPEECH DISORDERS

Motor speech disorders are a collection of communication disorders involving retrieval and activation of motor plans for speech, or the execution of movements for speech production (7). Subcategories include dysarthria and apraxia of speech. Motor speech disorders occur in both children and adults and may be acquired or developmental in nature (Table 4.3).

Dysarthria refers to a group of related motor speech disorders resulting from impaired muscular control of the speech mechanism, and manifested as disrupted or distorted oral communication due to paralysis, weakness, abnormal tone, or incoordination of the muscles used in speech (Table 4.4) (8). It affects the following:

- Respiration: respiratory support for speech, breathing/speaking synchrony, sustained phonation
- Phonation/Voice: loudness, quality
- Articulation: precision of consonants and vowels
- Resonance: degree of airflow through nasal cavity
- Prosody: melody of speech, use of stress and inflection

Movements may be impaired in force, timing, endurance, direction, and range of motion. Sites of lesion include bilateral cortices, cranial nerves, spinal nerves, basal ganglia, and cerebellum.

Associated characteristics of dysarthria include slurred speech; imprecise articulatory contacts; weak respiratory support and low volume; incoordination of the respiratory stream; hypernasality; harsh or strained/strangled vocal quality; weak, hypophonic, breathy vocal quality; involuntary movements of the oral facial muscles; spasticity or flaccidity of the oral facial muscles; and hypokinetic speech.

Some common etiologies for dysarthria in children include stroke, brain tumor, aneurysm, traumatic brain injury (TBI), encephalopathy, seizure disorder, cerebral palsy (CP), and high-level SCI.

Apraxia refers to a group of related motor speech disorders resulting from the impaired ability to produce movement in musculature in the absence of muscle weakness.

TABLE 4.3 MOTOR SPEECH DISORDERS

DEVELOPMENTAL	ACQUIRED
Developmental: no specific identifiable etiology to explain delays in speech acquisition	*Acquired*: an adverse event (usually neurologic) occurs that impedes continuation of previously normal speech acquisition
Phonological disorder	Dysarthria
Verbal apraxia	Verbal apraxia
Articulation disorder	Articulation disorder

TABLE 4.4 TYPES OF DYSARTHRIA

	SPASTIC	HYPOKINETIC	HYPERKINETIC	ATAXIC	FLACCID	MIXED
Site of lesion	Bilateral upper motor neuron	Extrapyramidal system	Extrapyramidal system	Cerebellum	Unilateral or bilateral lower motor neuron	Multiple sites of lesion
Associated characteristics	Spasticity of orofacial muscles Imprecise articulatory contacts Strained/ strangled voice quality Monopitch Reduced stress Reduced rate	Rigidity of orofacial muscles Imprecise articulatory contacts Hypophonia Monopitch Reduced stress and inflection Transient increased rate/rapid rate	Involuntary movements of orofacial muscles Imprecise articulatory contacts Harsh voice quality Incoordination of the respiratory stream Transient increased rate	Irregular articulatory breakdown Harsh vocal quality Incoordination of the respiratory stream Excess and equal stress pattern Reduced rate	Flaccidity of the orofacial muscles Imprecise articulatory contacts Breathy voice quality Low vocal volume Reduced stress and inflection Hypernasality	Characteristics dependent on site of lesion
Example of disorder	Cerebral palsy	Parkinson's disease	Dystonia	Friedreich's ataxia	Bulbar palsy	Amyotrophic lateral sclerosis

Source: Adapted from Ref. (8). Driver LE, Kurcz KB. Speech, language, and swallowing concerns. In: Brammer CM, Spires MC, eds. *Manual of Physical Medicine and Rehabilitation*. Philadelphia, PA: Hanley and Belfus, Inc.;2002:319.

Oral apraxia refers to an impairment of the voluntary ability to produce movements of the facial, labial, mandibular, lingual, palatal, pharyngeal, or laryngeal musculature in the absence of muscle weakness.

Verbal apraxia (also called apraxia of speech, or AOS) refers to an impairment of *motor speech* characterized by a diminished ability to program the positioning and sequencing of movements of the speech musculature for volitional production of speech sounds. Verbal apraxia may result in perceptual disturbances of breathing/ speaking synchrony, articulation, and prosody. Verbal apraxia is an acquired diagnosis related to an injury or illness. The site of lesion is generally the left precentral motor or insular areas.

Developmental verbal apraxia (also called developmental apraxia of speech, or DAOS) refers to a speech disorder resulting from delays or deviances in those processes involved in planning and programming movement sequences for speech. Associated characteristics of DAOS include receptive-better-than-expressive language, presence of oral apraxia (may or may not exist with DAOS), phonemic errors (often sound omissions), difficulty achieving initial articulatory configuration, increase in errors with increase in word length and/or phonetic complexity, connected speech poorer than word production, inconsistent error patterns, groping and/or trial-and-error behavior, and presence of vowel errors. DAOS is not related to a specific identifiable lesion and is defined primarily by speech symptoms.

Children with motor speech disorders may demonstrate impaired phonological systems because their ability to acquire the sound system of their language is believed to be undermined by difficulties in managing the intense motor demands of connected speech (9).

COMPONENTS OF SPEECH AND LANGUAGE

With regard to models of language, the prevailing school of thought follows Bloom and Lahey's philosophy, which proposes three main components of language: form, content, and use (Figure 4.4). According to Bloom and Lahey, language can be defined as "a knowledge of a code for representing ideas about the world through a conventional system of arbitrary signals for communication (10)."

These three components can be subdivided further into *phonology, morphology, syntax, semantics,* and *pragmatics*, as described in the following sections.

Form

Form with reference to language refers to the rule-based structure humans employ to formulate language, ranging from phonemes to sentences, and comprises *phonology, morphology,* and *syntax*.

Phonology refers to the rule-governed system by which sounds, or phonemes, are combined to create meaningful units, or words. The English language contains 44 recognized phonemes, which are classified as

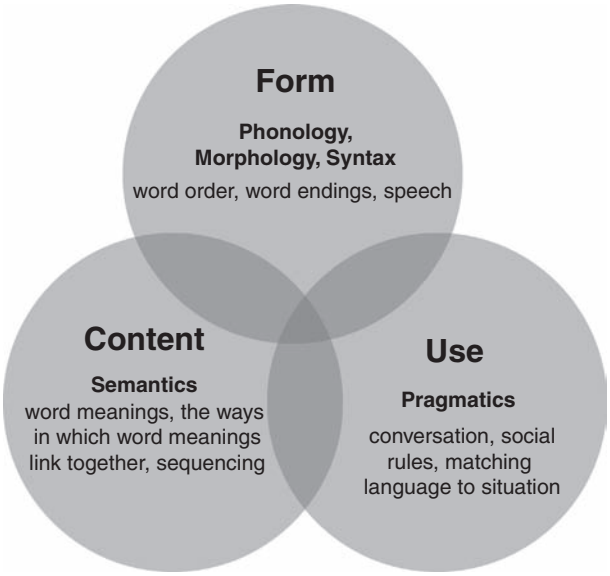


FIGURE 4.4 The three components of language.

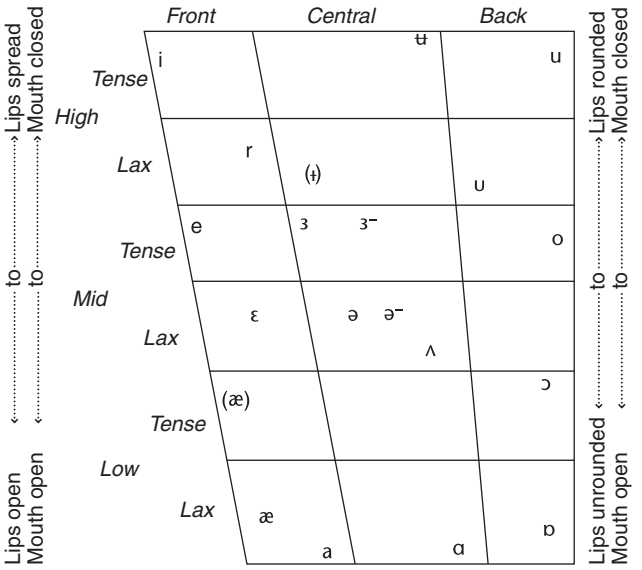


FIGURE 4.6 Tongue positions for vowel production. *Source*: Reprinted with permission from Ref. (12). Bronstein AJ. *The Pronunciation of American English*. New York, NY: Appleton-Century-Crofts, Inc., 1960.

consonants or vowels. This distinction involves the presence or absence of interruption of the airstream. Vowels are formed through modulation (without interruption) of the airstream via variation in position of the lips and tongue (Figure 4.5) (11).

Variations in tongue position for production of different vowels are systematically characterized as high, mid, or low, as well as front, central, or back, and can further be described as tense or lax (Figure 4.6) (12). For example, the vowel /i/, pronounced "ee," is considered a high, front, tense vowel, as the front of the tongue is high and the tongue is tensed. Diphthongs are combinations of vowels, and require movement of the tongue from one position to another during production.

Consonants are formed through a combination of varying degrees of interruption of the airstream and variations in tongue and lip posture (see Figure 4.3). Phonemic acquisition in children follows a systematic sequence, and it is believed that children acquire phonemes not in isolation, but rather in the context of their relationship to other sounds in a word (Table 4.5) (13).

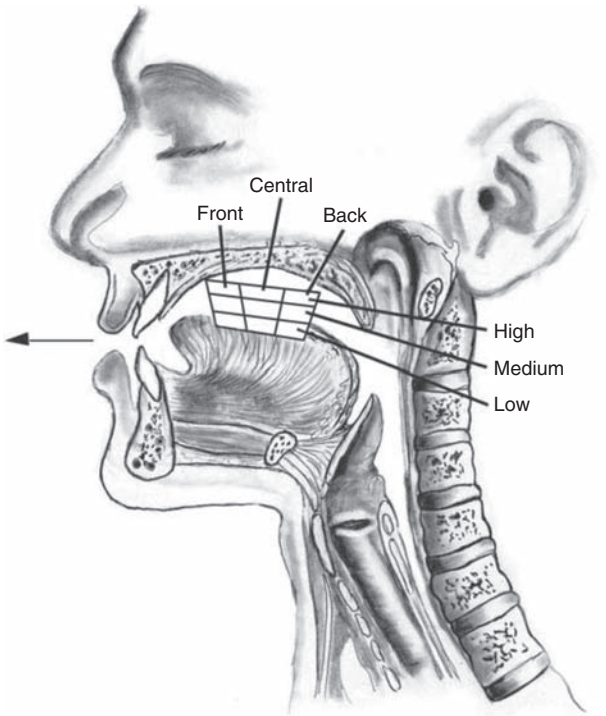


FIGURE 4.5 Vowel areas.

TABLE 4.5 PHONEMIC ACQUISITION: AGE AT WHICH 75% OF CHILDREN TESTED CORRECTLY ARTICULATED CONSONANT SOUNDS

AGE (YEARS)	SOUNDS
2	m, n, h, p, ŋ
2.4	f, j, k, d
2.8	w, b, t
3	g, s
3.4	r, l
3.8	š (she), tš (chin)
4	ð (father), Z (measure)
4+	dž (jar), θ (thin), v, z

Source: Adapted from Ref. (13). Soifer, LH. Development and disorders of communication. In: Molnar GE, ed. *Pediatric Rehabilitation.* 2nd ed. Baltimore, MA: Lippincott, Williams & Wilkins;1985.

TABLE 4.6 ACQUISITION OF CONSONANT SOUNDS

2	3	4	5	6	7	8
_____	p____					
_____	m____					
_____	h____					
_____	n____					
_____	w____					
_____	b____	____				
	____k	____				
	____g	____				
	____d	____				
	____t	____	____			
	____ng	____	____	____		
	____	f____		____		
	____	y____				
		____r	____	____		
		____l	____	____		
		____s	____	____	____	____
		____	ch____	____	____	
		____	sh____	____	____	
		____	z____	____	____	____
			____j	____	____	
			____v	____	____	____
			____	th____	____	
				(voiceless as in "**th**ink")		
				____TH	____	____
					____zh	____
				(voiced as in "**th**at")	(as in "trea**s**ure")	

Source: Adapted from Refs. (14) and (15).

Table 4.6 provides a graphic representation of the typical age ranges during which most children acquire consonant sounds (14,15). This is useful in determining at what age a child is considered outside of the norm for acquisition of a specific sound and when intervention might be indicated.

With regard to how well one can expect to understand a child's speech over the course of phonemic acquisition, Lynch et al. provide an estimate of speech intelligibility at different ages, summarized in Table 4.7 (16).

Phonological disorders are a subset of sound production disorders in which linguistic and cognitive factors, rather than motor planning or execution, are thought to be central to observed difficulties (common etiologic variables include otitis media with effusion, genetics, and psychosocial involvement) (17). Developmental phonological disorders result when children fail to progress in their acquisition of specific phonemes. The currently accepted theory regarding phonology in children proposes the existence of phonological processes that are present in the phonological systems of all children as they develop language,

TABLE 4.7 SPEECH INTELLIGIBILITY IN CHILDREN

By 18 months, a child's speech is normally 25% intelligible.
By 24 months, a child's speech is normally 50%–75% intelligible.
By 36 months, a child's speech is normally 75%–100% intelligible.

Source: Adapted from Ref. (16). Lynch JI, Brookshire BL, Fox DR. *A Parent-Child Cleft Palate Curriculum: Developing Speech and Language*. Oregon: CC Publications;1980:102.

and are systematically eliminated at predictable ages in a standard developmental progression. Failure to eliminate, or resolve, these processes, results in a phonological processing disorder. An example of a developmental phonological process is "stopping of fricatives," in which a child systematically substitutes a stop sound (a sound that stops airflow, such as /p, t, k/) for a fricative sound (a sound that produces friction through partial interruption of airflow, such as [th, s, z, f, v]), producing words such as "dum" for "thumb," "tun" for "sun," or "dip" for "zip." These sound substitutions are systematic and applied by the child in the same context each time that sound occurs. Nondevelopmental phonological processes are indicative of disordered versus delayed phonological development, and are rarely seen in normal development. An example of a nondevelopmental phonological process is initial consonant deletion, in which a child deletes the initial sound in a word, such as "ee"/"key" or "ake"/"make."

Table 4.8 illustrates the typical developmental sequence for resolving phonological processes (18).

TABLE 4.8 RESOLUTION OF PHONOLOGICAL PROCESSES: AGES BY WHICH PHONOLOGICAL PROCESSES ARE ELIMINATED

PHONOLOGICAL PROCESS	EXAMPLE	GONE BY APPROXIMATELY (YEARS; MONTHS)
Context sensitive voicing	pig = big	3; 0
Word-final devoicing	pig = pick	3; 0
Final consonant deletion	comb = coe	3; 3
Fronting	car = tar ship = sip	3; 6
Consonant harmony	mine = mime kittycat = tittytat	3; 9
Weak syllable deletion	elephant = efant potato = tato television = tevision banana = nana	4; 0
Cluster reduction	spoon = poon train = chain clean = keen	4; 0
Gliding of liquids	run = one leg = weg leg = yeg	5; 0
Stopping /f/	fish = tish	3; 0
Stopping /s/	soap = dope	3; 0
Stopping /v/	very = berry	3; 6
Stopping /z/	zoo = doo	3; 6
Stopping "sh"	shop = dop	4; 6
Stopping "j"	jump = dump	4; 6
Stopping "ch"	chair = tare	4; 6
Stopping voiceless "th"	thing = ting	5; 0
Stopping voiced "th"	them = dem	5; 0

Source: Adapted from Ref. (18). Bowen C. *Developmental Phonological Disorders: A Practical Guide for Families and Teachers*. Melbourne: ACER Press;1998.

Morphology refers to the rule-based system by which words are constructed and altered, often through addition of prefixes and suffixes, to reflect concepts such as number, possession, and verb tenses. For example, addition of the phoneme "-s" to the end of a word makes it plural. The "-s" in this instance is considered a morphological marker signifying the notion of "plural."

Disorders affecting morphology are most typically developmental and result when children have difficulty mastering the acquisition of rules for applying morphological markers. Difficulty with use of morphological markers can also be seen following certain types of focal brain injury, such as damage to Broca's area, when expressive language becomes telegraphic in nature, losing the nuances provided by morphological markers.

Syntax refers to the system of rules by which words are combined to create phrases, clauses, and sentences. The various parts of speech in English (eg, nouns, pronouns, verbs, adverbs, adjectives, etc.) serve different functions within these constructions, such as description, action, and attribute, and as such have specific rules for combination with each other. For example, the basic word order in English is subject–verb–object.

As with morphology, disorders affecting syntax are typically developmental and are the result of difficulty mastering the acquisition of rules for creating grammatically correct sentences.

Content

Content with reference to language refers to the *semantics*, or meaning, of words, as they relate to, or represent, objects, actions, and relationships. *Semantics*, or meaning, is conveyed through the use of words or other symbols within a given context. Development of semantics in children reflects growing and changing concepts related to experiences, culture, and cognitive level. An example of a changing semantic notion is that of overgeneralization. Children first learn the meaning of a word based on one representation of that word and initially overgeneralize it to apply to all similar representations. Hence, "dog" may at some point be applied to denote all four-legged creatures.

Child language disorders affecting semantics may be developmental and related to general cognitive development, or they may be acquired. Examples of disorders that involve semantics include specific language impairment (SLI), semantic–pragmatic language disorder, and Landau–Kleffner syndrome (19). In all these cases, children exhibit some degree of difficulty understanding the meaning of words and sentences. For children with semantic processing difficulties, the more abstract a concept is, the more difficult it is to understand. This holds true for things that require interpretation beyond the literal meaning, such as might be required in idioms or slang expressions. Deficits related to semantics can also result in difficulty identifying the key points in a sentence or story, which in turn may lead to problems with topic maintenance.

Use

Use with reference to language describes the function language serves within a social context, and is governed by pragmatics. Pragmatics refers to how we use the language we have acquired to communicate in social situations. Within a social interaction, language may be used in many different ways, such as to make comments, to ask questions, to acknowledge comments, and to answer questions. In 1976, Elizabeth Bates described three critical components of pragmatics: the ability to use speech acts to express intentionality in order to accomplish a given purpose (function), the ability to use social understanding and perspective-taking ability to make presuppositional judgments, and the ability to apply rules of discourse (eg, quantity, quality, relevance, clarity) in order to engage in cooperative conversational exchanges (20).

Child language disorders affecting pragmatics are most typically those associated with disorders on the autism spectrum. Recent changes presented in the *Diagnostic and Statistical Manual of Mental Disorders* (*DSM-5*) include a new diagnosis separate from autism spectrum disorder termed social communication disorder (SCD). This disorder is characterized by the presence of deficits in social communication that cannot be explained by cognitive impairment (21). Acquired injuries that may have an impact on pragmatics include TBI affecting the frontal lobes. Frontal lobe injury often impairs executive functioning and increases impulsivity, resulting in impaired judgment. This, in turn, may impair one's ability to understand perspective and to apply rules of discourse appropriately.

To summarize, language competence requires the successful intersection of form, content, and use. As simple as it may seem, having a successful conversation is a complex act requiring integration of many aspects of language and involving a blending of linguistic features with sociocultural understandings. "Conversation is not a chain of utterances, but rather a matrix of utterances and actions bound together by a web of understandings and reactions" (21).

SPEECH AND LANGUAGE ACQUISITION

Acquisition of speech and language skills follows a fairly systematic progression, with easily identifiable milestones associated with specific ages in each area, as briefly outlined here (22,23).

- *Birth to 3 months*
  - Makes pleasure sounds such as cooing
  - Develops differential cries for different needs
  - Develops social smile

- *3 to 6 months*
  - Increase in variety of vocalizations
  - Babbling sounds more speech-like, with increased consonant productions
  - Uses sounds and gestures to indicate wants
- *6 to 12 months*
  - Reduplicative babbling occurs (eg, dada, bibi, etc.)
  - Uses speech sounds to get attention
  - First words emerge (~10–12 months)
  - Responds to simple requests
  - Imitates speech sounds
- *12-18 months*
  - Imitates words overheard in conversation
  - Names familiar objects on request
  - Follows one-step commands during play
  - Finds a familiar object not in sight
- *18 to 24 months*
  - Uses words more frequently than jargon
  - Has expressive vocabulary of 50 to 100 words
  - Has receptive vocabulary of 300+ words
- *2 to 3 years*
  - Uses two- to three-word sentences
  - Points to pictures in books
  - Speech is understood by familiar listeners most of the time
- *3 to 4 years*
  - Uses simple sentences with negatives, imperatives, and questions
  - Talks about activities at school and home
  - Understands simple "wh-" question words
- *4 to 5 years*
  - Mean length of utterance (MLU) = 4.6 to 5.7 words
  - Uses grammatically correct sentences
  - Relays a long story accurately
- *5 to 6 years*
  - MLU = 6.6 words
  - Uses all pronouns consistently
  - Comprehends 13,000 words
- *6 to 7 years*
  - MLU = 7.3 words
  - Comprehends 20,000 to 26,000 words
  - Refines syntax

SPEECH AND LANGUAGE DISORDERS

Speech and language disorders in children can be conceptualized as falling into two categories: *developmental* and *acquired*. Within the category of *developmental*, we can also distinguish between developmental delay and developmental disorder. *Developmental language delay* refers to delay in the acquisition and development of age-appropriate language skills, typically across all domains. This can be due to medical or psychosocial factors. A *developmental language disorder* is characterized by atypical development of language skills in one or more domains, often with aberrant or interrupted development. As noted previously, there are specific milestones associated with each age as a child acquires speech and language skills. It is important to monitor development and watch for any signs that might indicate delay or disorder. The following is a list of danger signals of communication problems by age (24):

- *By 6 months*
  - Does not respond to the sound of others talking
  - Does not turn toward speaker out of view
  - Makes only crying sounds
  - Does not maintain eye contact with caregiver
- *By 12 months*
  - Does not babble
  - Does not discontinue activity when told "no"
  - Does not follow gestural commands, such as "want up" or "give me"
- *By 24 months*
  - Does not say a meaningful word
  - Does not refer to self by name
  - Does not follow simple directions
  - Does not talk at all at 2 years
  - Vocabulary does not seem to increase
  - Does not have any consonant sounds
  - Does not answer simple yes/no questions
- *By 36 months*
  - Does not say whole name
  - Does not seem to understand "what" and "where" questions
  - Uses jargon a great deal
  - Answers your question by repeating the question
  - Continues to echo statements made by others
  - Does not use two- to three-word utterances
  - Points to desired objects rather than naming them
  - Does not name any objects in pictures
  - Leaves off the beginning consonants of words
  - Cannot be understood even by parents
  - Does not respond when you call name

An *acquired language disorder* is characterized by language deficits in one or more domains secondary to neurologic insult. This can and often does result in aberrant development due to interruption in the normal course of language acquisition. When considering a speech and language disorder resulting from a congenital disorder such as cleft palate or Pierre Robin sequence, classification becomes more difficult. The disorder does not fit the definition of a developmental delay, in that the development is atypical secondary to structural deficits. The disorder is also not considered acquired, as the structural deficit leading to the disorder occurred at birth, before the child began to develop language.

Some common causes of loss or deterioration of language in childhood include head injury, unilateral cerebrovascular lesions, cerebral infections, brain tumors, seizure disorders, and cerebral anoxia. These disorders can result in acquired childhood aphasia (25), defined as a language disorder secondary to cerebral dysfunction in childhood appearing or occurring after a period of

normal language development. The cerebral dysfunction may be the result of a focal lesion of one of the cerebral hemispheres (eg, cerebrovascular accident (CVA)), a diffuse lesion of the central nervous system (CNS) above the level of the brainstem (eg, TBI, cerebral infection), a diffuse lesion related to convulsive activity, or lesion of unknown etiology (eg, Landau–Kleffner syndrome). In general, pediatric-acquired aphasia tends to be characterized by nonfluency, with primary deficits in verbal expression, with parallel deficits in written expression, and auditory comprehension relatively intact.

Pediatric TBI can result in more generalized dysfunction secondary to diffuse axonal injury caused by acceleration forces. Although such damage can have a significant impact on a variety of brain functions, the damage, sustained at the axonal or cellular level, is often not detected by brain scans. The definition of TBI, written by the Federal Division of Special Education as part of Public Law 101-476 (Individuals with Disabilities Act, or IDEA), was published in 1992 as the guideline for state departments of education to use in determining how to provide educational services to these children. It reads as follows (26):

> "Traumatic Brain Injury" means an acquired injury to the brain caused by an external force, resulting in total or partial functional disability or psychosocial impairment, or both, that adversely affects a child's educational performance. The term applies to open or closed head injuries resulting in impairments in one or more areas, such as cognition; language; memory; attention; reasoning; abstract thinking; judgment; problem-solving; sensory, perceptual, and motor abilities; psychosocial behavior; physical functions; information processing; and speech. The term does not apply to brain injuries that are congenital or degenerative, or brain injuries induced by birth trauma (Federal Register, Vol. 57, no. 189).

Other common acquired disorders that can affect speech and language development include high-level SCI and hearing loss. High-level SCI often affects some of the cranial nerves that are responsible for movement of the articulators necessary for speech production (Table 4.9) (27).

With regard to hearing loss, if children acquire hearing loss during the period of speech and language acquisition, they are at significantly increased risk for communication disorders.

There are many congenital disorders that can have an impact on speech and language development. Some of the most common include CP, cleft palate/craniofacial anomalies, hearing loss, and autism.

Cerebral palsy (CP) is defined as a group of disorders of development of movement and posture, causing activity limitation, that are attributed to nonprogressive disturbances that occurred in the developing fetal or infant brain. CP is the most common diagnosis associated with childhood disability, affecting 2% to 3% of the population. The motor disorders of CP are often accompanied by disturbances of sensation, cognition, communication, perception, and/or behavior, and/or by a seizure disorder (28). CP may significantly affect tone, which in turn affects ability to use those muscles appropriately to perform the necessary movements for speech production. As noted previously, speech production is a complex motor act requiring precise coordination of muscle groups, including respiratory, phonatory, and articulatory systems. When abnormal tone is present, either hyper- or hypotonicity, this interferes with coordination both within and across these systems, resulting in motor speech dysfunction, specifically dysarthria. The most common types of dysarthria associated with CP include spastic, ataxic, and hyperkinetic (see Table 4.4).

Children with spastic CP are more likely to exhibit imprecise articulatory contacts, strained/strangled voice quality, and reduced rate. Children with ataxic CP typically exhibit irregular articulatory breakdown, harsh vocal quality, incoordination of the respiratory stream, and reduced rate. Children with athetoid CP exhibit imprecise articulatory contacts, harsh vocal quality, incoordination of the respiratory stream, and transient increased rate.

Treatments for hypertonicity, such as intrathecal baclofen, selective dorsal rhizotomy, and various oral medications, may have an influence on speech and communication. These treatments frequently result in improvements, but in some cases may worsen impairment (29,30). The authors of this chapter report clinical observations of improved breath support for voice production and improved articulation with intrathecal baclofen therapy.

The presence of combined motor and cognitive impairments makes assessment of communication difficult. Efforts toward a more standardized classification system along the lines of the Gross Motor Function Classification System (GMFCS) (31) and Manual Ability Classification System (MACS) (32) have developed over recent years. The Communication Functional Classification System (CFCS) is a tool used to classify the everyday

TABLE 4.9 CRANIAL NERVES INVOLVED IN SPEECH AND SWALLOWING

Trigeminal (V)	**Vagus (X)**
Face (sensory)	Larynx (sensory and motor)
Head (sensory	Hypopharynx
	Soft palate
Facial (VII)	Cricopharynx
Taste (anterior 2/3)	
Ear (sensory)	**Spinal Accessory (XI)**
Facial expression (motor)	Soft palate (motor)
	Tongue (motor)
Glossopharyngeal (IX)	Pharynx (motor)
Pharynx (motor)	
Oropharynx (sensory)	**Hypoglossal (XII)**
Posterior tongue (sensory, taste)	Tongue (motor)
	Hyoid (motor)
	Extrinsic larynx

Source: Adapted from Ref. (27). Dodds WJ. *The physiology of swallowing*. Dysphagia. 1989;3:171–178.

TABLE 4.10 EXPRESSIVE PRODUCTION RATING SCALE (ExPRS)

Child's communication: Mark the item that best describes your child's typical abilities:

___ Speaks in a generally age-appropriate way; minor limitations, if any.
___ Speaks with some difficulty; speech may be slow or somewhat difficult to understand by a new listener.
___ Speaks with significant difficulty; speech is slow or quite difficult to understand by a new listener.
___ Communicates independently with limitations; individual uses adapted techniques such as signing or an augmentative communication device.
___ Communication is severely limited even with the use of augmentative technology.

Source: Adapted from Ref. (35). Albright KJ, Van Tubbergen M, Omichinski D, et al. Measurement of the Perceived Utility of Cognitive Assessments: Initial Reliability and Validity Testing of a New Research Tool. Poster presentation at: 8th Annual Conference Rehabilitation Psychology; March 2006; Reno, NV.

communication of individuals with CP using five descriptive levels for everyday communication performance. It has been found to have excellent test–retest reliability and moderate to strong interrater reliability depending on whether raters were professionals or parents (33). There has been some concern that by establishing a single rating to incorporate both receptive and expressive communication abilities, the ratings may be confounded by overall cognitive abilities. Vos et al. (2014) found that expressive language impairments are more closely related to motor impairments while receptive language impairments are more closely related to cognitive impairments among children with CP (34). Van Tubbergen and Albright developed a five-level ordinal scale to classify levels of expressive language: the ExPRS (Expressive Production Rating Scale) (35). Like the GMFCS and MACS, the ExPRS provides a descriptive classification system for expressive communication, including the use of alternative or augmentative communication (Table 4.10). Further investigation on the reliability and validity of the ExPRS is needed to enhance its potential in transdisciplinary settings.

Cleft palate and other craniofacial anomalies involving the oral cavity most typically affect a child's articulation as well as resonance. A cleft palate prevents the ability to valve the airstream at the level of the palate, making it impossible to close off the nasal passage during speech. This results in hypernasal speech. A number of other syndromes, such as velocardiofacial syndrome (also known as DiGeorge syndrome), affect the ability of the soft palate to function properly, resulting in velopharyngeal incompetence, in turn resulting in impaired resonance (hypernasality).

Congenital hearing loss can have a significant impact on the development of speech and language, depending on the severity of the loss. Speech and language disorders resulting from hearing loss may affect multiple areas of communication, including language comprehension, syntax, vocabulary, and articulation. The nature and extent of communication disorders in children with hearing impairment are influenced by type and degree of hearing loss, causative factors, age at onset, cognitive status, and environment. Early identification and intervention are critical to maximize potential for developing communication skills in children with hearing loss. Intervention can include provision of hearing aids, environmental modifications (eg, frequency modulation (FM) systems in the classroom), aural habilitation/rehabilitation, sign language, total communication (combination of auditory–vocal language, signs, gesture, and speech reading), or surgical implant (cochlear implant).

Autism is one of the fastest-growing childhood disorders in our nation today. The current estimate according to the Centers for Disease Control and Prevention (CDC) Autism and Developmental Disabilities Monitoring (ADDM) Network is that 1 out of every 68 children is diagnosed with autism. This represents a 30% increase from 1 in 88 reported 2 years ago. The term autism, or autism spectrum disorder (ASD), refers to a group of complex disorders of brain development characterized by deficits in social interaction, verbal and nonverbal communication, and restricted, repetitive behaviors (www.autismspeaks.org/what-autism/diagnosis/dsm-5-diagnostic-criteria). Until publication of the *DSM-5* diagnostic manual in May, 2013, the spectrum of autism disorders included distinct subtypes including autistic disorder, pervasive developmental delay—not otherwise specified (PDD-NOS), Asperger's syndrome, Rett's disorder, and childhood disintegrative disorder. All of these distinct diagnoses have now been subsumed under the diagnosis of ASD, with levels of severity (mild, moderate, severe) based on the level of support required to achieve success in social communication. The new diagnostic criteria for ASD include persistent deficits in social communication and social interaction across multiple contexts, with severity based on social communication impairments and restricted repetitive patterns of behavior. Severity levels range from level 1: "requiring support," to level 3: "requiring very substantial support." A separate, related diagnosis of social communication disorder (SCD) also appears in the fifth edition of the *DSM* (36). The diagnostic criteria for SCD include persistent deficits in the social use of verbal and nonverbal communication resulting in functional limitations in effective communication, social participation, social relationships, academic achievement, or occupational performance. The primary feature that distinguishes ASD from SCD is the presence of restricted repetitive patterns of behavior, such as inflexibility, difficulty coping with change, or

TABLE 4.11 PRIMARY DISORDERS OF SPEECH, LANGUAGE, AND SWALLOWING

	DEVELOPMENTAL	ACQUIRED
Motor speech disorders	Phonological disorder Verbal apraxia Articulation disorder	Dysarthria Verbal apraxia Articulation disorder
Language disorders	Language delay Language disorder	Aphasia
Voice disorders	Aphonia Dysphonia	Aphonia Dysphonia
Fluency disorders	Nonfluency Dysfluency/stuttering	Dysfluency/stuttering
Communicative–cognitive disorders	Learning disabilities Autism	Traumatic brain injury Aphasia
Memory disorders		Short-term memory deficit Long-term memory deficit Verbal learning deficit
Swallowing disorders	Oral aversion Discoordination of suck–swallow–breathe	Oral dysphagia Pharyngeal dysphagia Oropharyngeal dysphagia

Source: Adapted from Ref. (37). Driver LE, Kurcz KB. Speech, language, and swallowing concerns. In: Brammer CM, Spires MC, eds. *Manual of Physical Medicine and Rehabilitation*. Philadelphia, PA: Hanley and Belfus, Inc.; 317.

other repetitive behaviors that interfere with functioning in a variety of contexts (www.autismspeaks.org/what-autism/diagnosis/dsm-5-diagnostic-criteria).

ASSESSMENT AND TREATMENT OF SPEECH/LANGUAGE DISORDERS

Speech–language pathologists provide diagnostic, treatment, and educational services to children who are experiencing impairments of speech, language, voice, fluency, communicative–cognitive, memory, and swallowing skills. The primary disorders are outlined in Table 4.11 (37), divided into developmental versus acquired.

Assessment

In assessing language disorders in children, the normal developmental level associated with the chronological age of the child is a crucial starting point to identify the impact of a neurologic event or other interruptions in typical maturation. Early identification of children at risk is also critical, as we know that speech and language delays/disorders in infancy and toddlerhood can result in difficulties in academic learning, social interaction, and development of appropriate peer relationships throughout childhood (37,38).

Areas of assessment in pediatric communication disorders include pragmatics, cognition, orientation, attachment/interaction, prelinguistic behaviors, phonological development/intelligibility, oral motor function, language comprehension (auditory and reading), language production (verbal and written), fluency, voice, hearing, and feeding and swallowing. These areas are assessed formally through test batteries, objective procedures, and parent interview questionnaires, as well as informally through direct observation of and interaction with children in naturalistic contexts. Detailed descriptions of specific assessment materials and procedures in each of these areas is beyond the scope of this chapter. It should be noted that assessment is often done as part of a multidisciplinary evaluation, and input from other disciplines is often vital in providing the most comprehensive diagnosis and treatment plan. One area of common need for multidisciplinary input is augmentative and alternative communication (AAC). For children who are nonverbal or who have significant motor impairment, a reliable means of access to augmentative communication devices and to computers must be identified, and this process may require input from speech pathology, occupational therapy, rehabilitation engineering, and sometimes physical therapy. The rapid evolution of personal technology has led to an explosion of widely available devices, applications, and programs that can be incorporated into a child's therapy and home/community participation. The appsforaac.net community website maintains a current, curated list of useful resources. Once a child has undergone a thorough evaluation, results are carefully reviewed, a diagnosis is made, and treatment recommendations are formulated. A child's parents or caregivers are included as much as possible in the assessment process, as well as in the development of the treatment program.

With regard to the diagnosis, it is important to have a clear understanding of a child's medical history and any contribution that medical status may have made to the child's communication disorder. This will determine whether the deficit is considered developmental or acquired, and the diagnosis will then drive the treatment recommendations, including specific goals and objectives, treatment time frame, and projected outcome (prognosis). A clear understanding of a child's cognitive level is also crucial in making appropriate diagnoses as well as treatment recommendations. If a child's cognitive level is commensurate with the level of language ability, expectations for improvement and prognosis are different than for a child exhibiting a significant discrepancy between language and cognitive skills.

Individuals with speech and other impairments require accessible and sensitive cognitive assessment tools and strategies. Typical standardized tests assessing various cognitive abilities specify the modality in which information is presented to the child and the modality in which the child must respond. Many procedures assume clear and fluent speech for participation. For example, most tests of phonological awareness require the participant to verbally present words or sounds to demonstrate skills. For an individual with significant apraxia, it is difficult to determine whether errors are due to underlying deficits in phonological awareness, effects of apraxia, or other reasons. For individuals who use alternative or augmentative communication, most communication devices require them to make selections from preprogrammed arrays. This presents a further confound in that the ability to make choices of preference may be more developed than the ability to answer a factual question on demand if there are impairments in pragmatics (39).

Given the dearth of accessible speech, language, and cognitive assessment tools for individuals with communication impairments, especially if there are concurrent motor impairments, efforts to develop such instruments are a priority to optimize educational and medical interventions, as well as to provide accurate and meaningful diagnoses (40,41).

In addition to developing treatment recommendations, it is important to make any other referrals as appropriate. For example, if a child's history includes language regression, a referral to pediatric neurology may be indicated. If a child with documented speech and language delay has not had a formal hearing assessment, a referral to audiology is warranted. If a child is exhibiting characteristics consistent with a disorder on the autism spectrum, a referral to pediatric specialty clinic may be necessary to obtain a formal diagnosis. Due to recent legislation, some insurance companies require a multidisciplinary assessment through an approved autism evaluation center (AAEC), with consensual team diagnosis including a pediatrician or another appropriate developmental or behavioral physician (eg, behavioral pediatrician, pediatric neurologist, pediatric geneticist, pediatric psychiatrist), a speech–language pathologist, and a psychologist. This consensual, multidisciplinary diagnosis is required to access funding for ASD treatment.

Treatment

Once a child has been evaluated, recommendations for treatment are made. These include specific goals and objectives in the identified deficit areas. Treatment for children with developmental speech and language delay or disorder differs in a number of important aspects from treatment for children with an acquired speech and language disorder. Again, we distinguish between developmental *delay* and *disorder* in that *delay* implies typical but slowed or late development of communication skills. *Disorder* implies aberrant development of communication skills. For example, most typically developing children overgeneralize certain semantic concepts in the course of acquiring expressive vocabulary. At some point, they may use the word *dog* to refer to all four-legged animals, or *juice* to refer to all drinks. For children with developmental delay, they would be expected to persist in these overgeneralizations beyond predicted ages. In contrast, children with developmental disorders may exhibit atypical language patterns, such as reversing word order or leaving out certain parts of speech (eg, verbs) completely in their development of expressive language. These errors are not part of the typical pattern of language acquisition, and thus would be considered a disorder.

Treatment for children with developmental speech and language delay will typically focus on general language stimulation within the specific areas of delay. For example, for a child with delay in expressive language, a general goal might be for him or her to use language successfully to get daily needs and wants met. Objectives within that goal might be to increase expressive vocabulary, increase utterance length, ask and answer questions, or improve speech intelligibility. Treatment for children with developmental disorders will need to be more tailored to the specific errors exhibited, which will not necessarily fall within the typical acquisition of speech/language milestones. Augmentative communication forms, including sign, Picture Exchange Communication System (PECS), and other devices may be used in the treatment of developmental disorders. According to a recent study reported in *Language, Speech, and Hearing Services in the Schools*, AAC has been found to have positive effects on the natural speech and language development of some children with severe communication impairments, specifically with the use of an integrated multimodal intervention (IMI) technique that applies a variety of speech production and augmentative communication techniques to practice natural speech production within meaningful social communication contexts (42). The study demonstrated that the integration of speech-generating AAC devices and traditional speech intervention techniques did not impede development of natural speech, but in fact supported natural speech production. Children with the diagnosis of autism would fall under the

category of developmental disorder, in that their language development does not follow the typical developmental progression. There are a number of treatment programs for children with autism, ranging from applied behavioral analysis (ABA) (41,43) to the "floortime" (DIR—the Developmental, Individual Difference, Relationship-based intervention program created by psychiatrists Stanley Greenspan, MD, and Serena Wieder, PhD) approach (44). The decision regarding which treatment approach to use in part is determined by the severity of the communication disorder; children with more severe disorders are often referred to ABA programs due to the increased amount of structure. Children with milder disorders may benefit more from a play-based approach such as DIR.

Treatment for children with acquired communication disorders can be somewhat more complex, as it requires a detailed understanding of the specific deficits as well as how they relate to the child's development of communication as a whole. In addition, it requires the ability to distinguish between gains due to spontaneous recovery from injury, gains due to typical expected development, and gains due to treatment. One of the most common areas of treatment in acquired communication disorders is TBI.

Janet Lees proposes three stages of recovery in pediatric brain injury: acute period, lasting from emergency admission to reestablishment of stable conscious state; consistent recovery, lasting from reestablishment of stable conscious state to the point where progress begins to slow down, or plateau; and the slowed recovery, or plateau stage (45). The period during which a child makes the greatest progress is the second stage, in which intensive therapy and educational input can maximize recovery. The period where long-term residual deficits become apparent occurs during the third stage. The length of each stage varies, depending on the severity of the head injury. When treating children with acquired TBI, it is important to keep in mind the unique characteristics and needs specific to pediatric brain injury. For example, pediatric brain injury occurs on a moving baseline of normal development upon which further development is expected. For this reason, assessment tools need to be appropriate for the developmental age of the child; in young children, this means some functions will not be accessible. Plasticity in the developing nervous system may allow the preservation of certain functions, particularly those related to language. In addition, plasticity could theoretically involve relocation of function to the opposite hemisphere or elsewhere in the same hemisphere. Normal recovery may occur, can be a most dramatic and unexplained phenomenon, and should not be confused with plasticity. Finally, critical periods for the development of a particular function may exist, which, at most, cannot be retrieved. This may, for example, apply to the development of social communication in young children at relatively high risk of the development of autistic features (45).

When it is not possible to promote or maintain verbal communication in children, regardless of whether they have a developmental or acquired disorder, it may be necessary to provide augmentative or alternative options for communication. Numerous options are available for nonverbal children, ranging from sign language to high-tech augmentative communication devices. Common low-tech solutions include signing, pictures (eg, PECS) (46), and recordable devices with finite selections, such as the Cheap Talk Device (product information available at www.enablingdevices.com). Children in need of augmentative or alternative communication typically are evaluated by speech pathology first, and if a more comprehensive assessment is indicated, a second evaluation may be done as part of a multidisciplinary assessment, including occupational therapy and rehabilitation engineering. Children who have significant motoric impairments often need input from occupational therapy regarding access solutions. Children who have complex needs requiring more custom solutions often benefit from input from rehabilitation engineering.

FEEDING AND SWALLOWING PROCESSES AND DISORDERS

During the first 12 months, infants have a number of unique anatomic and physiologic characteristics that gradually diminish with growth and maturation (Figure 4.7) (47). For example, the larynx in infants is positioned higher in the neck than in older children and adults, with close approximation of the epiglottis and soft palate, resulting in added airway protection, as well as obligate nasal breathing (Figure 4.7A,B) (47,48). This is important in promoting the suck–swallow–breathe sequence, the most complex sensorimotor process undertaken by the newborn infant. Structural or functional abnormalities in the upper airway of infants put them at greater risk for feeding difficulties. Other unique features of infants include sucking pads in the cheeks to provide additional stability during sucking and a significantly larger tongue with respect to the oral cavity, which restricts tongue movement to the anterior–posterior direction characteristic of suckling.

Infants also exhibit a number of unique physiologic aspects that are important for successful feeding and swallowing. These include reflexes that assist with the development of feeding, such as the suck–swallow reflex, the rooting reflex, and the phasic bite reflex. As cortical development advances, these automatic reflexes gradually evolve into more volitional actions, beginning during the period from 4 to 6 months of age. For example, at about 6 months of age, the transition from suckling to sucking begins to occur, with anatomic and neurologic maturation resulting in gradual lowering of the jaw, allowing more space for tongue movement, and gradual increase in volitional control permitting increased refinement and control of movements. The development of motor milestones in infants and toddlers is accompanied by attainment of feeding and swallowing skills, as outlined in Table 4.12 (47,49).

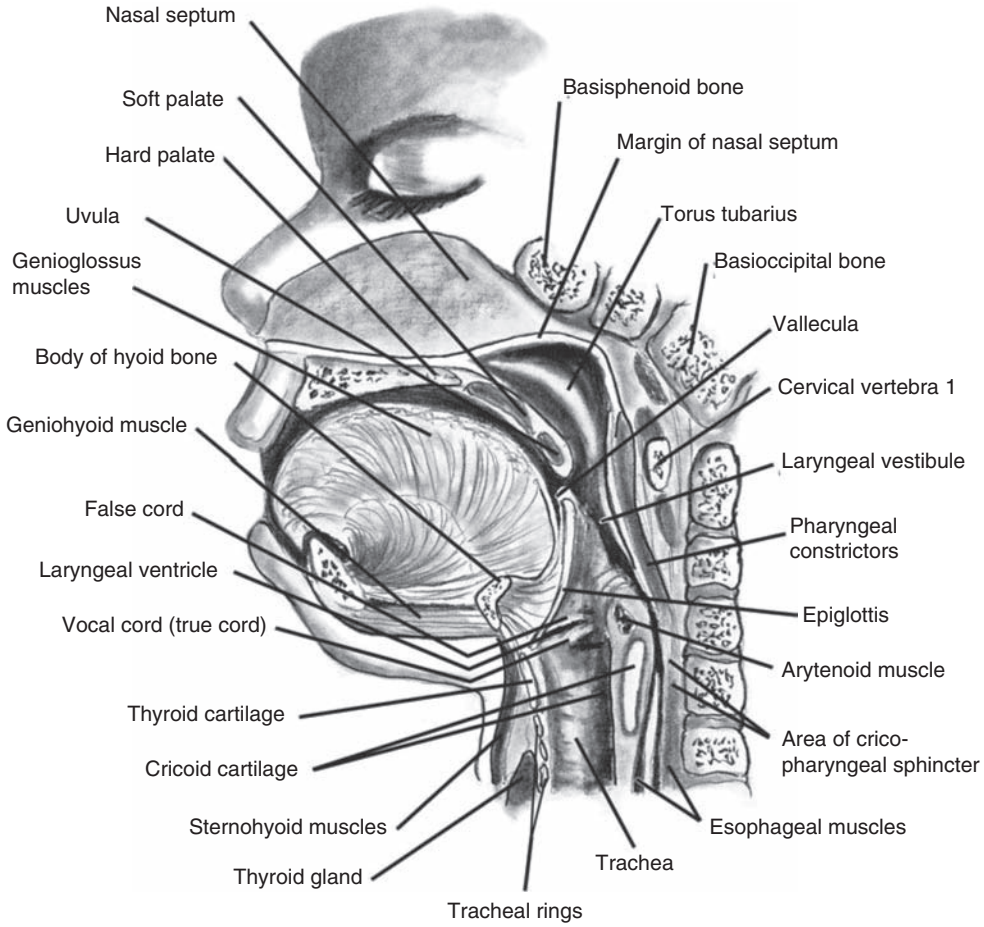


FIGURE 4.7A The pharynx: infant.

Critical periods are believed to exist in the development of normal feeding behavior. This can sometimes become problematic when caregivers are not sensitive to these critical stages. For example, caregivers may choose to maintain children on pureed foods due to apprehension regarding readiness to handle solid, chewable foods. However, research shows that delaying introduction of solid foods can result in food refusal and sometimes the development of food aversions (50). By the time children reach the age of 3 years, their ability to chew and swallow has matured and, with the exception of laryngeal position, their anatomy and physiology closely resemble those of the adult.

Infants with anatomic or physiologic abnormalities are at even greater risk for developing significant difficulty with establishing and maintaining oral feeding due to the inability to initiate oral feedings within age-appropriate time frames. It is crucial for clinicians to have a thorough understanding of normal anatomic and physiologic development for feeding and swallowing in order to understand the implications of disorders.

Feeding and swallowing abilities involve multiple, interrelated anatomic and physiologic components within the body (eg, oral motor, pharyngeal, esophageal, respiratory, gastrointestinal). For this reason, effective management of children with feeding and swallowing disorders typically requires input from many specialists. These specialists may work separately or ideally may work within an interdisciplinary feeding and swallowing team, providing the added benefit of coordinated care. An interdisciplinary approach is recommended at institutions where professionals evaluate and treat children with complex feeding and swallowing problems. Table 4.13 describes the members and functions of a comprehensive feeding and swallowing team.

Primary components of clinical assessment of pediatric feeding and swallowing skills include a thorough history, a prefeeding evaluation, and a feeding observation or trial feeding. If aspiration is suspected or risk of aspiration is a factor, instrumental assessments of swallowing, such as videofluoroscopic swallow study (VFSS) and fiberoptic endoscopic evaluation of swallowing (FEES) may also be necessary following the clinical evaluation.

Feeding and swallowing difficulties can occur within a broad range of disorders, including anatomic or structural defects, neurologic deficits, systemic conditions, or complex medical conditions. Congenital anatomic or structural defects commonly affecting swallowing include tracheoesophageal fistula (TEF), choanal atresia, and cleft palate. Acquired anatomic defects

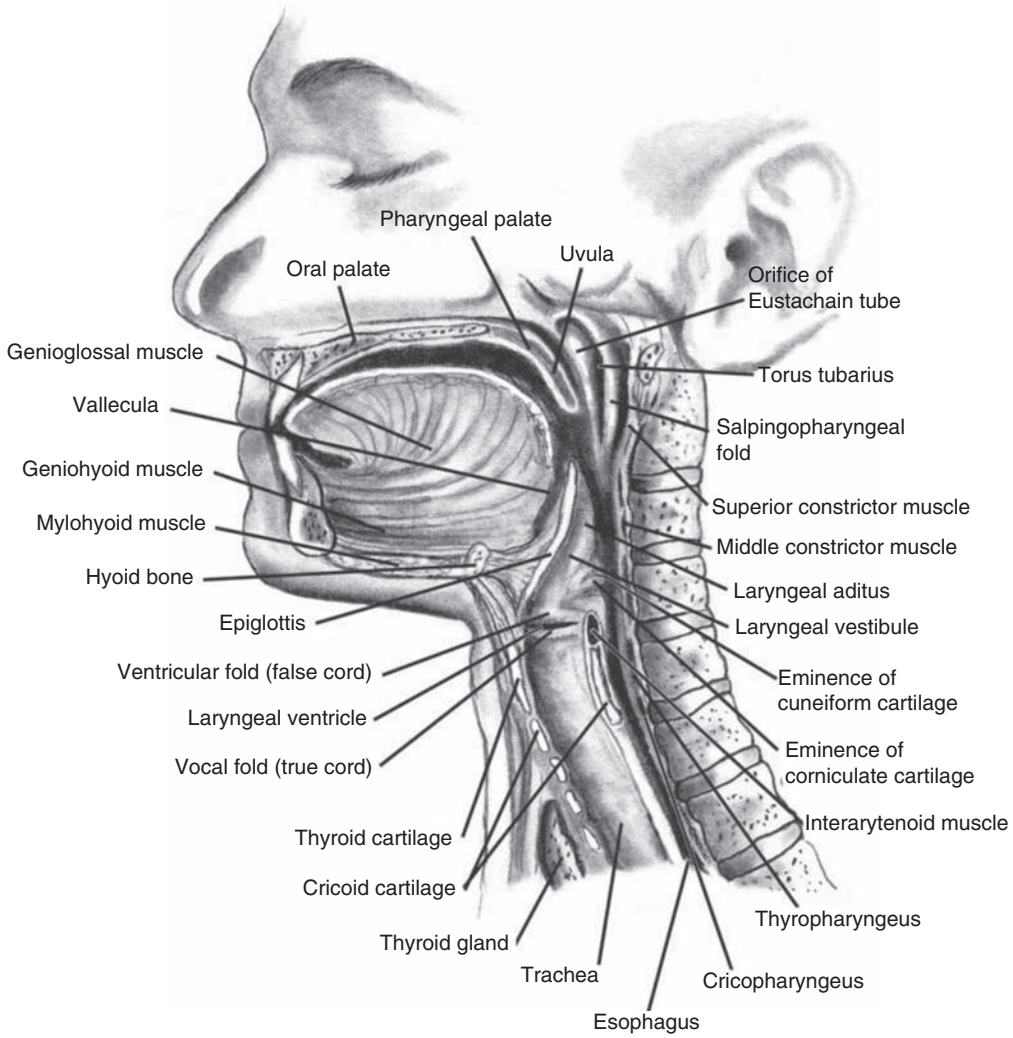


FIGURE 4.7B The pharynx: adult.

include laryngeal trauma. Neurologic deficits commonly affecting feeding and swallowing include CP, TBI, genetic syndromes, hypoxic/ischemic encephalopathy, meningitis, and Arnold–Chiari malformation. Systemic conditions typically associated with feeding and swallowing disorders include respiratory diseases such as BPD and reactive airway disease (RAD), and gastrointestinal disorders such as gastroesophageal reflux (GER). Complex medical conditions resulting in swallowing disorders include prematurity and cardiac abnormalities. Given the interrelated nature of systems contributing to swallowing function, abnormalities (congenital or acquired) in any one of these systems can result in a feeding or swallowing disorder. For example, premature infants or infants with cardiac abnormalities often have abnormally high respiratory rates. If respiratory rates are above 60 breaths per minute, successful feeding is often not possible because energy expended for breathing leaves no energy for feeding, resulting in breakdown in coordination and increased risk for aspiration (51). Infants and children with reflux are at increased risk for feeding difficulties, as reflux contributes to negative experiences associated with feeding (gastroesophageal pain/discomfort, aspiration), and subsequent feeding aversion may develop. Structural defects such as vocal fold paralysis, laryngeal cleft, tracheoesophageal fistula, glossoptosis, and choanal atresia can result in difficulty protecting the airway, leading to aspiration. Thus, obtaining a thorough medical history is crucial to understanding the etiology of a child's swallowing disorder.

In addition to medical history, a feeding history is important to obtain, as this will determine how to approach feeding assessment. If a child has been eating but his or her diet has been restricted to specific consistencies secondary to swallowing difficulties, this will be important to know. If a child has never been an oral eater, this is also critical information in subsequent clinical assessment decisions. Also, if a child has specific feeding utensils that he or she is accustomed to using, these should be used during the clinical assessment.

TABLE 4.12 ATTAINMENT OF FEEDING AND SWALLOWING MILESTONES

AGE (MONTHS)	DEVELOPMENT/POSTURE	FEEDING/ORAL SENSORIMOTOR
Birth to 4–6	Neck and trunk with balanced flexor and extensor tone Visual fixation and tracking Learning to control body against gravity Sitting with support near 6 months Rolling over Brings hands to mouth	Nipple feeding, breast, or bottle Hand on bottle during feeding (2–4 months) Maintains semiflexed posture during feeding Promotion of infant–parent interaction
6–9 (transition feeding)	Sitting independently for a short time Self-oral stimulation (mouthing hands and toys) Extended reach with pincer grasp Visual interest in small objects Object permanence Stranger anxiety Crawling on belly, creeping on all fours	Feeding more upright position Spoon feeding for thin, smooth puree Suckle pattern initially suckle—suck Both hands to hold bottle Finger feeding introduced Vertical munching of easily dissolvable solids Preference for parents to feed
9–12	Pulling to stand Cruising along furniture First steps by 12 months Assisting with spoon; some become independent Refining pincer grasp	Cup drinking Eats lumpy, mashed food Finger feeding for easily dissolvable solids Chewing includes rotary jaw action
12–18	Refining all gross and fine motor skills Walking independently Climbing stairs Running Grasping and releasing with precision	Self-feeding: grasps spoon with whole hand Holding cup with two hands Drinking with four to five consecutive swallows Holding and tipping bottle
18–24	Improving equilibrium with refinement of upper extremity coordination Increasing attention and persistence in play activities Parallel or imitative play Independence from parents Using tools	Swallowing with lip closure Self-feeding predominates Chewing broad range of food Up–down tongue movements precise
24–36	Refining skills Jumping in place Pedaling tricycle Using scissors	Circulatory jaw rotations Chewing with lips closed One-handed cup holding and open cup drinking with no spilling Using fingers to fill spoon Eating wide range of solid food Total self-feeding, using fork

Source: Adapted from Refs. (47) and (49).

In addition to indirect assessment through parent interview and thorough review of medical records, direct observation of the child prior to introducing food should address alertness, ability to tolerate oral stimulation, and presence of a non-nutritive suck or ability to manipulate a bolus. Oxygen saturation and respiratory rate during these activities may need to be monitored. Positioning restrictions secondary to physical limitations or medical interventions should also be identified, as these may have an impact on the child's ability to feed. A complete oral motor examination should also be completed to determine the presence of any structural or functional abnormalities of the oral musculature. The presence/absence of swallow response, laryngeal elevation, and vocal fold function should all be screened prior to the introduction of food.

With regard to the level of alertness, children with TBI and associated cognitive impairment are at increased risk for aspiration related to decreases in cognitive level. A retrospective study completed by the authors found a significant correlation between Rancho Los Amigos Level of Cognitive Functioning and swallowing ability (52).

Regarding oral presentation of materials, there are a number of aspects to consider. Until recently, the Evans

TABLE 4.13 FEEDING AND SWALLOWING TEAM MEMBERS

TEAM MEMBER	FUNCTION
Parents	Primary caregivers and decision makers for child
Physician	Medical leader
(Pediatric physiatrist, gastroenterologist, developmental pediatrician)	Team coleader Pediatric health and neurodevelopmental diagnosis Medical and health monitoring within specialty area
Speech–language pathologist	Team coleader (active in feeding clinic and coordinates programmatic activities) Clinic and inpatient feeding and swallowing evaluation VFSS with radiologist FEES (with otolaryngologist)
Occupational therapist	Evaluates and treats children with problems related to posture, tone, and sensory issues such as oral defensiveness Oral sensorimotor intervention program
Dietitian	Assesses past and current diets Determines nutrition needs Monitors nutrition status
Psychologist	Identifies and treats psychological and behavioral feeding problems Guides parents for behavior modification strategies Directs inpatient behavioral feeding program
Nurse	Organizes preclinic planning Reviews records and parent information Coordinates patient follow-up Changes gastrostomy tubes
Social worker	Assists families for community resources Advocacy for the child
Additional specialists	
Otolaryngologist	Physical examination of upper aerodigestive tract Detailed airway assessment FEES with speech–language pathologist Medical and surgical treatment of airway problems
Pulmonologist	Lower airway disease—evaluation and management
Radiologist	VFSS with speech–language pathologist CT scan of chest Other radiographic diagnostic studies
Pediatric surgeon	Surgical management of gastrointestinal disease
Cardiovascular surgeon	Surgical management of cardiac disease
Neurologist/neurosurgeon	Medical and surgical management of neurologic problems
Physical therapist and rehabilitation engineer	Seating evaluations and modifications to seating systems

Abbreviations: CT, computed tomography; FEES, fiberoptic endoscopic evaluation of swallowing; VFSS, videofluoroscopic swallow study.

blue dye test or modified Evans blue dye (MEBD) test was commonly used to detect aspiration at the bedside. Its use has recently become somewhat more controversial. A recent report in the literature of a retrospective study comparing results from the use of MEBD, FEES, and VFSS documents low sensitivity of this measure to aspiration and cautions the clinician regarding false negative results (53). Another study, reported by Tippett

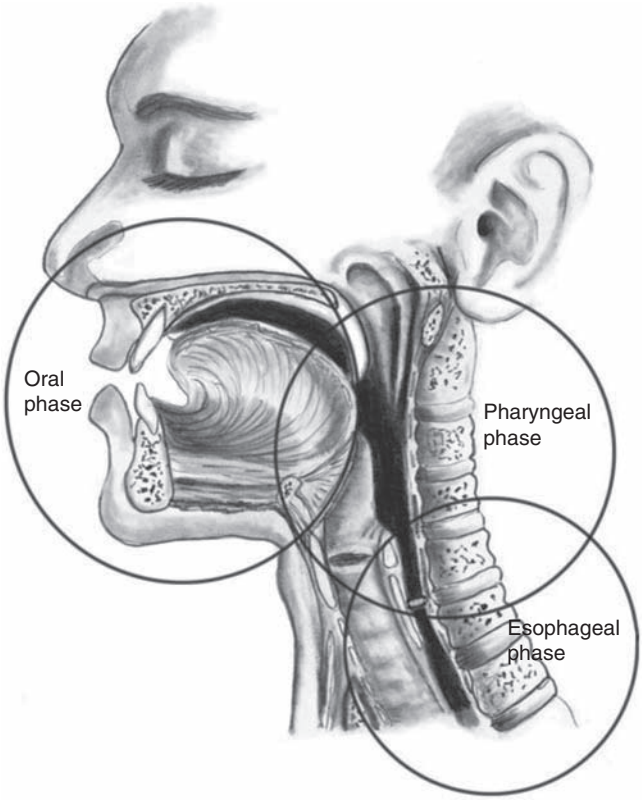


FIGURE 4.8 Phases of swallowing.

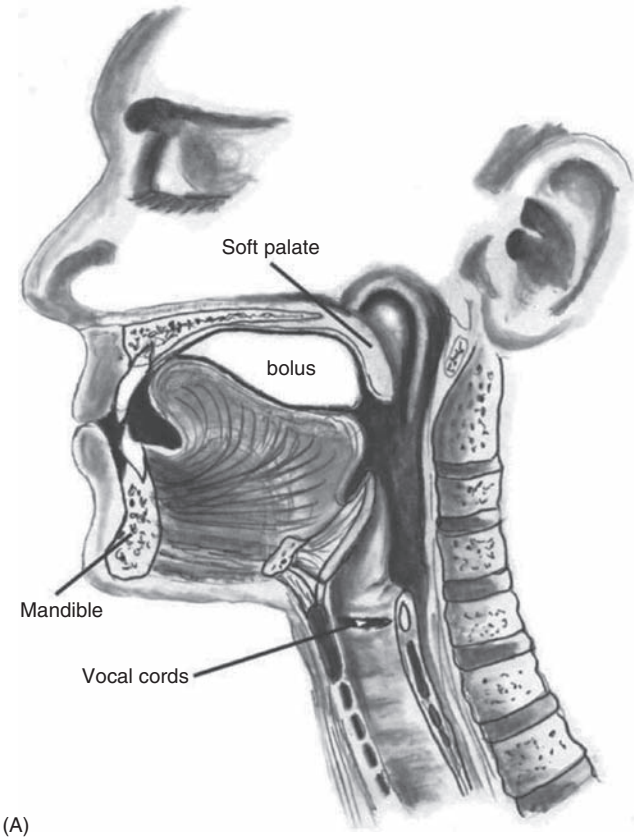
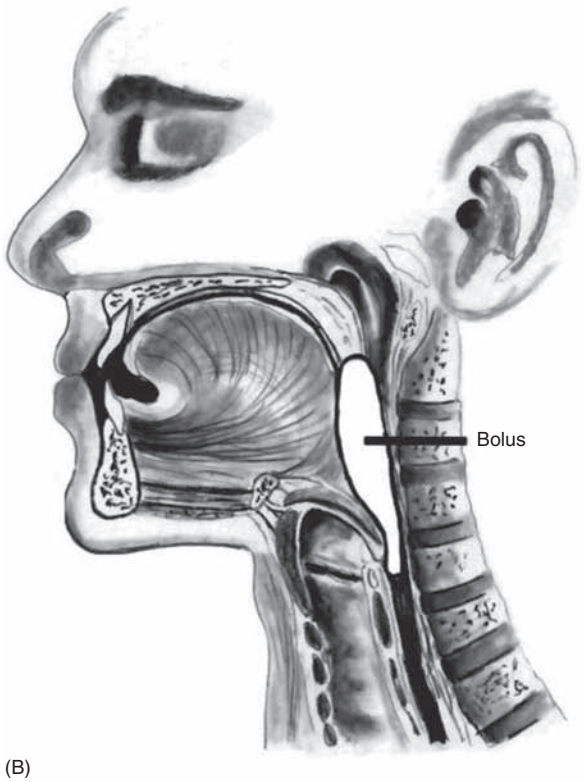


FIGURE 4.9 (A–C) Position of the bolus during phases of swallowing.



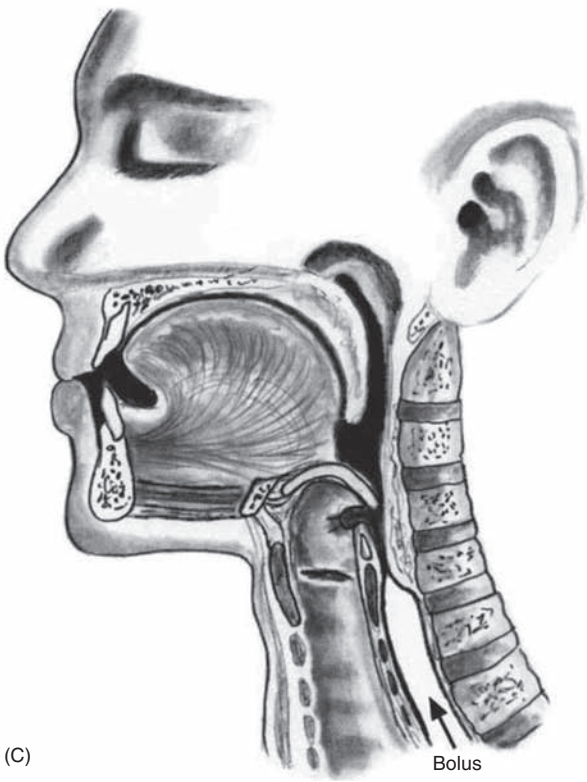


FIGURE 4.9 (*Continued*)

and Siebens in 1996, notes 90% sensitivity of the MEBD in detecting aspiration of dyed foods for a group of 34 consecutive patients with tracheostomies (54). Thus, although the validity of the study for determining aspiration remains controversial and requires further objective study, it remains a useful component of the bedside swallowing assessment for some children in determining safety for oral intake.

When using foods during the bedside assessment, a number of variables can be manipulated, including the presenter, the consistency, the mode of presentation, and the bolus size (55). Food can be presented by the clinician, the parent, or the child, depending on the readiness and medical stability of the child and the availability and willingness of the parent. The child's age, current oral motor status, and premorbid feeding abilities will all affect decisions regarding consistency, mode of presentation, and bolus size.

If aspiration is suspected during the bedside assessment (coughing/choking, drop in oxygen saturation, wet vocal quality), further instrumental assessment such as a VFSS is generally indicated. Instrumental studies will assist in providing more detailed information, such as when the aspiration occurs (eg, before, during, after the swallow), what factors caused the aspiration (eg, premature spillage, unprotected airway, cricopharyngeal dysfunction), and what compensations, if any (eg, food consistency, positioning, presentation), may improve the swallow. The VFSS assesses three phases of swallowing: oral, pharyngeal, and esophageal (Figure 4.8) (56). Figure 4.9 illustrates the position of the bolus during each of the three phases.

If there is no evidence of aspiration during the bedside assessment, recommendations are made for oral feeding based on the results of the trial feeding, the child's level of ability to feed orally, and the child's nutritional needs.

An alternative procedure, FEES, is sometimes recommended instead of VFSS (57). It involves the passage of a flexible fiberoptic endoscope transnasally to the area of the nasopharynx superior to the epiglottis, allowing observation of the swallowing mechanism from the base of the tongue downward. Use of FEES in the pediatric population has been established in the literature as a "practical and effective means of evaluating swallowing in children of all ages" (58,59). An advantage of VFSS is the ability to observe the actual aspiration event and to visualize the aspirated material in the airway. An advantage of FEES is the ability to observe amount and location of secretions and residue.

Instrumental examinations can be helpful in delineating pharyngeal and esophageal physiology as it pertains to swallowing. Decisions regarding when to perform an instrumental examination are guided by a number of factors, including risk for aspiration by history and clinical observation, documented incoordination of suck–swallow–breathe sequence during infant feeding, clinical evidence of pharyngeal or upper esophageal phase-swallowing deficits, prior aspiration pneumonia or similar pulmonary problems that could be related to aspiration, or etiology indicative of pharyngeal or laryngeal problem, such as neurologic involvement commonly associated with feeding and swallowing problems.

Factors determining which type of instrumental exam to use are outlined in Table 4.14.

Management decisions with regard to feeding may be complex, and a number of factors must be considered, including medical, nutritional, oral sensorimotor, behavioral, and psychosocial. Treatment may include direct and indirect strategies, depending on the swallowing deficit. Examples of direct strategies include use of positioning maneuvers such as chin tuck or supraglottic swallow. Examples of indirect treatment strategies include diet modifications (eg, thickening liquids), changes in feeding routine (eg, small amounts frequently throughout the day), or changes in presentation of food (eg, sippy cup versus bottle).

Diet texture modification is a common practice in management of dysphagia. Given the wide variation across clinicians and facilities, the American Dietetic Association attempted to establish some standard terminology

TABLE 4.14 INSTRUMENTAL SWALLOWING ASSESSMENT

FINDINGS BETTER VIEWED ENDOSCOPICALLY (FEES)	FINDINGS BETTER VIEWED FLUOROSCOPICALLY (VFSS)
Airway closure	Tongue control and manipulation of bolus
Amount and location of secretions	Tongue contact to posterior pharyngeal wall
Frequency of spontaneous swallowing	Hyoid and laryngeal elevation
Pharyngeal/laryngeal sensitivity	Cricopharyngeal opening
Residue build-up	Airway closure at level of arytenoid to epiglottal contact
Aspiration before the swallow	Epiglottic retroversion
Aspiration after the swallow	Esophageal clearing
Coordination of the bolus and airway protection	Aspiration during the swallow
Coordination of breathing and swallowing	Amount of material aspirated
Ability to adduct TVFs for supraglottic swallow maneuver	
Fatigue over a meal	
Altered anatomy contributing to dysphagia	
Effectiveness of postural change on anatomy	

Abbreviations: FEES, fiberoptic endoscopic evaluation of swallowing; TVF, true vocal folds; VFSS, videofluoroscopic swallow study.

and practice of texture modification through creation of the National Dysphagia Diet (NDD), published in 2002. The NDD was developed through consensus by a panel that included speech pathologists, dietitians, and food scientists. It proposes a hierarchy of four diet levels of semisolids and solids, as well as two levels for liquids (see Table 4.15) (60).

One treatment option for children that is somewhat controversial involves oral sensorimotor intervention. This treatment method is typically performed by either speech pathology or occupational therapy, and involves techniques that are directed toward improving a child's ability to accept, manipulate, and swallow foods successfully. These techniques may include work with the jaw, lips, cheeks, tongue, and palate, both with regard to desensitizing and improving function. Another more recent technique that has gained popularity is VitalStim® Therapy, a technique that uses external electrical stimulation on the anterior neck to promote improvement in swallowing function, and is used as an adjunct to more traditional swallowing treatment. Fairly extensive research has been completed in this area, but very little has been focused on pediatric patients, and what has been done has been inconclusive. According to Mary Christiaanse et al. in their study on neuromuscular electrical stimulation (NMES) for treatment of dysphagia in pediatric patients, NMES treatment in a heterogeneous group of pediatric patients with dysphagia did not improve the swallow function more than that seen in patients who did not receive NMES treatment (61).

The benefits of such treatment approaches are still inconclusive, with little evidence to date documenting efficacy, efficiency, and outcomes. Some children appear to improve oral function with variations in texture, tastes, and temperature of foods. Other children benefit from posture and positioning changes. To be most effective, treatment of swallowing disorders in children should ensure safety while promoting a pleasurable experience. Treatment should also include the primary caregiver in every session, as well as provide home programs and suggestions for how to work with children at home on a daily basis (62–64).

In conclusion, communication and swallowing are both complex acts that require coordination of multiple systems, and disruption in a single component in any one of those systems can and most often does result in some degree of communication or swallowing impairment. Assessment and treatment of these impairments requires thorough knowledge of development and disorders of relevant pediatric anatomy and physiology, as well as an understanding of how to apply that knowledge in evaluation and treatment to ensure the best possible outcome. As our field advances, and as we advocate for the most appropriate treatment for the children we serve, reliance on evidence-based practice has become, and will continue to be, a crucial component for success.

PEARLS AND PERILS

- Children with tracheostomies and those on ventilators are capable of oral communication and oral eating.
- Speech and language delay refers to typical development at a slower pace, while speech and language disorder refers to atypical development when compared with peers.

TABLE 4.15 DYSPHAGIA DIET LEVELS

DYSPHAGIA DIET CONSISTENCIES	EXAMPLES	INDICATIONS FOR USE
Thin liquids	Water, juice, soda	Adequate strength and coordination of lip and tongue musculature
Thick liquids	Nectars, milkshakes, cream soups, honey	Premature spillage of thin liquids with increased risk for aspiration
Mashed solids/purees	Yogurt, pudding, pureed meats and vegetables, cream of wheat	Mastication not required Child may have weak tongue/mandibular musculature or reduced mastication
Semisolid	Minced meats/fish, cottage cheese, scrambled eggs, soft mashed fruits or vegetables	Some mastication possible Fair oral motor control, although with some degree of oral weakness
Soft chunk solid	Poached or hard-boiled eggs, bananas, canned fruit, mashable vegetables, bread, cold cereal, pancakes, pasta, rice, noodles, cake, pie	Mastication necessary Appropriate for patients with adequate oral motor control but decreased endurance

Source: Adapted from Ref. (60). National Dysphagia Task Force. *National Dysphagia Diet: Standardization for Optimal Care*. Chicago, IL: American Dietetic Association; 2002.

- Autism spectrum disorder is four to five times more common in boys than girls, and has shown a 10-fold increase in prevalence in 40 years.
- Use of augmentative communication systems (devices, sign language, PECS) does not impede development of oral communication, and may, in fact, promote it.
- Liquids are the least safe alternative when initiating feeding following TBI due to delayed reaction times associated with cognitive level of recovery.

ACKNOWLEDGMENTS

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5

QUANTITATIVE ASSESSMENT OF GAIT: A SYSTEMATIC APPROACH

James J. Carollo and Dennis J. Matthews

Bipedal walking is a distinctive and uniquely human behavior that is celebrated in childhood and cherished across the life span. In the words of Dr. Aftab Patla, a renowned motor control expert who had a long and distinguished career at the University of Waterloo, "… nothing epitomizes a level of independence and our perception of a good quality of life more than the ability to travel independently under our own power from one place to another." Limitations in independent walking related to childhood disorders restrict education and employment options and access to social and cultural opportunities. Furthermore, a decline in walking ability directly affects the level of physical fitness so critical for health and well-being as we grow older, and contributes to a more sedentary lifestyle as an adult. Recent studies have linked habitual sedentary behavior to increased functional impairment and chronic disease risk in individuals with disability (1). This in turn can begin a cascade of chronic health issues and secondary conditions that can easily spiral out of control. Achieving and sustaining a functional level of walking ability is of the utmost importance to individuals with congenital or pediatric conditions and their families, and has a direct impact on their overall health status over their lifetime. Consequently, pediatric rehabilitation providers must be skilled in assessing walking and movement ability, and be comfortable utilizing all tools and techniques available to them to best manage their patients' care and expectations related to this most basic human function.

Over the last 25 years, *instrumented gait analysis* (IGA) has become more commonplace, evolving into an accepted, objective evaluation that can guide surgical and rehabilitation therapy planning for the child with personal mobility challenges. IGA provides a quantitative and comprehensive snapshot of one's characteristic movement pattern at a particular point in an individual's development or at discrete intervals in his or her treatment. Clinicians can use this information to describe the complex physiologic interactions that lead to abnormal movement and motor control, and better understand their impact on gait, movement, and other functional activities.

Observational gait analysis is a common topic during residency and most pediatric physiatrists are familiar with the procedures. However, training in the use of quantitative tools common to IGA and available from a modern clinical motion laboratory are frequently overlooked in postgraduate education, with the inclusion in the curriculum being highly dependent on the availability of such a facility during their training. Despite the advances in technology that have made IGA more commonplace in clinics around the world, only a handful of Physical Medicine & Rehabilitation (PM&R) residency programs are associated with motion analysis facilities. Even fewer are associated with clinical motion laboratories accredited by one of the two independent accreditation organizations, the Commission for Motion Laboratory Accreditation (CMLA) in North America, and the Clinical Movement Analysis Society (CMAS) of the United Kingdom and Ireland. Laboratories that have achieved accreditation conform to common standards of practice, quality, and information reporting that are the benchmark for IGA in clinical practice. This chapter can be used as a guide for those interested in utilizing data from accredited facilities in their practice regardless of their level of exposure to IGA in the past.

Mastery of IGA procedures along with the ability to perform meaningful interpretations that are clinically relevant remains challenging for many clinicians. We believe underutilization of modern gait analysis techniques in pediatric rehabilitation is related to the difficulty associating gait deviations seen in an IGA report with specific functional deficits during the walking cycle. Fundamental to making this connection is a clear understanding of the functional demands of normal gait. Armed with this understanding, the essential features of normal, efficient locomotion can be easily recognized, and this provides the basis for identifying the *absence* of these features in the child with gait dysfunction. When applied systematically,

this evidence-based approach can provide a logical strategy for clinical gait analysis and the management of gait disorders (2).

Therefore, the goal of this chapter is to familiarize clinicians with fundamental gait analysis principles by focusing on the inherent functional requirements of normal locomotion. This provides a framework for using specific measurements from an IGA report to pinpoint the joint and/or muscle system responsible for a particular functional deficit, and the subsequent target for clinical intervention.

NORMAL GAIT IS CYCLICAL AND SYMMETRIC

The principal goal of locomotion is to efficiently propel the body forward. The most natural way for humans to accomplish this task is to employ a bipedal gait pattern, where the base of support alternates between legs. Inman has described the cyclical alteration of each leg's support function and the existence of a transfer period when both feet are on the ground as *essential features* of normal locomotion (3). Since normal gait assumes no biomechanical advantage provided by either limb, a natural consequence of these essential features is the existence of a repeatable pattern that is both cyclical and symmetric. Figure 5.1 illustrates one complete gait cycle, or *stride,* and includes the *time periods* and *temporal events* associated with foot/floor contact that necessarily arise from changing the support limb. Temporal events are specific moments in time that divide the gait cycle into discrete time periods of finite duration, and are identified by the stick figures along the top of Figure 5.1. Typically, a cycle begins when one foot makes contact with the walking surface (initial contact) and ends when that same foot strikes again. This is the functional definition of a stride. Using such a convention allows a stride to be *time-normalized,* where a specific stride location is expressed as a percentage of the total cycle time or *stride period.* Time normalizing the gait cycle facilitates comparing subjects with different stride lengths, stride periods, and walking speeds on the same scale. Figure 5.1 illustrates the time periods and temporal events relative to the shaded ipsilateral side. If a subject's gait pattern is normal, the stride would be inherently cyclical and symmetric, with equal time periods on each side. The temporal event of foot-off (sometimes referred to as *terminal contact)* separates the gait cycle into stance and swing periods. Typically, the stance period accounts for 60% to 62% of the total gait cycle and the swing period takes the remaining 40% to 38%. We have intentionally refrained from using the terms "stance phase" and "swing phase" here to avoid confusing these intervals with the *phases* of gait to be introduced in a later section, although in common practice, the terms can be used interchangeably.

The stance period includes two intervals of double limb support at the stance/swing transitions, each lasting approximately 10% to 12% of the gait cycle at typical walking speeds. These are generally described as initial and final double support, but can also be identified in the context of the leading limb as right or left double limb stance period. The duration of the double limb support periods decreases with increasing walking speed, reaching zero at the moment running begins. The time interval between the initial and double support periods is defined as the single support period, and is the same duration as the swing period of the opposite limb. Assuming normal symmetry, any reduction in double limb support time is associated with a proportional increase in single limb support time, but since single limb support always corresponds to the contralateral swing period, the overall

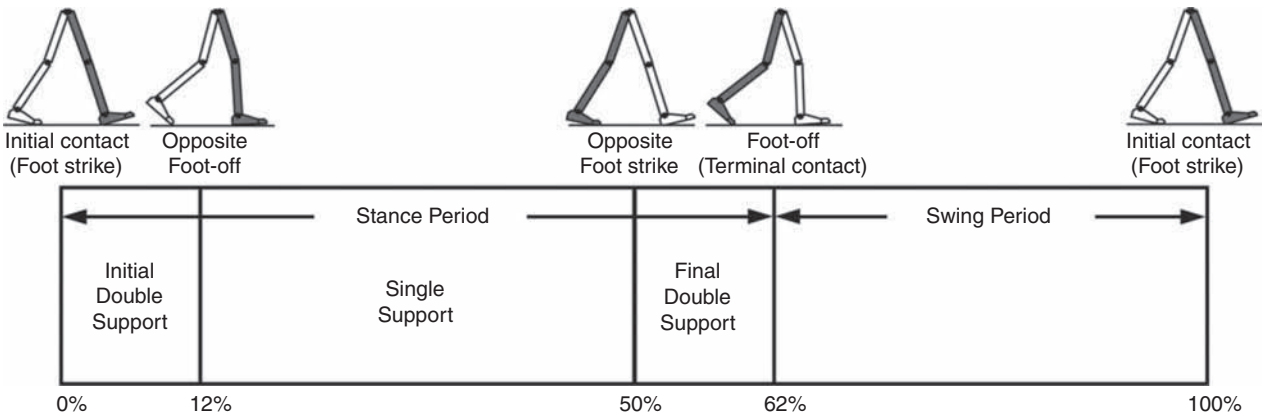

FIGURE 5.1 A typical gait cycle normalized in time, and represented on a linear scale from 0% to 100% of the total stride. This repeating cycle begins with initial contact and ends with the next initial contact of the same foot. The stick figures shown on top represent temporal events associated with foot-to-floor contact. They divide the cycle into swing and stance periods, one period of single support and two equal periods of double support.

stance period decreases, reaching 50% at the initiation of running when double limb support reaches zero. When both limbs' primary temporal events of foot strike (initial contact) and foot-off (terminal contact) are represented on the same time scale, the duration of each time period is easily illustrated.

These general terms for temporal events are applicable, regardless of gait pathology. Other terms are routinely used to identify temporal events marking the transition from the swing period to the stance period (heel strike, forefoot initial contact, foot-flat) and stance to swing (toe-off, push-off), but should only be used when they clearly describe the observed foot/floor contact pattern.

While period durations relative to a single side are easily described when the gait cycle is represented on a linear scale, left/right symmetry may be more easily conceptualized when the gait cycle is wrapped around a unit circle (2,4,5), as shown in Figure 5.2. For typically developing children and adults, ipsilateral and contralateral initial contact and foot-off will occur directly opposite each other around the circle, or 180 degrees out of phase. This graphically illustrates that the resulting time periods must be of equal duration for left and right single support, initial and final double support, and left and right swing periods. Any disruption in the natural sequence of temporal events anywhere along the cycle as a result of physical impairment, weakness, or spasticity will result in incorrect timing for the events that follow. This necessarily leads to a loss of symmetry that can be quantified by comparing the timing of temporal events between sides. Changes in symmetry reflected in the gait period durations are an index of gait pathology, and measuring this simple quantity can be quite useful for evaluating treatment performance over time.

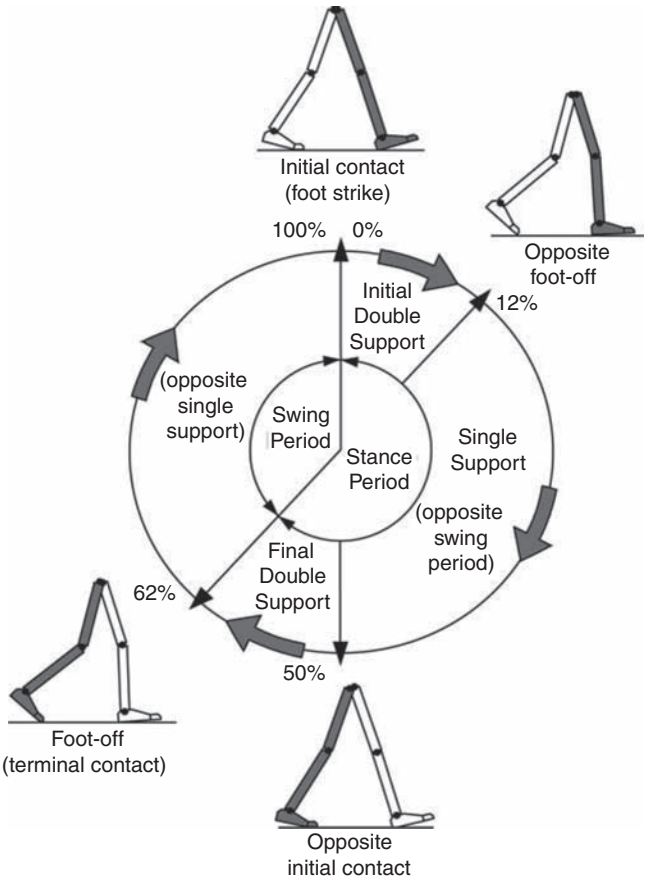

FIGURE 5.2 A typical gait cycle normalized in time, but wrapped around a continuous unit circle to illustrate symmetric phase relationships of temporal events and time periods. The beginning and end of the cycle occur at the 12 o'clock position. The temporal events of initial contact and foot-off for each leg are typically opposite each other on the unit circle, and the single support period of one limb is equal to the swing period of the opposite limb.

Since the duration of the swing period combined with leg length determines the distance covered by the swinging limb, deviation from normal symmetry and timing will give rise to differences in step length on each side, and subsequently the total distance traveled per gait cycle. By definition, *step length* and *stride length* are not synonymous. Step length is the distance (in the direction of progression) from a point of ground contact of the trailing foot to the next occurrence of the same point of ground contact with the leading foot. It is measured during double support and named for the leading limb. In contrast, stride length is the distance from initial contact of one foot to the next initial contact of the same foot, corresponds directly to the stride period, and is equivalent to the sum of successive left and right step lengths. Recognizing that speed is defined as the ratio of distance per unit time, the quantities of step length, stride length, cadence (steps per minute), and walking speed are mathematically related by simple formulas:

$$2\ steps = 1\ stride$$

$$\text{walking speed } (m/s) = [\text{cadence } (steps/min) \times \text{stride length } (m)]/120$$

$$\text{stride length } (m) = [\text{walking speed } (m/s) \times 120]/\text{cadence } (steps/min)$$

$$\text{step length } (m) = [\text{walking speed } (m/s) \times 60]/\text{cadence } (steps/min)$$

These basic outcome measures of overall gait performance, including the timing measures previously described and other quantities such as stance/swing ratio, are collectively known as *temporal–distance* or *temporal–spatial* parameters. They can provide considerable insight into the functional consequence of subtle gait abnormalities on walking performance. For example, children with cerebral palsy (CP) may experience foot clearance problems during limb advancement due to excessive ankle plantar flexion and/or decreased knee flexion during swing period. Evidence of this could be found in prolonged single support times on the more normal or less

involved side (a consequence of the longer swing time on the more involved side), and a reduced stance period, step length, and stance/swing ratio on the more involved side (6). If the source of the limb advancement problem can be attributed solely to the excess plantar flexion, the simplest intervention would be to prescribe a solid or posterior leaf-spring ankle foot orthosis (AFO) with a rigid plantar flexion stop to restrict excess plantar flexion during swing. Evidence that this intervention improved gait performance could be found in more symmetric single limb support times and step lengths, a more normal stance/swing ratio, and a higher walking speed.

While clinical motion laboratories routinely compare a patient's temporal–spatial measures to age-matched normative values, caution should be used when interpreting these results. Temporal–spatial parameters of cadence and stride length are directly related to walking speed (7), and since humans routinely walk at a variety of speeds, simple deviations from reference values alone may *not* be indicative of gait pathology. Rather, reduced values for these measures may simply reflect the need to adopt a speed appropriate to the terrain, the required task, or the size of the room (8). A person's natural gait is also dependent on the environment, with studies showing that subjects walk faster on a long walkway compared to a short one, and typically walk faster in outdoor studies compared to indoor studies (9). This lack of consensus regarding normal values supports the convention adopted by most clinical laboratories to compare patient results to their own laboratory-collected references, where these environmental factors can be consistent for all subjects. Nevertheless, while it is "normal" to walk at a variety of speeds, it clearly is abnormal to walk asymmetrically, so side-by-side differences in temporal–spatial measures within a particular patient should always be investigated.

When comparing temporal–spatial parameters in children, even greater care must be exercised, since several age-related differences arise from the close relationship of these measures to leg length and gait maturity (10). Sutherland has shown that in typically developing children, heel-first initial contact, sagittal plane knee flexion wave, reciprocal arm swing, and an adult joint angle pattern are acquired prior to the development of mature temporal–spatial parameters (11). All of these adult gait characteristics arise before the age of 3 years in most children (6). Because of this, Sutherland believes that gait maturity is best judged by the following five features, which he calls "determinants of mature gait" (11). These are: duration of single support, walking speed, cadence, step length, and ratio of pelvic span to ankle spread (P/A ratio). Notice that in addition to the first four measures that are fundamental temporal–spatial parameters, an anthropometric measure (P/A ratio) has been added, mainly to address the increased hip abduction and wider base of support common in the immature child's gait. In general, walking speed, step length, single support, and P/A ratio increase linearly with advancing age, with the greatest changes occurring during the first 4 years of life (6). Cadence decreases significantly between the ages of 1 and 2 years, after which it gradually continues to decrease (10). By age 4, the interrelationship between temporal and distance measures is fixed, although stride length and walking speed continue to increase with increasing leg length. Muscle phasic alterations in the early walkers are generally characterized by prolonged activation periods and subsequent longer periods of agonist/antagonist cocontraction around the joints of the lower extremities (12), most likely caused by neurologic immaturity associated with incomplete myelination (6). Despite these age-related differences that continue until skeletal maturity, the fundamental features and temporal–spatial parameter interrelationships associated with a normal, repetitive gait cycle are in place by age 4. For this reason, asymmetry detected using temporal–spatial measures can be used as indicators of gait pathology in both children and adults.

Because normal gait should be cyclical and symmetric, the existence of even small amounts of step-to-step variability may be an indication of gait pathology. Gait is most variable in the toddler, but gradually stabilizes as the child reaches adolescence (9). Hausdorff and colleagues have shown that the coefficient of variation for stride time in typically developing 3- to 4-year olds is approximately 6%, but decreases to 2% in 11- to 14-year olds (13). In older adults, increased variability is associated with increased risk of falling, with speed variability the single best predictor of falls (9). These examples provide further evidence of the importance of a cyclical and symmetric gait pattern and how variations in symmetry and cycle times reflected in the temporal–spatial parameters of gait may be associated with gait pathology.

TYPICAL COMPONENTS OF AN INSTRUMENTED GAIT ANALYSIS

As mentioned in the introduction, the phrase *instrumented gait analysis* (IGA) is often used to describe the application of computerized measurement technology to clinical gait analysis for the purpose of enhancing the interpretive power of the analysis beyond what can be discerned using observational and physical examination methods alone. The specialized nature of the systems used to perform an IGA typically requires a dedicated motion laboratory with specialists from clinical and technical disciplines to guide the patient through the testing procedures, make the required physical and anthropometric measurements, and capture and process all data (Figure 5.3). Analyses typically require 1.5 to 2 hours of patient contact time and between 8 and 12 hours of processing and analysis time, depending on the complexity of the patient referral and the number of measurements

FIGURE 5.3 A motion laboratory clinical specialist works to place reflective markers on a subject while the technical staff prepares for data capture.

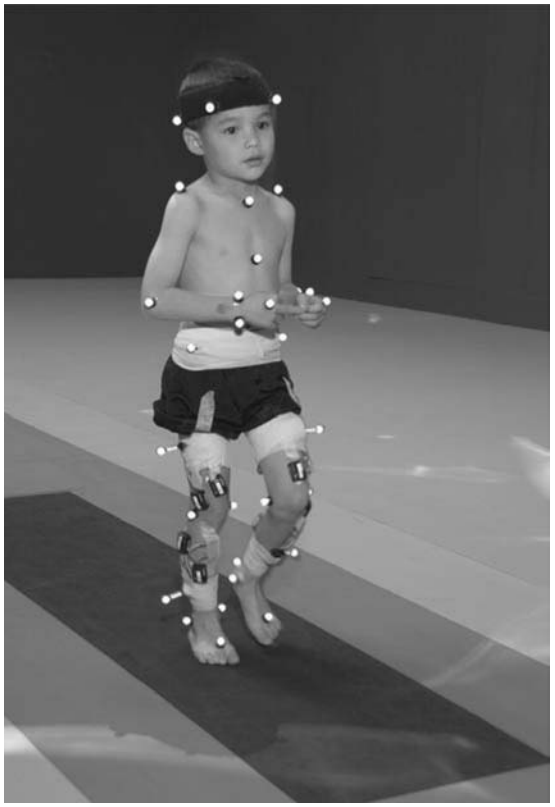

FIGURE 5.4 Subject with reflective markers or targets placed at strategic anatomic locations walks through a modern motion analysis laboratory. The location of the targets depends on the mathematical requirements of the limb-segment model used to calculate the kinematic values needed for analysis. This subject is using a full body marker set based on the Conventional Gait Model.

required to answer the clinical question. It is not within the scope of this discussion to comprehensively describe the full set of measurement tools available for clinical gait analysis in children. For this, the reader is referred to several excellent descriptions that are widely available (5,14–20). However, since it is important for the discussions that follow, we will briefly introduce the primary measures used, some tips for their practical application, and give examples of typical recordings as a reference.

In addition to the temporal–spatial parameters described in the last section, the primary measurements comprising IGA are gait *kinematics, kinetics, and dynamic electromyography* (d-EMG) (16). While there are certainly additional areas of measurement and many useful instruments that can be included in a comprehensive IGA, these three measurement categories are commonly accepted as the minimum necessary for clinical evaluation of the patient with gait dysfunction. They have also been identified as a specific requirement for accreditation by the CMLA and the CMAS of the United Kingdom and Ireland (21).

Gait kinematics is a general term that refers to measurement of the linear and angular displacements, velocities, and accelerations of body segments throughout the gait cycle. Generally expressed in terms of the joint angles between each limb segment, these quantities are most often described three-dimensionally using anatomical planes relative to the more proximal segment, but also includes the global position of the pelvis (pelvic tilt, obliquity, and rotation) and foot (foot progression angle) relative to a fixed laboratory coordinate system typically located in the middle of the walkway. Modern kinematic analysis systems use an assortment of markers or targets that are attached to the subject at strategic locations and can be tracked by specialized cameras or electromagnetic detectors (Figure 5.4). The kinematic measurement system identifies the position of the targets from multiple perspectives in three-dimensional (3D) space using a high sampling rate (>100 Hz) as the subject walks through a calibrated measurement volume. This determines a unique trajectory for each target, which can then be reconstructed by the computer utilizing a kinematic limb-segment model to produce a 3D animation of the walking subject within the virtual environment of the computer display (Figure 5.5). From this mathematical representation of the subject, kinematic graphs and interactive reports can be produced to facilitate the clinical analysis of the child's gait pattern.

Kinematic measurement systems rely heavily on motion-capture technology and specialized software that, fortunately, have found a major market in the video game and motion picture industry. This has had the positive effect of substantially lowering the startup cost of these systems in recent years, making the technology more available to the clinical community and improving the accuracy, precision, camera resolution, and processing speed. These advances have also increased the complexity of the kinematic models that can be implemented, which offers the promise of more comprehensive and anatomically correct descriptions of motion. However, it

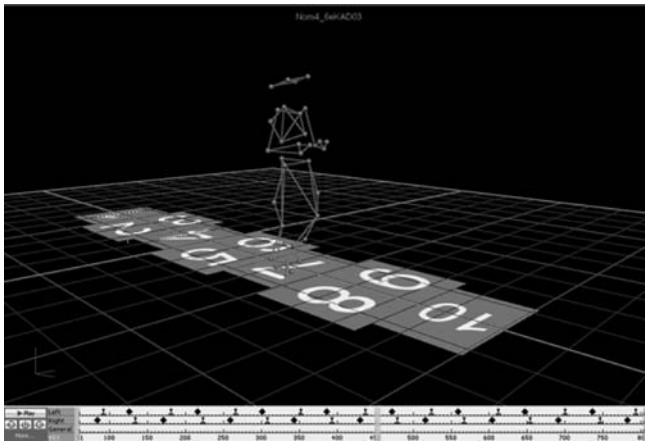

FIGURE 5.5 Three-dimensional animation of the walking subject within the virtual environment of a computer display.

may also introduce new challenges since increased model complexity necessitates greater software complexity. Furthermore, the requirement for model validation with each new software release necessitates regular laboratory procedural changes, and may introduce data discrepancies when patient results are compared over time using different models. Recognizing these potential technical concerns, gait kinematics represent an integral component of clinical movement analysis and are essential for analyzing the child with gait dysfunction. Figure 5.6 shows a set of 3D kinematic graphs associated with a sample of typically developing 12 to 13-year-old subjects used as a normal reference in our laboratory. We will discuss these kinematic graphs in more detail when discussing critical events in a later section.

While measurements of gait kinematics provide a quantitative description of body segment and joint movement during walking, *gait kinetics* describe the forces that

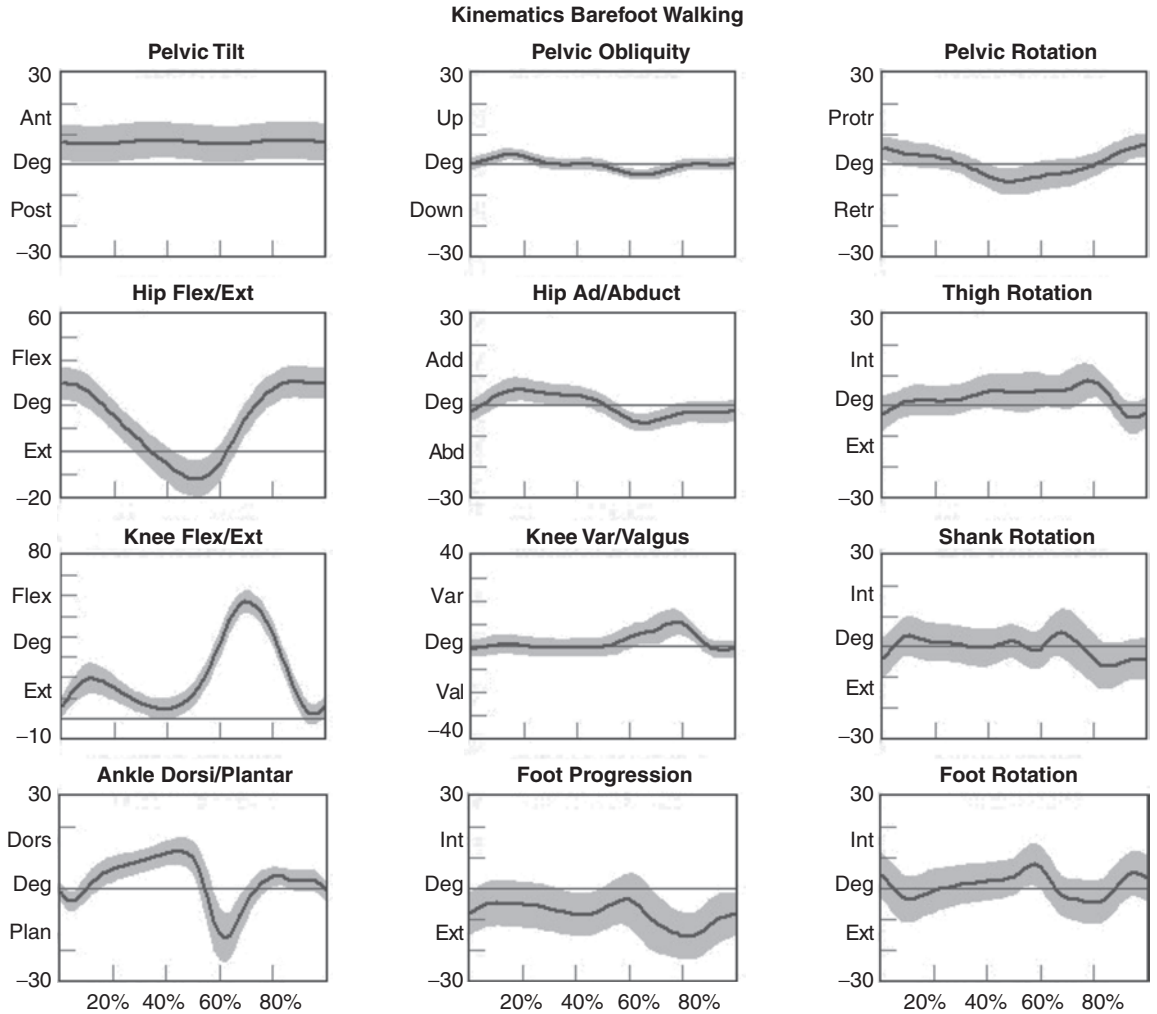

FIGURE 5.6 Normal three-dimensional kinematic graphs constructed using a sample of typically developing 12- to 13-year-old subjects. These data are used as a reference for comparing kinematic data from clinical subjects of the same age. The dark line is the average of all subjects and the gray band represents ± one standard deviation.

cause these movements and the calculated quantities that arise when a subject's reaction forces and 3D kinematics are applied to a mathematical model of the body. Since joint and muscle forces cannot be measured directly from the walking subject, the forces due to foot/floor contact are measured using a specialized instrument known as a *force platform* embedded in the walkway. The force platform measures the vertical, fore–aft shear, and medial–lateral shear components of the *ground reaction force* (GRF), which is the force vector acting at the supporting surface that is equal and opposite to the sum of all muscular, gravitational, and inertial forces generated by the body in motion. Since a force platform measures the magnitude and direction of the GRF as a single resultant vector quantity, only one foot can be in contact with the platform at a time for a valid measurement. In order to measure multiple foot strikes from both feet, the subject either needs to walk multiple times across a single platform or the laboratory needs to include a *force platform array* with multiple platforms in different orientations so that several clean foot strikes from both sides can be recorded in as few a number of passes as possible. A larger force platform array reduces alterations of gait characteristics in children with neuromuscular diseases in several ways. Installing multiple force platforms into the walkway reduces the number of trials required and thus minimizes the risk of fatigue. Furthermore, having multiple force platforms lessens the possibility of "targeting," which will alter the subject's characteristic gait pattern. Figure 5.7 shows the large 10-platform array of 60 cm × 40 cm force platforms currently used in our laboratory, and illustrates how sequentially rotating the long axis of each platform 90 degrees can accommodate a wide variety of stride lengths and step patterns for children and adults.

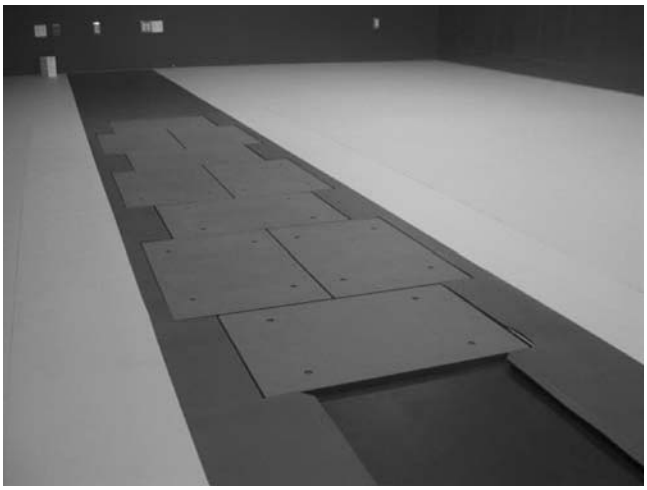

FIGURE 5.7 Large 10-platform array of 60 cm × 40 cm force platforms used in The Center for Gait and Movement Analysis at Children's Hospital Colorado in Aurora, Colorado. The "hopscotch" pattern of the platform array permits the recording of several individual foot strikes from both feet in a single walking pass. For illustration purposes, each platform is shown without its protective floor covering, which caused the platforms to blend into the surrounding walkway when applied.

The direct measurement of the individual force components and the vector sum of the GRFs has historically been used to evaluate gait kinetics and facilitate a more qualitative pre-/postsurgical comparison. The most useful clinical application of gait kinetics, however, is when it is combined with GRF measurement and a kinetic model of the lower extremities to calculate joint kinetics, specifically joint moments and powers (22). The most common way to accomplish this is to apply an "inverse dynamics" model of the lower extremity using the anthropometric dimensions of each segment (typically seven segments, including the pelvis and both thighs, shanks, and feet) and estimates of each segment's center of mass (COM) and inertial quantities. The forces at each joint can then be solved sequentially, starting from the GRF at the floor and working proximally, using the linear and angular forms of Newton's second law:

Linear: *force* = *mass* × *acceleration* (F = ma);
angular: *joint moment* = *moment of inertia* × *angular acceleration* (M = Ia).

By convention, joint moments can be considered either *external* or *internal*. External moments reflect the forces, acting on the body through the skeleton, that arise from the GRF, and since they reflect an external biomechanical load, are sometimes called *demand* moments. Internal moments describe the force generated by the muscles and ligaments acting on the skeleton to balance the external moments, and because they are counteracting an external load, are sometimes called *response* moments. Aside from their different functional descriptions, external and internal moments for the same joint are of equal magnitude and differ only in their mathematical sign. The joint moments described in a typical IGA report are internal moments, but this should always be confirmed since the sign and direction of the curves will be reversed if they actually describe external moments. Joint moments are vector quantities that describe the net torque around each joint but do not provide the individual force contribution from each agonist/antagonist pair or from individual muscles. Nevertheless, the net moment around the joint is quite useful because the magnitude and sign of the curve at any instance in the cycle can illustrate if one half of the agonist or antagonist pair is dominating at a specific point in the gait cycle. Net moment values can aid in clinical interpretation of gait by comparing them to reference values for typically developing children and by observing changes in the values before and after treatment. In addition, net moment values are helpful in understanding how a child may be compensating at a given joint for weakness or limited range of motion (ROM) at an adjacent joint. Figure 5.8 shows the sagittal plane kinematics,

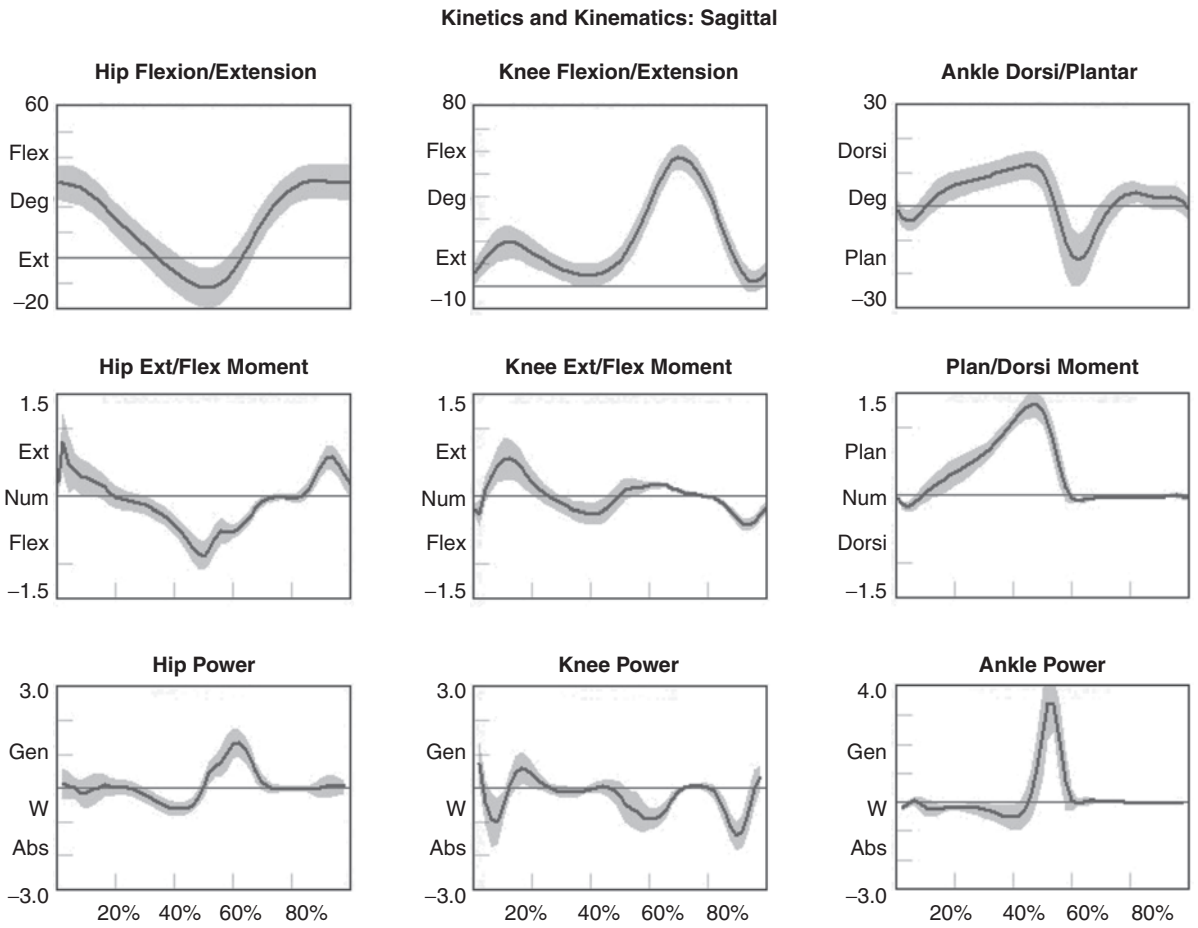

FIGURE 5.8 Graphs of sagittal plane kinematics, sagittal plane joint moments, and total joint power for the hip, knee, and ankle constructed using a sample of typically developing 12- to 13-year-old subjects. These data are used as a reference for comparing kinetic data from clinical subjects of the same age. The dark line is the average of all subjects and the gray band represents ± one standard deviation.

sagittal plane joint moments, and total joint power for the hip, knee, and ankle from a sample of typically developing 12 to 13-year-old subjects that we use as a normal reference.

Once the 3D moments at each joint have been calculated, joint power at any time in the gait cycle is the product of the joint moment and the corresponding angular velocity (instantaneous slope of the joint angular displacement curve from kinematics) at each percent interval of the gait cycle:

$$\text{joint power} = \text{joint moment} \times \text{joint angular velocity}$$
$$P(t) = M(t) \times \omega(t)$$

Similar to joint moments, joint power describes the net power at a joint rather than the individual power generated by a particular muscle or agonist/antagonist pair. Because this quantity is derived from both kinematics and ground reaction forces, joint power provides insight into the biomechanical mechanisms responsible for specific movements and, in a sense, quantifies the actual performance of the "motors" driving a particular gait pattern. In this way, joint power curves are extremely useful to identify when a particular joint is generating power (positive indicates concentric contraction) or absorbing power (negative indicates eccentric contraction) to analyze the transfer of power or energy from one joint to another and for understanding how one joint can compensate for disability at an adjacent joint. It should be pointed out here that although joint power is perhaps the single most informative biomechanical variable that can be obtained from an IGA, it does have limitations (23). For one thing, power is technically a single *scalar* quantity describing all planes of a joint combined, unlike displacement, velocity, and joint moment, which are directional vector quantities with individual component values for each anatomical plane. While in most cases the greatest contribution can be assumed to arise from the sagittal plane, the lack of a true directional component (especially at the hip) may lead to incomplete clinical interpretations. Another issue is that since extensive use of mathematical modeling is required to arrive at the joint power values, there are numerous assumptions made

in the process and great opportunity for errors or artifacts to influence the final curves. These issues should be considered when utilizing any kinetic variable for clinical decision making. However, they should not hinder the use of this information since these estimates cannot be obtained in vivo by any other means and still provide considerable insight into the functional causes of gait abnormalities.

Electromyography (EMG) is an important tool for evaluation of muscle and neurologic function and is well understood by the pediatric physiatrist. When used in the context of IGA, the purpose is slightly different from the conventional application. The primary objective of EMG in clinical gait analysis is to identify periods of muscle activation during walking so that decisions can be made regarding the appropriateness of muscle timing for agonist pairs as they selectively activate and deactivate during the gait cycle. This is the reason that we refer to this as *dynamic electromyography* or d-EMG, since the focus is on the phasic response of muscle during walking or some other functional activity. Since the subject will not be in a stationary position for the test, the instruments and technical procedures are also different from conventional diagnostic EMG. d-EMG requires a bipolar arrangement of electrodes and miniature differential amplifiers with a high common mode rejection ratio (CMRR) placed close to the site of the recording to ensure the EMG signal is not overwhelmed by motion artifact while the subject moves (Figure 5.9). Differential amplifiers with high CMRR (typically greater than 90) amplify voltage differences between the inputs and reject common voltages that may arise from movement of the electrodes or the soft tissue vibration that occurs with foot contact. Surface electrodes are the most commonly used electrode type for recording d-EMG from the pediatric patient to avoid the emotional trauma and change in gait pattern that indwelling electrodes often cause. Typically, the active portion of each electrode in the bipolar pair should be small and the pair should be placed as close together as possible along the long axis of the muscle (≤1 cm diameter, ≤2 cm separation) to minimize the effect of crosstalk from surrounding muscles. Unfortunately, surface electrodes are only suitable for recording muscle groups that are directly subcutaneous; if there is a need to evaluate deeper muscles individually, fine-wire electrodes made of a bipolar pair of 50-micron platinum wire must be inserted directly into the muscle of interest using a 25- to 28-gauge needle. When required, this is the most invasive aspect of an IGA, and should be used only when necessary in the pediatric patient and after all other data have been collected, since the level of patient cooperation and the likelihood of a typical gait pattern decrease considerably after a needle stick. In practice, most of the muscles of interest to the pediatric physiatrist can be successfully recorded using the surface electrode approach if proper procedures to minimize crosstalk and reduce motion artifact are followed.

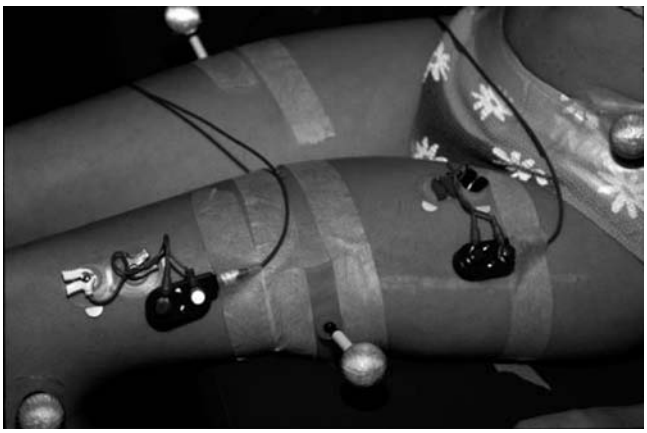

FIGURE 5.9 Patient with bipolar surface electrodes and small instrumentation amplifiers for recording dynamic EMG while he or she walks. This illustrates the electrode placement for the left vastus lateralis (distal location) and left rectus femoris (proximal location). Each electrode is connected to an instrumented backpack and then hardwired to the recording instruments. A wireless EMG recording system using similar electrodes but with individual transmitters for each muscle is shown in Figure 5.4.

Before the EMG recording can be used for clinical interpretation, the raw data must be filtered, processed, and time-normalized so that periods of muscle activation during the gait cycle can be identified. A good reference for processing guidelines is available from the International Society of Electrophysiology and Kinesiology, where they state that surface electrode recordings should be bandpass-filtered between 10 Hz and 350 Hz and fine-wire recordings filtered between 10 Hz and 450 Hz. This maximizes the signal, minimizes the noise, and reduces motion artifact. In modern systems, the filtered EMG data are sampled by analog-to-digital converters. Further processing is performed by computer using specialized software or in concert with the motion-capture system. Data can be presented as a continuous recording of "raw" EMG, an ensemble average of several cycles of EMG normalized to the gait cycle, or as linear envelopes reflecting the EMG magnitude throughout the gait cycle after rectification and integration of the raw EMG signal. In our laboratory, we have developed a system to superimpose the EMG signal over the observational video recording of the walking subject to screen for faulty EMG recording during the analysis and to better understand the interaction between observed movement and muscle activation (see Figure 5.10, right side). Regardless of how these data are presented, the goal is to use the EMG recording to identify periods of abnormal muscle activity and determine if this activity is responsible for abnormal movement patterns presented by the patient. Typically, the patient's activity is compared to a normal EMG reference, and deviations from normal values are scrutinized for their contribution to the

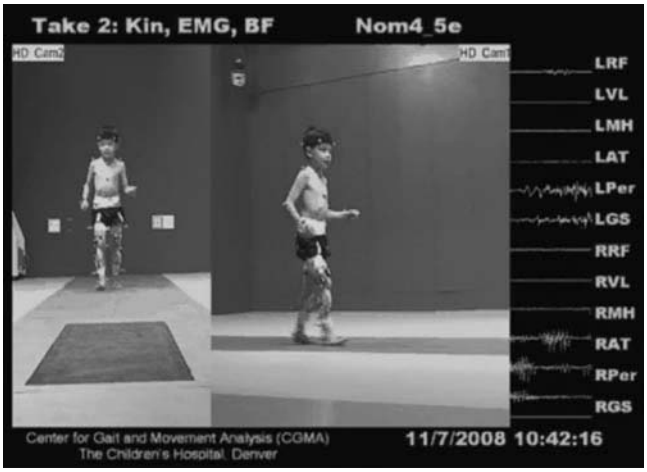

FIGURE 5.10 Biplane high-definition video recording with superimposed real-time EMG traces of six muscles bilaterally from a typically developing subject used as a reference at CGMA. The raw, unfiltered EMG recording provides immediate feedback on the quality of the EMG signal and the synchronization of muscle activity with observed movement.

overall movement pattern. Figure 5.11 shows filtered and time-normalized EMG for 12 muscles of the lower extremity from a 15-year-old typically developing subject used as a laboratory reference, along with published normal EMG activations represented as solid black bars at the bottom of each graph. The high-magnitude sections of the EMG recording for each muscle correspond to the published normal values, confirming that this typically developing subject has a normal adult activation pattern.

When EMG recordings are combined with kinematics, kinetics, temporal–spatial parameters, radiographs, and physical examination results, a comprehensive snapshot of the subject's walking pattern is revealed, providing an empirical basis for identifying the functional cause of a gait abnormality. However, to use these data successfully, we must return to the normal gait cycle to understand the functional requirements for walking, since these requirements are a natural consequence of subdividing the cycle on the basis of function.

IMPAIRMENT IDENTIFICATION IS FACILITATED BY SUBDIVIDING THE GAIT CYCLE

While a repetitive gait cycle arises from the alternating base of support used by all bipeds, the existence of this cycle provides great opportunity for clinical and biomechanical analysis of a child with gait dysfunction. In particular, a repetitive cycle lends itself to natural subdivision, which in turn, leads to a sequence of events that must be performed in order and with the correct timing for efficient walking to occur. The earlier section titled "Normal Gait Is Cyclical and Symmetric" discussed *temporal* subdivisions of the gait cycle delineated by foot/floor contact and their use in comparing limb symmetry, measuring outcomes, and the general characterization of overall gait performance. The focus of the current section is to describe another type of gait-cycle deconstruction, one based on *functional* subdivisions. For this approach, the functional prerequisites of walking are identified, providing a framework for subdividing the gait cycle into functional divisions (24). It is then possible to use the measurements available from IGA to identify quantitative differences at each joint and the specific functional abnormalities that occur at critical moments in the gait cycle (25,26).

FUNCTIONAL PREREQUISITES FOR WALKING

In their landmark paper published in 1953, Saunders, Inman, and Eberhart (27) described six gait subdivisions that they referred to as the "determinants" of normal gait. This treatise was significant in that it was perhaps the first formalized delineation of the gait cycle that explained how coordinated movements of the hip, knee, and ankle at specific points in the cycle led to efficient forward progression. Each determinant's influence on the 3D path of the whole-body COM was described using simple theoretical models, and the cumulative effect of all six determinants led to a smooth, low-amplitude trajectory that was assumed to be consistent with optimal, efficient locomotion. Specifically, Inman and colleagues believed that minimizing vertical and horizontal motion of the COM would maximize walking efficiency, since unnecessarily raising and lowering the COM would be wasteful from a potential energy perspective. By changing functional limb length with the addition of joints to an initially jointless model of the lower extremities and pelvis, each determinant served to smooth different portions of the COM trajectory, effectively raising the COM during double support and lowering it during single support. While it is true that unnecessarily large and abrupt movements of the COM reduce gait efficiency, some of the specific determinants identified by Inman and colleagues have now been discredited (28–30). The improved accuracy and temporal resolution of kinematic measurement instruments over the last 50 years have uncovered problems with the timing of some of the theoretical mechanisms described in the original paper, and in the case of longer step lengths, larger COM displacements are not necessarily associated with decreased gait efficiency.

While the relevance of specific determinants may now be in dispute, the real impact of this work is that it inspired generations of investigators to consider biomechanical explanations for gait dysfunction and led a few students of Dr. Inman to develop clinically applicable gait-cycle decompositions derived from the functional requirements of walking. In a later monograph (3), Inman

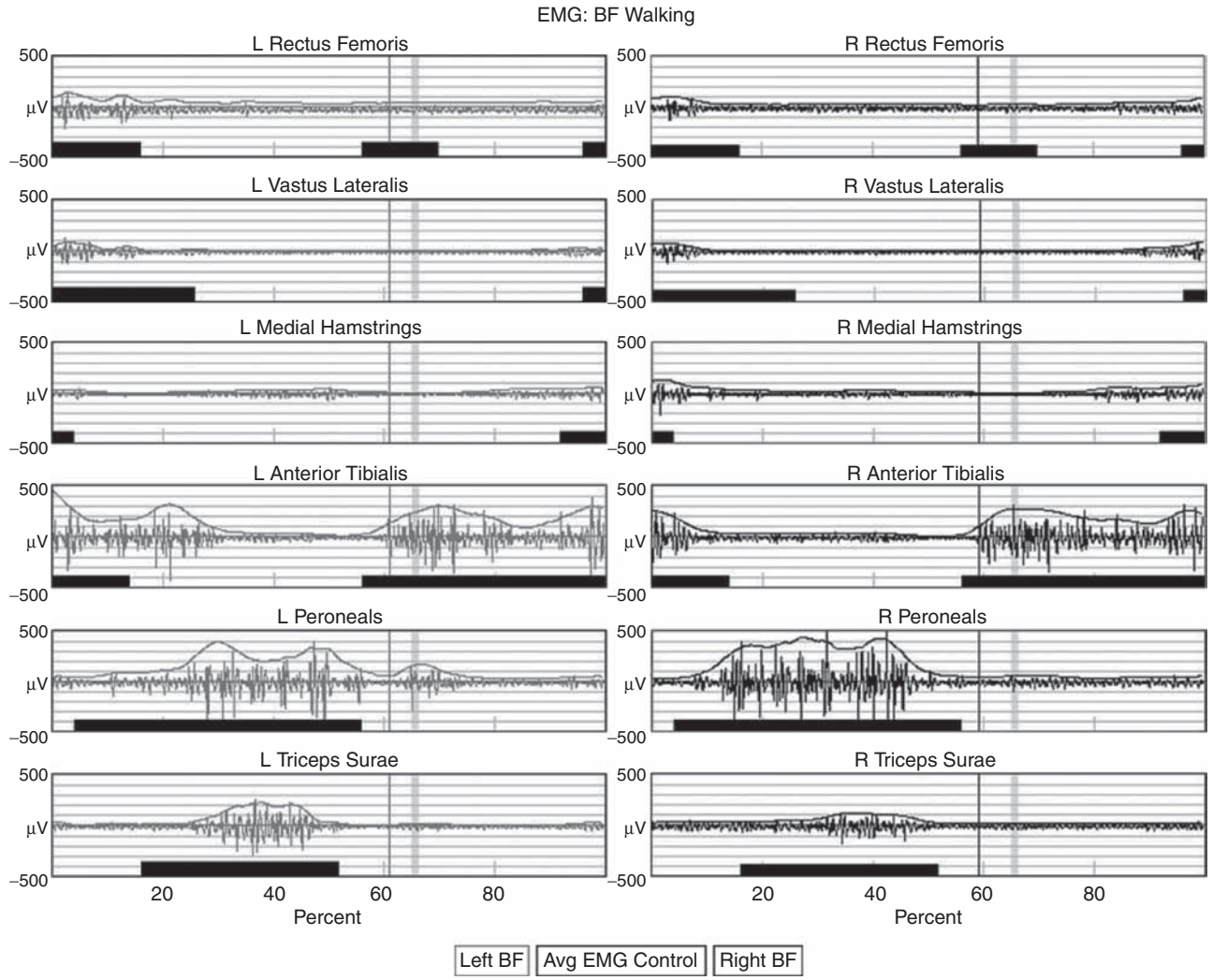

FIGURE 5.11 Filtered and time-normalized EMG for 12 muscles of the lower extremity from a typically developing 15-year-old subject. The black bars at the bottom of each graph are constructed from published normal EMG activations and are used as reference values. The smooth curve above the EMG activation is a processed EMG signal obtained by rectifying and integrating the raw EMG. Notice that each EMG activation pattern above a baseline level is contained within the reference bar indicating a normal EMG pattern. In this typically developing subject, there is very little EMG activity from the rectus femoris during initial swing.

described two basic functional requisites of walking that he deemed necessary for any form of bipedal gait, no matter how distorted by physical disability or assisted by prosthetic or orthotic devices. These are (a) continuing GRFs that support the body and (b) periodic forward movement of each foot from one position of support to the next.

These essential features of walking give rise to a periodic gait cycle that must always be present for continued locomotion. An orthopedic resident of Dr. Inman, Jacquelin Perry, recognized that the physical demands of supporting the body against gravity varied, depending on whether the stance limb was accepting the initial impact or continuing to carry the weight of the body during single support. To address this, she developed the notion of three functional gait *tasks* (31): weight acceptance, single limb support, and swing limb advancement. Dr. Perry considered weight acceptance the most demanding of the three functional gait tasks since it requires the stance limb's musculature and bony and ligamentous structure to provide shock absorption, initial limb stability (stiffness), and maintenance of forward progression. Preparation for the demands of weight acceptance begins late in the swing period, when pre-positioning of the leading limb occurs to correctly align the foot to accept weight at initial contact. The physical demands are lower for the task of single limb support, despite the fact that one leg alone has the complete responsibility for supporting body weight, maintaining whole-body stability (balance), and restraining forward momentum. This reduced physical demand during the task of single limb support is due to the inherent passive stability provided by the knee ligamentous structure and

the force balance at the hip as body weight moves forward (31). An essential functional requirement for this task is strong eccentric contraction of the calf musculature to control the tibia (and subsequently the rest of the stance limb) as it rotates over the fixed base of support provided by the foot. When the task of single limb support ends and swing limb advancement begins, the physical demands increase once again, since the three goals of weight transfer, limb advancement, and foot clearance must all be accomplished. Similar to the weight acceptance task, important preparatory actions must begin before the swinging limb is lifted from the supporting surface at the end of the stance period to meet the functional demands of swing limb advancement.

Findings from other investigators support the existence of these three fundamental gait tasks, although each investigator has used a somewhat different terminology when describing them (Table 5.1). Winter has characterized walking as an extremely complex motor control task that requires three elements: support control to prevent collapse against gravity (32); balance control of the head, arms, and trunk (HAT) acting as an inverted pendulum (33); and safe and coordinated lower limb movement during swing for minimum foot clearance and gentle heel contact (19). Dr. Winter and colleagues have also stated that the goals of these tasks can still be accomplished after disease, injury, or loss of function because of the inherent redundancy of lower extremity musculature and rapid adaptability of the central nervous system (34). It is interesting that Perry and Winter have identified essentially the same three gait tasks, despite approaching the study of gait from two different perspectives: clinical analysis of pathologic gait and biomechanics of human movement, respectively. This lends support to the existence of these three elements and warrants using them to understand functional deficits in subjects with gait pathology.

Gage has expanded on this description by identifying five elements essential to walking that he has referred to as "priorities" or "prerequisites" of normal gait (35). This functional subdivision of the gait cycle encompasses the three tasks described previously, but adds swing period elements necessary to ensure appropriate weight acceptance and the global task of energy conservation. In the order of functional priority, these are stability of the weight-bearing foot throughout stance, clearance of the non-weight-bearing foot during swing, appropriate pre-positioning of the swinging foot in preparation for initial contact, adequate step length, and energy conservation. This prioritization is influenced by Dr. Gage's interest in the gait of children with CP, and includes a gait efficiency task (energy conservation) to address the reduced functional capacity or endurance of many individuals with pathologic gait. He also identifies several physiologic and biomechanical mechanisms common to normal gait that can improve energy conservation. These are eccentric muscle contraction, return of "stretch energy" from prestretched muscles immediately prior to concentric contraction, biarticulate muscles functioning as energy transfer straps, and joint passive stability from the effects of GRFs whenever possible to spare muscle activation (36). While other investigators have addressed the functional prerequisites of gait (15,25,37), the contributions described previously form the basis of the strategy described in this chapter.

TABLE 5.1 FUNCTIONAL SUBDIVISIONS OF THE GAIT CYCLE ATTRIBUTED TO DIFFERENT INVESTIGATORS

INVESTIGATOR	INMAN	PERRY	WINTER	GAGE
Subdivision nomenclature	Requisites	Tasks	Motor control tasks	Prerequisites
Functional subdivisions	Continuing ground reaction forces that support the body	Weight acceptance	Support control to prevent collapse against gravity	Stability of the weight-bearing foot throughout stance
		Single limb support	Balance control of the HAT	
	Periodic forward movement of each foot from one position of support to the next	Swing limb advancement	Safe and coordinated limb movement during swing to achieve:	Clearance of the non-weight-bearing foot during swing
			• Minimum foot clearance • Gentle heel contact	Appropriate pre-positioning of the swinging foot in preparation for initial contact Adequate step length Energy conservation

Abbreviation: HAT, head, arms, and trunk.
Source: Adapted from Refs. (3), (19), (31)–(33), and (35).

PHASES OF THE GAIT CYCLE

Since the tasks of weight acceptance, single limb support, and swing limb advancement can only be accomplished successfully if appropriate limb movement patterns occur sequentially and with correct timing, Dr. Perry developed a systematic method of subdividing the gait cycle to simplify pattern identification and facilitate observational gait analysis (24). Now known as the *Rancho* classification, in honor of Rancho Los Amigos Medical Center where Dr. Perry and colleagues of the Pathokinesiology Service developed this method, it relies on eight subdivisions of the gait cycle, referred to as *phases* of gait. While in general, both phases and periods refer to specific time slices around the gait-cycle unit circle, Perry prefers to use the term "phase" for intervals that have specific functional significance and have a clear relationship to the three identified gait tasks described in the previous section. The Rancho classification provides a framework for functionally organizing the gait cycle harmoniously with the three fundamental gait tasks, and after 30 years of refinement, this approach has proven to be a powerful tool for identifying specific functional deficits or gait impairments during each phase of gait. IGA can be used to quantify the magnitude of a functional deficit at a joint by reviewing the kinematic and kinetic data, or abnormal muscle timing by reviewing the EMG. This provides evidence and helps pinpoint the specific region or system most responsible for the overall gait abnormality, and suggests interventions to directly correct the identified functional deficit or gait impairment in each phase.

TABLE 5.2 THE EIGHT PHASES OF GAIT AS DESCRIBED BY DR. JACQUELIN PERRY

PHASE OF GAIT	DESCRIPTION
Initial contact	The moment when the foot strikes the ground
Loading response	Initial double support period when the limb is accepting weight
Mid-stance	First phase of single support when the body advances over the stance limb ending ahead of the stance limb as weight is transferred to the forefoot
Terminal stance	Last phase of single support ending with opposite initial contact
Pre-swing	Final double support period when the knee rapidly flexes in preparation for swing and weight is shifted to the opposite limb
Initial swing	First third of swing period where maximum knee flexion occurs
Mid-swing	Middle third of swing period where maximum hip flexion occurs, ending with a vertical tibia
Terminal swing	Last third of swing period where final knee extension achieves maximum step length and the limb is properly positioned for weight acceptance

The eight phases described by Dr. Perry are identified in Table 5.2. Notice that all but the first phase (initial contact) represent separate time intervals between 0% and 100% of the gait cycle. Figure 5.12 illustrates the phases of gait in sequence around the unit circle, with stick figures signifying the temporal event delineating the beginning and end of each phase. Phases are shown equally spaced in this figure for clarity; typically, each phase will not be of the same time duration. Initial contact is considered a phase of gait since it marks an important transitional point between swing limb advancement and the challenging task of weight acceptance, although unlike the other phases, it is a single instant in time. The other important transitional period (from stance to swing) occurs during final double support and is known as the phase of *pre-swing*. In terms of temporal events, final double support (and therefore pre-swing) is considered part of the stance period because this interval ends with foot-off. However, in terms of gait phases, this interval also marks the first phase of the swing limb advancement task, highlighting the fact that from a functional standpoint, pre-swing has more to do with preparing the limb for moving forward than supporting the body during stance (24). Also notice that three new temporal events not associated with foot/floor contact have been introduced: heel-off, feet adjacent, and tibia vertical. These terms have been used by Whittle (5) and others (4,38) and are useful to delineate the phases in normal gait; however, there is not yet a consensus among gait investigators if these are the undisputed event markers. For example, Perry acknowledges that a rising heel usually marks the beginning of terminal stance in normal subjects, but in patients with weak ankle plantar flexors, this heel-off may be delayed into pre-swing, which would technically eliminate the terminal stance phase. Dr. Perry prefers to identify the beginning of terminal stance as the point where the body moves ahead of the limb and weight is transferred onto the forefoot (39). Similarly, the event marking the transition between initial swing and mid-swing has been identified as the point where the feet are adjacent (5), when the swing limb is directly under the body (4,38), when swing limb acceleration changes to deceleration in normal gait (36), where the knee begins to extend and the foot clears the ground (39), and mid-swing (4). Fortunately, most investigators agree that the temporal event marking the transition between mid-swing and terminal swing is the point where the tibia is directly vertical. These slight differences in the definition of the exact transition between phases are why some investigators report different phase durations for normal gait. This should not be a concern,

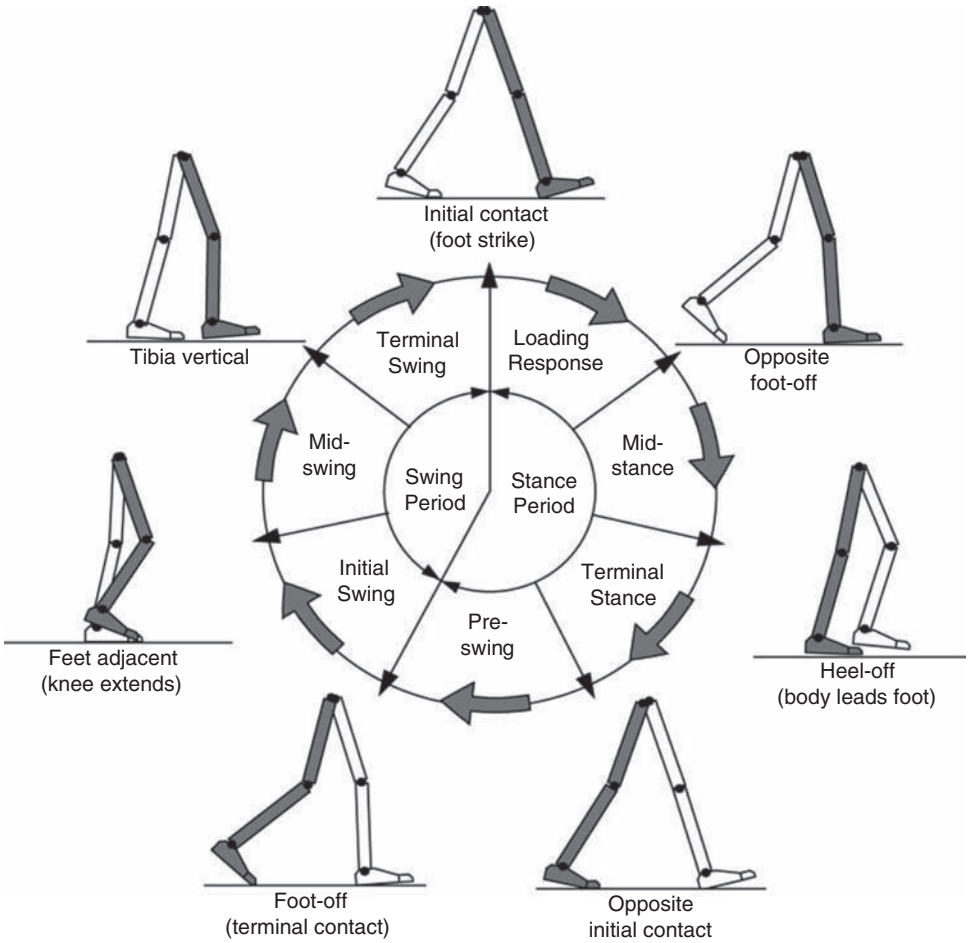

FIGURE 5.12 Typical gait cycle wrapped around a unit circle and subdivided into eight gait phases that have functional significance. The first phase is initial contact, and is equivalent to the temporal event by the same name. The phases are drawn in sequence and are equally spaced for clarity, but do not represent their usual duration. Refer to the text for a complete description of the phases and their functional significance.

however, because determining the exact transition point between phases and comparing phase durations to normal are less important than recognizing that there are distinct phases in gait that can be identified and that certain functional accomplishments must occur in each phase.

CRITICAL EVENTS LINK GAIT IMPAIRMENTS TO POSSIBLE INTERVENTIONS

With the gait cycle now subdivided both temporally and functionally into discrete phases, all that is left is to identify specific joint positions or motions in each phase that directly contribute to the accomplishment of the three functional tasks of weight acceptance, single limb support, and swing limb advancement. Dr. Perry and her colleagues at Rancho Los Amigos Medical Center refer to these specific joint positions, or motions, as *critical events* (39). They have identified 13 critical events over the entire cycle, with one or more critical events in each of the eight phases. Critical events occur at the foot, ankle, knee, or hip, and are largely focused on angular displacements in the sagittal plane. While there are other, more subtle motions occurring in all three anatomical planes, these 13 critical events are considered the most essential to producing a normal walking pattern, typically have the largest displacements, and are most easily observed from the walking subject, with or without the help of recording instruments. The significance of this approach is that once the critical events that are essential to producing a bipedal gait pattern are known, one can use the measures from IGA to determine functional reasons for why a particular critical event is absent, altered, or delayed. Interventions can then be focused on restoring critical events, leading to improved walking performance. The critical events for each phase of gait are shown in Table 5.3, including their relationship to stance and swing periods and each gait task. Notice that critical events and temporal events are quite different. As has been discussed throughout this

TABLE 5.3 RELATIONSHIP BETWEEN PERIODS, TASKS, PHASES, TEMPORAL EVENTS, AND CRITICAL EVENTS DURING THE GAIT CYCLE

STANCE PERIOD				**SWING PERIOD**				
TASKS	**WEIGHT ACCEPTANCE**		**SINGLE LIMB SUPPORT**		**SWING LIMB ADVANCEMENT**			
Phases	Initial contact (0%)	Loading response (0%–12%)	Mid-stance (12%–30%)	Terminal stance (30%–50%)	Pre-swing (50%–62%)	Initial swing (62%–75%)	Mid-swing (75%–87%)	Terminal swing (87%–100%)
Temporal events	Initial contact	B: Initial contact	B: Opposite foot-off	B: Heel-off (body leads foot)	B: Opposite initial contact	B: Foot-off E: Feet adjacent (knee extends)	B: Feet adjacent (knee extends)	B: Tibia vertical
		E: Opposite foot-off	E: Heel-off (body leads foot)		E: Foot-off			E: Initial contact
				E: Opposite initial contact			E: Tibia vertical	
Critical events	• Heel-first initial contact	• Hip stability	• Controlled tibial advancement	• Controlled ankle DF with heel rise	• Passive knee flexion to 40°	• Maximum knee flexion (>60°)	• Max hip flexion (25°)	• Knee extension to neutral
		• Controlled knee flexion for shock absorption		• Trailing limb posture			• DF to neutral	
		• Controlled ankle PF						

Abbreviations: B, beginning; DF, dorsiflexion; E, end; PF, plantar flexion.

chapter, temporal events are moments or instants in time used to delineate periods in the gait cycle, and critical events are important functional components that can be used to identify gait impairments.

CRITICAL EVENTS DURING THE WEIGHT ACCEPTANCE TASK

The two phases associated with weight acceptance, initial contact and loading response, coincide with the period of initial double support, and include four critical events. The first critical event, a heel-first initial contact, must be present if forward momentum is to be preserved (24) and the energy from the falling body COM is to be redirected in the direction of progression (32). It is also necessary to prepare the new support limb for the demands of the next phase. The next three critical events of hip stability, controlled knee flexion for shock absorption, and controlled ankle plantar flexion, must occur during the loading response phase. Hip stability requires dynamic joint stiffness in the sagittal and frontal planes at the hip to prevent unnecessary forward pelvic tilt or increased pelvic obliquity, respectively. This places a high demand on the torque (moment) production ability of the hip extensors and hip abductors of the forward load-bearing limb. This demand is reflected in increased muscle activation (recorded using dynamic EMG) in the gluteus maximus, gluteus medius, and hamstrings. If a patient has weak hip extensors and fails to compensate for this weakness, there will be an increased anterior pelvic tilt and/or forward trunk lean directly associated with the inability of the hip extensors to meet the demand of this critical event. Alternatively, if the subject is successfully compensating for the weakness, they will exhibit a posterior trunk lean to position the whole-body COM behind the hip joint center, thus reducing the torque production demand on the hip extensors. Evidence of either strategy is reflected in recordings of the hip sagittal plane angles (kinematics), the weight line (GRF vector), dynamic EMG of the hip extensors, or by calculating the hip extensor moments and powers (kinetics). Similar strategies are used when there is weakness in the hip abductors, leading to either uncompensated or compensated Trendelenburg's gait patterns in the frontal plane. All can be related to the loss of the critical event of hip stability during the loading response phase.

Controlled knee flexion for shock absorption must occur to prevent unnecessary knee flexion during loading response, which wastes energy and places a higher demand on the quadriceps. Similar to the hip, quadriceps weakness can also be either compensated or uncompensated. Uncompensated quadriceps weakness will present as abnormal or increased knee joint angular displacements in the sagittal plane (increased knee flexion) with possible collapse at the knee. Compensated patterns will display body postures that shift the COM forward (forward trunk lean) so that the GRF vector is closer to or in front of the knee joint, thereby reducing the quadriceps demand (quadriceps avoidance gait). Again, these patterns are reflected in kinematic, kinetic, and dynamic EMG measurements, within the context of meeting the needs of this critical event. Correctly controlled knee flexion during loading response is reflected in the slope and maximum knee flexion value during the first peak of the sagittal plane knee joint angular displacement curve, in the magnitude of the knee extensor moment and power absorption curve, and the EMG activity of different heads of the quadriceps.

The final critical event during loading response is controlled ankle plantar flexion. In this context, "controlled" refers to the ability of the ankle dorsiflexors to eccentrically contract and lower the initially neutral foot carefully to the ground to provide a more stable base of support than can be provided by the calcaneus alone. This action is referred to as the heel or first rocker (40), the first of three important mechanisms that occur at the foot and ankle and facilitate progression of the entire stance limb (31). These three rockers are illustrated in Figure 5.13. If the pretibial muscles have sufficient strength to restrain the rate of foot drop, the action of the first rocker pulls the tibia forward, which in turn is transferred to the femur by the active quadriceps that is attempting to restrain the rate of knee flexion. This is how the energy of the falling body COM is redirected to provide forward progression, an important energy-conserving mechanism often lost when the heel rocker is absent. If the pretibial muscles did not have sufficient strength but the subject was able to achieve a heel-first initial contact, a noticeable foot-slap would occur as the unstable lever at the ankle allows the foot to plantar-flex uncontrollably. This is reflected in a steep descending initial slope on the sagittal plane ankle kinematic curve and the absence of either a dorsiflexor moment or dorsiflexor power absorption on the corresponding sagittal plane kinetic curve (22). Experienced clinicians can also detect this event without all the modern conveniences by simply listening for the sound of the foot-slap! The effects of uncontrolled ankle plantar flexion at loading response are not as easy to detect when the subject fails to achieve a heel-first initial contact, as is the case with foot-flat, forefoot, or equinus initial contact. In this case, kinetic data are helpful, because as the point of foot/floor load bearing moves further in front of the ankle with progressively increasing plantar flexion at initial contact, there is a proportional increase in the magnitude of the incorrect plantar flexor moment during loading response. This plantar flexor moment (via the triceps surae) has the opposite of the desired effect on the knee, as occurs with eccentric contraction of the pretibial muscles when a true heel rocker is present; the knee extends when it should be flexing (41). This reduces the effectiveness of knee shock absorption and may eliminate it completely, increasing bone-on-bone forces at the knee. The amount of shock-absorbing energy transferred to the hip and ankle is reflected in the hip, knee, and ankle powers during this phase, and is useful for describing the potential degree of impairment associated with incorrect ankle

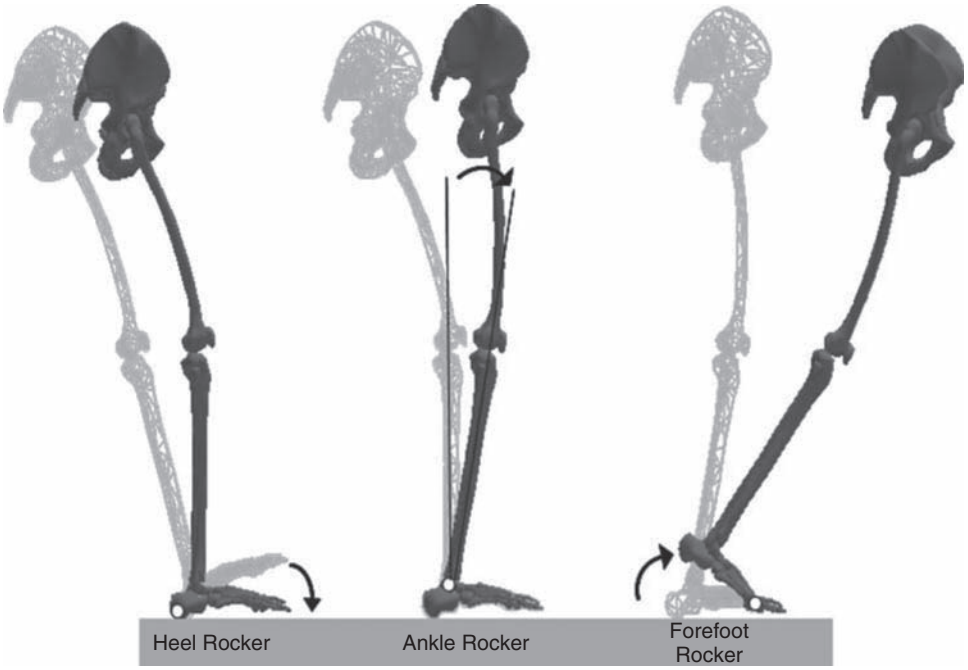

FIGURE 5.13 The three rockers representing normal ankle function in gait: These are the heel or first rocker, the ankle or second rocker, and the forefoot or third rocker. The lighter gray skeleton represents the beginning of each rocker, and the arrows signify the movement that is associated with each. Refer to the text for a full description of these important critical events.

function at loading response. Note that the magnitude of the EMG activity of the pretibial and posterior compartment muscles by themselves do not explain the moments at the ankle, since major force contributors arise from the inertial and gravitational forces that occur during the first rocker. The EMG activity does help sort out if the pattern of motion is due to weakness (pretibial muscles with first rocker present, triceps surae without it), neglect (often seen in traumatic brain injury [TBI]), or poor motor control (seen in CP or cerebrovascular accident [CVA]). This information can assist in determining whether a solid, leaf-spring, hinged, or floor-reaction AFO, Botox injections into the calf musculature, or tendon lengthening or transfer surgery would be the most appropriate intervention to use when the critical event of controlled ankle plantar flexion is abnormal or absent. Table 5.4 summarizes many of the gait measurements that are useful for identifying functional causes for absent or abnormal critical events during the weight acceptance task, and while not exhaustive, can help organize the array of measures available for assessing impairments during this task.

CRITICAL EVENTS DURING THE SINGLE LIMB SUPPORT TASK

As shown in Table 5.3, there are three critical events during the single limb support task. These are controlled tibial advancement during mid-stance, controlled ankle dorsiflexion with heel rise (heel-off) during terminal stance, and a trailing limb posture during terminal stance (39). During the two phases of this task (mid-stance and terminal stance), the responsibility of the stance limb is to simultaneously provide support against gravity without losing balance and contain the forward momentum built up by the contralateral swinging limb. Both of these objectives can be accomplished by controlling tibial advancement in the first half of single support and controlling ankle dorsiflexion in the second half. This will lead to the trailing limb posture (body COM forward of the base of support) necessary to permit a sufficient step length on the opposite side. If, at the end of loading response, the foot has achieved foot-flat, then during mid-stance, the ankle becomes the axis of rotation for the body's forward progression. This is referred to as the ankle or second rocker, and this mechanism continues until maximum dorsiflexion is achieved in terminal stance (Figure 5.13). With the heel and forefoot firmly planted, the tibia can rotate over the talus smoothly under the selective control of the soleus, later assisted by both heads of the gastrocnemius, which simultaneously limits knee extension. The slow-twitch, fatigue-resistant muscle fibers of the soleus are usually well suited to the sustained eccentric contractions required to control tibial advancement. Weakness in the triceps surae, however, results in the tibia advancing too quickly, which prematurely allows the tibia to move past vertical and leads to sustained knee flexion during mid-stance, and premature or excessive dorsiflexion and lack of knee extension at terminal stance. In this circumstance,

TABLE 5.4 GAIT MEASUREMENTS USEFUL FOR IDENTIFYING THE CAUSE OF AN ABSENT OR ABNORMAL CRITICAL EVENT DURING THE WEIGHT ACCEPTANCE TASK

GAIT PHASE	CRITICAL EVENT (ABNORMAL OR ABSENT)	PHYSICAL EXAM	TEMPORAL/DISTANCE MEASURES	KINEMATICS	KINETICS	DYNAMIC EMG
Initial contact	Heel-first initial contact: if absent, also consider mid-swing and terminal swing critical events	Strength • weak ankle DF ROM • tight hamstrings or triceps surae	• Reduced swing period, or reduced single support time on opposite side • Reduced step length	• Hip flexion > or < normal maximum of 30° • Knee flexion > 4° • Ankle not at neutral	• Refer to phases terminal swing or loading response	• Excessive hip extensor or hamstring activity • Reduced or absent ankle DF activity • Premature ankle PF activity
Loading response	Hip stability	Strength • weak hip extensors • weak hip abductors ROM • tight hip flexors, hip flexion contracture • femoral anteversion	• Prolonged initial double support time	• Hip flexion > 30° • Increased pelvic tilt and/or obliquity • Increased hip internal rotation • Pelvic retraction on side of weakness	• Large hip extensor moment with possible initial power absorption	• Excessive hip flexor and hip adductor activity • Decreased hip extensor and abductor activity
	Controlled knee flexion for shock absorption	Strength • weak quadriceps ROM • tight hamstrings or knee flexion contracture	• Prolonged initial double support time	• Knee flexion < 4° or > 20° • Abnormal knee flexion wave	• Initial knee flexor moment with no phase reversal (quad avoidance) • Large knee extensor moment with excessive power absorption	• Excessive knee extensor activity • Increased cocontraction at the knee with prolonged knee flexor activity
	Controlled ankle PF	Strength • weak ankle DF ROM • tight triceps surae or reduced DF • tight hamstrings or knee flexion	• Prolonged initial double support time • Reduced time to foot-flat, with possible foot-slap	• Tibia forward of vertical with ankle in DF • Abnormal first rocker (heel rocker) • Incorrect foot alignment with incorrect foot progression angle	• Large PF moment with high power absorption	• Reduced or absent ankle DF activity • Premature ankle PF activity • Premature tibialis posterior activity

Abbreviations: DF, dorsiflexion; PF, plantar flexion; ROM, range of motion.

a rigid AFO or, in extreme cases of weakness, a floor-reaction AFO can effectively supplement the weak plantar flexors, restore a more normal plantar flexor moment, and control tibial advancement during mid-stance and dorsiflexion during terminal stance. Gage also suggests using a rear-entry, hinged, floor-reaction AFO in these circumstances (12), which permits ankle plantar flexion but resists dorsiflexion in mid-stance and terminal stance.

In normal adults and typically developing children, the forward progression of the body causes the origin of the GRF vector (center of pressure or COP) to move forward to the metatarsal heads, causing the heel to rise at the beginning of terminal stance. Now the axis of rotation for the body's forward progression is the metatarsophalangeal (MTP) joint, giving rise to the forefoot or third rocker (31) (see Figure 5.13). While the first two rockers were constraining forward progression using eccentric plantar flexor contractions, the forefoot rocker is an accelerating rocker, as evidenced by the large ankle plantar flexor moment and transition from power absorption to power generation (36). With the help of strong concentric contraction of the fast-twitch fibers of the gastrocnemius, the ankle is stabilized and continued dorsiflexion in terminal stance is halted. By the end of terminal stance, the ankle is plantar flexing in preparation for initial contact on the other side, which yields the trailing limb posture necessary for maximum step length. When there is plantar flexor weakness, the third rocker is ineffective, which fails to control continued dorsiflexion, allows the knee to prematurely drop into flexion, reduces trailing limb posture, and shortens the opposite side step length. All of these factors reduce overall walking performance. AFOs that store energy in the structure of the orthosis as the ankle dorsiflexes (rigid, leaf-spring, floor-reaction) can provide a plantar flexion assist as the foot is unweighted in early pre-swing, depending on the amount of stiffness and energy storage built into the custom orthotic. This assist can return some of the reduced plantar flexor moment that would occur without orthotic use, and evidence of this can be found in the plantar flexor moment curve comparing orthotic and barefoot conditions.

If, at the beginning of mid-stance, the foot has either not achieved or is past foot-flat (equinus, early heel-off, spring-foot), the normal ankle and forefoot rocker mechanisms may not be effective, and the three critical events of single limb support will not be achieved. In toe–toe gait (equinus) or jump knee gait (forefoot initial contact and excessive knee flexion at loading response, followed by rapid knee extension and ankle plantar flexion in mid-stance), plantar flexors that are tight or have increased tone overly constrain forward tibial advancement in mid-stance and dorsiflexion in terminal stance, leading to excess knee extension and early heel rise. While the mechanism is different from the case of weak plantar flexors, the end result is the same: reduced effectiveness of second and third rockers and inability to achieve the three critical events. In these cases, Botox injections into the triceps surae, tendo-Achilles lengthening, or intramuscular triceps surae lengthening (Strayer procedure) can be effective in restoring second and third rockers, depending on severity. Ankle plantar flexion moments and powers, and dynamic EMG recordings are quite useful in selecting which procedure is most appropriate (22). Another example is crouch gait deformity, where hip and knee contractures combined with weak or overlengthened plantar flexors lead to early heel rise and premature forward advancement of the tibia in mid-stance, and premature and excessive dorsiflexion in terminal stance. In this case, the same impact on the second and third rockers described previously for weak plantar flexors will often occur. Dr. Gage has long been a proponent of performing single-event, multilevel (SEML) soft tissue and bony surgery for this deformity to restore the proper rocker mechanisms and, with the proper orthotics, the plantar flexion/knee extension couple that allows the patient to stand more erect and walk more effectively (12). Other centers have taken a more conservative approach of staging the procedures, which has the advantage of reducing the surgical impact at the time of the procedure, but may cause muscle imbalances at other joints, leading to additional surgeries down the road. In either case, or when nonsurgical interventions are warranted, the goal should be to restore the rocker mechanisms so that the three critical events of single limb support can be realized. Table 5.5 summarizes many of the gait measurements that are useful in identifying causes for absent or abnormal critical events during the single limb support task.

CRITICAL EVENTS DURING THE SWING LIMB ADVANCEMENT TASK

Swing limb advancement is the last task that must be accomplished to complete the gait cycle, and, as shown in Table 5.3, this task contains four phases and six critical events. There are two critical events in pre-swing: passive knee flexion to 40 degrees and rapid ankle plantar flexion. As previously discussed, pre-swing is an important transitional phase that, while still a component of stance period, is functionally more associated with preparing the trailing limb for the swing period to come. Achieving 40 degrees of knee flexion before the foot leaves the ground is essential. This is because once the foot is airborne, the leg acts as a compound pendulum, so further knee flexion is completely dependent on concentric contraction of the hip flexors, including the adductor longus, and the inertia of the lower leg and foot (36). At normal walking speeds, knee flexion during pre-swing requires no active muscle contractions around the knee (passive). It occurs by a complex mechanism that involves continuation of tibial

TABLE 5.5 GAIT MEASUREMENTS USEFUL FOR IDENTIFYING THE CAUSE OF AN ABSENT OR ABNORMAL CRITICAL EVENT DURING THE SINGLE LIMB SUPPORT TASK

GAIT PHASE	CRITICAL EVENT (ABNORMAL OR ABSENT)	PHYSICAL EXAM	TEMPORAL/ DISTANCE MEASURES	KINEMATICS	KINETICS	DYNAMIC EMG
Mid-stance	Controlled tibial advancement	Strength • weak ankle PF (advance too fast) ROM • with equinus, or excessive DF without equinus (advance too fast) • tight triceps surae or hamstrings (advance too slow) Neurologic • triceps surae tone	• With equinus (early heel-off) • Excessive DF without equinus (delayed heel-off) • Reduced single support time • Reduced opposite step length	• Tibia forward of vertical with ankle in excessive DF • Abnormal second rocker (ankle rocker)	• Abnormal slope (too flat) of the PF moment curve	• Prolonged ankle DF activity • Reduced or absent PF activity • Prolonged hip and knee extensor activity
Terminal stance	Controlled ankle DF with heel-off	Strength • weak ankle PF (DF too fast) ROM • excessive DF or reduced PF (DF too fast) • tight or increased tone in ankle PF (slow DF, early heel-off)	• With equinus (early heel-off) • Excessive DF without equinus (delayed heel-off) • Reduced single support time • Reduced opposite step length	• Abnormal second rocker (ankle rocker) • Excessive DF • Premature knee flexion	• Abnormal slope (too flat) of the PF moment curve • Reduced or absent PF power absorption	• Prolonged ankle DF activity • Reduced or absent PF, inverter, and evertor activity
	Trailing limb posture	Strength • weak ankle PF • weak opposite hip abductor ROM • excessive DF or reduced PF • tight hamstrings or tight hip flexors	• Reduced single support time • Reduced opposite step length	• Excessive hip flexion • Excessive knee flexion	• Prolonged hip external moment or delayed/absent hip flexor moment • Prolonged knee external moment or delayed/absent knee flexor moment	• Any activity in the hip or knee flexors, and hip or knee extensors (lack of passive stability)

Abbreviations: DF, dorsiflexion; PF, plantar flexion; ROM, range of motion.

advancement as the forefoot rocker continues from terminal stance; unloading of the limb as weight is transferred to the new stance limb; continued concentric contraction of the triceps surae, which produces rapid plantar flexion that propels the knee joint in front of the GRF vector, and concentric contraction of the adductor longus to initially accelerate the thigh forward (31).

All of these actions push the GRF vector so far behind the knee that it collapses in the absence of an equalizing knee extension moment produced by the quadriceps that normally are silent during pre-swing. So weakness in the plantar flexors, hip flexors, or adductor longus has an adverse effect on achieving the necessary knee flexion. Since a trailing limb posture with hip extension to 10 degrees past neutral amplifies the effect of the third rocker to shift the tibia forward, hip flexion contracture can also reduce the ability of the knee to passively flex to 40 degrees, despite the fact that such a contracture often prevents the knee from fully extending at terminal stance. Furthermore, it is interesting that because of the hip extensor component of the biarticulate hamstrings, inappropriate activation or tightness of these open-chain knee flexors can inhibit passive knee flexion in pre-swing by resisting the hip flexors and adductor longus as they attempt to accelerate the thigh. Problems with this critical event can be identified from the sagittal plane knee kinematics, the hip moments and powers, and the hip knee and ankle dynamic EMG. Problems with rapid plantar flexion, which is necessary to produce sufficient knee flexion in pre-swing, are also evident from the ankle kinematics, kinetics, and dynamic EMG. Since the critical events in this phase are so dependent upon concentric contraction and power generation at the hip and ankle, interventions to replace hip flexor and ankle plantar flexor strength are somewhat limited to AFOs that can return plantar flexion moment in pre-swing or enhance the third rocker, or stretching, lengthening, or weakening muscles that may be inhibiting hip flexion using neurolytic agents or surgical procedures (42).

In initial swing, the only critical event is to achieve maximum knee flexion of at least 60 degrees. If 40 degrees of knee flexion has been achieved at the end of pre-swing and the hip flexors and adductor longus stop firing before the end of initial swing, then in the absence of inappropriate quadriceps or hamstring activity, sufficient knee flexion should occur naturally during this phase. The point of maximum knee flexion must occur before the end of initial swing (not during mid-swing or terminal swing), since this is the point where the swinging limb must be at its shortest functional length to successfully clear the ground. The ankle dorsiflexors (pretibials) are firing concentrically at this time to bring the foot from its point of maximum plantar flexion at the end of pre-swing to at least neutral by the end of initial swing so that toe clearance can be ensured in mid-swing. In this phase, kinematics can be used to quantify the progress of the swinging limb, joint moments in the sagittal plane should be near zero, and dynamic EMG can be used to identify inappropriate muscle firing. Of particular interest in this phase is the activity of the rectus femoris. This biarticulate muscle initially is active in late pre-swing to assist with accelerating the thigh forward. At the moment the foot leaves the ground, continued activity of the rectus femoris may assist with hip flexion, but may have the negative consequence of providing open-chain knee extension through the patellar ligament. Since it has been shown that the brain uses the rectus femoris to accelerate the thigh to selectively control step length and cadence during swing (36), if hip flexor angular velocity is initially slow and 40 degrees of knee flexion was not achieved at the moment of foot-off, the rectus femoris may increase its activation in initial swing to serve as an auxiliary hip flexor. This abnormal compensatory activity of the rectus femoris is in an effort to produce increased thigh acceleration, but because of its biarticular structure, it yields the negative effect of producing a larger knee extension moment, exacerbating the problem of insufficient knee flexion in swing. Whether the rectus femoris is firing as a compensatory mechanism or because of incorrect motor control associated with upper motor neuron injury, if it continues to be active at the end of initial swing, it may contribute to the abnormality known as stiff-knee gait, the common name given to the gait abnormality of insufficient knee flexion in swing period. If kinematic and electromyographic evidence (reduced knee flexion peak and/or slope and prolonged activation) exists, then a rectus femoris transfer to the semimembranosus or sartorius may harness this inappropriate activity, or more likely, prevent concentric knee extension from limiting peak knee flexion in swing and thereby disrupt swing limb advancement. The rectus femoris transfer for the treatment of stiff-knee gait is a surgical procedure that was conceived as a direct result of using IGA techniques (43) and has been supported by a series of laboratory investigations and long-term follow-up (44,45,46,47,48,49). It is now considered the standard of care for the treatment of stiff-knee gait when evidence from IGA confirms that the rectus femoris is responsible for failure to achieve the critical event of obtaining maximum knee flexion of 60 degrees during initial swing.

The two critical events during mid-swing are related to achieving toe clearance as the limb swings through the lowest point in its arc of motion and it is at greatest risk to inadvertently make contact with the ground. These critical events are maximum hip flexion to 30 degrees and neutral ankle dorsiflexion. The hamstrings may fire near the end of mid-swing to begin decelerating the forward movement of the thigh or to slow down the cadence, but generally these muscles should be silent until terminal swing. Note that after the swinging limb clears the floor, there is typically no further need for hip flexion, and additional hip flexion will only decrease the likelihood of achieving the final critical event during terminal swing: knee extension to neutral. Children with CP and other patients with upper motor neuron disease have a difficult

time motor programming the previous two phases of motor activity, and often display excessive knee and hip flexion with peak values later than normal during mid-swing. Kinematics and dynamic EMG can help identify these incorrect patterns, and the usual procedures of stretching, lengthening, or injecting the offending muscles may be useful if they can permit the critical events in pre-swing and initial swing to occur. The critical event of neutral dorsiflexion is usually the responsibility of the ankle dorsiflexors, which typically initiate concentric activity in pre-swing. The pretibial muscles generally reduce their activity in this phase since they no longer need to concentrically contract from the plantar-flexed position and are only needed to hold the foot against gravity. If they are weak or if there is an upper motor neuron injury preventing normal motor control, foot drop will result, which will adversely affect toe clearance and necessitate compensatory mechanisms of circumduction at the hip, increased ipsilateral pelvic obliquity (hip hiking) or contralateral early heel-off (vaulting). The most common solution to this problem is to prescribe an AFO with a plantar flexion stop to hold the foot in the correct position throughout the swing period. In children and adults with TBI, dynamic EMG can be used to determine if the lack of dorsiflexion during swing is related to incorrect cortical control or an inability to correctly motor-plan the dorsiflexion activity. In the latter, training with biofeedback of muscle contraction may improve foot clearance during swing and eliminate the requirement of using an AFO.

The final critical event in the gait cycle is knee extension to neutral during terminal swing. This represents the last opportunity of the swinging limb to reposition the foot prior to weight acceptance, and if this critical event is achieved, a sufficiently long step length will result. In typically developing children and normal adults, hamstring activity will begin during this phase to decelerate the swinging lower limb so that a small amount of knee flexion (<4 degrees) is present at initial contact. In some cases during slow speed walking, the quadriceps will contract concentrically to assist with final knee extension, but in general, this is unnecessary if the subject displays proper motor control. In addition to kinematic recordings to confirm final position and dynamic EMG recordings to determine if there is excessive hamstring activity, large hip extensor and knee flexor moments with power absorption just prior to initial contact may be indicators that the hamstrings are tight or display increased tone. Lack of full knee flexion in terminal swing is one of the most common gait abnormalities in CP (50), and may require neurolytic injections or surgical lengthening of the hamstrings and adductors when the physical exam and gait measurements provide appropriate evidence. As in the previous sections, a table has been prepared to summarize the gait measurements that are useful in detecting abnormalities in the six critical events associated with swing limb advancement (Table 5.6). If all six of these critical events are performed successfully, the limb will be properly prepared for initial contact and ready to begin the cycle of gait events again.

The 8 phases and 13 critical events described in the previous sections and summarized in Table 5.3 complete the functional decomposition of the gait cycle. Armed with this analysis framework, the pediatric physiatrist can utilize IGA measurements, radiographs, and a comprehensive physical exam to better understand the complex interactions of body structure and function that produce abnormal gait patterns in the pediatric patient. We will conclude this chapter with a case study illustrating the use of this framework in a subject with a common pediatric diagnosis but a unique and challenging gait abnormality.

MOTION ANALYSIS CASE STUDY—SPASTIC DIPLEGIA

CP is the most common childhood disorder with new cases occurring in 2 to 2.5 of every 1,000 live births (51,52). The term CP is used to describe a group of chronic, but nonprogressive conditions affecting body movement and muscle coordination. It is caused by damage or injury to one or more specific areas of the brain, and usually occurs during fetal development, before, during, or shortly after birth, or during infancy (52). Among people with the four main types of CP (spastic, athetoid, ataxic, and mixed), more than 70% are classified as having spastic CP, as does the subject of this case study.

While the brain injury is not progressive and the primary impairments (increased muscle tone and spasticity) rarely change without treatment or surgery, the nature and severity of the disability can still worsen over time as the child matures (53). Mild forms result in slight limitations in walking or use of the hands; more severe cases can disrupt all coordinated movement and make everyday activities such as speech, walking, and eating virtually impossible to perform independently. Children with CP are especially at risk for reduced function as they transition to adulthood (54,55) since the secondary musculoskeletal impairments associated with the disorder (weakness, decreased joint ROM, pain and skeletal deformities) are more pronounced as they achieve their adult size and weight, and the demands of normal physical activity exceed their ability to perform them.

Since the original injury can affect any portion of the growing brain at any stage of development, the combination and severity of symptoms are highly variable, and no two children with CP display the same functional or cognitive status or ambulatory abilities. Deciding which intervention is most appropriate to reduce impairments and improve function in children with CP requires a comprehensive care assessment that should begin in a multispecialty clinic, and includes a physical examination and IGA for children at Gross Motor Function Classification System (GMFCS) levels 3 or higher.

TABLE 5.6 GAIT MEASUREMENTS USEFUL FOR IDENTIFYING THE CAUSE OF AN ABSENT OR ABNORMAL CRITICAL EVENT DURING THE SWING LIMB ADVANCEMENT TASK

GAIT PHASE	CRITICAL EVENT (ABNORMAL OR ABSENT)	PHYSICAL EXAM	TEMPORAL/ DISTANCE MEASURES	KINEMATICS	KINETICS	DYNAMIC EMG
Pre-swing	Passive knee flexion to 40°	Strength • Weak hip flexors ROM • Tight hip flexors (limit hip extension)	• Delayed foot-off • Prolonged stance period	• Insufficient hip extension at beginning of pre-swing (<10°) • Incorrect slope, second knee flexion peak	• Reduced peak hip flexor moment and power generation • Reduced knee extensor power absorption	• Reduced or absent adductor longus or hip flexor activity • Abnormal hamstring activity
	Rapid ankle PF	Strength • Weak ankle PF ROM • Excessive DF or reduced PF	• Delayed foot-off • Prolonged stance period	• Abnormal third rocker (heel rocker) • Excessive DF early or reduced PF late in pre-swing	• Reduced peak PF moment at start of pre-swing • Reduced peak PF power generation	• Prolonged ankle PF activity into late pre-swing • Inappropriate cocontraction of ankle PF, DF
Initial swing	Maximum knee flexion (>60°)	Strength • Weak hip flexors ROM • Tight rectus femoris	• Asymmetric stance/ swing ratio • High variability in swing period or step length	• Reduced or delayed peak knee flexion in swing • Incorrect slope in knee flexion wave • Slow ankle DF	• Nonzero moments and powers at hip, knee, and ankle by the end of initial swing	• Reduced or absent adductor longus and hip flexor activity • Reduced or absent ankle DF activity
Mid-swing	Maximum hip flexion to 30°	Strength • Weak hip flexors ROM • Tight hamstrings	• Asymmetric stance/ swing ratio • High variability in swing period or step length	• Maximum hip flexion late or in next phase • Abnormal pelvic obliquity (hip hike) or hip abduction (circumduction)	• Nonzero moments and powers at the hip • Premature knee flexor power absorption	• Prolonged rectus femoris activity • Premature hamstring activity
	Neutral DF	Strength • Weak ankle DF ROM • Tight ankle PF	• Asymmetric stance/ swing ratio • High variability in swing period	• Sagittal plane ankle curve with excess DF or PF	• Nonzero moments and powers at the ankle	• Reduced or absent ankle DF activity with foot drop • Any PF activity
Terminal swing	Knee extension to neutral	Strength • Weak hip flexors or knee extensors ROM • Tight hamstrings Neurologic • Hamstrings tone	• Asymmetric stance/ swing ratio • High variability in swing period or step length	• Knee flexion > 4° before initial contact • Compensatory and excess hip flexion for limb clearance	• Large hip extensor and knee flexor moment with power absorption just before initial contact	• Excessive knee flexor activity • Reduced or absent ankle DF activity

Abbreviations: DF, dorsiflexion; PF, plantar flexion; ROM, range of motion.

"LD" is a nonverbal 13.5-year-old male with spastic diplegia and developmental delays. His mother had an uncomplicated pregnancy and he was born full-term, but in his first year of life he demonstrated delayed developmental milestones and did not start walking until age 6. He is also hearing-impaired with cognitive, behavioral, and oral motor dysfunction. Previous treatments included oral baclofen, which had little effect on gait performance, and bilateral hinged AFOs. He had no neurologic or orthopedic surgical procedures performed prior to his visit to the CP clinic at our institution, after which LD was referred to our motion laboratory for IGA. His family reported an increased incidence of tripping and falling over the previous 15 months with fast walking and a perceived reduction in overall gait performance. They also reported that the left leg was now turning out more than in the past.

The physical examination performed on the day of the gait analysis measured LD's height as 165 cm, his weight as 63 kg, and he had equal leg lengths. No fixed joint contractures were found, but he showed a popliteal angle of -65 degrees bilaterally, consistent with hamstring tightness. We measured a thigh-foot angle of 35 degrees external on the left, 25 degrees external on the right, slight hindfoot valgus, moderate forefoot abduction, and moderate pes planus bilaterally. Ely and Thomas tests were normal, and there was no appreciable spasticity (1 on the Ashworth scale) in the hamstrings, quadriceps, peroneals, tibialis posterior, toe flexors, or triceps surae bilaterally. Strength information from manual muscle test of the major muscle groups was inconclusive due to the inability of the subject to perform an isolated muscle contraction and his difficulty understanding instructions due to cognitive limitations. However, the therapist performing the physical examination reported that most muscle groups should be at least in the range of 3 to 4 by observing other functional activities and by noting that the subject is an independent, limited community ambulator.

Radiographs taken at the time of the analysis showed slight adduction of the proximal femurs but no sign of femoral head uncovering and otherwise normal hip joints bilaterally. Standing anterior/posterior (A/P) and lateral radiographs of the foot showed forefoot abduction, uncovering of the talus, mid-foot collapse, and a reduced height of the medial longitudinal arch (see Figure 5.14). This was consistent with evidence of increased pressure at the navicular during the physical exam and redness caused by the orthotics in the same area.

Observational gait analysis using slow-motion, high-definition video recordings while the subject walked barefoot in the laboratory showed mild crouch gait deformity during stance period, a stiff-knee gait pattern during the initial swing period, reduced peak knee flexion in mid-swing, insufficient knee extension during terminal swing, and a reduced dynamic knee ROM throughout the gait cycle, all observed bilaterally. He also showed absence of a trailing limb posture due to limitations in hip extension and knee extension at terminal stance, contributing to his mild crouch gait. LD shows a foot-flat initial contact bilaterally, with no sign of a heel (first) rocker (see Figure 5.13). At the ankle, LD showed premature tibial advancement during loading response and mid-stance with delayed heel-off bilaterally. In the frontal plane, LD demonstrated moderate lateral trunk lean during loading response on both sides consistent with a compensated Trendelenburg's gait pattern, suggesting weakness of the hip abductors.

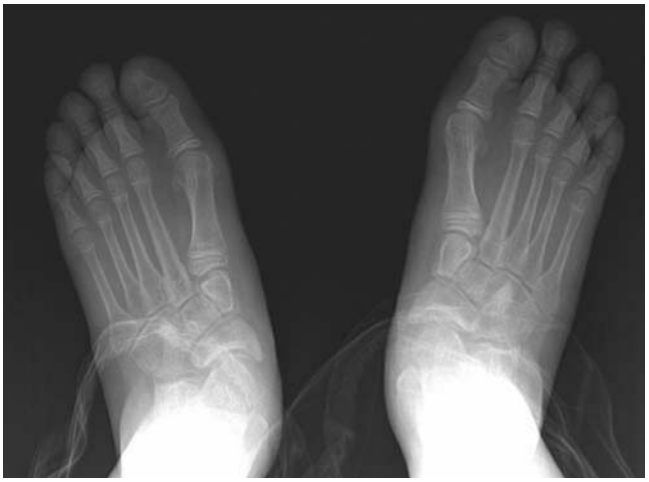

FIGURE 5.14 Bilateral A/P radiograph of the feet of the subject described in case study. This radiograph is commonly required when the subject presents with pes planus, to better understand the structural alignment of the foot.

The IGA included temporal–spatial measures, 3D kinematics, 3D kinetics, and dynamic EMG recorded from six muscles bilaterally using bipolar surface electrodes. Because of the report of pes planus, a plantar pressure recording was included to document the existence of excessive pressure in any area of the foot. The temporal–spatial recordings showed an average cadence of 103 steps/minute (88% normal) and an average walking speed of 51 meters/minute (65% normal). The left side average step length was 0.54 meters (80% normal) and the right side was slightly less at 0.47 meters (70% normal). There were no appreciable differences in gait symmetry or timing of gait events between the left and right sides, with the exception of a slightly longer single limb support time on the left that was not considered clinically significant.

Kinematic, kinetic, and dynamic EMG data from both legs for the barefoot trial are shown in Figures 5.15, 5.16, and 5.17, respectively. On the kinematic and kinetic curves, the right side is represented by a solid line and the left side uses a dashed line, and for comparison, a gray (shaded) band is included on each graph representing the ensemble averages from our typically developing child

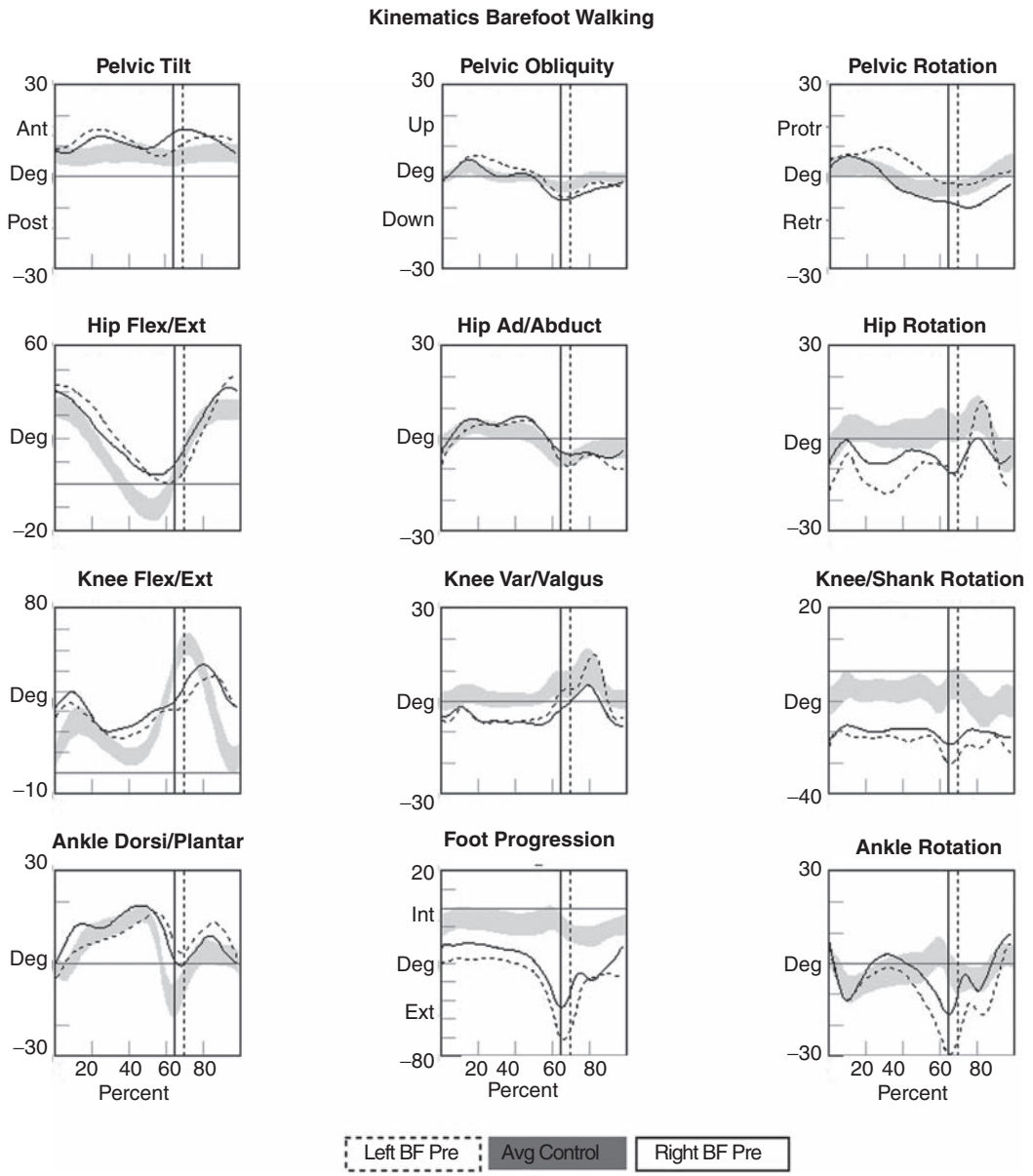

FIGURE 5.15 Three-dimensional kinematic graphs constructed from a representative trial from the instrumented gait analysis of the 13.5-year-old case-study subject "LD." The solid line describes the right side, the dashed line shows the left side, and the gray band (shaded) is from the age-matched normal database collected in the laboratory and used as a reference.

database for this age group. The gray bands correspond to +/-1 standard deviation across the ensemble average for that graph. For the EMG data shown in Figure 5.17, the right side is darker, the left side is lighter, and the black bar at the bottom of each graph is a normal timing reference.

There are a variety of ways to review these data systematically, including evaluating each joint or segment in sequence starting either proximally or distally, evaluating all graphs for a particular data type first and then moving on to the other categories, or reviewing all data for a particular phase of gait and then advancing to the next phase until the cycle is completed. Which of these procedures to follow is a matter of personal preference, and is sometimes dictated by the complexity of the case, but for the novice, it is a good idea to consistently follow the same procedure or review sequence until he or she is comfortable recognizing the significance of each graph individually. We typically start with the kinematic graphs and work distally from the pelvis, scanning the graphs across all phases of the gait cycle to identify deviations from the normal reference. Our goal is to find evidence to support the observational findings described previously, or detect subtle deviations in the movement pattern that were not obvious without quantification. We focus first on the portions of the curve that have the largest deviation from the reference data and then attempt to describe these deviations in the context of the 8 gait phases and 13 critical events described previously and summarized in

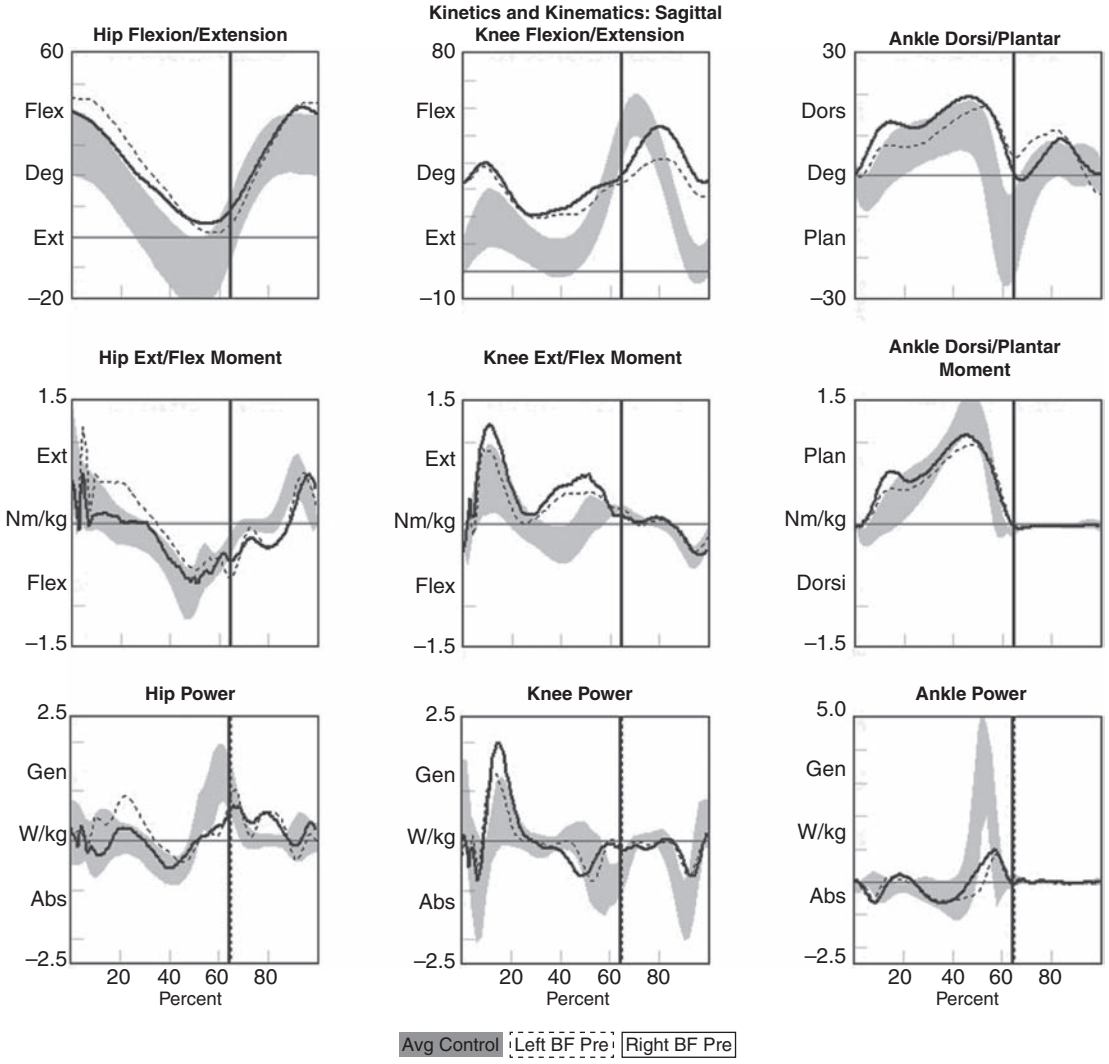

FIGURE 5.16 Three-dimensional graphs of sagittal plane kinematics, sagittal plane joint moments, and total joint power for the hip, knee, and ankle constructed from a representative trial from the instrumented gait analysis of the 13.5-year-old case-study subject "LD." The solid line describes the right side, the dashed line shows the left side, and the gray (shaded) band is from the age-matched normal database collected in the laboratory and used as a reference.

Table 5.3. As needed, we jump to the subject's kinetic and EMG data for additional evidence to explain the absence of a critical event at a specific phase of the gait cycle, and using all of the evidence gathered in the analysis, develop a logical rationale for the subject's unique gait pattern or abnormality.

In LD's case, we see evidence of slightly increased anterior pelvic tilt starting during loading response and reaching a peak of approximately 18 degrees at midstance on the left and 14 degrees on the right (Figure 5.15, first row). This gives rise to a pattern often seen in diplegia called a "double bump" as the pelvis tilts slightly forward during weight acceptance on each side. It is often associated with weakness of the hip extensors and lack of shock absorption more distally, and can be attributed to reduced performance in the critical event of hip stability during loading response. Another cause could be tight hip flexors, but this is unlikely given that the Thomas test from the physical examination was negative. The existence of the second bump in the pattern during pre-swing and initial swing comes from the same mechanism occurring at loading response on the contralateral side. It is reflected in the ipsilateral pelvic tilt because the pelvis is, of course, a single segment and the graphs of each hemipelvis section are 180 degrees out of phase. Pelvic obliquity and pelvic rotation are near normal bilaterally until foot-off and the beginning of initial swing, when the right hemipelvis drops and retracts slightly compared to the normal reference and the left side. This suggests that the compensated Trendelenburg's gait pattern observed is not completely effective at maintaining appropriate pelvic position on the right side during initial swing, possibly due to weaker hip abductors on the left side during loading response. All of these compensations can be

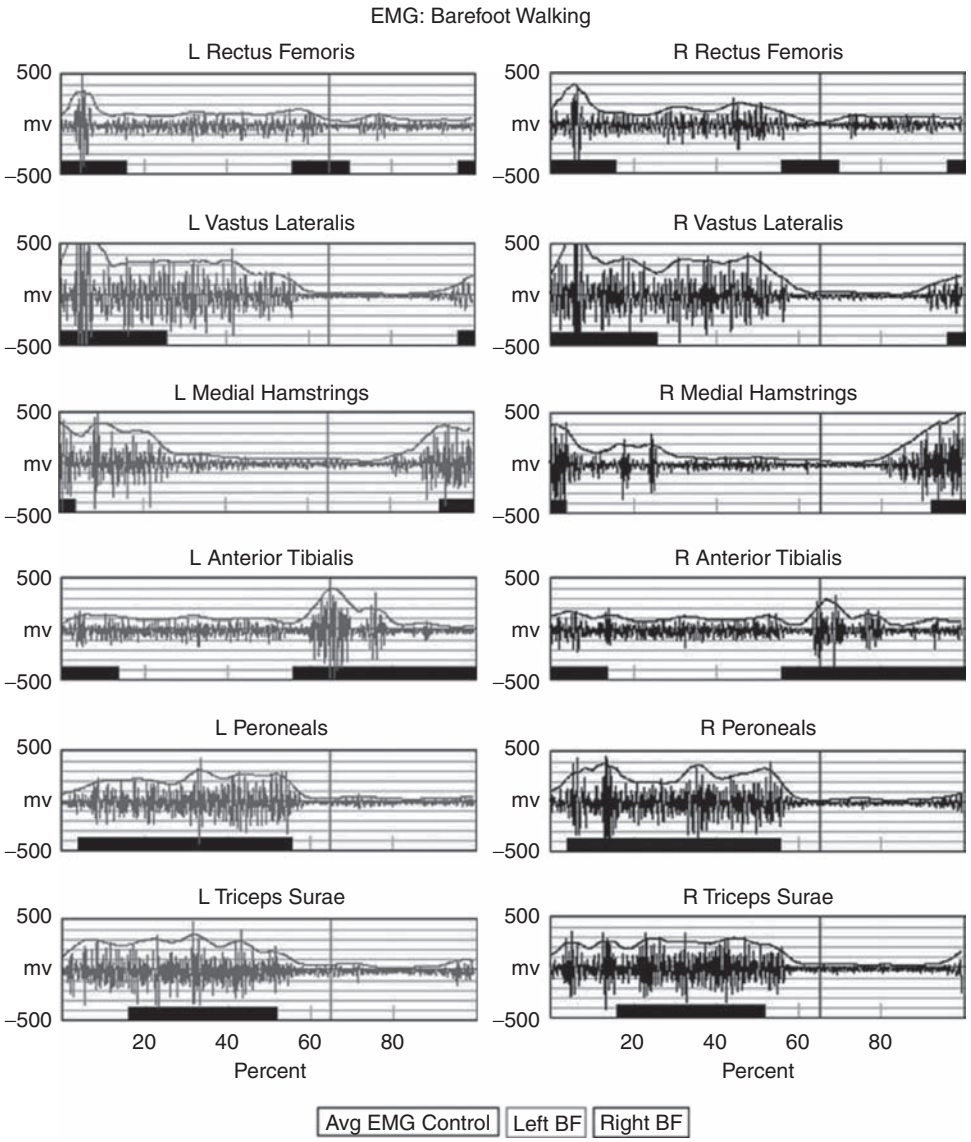

FIGURE 5.17 Filtered and time-normalized EMG for 12 muscles of the lower extremity for a representative trial from the instrumented gait analysis of the 13.5-year-old case-study subject "LD." The black bars at the bottom of each graph are constructed from published normal EMG activations and are used as reference values. The smooth curve above the EMG activation is a processed EMG signal obtained by rectifying and integrating the raw EMG.

attributed to difficulty in achieving the critical event of hip stability during loading response and are evidence of proximal weakness during the task of weight acceptance.

Moving to the hip joint, LD shows increased hip flexion during loading response, decreased hip extension during terminal stance and pre-swing, and increased hip flexion at terminal swing bilaterally. Notice that the shape and range of motion for the hip flexion curve is virtually the same as the average normal curve, except that it is shifted up toward increased flexion by about 10 degrees. This is approximately the same amount that the corresponding anterior pelvic tilt curve is offset from its normal value. These two graphs are always coupled since hip joint angles are calculated relative to the pelvis, the more proximal segment. The lack of hip extension at terminal stance is the most significant limitation here, since it negatively affects the ability to achieve a trailing limb posture during terminal stance, which is essential to achieving maximum stride length on the contralateral side. Moving to Figure 5.16 and the sagittal plane kinetics at the hip joint, we see no significant deficits in the hip moment curve bilaterally, but a reduced hip power generation at pre-swing, approximately 70% normal on the left and 50% normal on the right. Since sufficient power generation at the hip is necessary to achieve the critical event of passive knee flexion to 40 degrees during pre-swing,

and is also a necessary precursor to accomplish the task of swing limb advancement (50), reduced power generation at the hip may contribute to LD's increased incidence of tripping when trying to walk at higher speeds.

Returning to Figure 5.15, the transverse plane motion at the hip shows near-normal hip rotation on the right side, but increased hip external rotation of approximately 10 to 15 degrees on the left side. This suggests that some of the reported external foot position on the left can be attributed to the hip. Please note that because we have a rotational deformity it is often more systematic to review all the rotational curves from proximal to distal to gauge which segments or joints are most contributing to the foot position recorded in the foot progression angle curve. Looking distally down the kinematic chain, we see additional contribution to the final foot progression angle occurring at the knee (distal shank rotation, left approximately 20–25 degrees external, right approximately 10–15 degrees) and to a much lesser extent at the ankle bilaterally, yielding a final foot progression angle of approximately 40 degrees on the left and 20 degrees on the right during mid-stance and terminal stance. We use the term "distal shank rotation" here rather than knee rotation to highlight that the recording includes the external "twist" of the tibia or tibial torsion in the graph rather than just the amount of rotation occurring between the thigh and shank segment. The modified Helen Hayes marker set used to produce these curves assumes that the ankle joint axes and knee joint axes are offset in the transverse plane by the amount of the tibial torsion and normally would not include this offset. We prefer to include the tibial torsion in this curve to better understand the contribution of tibial torsion to the overall foot progression angle, and therefore, call it the distal shank rotation to avoid confusion. It is good practice to have a clear understanding of how the limb-segment model is calculating a particular quantity before utilizing it for clinical decision making, and this curve in particular is frequently affected by vague or unstated model assumptions.

To conclude the rotational assessment, we see a large peak in the external foot progression angle (left approximately 70 degrees external, right 50 degrees external) that corresponds to a lateral whip of the foot at foot-off, most likely as a compensatory mechanism to help with limb advancement. This large external foot progression angle with the left, about 20 degrees greater than right, is consistent with the parents' description, and from the kinematic analysis, can be attributed to both the thigh and shank on the left, and from compensatory mechanisms at the foot and ankle bilaterally.

The analysis now moves back to the knee, where some of the most significant gait deviations exist. In Figure 5.15, the bilateral knee flexion/extension curves show increased knee flexion (relative to the normal reference) during loading response, decreased knee extension during terminal stance, decreased and delayed peak knee flexion during initial swing (left more severe), and increased knee flexion during terminal swing and initial contact. This has the appearance of compressing the knee sagittal plane curve into the middle range of the normal reference, with a shallow rising slope from mid-stance through initial swing (left = 52 degrees/second, right = 65 degrees/second, normal = 240 degrees/second), and decreased dynamic range at the knee over the entire gait cycle (left = 24 degrees, right = 31 degrees, normal = 60–70 degrees). The existence of swing period gait deviations at the knee prevents the most important critical event in swing from being accomplished: achieving maximum knee flexion of at least 60 degrees. Their presence also provides evidence of a bilateral stiff-knee gait pattern that, as previously mentioned, adversely affects the task of swing limb advancement. But when taken together, this combination of excessive knee flexion in stance and insufficient knee flexion in swing has the effect of disrupting all other critical events associated with normal knee function, including controlled knee flexion during loading response, achieving trailing limb posture during terminal stance, passive knee flexion to 40 degrees during pre-swing, and finally reaching full knee extension during terminal swing. With this many critical events absent, altered, or delayed, all three fundamental gait tasks are compromised. Therefore, in order to see any significant improvement in walking ability, these critical events need to be restored, which by necessity prioritizes any intervention directly affecting knee ROM during the gait cycle. To find support for specific interventions, we return to the kinetic and EMG recordings shown in Figures 5.16 and 5.17, respectively. In the top row of Figure 5.17, we see that both the left and right rectus femoris EMG recordings show muscle activation beginning late in initial swing and continuing until terminal swing, with a small peak in initial swing slightly before peak knee flexion. This abnormal EMG activity in combination with insufficient peak knee flexion in initial swing and a shallow slope of the knee curve during pre-swing provide strong evidence to support the use of a rectus femoris transfer procedure bilaterally (46–49). When successful, this procedure can improve both the peak knee flexion in swing and the slope of the knee flexion wave starting in pre-swing, addressing two missing critical events at the knee. To address the other affected critical events, we must review the kinetics and EMG recordings during initial contact, loading response, mid-stance, and terminal swing phases. With greater than 20 degrees of knee flexion throughout the stance period, there is a significant force demand on the knee extensors during weight acceptance and single limb support. Evidence of this can be found in the large knee extensor moments that persist beyond loading response well into terminal stance, shown in the middle graph of Figure 5.16, and the prolonged stance phase EMG activity of the vastus lateralis and rectus femoris shown in Figure 5.17. These findings are consistent with a mild crouch gait deformity, set up by the limitation in knee extension at terminal swing.

While LD is able to overcome this biomechanically disadvantaged position and maintain an upright posture at this time, as he matures and grows heavier, any increase in knee flexion during stance may increase the demand to a level greater than he can withstand, which would severely limit his overall gait performance. It is reasonable here to consider the more aggressive surgical procedures that have been shown to improve knee function in cases of persistent crouch gait, namely a knee extension osteotomy to reduce knee flexion contracture and patellar advancement to treat patella alta and improve the function of the quadriceps (56). Since LD showed no significant knee flexion contracture on physical examination or radiographic signs of patella alta, and the crouch deformity was considered mild since he could achieve 20 degrees of knee flexion at terminal stance, these surgical procedures were deemed unnecessary at this time. However, since there was evidence of tight hamstrings on physical examination (popliteal angles of -65 degrees) and the EMG recording of the medial hamstrings (third row, Figure 5.17) showed premature onset in mid-swing, the team felt hamstring lengthening procedures would be appropriate. Nonsurgical techniques such as phenol injections to the hamstrings could be considered here, but with strong evidence for rectus femoris transfer and the ease of performing a hamstring lengthening at the same time as the rectus procedure, the surgical path seemed most appropriate for this patient. Furthermore, the combination of these two procedures has the best chance of restoring all missing critical events at the knee in the shortest amount of time to prevent continued progression of the crouch gait deformity as LD grows larger through adolescence.

To complete the IGA, we move distally once more to the remaining graphs describing the ankle and foot. The sagittal plane ankle kinematics graph in the lower-left corner of Figure 5.15 provides evidence of what was seen during the observational analysis: increased dorsiflexion at initial contact and no sign of a first (heel) rocker during loading response. This is consistent with the foot-flat initial contact observed, and is shown on the kinematic graph as an increasing slope in the first 10% of the gait cycle starting at 5 degrees of dorsiflexion, rather than a decreasing slope starting from a near-neutral ankle position for the normal reference. The right side shows increased dorsiflexion continuing into mid-stance, with a peak at about 15% of the gait cycle, after which the dorsiflexion stabilizes and then increases at a more normal rate (slope of the ankle curve) near the upper extreme of the normal reference until the beginning of terminal stance. After beginning in a dorsiflexed position at initial contact, the left side dorsiflexion increases at a normal rate, tracking closely the slope of the reference value and providing evidence of a near-normal second (ankle) rocker. Following peak dorsiflexion in terminal stance, the period of rapid ankle plantar flexion during pre-swing begins, which is associated with the third (forefoot) rocker. Unfortunately, maximum plantar flexion stops at a joint angle of approximately 8 degrees dorsiflexion on the left and 2 degrees dorsiflexion on the right—clearly insufficient compared to the normal reference. The ankle then maintains a dorsiflexed position throughout swing period bilaterally.

Considering these elements together and describing them in terms of fundamental gait tasks and critical events, we begin to see a clear picture of the impact of these gait deviations at the ankle. First, we have evidence that during weight acceptance LD is missing a heel-first initial contact and controlled ankle plantar flexion (first rocker) bilaterally. Second, during single limb support, controlled tibial advancement (second rocker) is altered on the right and controlled ankle dorsiflexion with heel rise is delayed bilaterally. Finally, while starting the task of swing limb advancement, rapid ankle plantar flexion (third rocker) in pre-swing is reduced bilaterally. Fortunately, LD does maintain sufficient dorsiflexion in mid-swing to clear his foot so as not to compound the lack of knee flexion and stiff-knee pattern already affecting swing limb advancement. As with the analysis at the knee, failure to achieve these critical events at the ankle represents significant gait dysfunction and must be addressed. Additional insight can be obtained from the kinetic graphs on the rightmost column of Figure 5.16 and the EMG recordings in the lower three rows of Figure 5.17. The combination of foot-flat initial contact, increased knee flexion, and increased ankle dorsiflexion during loading response places a greater than normal demand (external moment) on the ankle plantar flexors. They respond by increasing the net ankle plantar flexion moment (internal moment, shown in the middle right graph, Figure 5.16) during loading response and the early portion of mid-stance. This is most likely a compensatory response to the external demand, and is accomplished by prematurely activating the peroneals and triceps surae during terminal swing, initial contact, and loading response. Notice that the peak in right ankle dorsiflexion at approximately 15% of the gait cycle is accompanied by a peak in the plantar flexion moment and just preceded by a small peak of ankle plantar flexor power absorption, shown in the lower-right graph of Figure 5.16. The ankle power curve then reverses to generate a small amount of power at the point in the cycle (mid-stance) when continued ankle dorsiflexion is briefly reversed and the ankle plantar flexion moment returns to normal levels. This suggests that although biomechanically disadvantaged by foot position and excessive knee flexion, the ankle plantar flexors initially absorb energy during loading response as the tibia falls forward, but then limit excess dorsiflexion with a brief burst of power generation at the ankle at the beginning of mid-stance. Since the physical examination was inconclusive, it is not clear whether this is due to true ankle plantar flexor strength or simply the resistance or viscoelastic behavior of the musculotendon unit. Nevertheless, it does explain the early dorsiflexion peak in the ankle sagittal plane graph and suggests there is

some eccentric control over tibial advancement during loading response and mid-stance.

However, ankle function is not as good during terminal stance and pre-swing, when the powerful concentric contraction of the triceps surae muscles is needed to produce rapid ankle plantar flexion. The strongest evidence of this is shown in the reduced ankle power generation during pre-swing in Figure 5.16, where the power generation is approximately 25% normal on both sides. Since power generation is normally larger at the ankle than at any other joint, and substantial power generation from both the hip flexors and ankle plantar flexors is necessary to produce passive knee flexion during pre-swing, this is a profound deficit that affects both the knee and the hip, and is the strongest evidence of plantar flexor weakness in the analysis.

The experienced gait analyst might cite the delayed heel-off and short step length of this "calcaneal gait" pattern as obvious indicators of calf weakness. While this may be true, the ankle power data provides a strong quantitative justification for such a claim, and has the added benefit of gauging the degree of dynamic plantar flexor weakness that occurs at this critical point in the gait cycle. This evidence, along with the excess dorsiflexion during stance period and our concerns about more severe crouch gait deformity as LD matured, eliminated any thoughts of a tendo-Achilles lengthening or intramuscular lengthening of the gastrocnemius for this subject.

Based on the results of this IGA and the other physical examination and radiographic evidence, and following consultation with the patient and his family, our clinical team recommended that LD undergo bilateral rectus femoris transfers to the semitendinosus, bilateral hamstring lengthenings, bilateral Evans calcaneal lengthenings, and a left tibial osteotomy of approximately 20 degrees internal. The rectus femoris transfers were clearly indicated from both kinematic and dynamic EMG evidence and the presence of a stiff-knee gait pattern. The hamstring lengthenings were supported by physical examination and IGA data, and could be efficiently performed in conjunction with the rectus transfers. Since there were no previous surgical procedures performed on the hamstrings, our team prefers to transfer the rectus femoris to the semitendinosus, although we have found no evidence to rule out the other potential transfer sites of sartorius or gracilis (49). The Evans calcaneal lengthenings were primarily supported by the radiographic evidence that showed significant uncovering of the talus and forefoot abduction with mid-foot collapse. Further evidence to support this procedure was obtained using plantar pressure measurements from each foot recorded using a two-meter pressure plate mounted in the motion laboratory walkway after the force platform array. These recordings confirmed the existence of pes planus and showed an increased pressure under the first metatarsal heads and medial border bilaterally, with the pressures higher under the left foot. This information, combined with concerns expressed by the family regarding LD's flat feet, and the clinical team's expectation that a more rigid and properly aligned foot may improve the power generation capability of the ankle plantar flexors during pre-swing, convinced us to add this procedure to the recommendations. The left tibial osteotomy was warranted based on the rotational kinematic findings that showed a distal shank rotation of approximately 15 to 20 degrees greater than normal, a foot progression angle approximately 25 to 30 degrees greater than normal (including the contribution from the external hip rotation that was believed to be compensatory), and the family's concerns about the limb asymmetry and increasing external foot position.

Finally, bilateral posterior leaf-spring AFOs were prescribed to provide some plantar flexor assist and to help control tibial advancement in the presence of the weak plantar flexors that would most likely persist after LD recovered from his surgical procedures. However, we were hopeful that if we achieved our intended improvement in biomechanical position, increased knee flexion in swing period, increased knee extension at initial contact, and a more rigid foot following surgery, the physical demands of walking for LD would be sufficiently reduced to discontinue the AFOs once he fully recovered.

MOTION ANALYSIS CASE STUDY—POSTOPERATIVE ANALYSIS

The family accepted the recommendations for surgery and all procedures outlined previously were completed at our institution approximately 6 months after the first analysis. All procedures were performed at the same time and under a single anesthesia, in accordance with our single-event, multilevel protocol (SEMLS). Approximately 1 year after the procedure when LD was 15 years 2 months old, a repeat IGA was performed; the pre-/post-operation comparison graphs for kinematics and kinetics of the more affected left side are shown in Figures 5.18 and 5.19 respectively. Space limitations prevent us from presenting the complete results, although similar findings were obtained from the less affected right side.

Evidence of a marked improvement in LD's overall gait performance was seen in the temporal–spatial parameters and in specific features of the kinematic and kinetic graphs. Stride length increased by 20% to 97% of that normal for his age, with similar increases in step length on both sides. Cadence also increased by 12% to normal levels. Subsequently walking speed increased by 30% to 96% of the speed of a typically developing teenager. The left hand column of Figure 5.18 shows the sagittal plane kinematic pre-/post comparison for the left side. Here we see an improvement in hip extension and knee extension at terminal stance, a dramatic improvement in the peak knee flexion in initial and mid-swing, and a more normal ankle curve indicating the presence of a heel rocker, less dorsiflexion at terminal stance, and plantar flexion at pre-swing.

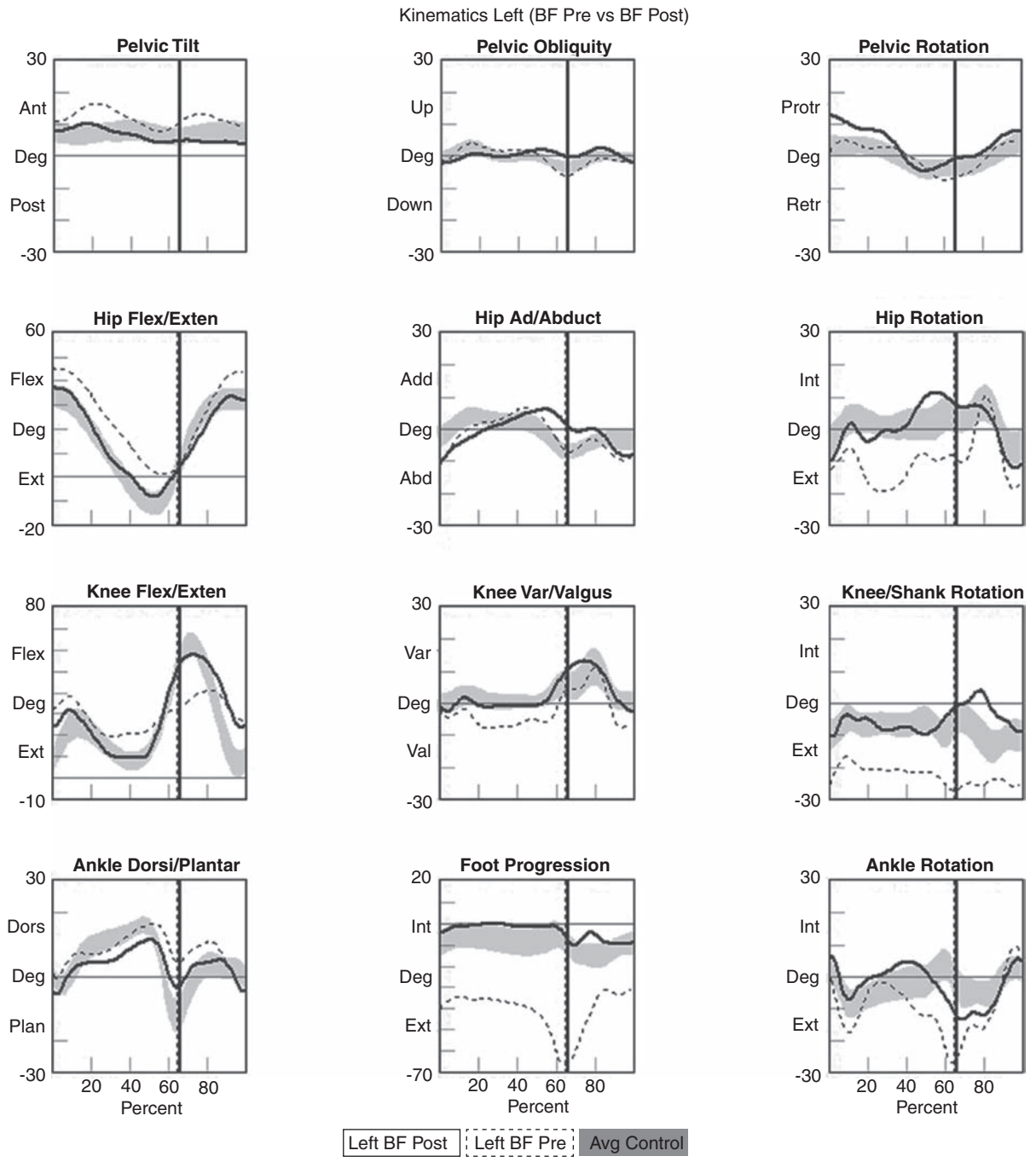

FIGURE 5.18 Three-dimensional kinematic graphs comparing the pre/post surgical outcomes on the left side of case-study subject "LD" approximately 1 year post surgery, at age 15 years 2 months. A representative IGA trial from each analysis is used for comparison. Post surgery graphs are represented as solid lines, pre-op values are shown as dashed lines, and the gray (shaded) band is from the age-matched normal database collected in the laboratory and used as a reference. Improvements in these curves compared to the pre-operative values are described in the text.

The improvement in hip extension and knee extension at terminal stance has the functional consequence of improving the trailing limb posture, which in turn explains why the stride and step lengths improved following the SEMLS procedures. Because knee extension at terminal stance, peak knee flexion in swing, and the slope of the rising edge of the knee curve all improved, this provides evidence that several critical events around the knee have been restored, and LD's stiff-knee gait pattern has been resolved (49). The sagittal plane knee curve also shows that knee extension at terminal swing and initial contact has not appreciably changed following the procedures, although with the improved knee extension at terminal stance and improved knee dynamic ROM the crouch gait pattern in stance has

been minimized further. We have found that hamstring lengthening is often not very effective at improving knee extension at terminal swing (50), but can be effective at increasing knee extension at terminal stance. We believe this is because swing period is a ballistic movement that relies on fine motor control to achieve the critical event of full knee extension during terminal swing, and because of this, requires a fundamentally different intervention from surgical lengthening. Physical therapy and gait training may be effective here, although the evidence is inconclusive at this time. Improvement in knee extension at terminal swing will be necessary to fully restore the first rocker and achieve the critical event of heel-first initial contact, which LD was able to achieve only occasionally during this assessment.

The most notable improvement seen in transverse plane kinematics is shown clearly in the foot progression graph of Figure 5.18. The 40 degrees of external foot progression during the stance period seen preoperatively has been reduced to about 5 degrees of external rotation

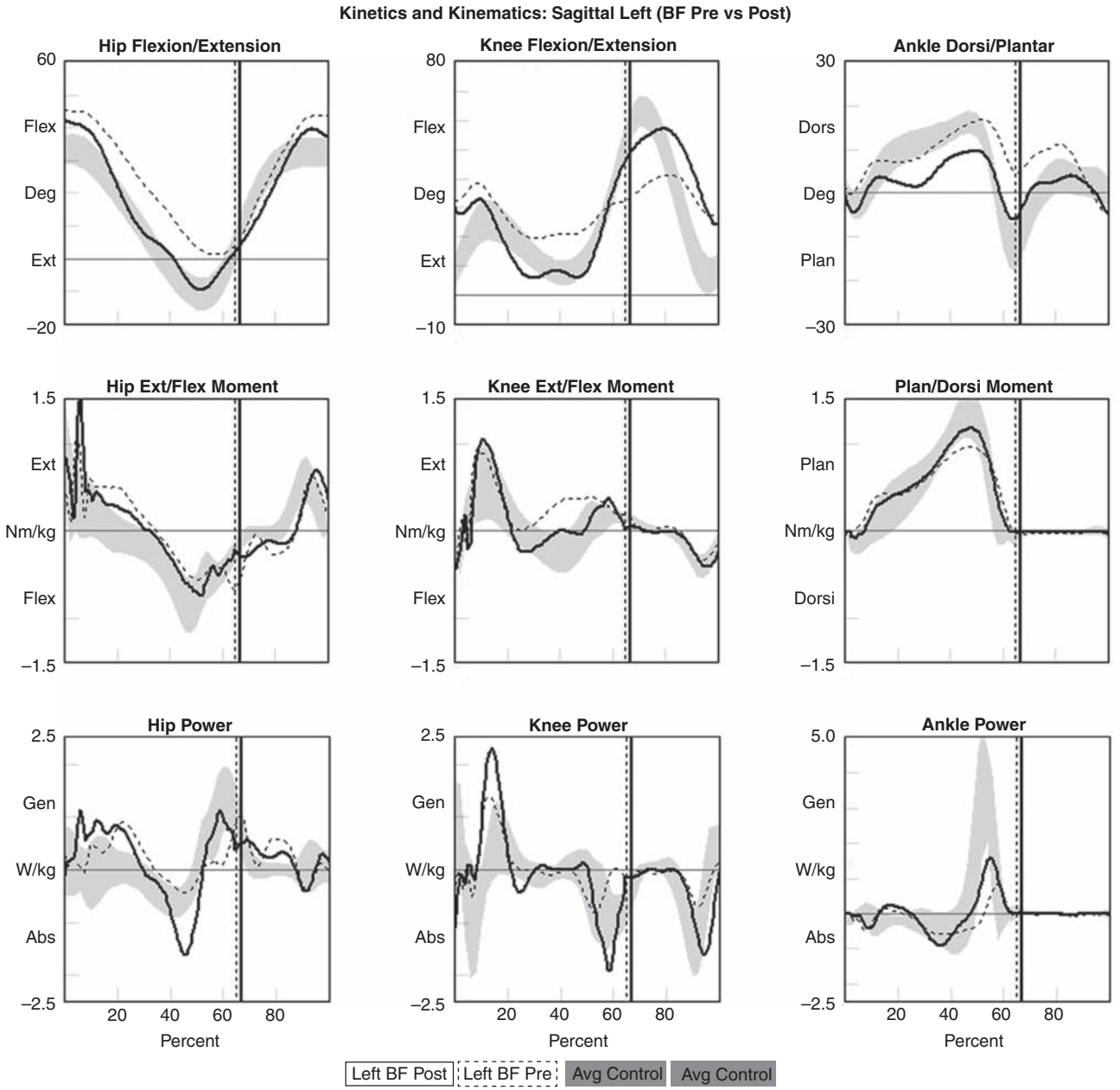

FIGURE 5.19 Sagittal plane kinematic and kinetic graphs comparing the pre/post surgical outcomes on the left side of case-study subject "LD" approximately 1 year post surgery, at age 15 years 2 months. A representative IGA trial from each analysis is used for comparison. Post surgery graphs are represented as solid lines, pre-op values are shown as dashed lines, and the gray (shaded) band is from the age-matched normal database collected in the laboratory and used as a reference. Improvements in these curves compared to the pre-operative values are described in the text.

following the SEMLS procedure. This change comes from an approximately 20 degree improvement in distal shank rotation following the internal tibial osteotomy, and a 10 degree reduction in external hip rotation. The remainder of the correction likely came from a small improvement in foot alignment following the calcaneal osteotomy, where the postoperative x-ray showed less pes planus than the preoperative film. Also notice that the large external whip evident on the preoperative foot progression angle curve at foot-off has been eliminated, most likely due to the improvement in the task of limb advancement and improved foot clearance now that knee flexion in initial swing has been improved following the rectus femoris transfer.

Figure 5.19 shows the sagittal plane kinematics and kinetics for the left side from the comparison study. Along with the improved hip extension at terminal stance (left column), there is a small increase in hip flexion moment at terminal stance, a large increase in hip flexion power absorption at terminal stance, and a subsequent increase in hip flexion power generation at pre-swing. All of these findings are consistent with the observation of an improved trailing limb posture, and provide evidence that there is functional importance in achieving this critical event. At the ankle we see a similar improvement in both plantar flexion power absorption and more importantly, plantar flexion power generation; this quantity has increased by more than 50%. It is likely due to the improved alignment of the foot in the direction of progression, which reduces the lever arm dysfunction associated with the externally rotated foot (35). With improved power generation at both the hip and ankle, knee flexion and power absorption at pre-swing must also improve, and evidence of this improved passive knee flexion can be found in the middle column of Figure 5.19. We also see a more normal knee flexion moment in the stance period, which is a direct consequence of the improved knee function and dynamic range achieved with the SEMLS procedures.

The last important findings from the postoperative analysis were reflected in the temporal–spatial data and post-operative orthotics versus barefoot trial (kinematics and kinetics graphs) that were included in the analysis but not reproduced here. With his posterior leaf-spring AFOs donned, LD's stride length and walking speed were about 10% better than in the barefoot trial. However, the kinematic graphs showed increased dorsiflexion at initial contact and throughout stance, and increased knee flexion at initial contact, terminal stance, and terminal swing consistent with a return of the mild crouch gait pattern during stance in the post-operative orthotics trial compared to the post-operative barefoot trial. There was also an internal foot progression angle with the AFO. Evidence of an excessive plantar flexion moment during loading response was also seen when wearing the AFO, along with a reduced power generation at pre-swing. Since his calves were still weak and we had evidence of improved walking speed and step length in the AFO trial, we elected to continue the orthotics prescription. However, all of these findings suggested that the posterior leaf-spring AFO had been molded in excessive dorsiflexion, or the trimlines around the malleoli had been excessive, so the intended purpose of providing plantar flexor assist during stance to prevent premature advancement of the tibia during the second rocker had not been achieved. Based on this information a new prescription for stiffer posterior leaf-spring AFOs molded with less dorsiflexion was recommended, along with physical therapy and gait training to improve calf strength and to try to improve knee extension at terminal swing. No additional surgical recommendations were made.

This case study illustrates how quantitative information on gait and movement in the hands of a clinician who understands the systematic approach outlined in this chapter can find evidence to support complex interventions and achieve reasonable outcomes in the pediatric patient with gait dysfunction. We hope this can be a guide to using IGA in your clinical practice, and demonstrates that functional decomposition of the gait cycle can be the basis for clinical decision making in these complex patients.

SUMMARY

In this chapter, we have attempted to provide an overview of the methods, procedures, and strategies for utilizing instrumented movement analysis to assist with the clinical interpretation of gait deformity in children. Focusing on the functional subdivisions that naturally occur during normal walking, and identifying the specific critical events that must be accomplished in each phase of gait, we have developed a framework that can be used for both instrumented and observational gait analysis and that can be applied to children and adults. By providing a brief description of modern computerized systems for movement analysis and linking measurements from these systems to functional tasks and critical events, we hope that IGA will be less intimidating and more clinically relevant to the pediatric physiatrist. Controversy remains regarding the value of IGA and its place on the modern rehabilitation service, with staunch advocates (8,10,35,57,58) and ardent detractors alike (59,60). It is our hope that armed with a solid background in the principles of gait analysis and an objective and impartial understanding of the benefits and limitations of current methodologies (61), every pediatric physiatrist can make the best clinical choices for the complex neuromuscular patients who rely on their decisions.

PEARLS, PERILS, AND RESOURCES

- IGA supports decision making for the child with walking problems, but does not replace a sound clinical and technical understanding of normal gait.

- Normal gait is naturally cyclical and symmetric, so any movement asymmetry should be investigated.
- There are 13 critical events that must occur during eight distinct phases of the gait cycle to produce a normal gait pattern. Each critical event has functional significance, and so provides a link between observed gait abnormalities and possible interventions.
- IGA provides evidence of absent, altered, or delayed critical events, and provides the framework for identifying treatments to directly address these functional limitations.
- While skill, experience, and practice are required to fully utilize IGA results for clinical interpretation, by following the strategy outlined in this chapter, the process can be less intimidating and more clinically relevant to the pediatric physiatrist.

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6

ELECTRODIAGNOSIS IN PEDIATRICS

Craig M. McDonald

INTRODUCTION

Electromyography (EMG), nerve conduction studies (NCSs), and evoked potentials, including somatosensory evoked potentials (SSEPs) and motor evoked potentials (MEPs), provide useful information to assist the clinician in the localization of pathology within the lower motor neuron and selected areas of the central nervous system. In the case of acquired or hereditary disorders of the lower motor neuron—anterior horn cell, peripheral nerve, neuromuscular junction (NMJ; presynaptic or postsynaptic region), or muscle—electrodiagnostic studies (EDSs) are a useful tool to be used as an extension of the clinician's physical examination. The information gained from EDSs may be invaluable in planning subsequent more invasive diagnostic studies (eg, muscle and nerve biopsy, cerebrospinal fluid (CSF) examination, MRI, which at times requires general anesthesia), allow for more cost-effective and specific molecular genetic testing, or aid in the surgical management of peripheral nerve trauma, compressive lesions, or entrapments. In the case of immune-mediated disorders such as myasthenia gravis or Guillain–Barré syndrome (GBS), EDSs may permit prompt treatment.

While many may assume a decreased role of EDSs in the era of molecular genetic diagnosis for children with neuromuscular disorders, EDSs continue to play an important role in the diagnosis evaluation of children with neuromuscular disorders, although the practice paradigm is evolving. Major advances in genetic analysis and neuroimaging have modified the traditional diagnostic approach to neuromuscular disorders. A recent study investigated the role of pediatric EMG in the modern molecular era (1). All EMG studies performed at Boston Children's Hospital from 2001 through 2011 were retrospectively reviewed and data were collected on study numbers, patient ages, referring provider subspecialty, study indication, electrophysiologic diagnosis, and study utility. A total of 2,100 studies were performed. The volume increased from approximately 160 to about 250 studies/year over a 10-year period. There was a trend toward studying older children. Neurologists, including neuromuscular specialists, constituted the major referral pool, whereas referrals from orthopedics increased steadily. Polyneuropathies followed by mononeuropathies were the most common diagnoses referred to the electrodiagnostic laboratory. Fifty-seven percent of studies were normal. EMG provided meaningful information in 94% of cases.

Another recent study assessed the diagnostic yield of electromyography in children with myopathic disorders (2), as the interpretation of pediatric electromyography in myopathic disorders is technically challenging. The investigators assessed the sensitivity and specificity of electromyography in pediatric myopathy cases through a retrospective chart review of patients 18 years and less, between 2009 and 2013. Two hundred twenty-four electromyographic studies were reviewed with the following referral diagnoses: myopathy, muscle weakness, neuromuscular disorders, myositis, myalgia, myoglobinuria, myasthenia, myotonia, cramps, periodic paralysis, hypotonia, and developmental delay. Only children who had an electromyography and muscle biopsy were included for analysis. Patients with neurogenic electromyography and NMJ disorders were excluded. Myopathic electromyography was defined as short-duration, low-amplitude, polyphasic motor unit potentials (MUPs) with rapid recruitment. Seventy-two patients were included (age range, 6 months–18 years) and they were grouped according to the following findings: group A: myopathic electromyography, and biopsy or genetically confirmed myopathy (32 cases); group B: myopathic electromyography but biopsy normal or nondiagnostic (12 cases); group C: normal electromyography but biopsy or genetically confirmed myopathy (three cases, all with metabolic myopathy); and group D: electromyography normal and biopsy normal or nondiagnostic (25 cases). The most common diagnoses were congenital myopathy (seven cases), metabolic myopathy (six cases), muscular dystrophy (six cases), genetically confirmed myopathy (five cases), myopathy, undefined (five cases), and inflammatory myopathy (four cases). The authors concluded that pediatric electromyography was 91% sensitive and 67% specific in myopathic

disorders. The metabolic myopathies were commonly missed by electromyography. In another center, the sensitivity and specificity of preoperative electromyography were assessed for the detection of muscle pathology in using muscle biopsy findings as a gold standard. The sensitivity and specificity were determined to be only 58% and 56%, respectively (3). Clearly the diagnostic utility of pediatric EDSs is somewhat dependent on the skills of the electromyographer and the nature of the referral problems.

Pediatric electrodiagnosis must be approached with knowledge of peripheral neuromuscular development and thoughtful planning of the study with regard to most likely diagnostic possibilities, developmental status of the child, and the likelihood that the pediatric electrodiagnostic practitioner will be able to provide clinicians and family with useful diagnostic information. The physical examination and developmental level of the infant or child direct the study. The examination requires patience and technical competence by an electrodiagnostic clinician experienced and skilled in the evaluation of children. This chapter will focus on considerations specific to the electrodiagnostic evaluation of infants and children with an emphasis on practical suggestions that may facilitate the completion of an accurate pediatric electrodiagnostic examination with the minimization of discomfort and distress to the child, parent, and pediatric electrodiagnostic specialist.

MATURATIONAL FACTORS IN PEDIATRIC ELECTRODIAGNOSIS

The normative neurophysiologic data relating to the maturation of peripheral nerves and muscle in children has greatly expanded in the recent past (4–13). The reader is referred to the volume by Jones, Bolton, and Harper (11) for an excellent review of neurophysiologic norms in pediatric populations. Peripheral nerve myelination begins at about the 15th week of gestation and continues throughout the first 3 to 5 years after birth (14). Conduction velocities (CVs) are determined by myelination, diameter of the fiber, and internodal differences. Myelination occurs at the same rate, whether intrauterine or extrauterine. CVs are directly related to gestational and postconceptual age and are unrelated to birth weight (15,16). CVs increase in direct proportion to the increase in diameter of fibers during growth. A direct relationship exists between the diameter of the axon and the thickness of the myelin sheath. The diameter of the fibers at the time of birth has been shown to be one-half of that in the adult. No unusual acceleration of myelination occurs subsequent to birth (17). Peripheral fibers reach their maximum diameter at 2 to 5 years after birth (17,18). The nodes of Ranvier continue to remodel with peak internodal distances being reached at 5 years of age.

NERVE CONDUCTION STUDIES

In general, normal standard adult values for CVs are reached by ages 3 to 5. In infancy, upper and lower extremity CVs are similar under age 1. Subsequently, faster conductions are maintained in the upper extremities, and comparatively slower conductions in the lower extremities as with adults. Unique values for expected CVs are observed for specific peripheral nerves.

MOTOR NERVE CONDUCTION

Motor conduction velocities (MCVs) in infants are found to be one-half of adult values. In infants, conduction studies should be at least greater than 20 M/S. At birth, mean MCVs for the median, ulnar, and peroneal nerves are 27 M/S. The median nerve may lag in maturation of CV relative to the ulnar and peroneal nerves. Ulnar MCV values reach the lower adult range by age 3 (17). The slight difference between ulnar and median MCV values present in the first 3 years of life disappears in children by 4 to 5 years of age. Careful and consistent measurements are necessary to achieve reliable and valid data. Normative values or selected motor nerve conduction velocities (NCVs) are shown in Table 6.1.

DISTAL MOTOR LATENCY (DML)

Distal motor latencies (DMLs) show maturational changes between infancy and 3 to 5 years of age, similar to MCVs. Normative data for distal latencies have generally been more incomplete with ranges of distances provided (from stimulation to active electrode). While the stimulation distance should always be recorded in the electrodiagnostic report, the specific distal latency is rarely of critical importance in determining a diagnosis in pediatric electrodiagnosis, as distal peripheral entrapments are relatively uncommon. Rather, reported distal latencies, which are either unusually fast or unusually slow in the setting of otherwise normal MCVs, should raise a suspicion regarding technical problems and identification of appropriate waveforms.

The *corrected DML* may be used as an alternative for young children, by applying the formula of Slomic and colleagues found in Wagner and Buchthal (19):

$$\text{Corrected DML} = \text{measured DML} - [L - X/\text{MCV}],$$

where L = actual distance between stimulating cathode to the active recording electrode, and X = standard distance (4 cm for nerves of upper limbs and 5 cm for nerves of the lower limbs). Garcia and colleagues (13) have reported the most complete data to date on corrected DML in children (see Table 6.2). The corrected DML in the neonatal

TABLE 6.1 NORMAL MOTOR CONDUCTION VELOCITIES (M/SEC)

	MEDIAN (REF.)	ULNAR (REF.)	PERONEAL (REF.)	TIBIAL (REF.)
24–72 hours	25.8 ± 3.40 (12)	28.0 ± 3.38 (12)	26.4 ± 3.58 (12)	23.9 ± 2.73 (12)
7 days–1 month	25.43 ± 3.84 (9) 26.17 ± 2.16 (13)	25.03 ± 2.7 (13)	22.43 ± 1.22 (9) 25.60 ± 3.68 (13)	25.30 ± 1.96 (4) 23.21 ± 2.79 (13)
0–3 months	32.3 ± 3.56 (12)	35.1 ± 3.40 (12)	30.8 ± 2.91 (12)	27.8 ± 3.89 (5)* 27.9 ± 2.27 (12)
4–6 months	37.0 ± 4.38 (12)	40.5 ± 4.24 (12)	36.1 ± 4.67 (12)	36.3 ± 4.98 (5)** 34.7 ± 2.92 (12)
1–6 months	34.35 ± 6.61 (9) 36.35 ± 3.66 (13)	36.33 ± 3.72 (13)	35.18 ± 3.96 (9) 36.69 ± 4.06 (13)	32.55 ± 4.05 (13)
6–12 months	42.3 ± 6.43 (12) 43.91 ± 3.44 (13)	47.2 ± 6.33 (12) 45.02 ± 2.93 (13)	40.8 ± 6.16 (12) 43.11 ± 4.13 (13)	38.5 ± 5.50 (12) 39.45 ± 4.29 (10)
1–2 years	48.23 ± 4.58 (9) 47.81 ± 2.33 (13)	48.95 ± 2.46 (13)	51.42 ± 3.02 (9) 47.43 ± 2.5 (13)	42.6 ± 3.80 (5) 42.42 ± 2.23 (13)
1–3 years	52.7 ± 4.70 (12)	53.8 ± 4.83 (12)	48.7 ± 4.86 (12)	44.9 ± 4.44 (12)
2–4 years	53.59 ± 5.29 (9) 52.71 ± 3.71 (13)	54.19 ± 3.49 (13)	55.73 ± 4.45 (9) 51.21 ± 3.95 (13)	49.8 ± 5.78 (5) 44.81 ± 1.91 (13)
4–6 years	56.26 ± 4.61 (9) 55.0 ± 5.20 (12) 56.48 ± 2.36 (13)	56.9 ± 4.34 (12) 56.51 ± 3.19 (13)	56.14 ± 4.96 (9) 49.6 ± 4.98 (12) 53.99 ± 3.74 (13)	50.0 ± 4.26 (5) 48.6 ± 4.25 (12) 48.43 ± 2.53 (13)
6–14 years	57.2 ± 3.71 (12)*** 57.32 ± 3.35 (9)	58.3 ± 5.76(12)***	49.6 ± 3.40(12)*** 57.05 ± 4.54 (9)	48.2 ± 2.76 (12)*** 52.4 ± 4.19 (5) ±

Data are presented as means ± standard deviation
*1 to 3 months; **3 to 6 months; ***7 to 14 years; ± 6 to 11 years

group is increased relative to other age groups, decreases over the first 12 months of life, remains unchanged between 12 months and 24 months, and slightly increases later. As most clinicians reading reports are not familiar with the corrected DML, an explanation of the calculation and normative interpretation should be included in the report, if this data is reported along with the actual DML and distance used.

TABLE 6.2 CORRECTED DISTAL MOTOR LATENCY (MSEC)

		MEDIAN	ULNAR	PERONEAL	TIBIAL
<1 month		3.00 ± 0.18	2.80 ± 0.43	3.33 ± 0.54	3.21 ± 0.36
1–6 months	2.47 ± 0.18	2.20 ± 0.38	2.51 ± 0.22	2.62 ± 0.41	
6–12 months	2.28 ± 0.21	1.98 ± 0.21	2.36 ± 0.31	2.55 ± 0.37	
1–2 years		2.34 ± 0.34	1.86 ± 0.16	2.35 ± 0.23	2.45 ± 0.46
2–4 years		2.34 ± 0.23	1.89 ± 0.17	2.57 ± 0.40	2.35 ± 0.24
4–6 years		2.56 ± 0.29	2.03 ± 0.25	3.02 ± 0.48	2.69 ± 0.46

Data are presented as means ± standard deviation

Corrected DML = measured DML – [L – X/MCV], where L = actual distance between stimulating cathode to the active recording electrode, and X = standard distance (4 cm for nerves of upper limbs and 5 cm for nerves of the lower limbs.

Source: Adapted from Ref. (13). García A, Calleja J, Antolín FM, Berciano J. Peripheral motor and sensory nerve conduction studies in normal infants and children. *Clin Neurophysiol*. 2000;111(3):513–520.

COMPOUND MUSCLE ACTION POTENTIAL (CMAP)

Compound muscle action potential (CMAP) amplitudes are important to consider in the evaluation of axonal loss, conduction block, and muscle fiber atrophy. CMAP amplitudes of lower extremity nerves are one-half to one-third adult values in infants and upper extremity CMAPs may be one-third to one-fourth adult values during infancy. As with MCVs, CMAP amplitudes increase in size with age but adult values are generally not reached until the end of the first decade. Normal values for CMAP amplitudes are shown in Table 6.3.

SENSORY NERVE CONDUCTION

Modern EMG equipment, which includes amplifiers and signal averaging capability, allows sensory nerve action potentials (SNAPs) to be routinely recorded in the absence of peripheral nerve pathology. Maturational changes for orthodromic and antidromic sensory conduction are similar to that for motor fibers (9,20,21). In infants and young children, two distinct peaks are often observed in the SNAP with proximal stimulation. This two-peak potential has been attributed to differences in maturation between two groups of sensory fibers (19) and often persists until 4 to 6 years of age. Sensory NCVs may be calculated from single distal antidromic or orthodromic stimulations by measuring the distance from the stimulation point to the active electrode and the distal latency. Normative values for sensory NCVs in selected nerves using orthodromic stimulation and proximal recording are shown in Table 6.4. Normal values for orthodromic and antidromic SNAP amplitudes are shown in Table 6.5.

F-WAVES

The F-wave is a late response that appears as supermaximal motor nerve stimulation and arises from the discharge of a small number of motor neurons, in response to antidromic stimulation of the motor axon. The F-wave latency is measured from hand and foot intrinsic muscles and is useful for evaluating the motor NCV and proximal nerve segments. With the F-wave, the speed of motor nerve conduction is measured over a long distance, and therefore is less subject to errors inherent in the calculation of MCVs over short distances (10 cm or less). F-waves can be recorded from most limb nerves in newborns and young infants. The minimum F-latency in normal children recorded from hand muscles with median or ulnar nerve stimulation at the wrist is generally less than 20 msec in children younger than 6 years of age (9,10,22). In the lower extremities, the F-wave latency recorded from intrinsic foot muscles with peroneal or posterior tibial nerve stimulation at the ankle is

TABLE 6.3 NORMAL CMAP AMPLITUDES (MV)

	MEDIAN (APB) (REF.)	ULNAR (ADM) (REF.)	PERONEAL (EDB) (REF.)	TIBIAL (AH) (REF.)
24–72 hours	3.60 ± 1.56 (12)	5.42 ± 2.21 (12)	3.43 ± 0.47 (12)	9.29 ± 1.93 (12)
7 days–1 month	3.00 ± 0.31 (9) 1.27 ± 0.74 (13)	1.88 ± 0.92 (13)	3.0 ± 1.26 (9) 1.77 ± 0.62 (13)	4.40 ± 1.73 (13)
0–3 months	4.06 ± 1.49 (12)	6.49 ± 2.83 (12)	4.52 ± 0.85 (12)	13.30 ± 2.86 (12)
1–6 months	7.37 ± 3.24 (9) 2.37 ± 1.27 (13)	3.11 ± 1.45 (13)	5.23 ± 2.37 (6) 2.68 ± 1.04 (13)	6.16 ± 2.44 (13)
6–12 months	5.47 ± 2.01 (12) 2.94 ± 1.17 (13)	6.97 ± 1.89 (12) 2.73 ± 1.09 (13)	5.86 ± 1.12 (12) 2.64 ± 1.32 (13)	14.06 ± 2.58 (12) 6.83 ± 2.69 (13)
1–2 years	8.90 ± 3.61 (9) 4.12 ± 1.90 (13)	4.55 ± 1.53 (13)	5.80 ± 2.48 (9) 3.69 ± 1.27 (13)	9.07 ± 2.12 (13)
1–3 years	5.88 ± 2.51 (12)	7.66 ± 2.23 (12)	6.42 ± 1.92 (12)	15.71 ± 1.79 (12)
2–4 years	9.55 ± 4.34 (9) 5.96 ± 2.01 (13)	5.48 ± 1.42 (13)	6.10 ± 2.99 (9) 4.25 ± 1.59 (13)	9.57 ± 3.54 (13)
4–6 years	10.37 ± 3.66 (9) 6.49 ± 1.83 (12)	8.80 ± 2.35 (12)	7.10 ± 4.76 (9) 3.78 ± 1.23 (13)	9.48 ± 2.39 (13)
6–14 years	12.37 ± 3.66 (9) 8.83 ± 1.87 (12)*	10.27 ± 2/02 (12)*	8.15 ± 4.19 (9) 7.22 ± 1.64 (9)*	15.75 ± 1.77 (12)*

Data are presented as means ± standard deviation

*7 to 14 years

Recording sites: abductor pollicis brevis (APB); adductor dididti minimi (ADM); extensor digitorum brevis (EDB); abductor hallucis (AH).

TABLE 6.4 NORMAL SENSORY CONDUCTION VELOCITIES (M/SEC)

	MEDIAN (REF.)	ULNAR (REF.)	SURAL (REF.)
24–72 hours	18.74 ± 2.64 (D2-W) (12)* 21.68 ± 2.43 (D2-E) (12)*	19.13 ± 0.29 (D5-W) (12)* 21.85 ± 1.37 (D5-E) (12)*	17.65 ± 2.43 (6 cm) (12)*
7 days–1 month	22.31 ± 2.16 (D2-W) (9) 24.09 ± 2.6 (D3-W) (13)	18.4 ± 3.97 (D5-W) (6)	20.26 ± 1.55 (4–8 cm) (9)
0–3 months	24.20 ± 3.51 (D2-W) (12)* 29.26 ± 4.14 (D2-E) (12)*	25.95 ± 2.49 (D5-W) (12)* 34.42 ± 4.13 (D5-E) (12)*	22.54 ± 2.28 (8 cm) (12)*
4–6 months	29.91 ± 2.17 (D2-W) (12)* 38.44 ± 5.35 (D2-E) (12)*	31.51 ± 2.70 (D5-W) (12)* 44.07 ± 4.12 (D5-E) (12)*	28.78 ± 2.98 (8 cm) (12)*
1–6 months	35.52 ± 6.59 (D2-W) (9) 35.07 ± 4.87 (D3-W) (13)	27.7 ± 6.37 (D5-W; 1–3 months) (6) 37.1± 5.25 (D5-W; 3–6 months) (6)	34.68 ± 5.43 (6–8 cm) (9)
6–12 months	40.31 ± 5.23 (D2-W) (9) 32.60 ± 3.15 (D2-W) (12)* 41.14 ± 4.43 (D2-E) (12)* 41.95 ± 2.68 (D3-W) (13)	40.0 ± 5.13 (D5-W) (6) 34.41 ± 3.11 (D5-W) (12)* 44.67 ± 3.45 (D5-E) (12)*	29.40 ± 3.55 (8 cm) (12)*
1–2 years	46.93 ± 5.03 (D2-W) (9) 45.12 ± 2.99 (D3-W) (13)	44.2 ± 7.79 (D2-W) (6)	49.73 ± 5.53 (8–10 cm) (9)
1–3 years	36.41 ± 3.93 (D2-W) (12)* 47.23 ± 3.74 (D2-E) (12)*	34.94 ± 2.92 (D5-W) (12)* 45.59 ± 4.26 (D5-E) (12)*	35.37 ± 4.32 (8 cm) (12)* 38.33 ± 4.49 (12 cm) (12)*
2–4 years	49.51 ± 3.34 (D2-W) (9) 48.82 ± 3.02 (D3-W) (13)	48.8 ± 3.01 (D5-W) (6)	52.63 ± 2.96 (8–10 cm) (12)
4–6 years	51.71 ± 5.16 (D2-W) (9) 41.04 ± 4.94 (D2-W) (12)* 51.22 ± 5.07 (D2-E) (12)* 50.72 ± 3.6 (D3-W) (13)	47.7 ± 6.75 (D5-W) (6) 42.94 ± 4.55 (D5-W) (12)* 51.58 ± 4.49 (D5-E) (12)*	53.83 ± 4.34 (8–10 cm) (9) 39.38 ± 4.58 (8 cm) (12)* 41.49 ± 4.41 (12 cm) (12)*
6–14 years	53.84 ± 3.26 (9) 43.71 ± 3.37 (D2-W) (12)* 53.44 ± 3.19 (D2-E) (12)*	46.6 ± 5.6 (D5-W) (6) 43.92 ± 3.91 (D5-W) (12)* 53.23 ± 3.58 (D5-E) (12)*	53.85 ± 4.19 (9) 40.60 ± 4.79 (8 cm) (12)* 42.75 ± 4.79 (12 cm) (12)* 46.71 ± 4.17 (14 cm) (12)*

Data are presented as means ± standard deviation

*Velocities based on peak latencies for Cai and Zhang (12); others based on onset latencies

D-W = finger to wrist using ring electrodes with orthodromic stimulation

D-E = finger to elbow using ring electrodes with orthodromic stimulation

D2 = index finger for median; D3 = middle finger for median; D5 = fifth finger for ulnar

Sural nerve studies use antidromic with recording electrodes behind the lateral malleolus with stimulus delivered at 6 cm to 14 cm above the malleolus as specified.

generally less than 30 msec (7,9). Normal values for F-wave latencies for children are shown in Table 6.6.

H-REFLEX

The H-reflex is present in both the upper extremity (median and ulnar) and lower extremity (with posterior tibial stimulation) in infancy. While the tibial H-reflex persists into adulthood, the upper extremity H-reflex responses are present in virtually all infants at birth and become suppressed in most children over the course of the first year. Normal values for H-reflex latencies in children are shown in Table 6.7.

NEUROMUSCULAR TRANSMISSION

The NMJ shows less stability with repetitive nerve stimulation in normal newborns. At low rates of stimulation (1–2 Hz), no significant incremental or decremental changes in CMAP amplitude are observed (23). At higher

TABLE 6.5 NORMAL SNAP AMPLITUDES IN CHILDREN (μV)

	MEDIAN (REF.)	ULNAR (REF.)	SURAL (REF.)
24–72 hours	6.76 ± 0.79 (D2-W) (12)	5.26 ± 0.57 (D5-W) (12)	5.29 ± 2.16 (6 cm) (12)
7 days–1 month	6.22 ± 1.30 (D2-W) (9) 4.86 ± 2.23 (D3-W) (10)	5.5 ± 3.1 (D5-W) (6)	9.12 ± 3.02 (4–8 cm) (9)
0–3 months	16.74 ± 1.47 (D2-W) (12)	7.83 ± 0.60 (D5-W) (12)	9.97 ± 1.24 (8 cm) (12)
4–6 months	17.72 ± 3.35 (D2-W) (12)	8.26 ± 1.00 (D5-W) (12)	13.58 ± 2.19 (8 cm) (12)
1–6 months	15.86 ± 5.18 (D2-W) (9) 10.66 ± 3.62 (D3-W) (13)	9.4 ± 3.2 (D5-W; 1–3 months) (6) 13.2 ± 3.23 (D5-W; 3–6 months) (6)	11.66 ± 3.57 (6–8 cm) (9)
6–12 months	16.00 ± 5.18 (D2-W) (9) 17.55 ± 1.70 (D2-W) (12) 9.00 ± 3.45 (D3-W) (13)	13.0 ± 5.6 (D5-W) (6) 10.87 ± 2.4 (D5-W) (12)	14.87 ± 4.67 (8 cm) (12)
1–2 years	24.00 ± 7.36 (D2-W) (9) 15.72 ± 4.50 (D3-W) (10)	16.3 ± 2.44 (D2-W) (6)	15.41 ± 9.98 (8–10 cm) (9)
1–3 years	19.51 ± 3.99 (D2-W) (12)	12.34 ± 2.1 (D5-W) (12)	18.02 ± 3.83 (8 cm) (12)
2–4 years	24.28 ± 5.49 (D2-W) (9) 12.02 ± 5.89 (D3-W) (10)	16.0 ± 3.6 (D5-W) (6)	23.27 ± 6.84 (8–10 cm) (9)
4–6 years	25.12 ± 5.22 (D2-W) (9) 19.78 ± 4.21 (D2-W) (12) 14.04 ± 5.99 (D3-W) (10)	14.2 ± 2.72 (D5-W) (6) 13.15 ± 3.6 (D5-W) (12)	22.66 ± 5.42 (8–10 cm) (9) 18.50 ± 3.89 (8 cm) (12)
6–14 years	26.72 ± 9.43 (9) 20.50 ± 3.49 (D2-W) (12)*	13.4 ± 4.2 (D5-W) (6) 14.30 ± 2.5 (D5-W) (12)*	26.75 ± 6.59 (9) 18.67 ± 4.39 (8 cm) (12)*

Data are presented as means ± standard deviation

*Amplitudes determined from baseline to peak

Amplitudes are determined peak to peak from positive to negative peak of the SNAP unless otherwise noted

D-W = finger to wrist using ring electrodes with orthodromic stimulation

D2 = index finger for median; D3 = middle finger for median; D5 = fifth finger for ulnar

Sural nerve studies used antidromic stimulation with recording electrodes behind the lateral malleolus with stimulus delivered at 6 m to 14 cm above the malleolus as specified.

rates of stimulation (5–10 Hz), normal infants may show slight facilitation. Decremental responses averaging 24% have been reported at high rates of stimulation (20 Hz) in normal newborn infants. At 50 Hz stimulation, normal newborns may show decrements of the order of 50% (20). In general, decremental changes greater than 10% at low rates of stimulation (2–5 Hz) and facilitatory changes greater than 23% at high rates of stimulation (20–50 Hz) are felt to be significant in the post-term infant (24). Some authors have utilized high rates of stimulation of the order of 50 Hz for 10 seconds to document facilitation greater than 20% to 23% (at times, over 100% increments are observed) in infantile botulism (24–26).

ELECTROMYOGRAPHY

MOTOR UNIT CONFIGURATION AND AMPLITUDE

Amplitudes of motor unit action potentials (MUAPs) are lower in infants with amplitudes ranging from 150 microvolts to approximately 2,000 microvolts. Generally, MUAPs more than 1,000 microvolts in 0 to 3-year-old children are rare (27,28). In infants, MUAPs are usually biphasic or triphasic.

MOTOR UNIT DURATION

Infantile MUAPs are often shorter in duration. De Carmo (27) found newborn infants to exhibit durations 17% to 26% shorter than those seen in adults. Durations of MUAPs are often shorter than 5 msec in infants.

MOTOR UNIT RECRUITMENT

In very young infants and children, it is difficult to assess the strength of voluntary contraction and determine when the interference pattern is full. In general, as the strength of voluntary contraction increases, there is an increase in MUAPs recruited. However, the recruitment pattern in

TABLE 6.6 NORMAL F-WAVE LATENCIES IN CHILDREN (MSEC)

	MEDIAN (REF.)	ULNAR (REF.)	PERONEAL (REF.)	TIBIAL (REF.)
24–72 hours	19.56 ± 2.44 (w) (12) 16.51 ± 1.74 (e) (12)	19.67 ± 2.74 (w) (12) 16.64 ± 1.30 (e) (12)	27.56 ± 3.82 (a) (12) 24.38 ± 3.74 (k) (12)	26.92 ± 3.27 (a) (12) 23.51 ± 2.45 (k) (12)
7 days–1 month	18.17 ± 2.17 (w) (13)	18.63 ± 1.6 (w) (13)	25.2 ± 4.82 (a) (13)	23.92 ± 1.62 (a) (13)
0–3 months	17.62 ± 1.39 (w) (12) 15.39 ± 1.46 (e) (12)	17.65 ± 1.39 (w) (12) 14.93 ± 1.80 (e) (12)	26.14 ± 2.84 (a) (12) 23.46 ± 2.76 (k) (12)	28.59 ± 2.41 (a) (12) 22.52 ± 2.10 (k) (12)
4–6 months	17.54 ± 1.96 (w) (12) 15.41 ± 1.56 (e) (12)	16.99 ± 1.24 (w) (12) 14.91 ± 1.28 (e) (12)	25.18 ± 4.37 (a) (12) 22.15 ± 2.68 (k) (12)	23.93 ± 1.85 (a) (12) 20.67 ± 2.30 (k) (12)
1–6 months	15.91 ± 1.22 (w) (13)	15.71 ± 1.6 (w) (13)	21.4 ± 1.78 (a) (13)	21.4 ± 1.35 (a) (13)
6–12 months	16.86 ± 1.50 (w) (12) 14.37 ± 1.17 (e) (12) 15.67 ± 0.89 (w) (13)	17.02 ± 1.45 (w) (12) 14.41 ± 0.88 (e) (12) 15.45 ± 1.37 (w) (13)	25.54 ± 2.04 (a) (12) 21.56 ± 3.36 (k) (12) 20.33 ± 1.1 (a) (13)	23.78 ± 1.83 (a) (12) 20.92 ± 1.59 (k) (12) 22.0 ± 2.05 (a) (13)
1–2 years	15.64 ± 1.08 (w) (13)	15.67 ± 0.78 (w) (13)	22.82 ± 1.66 (a) (13)	24.21 ± 1.63 (a) (13)
1–3 years	16.41 ± 1.13 (w) (12) 14.21 ± 0.77 (e) (12)	16.63 ± 1.88 (w) (12) 14.69 ± 1.35 (e) (12)	26.73 ± 2.87 (a) (12) 24.30 ± 2.46 (k) (12)	25.44 ± 2.20 (a) (12) 23.65 ± 1.71 (k) (12)
2–4 years	16.36 ± 1.45 (w) (13)	16.0 ± 1.41 (w) (13)	24.64 ± 2.21 (a) (13)	25.6 ± 2.53 (a) (13)
4–6 years	17.62 ± 1.62 (w) (12) 15.81 ± 1.17 (e) (12) 18.0 ± 1.27 (w) (13)	18.51 ± 1.74 (w) (12) 16.53 ± 1.48 (e) (12) 18.25 ± 1.48 (w) (13)	30.57 ± 3.82 (a) (12) 25.22 ± 3.44 (k) (12) 29.45 ± 2.58 (a) (13)	31.07 ± 3.10 (a) (12) 25.97 ± 2.32 (k) (12) 30.12 ± 2.52 (a) (13)
6–14 years	20.18 ± 1.61 (w) (12) 17.34 ± 1.52 (e) (12)	20.66 ± 1.92 (w) (12) 18.14 ± 1.46 (e) (12)	38.16 ± 4.43 (a) (12) 31.38 ± 4.75 (k) (12)	36.32 ± 3.72 (a) (12) 32.78 ± 3.89 (k) (12)
18–30 years	26.14 ± 3.03 (w) (12) 22.88 ± 1.34 (e) (12)	27.03 ± 2.14 (w) (12) 23.42 ± 1.90 (e) (12)	49.63 ± 7.74 (a) (12) 41.23 ± 7.63 (k) (12)	48.27 ± 3.09 (a) (12) 39.93 ± 2.73 (k) (12)
Side-to-side difference	1.03 ± 0.73 (2.5)	0.94 ± 0.69 (2.3)	1.19 ± 1.17 (3.5)	1.26 ± 1.01 (3.3)

Data are presented as means ± standard deviation

Minimum F-wave latency from 10 recordings

(w) wrist stimulation; (e) elbow stimulation; (a) ankle stimulation; (k) knee stimulation

Side-to-side difference shows mean ± SD (upper limits of normal)

infants may be disordered and chaotic. As with adults, the recruitment frequency, defined as the firing rate of an MUAP when a different MUAP first appears with gradually increasing strength of voluntary contraction, is helpful in differentiating a myopathic process (lower recruitment frequency values) from a neuropathic process (higher recruitment frequencies greater than 20–25 Hz). An example of neuropathic recruitment is shown in Figure 6.1.

TECHNICAL FACTORS WITH INFANTILE NERVE CONDUCTION STUDIES

TEMPERATURE

The maintenance of appropriate subject temperature is essential during NCSs. Neonates generally have difficulty with temperature homeostasis and low subject temperature may have profound effects on CVs. A skin temperature of 36 to 37°C produces near-nerve temperatures of 37 to 38°C and avoids spurious reductions in NCVs and prolongation of distal latencies. It is assumed that a 1°C drop in temperature produces a slowing of conduction of the order of 2 to 3 m/s. Every attempt should be made to maintain extremity temperature with infant warmers, heating lamps, or warm blankets.

VOLUME CONDUCTION

Volume conduction is defined as the current transmission from a potential source through a conducting medium, such as the body tissues. This may produce depolarization of peripheral nerves in proximity to the specific nerve being studied, and this is particularly problematic in smaller

TABLE 6.7 NORMAL H-REFLEX LATENCIES IN CHILDREN (MSEC)

	TIBIAL	MEDIAN	ULNAR
	17.37 ± 1.23	18.67 ± 1.71 (w) (25/25) 15.81 ± 1.15 (e)	18.66 ± 1.48 (w) (25/25) 16.24 ± 0.93 (e)
0–3 months	16.01 ± 1.23	17.15 ± 0.92 (w) (12/20) 14.99 ± 0.94 (e)	17.25 ± 1.93 (w) (9/20) 15.08 ± 1.85 (e)
4–6 months	15.73 ± 1.19	16.95 ± 1.12 (w) (12/20) 14.64 ± 0.82 (e)	16.74 ± 0.77 (w) (7/20) 14.68 ± 0.67 (e)
6–12 months	15.92 ± 1.28	15.75 ± 1.14 (w) (7/20) 14.23 ± 0.51 (e)	16.49 ± 1.01 (w) (4/20) 14.32 ± 0.50 (e)
1–3 years	16.91 ± 1.46		
4–6 years	18.76 ± 1.71		
7–14 years	22.00 ± 1.97		
18–30 Years	28.04 ± 1.68		
Side-to-side difference	0.56 ± 0.37 (1.3)		

Data are presented as means ± standard deviation

The tibial H-reflex was elicited by submaximal intensity of stimulus over the posterior tibial nerve at the knee with recording over the soleus distally measured one-half the distance from the stimulation point to the medial malleolus

The median and ulnar H-reflex was elicited in infants with proportions showing a response shown in parenthesis

Side-to-side difference shows mean ± SD (upper limits of normal)

Source: Adapted from Ref. (12). Cai F, Zhang J. Study of nerve conduction and late responses in normal Chinese infants, children, and adults. *J Child Neurol.* 1997;12(1):13–18.

children with less soft tissue separating nerves. For example, volume conduction can produce simultaneous stimulation of both the median and ulnar nerves at the wrist or at the elbow. Such volume conduction should always be suspected when higher stimulation intensities or durations are utilized and when CMAP configurations show an initial positive deflection or a multiple peak configuration.

SHOCK ARTIFACT

Shock artifact is a common problem with smaller subjects because of short distances between the stimulator and recording electrodes. This may be particularly problematic with distal stimulation. The ground electrode should

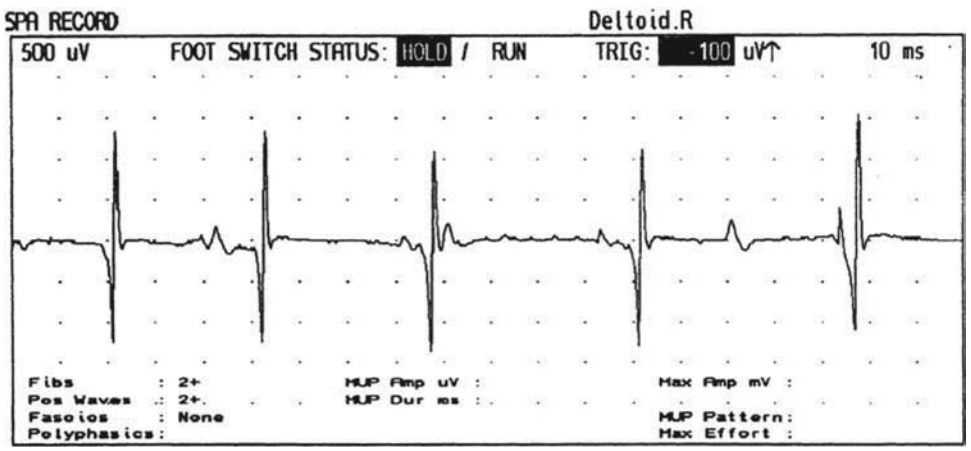

FIGURE 6.1 Neuropathic recruitment of the deltoid in a 12-month-old child with a brachial plexus injury sustained at birth. The initial recruited motor unit action potential is 2,500 μV, and it is firing at 25 Hz.

be placed between the stimulating and recording electrodes and, in infants, often a standard 6 mm silver disk or ring electrode can be placed around the wrist or ankle. Alternatively, the ground disk may be taped to the dorsal surface of the hand. Other approaches to minimize shock artifact in young children include the utilization of pumice paste to reduce skin impedance and permit suprathreshold stimulation with lower electrical currents, use of a minimal amount of conduction gel or cream, and rotation of the proximal anode in relation to the distal cathode.

MEASUREMENT OF DISTANCES/MEASUREMENT ERROR

Distance measurements must be extremely meticulous during pediatric electrodiagnostic evaluations. Segment studies are often of the order of 6 to 10 cm in length. A measurement discrepancy of only 1 cm may produce as much as a 10% to 15% conduction velocity error.

STIMULATING ELECTRODES

For neonates and young infants, small stimulators with short interelectrode distances are commercially available and simplify the testing of short nerve segments over small extremities (Figure 6.2). The stimulation intensity may be reduced by the use of a small monopolar needle electrode as the stimulating cathode with a more proximal surface anode in close proximity. For example, for ulnar orthodromic sensory studies, the author has utilized ring electrodes on the fifth digit and recording electrodes over the ulnar nerve at the elbow. Generally, a standard bipolar stimulator may be utilized for children 6 months of age and older.

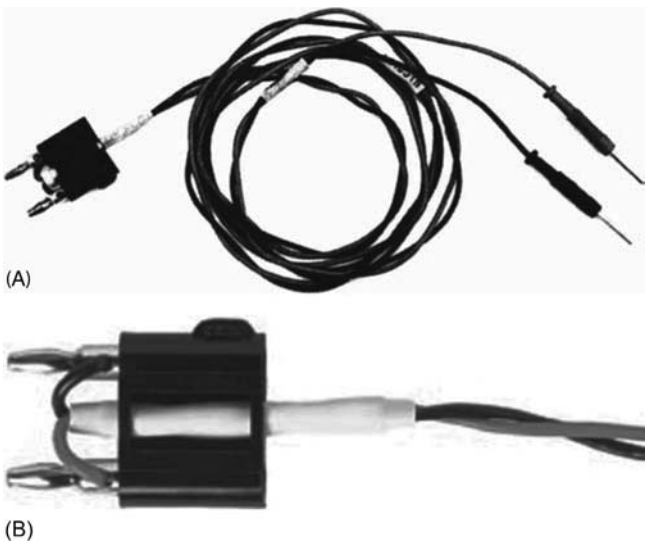

FIGURE 6.2 Pediatric nerve stimulator (A). The interelectrode distance between cathode and anode is less than 2 centimeters (B).

RECORDING ELECTRODES

Sensory Conduction

Generally, SNAPs are easily recorded in newborns. The standard ring electrodes, needle recording electrodes, and/or pediatric-size finger-clip electrodes may be used. While for adults, a 4 cm interelectrode distance is optimal, this is not possible in small children. Hence, the pediatric electrodiagnostic clinician should attempt to obtain as much distance as possible between active and reference electrodes. Every attempt should be made to obtain at least a 2 cm interelectrode distance. Stimulation of the digits, palm, or wrist with electrodes located more proximally at the elbow for median and ulnar sensory studies provides longer distance and less measurement error. In general, normative data for sensory NCVs are more readily available than normative data for distal latencies at specific distances.

Motor Conduction

Generally, standard 6 mm silver disk surface electrodes are used as active and reference electrodes for motor conduction studies. Some electrode diagnosticians prefer the use of ring electrodes on digits as the reference electrode and a standard surface electrode over the Moro point at the muscle as the active electrode (see Figure 6.3). Often 4 to 6 cm distances are used from the stimulator to the active electrode. CVs and CMAP amplitudes are generally more relevant data in infants than motor distal

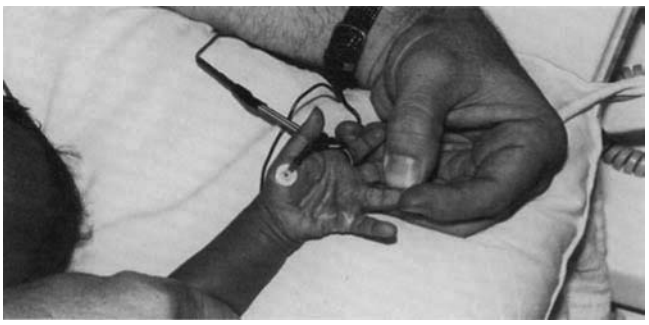

FIGURE 6.3 Recording electrodes for a median motor nerve conduction study in a small child. The active electrode is placed over the abductor pollicis brevis on the thenar eminence. The recording electrode is a ring electrode placed on the index finger. The ground electrode is a 6-millimeter silver disc electrode placed on the back of the hand.

latencies because distal nerve entrapments are rare. Thus, the distances used from distal stimulation to the active electrode are less critical.

SPECIAL CONSIDERATIONS FOR NERVE CONDUCTION STUDIES

The best normative data for pediatric NCSs are available for the median, ulnar, peroneal, tibial, facial, and phrenic motor nerves and the median, ulnar, and sural sensory nerves. Stimulation of the posterior tibial nerve (recording abductor hallucis brevis) produces a discrete CMAP more commonly than stimulating the peroneal nerve (recording over extensor digitorum brevis). The EDB muscle may be difficult to visualize or palpate in infants. Its CMAP configuration frequently has either an initial positivity or a low broad configuration. In addition, the CMAP amplitude may change substantially with slight changes in position for the active electrode over the extensor digitorum brevis.

The axillary and musculocutaneous motor NCSs may be helpful in the setting of infantile brachial plexopathy. Care should be taken to minimize volume conduction. Often, the intact side is used for amplitude comparisons.

Evaluations of proximal nerves such as the axillary spinal accessory musculocutaneous and femoral are often useful in the evaluation of severe demyelinating neuropathies (see Figure 6.4). The distal latencies of these nerves may be severely prolonged on the setting of severe reductions in the CMAPs of more distal nerves due to conduction block or axon loss.

Percutaneous stimulation of the phrenic nerve is performed with techniques similar to those utilized in the adult, with stimulation performed at the posterior border of the sternocleidomastoid at the level of the thyroid cartilage or alternatively just medial (or occasionally lateral) to the sternal head of the sternocleidomastoid. Recording electrodes may be placed in the fifth to sixth intercostal space 2 cm apart at the anterior axillary line or alternatively an active electrode may be placed immediately below the costal margin at the level of the nipple with the recording electrode at the xiphoid. The active electrode may need to be moved to adjacent positions to obtain an optimal M-wave (see Figure 6.5). Normative values for phrenic latencies have been reported in children (29,30). The author prefers to use ultrasound visualization of the diaphragm simultaneously with phrenic nerve stimulation to confirm downward deflection of the diaphragm. Volume conduction to the long thoracic nerve may produce a CMAP from the serratus anterior rather than the diaphragm. The downward deflection of the diaphragm spontaneously and with electrical stimulation may be confirmed, and the distance of diaphragmatic excursion quantitatively measured by ultrasound M-mode (31).

REPETITIVE NERVE STIMULATION STUDIES

Every attempt should be made to stabilize the extremity with an infant or pediatric-size arm board. The author prefers to use a block electrode or surface cathode and anode electrodes taped over the nerve as opposed to a handheld stimulator. This helps standardize each

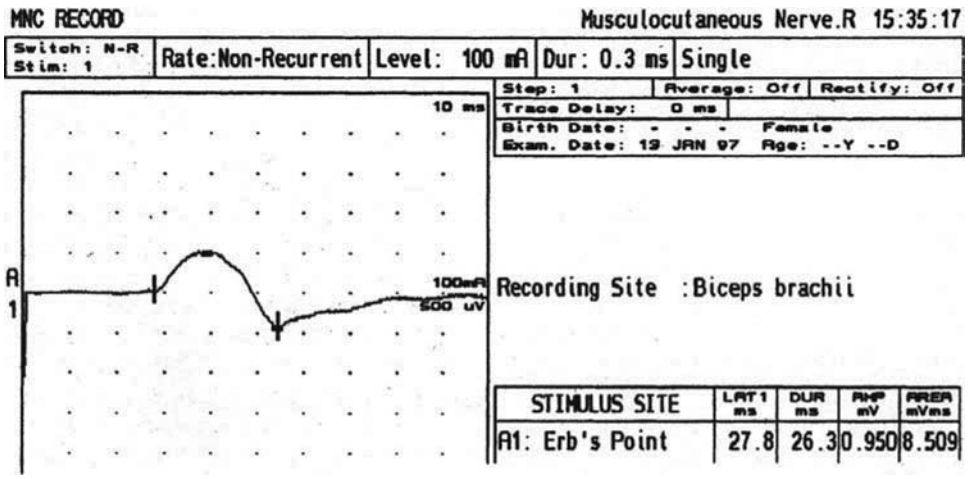

FIGURE 6.4 Nerve conduction study of the musculocutaneous nerve in Charcot-Marie-Tooth (CMT) type III. The nerve is stimulated at Erb's point and the recording electrode is placed over the biceps brachii. Distal latency is severely prolonged at 27.8 milliseconds. Note the reduced compound muscle action potential amplitude, presumably due to conduction block, and the relative lack of temporal dispersion, which is frequently seen in CMT.

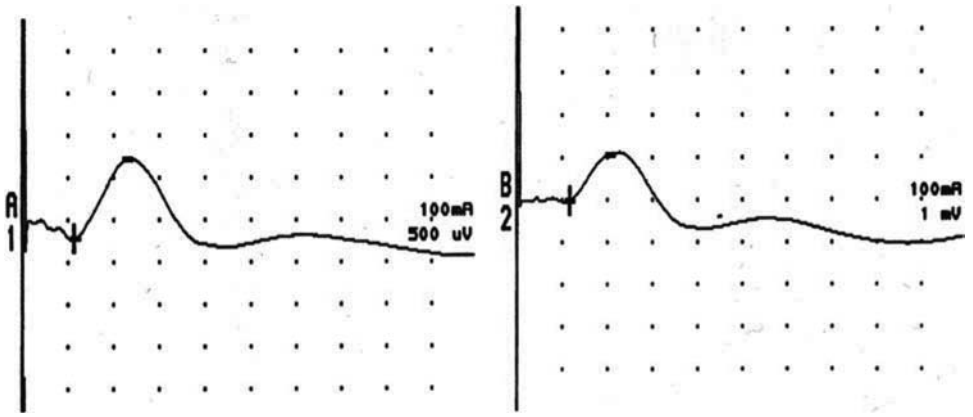

FIGURE 6.5 Phrenic nerve conduction study in a 13-year-old child with C2 traumatic spinal cord injury. A1 is the compound muscle action potential (CMAP) amplitude obtained on the right side and B2 is the CMAP obtained on the left side. Latencies are approximately 5 milliseconds and amplitudes from baseline to peak 1 mV. The viability of the phrenic nerves allowed placement of a phrenic nerve–diaphragm pacer for ventilation.

stimulation during a train of five stimuli at low or high rates of stimulation. In newborns, the author prefers to stimulate the median or ulnar nerve at the elbow to minimize shock artifact. Care should be taken to obtain a stable baseline between stimulations in a train. Decrements or increments in amplitude should be accompanied by similar decrements or increments in the area. If no concomitant area changes occur, then technical factors (changing baseline or changing temporal dispersion) may explain a decrement or increment in amplitude.

TECHNICAL FACTORS OF NEEDLE ELECTROMYOGRAPHY

ELECTRODES

Generally, 26 to 28 gauge Teflon-coated monopolar electrodes, usually 25 mm in length, are utilized. Some laboratories routinely use disposable concentric facial needle electrodes. These electrodes have smaller calibrated recording areas and hence provide more stability of MUAP configuration. In addition, concentric needle electrodes are more sensitive to changes in duration and amplitude than monopolar needle electrodes. Use of smaller electrodes (either small monopolar needles or small diameter concentric needle electrodes originally designed for the examination of adult facial muscles) provides considerable psychological advantages in children of sufficient developmental age to associate needles with pain. The instrumentation utilized for needle EMG of children is essentially the same as that used in adults. In the Intensive Care Unit, electrical interference may necessitate the use of either a facial concentric needle or a needle reference electrode. Long electrodes or long electrode leads can create problems with ambient electrical interference.

OPTIMAL MUSCLES TO STUDY FOR REST ACTIVITY

In evaluating an infant or young child for a generalized disorder, specific muscles are chosen to permit evaluation of insertional and spontaneous activity. The distal hand (first dorsal interosseous) and foot muscles of infants usually have minimal voluntary activity due to immature motor control at this developmental age, making them good sites to assess spontaneous activity. In addition, extensor muscles such as the vastus lateralis and gastrocnemius in the legs and the triceps in the upper extremities are useful sites for the evaluation of insertional and spontaneous activity.

In the neonate and young infant, foot and hand intrinsic muscles exhibit high levels of endplate noise because of the relatively larger endplate area in the immature muscle. This endplate activity may be confused with fibrillation potentials. Fibrillation potentials and positive sharp waves are not typically observed in the full-term normal newborn.

OPTIMAL MUSCLES FOR EVALUATION OF RECRUITMENT, MOTOR UNIT CONFIGURATION, AND INTERFERENCE PATTERN

In general, flexor muscles such as the tibialis anterior and the iliopsoas are useful for the evaluation of MUAPs and recruitment in the lower extremity. These muscles can be activated by tickling or pinching the bottom of the foot, producing a withdrawal response. In the upper extremity, the flexor digitorum sublimis and biceps muscles are frequently reflexively activated by the newborn or young infant. More proximal muscles can be activated by moving the extremity or positioning the extremity to produce antigravity stabilization of the limb by the firing

of proximal musculature. Alternatively, reflex posturing techniques such as the Moro response can be used to activate the shoulder abductors but are usually not necessary.

SEDATION

Pediatric physiatrists and neurologists performing pediatric electrodiagnostic evaluations have noted that extreme behavioral distress most frequently occurs among 2 to 6 year olds (32,33). Pain medications are occasionally or always prescribed by 50% of pediatric electromyographers (33). General anesthesia is occasionally utilized by 25% of electrodiagnostic practitioners (33). One study demonstrated that children exhibiting more behavioral distress during pediatric electrodiagnostic evaluations were younger, had been uncooperative with previous painful procedures, were more likely to have had more negative medical/dental experiences, and had mothers who themselves reported greater fear and anxiety about undergoing EMG/NCSs (32).

While some electromyographers never utilize sedation, there has been more interest in the use of analgesia, conscious and deep sedation, and more recently, general anesthesia with propofol or inhalational anesthetics. Traditional sedative choices include chloral hydrate (50–100 mg/kg), "DPT" (Demerol [meperidine hydrochloride], Phenergan [promethazine], and Thorazine [chlorpromazine]) and midazolam hydrochloride nasal spray. EMLA cream (lidocaine 2.5% and prilocaine 2.5%) has been used during electromyographic evaluations as a topical anesthetic (34). The mean duration of topical application in infants or older children was 45 to 145 minutes. Greater pain relief was obtained with the use of EMLA over the extensor forearm than the thenar eminence.

While general anesthesia is usually not necessary, the author has increasingly involved critical care and anesthesia colleagues who have utilized either propofol (2,6-diisopropylphenol), an intravenous sedative–hypnotic agent, or inhalational anesthetics with laryngeal mask anesthesia (LMA) airways for the electrodiagnostic evaluation of 18-month-old infants to 6-year-old children who exhibit substantial behavioral distress during an initial attempt at an electrodiagnostic evaluation without sedation. Propofol produces rapid onset of anesthesia (in 1–3 minutes) and sedation is maintained by either a continuous infusion or multiple boluses. Subjects usually awaken in less than 10 minutes from the time the infusion is discontinued. Sedation, analgesia, and particularly, general anesthesia have inherent risks and require appropriate monitoring. Propofol should be administered by an anesthesiologist or pediatric intensivist prepared to bag-mask ventilate or intubate the child if necessary. Adequate monitoring generally requires a sedation suite, pediatric ICU, and a recovery room or operating room. The author typically obtains all NCSs and a thorough examination of multiple muscle sites for abnormal spontaneous rest activity while the subject is deeply sedated or anesthetized with propofol. The level of sedation is then titrated to a point where appendicular movement is elicited with needle insertion or stimulation of the extremity. At this point, under lighter sedation, recruitment pattern and motor unit configuration are assessed. As the child awakens, interference pattern is evaluated with more vigorous motor activity. Children are usually amnestic to the EMG examination subsequent to propofol anesthesia.

The cost of anesthesia must be weighed against the importance of the acquisition of a thorough, technically precise, and accurate electrodiagnostic evaluation. An EMG obtained under anesthesia usually provides a suboptimal evaluation of motor unit configuration, recruitment pattern, and interference pattern with maximal effort, but better evaluation of quiet muscle for spontaneous activity and a more comprehensive acquisition of NCSs and repetitive nerve stimulation studies.

The key to successful data acquisition in most pediatric electrodiagnostic evaluations remains a well-organized, well-planned approach with distinct diagnostic questions prospectively considered. If the examination is planned to answer a specific question, it is usually possible to proceed expeditiously, completing the examination within a reasonable time (30 minutes). As children approach 6 years of age, it becomes easier to talk them through an evaluation and elicit their participation and cooperation.

NCSs are usually better tolerated than needle electromyography and many pediatric electromyographers perform NCSs first. Increased behavioral distress, subsequent to a needle examination, makes the motor nerve conductions and particularly, sensory NCSs, technically difficult due to excessive EMG background noise.

LIMITATIONS OF SINGLE-FIBER EMG

While normative data for fiber density, mean consecutive difference, and jitter have been reported for different muscles among different pediatric age groups (35), this procedure is difficult to use in younger children with limited ability to cooperate. Alternatively, a stimulated single-fiber EMG study may be obtained under general anesthesia in those suspected of a congenital myasthenic syndrome, and this technique has yielded excellent sensitivity and specificity for the identification of a neuromuscular transmission disorder (36–38).

SPECIFIC CLINICAL PROBLEMS IN PEDIATRIC ELECTRODIAGNOSIS

ELECTRODIAGNOSTIC EVALUATION OF THE FLOPPY INFANT

The most common referral for an electrodiagnostic examination in the infant is generalized hypotonia. The most common etiology for infantile hypotonia is central,

accounting for approximately 80% of cases. A differential diagnosis of infantile hypotonia is shown in Table 6.8 (39). Electrodiagnostic abnormalities in selected conditions producing infantile hypotonia are shown in Table 6.9.

Neurogenic causes of generalized weakness in infants are more accurately diagnosed with EDSs than are myogenic causes (40–42). A study of the predicted value of the electrodiagnostic examination in the hypotonic infant showed that EDSs accurately predicted the diagnosis in 65% of infants with spinal muscular atrophy (SMA) and only 10% of infants with myopathy. Seventy-five percent of the EDSs performed on infants with documented

TABLE 6.8 DIFFERENTIAL DIAGNOSIS OF INFANTILE HYPOTONIA

Cerebral hypotonia
Chromosome disorders
- Trisomy
- Prader–Willi syndrome

Static encephalopathy
- Cerebral malformation
- Perinatal CNS insult
- Postnatal CNS insult

Peroxisomal disorders
- Cerebrohepatorenal syndrome (Zellweger syndrome)
- Neonatal adrenoleukodystrophy

Inborn errors of metabolism
- Glycogen storage disease type II (Pompe disease)
- Infantile GM1 gangliosidosis
- Tay–Sachs disease (infantile GM2 gangliosidosis)
- Vitamin dependency disorders

Amino acid and organic acid disorders
- Maple syrup disease
- Hyperlysinemia
- Nonketotic hyperglycinemia
- Propionyl-CoA carboxylase deficiency

Other genetic disorders
- Familial dysautonomia
- Cohen syndrome
- Oculocerebrorenal syndrome (Lowe)

Benign congenital hypotonia

Spinal cord
Trauma (obstetrical; postnatal)
- Hypotonia early with acute paraplegia
- Hypertonia

Tumor or arteriovenous malformation (AVM)
- Hypertonia may occur later or with slow growing tumor

Anterior horn cell
- Spinal muscular atrophy (SMA) type I (Werdnig–Hoffmann)
- SMA type II
- Distal SMA with vocal cord paralysis and diaphragm weakness
- Poliomyelitis
- Neurogenic arthrogryposis

Polyneuropathies
Congenital hypomyelinating neuropathy
Chronic inflammatory demyelinating polyneuropathy
Acute inflammatory demyelinating polyradiculoneuropathy (Guillain–Barré syndrome)
Hereditary motor–sensory neuropathies
- Dejerine-Sottas
- Congenital hypomyelinating neuropathy

Toxic polyneuropathy
Leukodystrophies (Krabbe; Niemann–Pick)
Leigh's syndrome
Giant axonal neuropathy
Dysmaturation neuropathy

Neuromuscular junction
Presynaptic
- Infantile botulism
- Hypermagnesemia—eclampsia
- Aminoglycoside antibiotics
- Congenital myasthenia
  - Choline acetyltransferase (CHAT) deficiency
  - Paucity of acetylcholine synaptic vesicles
  - Congenital Lambert–Eaton-like syndrome
  - Decreased quantal release

Synaptic basal lamina defects
- Congenital myasthenic syndrome
  - Endplate acetylcholinesterase (AChE) deficiency

Postsynaptic
- Neonatal (autoimmune)
- Congenital myasthenia
  - Acetylcholine receptor (AChR) disorders involving α, β, δ, e receptor subunits
  - AChR deficiency causing kinetic abnormalities in function
  - AChR slow-channel syndromes
  - AChR fast-channel syndromes
  - Endplate rapsyn deficiency

Myopathies
Congenital myopathies
- Nemaline rod
- Central core
- Myotubular (centronuclear)
- Minicore (multicore)
- Congenital fiber type disproportion

Congenital myotonic dystrophy (DM1)
Congenital muscular dystrophy
- Fukuyama type (central nervous system [CNS] involvement)
- Merosin deficiency (with or without CNS involvement)
- Ullrich congenital muscular dystrophy (collagen VI deficiency, scleroatonic)
- Congenital muscular dystrophy with early spine rigidity
- Muscle–eye–brain disease
- Walker–Warburg Syndrome
- Undifferentiated

Inflammatory myopathies
- Infantile polymyositis

Metabolic myopathies
- Acid maltase deficiency (type II)
- Muscle phosphorylase deficiency (type V)
- Phosphofructokinase deficiency (type VII)
- Cytochrome c oxidase
- Carnitine deficiency

Endocrine myopathies
- Hypothyroidism
- Hypoparathyroidism

TABLE 6.9 INFANT HYPOTONIA: ELECTRODIAGNOSTIC ABNORMALITIES

DIAGNOSIS	MOTOR CONDUCTION	SENSORY CONDUCTION	SPONTANEOUS ACTIVITY	MOTOR UNITS
SMA	Decreased amplitude; may show decreased velocity	Normal	Fibrillation ±; spontaneous rhythmic motor unit firing	Decreased number; may show mild increase in amplitude, duration
HMSN III	Markedly prolonged	Prolonged or absent	0	Reported normal
Hypomyelinating neuropathy	Markedly prolonged; markedly decreased amplitude	Prolonged or absent	0	Reported normal or increased amplitude
Inflammatory polyneuropathy	Decreased amplitude; possibly decreased velocity; conduction block	±	Fibrillation may be present	Decreased number
Botulism	Decreased amplitude; normal velocity; incremental response to repetitive nerve stimulation studies with rates > 20 Hz	Normal	Fibrillations	Decreased amplitude, duration
Spinal cord injury	Normal motor velocity and amplitudes if nerves tested are not originating from area of injury; F-wave or H-reflex may be prolonged or absent	Normal	Fibrillations may be present in muscles innervated at level of injury	Decreased number at involved muscles; poor motor control below level of injury
Congenital myopathy	Normal velocity; amplitude may be decreased	Normal	Fibrillations may be present (in congenital myotubular myopathy)	Normal to decreased amplitude, duration; increased polyphasicity
Congenital myotonic dystrophy	Normal	Normal	Absent or few fibrillations	Poor activation; likely normal
Glycogen storage disease	Normal	Normal	Fibrillations (in types II, V, VII); frequency-varying; trains of positive waves	Decreased amplitude, duration
Metachromatic leukodystrophy	Decreased velocity; decreased amplitude	Slowed		

Abbreviation: HMSN III, Hereditary motor sensory neuropathy III
Source: Adapted from Turk, MA. Pediatric electrodiagnostic medicine. In: Dumitru D, ed. *Electrodiagnostic Medicine*. Philadelphia, PA: Hanley & Belfus; 1995:1133–1142.

myopathies were considered normal (43). The sensitivity of EMG improves after age 2 (42).

In arthrogryposis multiplex congenita and hypotonia, neither muscle biopsy nor NCS/EMG alone had consistently high sensitivities, positive predictive values, or specificities (44). When the clinical evaluation indicates a specific syndromic, developmental, or exogenous cause, NCS/EMG and muscle biopsy are not helpful and may not need to be performed. When the history, examination, and genetic evaluation are unrevealing, NCS/EMG and muscle biopsy together provide valuable diagnostic information.

In the evaluation of hypotonia, a complete electrodiagnostic evaluation is useful, including motor and sensory NCSs and appropriate needle examination with the highest yield muscles examined initially, and, if necessary, repetitive nerve stimulation. It should be emphasized that NCSs and electromyography are an extension of the clinician's physical examination. Electrodiagnostic findings need to be interpreted in light of clinical examination findings. Care should be taken not to overinterpret subtle findings on needle electromyography. Low-amplitude, short-duration, polyphasic MUAPs, which would be considered myopathic in adults, may be normal in young children. Motor unit amplitudes and durations may be reduced in the normal young child and mistaken for myopathic MUAPs. Endplate noise, abundant in the small intrinsic muscles of the hand and foot may be difficult to distinguish from fibrillation potentials. Thus, borderline findings on needle EMG should not be overinterpreted in the infant and young child.

Parents should be cautioned prior to an electrodiagnostic evaluation that definitive diagnostic information is often not obtained, and the results may help guide further diagnostic studies. For example, results from EMG may help to guide further studies such as muscle biopsy by providing information about the most appropriate muscle site for the biopsy. With SMA, an electrodiagnostic evaluation can allow the clinician to defer a muscle biopsy and proceed with molecular genetic studies of the survival motor neuron (SMN) gene. Often the SMN gene is ordered prior to any EDSs being performed, so fewer studies have been performed on this population over the past decade. EDSs in patients with hereditary motor–sensory neuropathy help to categorize the neuropathy as either primarily demyelinating or axonal, and such information may help focus subsequent molecular genetic analyses. In general, nerve conduction and electromyography still provide a useful tool for the localization of lesions within the lower motor neuron, but fewer studies have been required as genetic studies have become commercially available.

DIFFERENTIAL DIAGNOSIS FOR EARLY RESPIRATORY DISTRESS IN INFANCY

The differential diagnosis of lower motor neuron disorders with perinatal respiratory distress is fairly limited. Generally, respiratory distress within the first few days of life can be seen in SMA type I, congenital hypomyelinating neuropathy, congenital myasthenia, transient neonatal myasthenia, congenital myotonic muscular dystrophy, neurogenic arthrogryposis, and x-linked myotubular myopathy. These disorders are easily differentiated with EDSs and in some instances molecular genetic findings. For example, congenital myotonic muscular dystrophy may be definitively diagnosed with molecular genetic studies at the chromosome 19q13.3 locus. In congenital hypomyelinating neuropathy, sensory conduction abnormalities are unrecordable and motor NCVs are markedly slowed down (2–5 m/s) with temporal dispersion and low-amplitude evoked potentials (see Figure 6.6). SMA patients show normal sensory conductions, decreased CMAP amplitudes, occasional fibrillations, and decreased numbness of MUAPs. Congenital myasthenia patients show normal sensory conductions, normal motor NCVs, and abnormalities on repetitive nerve stimulation studies. X-linked myotubular myopathy patients show profuse fibrillations and myopathic MUAPs on EMG and diagnosis is confirmed by muscle biopsy.

ACUTE ONSET INFANTILE HYPOTONIA

Acute onset hypotonia in a previously normal infant should warrant an evaluation to rule out acute inflammatory demyelinating polyneuropathy (AIDP), infantile botulism, infantile polymyositis, an infantile form of myasthenia, a toxic process, or acute onset myelopathy. Repetitive motor nerve stimulation studies should be performed under the following circumstances: (a) there is constipation, bulbar involvement, and/or respiratory distress; (b) an infant presents with ptosis or extraocular muscle weakness; (c) CMAP amplitudes are severely reduced; (d) "myopathic" MUAPs are present; (e) a repetitive CMAP is observed after single supramaximal stimulation on routine NCS, suggestive of a diagnosis of congenital myasthenia with congenital acetylcholinesterase (AChE) deficiency or classic slow-channel syndrome.

MOTOR NEURON DISORDERS (EG, SMA)

SMA is perhaps the most common lower motor neuron disorder causing infantile hypotonia. The predictive value of needle electromyography (EMG) in the diagnosis of SMA has been established (40–43), but the need for EDSs has diminished over the years, given the 95% or greater sensitivity of SMN gene studies. As SMA remains an important consideration in infantile hypotonia, a review of the electrodiagnostic findings is useful.

The findings in this motor neuron disorder have largely been consistent with motor axonal loss, denervation, and (among persons less severely affected)

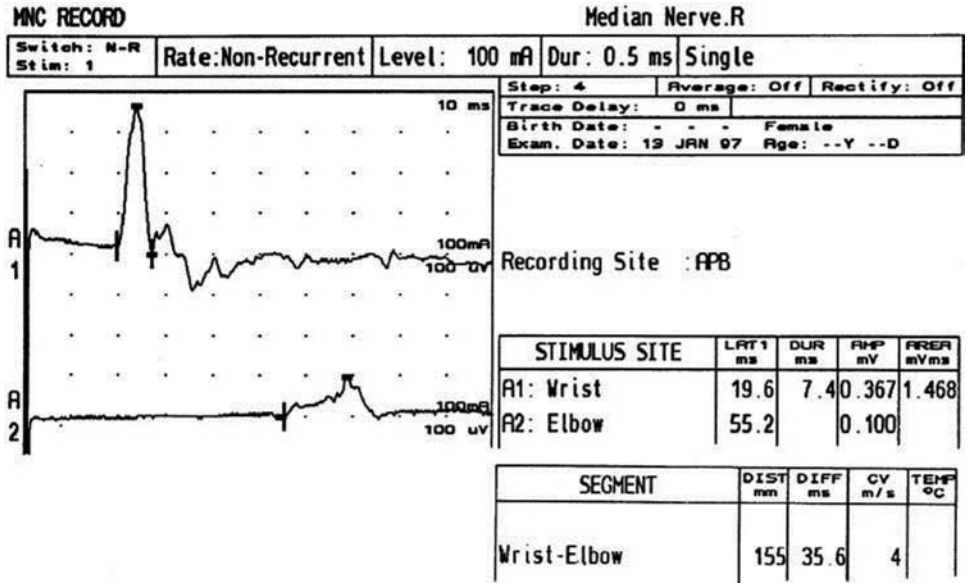

FIGURE 6.6 Median nerve conduction in a 5-year-old child with congenital hypomyelinating neuropathy documented by sural nerve biopsy and molecular genetic studies of the EGRF 2 gene. Distal latency is markedly prolonged at 19.6 milliseconds. There is reduced compound muscle action potential amplitude, at 0.367 mV, conduction block (note the drop in amplitude from distal to proximal), and conduction velocity at 4 m/s.

reinnervation. Traditional electrodiagnostic criteria for motor neuron disease are not suitable for patients with childhood SMA. For example, Buchthal (45) found that many infants with SMA did not meet strict criteria for motor neuron disease. If clinical findings suggest SMA, study of at least two muscles innervated by different nerve roots and peripheral nerves in at least three extremities is indicated (46). In the infant, spontaneous activity may be more readily determined with the study of muscles that are not as commonly recruited, such as the vastus lateralis, gastrocnemius, triceps, and first dorsal interosseous. Recruitment and motor unit characteristics can be assessed in muscles that are readily activated such as the anterior tibialis, iliopsoas, biceps, and flexor digitorum sublimis (45). The paraspinal muscles are usually not studied due to poor relaxation, and the experienced pediatric electrodiagnostic medicine consultant usually defers needle evaluation of the tongue in the hypotonic infant.

Although some authors (46) have described high-density fibrillation potentials in infants with poorer outlook, most studies have not demonstrated abundant fibrillation potentials in the infantile form (46–48). In SMA III, the incidence of fibrillation potentials ranged from 20% to 40% in one series (49) to 64% in another (50). The incidence of fibrillation potentials in SMA III does not approach the level seen in SMA 1. Additionally, spontaneous activity has been more frequently observed in the lower extremities than upper limbs, and proximal more than distal muscles in SMA III (49). The degree of spontaneous activity has not been found to be independently associated with a worse prognosis in SMA (43). Fasciculations are uncommonly observed in SMA Type I and appear more commonly in SMA II and III (46–48). In younger patients, fasciculations are difficult to distinguish from spontaneously firing MUAPs. In relaxed muscles, some motor units exhibit a spontaneous rhythmic firing (47,48,51).

Voluntary MUAPs frequently fire with an increased frequency although recruitment frequency may be difficult to determine consistently in infants. Compared to age-matched norms, MUAPs show longer duration, particularly in older subjects, and higher amplitude; however, a bimodal distribution may be seen with some concomitant low-amplitude short-duration potentials (47). Large-amplitude, long-duration MUAPs may be absent in many infants with SMA I but more commonly observed in SMA II and III (46). The percentage of large-amplitude MUAPs increase with the duration of the disease (48). Other signs of reinnervation, such as polyphasic MUAPs may be observed in more chronic and mild SMA. These polyphasic MUAPs may include late components such as satellite or nascent potentials. There may also be temporal instability of the waveform observed in individual MUAPs. Reduced recruitment (an incomplete interference pattern) with maximal effort is perhaps the most consistent finding in all SMA types (see Figure 6.7). In one series (43), the amplitude of MUAPs and degree of decrement in recruitment pattern were not individually associated with worse prognosis.

Motor NCVs and CMAP amplitude have been shown to be reduced in many patients with infantile SMA. The degree of motor conduction slowing (if present) tends to be mild and greater than 70% of the lower limit of normal (48,50,52–54). Reduction of motor conductions to

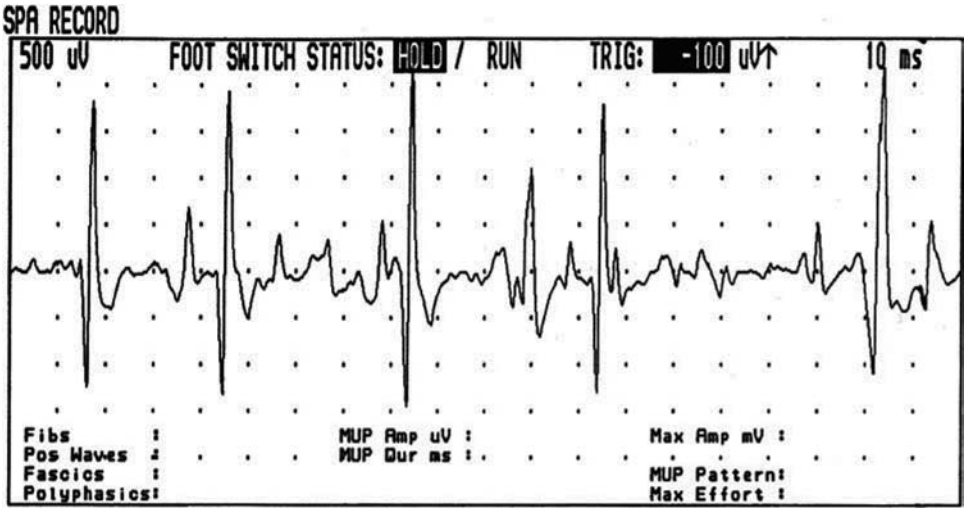

FIGURE 6.7 Incomplete or reduced interference pattern in spinal muscular atrophy type II. Note the large amplitude motor unit action potential (3,000 μV) firing at 25 Hz.

less than 70% of the lower limit of normal is described as an exclusionary criterion for SMA (55). The mild slowing of motor conductions is present to the same degree over distal and proximal segments as determined by M- and F-wave responses (53). The slowing of conduction is generally seen in those with correspondingly low-amplitude CMAPs and is thought to be due to selective loss of the fastest conducting fibers from large motor units. Alternatively, arrested myelination in utero has been proposed to explain this slowing in motor conduction noted in some SMA cases at birth (43). Survival has been found to be longer for those SMA infants with normal MCVs over a distal segment (43). Significant reductions in CMAP amplitudes have been frequently reported for SMAs I to III (43,46,50). Kuntz (50) reported a tendency toward greater reductions in CMAP amplitude among patients with earlier age of onset and shorter survival.

Sensory NCSs in SMA show essentially normal sensory CVs and SNAP amplitudes. Significant abnormalities in sensory studies exclude a diagnosis of SMA (55), while minor abnormalities in sensory CVs have infrequently been noted in SMA (52,56,57). Such rare sensory abnormalities have not been reported in SMA patients with diagnostic confirmation by molecular genetic studies.

CMAP amplitude and motor unit number estimation (MUNE) have increasingly been used in recent years as potential biomarkers for clinical trials in SMA. In one study (58), denervation was assessed in 89 SMA I, II, and III subjects via MUNE and maximum compound muscle action potential (CMAP) studies, and results correlated with SMN2 copy, age, and function. MUNE and maximum CMAP values of the ulnar nerve were distinct among SMA subtypes. Changes in MUNE and maximum CMAP values over time were dependent on age, SMA type, and SMN2 copy number. SMN2 copy number less than 3 (consistent with SMA I) was correlated with lower MUNE and maximum CMAP values and worse functional outcomes. As SMN2 copy number increased, so did functional status. Change in MUNE longitudinally over the time intervals examined in this study was not statistically significant for any SMA cohort. However, a decline in maximum CMAP over time was apparent in SMA2 subjects. Age-dependent decline in MUNE and maximum CMAP was apparent in both SMA I and SMA II subjects, with age as an independent factor regardless of type. Maximum CMAP at the time of the initial assessment was most predictive of functional outcome. Prospective longitudinal studies in four prenatally diagnosed infants demonstrated significant progressive denervation in association with symptomatic onset or functional decline. In another study (59), MUNE values correlated with Hammersmith Functional Motor Scale (HFMS) scores. Increased single motor unit action potential (SMUP) amplitude values correlated with decreased HFMS scores. The study confirmed that the MUNE method is a useful tool reflecting motor unit loss in SMA, and it is easy to perform and well tolerated.

In a recent longitudinal study, 62 children with SMA types II and III were observed prospectively for up to 42 months (60). Longitudinal electrophysiologic data were collected, including compound motor action potential (CMAP), SMUP amplitude, and MUNE. Significant motor neuron loss and compensatory collateral reinnervation were noted at baseline. Over time, there was a significant mean increase in MUNE (4.92 units/year), a mean decrease in SMUP amplitude (−6.32 μV/year), and stable CMAP amplitude. The unexpected longitudinal results differed from findings in amyotrophic lateral sclerosis studies, perhaps indicating that compensatory

processes in SMA involve new motor unit development. A better understanding of the mechanisms of motor unit decline and compensation in SMA will be important for assessing novel therapeutic strategies and for providing key insights into disease pathophysiology. MUNE and CMAP amplitudes seem to be sensitive parameters reflecting motor dysfunction in SMA but additional longitudinal studies are needed.

To investigate these measures as biomarkers of treatment response, MUNE and sciatic CMAP measurements were obtained in SMNΔ7 mice treated with antisense oligonucleotide (ASO) or gene therapy (61). ASO-treated SMNΔ7 mice were similar to controls at days 12 and 30. CMAP reduction persisted in ASO-treated SMNΔ7 mice at day 12 but was corrected at day 30. Similarly, CMAP and MUNE responses were corrected with gene therapy to restore SMN. SMN restoring therapies result in preserved MUNE and gradual repair of CMAP responses. This provides preclinical evidence for the utilization of CMAP and MUNE as biomarkers in future SMA clinical trials.

SPINAL CORD INJURY

Neonatal spinal cord injury (SCI) may occur as an obstetrical complication or as a result of a vascular insult to the spinal cord. Typical clinical presentation may include findings of diffuse hypotonia, possible respiratory distress, hyporeflexia, and urinary retention. Bilateral flaccid paralysis of the upper extremities may be a sign of neonatal SCI (62). An anterolateral SCI due to a vascular insult will produce EMG findings of severe denervation in diffuse myotomes. Typically, 2 to 3 weeks may lapse before fibrillations and positive sharp waves are elicited. Anterior horn cell and axonal degeneration will typically result in decreased CMAP amplitudes in multiple peripheral nerves. SNAP amplitudes are spared. Somatosensory evoked potentials may be spared if posterior columns are preserved.

Traumatic SCI often results in loss of anterior horn cells at a specific "zone of injury." For example, a child with C5 tetraplegia may have denervation present at the bilateral C6 and C7 myotomes. This zone of partial or complete denervation becomes particularly relevant in the evaluation of a patient for possible placement of an implanted functional electrical stimulation system for provision of voluntary grasp and release. The presence of denervation necessitates concomitant tendon transfers with electrical stimulation of the transferred muscle group.

Somatosensory evoked potentials (SSEPs) may help establish a sensory level in an infant or young child with SCI and is also useful in the evaluation of the comatose or obtunded child at risk for SCI without radiographic abnormality (SCIWORA). Somatosensory evoked potentials are discussed in the following.

Transcranial electrical MEPs to monitor the corticospinal motor tracts directly are now used routinely in addition to SSEPs for detection of emerging SCI during surgery to correct spine deformity or resect intramedullary tumors (63–67). Afferent neurophysiologic signals can provide only indirect evidence of injury to the motor tracts since they monitor posterior column function. Transcranial electrical MEPs are exquisitely sensitive to altered spinal cord blood flow due to either hypotension or a vascular insult. Moreover, changes in transcranial electrical MEPs are detected earlier than are changes in SSEPs, thereby facilitating more rapid identification of impending SCI.

BRACHIAL PLEXUS AND CERVICAL NERVE ROOT LESIONS

Traumatic obstetrical brachial plexopathy or "neonatal brachial plexus palsy" (NBPP) usually results from traction on the brachial plexus (predominantly upper trunk) and its associated spinal roots. This can lead to stretching or rupture of the trunks of the plexus and/or partial axonotmesis or avulsion of the spinal roots. The most common cause is a shoulder dystocia of the anteriorly presenting shoulder causing excessive lateral neck traction. Injury to the upper trunk of the brachial plexus, and/or C5 and C6 cervical roots is the more common injury known as Erb–Duchenne palsy. Damage to the lower trunk, and/or C8-T1 cervical roots, is referred to as Klumpke's palsy. Severe brachial plexus injuries may involve the entire plexus and C5-T1 nerve roots diffusely. Horner's syndrome due to injury of the C8 and T1 roots and the superior cervical sympathetic ganglion may be an associated clinical finding. An isolated Klumpke's palsy is rare in the setting of traumatic birth palsy and usually results from a fall onto a hyperabducted shoulder, penetrating trauma, or tumor.

EDSs help determine the location (root and/or plexus), extent, and severity of the brachial plexus injury. Examination should be deferred until at least 3 to 4 weeks after the injury to allow for abnormal spontaneous rest activity (fibrillations and positive sharp waves) to develop in the setting of denervation and axon loss (see Figure 6.8). Complete injuries are characterized electromyographically by absent MUAPs and absent CMAP amplitudes in peripheral nerves supplied by the transected axons. In the setting of total motor paralysis, motor NCSs with measurement of the amplitude of the CMAPs in distal and proximal muscles provide useful prognostic information. For example, the preservation of the CMAP amplitude 10 days or more after the injury with complete clinical paralysis suggests that the damage is, in part, a neuropraxic injury with better prognosis. In this setting, F-waves are absent. If motor function is absent and no MUAPs are observed, examination of the amplitude of the SNAPs in the dermatomal distribution

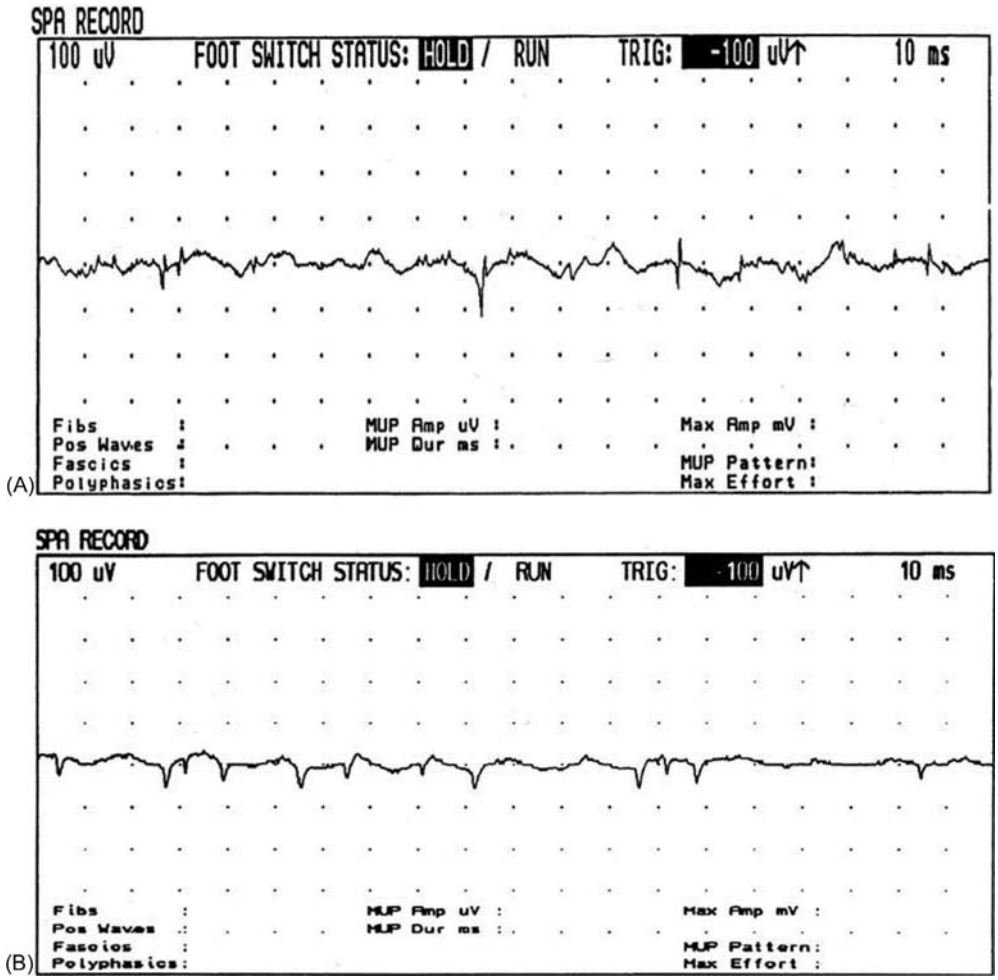

FIGURE 6.8 Fibrillation potential (A) and positive sharp waves (B) indicative of acute denervation and axon loss.

of the branches of the affected brachial plexus trunks can help distinguish injuries to the plexus from severe cervical root injuries or avulsions. The sensory dorsal root ganglion lies in the intervertebral foramen distal to the damaged segment with a root injury, leaving the sensory axon projection from the dorsal root ganglion to the limb intact. Thus, the SNAP is obtainable in the setting of a root avulsion with absent clinical sensation.

In the setting of Erb's palsy, assessment of a superficial radial sensory or median sensory response to the index finger is useful in making a distinction between a C6 root avulsion and a more distal lesion involving the trunk of the brachial plexus. The median SNAP to the middle finger provides information about the integrity of C7 axon projections distal to the dorsal root ganglion. The presence or absence of an ulnar SNAP can help distinguish a lower trunk injury from a C8 nerve root injury.

In perinatal traumatic brachial plexopathy, positive sharp waves and fibrillations, indicative of true denervation, can be found by 14 to 21 days after injury (68). The absence of fibrillations or positive sharp waves after this time frame suggests a neuropraxic lesion with intact axons. In this setting, the prognosis for recovery is favorable. Early in the course of recovery prior to reinnervation, the interference pattern usually is reduced or discrete and recruitment frequencies increased into the neuropathic range (often >20 Hz). A follow-up needle EMG evaluation 3 to 6 months after the injury is useful to determine subclinical evidence of reinnervation. Such reinnervation is typically characterized initially by "nascent" polyphasic MUAPs (see Figure 6.9). With reinnervation, the numbers of positive sharp waves and fibrillations decrease over time, amplitude of MUAPs increases as collateral spouting occurs, and with evaluation of the interference pattern, there is an observed increasing number of voluntary MUAPs.

The author prefers to initially obtain sensory NCSs (occasionally with sedation) consisting of a median sensory NCS recorded from the index finger (C6 dermatome), a median sensory NCS recorded from the middle finger (C7 dermatome), and an ulnar sensory NCS recorded from the fifth digit (C8 dermatome). Median and ulnar motor NCSs are useful to evaluate the integrity of axons traveling through the lower trunk. Axillary

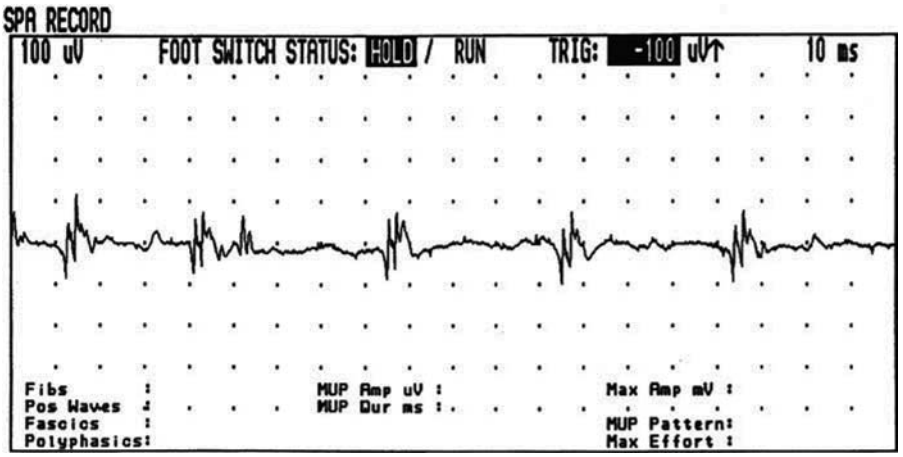

FIGURE 6.9 Polyphasic motor unit action potential (MUAP) with a neuropathic firing frequency at 25 Hz. These polyphasic MUAPs obtained 4 months after brachial plexus injury are indicative of reinnervation.

and musculocutaneous motor NCSs (with assessment of CMAP amplitudes) are useful if an upper trunk injury is suspected. These CMAP amplitudes may be compared to the intact side depending on patient tolerance of the study (69). A CMAP amplitude reduction of more than 90%, compared to the unaffected side, predicted severe weakness of the corresponding root level. During the EMG study of the deltoid, the examiner should assess the clinical sensation of the C5 dermatome. The use of dermatomal and mixed nerve SSEPs in brachial plexus injuries is discussed in the following.

In addition to a complete needle EMG screen of upper extremity muscles clinically affected, electromyographic examination of the infraspinatus or supraspinatus can help localize an upper trunk injury proximal to or distal to the takeoff to the suprascapular nerve. While the examination of the rhomboid can be difficult in the infant, a finding of fibrillations or positive sharp waves supports the presence of a C5 root injury. While in the adult, electromyographic evaluation of the cervical paraspinal muscles may help evaluate the extent and severity of cervical root injuries, generally these muscles are extremely difficult to study in the infant due to poor relaxation. In the young child, adequate relaxation of the cervical paraspinal muscles may be obtained with general anesthesia but this is usually not necessary and does not influence management. In addition, study of the serratus anterior and rhomboids (typically performed to assess involvement of C5 and C5–7 roots, respectively) may be technically difficult in the infant due to intact sensation, the presence of trapezius overlying the rhomboids, depth of the rhomboids and serratus anterior, and the risk that sudden movement may cause penetration of the needle into the pleural space. Usually a combination of needle EMG evaluation, sensory and motor conduction studies, and F-wave studies allows the electromyographer to determine the location and severity of the injury.

The natural history of conservatively managed brachial plexus birth palsy has been reported (69). Seventy-two percent of those referred for rehabilitation evaluation showed stable functional status at follow-up. There has been a resurgence of interest in surgical exploration of obstetrical brachial plexus palsy with external and internal neurolysis, neurotization, and in selected cases, nerve grafting (70–76). EMG evaluation at approximately 4 to 9 months postinjury may support the possible utility of a surgical exploration for neurolysis, neurotization, and/or nerve grafting if there is limited electrophysiologic evidence of reinnervation. Some authors suggest a repeat study within 3 months of the injury (77). Preoperative EDSs and intraoperative NCSs and somatosensory evoked potentials are helpful in the surgical decision making. Preoperative and/or intraoperative somatosensory evoked potentials may provide evidence of upper cervical root avulsion versus partial trunk and nerve root integrity as discussed in the following.

More recently, EMG and NCSs have been used in a more systematic fashion for surgical decision making. In two recent reports (78,79), infants with NBPP with severe lesions could be identified at 1 month of age by testing elbow extension and elbow flexion, and recording MUPs in the biceps muscle. Forty-eight infants were prospectively studied. The presence or absence of flexion paralysis at around 1 week (median 9 d; range 5–17 d), 1 month (median 31 d; range 24–53 d), and 3 months of age (median 87 d; range 77–106 d) was noted for clinical (shoulder external rotation, elbow flexion, extension, and supination) and EMG parameters (denervation activity, MUPs, and polyphasic MUPs in the deltoid, biceps, and triceps muscles). At 1 month, the absence of biceps MUPs had a sensitivity of 95% for later flexion paralysis, and the absence of deltoid MUPs had a sensitivity of 100% for flexion paralysis; the false-positive rates for the same findings were 21% and 33%

respectively. EMG at 3 months was highly misleading as MUPs were seen in 19 of 20 clinically paralytic biceps muscles. These authors have implemented a decision rule that children without active elbow extension at 1 month should be referred to a specialized center, while children with active elbow extension as well as active flexion should not. When there is active elbow extension, but no active elbow flexion, the authors concluded an EMG was needed; the absence of MUPs in the biceps muscle was an indication for referral.

In another retrospective study of preoperative EDSs and computed tomography myelogram (CTM; 80) conducted in 21 children, the sensitivity of EDSs and CTM for detecting a postganglionic rupture was 92.8% (95% CIs [0.841-0.969]) and 58.3% (95% CIs [0.420-0.729), respectively. The sensitivity for EDSs and CTM for preganglionic nerve root avulsion was 27.8% (95% CIs [0.125-0.509]) and 72.2% (95% CIs [0.491-0.875]), respectively. In cases in which both CTM and EDSs gave concordant results, the sensitivity for both modalities combined was 50.0% (95% CIs [0.237-0.763]) for avulsion and 80.8% (95% CIs [0.621-0.915]) for rupture. Overall, EDSs were most useful in identifying ruptures, particularly in the upper plexus, whereas CTM was most sensitive in identifying avulsions in the lower plexus. It was concluded that both EDSs and CTM scans must always be interpreted in the context of a comprehensive evaluation of the patient. They provide supplemental information (in addition to the physical examination) for early detection of nerve root rupture and avulsion injuries, aiding surgical decision making and preoperative planning for neonatal NBPP.

Chin and colleagues (81) recently reported on the prognostic significance of intraoperative EMG in NBPP. They investigated the predictive value of intraoperative neurophysiologic investigations in a total of 32 infants of 206 referred to their center who underwent exploration of the plexus, including neurolysis. The findings from intraoperative electromyography, sensory evoked potentials across the lesion, and gross muscular response to stimulation were evaluated. Outcomes were assessed with the modified mallet score at 3 to 4 years. The positive predictive value and sensitivity of the intraoperative EMG for C5 grafting and neurolysis were 100% and 85.7%, respectively, in infants without concurrent shoulder pathology. The positive and negative predictive values, sensitivity, and specificity of the three investigations combined were 77%, 100%, 100%, and 57%, respectively. In all, 20 infants underwent neurolysis alone for C6 and three had reconstruction. All of the former and one of the latter achieved biceps function of Raimondi grade 5. The positive and negative predictive values, sensitivity, and specificity of electromyography for C6 were 65%, 71%, 87%, and 42%, respectively. The method was concluded to be effective in evaluating the prognosis of C5 lesions. Neurolysis was preferable for C6 lesions.

FACIAL PARALYSIS IN THE NEONATE

Facial paralysis or an asymmetric facies is a common finding in the neonate. This may be due to acquired traumatic facial palsy (a common iatrogenic problem with forceps deliveries), central nervous system conditions, congenital facial palsy, and congenital hypoplasia of the depressor anguli oris muscle. Differentiating developmental from traumatic facial paralysis noted at birth is important for determining prognosis, but also for medicolegal reasons. Facial NCSs aid in diagnosis (82). Side-by-side comparisons of amplitudes and latencies are essential. CMAP amplitude reduction and prolonged latency on the involved side indicate facial nerve involvement. Brainstem auditory evoked potentials and blank reflexes may be helpful in determining central nervous system involvement. Axonal integrity can be determined by electromyographic evaluation for spontaneous activity and motor unit recruitment. Improvement on serial testing provides favorable prognostic information, particularly when improvement occurs over 1 to 2 weeks. Normal facial nerve distal latencies in the newborn are less than or equal to 12.0 msec; in children 1 to 12 months less than or equal to 10.0 msec; in children 1 to 2 years of age less than or equal to 6.3 msec; in children 2 to 3 years of age less than or equal to 4.5 msec; in children 3 to 4 years less than or equal to 4.0 msec; and less than or equal to 5.0 msec in children older than 4 years of age (83).

Asymmetric crying facies (ACF) is congenital hypoplasia of the depressor anguli oris muscle characterized by asymmetry of lower lip depression during crying. This has an overall incidence of 0.6%. One recent study determined the incidence of ACF in a large population of patients with 22q11.2 deletion to be 14% (84). Associated palatal anomalies were common (77%), as was congenital heart disease (78%). It is suggested that newborns with ACF be referred for further screening for the 22q11.2 deletion syndrome.

COMMON POLYNEUROPATHIES

Hereditary Neuropathies (Charcot–Marie–Tooth [CMT] Subtypes)

Clinical findings associated with hereditary neuropathies and the current classification of these disorders are described in Chapter 18. The demyelinating form (CMT 1) typically has onset in early childhood. Marked slowing of MCVs, usually to less than 50% of normal is usually present in early childhood (85–87). Generally, marked swelling of motor NCVs is present by 3 to 4 years of age (84). Distal latencies are usually severely prolonged. There is usually less temporal dispersion than observed in AIDP (GBS) due to fairly uniform demyelination of all axons. Needle EMG abnormalities include defibrillation with

positive sharp waves, decreased interference pattern, and large-amplitude polyphasic MUAPs resulting from reinnervation by collateral axonal sprouting.

CMT 2 is the axonal form. CMAP and SNAP amplitudes may be reduced, but NCVs are either in the low normal range or mildly reduced. Needle EMG shows evidence of chronic denervation and reinnervation. CMT 3 is also referred to as Dejerine-Sottas disease and congenital hypomyelinating neuropathy which usually present in infancy. CMAP amplitudes are reduced due to a combination of conduction block and axonal loss, motor NCVs are typically less than 10 m/s, and latencies may be three times the normal value (88).

Acute Inflammatory Demyelinating Polyradiculoneuropathy (GBS)

These children often present with an acute rapid ascending paralysis initially affecting the lower limbs. While pain is common, sensory symptoms are usually mild and objective sensory loss is fairly rare. Electrophysiologically, criteria for poor recovery in adults may not apply to children. One study documented good recovery in children with low median CMAPs and fibrillation potentials (89), while another study showed no difference in the incidence of reduced CMAP amplitude among ventilated and nonventilated children (74). It has been noted that gait disorder, leg pain, a high rate of distal conduction block (decreased distal CMAP amplitudes), and a good prognosis are among the main specific features of GBS in childhood (90).

Classic electrophysiologic findings in GBS include prolonged or absent F-waves early in the course of the disorder, slowing of CVs, both proximally and distally, prolonged distal latencies, reduced CMAP amplitudes with evidence of conduction block, and significant temporal dispersion (see Figure 6.10) (91). The electrophysiologic findings may lag behind the clinical signs and symptoms. In addition, electrophysiologic recovery may lag behind clinical recovery.

Chronic Inflammatory Demyelinating Polyradiculoneuropathy

This disorder has many features in common with AIDP. These patients typically show a subacute or chronic onset lasting more than 4 weeks, and the disorder continues with either a chronic or relapsing course. Electrophysiologic findings generally show more marked slowing of conduction velocity (often below 10 m/s) and elevated stimulation thresholds. As in AIDP, there is evidence of focal conduction block, temporal dispersion, prolongation of DMLs, and prolonged or absent H-wave and F-wave responses. These late responses may be absent due to proximal conduction block. Needle EMG may show a paucity of abnormal spontaneous rest activity and normal or slightly enlarged MUAPs, which exhibit a neuropathic firing pattern.

Axonal GBS/Acute Motor Axonal Neuropathy

In this disorder, children often present with rapid onset, quadriparesis, bulbar dysfunction, and respiratory insufficiency (92). The patients may have inexcitable motor nerves or very low-amplitude CMAPs. The

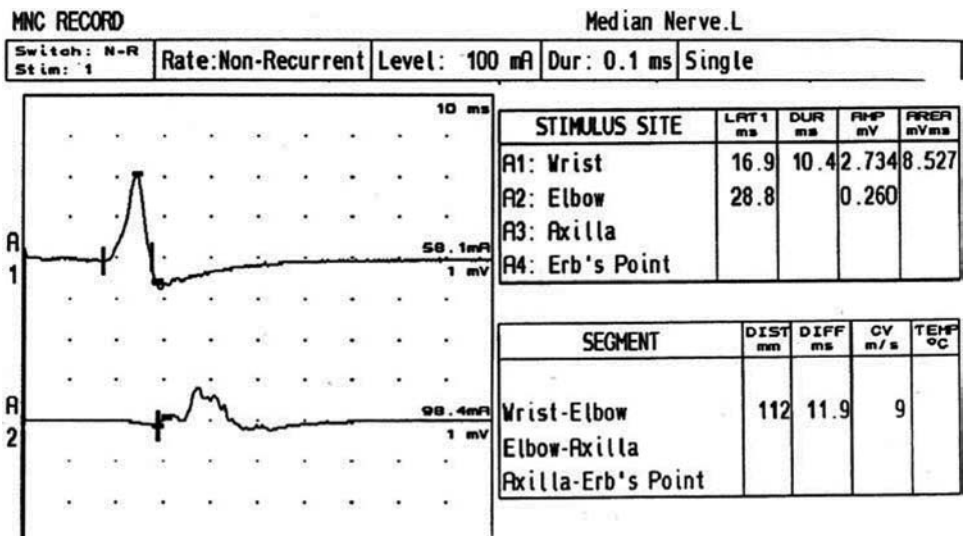

FIGURE 6.10 Median motor nerve conduction in a 4-year-old child with Guillain-Barré syndrome. Distal latency is prolonged at 16.9 milliseconds, and conduction velocity is slowed at 9 m/s. Note the conduction block (amplitude drop from 2.734 to 0.260 mV) and temporal dispersion.

author has observed such a case with clinical findings mimicking cerebral death (93). The child had combined demyelinating and axonal findings and eventually had near complete recovery over 18 months. In general, children with the axonal form of GBS are more likely to require assisted ventilation, develop severe quadriparesis, and require a much longer period of time to become ambulatory. Campylobacter jejuni has been implicated as a precipitating agent in many cases.

One recent study (94) evaluated an electrophysiologic classification of a GBS population into demyelinating and axonal subtypes, to investigate how serial recordings changed the classification and to underline the pitfalls in electrodiagnosis of GBS subtypes. In the first test, the electrodiagnostic findings resulted in 65% to 67% of patients classifiable as demyelinating AIDP, 18% classifiable as axonal GBS, and 14% to 16% equivocal. At follow-up, 24% of patients changed classification: AIDP decreased to 58%, axonal GBS increased to 38%, and equivocal patients decreased to 4%. The majority of shifts were from AIDP and equivocal groups to axonal GBS, and the main reason was the recognition by serial recordings of the reversible conduction failure and of the length-dependent CMAP amplitude reduction patterns as expression of axonal pathology. Axonal GBS is pathophysiologically characterized not only by axonal degeneration but also by reversible conduction failure at the axolemma of the Ranvier node. The lack of distinction among demyelinating conduction block, reversible conduction failure, and length-dependent CMAP amplitude reduction may fallaciously classify patients with axonal GBS as having AIDP. Serial electrophysiologic studies are mandatory for proper diagnosis of GBS subtypes and the identification of pathophysiologic mechanisms of muscle weakness. More reliable electrodiagnostic criteria taking into consideration the reversible conduction failure pattern should be devised.

Neuropathies Associated With Central Disorders

A variety of metabolic disorders produce abnormalities of both the central and peripheral nervous system. Abnormalities of lipid metabolism such as metachromatic leukodystrophy may produce a severe demyelinating peripheral neuropathy with electrophysiologic findings of high stimulation threshold and low CVs. Somatosensory evoked potentials may show both central and peripheral delay, and visual evoked potentials show central delay. Other disorders showing both central and peripheral nervous system involvement include Krabbe disease, Refsum's disease (phytanic acid storage disease), Tangier disease (hereditary high-density lipoprotein deficiency), A-beta lipoproteinemia (a vitamin E deficiency syndrome), Fabry's disease (alpha galactosidase A deficiency), Niemann–Pick disease (a variant of sphingomyelin lipidoses), peroxisomal disorders such as adrenoleukodystrophy, porphyria, which produces axonal degeneration of predominantly motor fibers, and tyrosinemia, which produces primary axonal degeneration with secondary segmental demyelination.

Krabbe disease is associated with marked central and peripheral demyelination and NCSs typically show a mixed sensorimotor demyelinating peripheral neuropathy. The peripheral neuropathy occurs very early in the neonatal period in Krabbe disease and affects the nerves uniformly. NCSs may provide a highly sensitive tool to screen this patient population (95).

In ataxia telangiectasia, there is a loss of large, predominantly sensory, myelinated fibers due to a primary axonal degeneration. In Friedreich's ataxia, an autosomal recessive condition, there is a primary axonal degeneration of peripheral nerve fibers producing reduced or absent sensory compound action potential amplitudes.

Acquired Toxic Neuropathies

Toxic polyneuropathies with predominantly axonal involvement include lead, mercury, and vincristine-induced neuropathy, among others. Predominantly demyelinating neuropathies may be caused by organophosphate poisoning and arsenic poisoning. While arsenic poisoning may clinically simulate GBS or chronic demyelinating polyneuropathy (CIDP), electrophysiologic studies have shown evidence of both axonal degeneration and severe demyelination.

Burn Associated Neuropathies

Children and adults with extensive burns are at increased risk for mononeuropathies and/or peripheral neuropathies (96–101). Mechanisms include direct nerve tissue destruction from the burn, extensive edema with compartment syndrome, critical illness polyneuropathy caused by systemic mediators, and entrapment neuropathies caused by scarring during and/or after healing. The incidence of neuropathy exceeds 10% in many series. Burn associated polyneuropathy (BAPN) is common after thermal injury, and the electrophysiologic manifestations of BAPN are usually present within the first week (98). Thermal injuries may induce an inflammatory cascade that results in alterations of nerve function. In one series, those with severe neuropathy had higher levels of C-reactive protein (98). Other risk factors associated with a significantly higher prevalence of neuropathy include age above 20 years, electrical burns involving full thickness of the skin, a surface area of more than 20%, history of alcohol abuse, and number of days in the intensive care unit (ICU). In animal models of burn injury, both functional and morphological deficits are produced in peripheral nerve axons at sites well removed from a full-thickness dermal burn injury (102). The neural deficits may contribute to changes in neuromuscular transmission and the development of limb and respiratory muscle weakness

that also accompanies burn injury. Further animal work has demonstrated that burn wound excision at 30 minutes but not at 3 hours prevented the nerve conduction deficits measured in mice with 20% body surface area burns (103). The cellular basis of burn-induced neuropathy is unknown, but nitric oxide and tumor necrosis factor-alpha (TNF-alpha) appear to play a role.

Vitamin D deficiency has been reported in pediatric burn patients (104), and the available literature on vitamin D status in burn patients has been reviewed. Vitamin D deficiency has been demonstrated to result in itching, muscle weakness, and neuropathy, all of which are common postburn sequelae. The major source of vitamin D is synthesis in the skin with a small amount being absorbed through dietary intake. Population groups are at higher risk of vitamin D deficiency if they have inadequate exposure to ultraviolet (UV) light or reduced biosynthetic capability due to skin damage. Burn patients fall into both risk groups and also suffer common complaints that overlap with those reported by patients with vitamin D deficiency.

Diabetic Polyneuropathy

NCV in the distal motor and sensory nerves, the motor nerve distal latency, and the SNAP amplitude were impaired in adolescent patients with type 1 diabetes. The deterioration in motor NCV, H-reflex latency, and SNAP amplitude became more conspicuous in late puberty and postpuberty and was related to poor metabolic control (105). In another study of children 7 to 20 years old with a duration of diabetes of more than 3 years, 57% of the patients had abnormal conduction, which was seen most often in the motor nerves, especially in the peroneal nerve (41%), followed by the median nerve (24%) (106).

Neuropathies Associated With Infections

HIV INFECTION. Children with HIV may develop a variety of neurologic sequelae including encephalopathy, progressive multifocal leukoencephalopathy, myelopathy, intractable seizures, optic neuritis, acute vasculitis, hemiplegia, paraspinal lymphoma, and peripheral nerve disease. The peripheral nerve dysfunction may present as distal symmetric sensory or sensorimotor polyneuropathy, carpal tunnel syndrome (CTS), lumbosacral polyradiculopathy, motor neuronopathy, AIDP and CIDP, autonomic neuropathy, sensory ganglionopathy, and toxic neuropathy (caused by antiretroviral medications) (107). In addition, polyradiculopathy and multiple mononeuropathies may be caused by other infections (eg, cytomegalovirus, hepatitis B or C, and herpes zoster). In one series, one-third of children 5 to 14 years of age had symptoms and signs of peripheral nerve involvement. Distal paresthesia and/or pain plus diminished ankle jerks and/or diminished vibration sense were the most common clinical findings. Symptoms were chronic and fluctuating, and pain was, in general, not severe. NCSs primarily revealed axonal changes (108). The issue of peripheral nerve involvement may be multifactorial. Children with HIV-1 infection are exposed to antiretroviral (ARV) drugs for an ever-increasing length of time throughout postnatal growth and development, and the cumulative toxicities are becoming progressively apparent. Evidence for nucleoside reverse transcriptase inhibitor (NRTI) associated mitochondrial toxicity is seen in vitro, in animal models and in NRTI-exposed adults and children (108). Peripheral neuropathy is associated with the chronic use of dual NRTI regimens in HIV-infected children, and regimens containing Zidovudine (Retrovir, AZT, ZDV) have less toxicity than do those containing d4T (109).

LYME DISEASE. Lyme disease is the most common tick-borne disease in the United States. Children and those spending extended time outdoors in wooded areas are also at increased risk. The spectrum of neurologic manifestations and the relative frequencies of different syndromes associated with North American Lyme disease caused by *Borrelia burgdorferi* infection have been reviewed in a series of 96 children referred for neurologic problems in association with the infection (110). The most frequent neurologic symptom was headache, and the most common sign was facial palsy. Less common manifestations were sleep disturbance, and papilledema associated with increased intracranial pressure. Signs and symptoms of peripheral nervous system involvement are infrequent. Cranial neuropathy is common (111) and children may present with only cranial neuropathy, both cranial neuropathy and other neurologic symptoms, and neurologic symptoms without cranial neuropathy. Patients with Lyme meningitis had statistically more frequent cranial neuropathy (73% vs. 4%) (112). In Lyme disease, the most common clinical syndromes include mild encephalopathy, lymphocytic meningitis, and cranial neuropathy (facial nerve palsy). In contrast with adult patients with neurologic Lyme disease, meningoradiculitis (Bannwarth's syndrome) and peripheral neuropathy syndromes are rare in children.

ENTRAPMENT MONONEUROPATHIES IN CHILDREN

Carpal Tunnel Syndrome in Children

CTS is a relatively rare complication in children with mucopolysaccharidosis types I, II, and III (eg, Hunter's and Hurler's syndromes), with mucolipidosis being the most common populations to manifest CTS during childhood (94). Treatment of the metabolic disorder does not necessarily reverse the symptoms and prompt surgical release is necessary. Other uncommon etiologies include congenital bone anomalies, mucolipidosis types II and III, hereditary neuropathies such as CMT 1 and hereditary neuropathy with liability to pressure palsies (HNPP), CIDP, treatment with growth hormone, hypothyroidism,

hemophilia with localized bleeding in the region of the carpal tunnel, autosomal-dominant acromicric dysplasia, Down syndrome, Schwartz–Jampel syndrome, multiple xanthomas associated with familial hypercholesterolemia, congenital macrodactyly in a median nerve territory, fibrolipomas and fibrolipomatous hamartoma of the median nerves, obesity, and Klippel–Trénaunay syndrome (113–119). Idiopathic CTS is rare in children. Hand clumsiness and thenar hypoplasia rather than sensory complaints are often presenting symptoms in children. In another pediatric series, all patients had hand pain, numbness, and paresthesias in their hands. Night pains and underutilization of the first three fingers are also described in cases of pediatric CTS (120). An infant with CTS presented with intermittent abnormal "pseudodystonic" posturing movement of both hands (121).

Ulnar Mononeuropathies in Children

Ulnar mononeuropathies are the most common upper extremity mononeuropathies seen in children (122). The most common etiology is acute trauma (eg, midshaft or proximal forearm fractures, elbow dislocation, etc.), compression from compartment syndrome, or entrapments in association with HNPP or other anomalous anatomy producing entrapment. Other etiologies include baseball throwing injuries in adolescents, Larsen syndrome with dislocations, congenital constriction band syndrome, insulin-dependent diabetes mellitus, ulnar nerve tuberculoma, leprosy, and so on. The location of the neuropathy is most commonly the cubital tunnel, but it may also localize to the forearm, wrist, or hand. The cause of ulnar nerve palsy in a series of children with cubitus varus deformity was constriction by a fibrous band and kinking in the proximal border of the flexor carpi ulnaris due to ulnar nerve dislocation and compression resulting from the forward movement of the medial head of the triceps brachii muscle (123). Cubital tunnel syndrome may occur in children and adolescents. Subjective complaints at the time of presentation included ulnar nerve instability at the elbow, pain at the elbow, and numbness and tingling in the ring and small fingers. Physical examination revealed a majority of extremities with a positive Tinel's sign and a positive elbow-flexion–compression test in approximately 50% of cases (124).

Radial Mononeuropathies in Children

Radial mononeuropathies are rare but do occur in children. In one series, 50% of radial neuropathies, including two in newborns with apparent prenatal onset, were atraumatic, primarily related to compression in six and entrapment in two. The other 50% were traumatic mononeuropathies related to fractures or lacerations (125). Electromyography documented the radial neuropathy to be localized to the proximal main radial nerve trunk in 13%, distal main radial nerve trunk in 56%, and posterior interosseous nerve in 31% of the children.

Peroneal Mononeuropathies in Children

The most common entrapment in the lower extremity is peroneal mononeuropathy at the fibular head. Children with peroneal mononeuropathy typically present with unilateral foot drop. Both distal branches are involved in the majority of cases; hence the level of the lesion is most commonly the common peroneal nerve at or above the fibular head, followed by the deep peroneal nerve and superficial peroneal nerve (126). Common etiologies include compression from a short leg cast, compression from prolonged surgical positioning, and trauma (eg, distal femoral physeal fractures, proximal tibial fractures, etc.). Contributing factors include hereditary neuropathies (CMT or HNPP), and significant rapid weight loss in an adolescent. Other etiologies may include compression from osteochondromas, neurofibromas, intraneural ganglions, or arthrogenic cyst of the fibula, and stretch during tibial limb lengthening.

Sciatic Mononeuropathies in Children

Sciatic mononeuropathies are uncommon in children. Etiologies in one series included compression, stretch injuries (eg, during closed reduction of a hip dislocation), lymphoma, vasculitis associated with hypereosinophilia, and penetrating trauma (127). The peroneal division is more commonly affected than the tibial division in the absence of penetrating trauma. The vascular supply to the peroneal division may be more susceptible to compromise from stretch or compression. Axonal sciatic lesions are more common than demyelinating lesions.

Femoral Mononeuropathies in Children

Wang and colleagues (128) from China reported electrodiagnostic findings in 22 pediatric patients with suspected femoral nerve injury. The investigators obtained normal values for pediatric femoral nerve motor and sensory conduction in all age groups, including proximal and distal CMAP latencies, proximal CMAP amplitude and duration, MCV, F-wave latency, and sensory conduction velocity. They measured proximal CMAP in all children in all age groups. The manifestation of femoral nerve injury in the 22 patients was primarily a clear decrease or absence of CMAP amplitude or a lengthened latency. Electromyographs revealed that 104 muscle parts were involved in the nerve function, in which 59 parts were found to be abnormal (56.73%). The development of the pediatric femoral nerve mainly began after the child was 1 years old and continued to 14 years. The proximal latency and CMAP amplitude of the pediatric femoral

nerve have clinical value. Detection of the femoral nerve is important in the diagnosis of lower limb monoplegia, especially for acute flaccid paralysis associated with nonpolio enterovirus infection.

Neuropathies With Limb Lengthening Procedures

Mononeuropathies in the setting of limb lengthening are not uncommon but are frequently subclinical. Patients undergoing tibial limb lengthening procedures are at risk for peroneal neuropathies, in particular, and rarely tibial mononeuropathies. Femoral lengthening can place a patient at risk for neuropathies affecting the sciatic nerve (particularly the peroneal division). Humeral lengthening can place upper extremity nerves at risk. Some have monitored for subclinical neuropathy of the upper and lower extremities using mixed nerve somatosensory evoked potentials during pin placement and serially during distraction (129,130).

NEUROMUSCULAR JUNCTION DISORDERS

Infantile Botulism

Infantile botulism primarily occurs in infants 2 to 6 months of age. Clinical findings include diffuse weakness, hypotonia, weak cry, poor feeding, constipation, and occasionally respiratory distress. The onset is fairly rapid. Electrophysiologic studies may show a reduced CMAP amplitude, preserved MCVs and snaps, and abnormal repetitive nerve stimulation findings at high rates of stimulation (see Figure 6.11). One study demonstrated an incremental response to repetitive nerve stimulation at rates of 20 to 50 Hz in 92% of infants with infantile botulism (25). The mean increment was 73% with a range of 23% to 313%. For lower frequency stimulation (2–5 Hz), variable changes occurred, but the majority of infants showed decremental responses. A recent study demonstrated that the isolation of clostridium botulinum from stool obtained by enema effluent

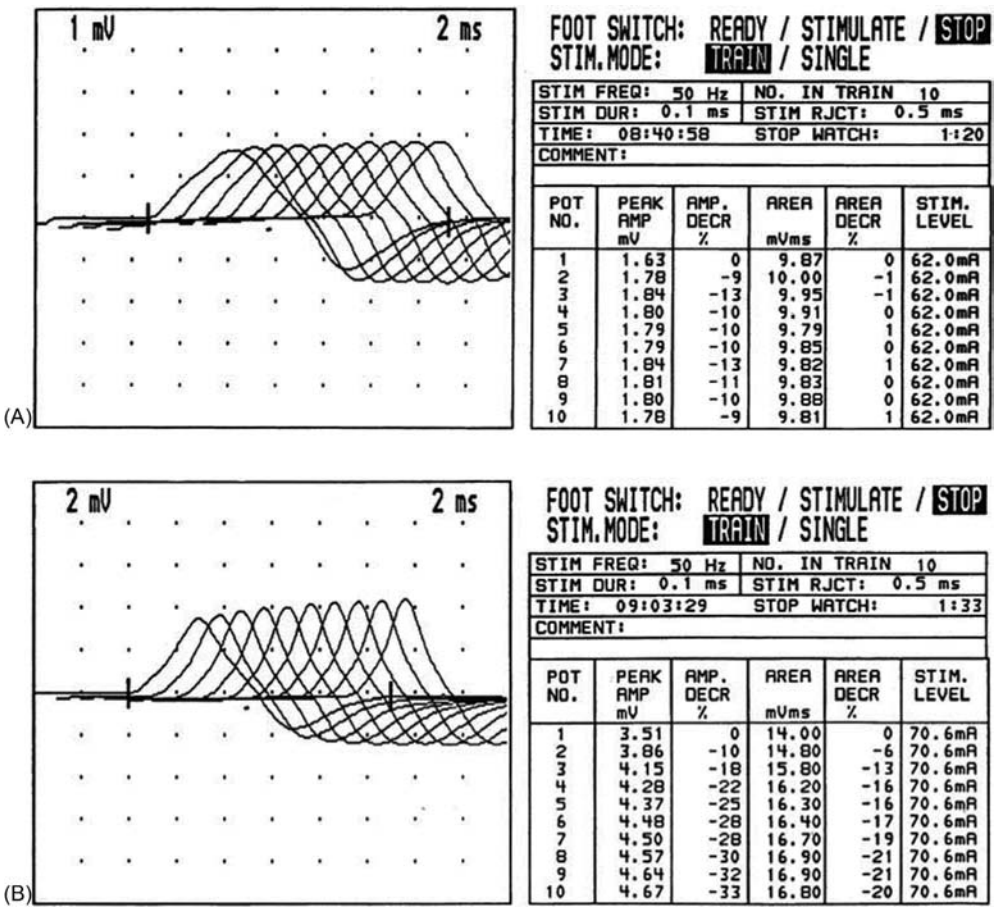

FIGURE 6.11 High frequency repetitive nerve stimulation in a 7-week-old infant with marked progressive weakness, respiratory failure, and botulism. (A) Several days into the course, the repetitive stimulation study of the ulnar nerve at 50 Hz is normal; however, the compound muscle action potential amplitude is severely reduced (1.63 mV). (B) Twelve days later, the infant is slightly improved clinically. A repeat study of the ulnar nerve at 50 Hz is diagnostic of infantile botulism with a 33% increment obtained between first and tenth stimuli. Clostridium botulinum was isolated from the stool.

was actually more sensitive for the diagnosis of infant botulism than EDSs (131).

EMG in infants with botulism demonstrates abnormal spontaneous rest activity with fibrillation potentials and positive sharp waves and short-duration low-amplitude MUAPs (25).

Transient Neonatal Autoimmune Myasthenia Gravis

This disorder is caused by the passage of antibodies from myasthenic mothers to their fetuses. Infants often present with hypotonia and respiratory distress. The diagnosis may be made by repetitive nerve stimulation studies. Given that normal infants exhibit less neuromuscular reserve than older children or adults, repetitive stimulation studies in this clinical setting utilize rates of 2 to 5 Hz almost exclusively. A decrement of greater than 8% to 10% between the first and fifth CMAP at low rates of stimulation is considered positive for myasthenia. The combination of repetitive motor nerve stimulation and edrophonium or neostigmine testing may improve the accuracy of the diagnosis (132). If a decremental response is obtained, the repetitive nerve stimulation may be repeated at 30 to 120 seconds after administration of edrophonium utilizing a stimulation rate of 2 to 5 Hz. Near complete repair of the decremental response may be evident in the myasthenic infant (see Figure 6.12). Serologic antibody testing may be helpful if the mother has documented antibodies. Transient neonatal myasthenia gravis is self-limited with a reported duration of 5 to 47 days with a mean duration of 18 days (133).

Toxic Neuromuscular Junction Disorders

Medications can interfere with neuromuscular transmission by inhibiting the release of acetylcholine, impairing the function of AChE, or binding directly to the acetylcholine receptor. Two drugs that may produce clinically

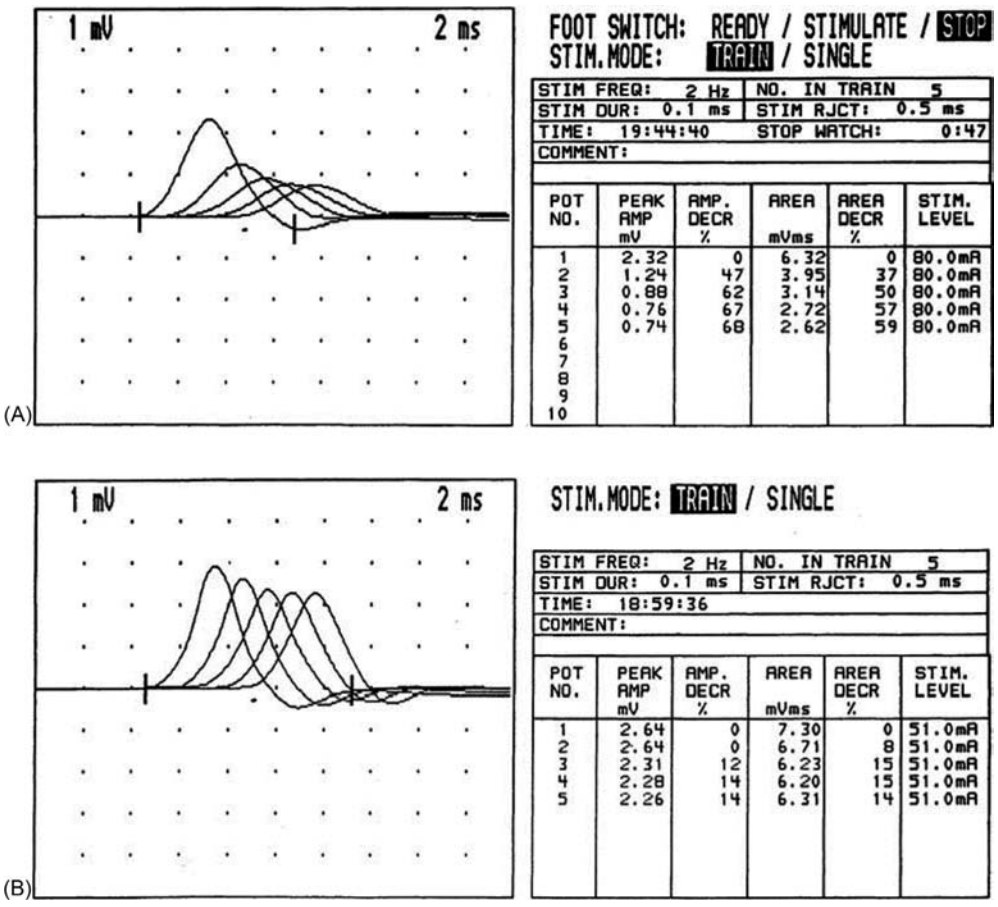

FIGURE 6.12 Low-frequency repetitive nerve stimulation study of the ulnar nerve in a 2-week-old infant with respiratory failure secondary to congenital myasthenia. (A) At baseline, a 68% decrement in amplitude and a 59% decrement in area is present between first and fifth stimuli with a stimulation frequency of 2 Hz. (B) Twenty minutes after intravenous neostigmine is given, the initial compound muscle action potential has improved from 2.32 to 2.64 mV and the decrement has improved to 14%. The infant was treated with Mestinon and later extubated.

significant weakness in normal children are magnesium and organophosphates (134,135).

Congenital Myasthenic Syndromes

Numerous presynaptic and postsynaptic congenital myasthenic subtypes exist, which are described in Chapter 18. These disorders often show decremental responses to high rates of stimulation whether they are pre- or postsynaptic. Typically, the decremental responses are greater at higher rates of stimulation. Standard repetitive nerve stimulation studies do not adequately distinguish presynaptic from postsynaptic subtypes but they do help diagnostically (see Figure 6.13). Based on clinical findings, repetitive nerve stimulation studies, and/or stimulated single-fiber EMG, a strong clinical suspicion of an NMJ disorder such as a congenital myasthenic syndrome might warrant further elucidation of the specific subtype of presynaptic or postsynaptic abnormality with application of a motor point biopsy. Ultrastructural evaluation of the NMJ with electron microscopy is usually performed on a biopsy of the deltoid or biceps, including the muscle region containing the NMJ (the "motor point"). For in vitro electrophysiologic and immunocytic chemical studies of the neuromuscular junction, a short muscle is usually removed from the origin to insertion along with its motor branch and NMJ. Muscles obtained have included the anconeus muscle near the elbow, the external intercostal muscle, and the fifth or sixth intercostal space near the anterior axillary line or the peroneus tertius muscle in the lower extremity. Often, patients undergo simultaneous biopsy of the deltoid (for electron microscopy [EM]) and motor point biopsy of the anconeus or intercostal muscle (for in vitro electrophysiologic studies). The in vitro electrophysiologic studies often allow specific delineation of the congenital myasthenic syndrome into one of the numerous unique subtypes. In recent years, many of the subtypes have been mapped to specific gene loci and increasingly molecular genetic studies are being used for diagnostic purposes.

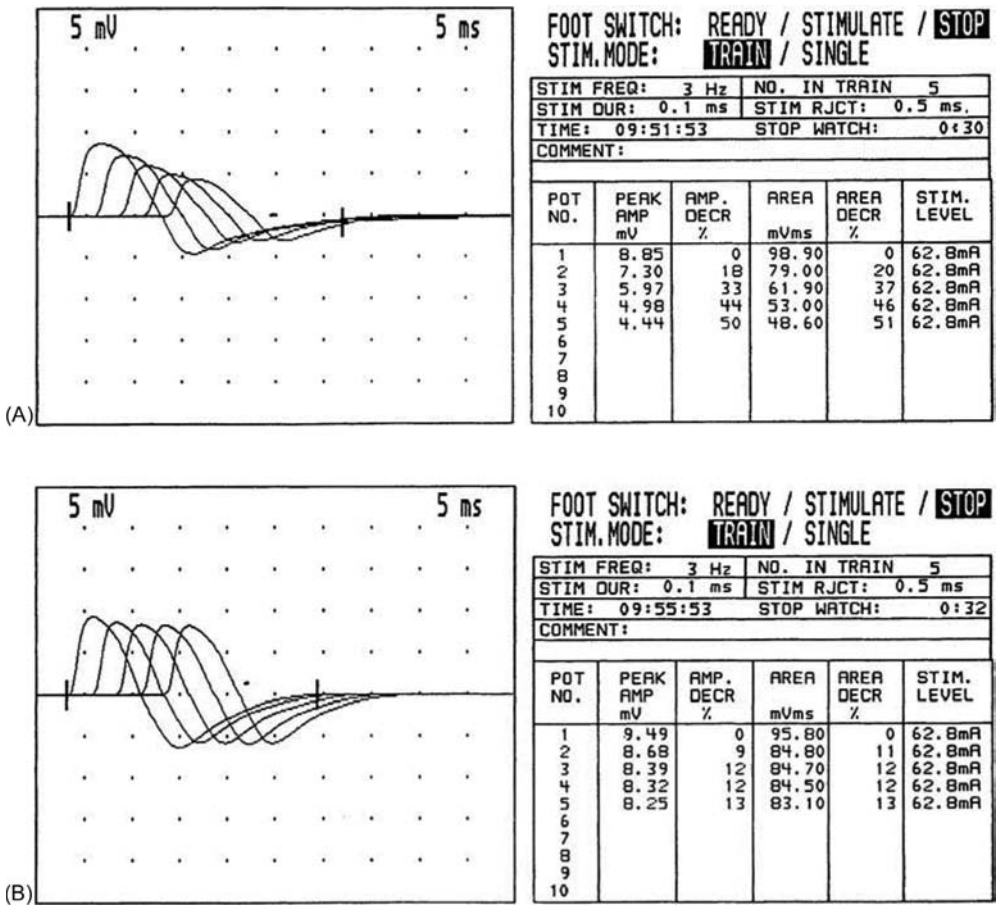

FIGURE 6.13 Low-frequency repetitive nerve stimulation study of the axillary nerve in a 12-year-old child with presynaptic congenital myasthenia. The active electrode is placed over the deltoid with stimulation at Erb's point using a block stimulator. (A) A 50% amplitude decrement is obtained between the first and fifth stimuli with 3 Hz stimulation frequency. (B) After a 30-second isometric contraction of the deltoid, the amplitude decrement has improved to 13%. The child was later confirmed to have a presynaptic congenital myasthenia by motor point biopsy of the anconeus muscle.

Myasthenia Gravis

Myasthenia gravis presents in adolescents more frequently than younger children. Muscle weakness typically increases with exertion but improves with rest and AChE medication. The disorder is an autoimmune etiology due to circulating antibodies, which bind on the postsynaptic membrane. While elevated acetylcholine receptor antibody levels may be diagnostic, a significant percentage of cases with autoimmune myasthenia gravis may have nondetectable circulating antibodies. Electrophysiologic studies demonstrate abnormal decremental responses at low rates of stimulation (2–3 Hz). The limb is well immobilized. A supramaximal train of 3 to 5 stimuli is applied. Typically, patients exhibit a smooth, reproducible decrement of the evoked synapse of greater than 8% to 10%. The defect in NMJ transmission can be enhanced by exercise, which results in postactivation facilitation. Often, there is an increased decremental response obtained 2 to 4 minutes after exercise with low rates of stimulation (2–3 Hz). This is due to postactivation exhaustion (24). Proximal muscles may show increased sensitivity versus distal muscles. Children with ocular myasthenia frequently exhibit normal responses with distal repetitive nerve stimulation studies and sensitivity of the repetitive nerve stimulation study is enhanced by the use of a more proximal shoulder girdle muscle (eg, axillary or spinal accessory nerve) or by the study of the facial nerve. Combining the diagnostic yield, patient comfort, and technical ease, the choice of muscle for RNS should be ulnar to the ADM followed by spinal accessory to the trapezius for a patient with predominant limb weakness; facial nerve to the nasalis and spinal accessory to the trapezius in oculobulbar; and facial to the nasalis in ocular myasthenia (136).

Lambert–Eaton Syndrome

This presynaptic NMJ disorder usually found in adults with small cell carcinoma of the bronchus has been described in children. Approximately 5% of all cases occur in children. The amplitude of the single evoked CMAP is low. With low rates of repetitive nerve stimulation, a decremental response is often obtained. After exercise or tetanic contractions, there is facilitation of the potentials by as much as 100% to 200%.

MYOPATHIES

Polymyositis/Dermatomyositis

Polymyositis/dermatomyositis has been described in children ranging in age from infancy to adulthood. Children may result with progressive proximal muscle weakness, dysphagia due to involvement of pharyngeal musculature, dyspnea, and muscle tenderness. A classic skin rash may or may not be present. Creatinine kinase values are often markedly elevated. Classic EMG findings include: (a) increased insertional activity with complex repetitive discharges; (b) fibrillations and positive sharp waves; and (c) low-amplitude, polyphasic, short-duration MUAPs recruited rapidly in relation to the strength of contraction.

Congenital Myopathies

Congenital myopathies are a heterogeneous group of disorders usually presenting with infantile hypotonia, normal cognitive status, and primary structural abnormalities of the muscle fibers, which are elucidated on histologic and electron microscopic evaluations of muscle biopsy specimens. Patients usually develop proximal rather than distal muscle weakness, which is nonprogressive and static. These myopathies are described in Chapter 18. NCSs are generally normal; however, there may be mild reductions in CMAP amplitudes. On needle EMG, findings are either normal or there may be mild, nonspecific changes, usually of a myopathic character (small-amplitude, short-duration polyphasic MUAPs). The only congenital myopathy consistently associated with abnormal spontaneous rest activity is myotubular (centronuclear) myopathy. In this disorder, the EMG reveals myopathic MUAPs with frequent complex repetitive discharges and diffuse fibrillation potentials.

Dystrophic Myopathies

The dystrophic myopathies are extensively described in Chapter 18. EMG is rarely used in present times for the diagnostic evaluation of a suspected dystrophic myopathy due to molecular genetic testing and the importance of muscle biopsy in differentiating among Duchenne muscular dystrophy, Becker muscular dystrophy, and limb girdle muscular dystrophies. EMG in dystrophic myopathies is characterized by low-amplitude, short-duration polyphasic MUAPs (see Figure 6.14). Recruitment is myopathic in nature with increased recruitment or "early" recruitment demonstrated with slight effort. The interference pattern is usually full. Complex repetitive discharges (see Figure 6.15) and abnormal spontaneous rest activity may be present, reflecting membrane instability.

Metabolic Myopathies

Nonspecific myopathic EMG findings may be demonstrated in metabolic myopathies. For example, absent maltase deficiency shows increased insertional activity, complex repetitive discharges, low-amplitude short-duration MUAPs, profuse fibrillations, and positive sharp waves. Carnitine deficiency, a disorder of lipid metabolism,

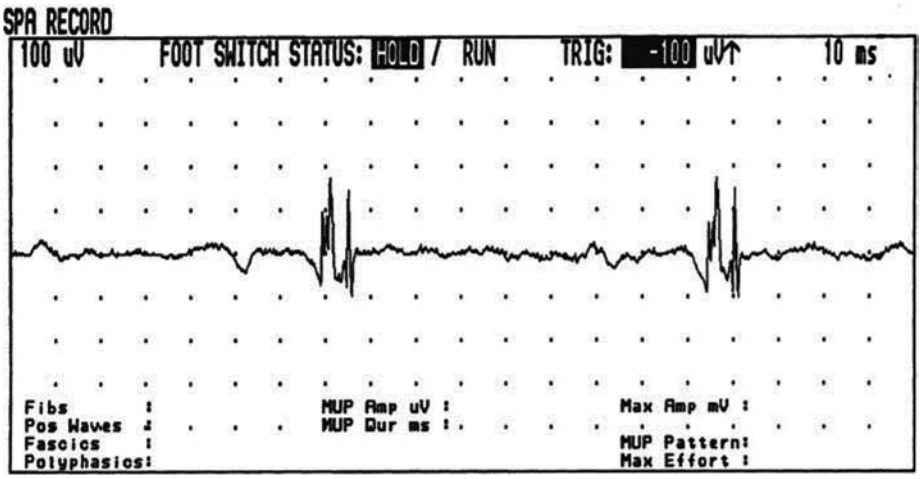

FIGURE 6.14 Low-amplitude short-duration polyphasic motor unit action potential in a 14-year-old girl with limb-girdle muscular dystrophy.

demonstrates increased recruitment for effort, decreased amplitudes of MUAPs and occasional fibrillations. EMG may be normal in many metabolic myopathies such as carnitine palmitoyltransferase I (CPT I) deficiency.

Myotonic Disorders

Myotonic disorders such as myotonic muscular dystrophy and Schwartz–Jampel syndrome may show myotonic discharges with either a positive sharp wave or fibrillation configuration, and a waxing and waning firing frequency. The myotonic discharges are often described as exhibiting the sound of a "dive bomber." There may be profuse fibrillations and positive sharp waves. MUAPs are often low amplitude and short duration. There may be more involvement of distal musculature than proximal musculature in myotonic muscular dystrophy. Again, with a known family history of myotonic muscular dystrophy, confirmation of the diagnosis in an individual with classic clinical features can be expeditiously and cost-effectively confirmed in the EMG laboratory. However, clinical trials frequently require molecular genetic confirmation of myotonic muscular dystrophy (DM1 versus DM2 and other myotonic disorders), so EMG is becoming less utilized diagnostically.

A recent study assessed the spectrum of disorders associated with electrophysiologic myotonia in a pediatric electromyography laboratory (137). Records of 2,234 patients observed in the Electromyography Laboratory at Boston Children's Hospital from 2000 to 2011 were screened retrospectively for electrophysiologic diagnoses of myotonia and myopathy. Based on electromyography, 11 patients manifested myotonic discharges alone,

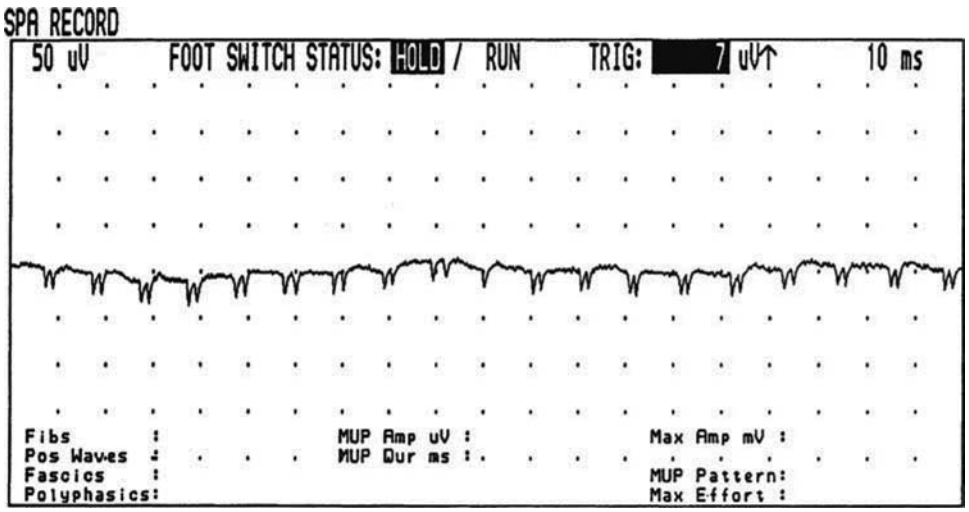

FIGURE 6.15 Complex repetitive discharges in a dystrophic myopathy.

8 exhibited both myotonic discharges and myopathic MUPs, and 54 demonstrated myopathic MUPs alone. The final diagnoses of patients with myotonic discharges alone included myotonia congenita, paramyotonia congenita, congenital myopathy, and Pompe disease (acid maltase deficiency). The diagnoses of patients with both myotonic discharges and myopathic MUPs included congenital myopathy and non-Pompe glycogen storage diseases. Myotonic discharges are rarely observed in a pediatric electromyography laboratory, but constitute useful findings when present. The presence or absence of concurrent myopathic MUPs may help narrow the differential diagnosis further.

Linear Scleroderma

Muscle atrophy and asymmetric extremity growth are common features of linear scleroderma (LS). Extracutaneous features are also common and primary neurologic involvement, with sympathetic dysfunction, may have a pathogenic role in subcutaneous and muscle atrophy. One recent study (138) investigated nerve conduction and muscle involvement by electromyography in pediatric patients with LS. A retrospective review of LS pediatric patients who had regular follow-up at a single pediatric center was conducted from 1997 to 2013. Electromyograms (EMG) were performed with bilateral symmetric technique, using surface and needle electrodes, comparing the affected side with the contralateral side. Abnormal muscle activity was categorized as a myopathic or neurogenic pattern. Nine LS subjects were selected for EMG, two with Parry–Romberg/hemifacial atrophy syndrome, seven with LS of an extremity, and two with mixed forms (linear and morphea). Electromyogram analysis indicated that all but one had an asymmetric myopathic pattern in muscles underlying the linear streaks. Motor and sensory nerve conduction was also evaluated in upper and lower limbs and one presented a neurogenic pattern. Masticatory muscle testing showed a myopathic pattern in the atrophic face of two cases with head and face involvement. Thus, in this small series of LS patients, the investigators found a surprising amount of muscle dysfunction by EMG. The muscle involvement may be possibly related to a secondary peripheral nerve involvement due to LS inflammation and fibrosis. Further collaborative studies to confirm these findings are needed.

SOMATOSENSORY EVOKED POTENTIALS

GENERAL PRINCIPLES

The somatosensory evoked potential (SSEP) is the sequence of voltage changes generated in the brain and the pathway from a peripheral sensory nerve following a transient electrical stimulus to the sensory cortex. Evidence suggests that these signals are related to large afferent fibers and the peripheral nerve, which ascend through the dorsal column pathways of the spinal cord, proceed to the thalamus, and arrive at the somatosensory cortex. These are the same pathways that mediate light touch two-point discrimination, proprioception, and vibration. Sensitive amplification and averaging techniques enable discrimination between the evoked response and other larger and more random physiologic potentials with which the signal is mixed. As a general rule, SSEP studies may be considered whenever the disease process in question can involve the somatosensory system. SSEPs reflect neurophysiologic activity in the posterior column, medial lemniscus pathways. SSEPs do not reflect activity in the anterolateral column of the spinal cord. Thus, SSEPs correlate better with clinical examinations of proprioception and vibration rather than pain or temperature sensation.

Individual components of the SSEP waveform are identified by their latency (ie, the time at which they occur following a peripheral stimulus), their polarity, the position at which they are observed to be maximal, and to a lesser extent by the amplitude and shape of the waveform. Individual components are referred to by a letter and number. The letter (N for negative or P for positive) refers to the polarity of the wave and the number either to the latency in milliseconds of the signal from the time of the stimulus (eg, N20) or alternatively, especially appropriate in pediatric SSEPs, the order in which the component was observed (eg, N1, P2). Examples of median and tibial SSEPs are shown in Figures 6.16A and 6.17A.

With mixed nerve stimulation, recording electrodes are placed over the peripheral nerve more proximally, thoracolumbar or cervical spine, linked mastoids, and scalp. For upper extremity stimulation, the likely generator source for the cervical spine response is the incoming root, as well as postsynaptic excitatory potentials generated at the dorsal root entry zone (139). For the lower extremity, the lumbar spine responses are similarly a reflection of the root or cauda equina activity and the postsynaptic activity of the cord. The linked mastoid response is generated at the brainstem level. The difference in the latency of scalp N1 and the cervical spine response with median nerve stimulation gives a central conduction time. Similarly, the difference in latency between scalp P1 for posterior tibial nerve stimulation and the spinal potential generated over T12 or L1 gives a central conduction time.

Filter settings vary from a low-frequency filter of 3 to 30 Hz to a high-frequency filter of 1.5 to 3 KHz. The peripheral nerve is typically stimulated with a rate of 3.1 Hz. Our lab utilizes a stimulation intensity of 1.5 times the motor threshold for mixed nerve stimulation and 2.5 times the sensory threshold for dermatomal stimulation. Electrodes are positioned according to a modified international 10 to 20 electrode system.

SEP latencies decrease with age until well into childhood (139–142). The maturation with the growth of SSEPs

is mainly associated with cell-growth processes such as myelination and with cell differentiation and synaptic development. CV along the central pathways progressively increases until 3 to 8 years of age, remains constant between 10 and 49 years of age, and slows thereafter. The N1 scalp latency of the median SSEP decreases until 2 to 3 years of age (owing to peripheral myelination) and then increases with body growth until adulthood. The cervical spine latency is relatively stable during the first 2 years (due to concomitant peripheral myelination and body growth), and then increases with age from 2 to 3 years until adulthood. The median SSEP central interpeak latency between cervical spine latency and scalp N1, which reflects central conduction time, decreases from a mean of 11.6 msec at 4 to 8 months of age to a mean of 7 msec at 6 to 8 years of age, and remains constant between 6.9 and 7.0 msec until adulthood (143,144).

Among infants less than 4 months of age, sleep can affect the cortical components and is best performed on the awake infant. With children greater than 4 months of age, sleep or sedation usually has little effect on the SEP waveform when performing mixed nerve stimulation. Indeed, the author has had no difficulty obtaining median nerve scalp responses in the pediatric ICU in comatose children with head trauma, or those heavily sedated. Dermatomal SSEPs, on the other hand, are state-dependent responses impacted by both sleep and sedation.

CLINICAL APPLICATIONS OF SSEPs IN CHILDREN

Brain Injury in SSEPs

Abnormalities of median SSEPs can be predictive of poor prognosis in the situation of brain injury due to head trauma or hypoxia. A loss of bilateral SSEP scalp waveforms, as shown in Figure 6.16B, portends a poor prognosis in comatose children (145–149). Asymmetric scalp responses in a comatose child may be associated with the development of motor abnormalities such as hemiparesis because of the proximity of the sensory cortex to the motor cortex (see Figure 6.17B). A recent study compared the predictive powers of clinical examination (pupillary responses, motor responses, and Glasgow Coma Scale [GCS]), EEG, and computed tomography (CT) to that of somatosensory evoked potentials (SEPs) in a systematic review. SEPs appear to be the best single overall predictor of outcome (116). Posterior tibial nerve SSEPs performed

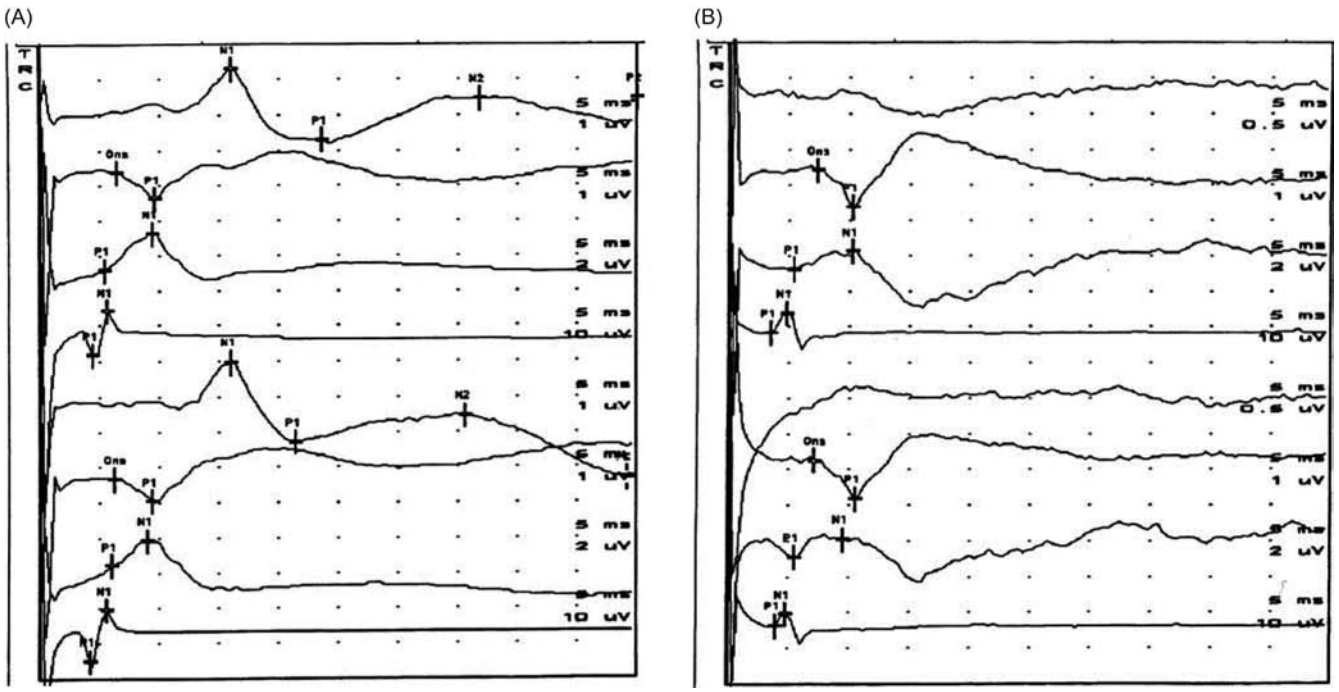

FIGURE 6.16 Median nerve somatosensory-evoked potentials (SSEPs) obtained in the pediatric intensive care unit. Channels 1–4 are responses with left median nerve stimulation, and channels 5–8 are responses with right median stimulation. Channels 1 and 5 are scalp responses (C4′ and C3′ referenced to Fz); channels 2 and 6 are brain (C4′ and C3′ referenced to linked mastoids); channels 3 and 7 are lower cervical spine responses (C7 spine referenced to Fz); channels 4 and 8 are peripheral responses obtained at the axillae. (A) Normal median SSEP responses obtained from a child with an epidural hematoma who was paralyzed with vecuronium for intracranial pressure control. There is no evidence of myelopathy. The child later recovered with minimal sequelae. (B) Abnormal median SSEP responses in a comatose child with severe brain injury and C1–C2 vertebral injuries. Note the bilaterally abnormal scalp reponses. Brainstem, C7 spine, and peripheral responses show no evidence of a spinal cord injury affecting posterior column pathways.

on neonates at high risk of future neurodevelopmental impairment have demonstrated a highly significant relationship between bilaterally abnormal posterior tibial nerve SSEPs and the presence of cerebral palsy at 3 years of age (150). Normal posterior tibial nerve SSEPs were associated with a normal outcome in 24 of 25 infants. In this study, posterior tibial nerve SSEPs were more predictive than cranial ultrasound. Another study of 43 children with hemiplegic cerebral palsy found a positive correlation between median nerve SSEPs and the affected side using the amplitude of the responses rather than the latency (151). Other studies have confirmed the prognostic value of SSEPs in infants at risk for neurodevelopmental impairment (152–156).

Traumatic Spinal Cord Injury

SSEP results combined with early American Spinal Injury Association (ASIA) motor scores have been shown to predict ultimate ambulatory capacity in patients with acute SCI (157,158). Other authors have shown that SSEP improvement over a 1-week interval during the first 3 weeks after SCI was associated with motor index score improvement over a 6-month period (159). Both ASIA scores and MEP recordings are similarly related to the outcome of ambulatory capacity and hand function in patients with SCI. Dermatomal somatosensory evoked potentials have also been shown to be more sensitive for the detection of sacral sparing and of more prognostic value than mixed nerve somatosensory evoked potentials (160). However, somatosensory evoked potentials and dermatomal SSEPs have been shown to add little or no useful prognostic information to the initial physical examination in either complete or incomplete SCI patient groups (161).

The author has a great deal of experience utilizing somatosensory evoked potentials in the pediatric ICU to evaluate for SCIWORA (162) in the situation where children are comatose, or too obtunded to cooperate with the examination, or the child's age precludes a detailed sensory examination. Figure 6.17A shows an example of a normal tibial SSEP, whereas Figure 6.17C demonstrates the impaired posterior column conduction between the lower cervical spinal cord and brainstem with a SCIWORA injury sustained by a 4-year-old child.

Tethered Cord Syndrome

Posterior tibial SSEPs have been shown in some studies to be a sensitive indicator of declining neurophysiologic status and a more sensitive diagnostic tool than the clinical testing of sensation in patients with tethered spinal cord post myelomeningocele repair (163–166). In addition, improvement of the evoked potentials after untethering has been documented (163,164,166). In the author's experience, the spine response is often caudally displaced in myelomeningocele. Absent or reduced amplitude lumbar spine potentials or prolonged lumbar spine or scalp latencies with tibial nerve stimulation in the setting of normal median somatosensory evoked potentials (normal spine latencies and amplitudes with median nerve stimulation, normal cervical to brain central conduction time, and normal median scalp latencies) have been suggested to be indicators of electrophysiologic impairment due to tethered cord syndrome.

In the most comprehensive study to date, 90 children were followed with serial peroneal SSEPs after a repair of their spinal dysraphic lesions with the objective of evaluating whether SSEPs were a useful way of monitoring these children to facilitate early detection of clinically significant retethering. Three hundred and nine studies were performed on these children yielding a mean of 3.4 studies per patient. The median time between SSEP studies was 13 months. A clinical examination was performed at the time each SSEP was done. There was a false-positive rate of 71% and a false-negative rate of 43%. It was concluded that serial SSEPs do not correlate well with clinical status and are not a useful modality for monitoring patients at risk for retethering (167). The author has followed a large population of children with myelomeningocele for decades and similarly has not found mixed nerve SSEPs to be useful in the evaluation of secondary tethered spinal cord after myelomeningocele repair.

Intraoperative Spinal Monitoring

There are many reports detailing the usefulness of intraoperative SSEP monitoring during scoliosis surgery (168–172), as well as during other surgical procedures of the spine. The limitation of SSEPs is the fact that they only monitor afferent pathways in the dorsal columns. Over the past decade, intraoperative spinal monitoring has evolved to include monitoring of the motor pathways. The corticospinal tracts are now being routinely monitored intraoperatively using transcranial electrical stimulation of the motor cortex (172), with motor evoked potentials recorded from either peripheral motor axons or as a CMAP from innervated muscles. Transcranial electrical MEPs to monitor the corticospinal motor tracts are now used routinely in addition to SSEPs for the detection of emerging SCI during surgery to correct spine deformity or resect intramedullary tumors (63–65).

Brachial Plexus Injury

The dermatomal SSEP can be a useful supplement to the assessment of the child with a brachial plexus injury (173). The child needs to be awake during the study. The C5 and C6 dermatomal SSEPs are generally most useful in the author's experience. The C5 dermatome is stimulated over the lateral proximal shoulder using a proximal disk as the cathode and a distal disk as the anode.

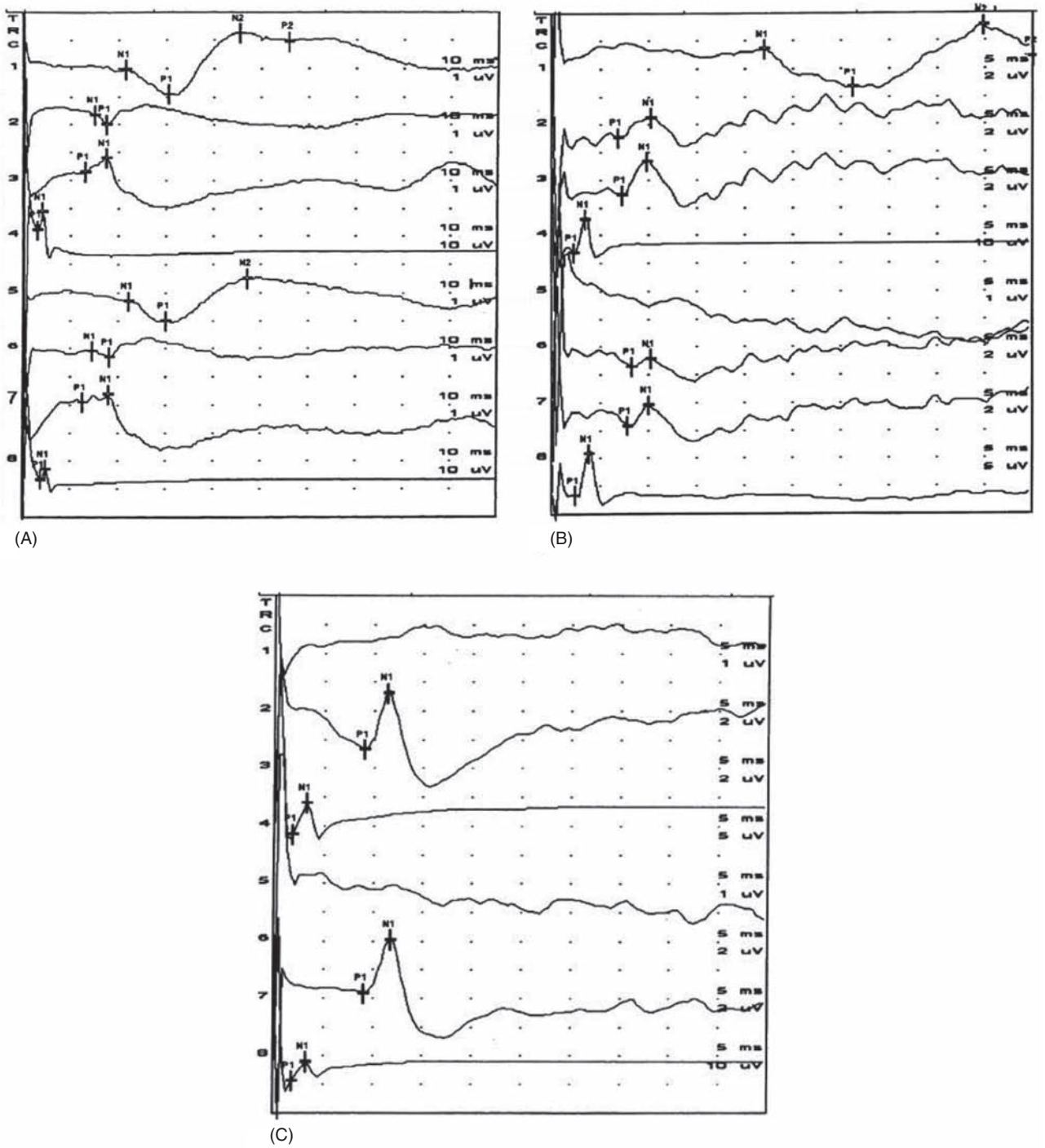

FIGURE 6.17 Tibial somatosensory-evoked potentials (SSEPs) obtained in the pediatric intensive care unit. Channels 1–4 are responses with left tibial stimulation, and channels 5–8 are responses with right tibial stimulation. Channels 1 and 5 are scalp responses (C2′ to Fz); channels 2–7 are spine responses (L2 spine referenced to flank); and channels 4 and 8 are peripheral responses obtained at the popliteal fossa. (A) Normal tibial SSEP study. (B) Abnormal tibial SSEPs in a child with left hemispheric brain injury. Peripheral and lumbar spine (L2 and T12 level) responses are normal bilaterally. The scalp response is normal with left tibial nerve stimulation (channel 1), but absent with right tibial nerve stimulation (Channel 5). (C) Abnormal tibial SSEPs bilaterally in an awake 4-year-old with low cervical spinal cord injury without radiographic abnormality. Peripheral (channels 4 and 8) and L2 spine (channels 2 and 7) responses are normal. Scalp responses (channel 2 and 5) are absent as a result of the low cervical spinal cord injury.

Intraoperative SSEPs with direct stimulation of exposed nerves may demonstrate incomplete injuries of upper cervical roots, a proximal stump of the ruptured C5 root with functional central continuity (thus, potentially suitable for grafting), or complete root avulsion. Preoperative diagnostic SSEPs, while a useful adjunct to conventional electrodiagnosis, do not enable one to discriminate incomplete cervical root avulsion from intact roots (174).

Demyelinating Diseases

Both SSEPs and brainstem auditory evoked potentials have been reported to be abnormal in children with, or carriers of, leukodystrophy (175,176). Peripheral and/or central abnormalities have been documented in metachromatic leukodystrophy Pelizaeus–Merzbacher disease, Krabbe disease, adrenoleulodystrophy, Canavan disease, Alexander disease, and multiple sulfatase deficiency (177).

Pediatric multiple sclerosis (MS), while relatively rare, does occur in preadolescents and adolescents (178). MRI has been shown to be slightly more sensitive than multimodal evoked potentials in confirming the clinical diagnosis of childhood MS (179). However, in suspected or probable MS, both SSEPs and visual evoked potentials may contribute to the determination of clinical diagnosis because of their capacity to demonstrate asymptomatic involvement in central somatosensory and central optic nerve pathways (180).

Acute transverse myelitis often results in severe myelopathy due to inflammation and demyelination. SSEPs have been shown to be abnormal in this condition and may provide prognostic information regarding the ultimate outcome (181).

The extent and location of nerve involvement in demyelinating peripheral neuropathies have been evaluated with SSEPs; however, SSEPs do not usually provide necessary additional information to standard nerve conductions. Hereditary motor–sensory neuropathy type I shows impaired peripheral conduction in both proximal and distal nerve segments with normal central conduction (182). AIDP patients have been shown to exhibit prolonged posterior tibial peripheral SSEP latencies in addition to prolonged or absent median F-waves. However, posterior tibial F-wave latencies and median nerve SSEPs were less sensitive studies for the detection of demyelination in AIDP (183). SEP can detect an abnormality and thus support the clinical diagnosis of GBS in the acute stage when the results of more conventional tests are inconclusive (184).

CONCLUSION

Pediatric EDSs are a useful diagnostic tool that aids in the localization of abnormalities within the lower motor neuron and often provide helpful prognostic information. EDSs have been less and less utilized in the diagnosis of many myopathic disorders and anterior horn cell diseases due to the importance of molecular genetic studies and/or muscle biopsy for the determination of disease subtypes. However, there remains a use for EMG and NCSs in many focal and generalized lower motor neuron conditions. For children suspected of having hereditary neuropathies, with no family member possessing genetic confirmation, a directed NCS may guide the acquisition of more specific and less costly molecular genetic studies. In other conditions such as GBS and focal neuropathic conditions, EDSs remain critical for diagnostic confirmation.

Practical suggestions relating to the pediatric electrodiagnostic evaluation have been provided. Study results must be interpreted in light of developmental and maturational issues affecting both clinical findings and electrophysiologic processes. A skilled electrodiagnostic evaluation utilizes careful strategic planning to provide the most important diagnostic information needed in an expeditious manner, with the least distress possible to the child and parent. Ongoing electrodiagnostic experience with the pediatric population provides increasing diagnostic acumen regarding pediatric lower motor neuron disease processes and sufficient technical skills to provide the referring physician with accurate diagnostic information.

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7

GUIDANCE TECHNIQUES FOR BOTULINUM TOXINS AND OTHER INJECTIONS

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Botulinum toxins (BoNTs) are an effective therapy for a wide variety of medical problems including muscle overactivity, pain, neurosecretory, urological, and ophthalmic disorders for patients of all ages. In pediatric patients, the most common clinical problems for which BoNTs are prescribed are muscle overactivity and sialorrhea associated with upper motor neuron syndromes (UMNSs) including cerebral palsy (CP) and acquired brain injury. BoNTs may also be recommended in children for conditions unresponsive to traditional treatments including muscle imbalance in congenital torticollis or obstetrical brachial plexus palsy, detrusor overactivity or dyssynergia, and migraine headaches.

To accurately select and target skeletal muscles, salivary glands, or other structures for injection with BoNT requires a fundamental knowledge of anatomy. To improve their accuracy in targeting when performing chemodenervation procedures, most physicians utilize one or more supplementary localization techniques in addition to relying on knowledge of surface anatomy, palpation and/or range of motion (ROM) (1). The goals of precisely targeting a structure for injection include optimizing the outcomes of the procedure, reducing BoNT dose required for treatment efficacy, and minimizing potential risks and/or adverse events associated with these procedures. Commonly used guidance methods include palpation, surface anatomy/reference guides, motor point/endplate targeting, electromyography (EMG), electrical stimulation (E-Stim) and image localization techniques (brightness mode [B-mode] ultrasound [US], fluoroscopy, computed tomography [CT]), and/or combinations of these techniques (2–5). To select the most appropriate guidance technique for a given patient and injection target, physicians must be familiar with the advantages and limitations of each of these targeting methods. What follows is a review of the advantages and limitations of the guidance techniques commonly used when performing BoNT injections.

PATIENT EVALUATION

When a patient arrives for a scheduled BoNT procedure, it is mandatory to take an interval history and examine the patient, even for a patient well known to the examiner. This is to rule out acute illness, infection, or other medical problems, which would necessitate rescheduling the procedure. Patients should also be questioned about their current medications (medications with anticholinergic effects may potentiate BoNT effects), anticoagulation levels, and whether they have received BoNT injections from another provider within the past 3 months. This last question has become of increased importance as multiple specialists who prescribe BoNT may be treating the same patient. When patients receive BoNT from more than one specialist, these procedures should be coordinated to avoid the use of an excessive number of units and/or to prevent "booster dosing."

TREATMENT PLANNING

Treatment planning prior to performing BoNT injections includes a thorough history and examination of the patient, functional assessment, and a review of the relevant anatomy. This evaluation is critical prior to performing any procedure as it will guide selection of the muscle or muscle groups or other targets. This assessment requires that a physician has an extensive knowledge of surface/cross-sectional anatomy of the region of interest including relevant muscles, salivary glands, vessels, nerves, bones, and nearby organs. In addition to structural anatomy, physicians must also be familiar with functional anatomy, kinesiology, and the biomechanics of movement. This knowledge is required for evaluation

of a patient's functional limitations and muscle selection. When performing BoNT injections, this knowledge of anatomy is the foundation upon which all other guidance techniques are based. Teaching tools that are available to clinicians to improve their knowledge of anatomy include various anatomy or biomechanics texts, print and/or electronic reference guides, anatomic simulators, and/or return to the gross anatomy lab (6–10).

ANATOMIC GUIDANCE METHODS

Anatomic guidance techniques rely on surface anatomy, knowledge of cross-sectional anatomy, palpation, and passive or active range of motion (PROM, AROM). The majority of anatomic reference guides used by physicians when performing BoNT injections were not developed for this purpose. These atlases were written to guide needle placement for diagnostic EMG procedures (11–13). While these texts may be useful for BoNT injections, they have limitations both for their original purpose and when used to guide BoNT injections. More recently, two anatomic atlases were published specifically for BoNT and/or other chemodenervation procedures (14–16).

TECHNIQUE

Once a muscle or muscles have been localized using a combination of surface anatomy, palpation, and PROM or AROM, the skin is disinfected and cleansed as per the physician or institution's protocol. Standard single-use hypodermic needles are used for the injection. Needle size and length are determined by the estimated depth of the muscle. For superficial muscles, a 30 g, 1-inch needle may be sufficient, for deeper muscles 26 to 27 g, 1- to 1.5-inch needles or even a 25 g, 2.5- to 5-inch spinal needle may be required.

EQUIPMENT

Surface anatomy and/or cross-sectional reference guides, hypodermic needles of various lengths, and other injection supplies (gloves, skin cleansers, gauze, band aids/plasters).

ADVANTAGES AND LIMITATIONS OF ANATOMIC GUIDANCE FOR BoNT THERAPY

Advantages

- All physicians receive training in anatomy in medical school.
- Most physicians have access to a variety of anatomic reference guides and are familiar with their use, and the guides are relatively inexpensive.
- Anatomic simulators can provide physicians with detailed information about orientation and function of muscles.

Limitations

- A physician's training in the anatomy lab and therefore his or her last review of gross anatomy may have been many years ago.
- Positioning: When treating patients with spasticity, positioning the patient as described in reference guides may be challenging, at best or may be unfeasible. When the patient is not positioned as described in the reference guide, the recommended site for needle insertion into the target may be incorrect. This limits the use of reference guides when performing BoNT injections in many patients.
- Even when used to guide diagnostic EMG, as they were designed to do, studies of the accuracy of surface anatomy and/or reference guides has been called into question, see Evidence discussion.
- Palpation and surface anatomy: While a few muscles may be easily identified by their surface anatomy and/or by using palpation, it may be difficult or impossible to correctly identify many muscles including:
  - Muscles of the neck or forearm where complex overlapping may make it difficult to correctly identify a target muscle
  - Deeply situated muscles in the limb or neck; where it may be impossible to palpate the target muscle and/or estimate muscle depth
  - Obesity obscures surface landmarks or palpation of the target muscle and limiting depth/location estimation
  - Disuse atrophy or atrophy caused by repeated BoNT injections may limit estimation of muscle depth
  - Patients in whom spasticity has caused anatomic rearrangements or deformities
  - Postoperative muscle changes following lengthening or transfers
  - Patient cooperation

Evidence supporting or refuting the use of anatomic guidance for BoNT injections: There is an increasing body of evidence that calls into question the practice of relying solely on anatomic guidance when performing BoNT injections.

CADAVER STUDIES

- A 2012 study evaluated the accuracy of injections into the gastrocnemius muscles of 30 cadavers using palpation and surface landmarks. 121 physicians injected ink into the gastrocnemius muscle followed by dissection to evaluate the accuracy of ink injections (17). Only 43% of the injections were successful with 57% of the

injections placed outside of the gastrocnemius muscle, either in the soft tissue (19.8%) or deep to the gastrocnemius, in the adjacent soleus muscle (37.2%).

- A 2011 blinded study compared "blind" (e.g., manual) versus US placement of a wire into 14 lower limb muscles in fresh cadavers. Two clinicians (a resident with 6 months' EMG experience and an attending ≥ 10 years' experience) performed the needle insertions and the accuracy was then verified by CT, and assessed by a third clinician (18). The overall accuracy in the 14 tested muscles with blind wire placement using anatomic guidance was 39% (range 0%–100%) whereas the accuracy for US-guided wire placement was 96% (range 50–100%). The only muscles in which blind placement was 100% accurate were the tibialis anterior and short head of the biceps femoris, whereas with US guidance the only muscle with less than 100% accuracy was the semitendinosus muscle. Unexpectedly, the accuracy of anatomic guidance/blind placement was 0% for needle insertions into semitendinosus, rectus femoris, and extensor hallucis longus. When comparing the less and more experienced clinicians, there was no significant difference in accuracy of needle placement in the target muscle. The experienced clinician was only more accurate in the trajectory of needle insertion toward the target muscle.
- A 2003 study evaluated the accuracy of fine-wire insertions by three physicians (with varying degrees of EMG experience) into 36 lower limb muscles in 10 cadavers (263 muscles) using standard EMG anatomical reference guides (11,19). The accuracy of wire placement was checked by anatomical dissection by an anatomist. In this study, 57% of wire insertions penetrated the target muscle; however, the wire tip was only in the target muscle 45% of the time. As with the previously mentioned 2011 study, there was significant variability in the accuracy of targeting different muscles from 100% for vastus medialis to 0% for 12 attempts in the hip flexors. The proximity of the wire to other structures was also reported with 17% of insertions either penetrating or passing within 5 mm of an important structure. The authors concluded that the accuracy of blind wire placement using EMG reference guides was quite variable and the development of safer strategies was recommended.

CLINICAL STUDIES

Comparison of Anatomic Localization With Other Localization Techniques for BoNT in Limb Muscles

The results from anatomic/cadaver studies are supported by clinical studies evaluating the accuracy of needle placement using anatomic techniques in upper and lower limb muscles.

- Manual placement versus EMG: A 2013 prospective study describes the development of a structured protocol using surface anatomy and PROM to localize lower limb muscle injection sites for BoNT injections. The described manual localization protocol details the origin, insertion, innervation, and function of the muscles; how to position the patient, localize the muscle belly, and support the limb, the site, and direction for needle insertions; and the PROM procedure to verify needle location. The authors report that the accuracy of needle insertion using this protocol will be verified using E-Stim. All patients were sedated for the BoNT procedure. While this study provides a description of the protocol, it provides no results from data collection. Therefore the accuracy of this manual/PROM localization protocol is unknown (20).
- Manual placement versus EMG: A randomized controlled trial (RCT) in 27 adult patients with spasticity from UMNS (brain injury, spinal cord injury) compared the effectiveness of BoNT injections in upper and lower limb muscles guided by EMG versus injections guided by landmark-based systems. Outcome measures included evaluation of the reduction of spasticity using the Modified Ashworth Scale (MAS) and the functional outcome using the Modified Barthel Index. While a reduction in MAS score and improved Barthel Index score was noted in all subjects, the degree of improvement in spasticity and function was greater in patients where BoNT injections were guided by EMG. The authors concluded that when performing BoNT injections for the treatment of spasticity, the use of EMG to guide injections was superior to injections guided by anatomic/landmark-based reference guides (21).
- Manual versus EMG: A 2003 study of the efficacy of BoNT injections in adult patients with focal hand dystonia reported superiority of EMG compared to anatomic guidance (22).
- Manual placement versus E-Stim: A 2009 study in children with CP (hemiplegia or diplegia) compared the efficacy of BoNT injections guided either by palpation or E-Stim. At 3 months, patients who had injections guided by E-Stim had a statistically greater reduction in MAS scores, PROM, Composite Spasticity Scale scores, and Gross Motor Function Measure scores than those patients injected using manual guidance alone (23).
- Manual placement checked by E-Stim: A 2005 study in 226 children with CP investigated the accuracy of manual needle for BoNT injections checked with E-Stim to confirm needle position in upper and lower limb muscles (1,376 needle insertions) (24). The needle insertion site was determined by surface anatomy/manual placement and depth estimated by limb size and using PROM. Once the clinician was satisfied with the position of the needle, the accuracy was checked by using E-Stim. During stimulation, the clinician observed the pattern of muscle twitch and whether the twitch response was in the target muscle, or other muscles.

The reported accuracy of manual placement was as follows: gastrocsoleus 78%, hip adductors 67%, medial hamstrings 46%, tibialis posterior 11%, biceps brachii 62%, pronator teres 22%, flexor carpi radialis (FCR) 13%, flexor carpi ulnaris (FCU) 16%, and adductor pollicis 35%. The authors concluded for the muscles tested that manual placement was adequate only in the gastrocnemius. They also postulated that inaccurate muscle targeting could be responsible, at least in part, for a lack of response or insufficient clinical response following BoNT injections in children with CP.

- Manual placement, E-Stim and US: A 2012 RCT of 49 adult patients evaluated the efficacy of BoNT injections in the gastrocnemius muscle of 49 adult patients with poststroke spasticity (PSS) (25). The study compared the efficacy of three localization techniques (manual, E-Stim, US) using a fixed dose and dilution protocol (onabotulinumtoxinA 100 units medial head, 100 units lateral, dilution 100 units reconstituted with 2 mL preservative free normal saline). At 4 weeks, the patients injected with US guidance had a greater reduction in MAS score than in patients injected with manual needle placement. The US injection group also had a greater increase in PROM compared with the E-Stim and manual injection groups. There was no difference in the Tardieu Scale score between the 3 groups. The authors concluded that when performing BoNT in the gastrocnemius muscle of adult patients with PSS, US guidance provided a greater reduction in MAS and clinical benefit than injections guided with manual needle placement or E-Stim.
- Manual placement checked by US: In a 2009 study of 39 children with CP, the accuracy of blind needle placement in the gastrocnemius muscles was checked using US by a blinded clinician (26). The authors reported an overall accuracy of blind placement in the gastrocnemius of 78.7%. The accuracy of needle placement in the thinner lateral gastrocnemius was 64% overall (46% in younger patients), whereas accuracy of needle placement in the medial gastrocnemius was 93% overall (87% in younger patients). The authors concluded that supplementary localization should be considered for the medial gastrocnemius muscle in younger patients and for the lateral gastrocnemius for all patients.
- Manual placement checked by US: In a second 2009 study of 54 children with CP, the authors evaluated the effect of a number of variables on the effectiveness of lower limb BoNT injections (27). Variables included patient age, BoNT dose, BoNT dilution, muscles injected, and guidance method for injections. BoNT injections were guided by manual needle placement in 44% of patients and by US in 56% of patients. The authors reported a greater efficacy of injections guided by US, in patients less than 6 or greater than 12 years of age, when the muscles injected were hamstrings or gastrocnemii and when the dose/muscle was greater than 0.8 units/kg of onabotulinumtoxinA. Dilution had no effect on efficacy. The authors concluded that this study confirmed the usefulness of US guidance for BoNT injections in lower limb muscles.
- Location of muscle/muscle fascicles using recommendations from reference guides checked by US: In a 2010 study of forearm flexor muscles in patients with spasticity, the location of the muscle or muscle fascicles using anatomic reference guides was compared to the position of the muscle/muscle fascicle using US. There were significant differences between the estimated position of the muscle or muscle fascicles compared to US for the FCR, flexor policis longus (FPL), and for the fascicles of the flexor digitorum superficialis (FDS) (28).

CONCLUSIONS

Although a thorough knowledge of surface and functional anatomy is a requirement when performing BoNT injections and critical for muscle selection, the use of this knowledge alone may not be the optimal method to guide BoNT injections. While many clinicians report a good effect with BoNT using blind needle placement, no study to date has shown this technique to be superior to using a supplemental guidance method. Due to the limitations of anatomic guidance techniques, many if not most clinicians choose to use one or more supplementary localization techniques when performing BoNT injections.

MOTOR ENDPLATE TARGETING OR LOCALIZATION TECHNIQUE

It is well established that to exert its action BoNT avidly binds to and is internalized at cholinergic nerve terminals including at the neuromuscular junction. Knowing this, clinicians and researchers have suggested (or investigated) that targeting the endplate zones or motor points may enhance the uptake of the toxin (29–32). The technique of motor point or endplate targeting requires knowledge of the location and distribution of motor endplates (MoEPs) in human skeletal muscle.

The location of motor points or endplate zones in mammalian muscles has been studied in animal models and in humans using histochemical staining and electrophysiologic methods (32–35).

STUDIES ON THE LOCATION OF MOTOR ENDPLATE LOCATION AND DISTRIBUTION

In the 1950s, two researchers published data on the location and distribution of MoEP in human skeletal muscles. Coers described three types or arrangements of MoEP in human muscle: (a) muscles having a single innervation band, (b) muscles with multiple innervation bands, and (c) muscles where the innervation bands were scattered

throughout (35). Christensson published data on the distribution and pattern of MoEP in stillborn infants. She reported that MoEPs were distributed in a single transverse band at the midpoint of unipennate muscles and in a concave band in bipennate muscles (gastrocnemius) (34).

In the last decade, additional anatomical studies detailing the location of MoEP have been published (29,36,37). Kim et al. reported that the MoEPs in gastrocnemius and soleus muscles were distributed along the length of the muscle. They reported the location of the most proximal of MoEPs in the medial gastrocnemius, lateral gastrocnemius, and soleus at 9.6% (+/- 3.5%), 12.0% (+/- 3.4%), and 20.5% (+/- 3.9%, of the lower leg length. The most distal MoEPs were reportedly located at 37.5% (+/- 5.5%), 37.9% (+/-2.3%), and 46.7% (+/- 3.6%) of lower limb length, respectively (36).

In the biceps brachii muscle, an inverted V arrangement of MoEP has been reported (37). The authors reported the MoEP zone to be 1 cm in width, laterally located 7 cm superior to the olecranon, in the midline located 11 cm superior to the olecranon, and medially 8 cm proximal to the olecranon. The authors also reported the ratio of MoEP location to total olecranon–acromion length: 0.25 at the lateral edge, 0.39 at midline, and 0.28 medially.

The location and distribution of MoEP in the psoas muscle of adult cadavers were reported in a 2010 study. The authors reported an average of 3.7 (range 2–7) nerve branches from the lumbar plexus innervating the long pennate psoas muscle, which was made up of converging fibers of variable length. The area of the MoEP zone was reported to correlate with a zone between 30.83% and 70.25% of the distance from T12 to the inguinal ligament. Therefore the majority of the MoEPs were proximal to the sacral promontory (29).

MoEP TARGETING FOR BoNT INJECTIONS

Clinicians have suggested or recommended targeting MoEP for BoNT injections, for decades (30,31,38). There is limited data from trials comparing MoEP targeting to other techniques.

LOWER LIMB

In a 2014 double-blind randomized controlled trial (DB-RCT), Im et al. compared the efficacy of a fixed dose of BoNT with injections placed within the MoEP zone suggested by anatomical studies (2/10 and 3/10 of calf length) to injections below the mid-belly of the muscle. Both groups improved and there were no statistical differences between the two groups in either clinical or electrophysiologic measures (39). In a 2011 review, Van Campenhout et al. published the location of MoEPs in lower limb muscles and anatomical guidelines for the use of MoEP targeting for BoNT injections. Based on the location of MoEP, the authors recommended an optimal injection zone for the gastrocnemius, soleus, tibialis posterior, semitendinosus, semimembranosus, biceps femoris, gracilis, rectus femoris, adductors longus, brevis, magus, and psoas muscles.

UPPER LIMB

In a 2009 DB-RCT, Gracies et al. used published information on the location of the MoEP in the biceps brachii to compare the effectiveness of a fixed dose of onabotulinumtoxinA using MoEP targeting, standard dilution (100 units in 1 ml), and high volume dilution (100 units in 5 ml). The authors reported a greater benefit with injections using MoEP targeting and those with high volume dilution of onabotulinumtoxinA (40).

MoEP TARGETING TECHNIQUES

The use of MoEP targeting can be incorporated into conventional targeting techniques used to guide BoNT injections. When using manual or blind needle guidance for BoNT injections, physicians can make use of published motor point maps or the information (where available) as to the location of MoEPs or MoEP zones (29,36,37). When using EMG, physicians can target MoEP by listening for endplate noise and injecting toxin in this zone/location (30). If using E-Stim, MoEPs are targeted by repositioning the needle within the muscle and maintaining a maximal or visible twitch while reducing the stimulation intensity (40). While MoEP cannot be visualized with US or other imaging-based guidance techniques, the published information on the location of MoEPs in muscles can be used when using US to guide BoNT injections.

ADVANTAGES AND LIMITATIONS OF MoEP TARGETING FOR BoNT THERAPY

Advantages of MoEP Targeting

Targeting of MoEP when performing BoNT chemodenervation procedures has at least a theoretical advantage over injections placed in areas away from the MoEP. The data on the superiority of MoEP targeting is limited and additional controlled trials are needed to determine whether all BoNT injections should be performed using this technique.

Disadvantages of MoEP Targeting

A potential disadvantage of MoEP targeting is that it may increase the time required to perform a BoNT procedure.

Using this technique requires measuring the limb and marking MoEP zones. However, this information is memorized or can be transcribed to a template or form, which can be used in clinical practice to speed up this process.

If MoEP targeting is not superior, then the extra time and effort required for this method is unnecessary.

MoEP targeting, as currently described, is not useful for nonmuscle targets. Studies of the location of other cholinergic nerve terminals in organs targeted for BoNT injections may become available in the future.

CONCLUSIONS

While there is limited data on the superiority of MoEP targeting over injection at other sites in skeletal muscles, this practice requires minimal time or effort. Therefore, clinicians should consider incorporating this technique into whatever guidance technique or techniques that they currently use to guide BoNT injections into muscles.

ELECTROMYOGRAPHY GUIDANCE FOR BoNT INJECTIONS

EMG is the most commonly reported or recommended localization technique used for BoNT injections in muscle targets, other than anatomic guidance (1, 22, Botox PI, Dysport PI, Xeomin PI, Myobloc PI). Obviously, EMG is not helpful for localizing or injecting nonmuscle targets and is not necessary for most muscles of the face.

When using EMG to guide BoNT injections, available equipment options include a standard electrodiagnostic machine or one of the inexpensive commercially available small portable EMG audio or audiovisual amplifier units. EMG-guided injections require the use of sterile, single-use, insulated injecting needle electrodes. These needles are available in a range of lengths (25–75 mm). The choice of needle length/size is determined by the depth of the target muscle(s).

TECHNIQUE FOR EMG-GUIDED BoNT PROCEDURES

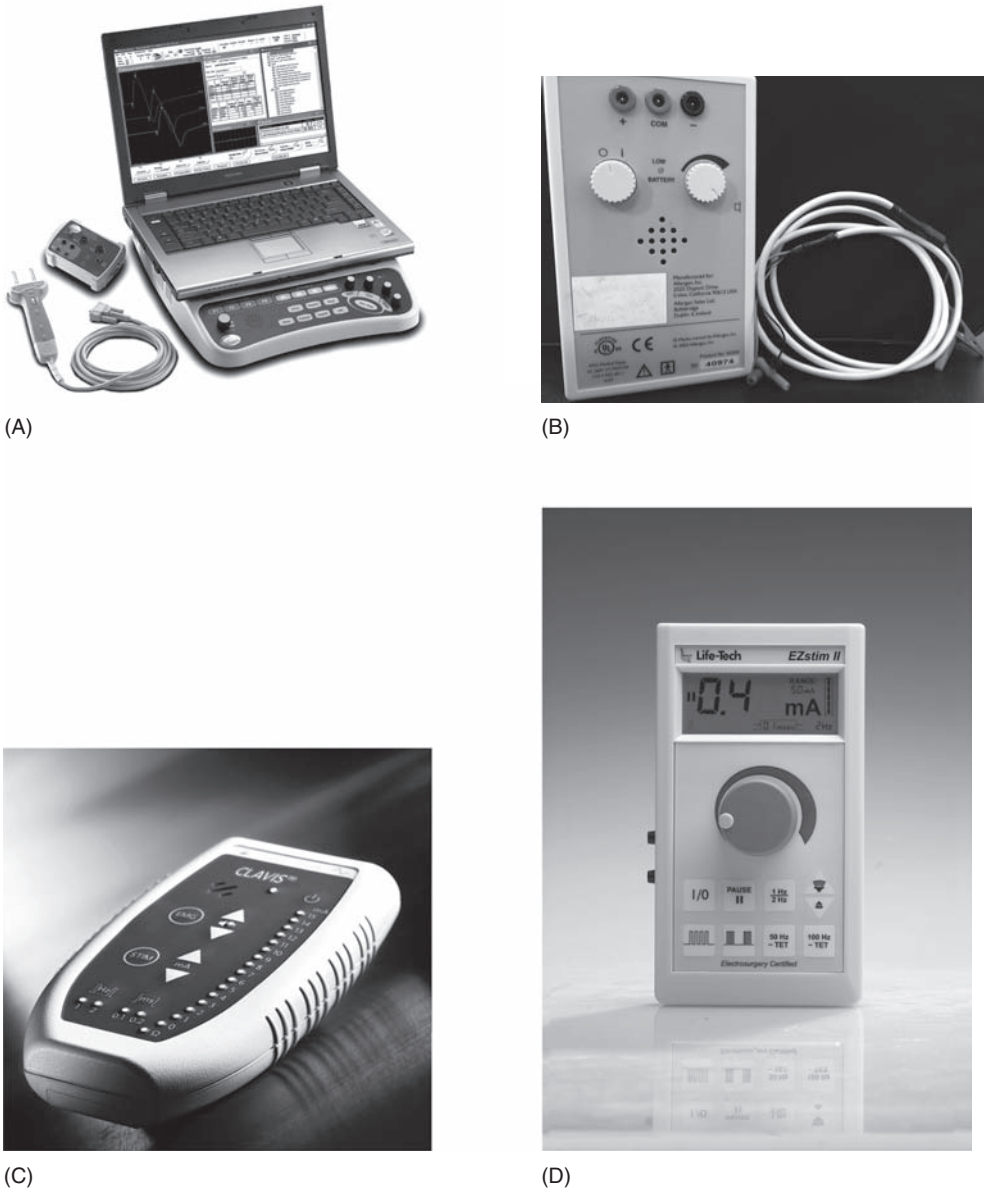
When EMG is used to guide BoNT injections, identifying the optimal site for needle insertion begins with inspection of the limb followed by palpation and ROM. PROM or AROM while palpating the limb or body part may facilitate muscle localization. Once the site for needle insertion has been selected, the skin is then disinfected or cleansed and the needle electrode is inserted through the skin, advancing the needle to the target while listening for audible EMG activity. To facilitate muscle localization, the patient should be instructed to either relax or contract the target muscle (if he or she has adequate selective motor control or can cooperate with this request). If the patient has impaired selective motor control, asking him or her to contract (or relax) a given muscle may result in mass synergy or cocontraction in multiple muscles, which may limit the accuracy of EMG guidance. In patients with impaired motor control, another option is for the examiner to perform PROM while recording EMG. The movement of the muscle may trigger activation of motor units or insertional activity as the needle moves within the muscle during passive motion (2).

When using EMG to guide BoNT injections, the clinician listens as the recording needle electrode is advanced into the target muscle. As the needle nears an actively contracting muscle fiber, the tone associated with EMG activity will change from a dull or low-pitch to the crisp high-pitched sound, which is characteristic when recording nearby motor unit action potentials (MUAPs). This tonal change from dull to crisp indicates that the needle electrode is near the firing muscle fiber. If the tone remains dull, then the needle position should be adjusted until a crisp tone is heard.

Localizing a target muscle using EMG requires simultaneous contraction in the target and relaxation in antagonist muscles and/or adjacent muscles, which may have a similar action. While this process of contraction and relaxation may be facile for some patients, it is often difficult for many patients, particularly those with UMNS, impaired motor control, or cognitive impairments. Patients with UMNS and spasticity may have impaired reciprocal inhibition. This may lead to firing in muscles at rest or when the muscle is moved in the direction of the antagonist and firing in multiple muscles. Cocontraction, mass synergy, and the loss of reciprocal inhibition often limit the usefulness of EMG to isolate a specific muscle for injection. For example, when a needle electrode is inserted into the forearm to target the FDS, there may be cocontraction or firing in the other flexor digitorum profundus (FDP) or wrist flexors. Recording EMG activity in this circumstance only indicates the needle is in an active muscle but it may not be possible to determine which muscle is firing. PROM of the various muscles may be useful in this circumstance as the needle electrode may reveal increased firing in the stretched muscle. E-Stim may also be more useful than EMG for muscle localization when cocontraction is present. Careful inspection of the patient's abnormal posture may help determine which muscle is contributing to the pattern (1,3).

EQUIPMENT FOR EMG GUIDANCE

Electrodiagnostic machines, EMG audio amplifiers, and EMG audio/liquid-crystal display (LCD) units (Figures 7.1A–C) are used as equipment. Other supplies include monopolar insulated injection electrodes/needles of various lengths, surface electrodes, gloves, skin cleansers, gauze, and band aids/plasters.



(A) (B) (C) (D)

FIGURE 7.1 (A) EMG machine; (B) EMG audio amplifier; (C) EMG E-Stim combined; (D) E-Stim unit.

ADVANTAGES AND LIMITATIONS OF EMG FOR BoNT PROCEDURES

Advantages

- One of the main advantages of EMG is that it provides auditory feedback indicating the level of activity or overactivity in a muscle.
- In adult patients with cervical dystonia, EMG is often useful to determine whether a muscle is active and therefore contributing to the patient's abnormal head posture (21,25).
- In adult patients with focal limb dystonia, EMG is often useful to isolate individual muscle fascicles.

Limitations

- While EMG may be useful to isolate a muscle or muscle fascicle in patients with focal dystonia, it may be less useful in patients with UMNS-related muscle overactivity (spasticity, dystonia) where;
  - The presence of cocontraction, mass synergy, and loss of reciprocal inhibition may lead to diffuse activation of multiple muscles.
  - EMG activity in muscles surrounding the target may make it impossible to correctly isolate a muscle. For example, when a needle electrode is inserted into the forearm of a patient with flexion synergy, it may only be possible to state that the needle is in an active muscle, but it may not be possible to determine which muscle.

- The usefulness of EMG may also be limited when patients are sedated for BoNT injections, particularly when general anesthesia is used.
- Patients often report more pain with the insertion of Teflon-coated needle electrodes than with standard hypodermic needles.
- Insulated needles are more costly than hypodermic needles.

EVIDENCE

There is limited evidence, particularly in pediatric patients, to support or refute whether EMG improves the efficacy or safety of BoNT injection or of its superiority over other techniques.

- See the previous section, "Clinical Studies; Comparison of Anatomic Localization With Other Localization Techniques for BoNT in Limb Muscles," for details from several studies, which showed superiority of BoNT injections guided by EMG compared to anatomic guidance in adult patients with spasticity and focal hand dystonia (21,22).
- In a 2012 study, Hong et al. compared the incidence of adverse events (dysphagia) when BoNT injections were guided by EMG versus US. The incidence of dysphagia was 34.7% in patients where the procedure was guided by EMG and 0% in the same patients when the procedure was guided by US (41).
- In a 1996 methodological study comparing EMG to E-Stim for adults with focal hand dystonia, there was no significant difference in outcome measures between the two techniques (42).
- In a study of adults with cervical dystonia, the authors reported that EMG guidance may reduce the number and severity of side effects following BoNT injections (43). Additional studies comparing the EMG and other guidance are reviewed in the sections following.

SUMMARY

EMG may provide useful information as to whether a muscle is active and/or contributing to the clinical problem. It may be challenging or difficult to determine if the EMG needle is actually in the target muscle. The combination of US and EMG may be useful as US can direct the needle to a target muscle and EMG will determine if the muscle is active.

ELECTRICAL STIMULATION GUIDANCE FOR BoNT INJECTIONS

E-Stim is only useful for BoNT injections in muscles and is not required when targeting superficial muscles of the face. When using E-Stim for chemodenervation procedures, a nerve trunk can be stimulated, which will activate the entire muscle or group of muscles innervated by the stimulated nerve. Alternately, E-Stim can be used to stimulate distal motor nerve branches to activate muscle fascicles, for example, motor point stimulation or for MoEP targeting (1,40). Nerve trunk stimulation is typically used for diagnostic nerve blocks using local anesthetics or for phenol chemoneurolysis procedures. When using E-Stim to guide BoNT injections, physicians typically use motor point or endplate targeting techniques. These techniques are also used for phenol chemoneurolysis.

TECHNIQUE

When using E-Stim to guide BoNT injections, selection of the most appropriate site for needle insertion begins with inspection of the limb or body part, PROM, and AROM. To determine this site, many physicians also use published information on the location of endplates within a muscle, if this information is available (see the previous section on endplate location/targeting).

Once the location of the needle insertion is identified, the skin is cleaned or disinfected using the physician or institution's standard protocol. The physician then inserts the needle electrode through the skin advancing the needle toward the muscle target. When the physician estimates that the needle is near or within the muscle target, the stimulator is turned on and the intensity of stimulation is adjusted up to produce a muscle twitch, typically 1 to 3 mA. The needle is then advanced, redirected, or repositioned so that successive reductions in stimulation intensity continue to produce a maximum twitch in the desired muscle. When performing motor point blocks, the reported goal for stimulation intensity is 0.025 to 0.5 mA (1). If upon stimulation, contraction occurs in several muscles or a muscle other than the target, the clinician should reposition the needle to isolate the target muscle. Care must be taken to avoid overstimulation, which may lead to volume conduction.

EQUIPMENT FOR E-STIM GUIDANCE

A nerve stimulator (either a portable handheld unit or the stimulator from an electrodiagnostic machine) is used (Fig 7.1A, C, D). The stimulator unit must have a port to which the cable of the insulated injecting electrode (needle) is connected. Other supplies include monopolar insulated injection electrodes/needles of various lengths, surface electrodes, a cable to connect the reference electrode to the stimulator, gloves, skin cleansers, gauze, and band aids/plasters.

ADVANTAGES AND LIMITATIONS OF E-STIM FOR BoNT INJECTIONS

Advantages

- The primary advantage of E-Stim when compared to using anatomic or EMG guidance is that E-Stim produces a direct visual feedback confirming that the needle is in the target muscle (i.e., muscle twitch/contraction and/or joint movement).
- E-Stim can be used in a patient for whom mass synergy or cocontraction limits the usefulness of EMG.
- E-Stim can be used when patients are sedated, where EMG is often not helpful.
- E-Stim may help isolate muscles that are difficult to isolate with EMG (deep or overlapping muscles, thin muscles such as the rectal sphincter).
- Studies have shown that E-Stim is more accurate than manual needle placement (24,44).

Disadvantages

There are a number of disadvantages to E-Stim not the least of which is that electrical stimulation increases the pain of the procedure (3,25,44,45).

- The stimulation and repeated insertions and adjustments of the needle that are necessary with E-Stim often cause pain and may be difficult for patients to tolerate, even cooperative adults.
- Most if not all pediatric patients will require general sedation and analgesia.
  - Sedation may increase the risk of BoNT procedures, particularly in compromised patients.
  - Sedation increases the time and cost of the BoNT procedure.
  - Sedation increases the time away from work or school for the patient and/or parent.
- When the needle is outside of the target muscle, overstimulation (excessive current) can lead to volume conduction and therefore contraction in the target muscle. The resulting muscle twitch may lead the physician to falsely conclude that the needle is in the target muscle when in fact it is located elsewhere.
- When stimulation occurs for a motor nerve branch prior to its insertion in the targeted muscle, stimulation will lead to a muscle twitch but the needle tip is outside of the muscle. The physician may falsely conclude that the needle is in the target muscle and toxin may be injected at the wrong site.

EVIDENCE

Although E-Stim is commonly used to guide chemodenervation procedures, there is limited evidence from controlled trials to support or refute the superiority of E-Stim over other guidance techniques for BoNT procedures.

- See the previous section, "Clinical Studies; Comparison of Anatomic Localization With Other Localization Techniques for BoNT in Limb Muscles," for studies comparing E-Stim to manual, EMG, and US guidance techniques.
- Surface E-Stim, following BoNT injections in calf muscle, has been shown to potentiate or increase the effects of the toxin in the stimulated muscle (46,47).

IMAGING-BASED GUIDANCE FOR BoNT INJECTIONS

Fluoroscopy, CT, MRI, and US all have been reported for guiding BoNT injections (4,5,45,48–50). Because of the exposure to ionizing radiation with CT and fluoroscopy, these techniques are used much less frequently than MRI and US. US is the most commonly reported image guidance technique for BoNT injections because it provides continuous real-time, high-resolution images and because it is less costly and more accessible and convenient than MRI, CT, or fluoroscopy (5,45,48). MRI-, CT-, or fluoroscopy-guided procedures are typically performed by interventional radiologists. Because most clinicians who perform BoNT injections do not use these techniques, only US guidance will be reviewed (2,45).

US is used to guide an array of invasive procedures including those targeting musculoskeletal structures (51). In the past 10 years, the reports of US used to guide BoNT injections and nerve blocks have grown substantially (4,25,26,28,41,48,52–,57). This growth is in part related to the development of high-frequency transducers, which provide exquisitely detailed images with a resolution similar to MRI and the increased access to lower cost portable US machines (14,15). In addition, unlike EMG or E-Stim, US can be used to guide BoNT injections in muscle and nonmuscle targets, including the salivary glands (45,57).

US PHYSICS AND SONO-ACOUSTIC PROPERTIES OF TISSUES

A full discussion of the physics of US and scanning techniques is beyond the scope of this chapter and the reader is referred to several reviews and a text (4,15,45,48). US scanning is made possible by piezoelectric crystals, which are responsible for generating and receiving returning soundwaves or echoes. These crystals are placed into arrays within a transducer, which is used when scanning with US. Sound waves are generated when piezoelectric crystals convert electrical pulses (generated by the US machine) into mechanical vibrations. When a transducer is placed in contact with a patient, these mechanical vibrations or sound waves are transmitted through the patient's skin and on to deeper tissues. Sound waves transmitted

through the body are then scattered, refracted, or reflected off various interfaces within the body. The sound waves that are reflected back to the transducer are converted by the piezoelectric crystal back into electrical pulses. These pulses are processed by the central processing unit (CPU) of the US machine (using time–distance coefficients) to create real-time grayscale US images viewed on the US display screen (58).

The *frequency of the transducer* determines the resolution of the image and the depth of penetration of the US beam. Higher frequency sound waves have a higher sampling rate and therefore provide a higher resolution images. This detail is at the expense of depth of penetration as soft tissues act as a high-frequency filter and limit the transmission of higher frequencies to deeper structures. Most transducers have a mix of frequencies, for example, 5 to 12 MHz, 3 to 5 MHz, and so on. This allows scanning of tissues at various depths (45,58).

The *echogenicity of a tissue*, and therefore its appearance on the US display screen, is determined by its impedance to sound waves, which is termed its acoustic impedance. When a tissue has no impedance to sound wave transmission, sound will travel through a tissue without being reflected. When sound waves encounter tissue interfaces of differing acoustic impedances, sound waves are reflected, refracted, or scattered off these interfaces. When scanning, if no sound waves return to the transducer, then the image on the screen will be black, or anechoic. When most of the sound waves pass through tissues without being reflected and only a few sound waves return to the transducer, the image on the display screen will appear dark or hypoechoic. When a tissue is highly reflective of US, most waveforms are reflected back to the transducer and the image on the display screen will appear bright or hyperechoic. Tissues with higher water content are relatively hypoechoic. Those with low water content and those with a higher content of fibroconnective tissue or calcium (bone cortex) will appear hyperechoic.

When scanning with US, tissues are described by their internal echotexture and echogenicity. Most, but not all, organs in the human body are of a mixed echogenicity. For example, skeletal muscle is composed of contractile fascicles, which are relatively hypoechoic, and noncontractile fibroconnective tissue, which in contrast to the fascicles appears relatively bright or hyperechoic (Figures 7.2A,B). In contrast, glandular tissues such as salivary gland or thyroid are homogenous in composition and echotexture. Therefore its appearance on the display screen is a uniform gray scale (Figure 7.2C). The appearance of some tissues may also vary with the scanning plane, for example, in long axis versus transverse planes. For example, when scanned in a longitudinal plane, muscle has the appearance of long thin hypoechoic bands (contractile fascicles) surrounded by and interspersed with linear hyperechoic bands (noncontractile fibroconnective tissue) (Figure 7.2A). In a cross-sectional or transverse view, muscle will have a specked appearance made up of hypoechoic fascicles and hyperechoic connective tissue (Figure 7.2B). On B-mode scans, blood vessels will appear hypoechoic, tendons are highly echogenic/hyperechoic, and nerves have a mixed hyperechoic, hypoechoic appearance (Figure 7.3A).

TECHNIQUE FOR US GUIDED FOR BoNT INJECTIONS

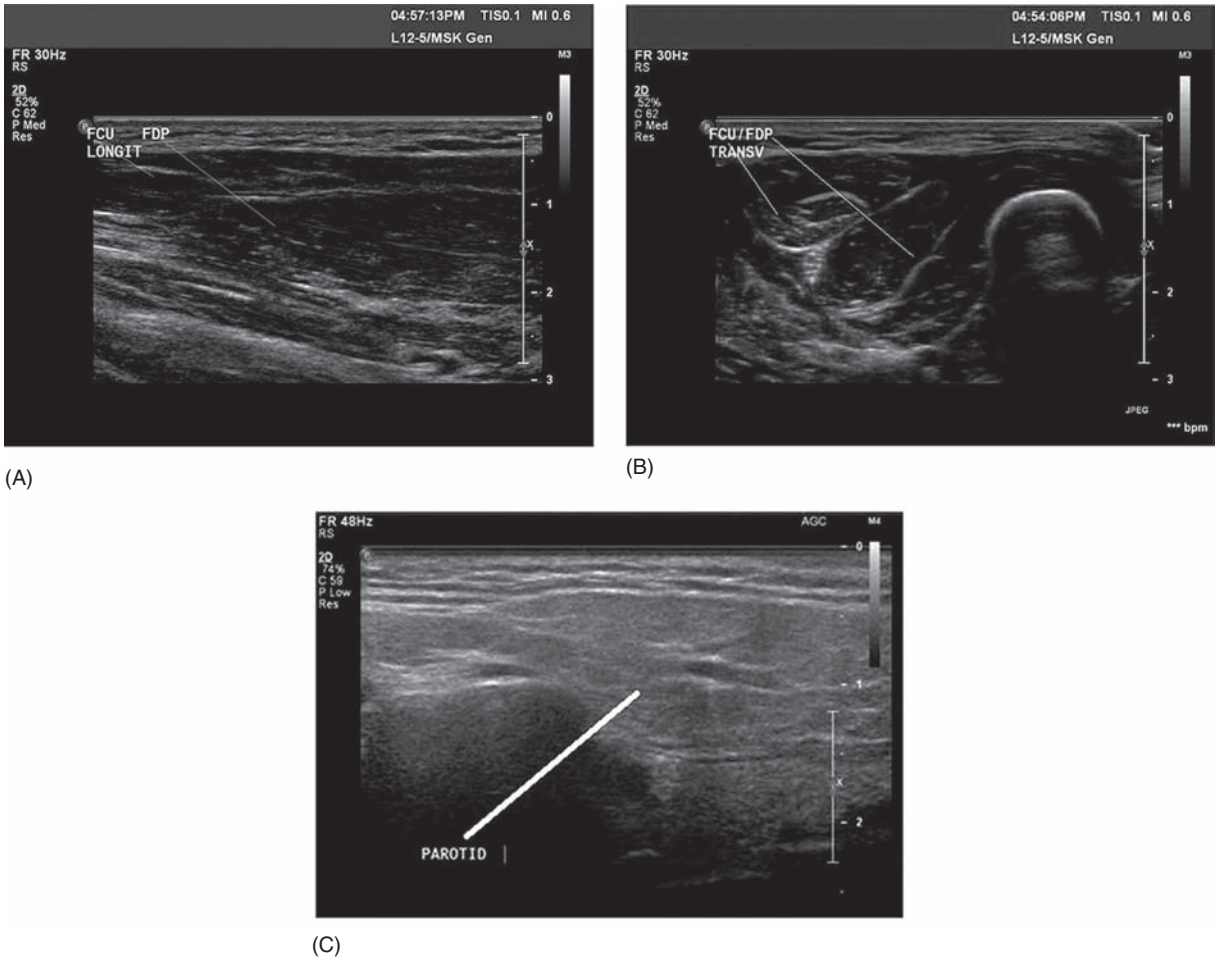
When using US to guide BoNT injections, as with other supplemental guidance techniques, the procedure begins with inspection of the limb/body part, palpation, spasticity assessment, PROM, and AROM. The patient is then positioned to expose the area to be scanned and then injected. This may require the assistance of another clinician in patients with UMNS, involuntary movements, dynamic contractures, or with less cooperative patients.

After turning on the US machine, the physician enters the patient data into the machine, selects the most appropriate transducer and desired machine preset program (Musculoskeletal/MSK for muscle injections, thyroid for salivary gland nerve block for diagnostic/therapeutic nerve blocks). When selecting a transducer for a procedure the sonographer should choose the transducer of the highest frequency, which provides an adequate depth of field view as this will provide the highest resolution image. To optimize imaging of the structure of interest, the physician must adjust the depth, number of, and position of focal zones, and gain (14). Some US instruments have automatic settings for the number of and position of focal zones, but may allow the physician to adjust these settings. Following this, the desired scanning mode is selected. B-mode and color Doppler are the most frequently used scanning modes when performing *US-guided chemodenervation procedures*. B-mode provides real-time grayscale images of structures and continuous visualization of the needle, target of injection, the toxin as it is injected, and other structures within the field of view. Color Doppler is useful in identifying blood vessels in the field of view so as to avoid them when inserting the needle and directing it into the muscle (Figure 7.3B).

Gel is applied to the transducer and the transducer is placed in contact with the patient to reduce impedance to sound waves. The transducer is then placed in contact with the patient's skin and the entire region of interest is scanned in transverse and longitudinal imaging planes (Figures 7.4A,B). The US beam created by the transducer is quite thin (the width of a credit card) (45,58). As a result of the limited field of view of the US beam, the entire region must be scanned thoroughly to identify not only the muscle or muscles of interest, but also structures such as nerves, vessels, and adjacent organs.

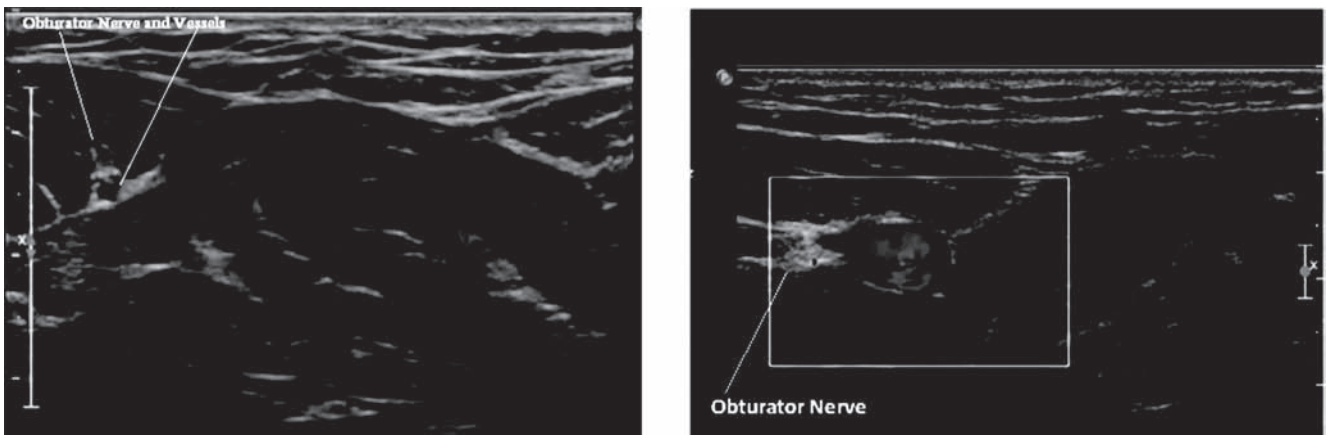
Muscles are identified based on pattern recognition, for example, their characteristic shape and by nearby structures (Figures 7.2B, 7.3A,B, 7.5A,B).

Once the muscle position and depth have been identified, the physician chooses the most appropriate



(A) (B) (C)

FIGURE 7.2 (A) Flexor carpi ulnaris and digitorum profundus, longitudial B mode ultrasound scan; (B) flexor carpi ulnaris and digitorum profundus, transverse B mode ultrasound scan; (C) Parotid gland.



(A) (B)

FIGURE 7.3 (A) Adductor muscles oburator nerve and vessels, transverse B mode US scan; (B) adductor muscles, obturator nerve, and vessels transverse color Doppler scan.

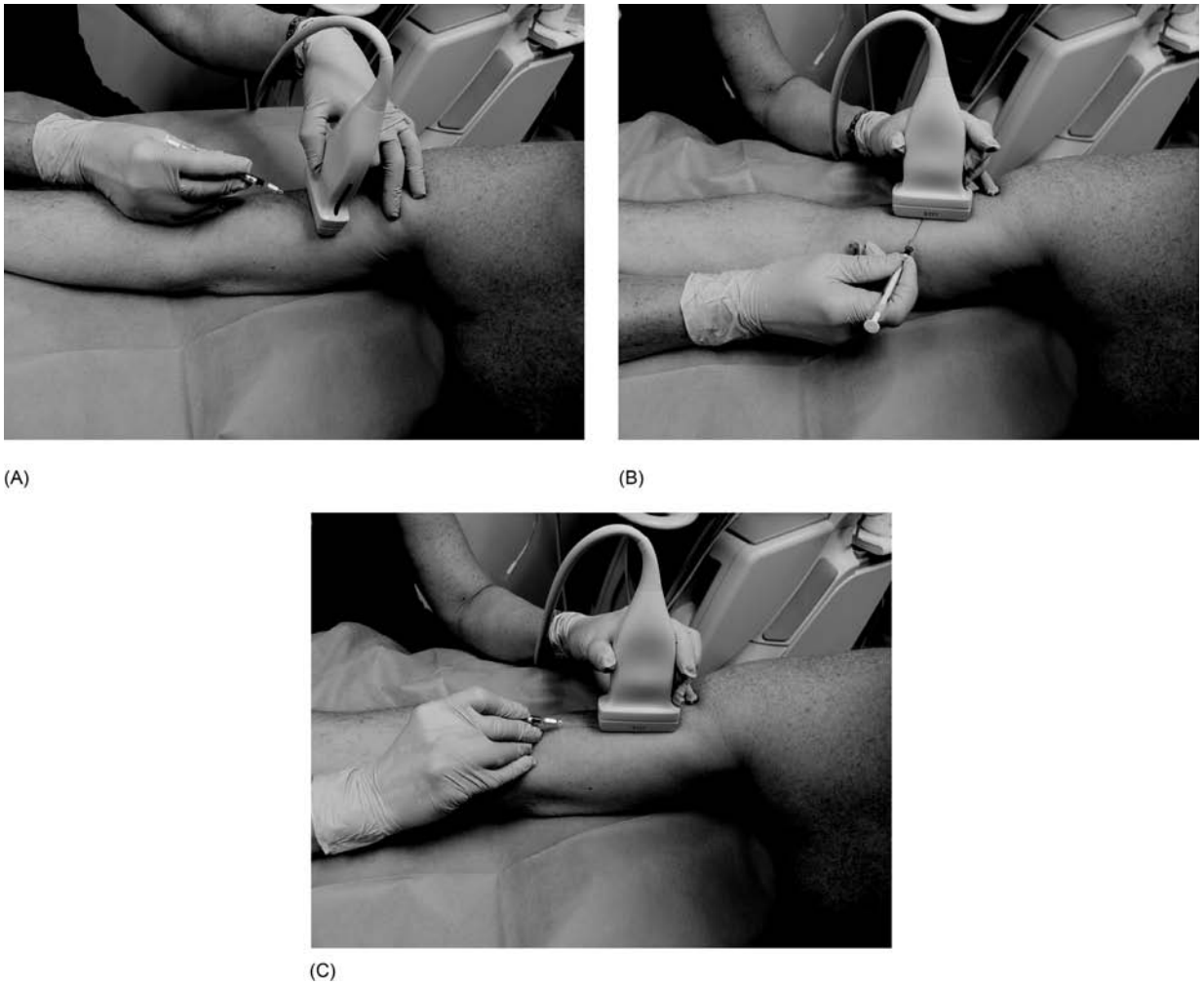


FIGURE 7.4 (A) Biceps transverse scan, out-of-plane needle insertion (2); (B) biceps longitudinal scan, out-of-plane needle insertion (1); (C) biceps longitudinal scan, in-plane needle insertion.

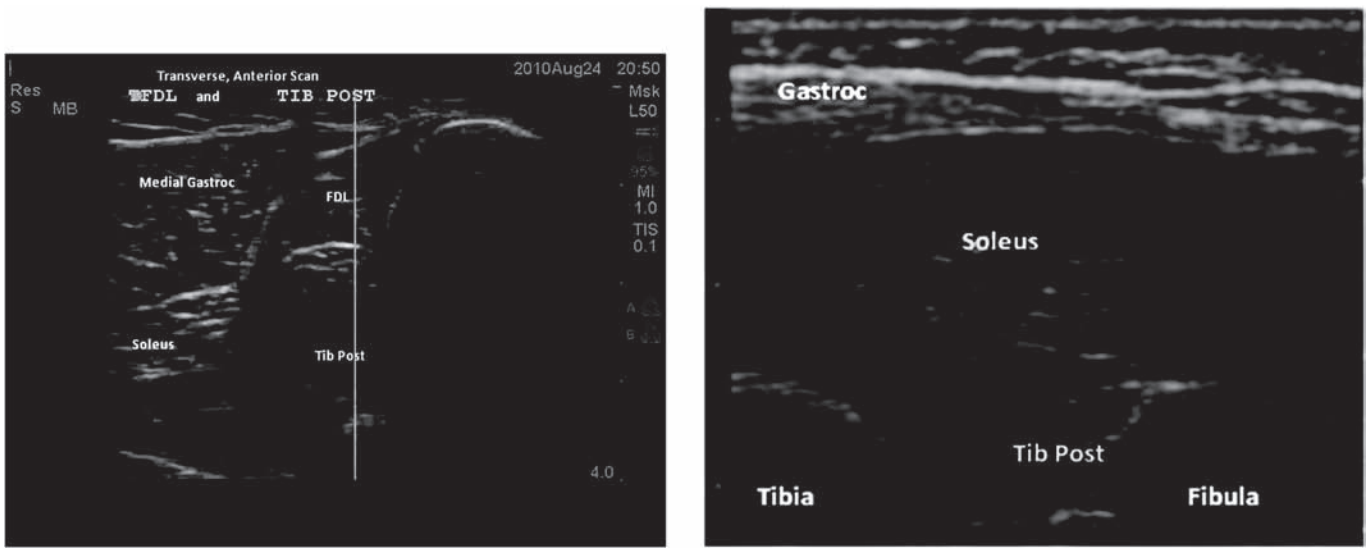
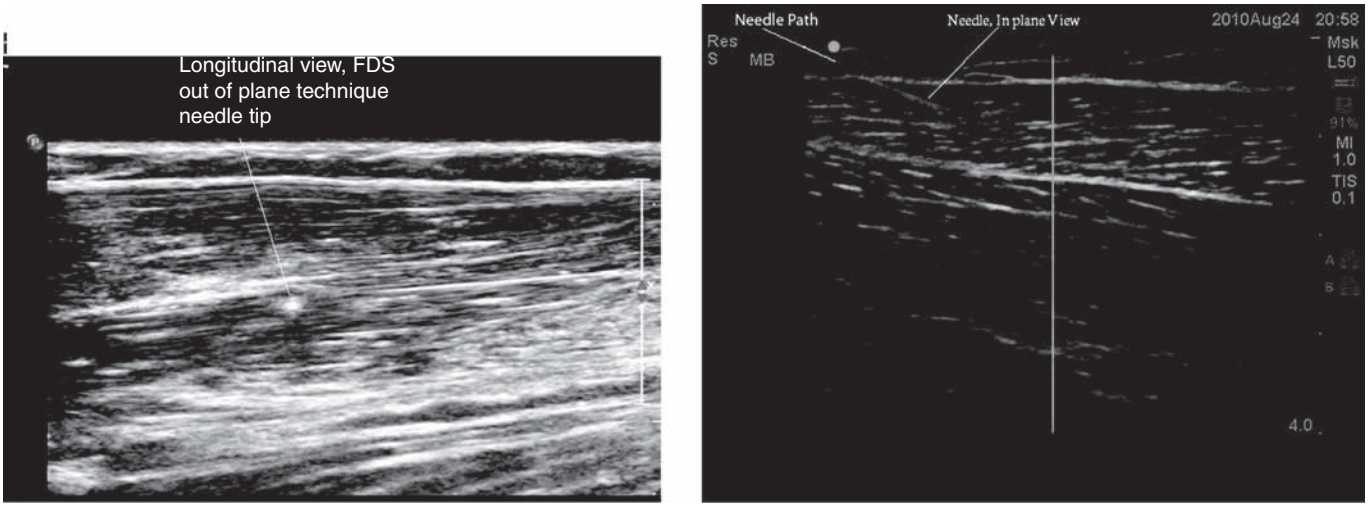


FIGURE 7.5 (A) B mode scan, antero-medial calf, 5 y.o.; (B) B mode scan, posterior calf, 12 y.o.



(A) (B)

FIGURE 7.6 (A) Out-of-plane view of needle in muscle; (B) in-plane view of needle in muscle.

needle and injection technique. Using an out-of-plane technique, the needle is inserted across the short axis of the transducer (Figures 7.4A,B). When using an in-plane technique, the needle is inserted down the long axis of the transducer (Figure 7.4C). Using the out-of-plane technique, the needle is visualized as a hyperechoic dot (Figure 7.6A). With the in-plane technique the entire needle is visualized, including the needle tip (Figure 7.6B). When using an out-of-plane technique, only the tip of the needle is visualized. Therefore the physician tracks the needle to its target using a walkdown technique (4,45). Using this technique, the physician observes the needle tip or the movement it creates within the muscle as the needle is moved from superficial to deep. Once the target muscle is reached, the toxin is injected. If desirable, the needle may be repositioned in the muscle to allow for multiple injection sites.

EQUIPMENT

US-guided BoNT injections require a US instrument or machine and one or more transducers (Figure 7.7A–C). A linear transducer with a lower range of frequencies of 4 to 5 mHz (for deep muscles in larger patients) and a higher frequency range of 12 to 17 mHz will allow visualization of most muscles in pediatric and adolescent patients. A lower frequency range curvilinear transducer may be needed for deeply seated muscles in larger teenagers or adult patients. A hockey stick transducer is useful when imaging small patients, irregular surfaces, and for salivary gland injections. Other US supplies include US gel and transducer cleansers. Sterile transducer covers and gel packets are available, if desired. Other procedural supplies include alcohol or another skin cleanser, hypodermic needles of various lengths, gloves, skin cleansers, gauze, and band aids/plasters. If the physician is also using EMG or E-Stim then monopolar insulated injection electrodes/surface electrodes, a cable to connect the reference electrode to the stimulator, and a stimulator or EMG machine are also required.

CLEANING AND MAINTENANCE

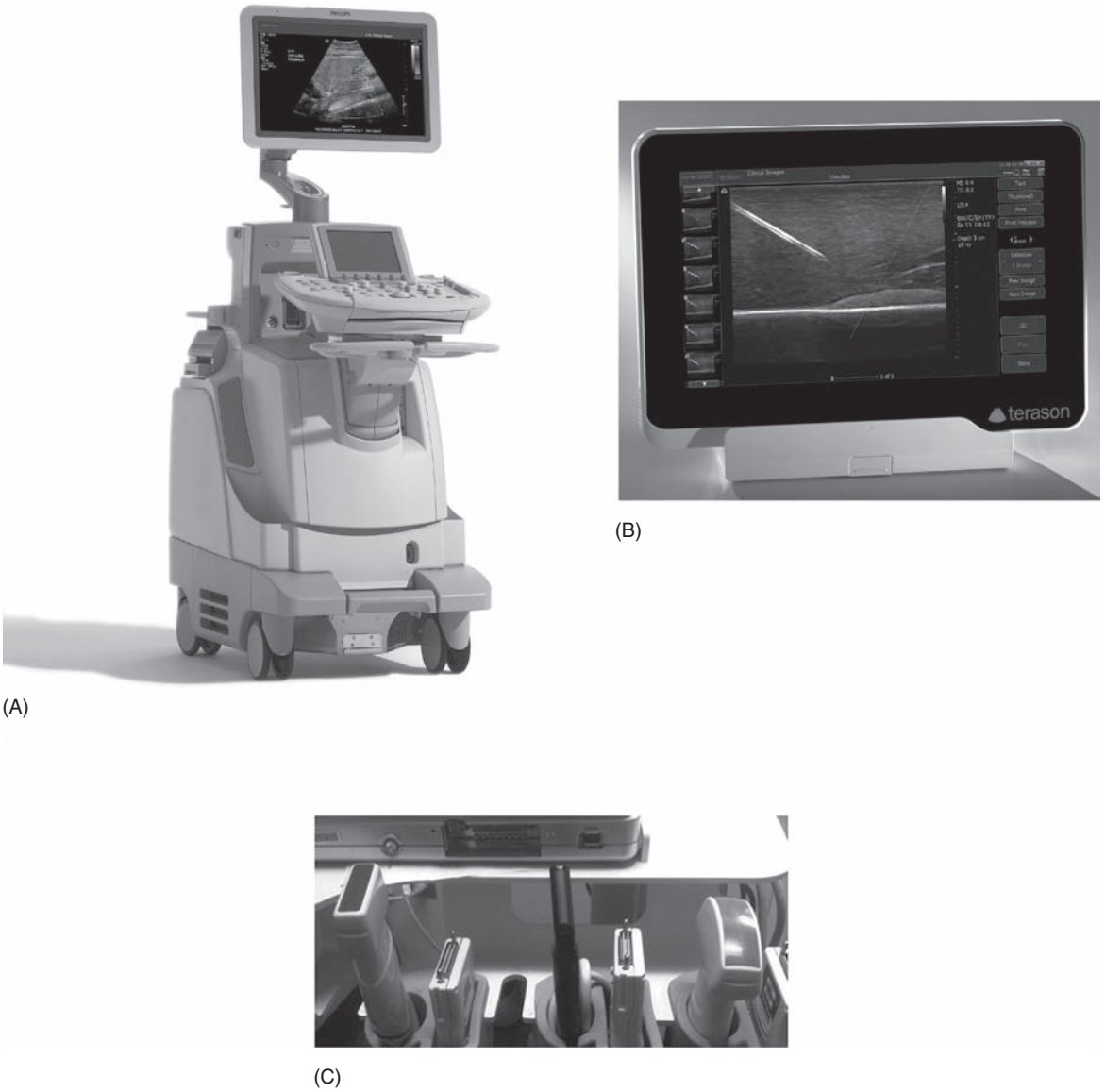
US machines and transducers should be cleaned as per the manufacturer's recommendations. Alcohol-based cleaners should not be used to clean transducer heads as this will damage the membrane (14).

ADVANTAGES AND LIMITATIONS OF US TO GUIDE BoNT INJECTIONS

As with the other localization techniques used to guide BoNT injections, there are advantages and disadvantages to using US guidance (2,4,45,48).

Advantages

- US provides a detailed view of the muscle location, depth, and structures in the region of interest.
- The needle can be tracked continuously during the procedure to a position within the target muscle.
- The volume of injectate can be visualized during the procedure preventing overdistention or injection at one site.
- US decreases the risk of inadvertent needle penetration and/or injection of untargeted muscles, vessels, nerves, or other structures.



(A)

(B)

(C)

FIGURE 7.7 (A) Phillips IU 22 ultrasound; (B) Terason 3200 uSmart ultrasound; (C) ultrasound transducers.

- US localization/visualization of muscles can be performed quickly and is painless.
- Standard hypodermic needles, which may be used with US-guided procedures, may be less painful to insert than insulated monopolar needles.
- Viewing the US image may provide distraction to the patient during the procedure.
- Many procedures can be done with the patient unsedated.

Disadvantages of US Guidance for BoNT Injections

- Cost of the equipment is high.
- Physicians unfamiliar with US have a steep learning curve.
- Access to hands-on training, particularly training specific for BoNT injections may be limited.
- Until the physician is familiar with and facile with this technique, using US for BoNT injections may increase the time required for the procedure.
- US may not provide information on the activity of a muscle, for example, whether it is contributing to the patient's problem. However, US can be combined with EMG to obtain this information, if desired.

EVIDENCE SUPPORTING OR REFUTING THE USE OF US TO GUIDE BoNT INJECTIONS

There is increasing evidence that US is more accurate at localizing targets for BoNT as well as reducing adverse events and outcomes with BoNT injections. See the section "Clinical Studies; Comparison of Anatomic Localization With Other Localization Techniques for BoNT in

TABLE 7.1 ADVANTAGES AND DISADVANTAGES OF VARIOUS GUIDANCE TECHNIQUES FOR CHEMODENERVATION PROCEDURES

METHOD	PALPATION	EMG	STIMULATION	ULTRASOUND
Accuracy	+/-	+/-	+	+++
Practicability	+	-	+/-	++
Availability	+/-	+/-	+/-	+
Pain	+	-	+/-	+++
Speed	+/-	-	+/-	++
Evaluation	+/-	-	+/-	+++
Future Research	-	-	-	+++

Limb Muscles," for evidence reviews for manual, EMG, and E-Stim guidance.

SUMMARY

US guidance provides detailed information on the location of the target structure, the position of the needle, and location of the injectate during interventional procedures including BoNT injections. The evidence to support the superiority of US over other guidance techniques is increasing. While there is a steep learning curve when learning to use this technology, it is a technique physicians should consider learning and adding to their tool boxes.

CONCLUSIONS

When selecting a guidance or localization technique for BoNT procedures, physicians should be aware of the advantages and disadvantages of each of the available techniques (Table 7.1). This knowledge allows the physician to select the most appropriate guidance method for a given patient and procedure. Optimally physicians should be trained in and have access to all of these techniques. Additional head-to-head comparative studies are needed to further define and determine which of the discussed techniques is superior, and if so, for which muscles and/or procedures (59–61).

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8

ORTHOTICS AND ASSISTIVE DEVICES

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Knowledge of orthotic and assistive devices is an important component of rehabilitation practice. Having an understanding of normal upper and lower body movement is fundamental for appropriate recommendation and fabrication of an orthosis. Likewise, clinicians' understanding of typical language and communication behaviors, literacy skills, and socialization needs is a prerequisite to the recommendation of augmentative and alternative communication (AAC) devices, assistive reading and writing aids, and social media tools.

Similarly, the role of orthotic and assistive devices for patients also relates to the overall rehabilitation goal of eliminating, minimizing, or helping them to overcome the limitations imposed by their underlying disorders across areas of physical, cognitive, and psychosocial functions. While some orthotic and assistive devices allow a patient to achieve a degree of independence in a single area of function (such as mobility), pediatric rehabilitation seeks to treat that patient as a whole person across his or her activities of daily living, learning, leisure and recreation, and/or vocational functions. Thus, the role of orthotic and assistive devices needs to be seen not just within the context of *independence* but also within the context of *interdependence* with others in a person's surroundings.

Interdependence recognizes that all people have strengths and weaknesses and that all people function most effectively when they do so within social networks that consist of life partners (parents, siblings, children and, often, other relatives); close personal and family friends; teachers and schoolmates; paid workers (such as caregivers, aides, nurses, therapists, physicians, and other health care providers); coworkers; acquaintances; and unfamiliar people. *Interdependence occurs when each of us uses our strengths to build relationships that help support our weaknesses*. Dr. Al Condeluci in Community & Social Capital asks that we "consider the notion of reciprocity (1). The more you become connected with your community, the more people begin to watch out for each other." Dr. Condeluci also applies literature from the field of social capital to the rehabilitation vision for people with disabilities: "Social capital refers to the connections and relationships that develop around community and the value these relationships hold for the members… those tangible substances that count for most in the daily lives of people: namely good will, fellowship, sympathy, and social intercourse among the individuals and [others] who make up a social unit." Thus, an adolescent who has a spinal cord injury (SCI) but can use her head control to independently drive a wheelchair at home, in school, and in the community still cannot participate and contribute to planning the eighth grade class yearbook if her family has not yet been able to buy an adapted van to take her to the Starbucks where her classmates meet to work on the project on Thursday evenings, unless she is perhaps able to participate from home through her head- or speech-controlled computer via Skype, FaceTime, or social media.

The key to identifying the most appropriate orthosis, augmentative communication, or assistive technology system is being creative and having a proper understanding of the anatomical, biomechanical, language and communication, and social networking needs of the patient and being sensitive to the patient's (or the parents') preferences and desires.

The pediatric population adds a further challenge. Early development is heavily based on fine and gross motor skills. Infants and children use these skills to explore and manipulate their environments. Studies have indicated that the inability to master the environment independently may lead to decreased socialization, learned helplessness, and a delay in normal development (2,3). Therefore, an orthosis, augmentative communication device, or assistive learning system should allow for and assist in the growth of the child.

Several team members are involved in prescribing, fabricating, and fitting the orthosis, augmentative communication device, or assistive technology system

option. The physician, often with input from the therapist, provides patient assessment and a prescription of the recommended device (4). The therapist and/or orthotist are instrumental in its fabrication and fitting. A team including a speech–language pathologist, an occupational therapist, a special educator, and/or a rehabilitation engineer is often beneficial for AAC device recommendations. Finally, the patient and family play an important role in its acceptance and use. If the device is cumbersome and difficult to manage, it will be rejected and find a home on the top shelf in the closet (5).

UPPER AND LOWER LIMB ORTHOSES

When choosing an orthosis, there are a few key principles to keep in mind. The orthosis should enhance normal movement while decreasing the presence of abnormal postures and tone. It should be simple, lightweight, durable, and strong. It should be easy for the child to use and maintain. Finally, it should augment functional independence. An orthotic device is not successful unless it assists in improving a child's quality of life. In 2004, a manufacturer new to the rehabilitation field designed a medical device for the hand. The Hand Rehabilitation System was meant for upper limb impairment and paralysis therapy. In addition, the Foot Drop System was presented for those with gait disturbance specific to foot drop impairment. In 2011, Bioness Incorporated developed a device for thigh weakness. The system compliments the foot drop system by aiding and assisting the thigh and knee movement for improved gait performance. Their devices are neuromodulation products that are designed to service populations with multiple sclerosis, traumatic brain injury (TBI), cerebral vascular accident, SCI, and cerebral palsy, and aid in their recovery. Their product can assist both the upper and lower extremity using stimulation to aid in regaining mobility and functional skills so that they can achieve optimal self-care independence, play, and/or work productivity. The upper limb orthosis addresses neurologic impairments, while the lower extremity orthosis focuses on regaining the associated foot drop commonly seen in those clients with central nervous system disorders. Both upper and lower extremity orthoses use mild functional electrical stimulation (FES) to improve loss of function from injury associated with a central nervous system disability. The orthoses can be used in the clinic setting or at home. The overall goal of its use with the involved extremity is to reduce the spasticity, minimize the pain and discomfort during use, increase local blood circulation, prevent muscle atrophy, improve or maintain range of motion in the limb, and reeducate muscle use to enhance functional movement (see the following website for additional information: www.bioness.com/Bioness_for_Hand_Rehab.php).

Tables 8.1, 8.2, and 8.3 list some of the more common upper and lower extremity orthoses. Special considerations and limitations are also listed.

SHOE INSERTS

Many orthopedic and neurologic pediatric disorders have sequelae that require orthotic management. Shoe inserts may be a viable option in many circumstances. There are many commercially available products to control differing levels of impairment in the hindfoot, midfoot, and forefoot. Heel cups help with shock absorption for joints, heel spurs, bursitis, and tendonitis. In the mid-foot, orthoses assist to maintain the arch of the foot in varying degrees of firmness. Numerous products are also available to control disorders of the forefoot and toes. To relieve metatarsalgia, metatarsal bars are available to unload pressure from the metatarsal arch. Pads are available to help to realign hammer and claw toes, cushion bunions and calluses, and to protect toes from friction and irritation. A limitation to these commercially available products is that many times they do not come in pediatric sizes and must be modified to fit.

ORTHOSES FOR POSITIONING, RANGE OF MOTION, AND HEALING

Due to immobility, spasticity, and/or abnormal postures, many children are at risk for joint contractures, musculoskeletal deformity, and skin breakdown. Traditionally, caregivers have used pillows and towel rolls to maintain more appropriate postures. Bony areas such as the occiput, scapular spine, coccyx, femoral head, fibular head, and calcaneus are at greatest risk for skin breakdown from prolonged bed rest or maintenance of one position. Gel pads may be used to distribute weight over a larger area. The child may benefit from positioning pieces to maintain neutral positions and decrease pressure on parts of the body. Foam wedges in various lengths and sizes are commonly used for back support to position a child in side-lying. An abduction pillow may be used to decrease scissoring and increase hip abduction. Foam arm and leg elevators help to reduce edema, and foot splints/boots are available to maintain the foot in a dorsiflexed position with relief for the calcaneus to prevent pressure sores.

The Versa Form pillow is a semipermanent positioning support. These styrene bead bags are available in a variety of sizes and allow for molding to a child in any position. A vacuum pump is required to remove air from the pillow to make it firm. The bead bags need to be reformed after several weeks of use. This technology gives the practitioner flexibility to change a child's positioning frequently. It is also a "system" that can be used in multiple environments.

TABLE 8.1 UPPER EXTREMITY ORTHOSIS

UPPER EXTREMITY ORTHOSIS	COMMON NAME	FUNCTION (REF.)	SPECIAL CONSIDERATIONS (REF.)
STATIC			
Finger/thumb	Neoprene thumb abductor	Places thumb in abduction to promote functional use of the hand.	Will not overcome severe cortical thumb position.
Hand	Short opponens	Places thumb in a functional position between palmar and radial abduction. Wrist and fingers are freely mobile.	Allows for full wrist flexion and extension. Should be worn at all times, removing only for hygiene and exercise.
Wrist–hand	Thumb spica	Immobilizes and protects the thumb, positioning it in opposition. In this type of splint, unlike the short opponens, the wrist is immobilized. Provides a stable post against which the index finger can pinch.	Need to allow for full MCP flexion of the fingers, especially the index finger, and full IP flexion of the thumb.
	Resting hand	Preserves a balance between extrinsic and intrinsic musculature and provides joint support when the hand is put at rest. Prevents deformity.	Should preserve the MCP joint descent and longitudinal arch following the contour of the distal palmar crease. Pressure at the MCP joint or proximal phalanx should be avoided, as this could cause injury to the MCP joint.
	Wrist cock-up	Supports, immobilizes, or stabilizes the wrist in extension. Increases mechanical advantage for grasp.	Must maintain full MCP flexion and CMC motion of the thumb. Monitor the area over the styloid process for pressure changes if a dorsal splint is used.
	Antispasticity ball	Positions the wrist, abducts the fingers and thumb, and maintains the palmar arch in a reflex-inhibiting position.	Should not be used for minimal spasticity (6).
Elbow	Elbow extension	Increases extensor range of motion and prevents flexion.	Not recommended for severe flexor contracture or fluctuating tone in either flexor or extensor patterns.
Elbow–wrist–hand	Full elbow/hand	Promotes supination at the forearm and provides a long stretch of the limb near end range to decrease tone.	Not recommended for flexor tightness.
Shoulder	Humeral orthosis	Used for humeral fractures. Stabilizes the shaft of the humerus circumferentially.	May shift position if not appropriately anchored by straps.
	Gunslinger	Used for rotator cuff repairs; positions the shoulder girdle in to slight abduction and prevents excessive tension on the repair.	Make sure the edges around the base of the splint do not cut into the hip area. Check the fitting both in standing and supine positions to accommodate the shift of the splint.
Clavicle	Harness strap	Used for displaced clavicular fractures; proximally stabilizes shoulder girdle movement and limits shoulder flexion and abduction movement beyond 90 degrees.	Must mark settings for appropriate fit due to increased adjustability. Keep a check on skin integrity around the underarm area.

(*continued*)

TABLE 8.1 UPPER EXTREMITY ORTHOSIS (*CONTINUED*)

UPPER EXTREMITY ORTHOSIS	COMMON NAME	FUNCTION (REF.)	SPECIAL CONSIDERATIONS (REF.)
DYNAMIC			
Hand	MCP flexion assist splint	Gradually lengthens or gently stretches soft tissue structures that limit joint flexion; generally not used for fixed contractures.	Ensure that the traction applied is gentle to guard against soft tissue hemorrhages; microtrauma around the joints, which can cause edema, pain, and increased scarring.
	MCP extension assist splint	Passively pulls the proximal phalanx into extension while allowing active flexion; generally used for radial nerve palsy or the hemiplegic hand.	Do not position the proximal phalanx in either radial or ulnar deviation when using dynamic traction. Avoid hyperextension of the MP joint, which can cause microtrauma and hyperlaxity to the MP volar plates.
	Finger spring—PIP extension assist	Gives dynamic traction of the PIP joint without limiting motion at the MCP joint. Assists in reducing tightness or contractures of the PIP joint.	Not recommended for severe spasticity.
Elbow	Dynasplint	Brace adjusts to lock out undesired flexion and extension. Settings are adjusted in increments of 10 degrees.	Not recommended for severe spasticity or fixed contractures.
Power	Smart-WHO	Flexor-hinge hand orthosis that immobilizes the thumb in opposition and semiflexes the IP joints of the index and middle fingers to allow the index and middle fingers to move simultaneously toward the thumb. Variations include using an external power battery pack, SMA actuators, ratchet hand position, and shoulder-driven cables (7).	Although design is lightweight and simple, a disadvantage can be the actuator's bulkiness as well as the unsightliness of the orthosis.

Abbreviations: CMC, carpometacarpal; IP, interphalangeal; MCP (or MP), metacarpophalangeal; PIP, proximal interphalangeal; SMA, spinal muscular atrophy; WHO, wrist–hand orthosis.

TABLE 8.2 LOWER LIMB ORTHOSES

ORTHOSIS	COMMON NAME	FUNCTION (REF.)	LIMITATIONS
Solid ankle foot orthosis	AFO, MAFO	Reduces tone, prevents joint contracture, and provides knee and ankle stability. Most appropriate for a child with severe tone, ankle joint hypermobility, and rigid deformities.	Does not allow any ankle movement and therefore limits smooth progression from heel strike to push off.
Hinged or articulated ankle foot orthosis	HAFO	A hinged AFO with a plantarflexion stop and free motion into dorsiflexion allows the tibia to translate over the foot in stance. This orthosis allows the foot to dorsiflex for balance reactions and improves ambulation on uneven surfaces and stairs. Posteriorly, a dorsiflexion stop strap can be added to limit the amount of dorsiflexion. A plantarflexion stop in 2–5 degrees of dorsiflexion may assist to control genu recurvatum at the knee.	Does not control "crouched" posture allowing increased dorsiflexion and knee flexion. Children with strong extensor posturing may break the ankle joint. May allow hindfoot to slip, causing mid-foot break if insufficient hindfoot dorsiflexion is present.

(*continued*)

TABLE 8.2 LOWER LIMB ORTHOSES (*CONTINUED*)

ORTHOSIS	COMMON NAME	FUNCTION (REF.)	LIMITATIONS
Anterior floor-reaction or ground reaction ankle foot orthosis	GRAFO	Limits a "crouch" posture (stance posture with hip flexion, knee flexion, and ankle dorsiflexion). At heel strike, it encourages a force up through the anterior cuff of this orthosis, giving the knee an extension torque. Knee extension is maintained throughout stance.	A child with significant hamstring or hindfoot tightness or tone will not benefit from this orthosis.
Rear-entry hinged floor-reaction AFO		Dorsiflexion stop limits a "crouch" posture while allowing for plantarflexion during the loading phase of stance and at pushoff.	Active dorsiflexion is required to restrict foot drag during swing.
Posterior leaf spring	PLS	The trimlines of this solid AFO are posterior to the malleoli. The slender posterior portion of this AFO gives it flexibility to allow for some dorsiflexion in stance and plantarflexion at pushoff.	Does not allow full motion into dorsiflexion or plantarflexion. For medial–lateral ankle stability and arch control, another orthosis may be more appropriate. Does not control foot deformity or extensor tone. Excessive torque on spring may cause skin problems.
Dynamic ankle foot orthosis	DAFO	A supramalleolar orthosis that uses a footboard to support the arches of the foot. Provides medial–lateral ankle stability with control for pronation/supination. Allows some ankle dorsiflexion/plantarflexion.	Difficult to fit into shoes. Difficult for self-donning. Child may quickly outgrow this splint, since it is finely contoured to the foot.
Knee hyperextension splint		Maintains neutral knee and limits knee hyperextension. Uses three points of pressure: superior–anterior surface of the knee, inferior–anterior surface of the knee, and posterior to the knee joint (8,9).	Controls only the knee. Does not control extensor posturing well. It is bulky under clothes and difficult to sit with.
Swedish knee cage	KO	Controls genu recurvatum with the same three points of pressure as a knee hyperextension splint and works the same. Uses metal uprights and straps instead of plastic material (10).	Controls only the knee. It is difficult to fit to smaller children, and it is difficult to maintain correct positioning.
Knee ankle foot orthosis	KAFO	Molded plastic upper and lower leg components, usually with a locked or unlocked hinged knee joint. Four most common knee locks are free, drop lock, bail lock, and dial lock. Free knee allows full motion at the knee axis. Knee axis may be straight or offset. Offset axis has an increased extensor moment at the knee joint. The drop lock is a metal collar that slides into place to maintain the knee in extension. The bail lock is a spring-loaded lock that has a trip mechanism to unlock the knee. The dial lock is a lock that may be set in varying degrees of flexion, used to accommodate or decrease a knee flexion contracture (11).	It is bulky and difficult to don/doff. Free knee at times allows too much motion. Drop lock requires fine motor control to lock and unlock. The child must be able to get the knee fully extended to engage the drop lock. Bail locks at times become easily disengaged. Dial locks do not allow free movement through the available range.
Hip knee ankle foot orthosis	HKAFO	Hip belt and joint. Hip and knee joints may be locked or unlocked. Able to progress child to an increasing number of free joints at a time.	Bulky, difficult to don/doff. Difficult to manage clothing for toileting.

(*continued*)

TABLE 8.2 LOWER LIMB ORTHOSES (*CONTINUED*)

ORTHOSIS	COMMON NAME	FUNCTION (REF.)	LIMITATIONS
Reciprocating gait orthosis	RGO	HKAFOs that are connected by a cable system that links hip flexion on one side with hip extension on the other. This device assists children with active hip flexion and no hip extension to advance legs with a more normalized gait. Allows the child to ambulate with a reciprocal or swing-through gait (11).	Bulky, expensive. Difficult to don/doff. Not appropriate for a child with hip and/or knee flexion contractures. Difficult to manage clothing for toileting.
Hip spica/hip abduction splint		An orthosis made of thermoplastic material and Velcro to position a newborn's legs in abduction and flexion. This splint is used to maintain the femoral head in the acetabulum to mimic normal hip formation. Use of this splint helps to avoid hip subluxation and dislocation. Used from birth up to a year.	Requires frequent repositioning. May need frequent adjustments for growth. Difficult for caregivers to maintain appropriate fit.
Pavlik harness		A soft splint used for children with the diagnosis of congenital hip dislocation. This splint is generally used in the first 9 months of age. Bilateral lower limbs are positioned with hips abducted and flexed to 90 degrees in an attempt to maintain the hips in a reduced position.	Careful positioning required. Caregivers must be vigilant in checking splint positioning.
Parapodium or Variety Village Stander		This device allows the child to stand without upper extremity support, freeing bilateral arms to do activities. Walking with this device and crutches can be quicker than with the Parapodium with a swivel device (11).	More energy expenditure than with the swivel device. Children are unable to independently don/doff or to independently transfer supine to stand and stand to supine. Device is heavy.
Parapodium with ORLAU swivel modification		This orthosis allows the child to walk without use of a gait aid and to use the arms for other activities. Less energy expenditure than with a Parapodium and crutches (6,12).	Slower than walking with Parapodium and crutches.
Twister cables		Cables are attached to a pelvic band and traverse the lower limbs to attach on shoes or AFOs. These cables provide control for increased internal rotation. Work well with children with normal to floppy tone to control internal rotation (11).	Do not work well with children with extensor spasticity. They may need to be frequently readjusted as the child grows.

Abbreviation: ORLAU, orthotic research and locomotor assessment unit.

MOBILITY AIDS

TRANSFER AIDS

There are a number of commercially available patient care lifts to assist caregivers and/or health care professionals with performing safe transfers for children. The Trans-Aid and Hoyer lift are two examples of patient care lifts. They are designed to transfer children from bed to wheelchair, off the floor, onto a toilet, into a car, or from any one room to another. Slings are available with heavy-duty support options to further minimize the effort of the caregiver while maximizing safety during the transfer. There are also institutional lifters available, which offer a 400-pound and 600-pound weight capacity, as well as portable home-care lifts, which are lightweight, portable, and designed for home doorways and narrow halls.

Powered overhead transfer lift systems provide families with a unique transfer method. This system enables users to transfer from bed to wheelchair, toilet, or bath using a motorized lift and either manual or motorized lateral movement along a permanent ceiling-mounted track or a free-standing semipermanent rack. However, this transfer system is expensive and usually not covered by insurance.

TABLE 8.3 TRUNK ORTHOSES

TRUNK ORTHOSIS	COMMON NAME	FUNCTION	SPECIAL CONSIDERATIONS
STATIC			
Trunk	Thoracic–lumbosacral orthoses	This device is used to stabilize the spine after surgery, fractured vertebrae, or used therapeutically to provide the trunk with upright support during static or dynamic activities.	Can affect respiratory function. Brace will not correct spinal deformity, but may alter the progression of the curve.
	Theratogs/Benik	This orthotic undergarment and strapping system gives users with sensorimotor impairment tactile positioning cues for improving postural alignment, postural and joint stability and movement, skill and precision.	Can cause pressure along axillary line, breast bone, or ASIS area. Monitor wear time secondary to heat intolerance.

Abbreviation: ASIS, anterior superior iliac spine.

In addition to patient lifts, there are other smaller devices that can assist children with ease of transfers. One option is a transfer board, while another is an overhead trapeze bar attached to an over-bed frame. The most commonly used transfer board is constructed of maple wood measuring approximately 8 inches wide by 24 inches long. It is ideal for all types of transfers (bed, car, bath bench, commode, etc.). Trapeze bars may be attached overhead to bed frames to assist the child with bed mobility skills and positioning changes. The position is individually set and can be altered as needed. Typically, trapeze bars assist with supine to sitting transfers and initiating rolling side to side. They are often appropriate for use initially, but are soon removed after the child's strength and bed mobility skills improve.

STANDERS

Numerous passive standing devices are available. These devices offer many potential benefits for the child, including the provision of a sustained muscular stretch, maintenance of trunk and lower limb passive range of motion, facilitation of cocontraction of muscles, decreasing tone, and improvement in trunk and head control. Standers should be used a couple of times a day for up to a total of 1 hour. The child should progressively work to increase tolerance in the standing position. However, passive standing should not take the place of the child exploring his or her environment and body.

Three types of standers will be discussed here: supine, prone, and upright. Supine standers go from a horizontal position to approximately 90 degrees upright, depending on the model chosen. Laterals, kneepads, adduction/abduction supports, and head supports all assist to maintain the child's posture while in this stander. Bilateral upper extremity strengthening can be performed in this position, with or without a tray. However, it does not provide for any upper extremity weight-bearing. A further limitation is that it will not work to improve head and trunk control. This stander is recommended for a child with significant extensor tone and posturing and/or a child with poor or absent head control. It is also preferred over the prone stander for the larger child due to the increased ease in positioning.

Prone standers support the child anteriorly. Postural support is supplied through trunk laterals, hip guides, abductor blocks, knee blocks, and shoe holders. These standing devices do come with a chin support to aid children who have limited head control or fatigue easily. However, the child should not be permitted to "hang" on this support; a supine stander is more appropriate if the child lacks fair head control. The stander can be used to improve antigravity head control and promote bilateral upper extremity weight-bearing. Its tray may serve as a functional surface for stimulation. This stander may not be appropriate for some children with increased extensor tone. In these cases, gravity increases the work required for neck and trunk extension as well as shoulder retraction, thus feeding into primitive posturing.

Upright standers, such as the Evolv by EasyStand (Figure 8.1) maintain the child in an erect position through supports at the hips, knees, and trunk. Certain standers are available with a hydraulic or manual lift, making positioning of the larger child easier. This stander mimics a normal standing position and permits the child to work on head control and upper extremity strengthening. The seat swings to the side for ease of transfer.

FIGURE 8.1 EasyStand Evolv.

GAIT AIDS

Gait aids are assistive devices designed to improve functional independence and/or expand exercise options through standing and walking. In pediatrics, gait aids assist children to explore and interact with their environment. Improved balance, decreased energy expenditure, decreased impact on joints, improved posture, and decreased pain are all potential benefits of gait aid use. The most common gait aids are canes, crutches, and walkers.

Canes are available in different sizes with a variety of handles and supports (ie, straight cane versus quad cane). A quad cane provides a better base for support, but a normal gait cycle is more easily mimicked using a straight cane. A hemicane is a combination of a cane and a walker. It has a four-point base and the largest base of support of all the canes. It gives the greatest amount of stability among the canes, but also encourages the child to lean laterally when ambulating.

Crutches generally fall into two categories: axillary and Lofstrand. Axillary crutches are usually constructed of wood or aluminum and have limited adaptability. Some crutches may be modified to offer forearm support to decrease weight-bearing through wrists and hands. The child and family should be cautioned about possible nerve impingement from sustained axillary pressure with improper use. A "Kenny Stick" is an axillary crutch without the underarm piece. In place of the underarm support is a leather armband that fits around a child's forearm.

Lofstrand crutches are much more flexible. They have a variety of forearm cuff styles, including circumferential or half cuff. Functional independence is increased with the use of Lofstrand crutches because the child is able to reach with his or her hands and the circumferential cuff will stay on the forearm. Half cuffs require less reliance on the cuff for balance, but they will not stay on the forearm if the handgrip is released. Handles may be wide and flat, pistol, or rounded. Rounded handles are the most commonly prescribed. The flat, wide handles may be helpful with tonal issues as well as with carpal tunnel inflammation. Pistol grips provide grooves for finger placement. Newer varieties of Lofstrand crutches are lightweight for children who have limited strength or need shock absorption for their joints.

Another adjustable option for all crutches is the crutch tip. Crutch tips may be constructed with materials of various flexibilities and in different widths to make the crutches more stable. Tips may include a gel, providing some shock-absorbent qualities. In addition, studded cups, which cover crutch tips, are available to make ambulation in rain and snow easier.

Three varieties of walkers are appropriate for the pediatric population: forward, reverse, and gait trainers. Forward walkers are the traditional type of walker. They can be purchased with or without wheels. Children can grip flat handles or use platforms on one or both sides to weight-bear through the elbows and forearms. It should be remembered that forward walkers promote trunk flexion in many children.

Reverse walkers, also called posture control walkers, promote an erect posture. The child has increased extension at the trunk and hips when his or her hands are positioned to the sides or slightly in front. A pelvic support can be added to assist with lateral pelvic control and to facilitate trunk extension. Platforms can also be attached to allow forearm weight-bearing. These walkers are widely used in the pediatric population. However, due to increased width, adult-sized children may have difficulty with accessibility. Other accessories available with some walkers are swivel wheels, forearm attachments, hip guides, handwheel locks, baskets, and seats.

Gait trainers make ambulation a viable option for children who are unable to ambulate with other aids. Intensive body-weight-supported treadmill training may be an effective intervention for some children with cerebral palsy who are ambulatory (12). A gait trainer is an assistive device that provides significant trunk and pelvic support (Figure 8.2). It consists of a metal frame with adjustable-height metal uprights that support the trunk and arms. Adjustable-height seats, which are either slings or a bicycle-type seat, are attached. The seat is not used to support the entire body weight, but rather to keep the

FIGURE 8.2 Rifton gait trainer.

child erect. This gait device has been used to teach a more normal reciprocal gait pattern. It may function as a stepping stone to walking with a walker or crutches. Some limitations of gait trainers include decreased transportability, difficulty with positioning, and decreased accessibility. They are wider and longer than traditional walkers are. Gait trainers do have a place in therapeutic rehabilitation—to provide a child with independent means of ambulation when no other assistive device is appropriate and as a therapeutic tool toward ambulation with a more accessible assistive device. Accessories available with gait trainers are trays, wheel locks, harnesses, forearm supports, and differing lower extremity supports.

For facility use, weight-bearing and ambulation aids are available. The Lite Gait is a partial weight-bearing gait therapy device. It allows the therapist to control the amount of weight-bearing by supporting the patient in a harness system over a floor treadmill. With other therapeutic modalities, this has been shown to improve ambulation and endurance levels (13).

The EVA Walker is a heavy-duty walker that has a manual or hydraulic lift that is easily adjustable for a variety of patients. It allows for significant upper extremity weight-bearing to assist and improve ambulation for more moderately dependent patients.

WHEELCHAIRS AND SEATING

The degree of limitation in mobility varies across a broad range for people with physical disabilities (5). Over the years, technology related to wheelchair seating and mobility has enhanced the opportunities for people with disabilities. Many more options exist to match technology with the user than ever before.

In order to begin the process of matching the child's needs to a particular wheelchair, it is recommended that a thorough evaluation be made. Many factors contribute to deciding on a particular seating and mobility system for the pediatric population. These include growth, specific disability, medical interventions, and prognosis of future functional and cognitive abilities. Assess the particular needs of the child, collect medical and surgical history, and perform a physical assessment. A multidisciplinary team approach usually works best. Once the assessment is performed, educate the family on various wheelchairs relative to the child's goals. If possible, simulate the child in as close to the recommended equipment as possible. Finally, determine the particular seating objectives for the child as well as the type of mobility base (4).

Every child has a unique set of challenges that will dictate how his or her rehabilitation needs will be met. Proper seating provides stability and support, decreases the likelihood of postural deformities, and enhances upper extremity control. Proximal stability allows for distal control. Within a wheelchair seating system, maintaining proper body alignment is achieved by using various seating and positioning components (14). Seating systems, including both the seat and the back, can be linear, contoured, or molded. Of the three, linear seating systems provide adjustability that allows the seating system to grow as the child grows. Linear seating systems are the most adaptable as the person's orthopedic needs change. The basic materials consist of plywood for the base, foam (usually viscoelastic, which can vary in density and be combined in layers for pressure relief and distribution) for comfort and pressure relief, and a covering, usually Lycra, Rubatex, or Dartex. Positioners such as laterals, abductors, and adductors are easy to mount on these systems.

Contour systems, in contrast to linear systems, conform closer to the actual shape of one's body. When recommending a contour system, close attention should be given to the growth rate and potential medical interventions, as the shape of the contour may not be an appropriate choice. Custom molded systems provide maximal support and should be considered for children with fixed deformities. Molded systems do not change as the child grows, unless remolding is performed, which is potentially time-consuming and costly. Although this system aids in controlling tone and nicely contours to most deformities, it has the reverse effect of limiting the amount of freedom children have in their seating system.

For patients who lack sensation, a variety of cushions exist that assist in alleviating pressure, which will decrease the likelihood of skin breakdown. Cushions fall under several categories, including foam, gel, air, and water (Table 8.4). Cushions should provide pressure relief under bony prominences, provide a stable support surface for the pelvis and the thighs, and function effectively in different climates. They should be lightweight, especially if a person is transferring independently or is a self-propeller, and be durable. Each type of cushion has advantages and disadvantages.

TABLE 8.4 CUSHION TYPES

FOAM	GEL	AIR
• Lightweight	• Lightweight	• Provides extremely good pressure relief
• Provides a stable base of support	• Provides a stable base of support	
• Various densities available that can improve pressure-relieving qualities	• Various densities available that can improve pressure-relieving qualities	• Lightweight
• Heavy	• Heavy	• Can be unstable
• Conforms to individual shape	• Conforms to individual shape	• Requires careful monitoring and maintenance

Pressure mapping systems are tools used by clinicians to measure interface pressures between two surfaces, such as a seated person and the cushion he or she is sitting on (see an example of a pressure mapping system by Vista Medical at www.pressuremapping.com). A visual output on a computer monitor allows easy viewing and understanding. Using this tool allows clinicians to "diagnose" potential causes of skin ulcers as well as to select a cushion that will provide the most appropriate pressure relief for that patient. Keep in mind that all risk factors, both extrinsic and intrinsic, should be considered before assuming a particular cushion or piece of equipment is causing skin breakdown.

POSITIONING COMPONENTS

Within a wheelchair seating system, maintaining proper body alignment is achieved by using various positioning components. Evidence supports that children with cerebral palsy should be fitted for wheelchairs that place them in a functional sitting position (15). Lateral supports can be used to encourage midline trunk position when trunk control is poor. Support for those with spinal deformities, such as scoliosis and kyphosis, is carefully evaluated to ensure that corrective forces applied to the individual are tolerable. Scoliosis is managed with the three-point pressure technique. Support pads are placed under the apex of the curve, high on the opposite side and bilaterally at the pelvis. Severe deformities should always be accommodated comfortably, using a seating technique that allows for contact with as much surface area as possible. Usually, when supporting a scoliosis, some degree of tilt (maintaining 90-degree hip angle) is necessary to alleviate some of the effects of gravity on the spine. Also, if the client is not forced to counteract gravity, the lateral supports do not need to be as aggressive and can be made more comfortable and tolerable (16). Chest harnesses assist in stabilizing the trunk by anterior support as well as by preventing forward trunk flexion.

Positioning belts are used for pelvic alignment and stabilization. An improperly placed pelvic positioner is more detrimental than no positioner at all. The standard angulation of a pelvic positioning belt is at a 45-degree angle to the sitting surface (3). There are many pelvic positioners available to maintain proper pelvic alignment, such as a double pull padded hip belt, a Hip Grip Pelvic Stabilizer by BodyPoint Inc., and Pelvic Harness by Rifton, to name a few. Subasis bars are used primarily for high-tone patients. Proper placement and position of the bar is critical to the success of the product. Improper positioning can potentially lead to skin breakdown.

Additional positioners include abductor pads that reduce or prevent increased adduction and assist in providing proper leg alignment. It should be remembered that abductors are not to be used to block a child from "sliding" out of the wheelchair. This may cause injury to the perineal area (2). Adductors decrease hip abduction and assist in providing proper leg alignment. Shoe holders and ankle positioners help control increased extension or spasms in the lower limbs and correct or prevent excessive internal or external foot rotation.

Head position is important for many reasons, including proper visual input, control of tone, and proper alignment for feeding and swallowing. Headrests provide support and positioning for a patient with poor head control due to low tone, active flexion, or hyperextension. They provide posterior and, if necessary, lateral support. They also furnish safety in transport. The size and shape of the headrest depend on individual needs. Total head support can be achieved with the same headrest that allows the child to freely move his or her head to explore his or her environment.

When proper seating and positioning components are in place, pediatric wheelchairs provide users with the opportunity to explore and experience the world around them. It encourages social integration as well as enhances the level of involvement in various school and home activities. The majority of wheelchairs can be divided into two main categories: dependent mobility and independent mobility. These categories represent the level of functional mobility the child can achieve. Strollers,

recliner wheelchairs, and tilt-in-space wheelchairs typically make up the types of chairs recommended for people who need a temporary means of mobility or who are incapable of independent mobility. Tilt-in-space chairs, such as the Quickie or Zippie IRIS (Figure 8.3) (see website for additional information: www.sunrisemedical.com), are recommended for people who need moderate to maximum positioning when there is little tolerance for an upright position. A reduction of pressure readings at the ischial tuberosities with tilt and recline positioning was shown as a general trend in a study by Pellow (7). Tilt-in-space wheelchairs provide pressure relief by redistributing body weight. The tilt also can assist the caregiver in properly positioning the child in the wheelchair by allowing gravity to assist. Positioning strollers, such as the Kid Kart Xpress, are typically used for younger children in whom independent mobility is less of an issue. Most strollers are also easily transportable.

Independent mobility can be achieved by using a manual wheelchair or a power wheelchair. Functional abilities and mobility goals dictate the type of wheelchair recommended. Manual wheelchairs can range from providing minimal support to complete postural support. Manual wheelchairs are lightweight in nature and have a multitude of features that can be adjusted or added to enhance efficient and effective use. Table 8.5 offers a comparative look at the various wheelchair components. Although this is a list of manual wheelchair components, many features can be considered for power wheelchairs as well.

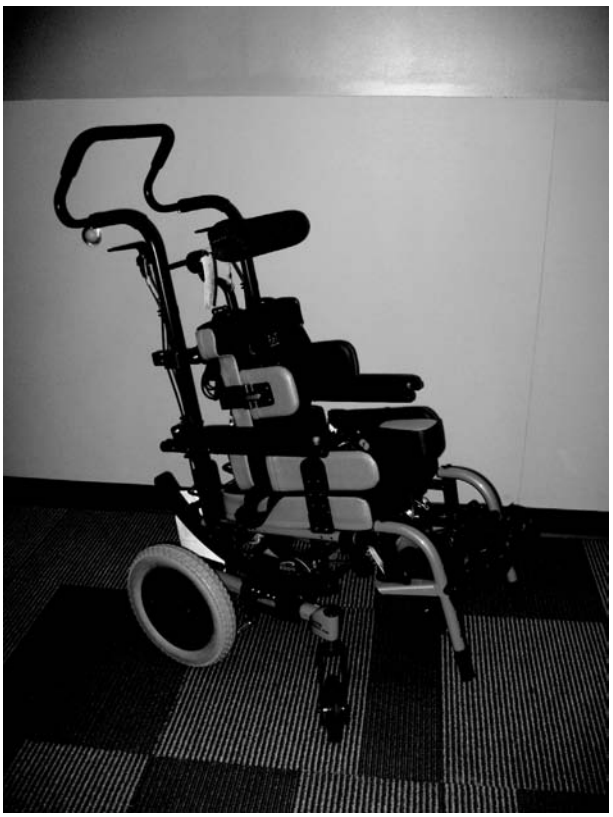

FIGURE 8.3 Zippie IRIS.

Power wheelchairs provide independent mobility when manual wheelchairs cannot be used. Independent mobility is believed to be essential for perceptual motor and social skill development. Self-produced locomotion also is believed to have an impact on cognition, communication, and psychosocial development (9). Technological advances in electronics have enabled people with severe physical disability to operate a motorized wheelchair. Power wheelchairs can incorporate unique features that enhance function critical to health maintenance, as well as social development. The children who received power mobility had significantly greater improvement in receptive language on the Beck Depression Inventory (BDI). Children with power mobility also score higher in social-function, functional skills and self care assistance on the Pediatric Evaluation of Disability Inventory (PEDI) than the children who did not use power mobility (17). Power wheelchairs have pediatric sizes that are capable of raising the child from a seated to a standing position (for an example, see the Permobil website at www.permobilus.com), as well as elevating in the seated position using a "seat elevator." Table 8.5 shows an overview of powered mobility systems.

Some power wheelchairs lower to floor level to allow the child to socially interact with peers. However, there may be constraints to using a power wheelchair. The family may not have the means to transport the wheelchair, or the power wheelchair cannot be used in the home due to limited physical space and accessibility. Funding may also prohibit the ability to acquire a power wheelchair. Another option for powered mobility for children may lie in three- or four-wheeled scooters. Scooters are usually less expensive than a power wheelchair, but do not offer a great deal of positioning options. Although choices are limited for pediatric-sized scooters, several do exist that can accommodate small children.

CAR SEATS

Conventional restraint devices may not always be the option for safety in transportation (18). Alternative car seats can be purchased for children with special needs. There are several commonly used types of special needs car seats: the Britax Traveller Plus (for more information, see www.snugseat.com) offers the most seat width in a special needs car seat. The Convaid Carrot Car Seat offers a modular system that can grow as the child grows. Both include seat depth extenders, adequate positioning pads, five-point safety straps, and an appropriate restraint system. The Carrie Car Seat comes complete with head support, harness and safety belt straps, and foot supports. At times, a child is sent home from the hospital in a spica cast or one that limits the fit in

TABLE 8.5 WHEELCHAIR CHARACTERISTICS

FRAMES

Rigid	Folding	Hemiheight	Tilt in Space	Recliner	One-Arm Drive
(+) Efficient ride (+) Durable (+) Lightweight (–) Decreased shock absorption	(+) Shock absorption (+) Ease of transport (+) Ability to narrow chair (–) Less efficient propulsion	(+) Allows LE propulsion (+) May make transfer easier (+) Optimal height for peer interaction (–) May make transfers difficult (–) Compromise height at tables	(+) Pressure relief (+) May assist to help balance (+) head control (+) Change position for respiration (–) Heavy (–) Difficult to break down	(+) Pressure relief (+) Seating for hip contractures (+) Limited tolerance for upright posture (+) Ease of breathing/feeding (–) Difficulty changing position with spasticity (–) Laterals and headrest move with changing position	(+) One functional UE (+) Sometimes difficult to manipulate (–) Additional steps for folding a chair

ARMRESTS

Conventional	Height Adjustable	Flip-Up	Swing-Away	Arm Troughs
(+) Offers protection (–) Heavy (–) Hand function (–) Cosmesis	(+) Positioning assist (+) Offers protection (+) Ease of transfers (–) Bulky	(+) Hand function varies (+) Remains attached for quick availability (–) May be in bad position	(+) Durable (+) Cosmesis (+) Easiest to operate (+) Can change width via cushion (–) No protection (–) Must order side guards for protection	(+) Alignment of UEs with minimal AROM (–) Bulky (–) Difficult to support tray

FOOTRESTS

Hanger Angle			Types		
60 degrees	*70 degrees*	*90 degrees*	*Tapered*	*Standard*	*Elevating*
(+) Able to have large casters (+) Limited ROM (+) Increase depth without length (+) Taller person (–) Increased length of chair	(+) Reduces spasticity problems (+) Compromise	(+) Reduces turning radius (+) Reduces chair length	(+) Increased accessibility (+) Positioning (–) Decreased calf space	(+) Adequate calf space (–) Decreased accessibility	(+) Positioning—contractures (+) Edema (–) Increased chair weight (–) Increased length (–) Decreased accessibility (–) Elevating mechanism (–) Cumbersome

(*continued*)

TABLE 8.5 WHEELCHAIR CHARACTERISTICS (*CONTINUED*)

FOOTPLATES	FRONT RIGGING				
Solid/Platform	**Angle Adjustable**	**Hemimount**	**Flip-Up**	**Fixed**	**Swing-Away**
(+) Folding frame more stable (+) Durable (–) Must remove to fold on folding frame	(+) Best positioning-ankle contractures (+) Reduce extensor thrust in lower limbs (–) Heavier	(+) Positioning for shorter legs	(+) Easier to move out of way (–) Not as durable	(+) More durable (+) Change seat depth without length (–) Transfers more difficult (–) Cannot reduce chair length	(+) Facilitate transfers (+) Greater accessibility (–) Must manipulate release mechanism
LEG STRAPS					
Toe loop, heel loop, calf strap	**Shoe holders**				
(+) Maintain feet on footplates (+) Straps maintain position even with flexor spasticity (+) Straps may be used for WC/floor/WC transfer (–) May make transfer difficult	(+) Control increased extension or spasms in lower limbs (+) Excessive internal, external rotation (+) Prevent aggressive behavior for safety (–) Heavy (–) Cumbersome				
CASTERS					
Solid	**Pneumatic**	**Semipneumatic**	**Size 6–8 Inches**	**Size 3–5 Inches**	
(+) No maintenance (+) Least rolling resistance (+) Energy-efficient	(+) Most shock absorbent (+) Easier to maneuver over small objects (–) Requires maintenance	(+) No maintenance (+) A good compromise between solid and pneumatic	(+) Less rolling resistance (+) Increase footplate/ground clearance (+) Good on rough terrain (+) Tilt (+) Rugged terrain (+) Smoother ride (–) Larger caster—hard to turn on carpet	(+) Less shimmy (+) More responsive to quick turns (+) May aid in curb maneuverability (+) Increase footplate/caster clearance (+) Indoor use—tighter turns (–) Poor maneuverability over rough or uneven terrain	

TABLE 8.5 WHEELCHAIR CHARACTERISTICS (*CONTINUED*)

AXLES

Axle position		**Axles**			
Single Position	*Multiposition*	*Amputee*	*Standard*	*Quick Release*	*Quad Release*
(+) Durable (−) No adjustability	(+) Adjustability (−) Decreased durability	(+) Fits special population (−) Decreased durability	(+) Threaded (−) Cannot remove rear wheels	(+) Can remove rear wheels (+) Reduce size weight for transportability (−) Need good hand function (−) Durability	(+) Can remove rear wheels (+) Lower hand function (−) Durability (−) May accidentally disengage

REAR WHEELS

Spoked	**Mag**
(+) Shock absorption (+) Lighter (−) Maintenance	(+) No maintenance (+) Decreased chance of finger injury (−) Heavier

TIRES

Urethane	**Pneumatic**	**Kevlar**	**Knobby**	**High Pressure**	**Airless Inserts**
(+) Good Indoors (+) No maintenance (+) Durable (−) Rougher ride (−) Heavier	(+) Rough terrain (+) Good traction (+) Lighter (−) Maintenance	(+) Reinforced tire	(+) All-terrain (+) Increased traction (+) Added flotation (−) Squeaks when new	(+) High pressure (+) Lighter (−) Need Presta valve	(+) Flat-free (+) Compromise (+) Low maintenance (−) 1 pound heavier than pneumatic tires

PUSH RIMS

Aluminum	**Friction-Coated**	**Projection "Quad Knobs"**
(+) No friction (+) Fine control (−) Cold in cold weather (−) Slippery if wet	(+) Impaired hand function (−) Chair width increased (−) Slippery if wet (−) Can cause burns (−) Coating wears away	(+) Angle varies (+) Length varies (+) Number varies (−) Angle increases width (−) Decreased efficiency if pegs do not end up in right position (−) Difficult to descend

(*continued*)

TABLE 8.5 WHEELCHAIR CHARACTERISTICS (*CONTINUED*)

WHEEL LOCKS				
Push to Lock	**Pull to Lock**	**Scissors**	**Extensions**	**Grade Aids**
Attendant Wheel Lock	(+) Ease of access for caregiver	(-) Inaccessible for client	(+) Increased leverage lessens force	
(–) May hit transfer surface and unlock (–) May hit hand when propelling	(+) Not as likely to unlock during transfer (+) Closer for transfers (+) Clear for propulsion	(+) Clear for propulsion (+) Clear for transfers (–) Difficult to manipulate (–) Less surface contact with camber	(+) Easier to reach (+) Easier to operate (–) Decreased brake durability (–) In the way (–) Toggle	(+) Prevents chair from rolling backwards (–) Difficult to propel forward (–) May engage inadvertently (–) Requires treaded tire (–) Prevents recovery from backward fall (–) Low durability
Power wheelchair	(+) Durable (+) Allows power seat functions (+) Allows higher speed (+) Allows adjustable electronics for alternate interface drive controls or ECU control (+) Improved shock absorption (+) Negotiates small curbs and uneven terrain (+) Adaptable for progressive diseases	(–) Heavy (–) Difficult to transport (–) Cost (–) Lengthy repairs		
Scooter	(+) Car transportable	(–) Not bus transportable		
Power Assist	(+) Manual wheelchair frame with power assist add-on components (+) Transportable and collapsible	(–) Cannot utilize power seat functions		

Abbreviations: AROM, active range of motion; ECU, environmental control unit; LE, lower extremity; UE, upper extremity; WC, wheelchair.

a safe manner for travel. The Hippo Car Seat is for transporting children with hip spica casts, broomstick casts, and Ilfeld splints. For those children whose postures require more than a lap belt and shoulder harness, an EZ On vest (http://ezonpro.com) is recommended. It can be used in upright sitting in the rear seat or in side-lying in the back seat. EZ On vests can accommodate children aged 2 to 12, depending on size and weight.

Children with tracheostomies should avoid using child restraint systems with a harness tray/shield combination or an armrest. Upon sudden impact, the child could fall forward and cause the tracheostomy to contact the shield or armrest, possibly resulting in injury and a blocked airway. Five-point harnesses should be used for children with tracheostomies (13).

Transporting wheelchair occupants can be a challenge for many, especially school bus supervisors. Research and accident data show that wheelchair tiedowns and occupant restraint systems (WTORSs) can reduce the possibility of injury by preventing the wheelchair occupant's head from hitting the vehicle interior (19). Several commercially available systems exist that secure the wheelchair to the vehicle, including a four-point belt system, a "docking" station, and a "T" bar configuration. It is also recommended that wheelchairs face forward to avoid collapsibility should there be a collision. In addition to the wheelchair seatbelt and shoulder or chest harness, the standard lap and shoulder belt anchored to the vehicle or the restraint system should be used (5,20).

VEHICLE ACCESSIBILITY: RAMPS AND LIFTS

The safest strategy is to transfer the child into an appropriate car seat; if they need to stay in a chair, there are two options. The first option is two four-point restraint straps in the front and two at the back, which buckle to the floor so that the chair is securely fixed to the floor of the vehicle. If the chair has a solid base, as in a power chair, one can install an "EASY LOCK" (Figure 8.4); a plate with a steel peg is bolted to the bottom of the chair. The chair is steered over a box, the EASY LOCK docking station (Figure 8.5), which is bolted to the floor of the vehicle. This box has a "v"-shaped notch, and when the peg reaches the bottom of the "v," a latch locks it in place. When the chair is to leave the vehicle, a switch releases the latch and the peg is backed out of the "v"-shaped notch in the box.

Getting the child and chair into the vehicle requires either a lift, which is fit to full size vans, or a ramp, which can be fit to minivans with both side and rear entry. There are also several companies that modify vehicles, including non-minivans, such as a Honda Element for side entry. The minivans have adjustments that allow the vehicle to have a kneeling capability. Since the vehicle is closer to the ground, the ramp does not need to be very steep. This is accomplished with a hydraulic suspension system, which is vulnerable to leaks. The non-minivan conversions have the floor lowered, so they may be vulnerable to speed bumps. Ramps come with a hydraulic feature to raise and deploy them or they can be manually raised and lowered. Some families use a minivan, and they use folding ramps, which they set up and take down manually. Recently there has been a side accessible vehicle, the MV-1, which is fully ADA compliant with a ramp that slides out from under the floor and is not a conversion but a vehicle warrantied and designed bumper to bumper for this function.

FIGURE 8.4 Steel peg of EASY LOCK.

RECREATIONAL EQUIPMENT

An integral part of a child's life should be learning and self-exploration through recreational activities and play. Many tricycles now fit the needs of some physically

FIGURE 8.5 Docking station of EASY LOCK.

challenged children. Special features include hand propulsion, wider seats, seatbelts, trunk supports, and chest straps. The Step-N-Go bicycle allows a rider to stand and pedal, making propulsion easier for children with extensor tone. The Rifton Adaptive Tricycle (see www.rifton.com/products/mobility/adaptivetricycles/index.html) provides the user with the ability to sit and pedal. This bike provides multiple positioning supports and the capability to grow. A "roller racer" is a riding toy for children with lower extremity dysfunction. It sits close to the ground and is propelled by moving the handlebars from side to side. Electronic cars can be adapted with switches or a proportional joystick. Scooters can be propelled with arms or legs. Many commercially available mobility devices are on the market today. Further information on recreational equipment is available in the adapted sports and recreation chapter (Chapter 9).

ADAPTIVE INTERFACES

With the level of human interaction incorporated into today's technology, the interface between a device and the child takes on a new meaning and new challenges. The success of a device can be determined by the interface, which takes the form of various technologies. Depending on the physical abilities of the user, the interface between the child and the product can look quite different. A significant proportion of severely disabled people need to use head movements to control assistive equipment such as speech-generating devices, environmental control systems, *assistive reading and writing tools*, and powered wheelchairs (10). Other adaptive interfaces include eye gaze, "sip and puff" systems, chin control devices, and other various switches configured to provide a specific output, depending on the device to be controlled.

Other adaptive interfaces used typically to control one's environment or access to a computer include voice activation. Speech recognition may use software such as Dragon Naturally Speaking *or software that is included in operating systems of many computers, tablets, and smart phones*. Eye gaze *tracking* technology continues to improve *and often can be set to track the coordinated movements of two eyes or more specifically the movement of one eye when a person does not have normal binocular movements*.

As electronics have advanced, particularly in powered mobility systems, so has the ability to integrate controls. Therefore, it is possible to have a power-wheelchair user also control his or her communication device using the same interface that allows him or her to control the power wheelchair. Although these technologies are sophisticated, they offer another means of accessing the environment and maximizing independence. The advantages of integrated control are that persons with limited motor control can access without assistance several devices with one access method and tool (10).

AUGMENTATIVE AND ALTERNATIVE COMMUNICATION

Children who have physical, cognitive, and developmental disabilities and are as young as 2 years arrive for medical and therapy appointments as already competent users of their parents' smart phones and tablets for leisure/entertainment and with early learning apps. Five-year olds with visual, visual–motor, and fine motor planning disorders can be introduced to keyboarding as a possibly more functional alternative to handwriting—but may need to be retrained from swiping on smart phones and tablets as an access method before they learn to point and touch keys.

All children, whether disabled or not, utilize a complex communication system that integrates spoken, written, and pragmatic social language skills. AAC includes low- and high-technology devices that supplement these skills and facilitate language learning. Augmentative communication options are appropriate for any child whose natural speech and writing does not enable him or her to express himself or herself to all listeners in all environments and for all pragmatic communication purposes. In addition, AAC devices should be considered when natural speech and writing does not sufficiently support continued speech, language, and academic learning and success. Rather than slowing down the development of more competent oral speech and articulation skills, using an AAC system to support expressive language (vocabulary, syntax, and pragmatic) learning and teach a child the interpersonal power of "spoken language" can help support speech development and provide more competent language skills when oral speech capabilities catch up.

The communication impairments of children with complex communication needs may be caused by motor speech disorder (such as dysarthria or dyspraxia); a cognitive and language disorder (such as global developmental delay); a pervasive developmental disorder or autism spectrum disorder; a chromosomal abnormality (including Down syndrome); mental retardation; a brain injury; cerebral palsy; or a neuromuscular disorder (such as muscular dystrophy or SCI). Many children from age two through adolescence who can benefit from AAC have a multiplicity of complex diagnoses that may include hypotonia, spasticity, attention deficit disorder (ADD) or attention deficit hyperactivity disorder (ADHD), and/or cortical visual impairment.

Communication behaviors develop spontaneously in all children, regardless of the severity and multiplicity of their disabilities. Nonverbal communication behaviors may manifest as vocalizations for satisfaction and dissatisfaction; eye gaze and eye contact; looking away from a person, place, or thing; idiosyncratic gestures; and physically leading adults to desired objects and places. Even when such communication behaviors are more "reflexive" or self-directed than intentionally interactive,

parents, caregivers, and familiar listeners typically learn to recognize communicative information from their children's behaviors.

One goal of AAC intervention includes introducing communication strategies that help a child develop systematic language and communication behaviors. Systematic communication helps listeners to more readily understand a child's communicative intent, helps to reduce the "20 questions" guesses that parents and caregivers typically engage in, and helps the child and his or her listeners form a communication dyad. With regard to the psychosocial development of children and adolescents with disabilities, the use of a speech-generating device may enable them to shift social and communication control of interactions from parents, teachers, and caregivers to the child—just as happens with typically developing children as they develop *independence* and *interdependence* within the social interactions that occur throughout their home, school/daycare, and community experiences.

Not all augmentative communication devices need to be speech-generating. Low-tech aids can include communication notebooks, communication boards, and Picture Exchange Communication System (PECS) displays. They may be even simpler, including low-tech systems, such as refrigerator magnets or homemade picture magnets displayed on the refrigerator or on a cookie sheet for portability.

It is important that all people utilize their residual speech whenever functional, although many children and adolescents may benefit from an AAC device to augment that speech. Natural speech may be used primarily for initiation and getting attention, with a supplementary device used to communicate specific or complex information (eg, "I want + go + grandma + house" rather than just "go" or "grandma"). Unaided natural speech may be one's primary communication technique, but supplemented by a speech amplifier or a speech-generating device in noisy environments.

AUGMENTATIVE AND ALTERNATIVE COMMUNICATION (AAC) DEVICES

Today's AAC devices typically are hosted by Windows tablet computers, iPad tablets, or Android tablets; some apps are available for smart phones as well. AAC devices should be thought of as having features that support their users' achievement of success across their home, school, community, and leisure (and eventually work) environments. Like all technologies, neither the features themselves nor the functions that open the doors to success can be seen as static. A 2012 holiday gift giving guide about cameras referred to considering "which cameras use their newfound Wi-Fi capabilities." Within a year, it was noted that "new models can back up straight to cloud services or networked computers as well as connect directly to a mobile device, so you can view, transfer, and edit shots, and then upload to sharing sites over your devices mobile broadband. Some models use Wi-Fi to remotely control the camera, too, using your mobile device's display as a viewfinder." The same rapid shift is true—and will continue to be true—of features and capabilities in AAC devices.

The most important features of AAC devices can be thought of as (a) access options; (b) vocabulary and syntax organization; (c) pragmatic language function supports; and (d) language output. Consequently, the most important team members to determine an effective match of an AAC device to an individual are often a speech–language pathologist and an occupational therapist, often in conjunction with a seating specialist for individuals who have positioning and wheelchair needs. Parents as well as adolescents and younger children and other team members (whenever possible) should be active participants in decision making regarding a specific AAC system for an individual. At the very least, determination of a child's need for and development of a recommendation for a specific AAC device should include a detailed parent and team member education process so that they can help make informed decisions about a device and a treatment plan for implementation of that device in a child's natural environments.

Most AAC devices currently use dynamic display technology in order to provide efficient access to expressive language. Dynamic display technology changes its display of vocabulary keys (that contain letters, words, pictures, commands, etc.) according to what has been selected before. The most common version of this is the word prediction displays that smart phones and tablet computers use to make typing e-mails and text messages easier for those who are not efficient users of their small keyboards. A less familiar version of dynamic display language organization includes syntactical prediction of vocabulary so that selection of the verb "go" automatically links to a page of vocabulary that represents different places (perhaps with its own link to a page of "People" since many of us are more likely to talk about going to grandma's house for dinner than to a fast food restaurant).

As modes of communication have been changing throughout society, the modes of communication that AAC devices support also have needed to change—and will need to continually change. High schools and junior high schools are providing access to curriculum materials online and allowing students to submit homework through e-mail. This makes access to written output and electronic communication through the same language system a person uses for face-to-face communication more important to children with disabilities who are even partially included in inclusive education settings. Text messages frequently are the most common way that adolescents socialize, plan, and even schedule school-centered work group meetings. Adolescents who use AAC devices need to have access to receive and send text messages through

their "communication devices" to fully participate in activities with their peers. As electronic health records are more universally used among health care providers and networks, more patients have easier access through secure patient portals to their health information, communication with their health care team members, scheduling appointments, and so on. This can provide adolescent AAC users with more independent and private communication with their health care team members, without being constantly dependent on their parents for communicating their physical statuses, experiences, and feelings with doctors, nurses, and other health care providers.

FIGURE 8.6 Tracker Pro Head Tracking camera from AbleNet Inc. (www.ablenetinc.com).

Access Options

The most common way for people, including children, to access AAC devices is through direct selection—reaching and touching vocabulary keys. AAC devices typically contain options for different numbers and sizes of keys on a screen for people who have different degrees of visual and fine motor patterns. Most of them also have settings to customize how long one has to hold or release keys in order to account for different degrees of spasticity or tremors. Options for different color contrasts between page background, key backgrounds, and key symbols are important for children with some types of visual impairments. Even with customization of such settings, direct selection typically is the fastest and most reliable way for people to access AAC devices. This may require additional wheelchair or desktop mounting equipment to be ordered with the devices in order to optimally position them for children who have neuromuscular or visual impairments.

Mouse, head tracking (Figure 8.6), and eye gaze tracking systems can be considered directed selection and rely on the mouse pointing options of AAC devices that are hosted by computer or tablet systems. This can be accomplished through a standard mouse or trackpad or with "mouse emulators." While targeting desired keys is done through mouse pointing, selection typically is accomplished by pressing a switch (or "mouse button") or by leaving the cursor on the target for a custom-set "dwell time." Head and eye tracking systems are "mouse emulators." Some head tracking systems use specialized cameras that track the movements of the user's head or other body parts, often with a small reflective dot applied to the target body part, and do not require special software. Others use the built-in cameras of an AAC device's host computer and may require special software. Some eye gaze tracking systems most effectively focus their tracking sensors on the eye movements of both eyes together; some can competently track the position of a single eye and can be expected to be more effective for people who have poorly coordinated bilateral eye movements.

Finally, scanning selection techniques are available in many AAC devices for people whose motor control is limited to one, two, or three body parts or movements. These require that one, two, or three switches be placed in effective proximity to the user's movements, but still typically remain the slowest and most physically effortful technique to accessing technology. Customizable settings that give users more direct control and speedier access rates include two-switch scanning (in which activation of Switch #1 moves the scanning cursor to a target and then activation of Switch #2 makes the selection), inverse two-switch scanning (in which holding Switch #1 moves the cursor to a target, release of the switch stops the cursor movement, and activation of Switch #2 makes the selection), and block/row/column scanning (in which the user moves the cursor to a section of the vocabulary display, then to a smaller group such as a row within that portion of the display, and then to an item within the row or even to a section of a digital photograph or "scene") (Figure 8.7).

Vocabulary and Syntax Organization

The most functional AAC devices are those that have complex vocabulary and syntax options as out-of-the-box programming. The ways in which vocabulary is organized are critically important for anybody who cannot access all of the words that he or she might want to say through spelling and typing. On the other hand, there are few of us who can keep up with the pace of fluent conversation by typing. Therefore, language organization systems that focus on phrase and whole word organization (complemented by less frequent use of whole messages) should be considered.

Language organization software and apps may represent phrase, word, or whole message vocabulary through printed words, picture symbols, multimeaning icons, or digital photographs. The most robust language organization may include self-prompting syntactical sequencing features (eg, selecting a phrase such as "I want" at the beginning of a sentence or utterance automatically links

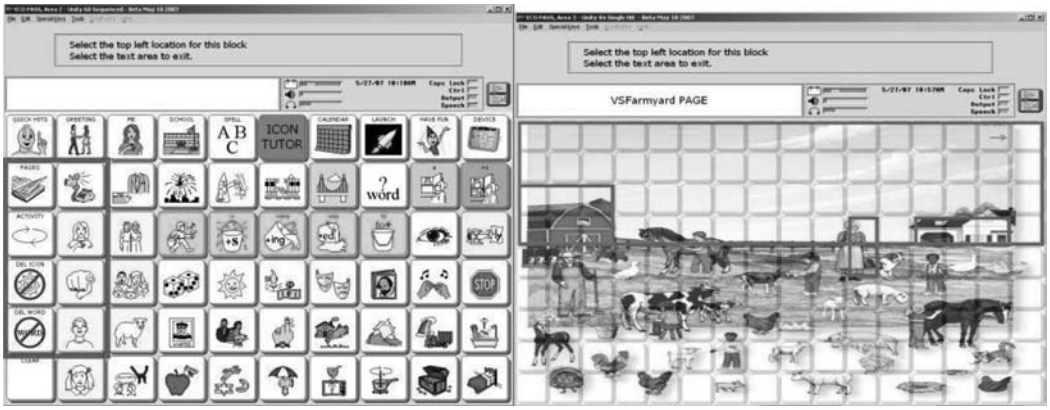

FIGURE 8.7 Examples of block/row/column scanning set-ups.

to a display of verbs; selecting "to play" as a verb results in the visual suggestion of "toys," "games," and "sports" for categories of nouns that might follow) (Figure 8.8).

Children with complex communication needs may use customized whole message vocabulary keys for a range of pragmatically and appropriately socially interactive purposes. For example, telling others about personal information as a personal safety skill can be important but is more frequently used as the basis of introducing oneself to adults and peers and engaging with them in reciprocal, turn-taking conversations (Figures 8.9 and 8.10).

Most young learners, including children with complex communication needs, use early literacy experiences to learn joint attention and language skills. In fact, home assessments by many early intervention specialists include an assessment of how many "literacy artifacts" are visible in the home (books, magazines, crayons, tablet computers with book apps, etc.). When children are verbal or have emerging verbal language skills, adults at home and in school typically read *with* them; when a child is nonverbal, fewer people expose them to story books and, when they do, read *to* them. Most of us find that our attention is shorter when we are talked (or lectured) to without our active participation in the experience. AAC devices can give children with complex communication needs the ability to actively participate in age-appropriate story book reading (and song singing) and access to learning of picture associations, vocabulary, sequential and syntactical language, turn-taking communication interactions, and visual processing with vocabulary pages that largely include pictures that match those in story books (Figure 8.11).

Additionally, digital photographs that are personally meaningful and contextually relevant—such as photos of personal experiences—can form the foundation of personal stories that people of many ages can share with others (and tell multiple times with different communication partners in different conversational contexts) (Figure 8.12).

Finally, language organization options include the uniquely linguistically organized system of Unity software with Minspeak icons. Unity's icons are more conceptual and multimeaning than the more pictographic symbols or printed words of most other AAC systems.

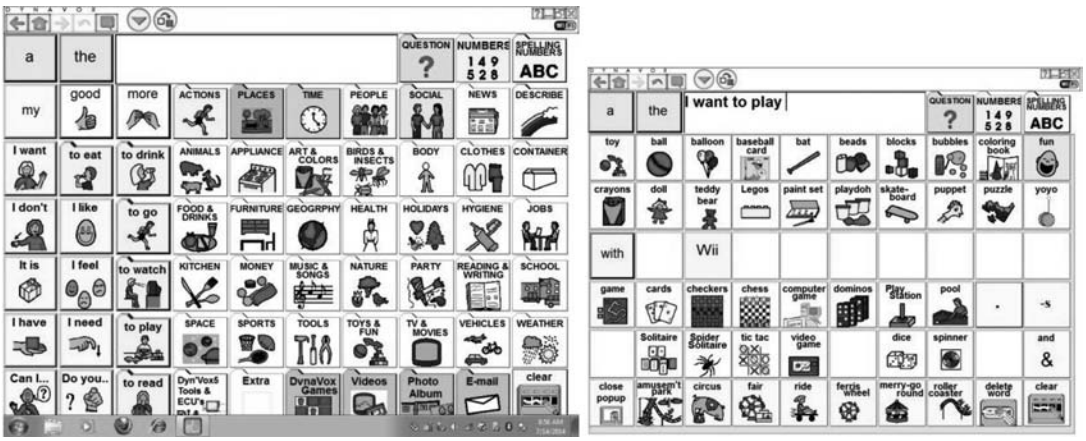

FIGURE 8.8 Picture Word Power with Phrases and Categories (www.inmaninnovations.com).

FIGURE 8.9 Telling others personal information.

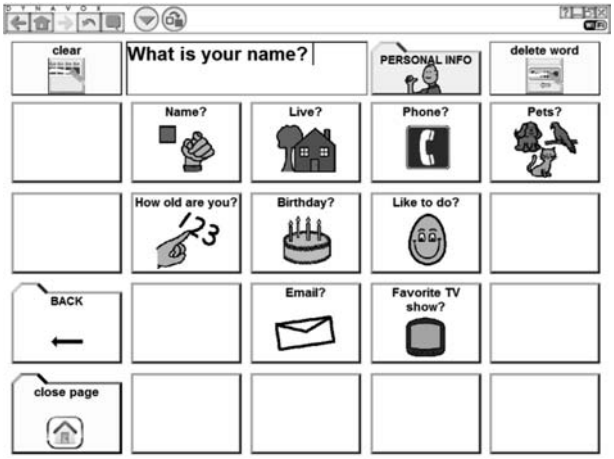

FIGURE 8.10 Asking others for their personal information.

For example, the [apple] icon refers to verbs (including eat, feed, chew, and taste), noun categories (including foods and breakfast foods, lunch foods, dessert, fruits/vegetables), and attributes/adjectives (including delicious and awful) (Figure 8.13). Similarly, the [rainbow] icon refers to verbs (including draw and color), the category of art supplies, name of colors, and attributes/adjectives (including pretty, beautiful, and ugly) (Figure 8.14).

Unity also uses a color coding and layout system that supports learning of its language and vocabulary by the color and the location of vocabulary keys on the devices' touch-sensitive screens. Pronouns and sentence starter phrases are always yellow and always on the left side of the screen; verbs are always green and located in the same two locations of the screen; attributes across topics and concepts are always in the same two or three locations of the screen; and so on. This system minimizes the amount of visual scanning and visual searching needed to learn and access expressive language and instead relies on the development of visual and motor memory patterns. It led to an approach called Language Access through Motor Planning.

While Unity seems like a cognitively complex system, this author has worked with children under 3 years of age who have learned it. One 2.5-year-old nonverbal girl who has significant disabilities due to a chromosomal abnormality had a 4-week trial of the system and learned a great variety of vocabulary and sentence structures. At the end of the trial, her mother commented that they were beginning to understand how much knowledge her daughter had about her world. This author, in re-evaluating her at the end of her trial period, was exploring her ability to use Unity to describe items by combining color and noun vocabulary. When he presented her with the "Brown Bear" story book as one activity structured for this expressive vocabulary and language task, she independently navigated to her "Reading" and "Books categories, opened the "Brown Bear" story page and giggled since taking turns reading the story was a much more fun activity for her.

FIGURE 8.11 Vocabulary page for actively participating and exchanging communication turns when reading "Brown Bear" story book.

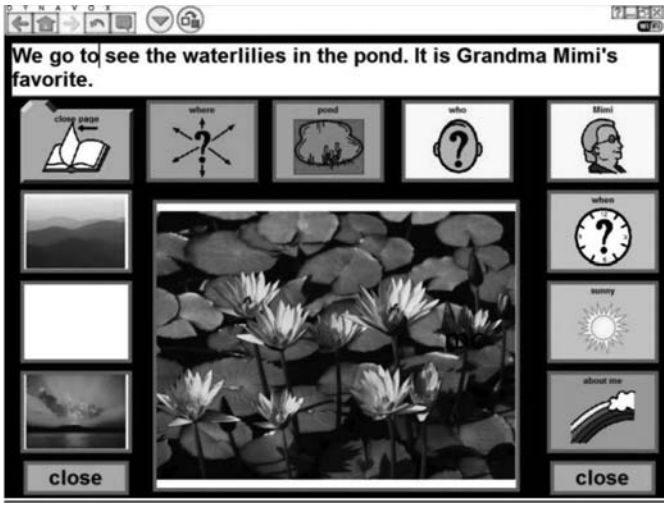

FIGURE 8.12 Vocabulary page to support telling of a Personal Story about going to see water lilies with grandma.

Pragmatic Language Function Supports

Pragmatic language functions refer to the purposes for which people communicate. Children and adolescents with complex community needs and other challenges have as much need to engage in a range of socially appropriate communication interactions as anybody else does. In fact, their need may be greater because until they can engage in socially appropriate communication and conversational interactions that are of as much interest to and valued by their communication partners, their inclusion in activities with nondisabled peers and adults can be expected to be limited.

According to the American Speech–Language–Hearing Association (ASHA), successful use of Pragmatics involves three major communication skills:

1. Using language for different purposes, such as
   - greeting (eg, hello, goodbye)
   - informing (eg, I'm going to get a cookie)
   - demanding (eg, Give me a cookie)
   - promising (eg, I'm going to get you a cookie)
   - requesting (eg, I would like a cookie, please)
2. Changing language according to the needs of a listener or situation, such as
   - talking differently to a baby than to an adult
   - giving background information to an unfamiliar listener
   - speaking differently in a classroom than on a playground
3. Following rules for conversations and storytelling, such as
   - taking turns in conversation
   - introducing topics of conversation
   - staying on topic
   - rephrasing when misunderstood
   - how to use verbal and nonverbal signals
   - how close to stand to someone when speaking
   - how to use facial expressions and eye contact

Source: http://www.asha.org/public/speech/development/Pragmatics/

FIGURE 8.13 Unity's vocabulary related to eating and foods.

Note: Unity is a registered trademark of the Prentke Romich Company (www.prentrom.com).

FIGURE 8.14 Unity's vocabulary related to art and colors.

Note: Unity is a registered trademark of the Prentke Romich Company (www.prentrom.com).

Active participation in one's health care services and treatment planning should be added to this list. AAC devices are more and more commonly used not only by people with chronic disorders and disabilities but also by people who have temporary speech impairments due to postsurgical tracheotomies, other short- or long-term acquired oral speech impairments, or ventilator dependence. The Joint Commission in its 2010 *Roadmap for Hospitals: Advancing Effective Communication, Cultural Competence, and Patient- and Family-Centered Care* stated that "For patients that experience sensory or communication impairment due to their current medical condition, it may be necessary for the hospital to provide auxiliary aids and services or augmentative and alternative communication (AAC) resources to facilitate communication" (www.jointcommission.org/roadmap_for_hospitals).

Language Output

Most current AAC devices utilize synthesized speech output. Although almost all synthesized speech output inherently has a measure of a "robotic" speech quality, it also provides more intelligible speech output when a person syntactically sequences vocabulary selections into original utterances. Digitized (eg, recorded) speech output may be more intelligible for expressing preprogrammed whole messages but the same speaker should then be programming all vocabulary for an AAC device user so that he or she does not speak in a variety of voices. When used for syntactically sequencing phrases and words in originally generative messages, digitized speech output contains the uneven rhythm and intonation with which each individual word and phrase was recorded, thus disrupting the intelligibility of the complete message.

As more and more AAC devices have become hosted by Windows computers, speech synthesizer

software such as ModelTalker has become more commonly thought of. Such software uses "voice banking" (the recording and saving of samples of a person's natural speech or vocalizations) as the basis for processing the live voice into synthesized speech output that has much of the tone and quality of the original natural speech (www.modeltalker.com).

Written output, e-mail output, texting, and posting messages and pictures to social media have assumed progressively more important places in pragmatic, social, and interdependent communication interactions among children as well as adolescents. AAC devices can provide access to these communication techniques even if people are not fluent typists and spellers or cannot effectively access the keyboards (or speech recognition) of standard computing devices. At the time of writing, few insurance companies have considered "computers" and "computing" to be a medical necessity and only cover funding of "dedicated speech-generating devices." Many AAC manufacturers offer the option to have the underlying Windows computer, Android tablet, or iPad tablet "opened" later for a small fee. Perhaps by the time this book is published, those policies may have changed along with the understanding that electronic communication through patient portals in hospitals and doctors' offices; written output for educational successes; and electronic communication through text messages and social media are as critical a necessity as is traditional speech output for helping children and adolescents who have medical diagnoses that create complex communication needs overcome the barriers associated with their disabling conditions.

That day, however, can be expected to come faster only if a growing number of health care providers become advocates for such patients' achievement of *interdependence with others in their immediate and extended social networks*.

Resources

These organizations offer some combination of information, educational programs, and a range of other resources in AAC:

- American Speech–Language–Hearing Association (ASHA): www.asha.org
- Assistive Technology Industry Association (ATIA): www.atia.org
- Association of Assistive Technology Act Programs (ATAP): www.ataporg.org
- International Society for Augmentative and Alternative Communication (ISAAC): www.isaac-online.org
- Patient Provider Communications: www.patientprovidercommunication.org
- RERC on Communication Enhancement: www.aac-rerc.com

ASSISTIVE ROBOTS AND ORTHOSES

AUTOMATED FEEDERS

Task-specific devices exist for feeding such as the Winsford feeder—sold through North Coast Medical and Rehabilitation Products for approximately $3,500. This is a motorized device intended for people without available arm function. They activate a chin switch, which sends a signal to scoop up the food off a mechanized plate and presents it to the user. The Neater Eater (Neater Solutions, Buxton, UK) is a table-mounted feeding device that comes in two versions. The first is a motorized feeding arm that can be controlled by a user with little arm function and retails for about $4,000 (Figure 8.15). It is attached to a tabletop and can be controlled by a foot switch. A manual version is also attached to a tabletop and is for someone with some arm movement but whose movement may be erratic or tremulous. The arm has a built-in damper that filters out unwanted movement.

ORTHOSES

This area has seen a few new devices that have become commercially available. What makes this segment unique is that these are devices that are attached to an appendage (typically the arm) and provide assistance to accomplish activities of daily living. They utilize the remaining residual strength of the individual to allow voluntary movements. They act to amplify weak movements of the arm and negate the effect of gravity for the user so that they can perform tasks such as feeding themselves very easily.

Among the earliest and most accepted devices is the balance forearm orthosis (BFO), also called the mobile arm support (Figure 8.16). The BFO (JAECO Orthopedics, Hot Springs AR), which is a passive (body-powered) device, was developed in 1965. It provides a person with weak

FIGURE 8.15 The Neater Eater.
Courtesy Neater Solutions, UK.

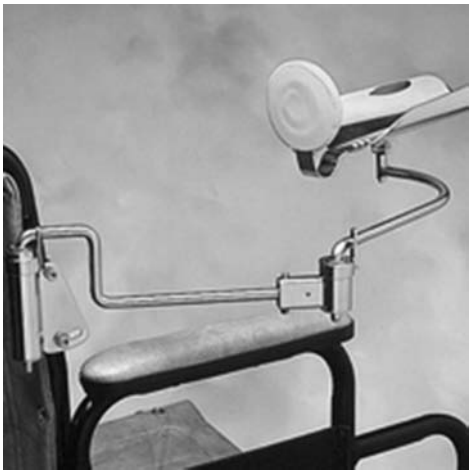

FIGURE 8.16 Balanced forearm orthosis (BFO). Courtesy Jaeco Orthopedics, Hot Springs, Arkansas.

FIGURE 8.18 WREX mounted to a body jacket and constructed using 3D printed parts.

musculature with the ability to move his or her arms in a horizontal plane. Two linkages that have joints along the vertical axes accomplish this. One end of the BFO is attached to a wheelchair; the other end is connected to a trough into which a person places his or her forearm. The trough uses a fulcrum at the forearm that permits the hand to elevate if the shoulder is depressed. The BFO allows a person to move horizontally, for example, over a lap tray and to use compensatory movements to attain limited movement in the vertical direction. The BFO retails for approximately $600 and is available through Patterson Medical.

The Wilmington Robotic Exoskeleton (WREX) is a body-powered orthosis that is modular and mounted to a person's wheelchair or to a body jacket (Figure 8.17). It is a two-segment, four-degree-of-freedom exoskeletal arm, energized by elastic bands that aid in moving the arm in three-dimensional (3D) space. The WREX allows full passive range of motion of the arm and provides a sense of flotation that assists in voluntary movement (21). WREX can easily be adjusted to accommodate subjects of different size weights and arm lengths by changing the number of bands or sliding the telescoping links. WREX can be mounted to a wheelchair (Figure 8.17) or a body jacket (Figure 8.18), and is intended primarily for people with muscular weakness such as muscular dystrophy, spinal muscular atrophy, and arthrogryposis (22). The WREX was conceived and developed at the Alfred I. duPont Hospital for Children and is now manufactured and sold by JAECO Orthopedics, Hot Springs, AR and Patterson Medical, Bolingbrook, IL for approximately $2,000.

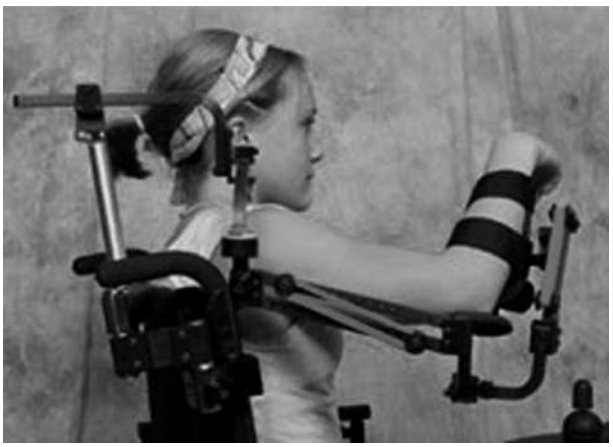

FIGURE 8.17 Wilmington Robotic Exoskeleton (WREX) mounted to a wheelchair.

Two other passive upper extremity orthoses have recently been commercialized and both emanate from the Netherlands. The first is the Armon made by Microgravity Products and is powered by springs. It retails for $3,000 and is for people with arm weakness. It attaches to the forearm of the user and provides gravity balancing. It can be attached to the wheelchair or a tabletop. The Armon does not follow the contours of the arm. It can be adjusted by a motor to compensate for the weight of a person. The second device is called the Dynamic Arm Support (DAS) made by Exact Dynamics. It is similar to the Armon but has a vertical movement that provides the elevation. It too can be adjusted for different sized people with the aid of a motor. It can be attached to a wheelchair.

ROBOTS

The Assistive Robotic Manipulator (iARM; see Figure 8.19) is a six-degree-of-freedom wheelchair-mounted robotic device developed in the Netherlands (Exact Dynamics Inc. Netherlands). As a result of its functionality and mobility, the iARM offers its users a wide range of manipulation possibilities. Example tasks include eating, pouring and drinking, playing board games, operating switches, and opening doors. The iARM manipulator features a

FIGURE 8.19 iARM.

programmable user interface and flexible input/output for interfacing with electrical wheelchairs. The iARM folds into an unobtrusive position at the side of the wheelchair when not in use and folds out when commanded. Its present inputs include a 16-button keypad, trackball, and joystick, which perform individual joint control, integrated hand control, or programmed modes of control. There are currently approximately 400 users of the iARM worldwide. The iARM robot costs approximately $20,000.

THERAPY ROBOTS

The term "rehabilitation robot" has been around for about 35 years when it was applied to assistive motorized devices that performed tasks of daily living for people with physical impairments. As shown previously, these applications are continually being developed; however, the term is being increasingly applied to machines that assist in the recovery from a condition such as stroke. This shift in emphasis from assistive to rehabilitation in robotics is largely driven by an aging population resulting in a far greater number of potential beneficiaries. There are approximately 800,000 new cases of stroke in the United States every year. The "graying" of the population is more pronounced in other countries such as Japan. Patients undergo physical therapy to restore lost function. The therapy tends to be repetitive and evidence suggests that the duration, intensity, and quality of therapy all play a role in recovery. Although functional gains still remain small, the potential of machines assisting in therapy is enormous. These machines are ideally suited to the rigorous and repetitive nature of therapy. The following paragraphs list some of the devices that are currently on the market.

Manually assisted treadmill walking is commonly used for regular therapy for patients with neuromuscular impairments. This type of therapy is performed with some type of harness system that supports the patient's weight. There are two main limitations to this type of therapy; it is labor intensive, as it requires two therapists to move the patient's legs. This causes therapist fatigue and back pain due to awkward ergonomic positions for therapists. Second, manual therapy lacks repeatability and a way to objectively measure performance. The Lokomat (Hocoma AG, Volketswil, Switzerland) is a bilateral robotic gait trainer that is used along with a weight-supported system. It can replace some of the functions of therapists and free them from performing the arduous task of leg movement. The Locomat can provide customized gait training to an individual patient by defining the optimal trajectory of leg movements and creating a specified set of force interactions between the device and the patient. The Locomat has been commercially available since 2000 and is used in numerous clinics for SCI, stroke, and TBI populations. There are over 400 Locomat systems in use worldwide.

Hocoma AG features a line of products for upper extremity therapy. ArmeoSpring is a device based on the WREX. It furthers the WREX by adding an interactive component that contributes to the therapeutic process. Over 500 have been sold by Hocoma AG. ArmeoPower is newer and motorized. It aids in conditions such as stroke, TBI, and other neurologic disorders (23).

InMotion Robots (Interactive Motion Technologies Inc., Cambridge, MA) are a suite of table-mounted robotic systems that provide therapy for the shoulder, elbow, wrist, hand, and overground ankle training. The robots are combined with a video screen to provide a fun and therapeutic environment for exercise. These robots can be programmed to vary the relative effort between the user and the robot. If, for instance, the user is very weak the robot can do most of the work. As the patient gains strength, the robot effort can be decreased appropriately. The InMotion system has been developed over the past 20 years and its strength is that it offers a very low impedance system, so the effect of the robot can be imperceptible to the user. It is used for stroke and other neurologic disorders.

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ADAPTIVE SPORTS AND RECREATION

Michelle Miller and Ellen S. Kaitz†

Adapted sports for the disabled (DA) were introduced in the mid-20th century as a tool for the rehabilitation of injured war veterans. They have developed to encompass all ages, abilities, and nearly all sport and recreational activities, from backyards to school grounds to national, international, and Paralympic competitions. The trend in recent years has led sports away from its medical and rehabilitation roots to school- and community-based programs focused on wellness and fitness, rather than on illness and impairment. However, rehabilitation professionals remain connected in a number of important ways. Sports and recreation remain vital components of a rehabilitation program for individuals with new-onset disability. Furthermore, rehabilitation professionals may be resources for information and referral to community programs. They may be involved in the provision of medical care for participants or act as advisors for classification. As always, research to provide scientific inquiry in biomechanics, physiology, psychology, sociology, technology, sports medicine, and many related issues is a necessary component.

HISTORY

Sports and exercise have been practiced for millennia. Organized activities for adults with disabilities have more recent roots, going back to the 1888 founding of the first Sport Club for the Deaf in Berlin, Germany. The International Silent Games, held in 1924, was the first international competition for DA athletes. Deaf sports were soon followed by the establishment of the British Society of One-Armed Golfers in 1932. Wheelchair sports are younger still, having parallel births in Britain and the United States in the mid-1940s. Sir Ludwig Guttman at the Stoke Mandeville Hospital in Aylesbury, England, invented polo as the first organized wheelchair team sport. "It was the consideration of the over-all training effect of sport on the neuro-muscular system and because it seemed the most natural form of recreation to prevent boredom in hospital . . ." (1). Within a year, basketball replaced polo as the principle wheelchair team sport. In 1948, the first Stoke Mandeville Games for the Paralyzed was held, with 16 athletes competing in wheelchair basketball, archery, and table tennis. This landmark event represented the birth of international sports competition for athletes with a variety of disabilities. The games have grown steadily, now comprising more than two dozen different wheelchair sports. The competitions are held annually in non-Olympic years, under the oversight of the International Stoke Mandeville Wheelchair Sport Federation (ISMWSF).

While Guttman was organizing wheelchair sports in Britain, war veterans in California played basketball in the earliest recorded U.S. wheelchair athletic event. The popularity flourished, and, a decade later, the first national wheelchair games were held. These games also included individual and relay track events. With the success of these games, the National Wheelchair Athletic Association (NWAA) was formed. Its role was to foster the guidance and growth of wheelchair sports. It continues in this role today under its new name, Wheelchair Sports USA.

The U.S. teams made their international debut in 1960 at the first Paralympics in Rome. The term "Paralympic" actually means "next to" or "parallel" to the Olympics. In the 54 years since, the number and scope of sport and recreational opportunities has blossomed. The National Handicapped Sports and Recreation Association (NHSRA) was formed in 1967 to address the needs of winter athletes. It has more recently been reorganized as Disabled Sports USA (DS/USA). The 1970s saw the development of the United States Cerebral Palsy Athletic Association (USCPAA) and United States Association for Blind Athletes (USABA). In 1978, Public Law 95–606, the Amateur Sports Act, was passed. It recognized athletes with disabilities as part of the Olympic movement and paved the way for elite athletic achievement and recognition.

In the 1980s, a virtual population explosion of sport and recreation organizations occurred. Examples of these organizations include the United States Amputee Athletic Association (USAAA), Dwarf Athletic Association of

† Deceased.

America (DAAA), and the United States Les Autres Sports Association (USLASA; an association for those with impairments not grouped with any other sports organizations), the American Wheelchair Bowling Association (AWBA), National Amputee Golf Association (NAGA), United States Quad Rugby Association (USQRA), and the Handicapped Scuba Association (HSA). Internationally, the International Paralympic Committee (IPC) was created in 1989 as a nonprofit organization, headquartered in Bonn, Germany, to organize the summer and winter Paralympic games, promote the Paralympic values, and encourage participation in disabled sports from the beginner to the elite level. It also acts as the international federation for nine sports and oversees worldwide championships in these sports. It recognizes four additional International Organizations of Sports for the Disabled (IOSDs) including the Cerebral Palsy International Sports and Recreation Association (CPISRA), the International Blind Sports Federation (IBSA), the International Sports Federation for Persons with an Intellectual Disability (INAS), and the International Wheelchair and Amputee Sports Federation (IWAS). The awareness of and appreciation for disabled athletes was especially evident in the 2012 Paralympic Games held in London. There were 4,237 athletes present from 164 countries who participated in a total of 503 events in 20 sports. During this event, 251 world records and 314 Paralympic records were set. There was a record 2.7 million spectators and millions more who watched the televised events or accessed the events through the Internet. Surveys of the British noted a significant positive change in attitude toward athletes with disabilities and disabled sports.

While the history of sports for the DA can be traced back a century, the development of junior-level activities and competition can be measured only in a few short decades. The NWAA created a junior division in the early 1980s that encompassed children and adolescents from 6 to 18 years of age. It has since established the annual Junior Wheelchair Nationals. Junior-level participation and programming have been adopted by many other organizations, including the National Wheelchair Basketball Association (NWBA), DS/USA, and American Athletic Association of the Deaf (AAAD). Sports for youth with disabilities are increasingly available in many communities through Adapted Physical Education (APE) programs in schools, inclusion programs in Scouting, Little League baseball, and others.

EXERCISE IN PEDIATRICS: PHYSIOLOGIC IMPACT

It is widely accepted that exercise and physical activity (PA) have many physical and psychological benefits. Much research has been done to support this in adults. More recently, data has been presented to describe the benefits of exercise in both healthy children and those with chronic disease.

Exercise programs in healthy children have resulted in quantifiable improvements in aerobic endurance, static strength, flexibility, and equilibrium (2). Regular PA in adolescence is associated with lower mean adult diastolic blood pressures (3). However, a survey of middle school children showed that the majority are not involved in regular PA or physical education (PE) classes in school (4). Despite this, school days are associated with a greater level of PA in children at all grade levels than free days (5). Requiring PE classes in school improves the level of PA in children, but does not lower the risk for development of overweight or obesity (6) without dietary education and modification (7). Children attending after-school programs participate in greater amounts of moderate and vigorous PA than their peers (8).

Obesity is increasing in epidemic proportions among children in developed countries. It has been linked to development of the metabolic syndrome (defined as having three or more of the following conditions: waist circumference ≥ 90th percentile for age/sex, hyperglycemia, elevated triglycerides, low- and high-density lipoprotein [HDL] cholesterol, and hypertension) (9); both obesity and metabolic syndrome are more common in adolescents with lower levels of PA (10). Insulin resistance is reduced in youth who are physically active, reducing the risk of developing type 2 diabetes (11). Exercise in obese children can improve oxygen consumption and may improve cardiopulmonary decrements, including resting heart rate (12). An 8-week cycling program has been shown to improve HDL levels and endothelial function (13), though in the absence of weight loss, had little effect on adipokine levels (14).

Exercise has positive effects on bone mineralization and formation. Jumping programs in healthy prepubescent children can increase bone area in the tibia (15) and femoral neck, and bone mineralization in the lumbar spine (16). The effects of exercise and weight-bearing may be further enhanced by calcium supplementation (17). The effects on postpubertal teens are less clear.

In children with chronic physical disease and disability, the beneficial effects of exercise are beginning to be studied more systematically. Historically, it was believed that children with cerebral palsy (CP) could be negatively impacted by strengthening exercises, which would exacerbate weakness and spasticity. Recent studies show this to be untrue. Ambulatory children with CP who participate in circuit training show improved aerobic and anaerobic capacity, muscle strength, and health-related quality-of-life scores (18). In ambulatory adolescents with CP, circuit training can reduce the degree of crouched gait and improve perception of body image (19). Performing loaded sit-to-stand exercises results in improved leg strength and walking efficiency (20,21).

Percentage body fat is greater, and aerobic capacity (VO_2/kg) is lower in adolescents with spinal cord

dysfunction than healthy peers. Their levels mirror those in overweight peers. They also reach physical exhaustion at lower workloads than unaffected controls (22). Participation in programs such as BENEfit, a 16-week program consisting of behavioral intervention, exercise, and nutrition education, can produce improvements in lean body mass, strength, maximum power output, and resting oxygen uptake (23).

Supervised physical training can safely improve aerobic capacity and muscle force in children with osteogenesis imperfecta (24). Patients with cystic fibrosis who participate in stationary cycling for aerobic conditioning dislike the tedium of the exercise, but improve their muscle strength, oxygen consumption, and perceived appearance and self-worth (25). Pediatric severe-burn survivors have lower lean body mass and muscle strength compared with nonburned peers; however, both are significantly improved following exercise training (26).

Children with polyarticular juvenile idiopathic arthritis have safely participated in aerobic conditioning programs, with improvements noted in strength and conditioning. Those with hip pain may be negatively impacted, having increased pain and disability (27). The exercise prescription in children experiencing hip pain should be modified to reduce joint forces and torques.

Joint hypermobility and hypomobility syndromes commonly result in pain. These patients demonstrate lower levels of physical fitness and higher body mass indexes, likely secondary to deconditioning (28). These and other children with pain syndromes benefit from increased exercise and PA.

EXERCISE IN PEDIATRICS: PSYCHOSOCIAL IMPACT

The U.S. Department of Health and Human Services published in 2010 that 31% of children (ages 4–11) were reported to be sad, unhappy, or depressed compared with 17% of children without disabilities (29). Regular PA in early childhood through adolescence fosters not only improvements in physical health, but also psychosocial health and development (30,31). The amount and quality of PA have significantly declined over the past several decades and even able-bodied (AB) children are no longer meeting the recommended guideline of 1 hour or more of moderate-intensity PA on 5 or more days a week (32). In disabled children, the amount of PA is even more restricted due to a variety of factors, including the underlying disability, physical barriers, and availability of resources (33). Sit et al. noted that the amount of time spent by children in moderate PA at school during PE and recess was lowest for children with a physical disability, at 8.9%, and highest for children with a hearing loss, at 16.6% of recommended weekly minutes (34). Studies involving AB children have demonstrated that providing game equipment and encouragement from teachers can significantly increase moderate activity levels during recess (35). Deviterne et al. reported that providing participant-specific written and illustrated instruction concerning sporting activities such as archery to adolescents with motor handicaps improves their skill performance to a level similar to an AB adolescent at the end of the learning session that can foster increased self-esteem (36).

Many studies have demonstrated increased social isolation with fewer friendships among disabled children and adolescents. The Ontario Child Health Study revealed that children with a chronic disability had 5.4 times greater risk of being socially isolated and 3.4 times greater risk of psychiatric problems (37). Mainstreaming seems to have a positive impact, although concerns regarding AB peer rejection are still pervasive (38). Children in integrated PE programs were more likely to view their disabled peers as "fun" and "interesting" compared to children who were not integrated (39). One study of teacher expectations in mainstreamed PE classes revealed significantly lower expectations for the disabled student's social relations with peers (40). The attitude toward mainstreamed PE among high school students was significantly more positive in the AB group as opposed to the disabled population (41). Disabled children often view their lack of physical competence and, second, the status among their peers as the major barriers in social competence (42).

In addition to regular PA, play is a major component of childhood and important in psychosocial development of children. In preschool children with developmental delay or mental retardation, they were more likely to play on their own or not participate in play compared to the typically developing peers. Placing them in an integrated playgroup increased peer interactions compared to a nonintegrated playgroup, but did not correct the discrepancy in sociometric measures (43). There have also been discrepancies noted in the type of play for children with developmental delays. These children are less likely to participate in imaginative or constructive play (ie, creating something using play materials) and more likely to participate in functional (ie, simple repetitive tasks) and exploratory play (44). It has been suggested that play should be taught, and one study by DiCarlo demonstrated that a program that taught pretend play increased independent pretend toy play in 2-year-old children with disabilities (45).

Play for children with physical disabilities is also impaired. Children rely on technical aids such as bracing, walkers, wheelchairs, or adult assistants to access play areas and play equipment. Studies have shown that they are seldom invited to spontaneous playgroups and rarely take part in sporting activities unless the activity is geared toward children with disabilities (46). In a study by Tamm and Prellwitz, preschool and school children in Sweden were surveyed about how they viewed children in a wheelchair. They were willing to include disabled children in their games, but saw barriers to participation

in outdoor activities due to the inaccessibility of playgrounds and the effect of weather. They did not feel children with disabilities would be able to participate in activities like ice hockey, but could play dice games. They felt sedentary and indoor activities were more accessible. The children also felt that disabled peers would have high self-esteem, although most literature has documented that disabled children have low self-esteem (47).

In another study, children with motor disabilities were surveyed regarding how they perceived their technical aids in play situations. Younger children viewed their braces, crutches, walkers, or wheelchairs as an extension of themselves and helpful in play situations. Older children also saw the equipment as helpful, but a hindrance in their social life, as it made them different from their peers. Both older and younger children saw the environment as a significant barrier to play. Playgrounds often had fencing surrounding the area, sand, and equipment such as swings or slides that were not accessible without the assistance of an adult. The weather impacted accessibility due to difficulty maneuvering on ice or through snow. Children often took on an observational role on the playground or stayed inside. It was noted that the lack of accessibility sent the message that children with DA were not welcome and further isolated the DA group. As far as adult assistance, the younger children often incorporated the adult as a playmate. As children became older, they viewed their adult assistants as intrusive and a hindrance in social situations. Older children often chose to stay at home and be alone rather than going somewhere with an adult (46).

The research has highlighted many areas for improvement in accessibility for play and social interaction. Several articles detail ways to create accessible playgrounds, and these playgrounds are now becoming more prevalent in the community (Figure 9.1). Playground surfaces can be covered with rubber, and ramps can be incorporated throughout the play structure to allow access by wheelchairs, walkers, and other assistive devices. Playground equipment can include wheelchair swings and seesaws that allow a wheelchair placement (48).

ADAPTED SPORTS AND RECREATION PROFESSIONALS

A variety of fields provide training and expertise in adapted sports, recreation, and leisure. They include APE teachers, child life specialists, and therapeutic recreation (TR) specialists. Physical and occupational therapists often incorporate sports and recreation into their treatment plans as well. However, their involvement remains primarily within a medical framework and will not be discussed here.

APE developed in response to the Individuals with Disabilities Education Act (IDEA), which states that children with disabling conditions have the right to free, appropriate public education in the least restrictive environment. Included in the law is "instruction in physical education," which must be adapted and provided in accordance with the Individualized Education Program (IEP). APE teachers receive training in identification of children with special needs, assessment of needs, curriculum theory and development, instructional design, and planning, as well as direct teaching (49,50). The APE National Standards (51) were developed to outline and certify minimum competency for the field in response to only 14 states developing standards for APE following the passage of the IDEA. APE teachers provide some of the earliest exposure to sports and recreation for children with special needs, and introduce the skills and equipment needed for future participation.

TR has its roots in recreation and leisure. It provides recreation services to people with illness or disabling conditions. Stated in the American Therapeutic Recreation

FIGURE 9.1 Playground equipment can be adapted to include children of all abilities, including pathways for wheelchair and walker access.

Association Code of Ethics, the primary purposes of treatment services are "to improve functioning and independence as well as reduce or eliminate the effects of illness or disability" (52). Clinical interventions used by TR specialists run the gamut, from art, music, dance, and aquatic therapies to animal, poetry, humor, and play therapy. They may include yoga, tai chi chuan, aerobic activity, and adventure training in their interventions. While some training in pediatrics is standard in a TR training program, those who have minored in child life or who have done internships in pediatric settings are best suited for community program development. TR specialists are often involved in community-based sports for those with DA, serving as referral sources, consultants, and support staff.

Child life is quite different from TR. Its roots are in child development and in the study of the impact of hospitalization on children. Its focus remains primarily within the medical/hospital model, utilizing health care play and teaching in the management of pain and anxiety and in support. Leisure and recreation activities are some of the tools utilized by child life specialists. Unlike TR specialists, child life workers focus exclusively on the needs and interventions of children and adolescents. There is often overlap in the training programs of child life and TR specialists. The role of the child life specialist does not typically extend to community sports and recreation programs.

PARTICIPATION IN PHYSICAL ACTIVITY

A number of scales have been developed to measure participation in activities. One example is the World Health Organization Health Behavior in School-Aged Children (WHO HBSC) survey. It is a self-reported measure of participation in vigorous activity that correlates well with aerobic fitness and has been shown to be reliable and valid (53). The Previous Day PA Recall (PDPAR) survey has been shown to correlate well with footsteps and heart rate monitoring, and may be useful in assessing moderate-to-vigorous activity of a short time span (54).

The PA Scale for Individuals with Physical Disabilities (PASIPD) records the number of days a week and hours daily of participation in recreational, household, and occupational activities over the past 7 days. Total scores can be calculated as the average hours daily times a metabolic equivalent value and summed over items (55).

The Craig Hospital Inventory of Environmental Factors (CHIEF) is a 25-item survey that identifies presence, severity, and frequency of barriers to participation, and is applicable to respondents of all ages and abilities. A 12-item short form, CHIEF-SF is also available. When applied to a population with diverse disabilities, the CHIEF measure revealed the most commonly identified barriers to participation are weather and family support (56).

Pediatric measures include CAPE, which stands for Children's Assessment of Participation and Enjoyment. This tool has been validated in AB and DA children aged 6 to 21 years. It is used in combination with the PAC, the Preferences for Activities of Children. Together, they measure six dimensions of participation (ie, diversity, intensity, where, with whom, enjoyment, and preference) in formal and informal activities and five types of activities (recreational, active physical, social, skill-based, and self-improvement) without regard to the level of assistance needed. The scales can be used to identify areas of interest and help develop collaborative goal setting between children and caregivers. Identification of interests and barriers can facilitate problem solving and substitution of activities fulfilling a similar need (57). The European Child Environment Questionnaire (ECEQ) has been used to show that intrinsic and extrinsic barriers are equally important in limiting PA among DA youth (58).

Using these and other measures, one finds that participation in PA varies widely, even among nondisabled populations. The Third National Health and Nutrition Examination survey found that the prevalence of little to no leisure-time PA in adults was between 24% and 30% (59). The groups with higher levels of inactivity included women, older persons, Mexican Americans, and non-Hispanic Blacks. A number of factors have been positively associated with participation in healthy adults, including availability and accessibility of facilities, availability of culture-specific programs, cost factors, and education regarding the importance of PA. Likewise, in healthy adolescents, PA is less prevalent among certain minorities, especially Mexican Americans and non-Hispanic Blacks. Participation in school-based PE or community recreation centers is positively correlated with PA, as are parental education level and family income (60). Paternal PA, time spent outdoors, and attendance at nonvocational schools are more common among children with higher levels of PA (61). Access to parks increases participation, especially in boys. Lower levels of moderate or vigorous PA are seen in those who reside in high-crime areas (62).

When followed over time, adolescents tend to decrease their participation in PA from elementary to high school. Boys who are active have a tendency to pursue more team sports, whereas girls are more likely to participate in individual pursuits (63). Coaching problems, lack of time, lack of interest, and limited awareness have been cited as other barriers to PA (64). Overall, however, informal activities account for more participation in children and teens than formalized activities (65).

Ready access to technology is associated with a decline in healthy children's participation in PA. Television viewing is inversely related to activity levels and positively correlates with obesity, particularly in girls (66). Increased computer time is also related to obesity in teenage girls (67). Interestingly, playing digital games

has not been linked with obesity, and active video games have, in fact, increased levels of PA among children and adolescents (68–70).

It is not surprising to learn that many of the barriers to PA identified by AB are the same as those experienced by children with DA. The most commonly cited are lack of local facilities, limited physical access, transportation problems, attitudinal barriers by public and staff, and financial concerns. Lack of sufficiently trained personnel and of appropriate equipment have also been identified (33,71,72). Among those children with severe motor impairments, the presence of single-parent household, lower family income, and lower parent education are significant barriers (65). Pain is more frequently reported in children with CP and interferes with participation in both activities of daily living (ADLs) and PA (73). The presence of seizures, intellectual impairment, impaired walking ability, and communication difficulties predicts lower levels of PA among children with CP (74). Many children are involved in formal physical and occupational therapy.

Therapists as a whole have been limited in their promotion of recreation and leisure pursuits for their pediatric clientele (75). Therapy sessions and school-based programs provide excellent opportunities for increasing awareness of the need and resources available for PA. Policy and law changes related to the Americans with Disabilities Act are resulting in improved access to public facilities and transportation. Many localities are providing adapted programs and facilities that are funded through local taxation (Figure 9.2). Impairment-specific sports have grown from grassroots efforts, often with the assistance or guidance of rehabilitation professionals. Organizations such as BlazeSports (www.blazesports.org) have developed programs throughout the United States. The bedrock of BlazeSports America is made up of the community-based, year-round programs delivered through local recreation providers. It is open to youth with all types of physical disabilities. Winners on Wheels "empowers kids in wheelchairs by encouraging personal achievement through creative learning and expanded life experiences that lead to independent living skills." Chapters exist in many cities across the United States and incorporate PA into many of the activities they sponsor.

The American Association of Adapted Sports Programs (AAASP) employs athletics through a system called the adaptedSPORTS model. "This award-winning model is an interscholastic structure of multiple sports seasons that parallels the traditional interscholastic athletic system and supports the concept that school-based sports are a vital part of the education process and the educational goals of students" (www.adaptedsports.org). The sports featured in the adaptedSPORTS model have their origin in Paralympic and adult disability sports. The program provides standardized rules for competition, facilitating widespread implementation. Application in the primary and high school levels can help students develop skills that can lead to collegiate-, community-, and elite-level competition.

FIGURE 9.2 Many public facilities have wheelchairs available for rent or use that are designed for use on the beach.

In some communities, AB teams or athletes have partnered with groups to develop activity-specific opportunities. Fore Hope is a nationally recognized, nonprofit organization that uses golf as an instrument to help in the rehabilitation of persons with disabilities or an inactive lifestyle. The program is facilitated by certified recreational therapists and golf professionals (www.forehope.org). A similar program known as KidSwing is available to DA children in Europe and South Africa (www.kidswing-international.com). Several National Football League (NFL) players have sponsored programs targeting disabled and disadvantaged youth. European soccer team players have paired with local organizations to promote the sport to DA children.

Financial resources are also becoming more available. The Challenged Athletes Foundation (CAF) supports athletic endeavors by providing grants for training,

competition, and equipment needs for people with physical challenges. Athletes Helping Athletes (www.athleteshelpingathletesinc.com) is a nonprofit group that provides handcycles to children with disabilities at no cost. The Golden Opportunities fund (www.dsusa.org) provides support and encouragement to DA youth in skiing. More resources can be found at the Disaboom website (vcelkaj.wix.com/disaboom).

INJURIES IN DISABLED ATHLETES

With more DA athletes come more sports injuries. The field of sports medicine for the disabled athlete is growing to keep pace with the increase in participation. Among elite athletes in the 2002 Winter Paralympics, 9% sustained sports-related injuries. Sprains and fractures accounted for more than half of the injuries, with strains and lacerations making up another 28% (75). Summer Paralympians sustained sprains, strains, contusions, and abrasions rather than fractures or dislocations (76). Retrospective studies have shown a 32% incidence of sports injuries limiting participation for at least a day. Special Olympics participants encounter far fewer medical problems than their elite counterparts. Of those seeking medical attention during competition, overall incidence is under 5%, with nearly half related to illness rather than injury. Knee injuries are the most frequently reported musculoskeletal injury. Concerns regarding atlantooccipital instability and cardiac defects must be addressed in the participant with Down syndrome.

Among elite wheelchair athletes, upper limb injuries and overuse syndromes are common; ambulatory athletes report substantially more lower limb injuries. Spine and thorax injuries are seen in both groups (77). Wheelchair racers, in particular, report a high incidence of arm and shoulder injuries. The injuries do not appear to be related to distance, amount of speed training, number of weight-training sessions, or duration of participation in racing (78). A survey of pediatric wheelchair athletes reveals that nearly all children participating in track events report injuries of varying degrees. Blisters and wheel burns are most frequent, followed by overheating, abrasions, and bruising. Shoulder injuries account for the majority of joint and soft tissue complaints. Injuries among field competitors are less frequent, with blisters and shoulder and wrist problems reported most often. Swimmers report foot scrapes and abrasions from transfers, suggesting opportunity for improved education regarding skin protection (79).

An important factor in injury prevention for the wheelchair athlete is analysis of and instruction in ergonomic wheelchair propulsion (80). Proper stroke mechanics positively affect pushing efficiency. Push frequency also affects energy consumption and can be adjusted to improve athletic performance (81). Motion analysis laboratories and Smartwheel technology can be utilized to objectively analyze and help improve pushing technique, thus reducing injury (82).

While some injuries are sport-specific, others may be more common among participants with similar diagnoses. Spinal-cord-injured individuals are at risk for dermal pressure ulcer development, thermal instability, and autonomic dysreflexia. In fact, some paralyzed athletes will induce episodes of dysreflexia, known as "boosting," in order to increase catecholamine release and enhance performance (83). Education regarding the risks of boosting is essential, as are proper equipment and positioning to protect insensate skin.

Athletes with limb deficiencies may develop painful residual limbs or proximal joints from repetitive movements or ill-fitting prostheses. The sound limb may also be prone to injury through overuse and asymmetric forces (84). Participants with vision impairments sustain more lower limb injuries than upper limb injuries, while those with CP may sustain either. Spasticity and foot and ankle deformities in children with CP may further predispose them to lower limb injury. As with all athletes, loss of range of motion, inflexibility, and asymmetric strength further predispose the DA participant to injury. Instruction in stretching, strengthening, and cross-training may reduce the incidence and severity of injury.

"EVENING THE ODDS": CLASSIFICATION SYSTEMS

Sport classification systems have been developed in an attempt to remove bias based on innate level of function. In theory, this would allow fair competition among individuals with a variety of disabilities. Early classifications were based on medical diagnostic groupings: one for athletes with spinal cord lesion, spina bifida, and polio (ISMWSF); one for ambulatory amputee athletes and a separate one for amputee athletes using wheelchairs; one for athletes with CP; one for les autres (International Organizations of Sports for the Disabled [IOSD]), and so forth. These early attempts reflected the birth of sports as a rehabilitative tool. This form of classification continues to be used in some disability-specific sports, such as goalball for blind athletes and sit volleyball for amputee athletes. Other older systems took into account degree of function. This system unfairly penalized athletes who were more physically fit, younger, more motivated, and so forth.

The most recent IPC regulations recognize ten specific impairment types as eligible for participation. They are as follows:

- **Impaired muscle power:** Impairments in this category have in common that there is reduced force generated by the contraction of a muscle or muscle groups (eg, muscles of one limb, one side of the body, the lower half of the body). Examples of conditions included in this category are para- and quadriplegia, muscular dystrophy, postpoliomyelitis, and spina bifida.

- **Impaired passive range of movement:** Range of movement in one or more joints is reduced in a systematical manner. Note that hypermobility of joints, joint instability (eg, shoulder dislocation), and acute conditions of reduced range of movement (eg, arthritis types of impairment) typically will be excluded as "eligible impairment."
- **Limb deficiency:** There is a total or partial absence of the bones or joints as a consequence of trauma (eg, traumatic amputation), illness (eg, bone cancer), or congenital limb deficiency (eg, dysmelia).
- **Leg length difference:** Due to congenital deficiency or trauma, bone shortening occurs in one leg.
- **Short stature:** Standing height is reduced due to aberrant dimensions of bones of upper and lower limbs or trunk (eg, achondroplasia).
- **Hypertonia:** A condition marked by an abnormal increase in muscle tension and a reduced ability of a muscle to stretch. Hypertonia may result from injury, disease, or conditions that involve damage to the central nervous system. When the injury occurs in children under the age of 2, the term cerebral palsy is often used, but it also can be due to brain injury (eg, stroke, trauma) or multiple sclerosis.
- **Ataxia:** Ataxia is a neurologic sign and symptom that consists of a lack of coordination of muscle movements. When the injury occurs in children under the age of 2, the term cerebral palsy is often used, but it also can be due to brain injury (eg, stroke, trauma) or multiple sclerosis.
- **Athetosis:** Athetosis can vary from mild to severe motor dysfunction. It is generally characterized by unbalanced, involuntary movements of muscle tone and a difficulty maintaining a symmetrical posture. When the injury occurs in children under the age of 2, the term cerebral palsy is often used, but it also can be due to brain injury (eg, stroke, trauma).
- **Vision impairment:** Vision is impacted by either an impairment of the eye structure, optical nerves or optical pathways, or visual cortex of the central brain.
- **Intellectual impairment:** The IPC further recognizes the necessity of sport-specific classification systems. To ensure competition is fair and equal, all Paralympic sports have a system in place, which ensures that winning is determined by skill, fitness, power, endurance, tactical ability, and mental focus, the same factors that account for success in sport for AB athletes.

The purpose of classification is to minimize the impact of impairments on the activity (sport discipline). Thus having the impairment is not sufficient. The impact on the sport must be proved, and in each Paralympic sport, the criteria of grouping athletes by the degree of activity limitation resulting from the impairment are named "Sport Classes." Through classification, it is determined which athletes are eligible to compete in a sport and how athletes are grouped together for competition. This, to a certain extent, is similar to grouping athletes by age, gender, or weight. Classification is sport-specific because an impairment affects the ability to perform in different sports to a different extent. As a consequence, an athlete may meet the criteria in one sport, but may not meet the criteria in another sport (IPC, 2007).

ADAPTING RECREATION OPPORTUNITIES

Camping

Camping, mountaineering, and hiking are among the many outdoor adventure activities available to children with disabilities. The National Park Service maintains information on park accessibility and amenities across the United States. The America the Beautiful—National Parks and Federal Recreation Lands Pass is available to any blind or permanently disabled U.S. citizen/permanent resident, and allows free lifetime admission to all national parks for the individual and up to three accompanying adults. Accompanying children under the age of 16 are admitted free of charge. It is obtained at any federal fee area or online at http://store.usgs.gov/pass and allows a 50% reduction in fees for recreation sites, facilities, equipment, or services at any federal outdoor recreation area.

Boy and Girl Scouts of America each run inclusion programs for children with disabilities. Opportunities also exist in dozens of adventure and specialty camps across the United States. Some are geared to the disabled and their families, allowing parallel or integrated camping experiences for disabled children. Participation requires few adaptations, and the Americans with Disability Act has been instrumental in improving awareness in barrier-free design for trails, campsites, and restrooms. Parents should evaluate the camps in regard to the ages of the participants, medical support, and cost. Often, camps are free or offer scholarships and may provide transportation. Some camps will have diagnosis-specific weeks, such as CP, spina bifida, muscular dystrophy, and so on. A nice summer camp resource is www.mysummercamps.com.

There are accessible recreational vehicles (RVs) available for rent as well as purchase. Many manufacturers will customize their RVs during the production process. A number of travel clubs exist across the United States and have websites giving information on accessible campsites with an RV in mind. In addition, many have annual gatherings of their members at a chosen campsite. One good website is www.handicappedtravelclub.com.

Fishing

Fishing can be enjoyed by virtually anyone, regardless of ability. One-handed reels, electric reels, and even sip-and-puff controls allow independent participation. A variety

of options exist for grasping and holding rods as well. These range from simple gloves that wrap the fingers and secure with Velcro or buckles to clamps that attach directly to the rod, allowing a hand or wrist to be slipped in. Harnesses can attach the rod to the body or to a wheelchair, assisting those with upper limb impairments. There are devices that assist with casting as well for individuals with limited upper body strength or control. Depending on the level of expertise and participation of the fisher, simple or highly sophisticated tackle can also be had (85,86).

Both land and sea fishing opportunities are accessible to the disabled. Piers are usually ramped and may have lowered or removable rails for shorter or seated individuals. Boats with barrier-free designs offer fishing and sightseeing tours at many larger docks. These offer variable access to one or all decks, toilet facilities, and shade (85).

Hunting

Adaptations to crossbows and rifles have made hunting accessible for many. The crossbow handle and trigger can be modified for those with poor hand function. Stands for rifles and crossbows are also available for support. Many hunting ranges have incorporated wheelchair-accessible blinds.

Dance

Dancing has become more popular in the AB and disabled populations over the past 15 years. The wheelchair is considered an artistic extension of the body, and many dances have been adapted for the movement of the wheels to follow the foot patterns of classical ballroom dancing. Wheelchair dancing was first begun in 1972 and pairs DA and AB individuals in a variety of dances. Recreational opportunities and competition are available in many states, with classes including duo dance featuring two wheelchair dancers together, group dancing of AB and wheelchair competitors in a synchronized routine, and solo performances. Wheelchair dance sport has been a recognized sport within the Paralympics since 1998, although it is not currently included in the program. International competition in wheelchair dance has been around since 1977. In addition, ballet, jazz, and modern dance companies offer inclusion for children with disabilities.

Martial Arts

Martial arts classes include children with a variety of disabilities. The classes can be modified to allow skills at the wheelchair level in forms, fighting, weapons, and breaking. Children are taught self-respect, control, and can advance through the belt system. They are also taught basic self-defense in some settings. There are many different styles of martial arts, and parents should check within their communities for available resources. Equipment adaptations are not needed for this activity.

Scuba and Snorkeling

Freedom from gravity makes underwater adventure appealing to individuals with mobility impairments. Little adaptation to equipment is needed to allow older children and adolescents with disabilities to experience the underwater world. Lower-limb-deficient children may dive with specially designed prostheses or with adapted fins, or may choose to wear nothing on the residual limb. Similar to those with lower limb weakness or paralysis, they may use paddles or mitts on the hands to enhance efficiency of the arm stroke. Of particular importance is the maintenance of body temperature, especially in individuals with neurologic disability, such as spinal cord injury or CP. Wet or dry suits provide insulation for cool or cold water immersion. They also provide protection for insensate skin, which can be easily injured on nonslip pool surfaces, coral, and water entry surfaces.

It is crucial that individuals receive proper instruction by certified dive instructors. Most reputable dive shops can provide information and referral. The HSA (www.hsascuba.com) is an excellent reference as well. Disabled divers are categorized based on the level of ability. They may be allowed to dive with a single buddy (as with AB divers), two buddies, or two buddies of whom one is trained in emergency rescue techniques. Although there is no particular exclusion from diving based solely on disability, a number of medical considerations may preclude scuba diving, including certain cardiac and pulmonary conditions, poorly controlled seizures, and use of some medications. Discussion with the primary care physician and with dive instructors should precede enrollment or financial investment. Scuba diving has also been used as adjunctive therapy in acute rehabilitation programs (87).

Music

Music has been used both as a therapeutic tool and as a means of artistic expression. Attentive behavior was increased in children with visual impairments who participated in a music program (88). There are many options for children who want to play music. Adaptations may be as simple as a universal cuff with a holder for drumsticks or as sophisticated as a computer program to put sounds together to form a musical piece. Two such computer programs are Fractunes and Switch Ensemble. Adaptive use musical instruments (AUMI) software allows the user to compose music with gestures and movements. Drumsticks can have built-up

FIGURE 9.3 Musical instruments and their video game likenesses may be adapted for use by those with limited strength.

rubberized grips. Straps or a clamp may be used to hold a smaller drum onto a wheelchair for a marching band. Woodwind and brass instruments can be fitted with stands and finger pieces adapted for one-handed playing. Mouthpieces may have different angulations to allow easier access for those who have trouble holding the instrument. Some musical instrument makers, including Flutelab (www.flutelab.com), have become quite creative in how they can adapt their instruments. Other individuals have learned to play instruments such as the guitar with their feet (Figure 9.3).

Hippotherapy and Horseback Riding Therapy

Therapeutic horseback riding, or hippotherapy, has been popular in Europe since the 1950s and spread to the United States in the late 1960s. It uses the rhythmic motions and warmth of the horse to work on the rider's tone, range of motion, strength, coordination, and balance. The movement of the horse produces a pattern of movements in the rider that is similar to human ambulation (89). The rider may sit or be placed in various positions on the horse's back or, alternatively, may perform active exercises while on horseback.

There are two recognized treatment options: instructor-directed, recreational horseback riding therapy (HBRT) and licensed-therapist-directed hippotherapy. HBRT is directed by nontherapist riding instructors and assistants, and follows the North American Riding for the Handicapped Association (NARHA) curriculum for riding therapy. It encourages the development of sensorimotor and perceptual motor skills, utilizing the developmental riding therapy methods described by Spink (90). Children are challenged to maintain balance and posture in all body positions as the horse walks and the instructor encourages them to reach and use their upper limbs in a variety of exercises (91). Hippotherapy is directed by a licensed health professional and focuses treatment based on the impairment and functional limitations of children with neuromuscular dysfunction. The horse is considered a therapeutic tool to improve language or gross motor function, including walking, posture, balance, and mobility (92).

Children with any of a variety of disorders that affect muscle tone, strength, or motor skills may benefit from this form of therapy. These disorders include but are not limited to CP, myelodysplasia, cerebral vascular accident, traumatic brain injury, spinal cord injury, amputations, neuromuscular disorders, and Down syndrome. A careful screening of individuals with spinal pathology should be performed to rule out instability prior to participation. This screening includes the Down syndrome population, in whom 15% to 20% has atlantoaxial instability (93). In addition, children with a poorly controlled seizure disorder may be excluded. Cognitive or behavioral impairments should not be so severe that they place the rider or others at risk.

Many potential physical, cognitive, and emotional benefits of hippotherapy have been reported. These include improvements in tone, posture, balance, strength, gait, hygiene, attention, concentration, language skills, self-confidence, and peer relations (89,94). Most studies have evaluated the effect on the CP population and children with developmental disabilities. Benda et al. noted improvements in back and hip muscle symmetry using remote surface electromyography in children with CP following an 8-minute training session on the horse, as compared to children who sat for 8 minutes on a barrel. Unfortunately, the study did not evaluate if these improvements persisted once therapy was completed (95). Sterba studied the effect of an 18-week training session of riding three times a week on children with different types of CP. Significant improvements in the Gross Motor Function Measure (GMFM) were reported. Progress was noted in all dimensions of the GMFM: lying and rolling; sitting; crawling and kneeling; standing; and walking, running, and jumping during therapy. At 6 weeks following completion of the program, only dimension E (walking, running, and jumping) had continued improvement, with the other domains returning to baseline (96). In a separate area of study, boys with attention deficit hyperactivity disorder (ADHD) and/or learning difficulties demonstrated decreased frustration, physical aggression, and difficulties with authority relations after participating in HBRT (97).

Resource

North American Riding for the Handicapped Association (NARHA)—www.narha.org

Aquatic Therapy

Water has been an important therapeutic medium for centuries. In pool therapy, the water's intrinsic buoyancy nearly eliminates the effects of gravity. Therefore, less effort is required for movement and the weight borne on the limbs is minimized. As recovery progresses, activity in the water can be graded to provide varying amounts of resistance. The water temperature can also be therapeutic, with warmer water producing muscle relaxation. Finally, children often view the pool as fun rather than therapy and are often encouraged by the ability to perform movements in the water that they are unable to do on land (98).

The most common indication for pool therapy is muscle weakness, although gains are also noted in range of motion, coordination, endurance, and normalization of tone. It has been recommended for children with CP, neuromuscular disorders, spinal cord injuries, myelodysplasia, arthritis, brain injury, stroke, burns, fractures, and even asthma (98). Children as young as neonates may benefit (99). Aquatic therapy, however, is not indicated for everyone. Caution should be used in children with hypertension or hypotension, open wounds, infective skin lesions, fever, or temperature instability (98). It is contraindicated for children with uncontrolled seizures or excessive fear of the water, or whose cognitive status poses a safety risk for themselves or others.

There are a variety of approaches in aquatic therapy, including Bad Ragaz, Watsu, Halliwick method, sequential swim techniques (SST), and task-specific approaches (100,101). Bad Ragaz is based on proprioceptive neuromuscular facilitation using active and passive techniques (102). The Watsu approach is an energy-release technique in which a body segment is moved while the rest of the body is allowed to drag through the water, thus providing stretch (103). The Halliwick method and SST work on distinct movement patterns with a specific goal, such as swimming. The task-specific approach includes activities such as ambulation (104).

A review of the literature supporting aquatic therapy in children contains little Class 1 evidence. Most studies are small in sample size and fall within level 4 and 5 evidence (105). One study with Class 2 evidence demonstrated improved vital capacity and water orientation skills (standing in the water, floating, and swim positions) in kindergarteners with CP who participated in a 6-month aquatic program compared to controls in a land-based program (106). In a recent study by McManus et al., children between the ages of 6 and 30 months with delayed functional mobility completed an aquatic therapy program as part of early intervention (EI). There was a significant improvement in motor skills compared to the control group, who received traditional EI therapy services based on the Gross Motor Subsection of the Mullen Scales of Early Learning. The study was limited by the sample size, variety of diagnoses, and the lack of more accepted testing as accomplished by the GMFM or Peabody (107). The adult literature has more evidence-based support of aquatic therapy, and the same types of studies will need to be replicated in the pediatric population.

Aquatic therapy programs are now offered through many hospital programs as well as local facilities such as the Young Men's Christian Association (YMCA) and Young Women's Christian Association (YWCA). Fragala-Pinkham et al. piloted an aquatic exercise program for children with disabilities in conjunction with the local YMCA. They limited participation to ambulatory children with or without an assistive device who did not need constant individualized attention. The program lasted 14 weeks and included swimming laps, strengthening, and pool games. Improvements were noted in swimming ability, endurance, self-esteem, and PA levels. Some of the children were able to participate in inclusive programming at the YMCA following completion of the program and others continued to participate in swimming activities on their own (108).

Yoga/Tai Chi Chuan

Yoga is a mind–body movement therapy with the following components: body mechanics, including breathing skills (pranayama) and simple postures (yogasanas); fitness (sithilikarana, vyayama, and suryanamaskar); and meditation. It has been demonstrated that physiologic changes in the body can be achieved through breathing manipulation, postures, and cognitive control (109,110). There are many different types of Hatha yoga currently being practiced in the United States, each with a different emphasis on the various components.

Studies in the pediatric population have focused primarily on typically developing children, although some have evaluated the effect on those with mental retardation, ADHD, visual impairment, physical impairment, and asthma. The current research has been classified at the 2B level or lower. Primary drawbacks in the studies have been the lack of randomized controlled studies, absent or poor reporting of adverse events, and the wide variety of Hatha yoga protocols used for treatment (111). The existing literature suggests that there can be improvements in mental ability, such as attention, motor coordination, emotional control, and social skills, in children with ADHD or mental retardation (108,112). There was a positive impact in typically developing children on spatial memory, reaction time, motor planning, motor speed, heart rate, and focused attention (111). Children with visual impairments demonstrated less anxiety and children with physical impairments regained some functional ability, with improved flexibility and balance (113). Children with asthma improved their forced expired volume (FEV), peak flow rate, and distance walked in a 12-minute time period, as well as reported decreased symptoms and medication use (114,115).

Tai chi chuan, or tai chi, has been practiced in China for centuries and has recently gained popularity in the United States. It is a low-intensity exercise with flowing, controlled movement patterns emphasizing semisquatting postures, balance, relaxation, flexibility, and regulated breathing. Like yoga, it works to balance the mind and body. There are various styles, including Chen, Yang, Wu, and Sun (116).

Most studies of tai chi have been completed in the elderly population and suggest some benefit for overall balance and prevention of falls, strength, flexibility, reduction of blood pressure, memory, and emotional well-being, with decreases in depression and anxiety (116,117). Studies in the treatment of rheumatoid arthritis have been limited by poor methodological quality, and do not definitively support the use of tai chi as a treatment (118). In their review of the literature, Lee et al. discussed the possible adverse effects of increased pain in the knee, shoulder, and back, yet acknowledged possible improvements in disability index, quality of life, depression, and mood in the rheumatoid population. There are few studies in the pediatric population. One study presented by Yu-Feng Chang et al. noted improvements in asthmatic children in their forced vital capacity (FVC), FEV1, and peak expiratory flow at rest and postexercise after completing a 12-week tai chi program. There was no significant change in their reported symptoms when compared to the control group (119). Further studies are needed to delineate the benefit of this therapy in the pediatric population.

SPORTS FOR FUN AND COMPETITION

Archery

With the exception of the adaptive equipment, archery is essentially unmodified. It is a popular recreational and competitive activity in which individuals with virtually any disability can participate (Figure 9.4).

Equipment

Trigger release or release cuff: Designed for individuals with a poor grasp or weakness, it assists in the smooth draw and release of the bowstring. Its use is permitted in sanctioned competition only by those with tetraplegia from CP or a spinal cord injury.

Wrist and elbow supports: Provide support and stability for the bow arm.

Standing supports: Give the wheelchair user a choice between sitting and standing while shooting.

Bow supports: Provide support and stability of the bow for individuals with weakness or a poor grasp. Its limited use is permitted only in USCPAA competition.

Crossbows and compound bows: For recreational use primarily, although compound bows are allowed in USCPAA competition.

FIGURE 9.4 Minor adaptations allow participation in bow sports.

Mouthpieces: Allow archers with upper extremity impairments to draw the bowstring with the mouth (36).

Resources

Physically Challenged Bowhunters of America, Inc.: http://pcba-inc.org

Grand National Archery Society (United Kingdom): www.archerygb.org

United Foundation for Disabled Archers: www.uffdaclub.com

Baseball

Miracle League is a program facilitating participation of disabled children in a baseball-like activity. In Miracle League play, every player bats once per inning, all base runners are safe, each player scores a run before the inning is over, and the last batter up gets a home run. AB peers and community volunteers assist DA players. Each team and each player wins every game. Another form of the sport is Push N Power Baseball, which utilizes hockey sticks and balls in combination with traditional baseball rules. When unable to catch, pass, or pick up the ball, verbal responses are substituted. Little League baseball also has a division called Challenger, which encourages participation by cognitively and physically challenged children. Teams may have up to 20 players, and may be played as tee ball, coach-pitched, or player-pitched.

Equipment

Sports wheelchair, baseball, glove

Super Sport: Upper extremity prosthesis designed for ball sports (37)

Unihoc hockey sticks and balls

Resources

Miracle League: www.miracleleague.com
Push N Power Baseball rules: www.reocities.com/CollegePark/Lab/5515/BASEBALL.html
Little League: www.littleleague.org

Basketball

Basketball may be played either as an ambulatory or a wheelchair sport. Teams of five play on a regulation basketball court following National Collegiate Athletic Association (NCAA) rules, with only slight modifications to accommodate the wheelchairs. The NWBA uses a classification point system during competition. A junior program was developed by NWBA with four divisions, each having different age requirements, ball sizes, court measurements, time restrictions, and basket heights. It is a popular sport spanning all disabilities. Adapted versions with no contact, no running, no dribbling, and/or lower baskets are useful for developing skills (36).

Equipment

Sports wheelchair, basketball
Super Sport: Upper extremity prosthesis designed for ball sports

Resources

National Wheelchair Basketball Association: www.nwba.org
International Wheelchair Basketball Federation: http://iwbf.org

Bowling

Recreational bowling may include the use of standard lanes with gutter guards (bumpers) and the use of lighter-weight balls. Rules for competitive bowling may be divided into three divisions: AWBA, Special Olympics, and USCPAA. Lane measurements, rules, and bowling balls are the same as in the AB population under the AWBA. However, assistive devices, such as a handle ball, bowling stick, and bowling prosthesis, are allowed. Under the Special Olympics, target bowl and frame bowl are also allowed. Target bowl uses regulation pins, a 2-pound bowling ball, and a carpeted lane that is half the regulation length. Frame bowl uses plastic pins and a ball and a shortened lane. Under the USCPAA, there are four divisions with a ramp or chute allowed. Other rules follow the AWBA recommendations.

Equipment

Handle ball: A bowling ball with a spring-loaded retractable handle for individuals with poor finger control.
Bowling stick: A two-pronged stick similar in appearance to a shuffleboard stick.
Bowling ramp/chute: A wooden or metal ramp from which bowlers can push the ball down using their hands, feet, or a head stick.
Bowling prosthesis: Attaches to a standard prosthetic wrist and fits into one of the holes of the bowling ball. It has a release mechanism activated by stretch on the expansion sleeve.

Resource

American Wheelchair Bowling Association: http://awba.org

Cycling

Cycling is immensely popular as both a recreational and competitive activity. A variety of adaptations are possible to make cycling accessible to a whole range of abilities. Children's tricycles may have blocks, straps, or shoe holders attached to pedals. Backrests and harnesses can be added to the seat to aid in positioning and stability. Adult-sized tricycles can be similarly adapted (Figure 9.5). Specialized terminal devices for upper limb prostheses make grasping handlebars easier, and both brakes can be controlled by one hand for safety. Recumbent cycles afford

FIGURE 9.5 An adult-sized tricycle allows a disabled child to join on family rides.

maximum trunk support for recreational use by those with poor balance as well as by AB riders. Arm-driven units, which attach to the front of a wheelchair frame, are available with anywhere from 3 to 48 speeds. Finally, a variety of tandem cycles or tandem conversion kits are on the market. These range from simple tandems to hybrid hand and leg cycles that allow DA and AB to ride together.

Handcycles are arm-driven cycles with rowing or push–pull drives that assist individuals with lower limb impairment or absence. While used for recreation as well, competitive cycling is a rapidly growing sport. Handcycle races may be held in isolation or in combination with bicycling races. In 2004, handcycling was introduced as a Paralympic sport; triathlons that combine swimming, wheelchair racing, and handcycling are increasingly including junior competitors.

Resources

United States Handcycling Federation: www.ushandcycling.org
Adaptive Adventures: www.adaptiveadventures.org
US handcycling: www.ushf.org
Union Cycliste International: www.uci.ch

Football (American)

Rules for wheelchair football vary from league to league. There is one national competition, the Blister Bowl, which is held in California. There are six players per team, one of whom must be female or tetraplegic. The asphalt field measures 60 by 25 yards and is divided into 15-yard segments. Play follows NCAA rules and is similar to touch football, with players advancing the ball by running or passing. All players are eligible receivers. Four 15-minute quarters are played. Participants primarily include individuals with amputations, CP, spinal cord injury, and les autres. Wheelchair football is not yet recognized as an "official" sport. The game may be also played on a basketball court indoors.

Equipment

Sports wheelchair, regulation football

Resource

Universal Wheelchair Football Association:
www.mobility-Advisor.com/wheelchair-football.html

Goalball

Goalball is played on a gym floor measuring 60 x 40 feet by blind or visually impaired players. All individuals are blindfolded since there are varying degrees of visual impairment. Each team of three players takes opposite ends of the court and plays two 10-minute halves. The goal is to roll the goalball, a 3-pound ball with bells inside, past the opposing team and across the end line. Players listen for the ball and dive to block it from advancing further. If blocked successfully, they may throw it back or pass it to another teammate to throw.

Equipment

Goalball

Resources

USA Goalball: www.goalball.us
www.ibsasport.org/sports/goalball

Hockey

Floor hockey is, in some respects, similar to ice hockey. It is played in a gymnasium with a minimum playing area of 12 × 24 meters and a goal at each end. Teams are composed of six players, who play three 9-minute periods. The puck is a felt disc, and hockey sticks are wood or fiberglass rods. Games may be either ambulatory or played from wheelchairs. A similar sport, poly hockey, uses a hard plastic puck, a smaller plastic version of the conventional ice hockey stick, and a playing area measuring 12 × 24 meters at a maximum. Canada has further developed a version for power-wheelchair users using a 3-inch plastic ball rather than a puck and following National Hockey League (NHL) rules. Sledge hockey (sled hockey in the United States) is played on a regulation-size ice rink using a standard puck or small ball and short sticks called pics. Players are seated on a sledge, which is an oval-shaped frame with two skate-like blades and a runner. Pics are used to propel as well as to advance the puck or ball (Figure 9.6).

Equipment

Hockey sticks/pics, puck/ball, goals, helmet, kneepads, elbow pads, shin guards, sled

Resource

United States Sled Hockey Association:
www.usahockey.com/sledhockey

Quad Rugby

Quad rugby combines aspects of basketball, hockey, and soccer into an exciting sport developed for tetraplegic individuals. It is played with a volleyball on a regulation-size

FIGURE 9.6 Sled hockey is as fast-paced and thrilling as its able-bodied counterpart.

basketball court with goals at both ends measuring 8 × 1.75 meters. Teams consist of four players in manual wheelchairs, who play four 8-minute quarters. Players are classified from 0.5 to 3.5 in 0.5 increments, based on increasing arm function and trunk control. The combined point value of players on the floor may not exceed 8.0 at any time. The ball must be advanced over midcourt within 15 seconds of possession, and the ball must be bounced or passed within 10 seconds. A goal is scored when two of the players' wheels cross the goal line with the volleyball under control. Penalties may result in loss of possession or a trip to the penalty box, depending on the infraction.

Equipment

Volleyball, gloves, straps (trunk, legs, feet), quad rugby wheelchair: must have antitippers

Resources

International Wheelchair Rugby Federation: www.iwrf.com
Canadian Wheelchair Sports Association: www.cwsa.ca
United States Quad Rugby Association: www.usqra.org

Racquetball

Racquetball may be either an ambulatory or a wheelchair sport. It is played on a regulation-size racquetball court and follows the rules of the American Amateur Racquetball Association. There are novice, intermediate, open, junior, two-bounce, and multiple-bounce divisions. It is recommended that players using wheelchairs equip their chairs with roller bars or wheels under the footrest and with nonmarking tires. Racquetball is another of the sports in which DA and AB players can play side by side.

Equipment

Standard racquet: A built-up grip or wrapping the handle to the player's hand may be required for those with grip difficulties.
Standard balls, lightweight sports wheelchair

Resource

USA Racquetball: www.usra.org

Road Racing

As running has increased in popularity as a recreational and competitive sport, DA athletes have formed their own running clubs and begun to participate in a variety of road races. Training is usually done on the road or a track. For the wheelchair road racer, rollers are also available. The racing chair is placed on the rollers allowing for freewheeling and training indoors. The rules for road racing are no different between the AB and DA populations: Whoever crosses the finish line first, wins. DA athletes are placed in functional classes to make the competition more equitable. Power wheelchairs are not permitted in competition. Distances range from the 1-mile fun runs to full marathons. Many of the well-known AB marathons now include one or more wheelchair divisions. The longest wheelchair race to date is the Midnight Sun Wheelchair Marathon, which covers 367 miles from Fairbanks to Anchorage, Alaska.

Equipment

Sports wheelchair: Customized racing wheelchairs are available for serious athletes; three-wheelers are most popular.
Gloves

Resources

DS/USA: www.dsusa.org
Cerebral Palsy International Sports and Recreation Association: www.cpisra.org
BlazeSports: www.blazesports.org
Wheelchair Sports USA: www.wsusa.org
Adaptive Adventures: www.adaptiveadventures.org

Skiing: Alpine

In the past 30 years, adaptive skiing has grown immensely in popularity. With the advances in adaptive equipment, all disability groups can participate in this sport. Skiing techniques include three-track, four-track, and sit skiing. Three-trackers use one ski and two outriggers, thus creating three tracks in the snow. Outriggers are essentially modified Lofstrand crutches with short skis attached with a hinge. They provide additional balance and steering maneuverability. Single-leg amputees and individuals with hemiplegia are often three-trackers. Four-trackers use two skis and two outriggers. In those with spasticity or poor leg control, a ski bra can be attached to the ski tips. This will prevent the ski tips from crossing. Individuals with muscular dystrophy, spina bifida, paraplegia, and CP typically use four-track skiing. Sit skiing utilizes a mono-ski or bi-ski and two outriggers. All disability groups can sit ski. A tether, which allows the instructor to slow the skier down, is required until the sit ski is mastered. Tethers can also be beneficial during instruction in the ambulatory population. Competitive racing includes slalom and downhill courses.

Equipment

Outriggers, skis, ski bra, ski boots
Ski hand/All-Terrain Ski Terminal Device: specialized terminal device for upper limb amputees
Ski leg: A variety of ski-specific lower extremity prostheses are available

Resources

United States Ski and Snowboard Association: www.ussa.org
U.S. Ski Team: www.usskiteam.com
Ski Central: http://skicentral.com
Sitski: www.sitski.com

Skiing: Nordic

Standing skiers can often participate in Nordic (cross-country) skiing with standard equipment, sometimes modified to accommodate prostheses or braces. Sit skis are also available as in alpine skiing, although the ability of the participant to self-propel is often limited by the weight of the equipment. Tethers may be used to assist in forward movement. Biathlon is a sport consisting of cross-country skiing and target shooting.

Equipment

Outriggers, skis, ski boots, sit ski
Ski hand/All-Terrain Ski Terminal Device: specialized terminal device for upper limb amputees
Ski leg: A variety of ski-specific lower extremity prostheses are available

Resources

United States Ski and Snowboard Association: www.ussa.org
U.S. Ski Team: www.usskiteam.com

Soccer

There are very few modifications to the actual game, and the rules of the United States Soccer Federation are followed (Figure 9.7). The modifications include seven players on a team, a smaller field measuring 80 × 60 meters, and occasionally, a smaller goal. These modifications result from fewer participants in a given area. A smaller goal is indicated in the CP population in whom mobility impairments make a larger goal more difficult to defend. Crutches have been allowed for some competitors with lower extremity amputations who do not use a prosthesis.

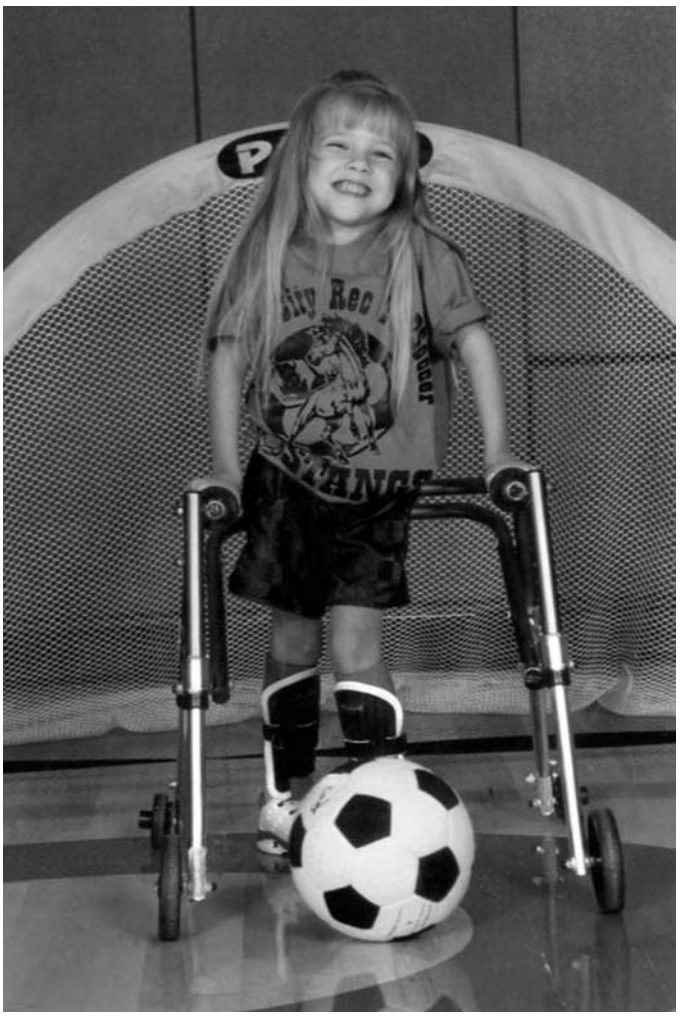

FIGURE 9.7 Soccer can be played by ambulatory children with gait aids, or by power-wheelchair users utilizing larger balls at indoor facilities.

Equipment

Regulation-size soccer ball
Super Sport: Upper extremity prosthesis designed specifically for ball handling

Resources

American Amputee Soccer Association: www.ampsoccer.org
TopSoccer: www.usyouthsoccer.org/programs/TOPSoccer.asp

Softball

Dwarf softball is played according to the rules of the Amateur Softball Association without any modifications. The Special Olympics offers a variety of competitive events, including slow-pitch softball and tee ball. Wheelchair softball is also available primarily for individuals with spinal cord injuries, amputations, CP, or les autres conditions. It is played on a hard surface with the pitching strip 28 feet from home base and other bases 50 feet apart. Players must use a wheelchair with a foot platform and are not allowed to get out of their chairs. Ten players make up a team, and one of the players must be tetraplegic. The WS/USA point classification is used, and total team points on the field may not exceed 22. A larger ball is used, eliminating the need for a mitt, which would interfere with propelling.

Equipment

Softball, mitt
Prostheses: Upper extremity terminal devices that fit into a mitt or substitute for a mitt are available. A set of interlocking rings can also be attached to the bottom of a bat, allowing an adequate grip by a prosthetic hand.

Resources

National Wheelchair Softball Association: www.wheelchairsoftball.org

Swimming

Swimming is a universal sport in which all disability groups may participate. Numerous competitive events are offered across the United States. These include races of a variety of distances in freestyle, breast stroke, backstroke, butterfly, individual medley, freestyle relay, and medley relay. Classification systems have been developed by each DA sports organization to divide participants into classes based on impairment. In addition, swimmers are grouped according to gender and age. Flotation devices are often recommended, although only allowed in competition in two USCPAA classes. Flotation devices include tire tubes, inflatable collars, waist belts, life vests, head rings, water wings, and personal flotation devices. The use and choice of device is dependent on swimming ability, swimming style, and experience (Figure 9.8).

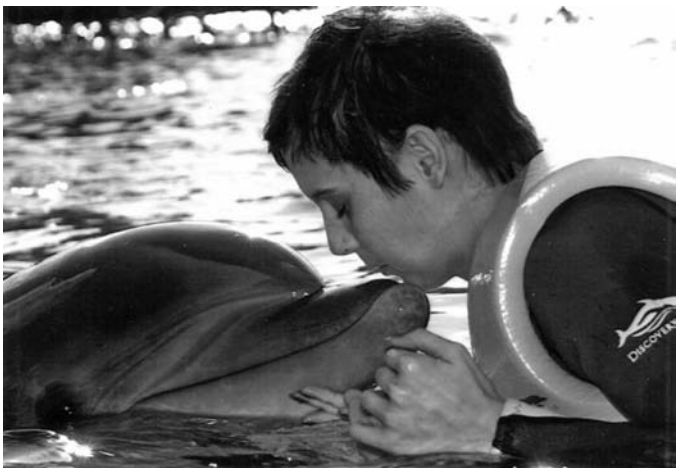

FIGURE 9.8 Water sports are made easier with flotation devices supporting weak limbs.

Equipment

Flotation device, lift, or ramp
Prosthetics: Includes swim fins attaching to lower extremity prosthetic sockets and swimming hand prostheses. These are generally not allowed in sanctioned competition.

Resource

USA Swimming's Disability Swimming Committee: www.usaswimming.org

Table Tennis

Only slight modifications involving the delivery of the serve differentiate this sport from AB competition, which follows United States Table Tennis Association rules. The only equipment modifications allowed are to the paddle and, in the case of dwarf competition, floor raisers to make up for height differences. In recreational play, side guards may be added to the table to keep the ball in play longer.

Equipment

Velcro strap or cuffs: Allow correct placement of the paddle in the player's hand
Regulation-size table, paddles, ball

Resources

U.S. Disabled Athletes Table Tennis Committee: www.midy.com/~usatt/parapong
ITTF Para Table Tennis: http://ipttc.org

Tennis

Wheelchair tennis is played on a regulation-size tennis court as either a singles or doubles game. Players are allowed a maximum of two bounces before the ball must be returned. Scoring and other rules follow the United States Tennis Association guidelines. Players are broadly divided into two groups: paraplegic and tetraplegic. Within these divisions, players compete in subdivisions based on their skill. This sport is open to all disability groups. When a wheelchair user plays against an AB opponent, the rules of each one's sport applies to their respective side of the court.

Equipment

Sports wheelchair, tennis racquet, straps (trunk, legs, feet)
Racquet holder: Ace wrap or taping may provide additional support of grip strength if needed. Alternatively, a racquet holder orthosis may be beneficial.

Resources

United States Tennis Association: www.usta.com
International Tennis Federation: www.itftennis.com/wheelchair

Track and Field

Track and field events are some of the most popular of the adapted sports competitions and involve individuals from all disability groups. Track events may be ambulatory or at the wheelchair level. Ambulatory and wheelchair events range in distance from 10 meters to a full marathon, and take place on a typical track. Running, walking, and hurdles are all included in the ambulatory division. Power and manual wheelchair slalom races are available in the Special Olympics.

Field events typically include shot put, discus, javelin, long jump, and high jump. The USCPAA has also developed seven events for those athletes who are more physically impaired. These include the distance throw, soft discus, precision event, high toss, thrust kick, distance kick, and club throw. In the distance throw, athletes throw a soft shot as far as possible. The soft discus is similar to the conventional discus, except that the discus is made of a cloth material. For the precision event, six soft shots are thrown at a target, with points awarded for accuracy. The high toss involves throwing a soft shot over a progressively higher bar. Athletes have three attempts to clear the height. In the thrust kick, athletes kick a 6-pound medicine ball away from them, with their foot in constant contact with the ball. The distance kick is similar; however, it uses a 13-inch rubber ball and allows the athlete to initiate a back swing with the foot prior to striking the ball. For the club throw, an Indian club is thrown as far as possible.

Equipment

Racing gloves
Sports wheelchair: Custom-designed racing chairs are available for the serious athlete
Throwing chair: Provides a stable platform from which athletes may throw

Resources

BlazeSports: www.blazesports.com
Wheelchair Sports USA: www.wsusa.org
Special Olympics: www.specialolympics.org

PEARLS AND PERILS

- Major barriers to participation for children and adolescents with disabilities include lack of transportation, financial constraints, and physical and attitudinal barriers. The presence of an adult assistant further distances disabled children from their AB peers.
- Strengthening exercises in children with spasticity are not contraindicated, and often result in improved strength, aerobic capacity, and quality of life.
- While active weight-bearing exercises such as jumping result in increased bone density, the osteogenic benefits of passive weight-bearing are less clear.
- Access to technology such as video games and computers has resulted in a trend of lowered PA and increased obesity among AB and DA youth. However, use of active video games is resulting in increased levels of PA. This technology is also being implemented in habilitative and rehabilitative therapy programs.
- Sport- and disability-specific injury patterns are being recognized among disabled youth, leading to a new field of sports medicine for the disabled. Prescription of appropriate training and equipment are among the tools necessary for the pediatric rehabilitation professional.

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10

ORTHOPEDICS AND MUSCULOSKELETAL CONDITIONS

Kevin P. Murphy, Colleen A. Wunderlich, Elaine L. Pico, Sherilyn Whateley Driscoll, Elizabeth Moberg Wolff, Melanie Rak, and Maureen R. Nelson

GROWTH AND DEVELOPMENT OF THE BONY SKELETON

The skeletal system develops from mesoderm and neural crest cells (1). Somites form from paraxial mesoderm and differentiate into sclerotomes, dermatomes, and myotomes. Sclerotome cells migrate from the somite and ultimately become chondrocytes. The remaining dermatome cells form the dermis. Myotome cells give rise to striated muscles of the backs of limbs (Figure 10.1).

Limbs and respective girdles, the appendicular skeleton, are derived from cells of the lateral plate mesoderm. Limb buds appear in utero approximately on day 26 for the upper extremities and day 28 for the lower extremities (2). The hand plate forms in the fifth week, with digitization of rays in the sixth week. Notches appear between the rays in the seventh week, the failure of which results in syndactylism. During the seventh week, the limbs also rotate laterally in the upper extremities and medially in the lower extremities.This brings the thumb to the more lateral position in the upper extremity and the great toe to the more medial position in the lower extremity. Chondrification begins in the sixth week, followed by early ossification in the seventh week and subsequent joint cavity formation in the sixteenth week. By the eighth week, definite muscle formation is noted, as the embryo assumes a human appearance and basic organ systems are completed. The fetal period begins at 9 weeks with rapid growth and changes in body proportion (3).

Knowledge of the normal proportions and growth and development of the musculoskeletal system allows a firm foundation for the understanding of both congenital and acquired conditions requiring care in the developmental years.

Figure 10.2 displays the growth rates for boys and girls by age. About half of the individual's height is reached by age 2 and three-fourths by age 9. Prediction of adult height can be obtained by plotting bone age against current height to determine percentile value (Figure 10.3). Following the percentile to skeletal maturation is a method for estimating final adult height. Paley height multipliers offer an even simpler way of estimating adult height at any child age (4). Predictions are less accurate for the younger child (Figure 10.4). The reader is referred to more detailed references for tables displaying differences over time and growth and rates for standing, sitting, and subischial lengths in boys and girls (5). The measurement of arm span provides an indirect control parameter for the measurement of standing height, particularly useful in those who are nonambulatory. To measure arm span, the patient simply raises the arms to a horizontal position, and the distance between the tips of the middle fingers is measured with a tape measure (6,7). The standing height is about 97% of arm span. In children with spinal deformity, arm span is a good estimate of what standing height would be if there were no abnormal curvatures. It is well known that different proportions of the body grow and change at different percentages over the developmental years (Figure 10.5).

CONGENITAL CONDITIONS

Minor limb deficiencies are relatively common in the upper and lower extremities. Syndactyly occurs in approximately 1 in 2,200 births, either as cutaneous with simple webbing of the fingers or osseous with fusion of the bones when the digital rays fail to separate between the fifth to eighth weeks of gestation (8). It is most frequent between the third and fourth fingers and between the second and third toes, and is inherited as a simple dominant or simple recessive trait. It can occur in isolation or as part of a syndromic condition. Surgical separation of

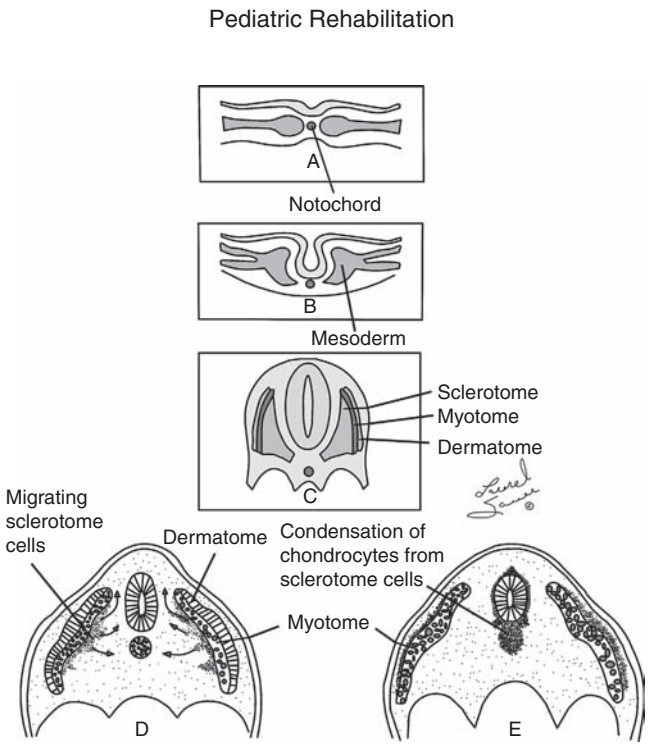

FIGURE 10.1 Trilaminar disk. Neural tube closure. Mesoderm differentiates into dermatome, myotome, and sclerotome. Migrating sclerotome cells become chondrocytes. Chondrocytes ultimately form vertebral bodies and arches.

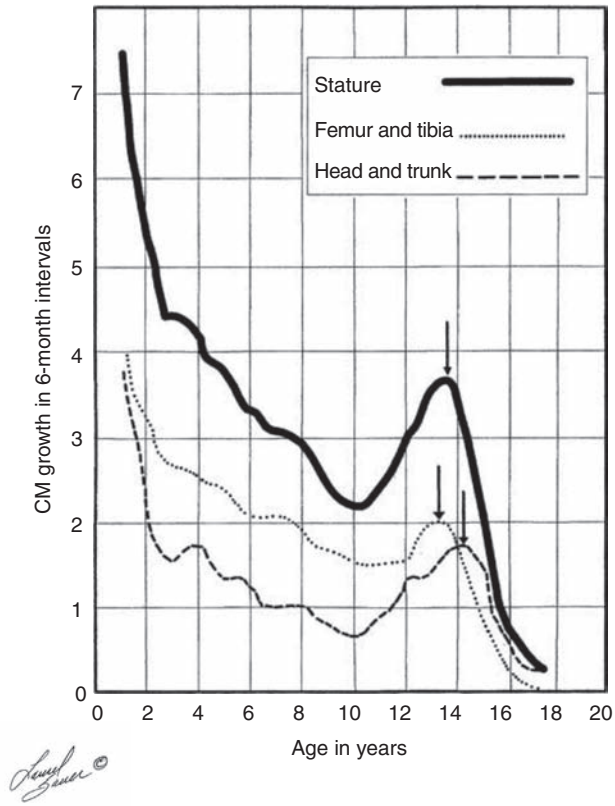

FIGURE 10.2 Greene and Anderson growth curve.

Source: Greene W, Anderson M. Skeletal age and the control of bone growth. *Instr Lect Am Acad Orthop Surg*. 1960;17: 199–217.

the digits is more common with complete syndactyly for functional and cosmetic reasons. Polydactyly has an incidence of approximately 1 to 1.5 per 1,000 live births and is the most common congenital toe deformity (5). Eighty percent of polydactyly in the foot occurs with the fifth toe. Most often an isolated trait, an autosomal-dominant inheritance pattern has been identified with variable expressivity. Radiographic evaluation is necessary to define duplicated structures. Deferring radiography until after 6 months of age allows phalanges to ossify. Surgery around the age of 1 not only improves cosmesis, but also is helpful in facilitating shoe fitting.

Camptodactyly, translated from Greek, means "bent finger." The proximal interphalangeal (PIP) joint is flexed, most commonly digit 5. Incidence is felt to be less than 1% of the general population with equal gender distribution (8,9). Appearance in adolescence, often girls, is less common. Surgical reconstructions are for functional and cosmetic reasons.

Malformations of the radius are more common than those of the ulna and are associated with numerous syndromes (5,10,11,12). In children with limb anomalies, a multisystemic review is generally indicated because abnormalities in other systems are often present. Simple and multifactorial inheritance may all be causative in addition to teratogenic effects, such as maternal exposure

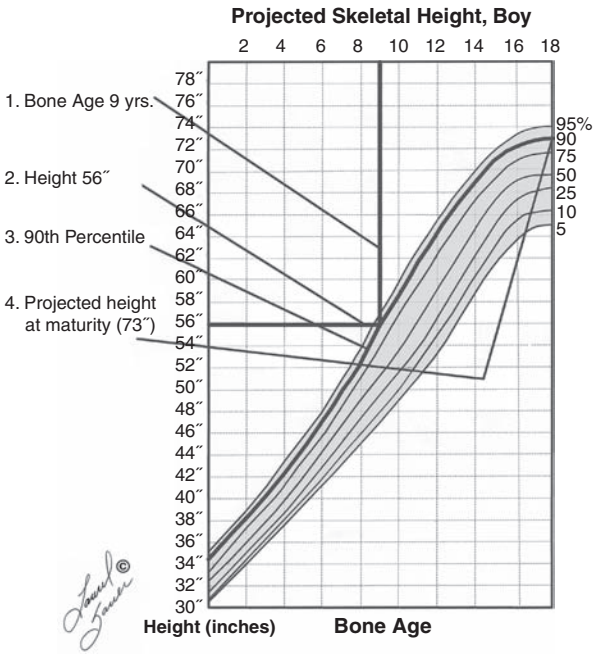

FIGURE 10.3 Prediction of adult height. Adult height predicted by plotting child's bone age versus current height to determine percentile value. Follow the percentile to skeletal maturation for estimate of final adult height.

Height Multiplier: GIRLS, Birth to 18 Years

Age (year + month)	M	Age (year + month)	M
Birth	3.290	8 + 6	1.254
0 + 3	2.759	9 + 0	1.229
0 + 6	2.505	9 + 6	1.207
0 + 9	2.341	10 + 0	1.183
1 + 0	2.216	10 + 6	1.160
1 + 3	2.120	11 + 0	1.135
1 + 6	2.038	11 + 6	1.108
1 + 9	1.965	12 + 0	1.082
2 + 0	1.917	12 + 6	1.059
2 + 6	1.815	13 + 0	1.040
3 + 0	1.735	13 + 6	1.027
3 + 6	1.677	14 + 0	1.019
4 + 0	1.622	14 + 6	1.013
4 + 6	1.570	15 + 0	1.008
5 + 0	1.514	15 + 6	1.009
5 + 6	1.467	16 + 0	1.004
6 + 0	1.421	16 + 6	1.004
6 + 6	1.381	17 + 0	1.002
7 + 0	1.341	17 + 6	–
7 + 6	1.309	18 + 0	–
8 + 0	1.279	Jonathan Paley et al., JPO 2004	

Height Multiplier: BOYS, Birth to 18 Years

Age (year + month)	M	Age (year + month)	M
Birth	3.535	8 + 6	1.351
0 + 3	2.908	9 + 0	1.322
0 + 6	2.639	9 + 6	1.298
0 + 9	2.462	10 + 0	1.278
1 + 0	2.337	10 + 6	1.260
1 + 3	2.239	11 + 0	1.235
1 + 6	2.160	11 + 6	1.210
1 + 9	2.088	12 + 0	1.186
2 + 0	2.045	12 + 6	1.161
2 + 6	1.942	13 + 0	1.135
3 + 0	1.859	13 + 6	1.106
3 + 6	1.783	14 + 0	1.081
4 + 0	1.731	14 + 6	1.056
4 + 6	1.675	15 + 0	1.044
5 + 0	1.627	15 + 6	1.030
5 + 6	1.579	16 + 0	1.021
6 + 0	1.535	16 + 6	1.014
6 + 6	1.492	17 + 0	1.010
7 + 0	1.455	17 + 6	1.006
7 + 6	1.416	18 + 0	1.005
8 + 0	1.383	Mature Height = Ht x M	

FIGURE 10.4 Paley height multipliers. Charts provide a simple method of predicting adult height for boys and girls.

to viral infections and chemical dependency such as alcohol (10). A failure of the scapular to descend from its cervical region overlying the first through fifth ribs results in *Sprengel's deformity* (6,13–16). Children often present with a shortened neckline (Figure 10.6). Lack of normal scapulothoracic motion and malpositioning of the glenoid causes limited forward flexion and abduction of the shoulder. An *omovertebral bar* is present in up to 50% of cases (17). The bar connects between the superior medial angle of the scapula and the cervical spine, and consists of fibrous cartilaginous tissue or bone. It is not uncommon to see other abnormal regional anatomy and syndromes that need to be screened for carefully, including scoliosis, spina bifida, rib anomalies, and Klippel–Feil syndrome (18,19). Renal and pulmonary disorders can also be present, and a renal ultrasound, if not already completed, is indicated. The condition can be bilateral in up to 30% of cases.

Congenital dislocation of the radial head unaccompanied by other congenital abnormalities of the elbow or forearm is rare (5). Congenital *radioulnar synostosis* is also a rare condition caused by failure of the radius and ulna to separate, usually proximal (Figure 10.7). The forearm is usually left in significant pronation with the condition bilateral 80% of the time (20). This condition is also associated with multiple other syndromes, which need to be carefully screened for (10). Children present for evaluation depending upon the degree of functional deficit. Radiographs can be helpful when ossification is present. MRI scans of the proximal radius and ulna can reveal more of a cartilaginous synostosis or a fibrous tether that has not ossified. Children with radioulnar synostosis without functional limitation should be observed. Success with *surgery* to resect the synostosis is often limited with minimal functional gain (5). Rotational osteotomies for pronation deformities greater than 45 degrees can be helpful. Postoperative compartment syndrome of the forearm needs to be watched for (21,22).

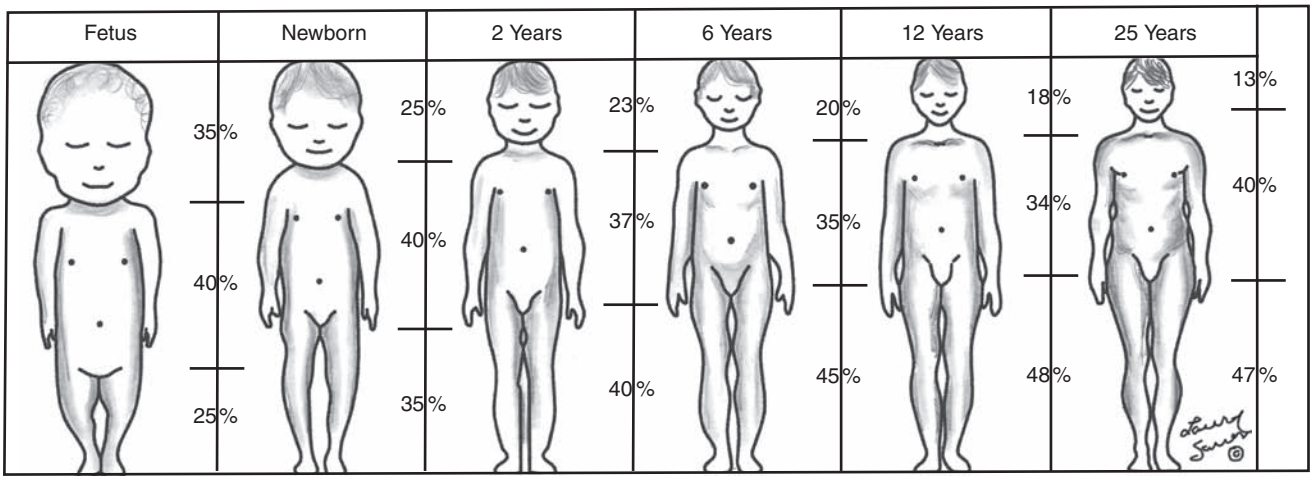

FIGURE 10.5 Proportions of the body as they change during growth. (Source: Lowrey GH. Growth and Development of Children. 6th ed. Chicago, IL: MYB; 1973.)

In about 5% of humans, there are minor variations in the number or proportions of vertebra (23). Osseous anomalies are felt to account for up to 6% of children who present with signs of torticollis. Individuals with cervical fusion are generally apparent on plain cervical radiographs, including flexion and extension views. The *Klippel–Feil syndrome*, sometimes called *brevicollis*, is characterized by short neck, low hairline, and restricted neck movement. Consisting of congenital fusions of the cervical vertebra, its incidence is approximately 0.7% (5,11). Failure of segmentation in the cervical spine most often characterizes the Klippel–Feil syndrome. Patients with Klippel–Feil syndrome or related conditions should have a renal ultrasound and cardiac evaluation (echocardiogram). *Contact sports* are *contraindicated*, as are similar, more aggressive activities.

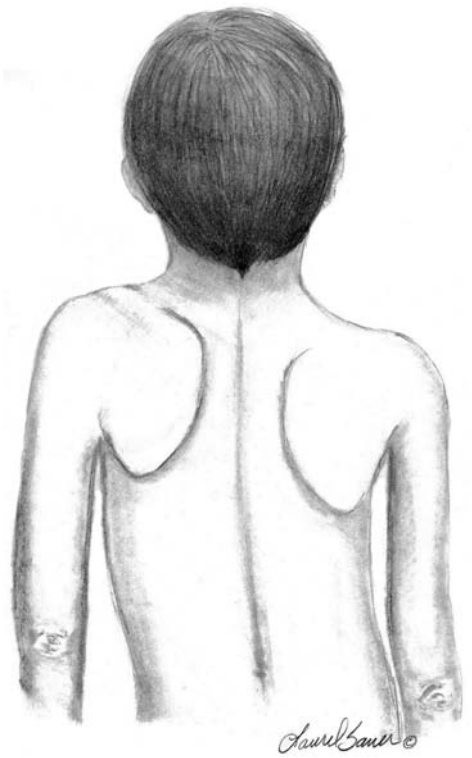

FIGURE 10.6 Sprengel's deformity.

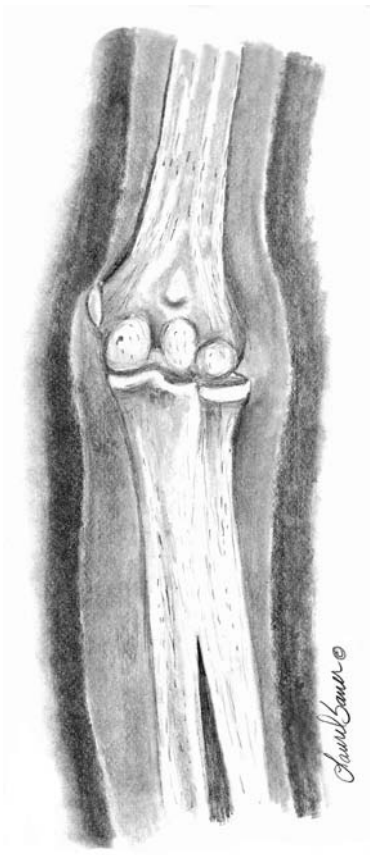

FIGURE 10.7 Radial ulnar synostosis.

Intraspinal anomalies need to be considered, especially in the presence of hairy patches, dimples, nevi tumors, or asymmetric or absent abdominal reflexes. In children with *Down syndrome*, atlantoaxial instability may be identified in up to 13% (11), but only 1% to 2% has symptomatic instability that requires surgery. X-ray examination of the cervical spine in children with Down syndrome is recommended at about the age of 3 years and before such children enter competitive sports such as the Special Olympics. Repeat x-rays are obtained after the cervical spine has been completely formed, at around the age of 8 years and every decade thereafter across the life span, as recommended by the American Association of Down Syndrome. The *atlantodens interval (ADI)* should be no greater than 4 mm in children 7 years of age and younger and no greater than 3 mm for children 8 years and older (5,8). ADI up to 5 mm has been accepted in the more traditional sense (24).

Clubfoot, talipes equinovarus, is a common term used to describe several kinds of ankle or foot deformities present at birth. The foot is generally in equinus, with forefoot and hindfoot varus and severe adduction (Figure 10.8). As the most common birth defect, it carries an incidence ranging from 1:250 to 1:1,000 live births, depending on the population (8). The condition is one of the most treatable of birth defects, often leading to normal or near-normal athletic activities later in life (5). *Multifactorial genetic inheritance*, along with poorly understood environmental factors, may explain the bulk of etiology. Some clubfoot disorders are transient or apparent in nature and result simply from intrauterine crowding. Other conditions may occur in association with myelodysplasia, arthrogryposis, and particularly hip dislocation. Prenatal ultrasound can be effective in diagnosing intrauterine clubfoot, with no false-negative prediction and a true-positive predictor rate of 83% (8). Recent treatment has focused primarily on the *Ponseti technique* (25,26,27,28). The range of motion (ROM) should be maintained by passive exercise and therapeutic play, particularly into dorsiflexion and eversion. Persistent deformity into adulthood can result in unstable ankles, lateral sprains, and difficulty with weight-bearing and other gross mobility tasks.

Metatarsus adductus can be seen in up to 12% of full-term births (8). Intrauterine crowding or positioning may be causative. Flexibility can be determined by fixing the hindfoot in a neutral position and gently manipulating the mid-foot and forefoot to a more lateral position. Internal tibial torsion may be associated, making the thigh-foot angle worse. Serial casting may be helpful in children under 1 year of age. Careful attention should be given not to place the hindfoot in valgus or create a skew foot deformity. Surgery is rarely indicated, but can be done in the more rigid persistent deformities after the age of 5. Various forms of posterior medial release are available (5).

Flat feet or pes planus may be flexible or rigid (5). Flexible pes planus is usually asymptomatic, at least in the early years, and is the most common type found in children. Inexpensive scaphoid pads or medial inserts may help to create more plantigrade weight-bearing in the child, but they do not correct the deformity. Extreme cases, such as in children with hypotonia, may require surgery after the age of 5 years in the form of a calcaneal lengthening once the bony cortices are more solid. Untreated progression may occur with compensatory hallux valgus, planovalgus, and secondary bunion and toe deformities. Pes planovalgus is associated with more active or shortened peroneal musculature, progressing over time, with the development of pain, particularly in later years. Rigid pes planus is a congenital deformity associated with other anomalies in 50% of cases (29). It is caused by failure of the tarsal bones to separate leaving a bony cartilaginous or fibrous bridge or coalition between two or more tarsal bones (30,31). *Talocalcaneal coalitions* tend to become symptomatic earlier, between 8 and 12 years, whereas *calcaneonavicular coalitions* are more likely to be symptomatic between 12 and 16 years. Symptoms are insidious with occasional acute arch, ankle, and mid-foot pain. The hindfoot often does not align in its normal varus position on tiptoe maneuvers (5). Patients are predisposed to ankle sprain secondary to the limited subtalar motion, and stress to the subtalar and transverse tarsal joints frequently causes pain. *Computed tomography* (CT) scans are diagnostic, and

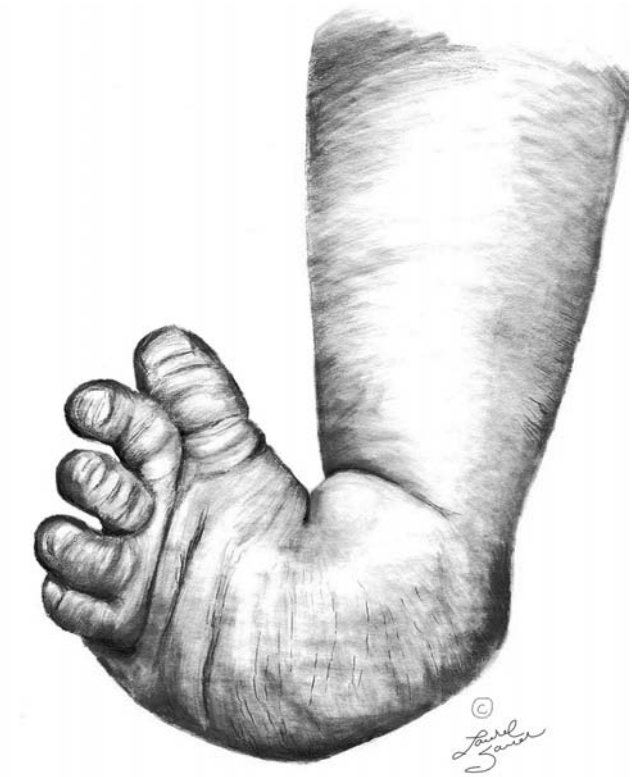

FIGURE 10.8 Clubfoot deformity. Associated forefoot supination, deep medial crease, and equinovarus of the hindfoot.

initial treatment is conservative with short-leg casting or molded orthosis and rest. If conservative care fails, surgical intervention is usually necessary. With all symptomatic pes planus, accessory navicular bones need to be considered (8). Rigid cavus feet may be associated with metatarsalgia, clawing, and intrinsic muscle atrophy. With a cavus foot, stresses are increased across the joints, along with pressures on bony prominences, muscle strength being required to maintain posture. The result is pain, fatigue, and instability. The *cavus foot* may be caused by an underlying neurologic condition such as Charcot-Marie-Tooth disease, spinal dysraphism, Friedreich's ataxia, or spinal tumor. Custom-molded inserts or orthosis may be helpful in providing arch support and decreased pain by relieving pressure off bony prominences and providing a shock-absorber effect. Cavus feet can often run in families, making family history critical. Clinical exam for flexibility with localization of the deformity to the forefoot or hindfoot should be completed. The *Coleman block test* for determination of hindfoot flexibility can be critical, particularly for any surgical repair in the more rigid and symptomatic deformity (32). Plantar fascia release is standard for all cavus foot procedures (33,34).

Congenital vertical talus is exceedingly rare (5). The navicular bone is dislocated dorsolaterally on the head of the talus (Figure 10.9). It is commonly associated with neuromuscular and genetic disorders, including trisomy 13, 14, 15, and 18 (35). Clinical features include a rigid convex plantar surface (rocker bottom) with hindfoot equinus and hypoplastic laterally deviated forefoot. Casting can initially have some benefit for contracted dorsolateral soft tissues, but only as a prelude to surgical intervention. A single-stage procedure is generally the consensus (5) and can involve talectomy, naviculectomy, subtalar arthrodesis, and triple arthrodesis.

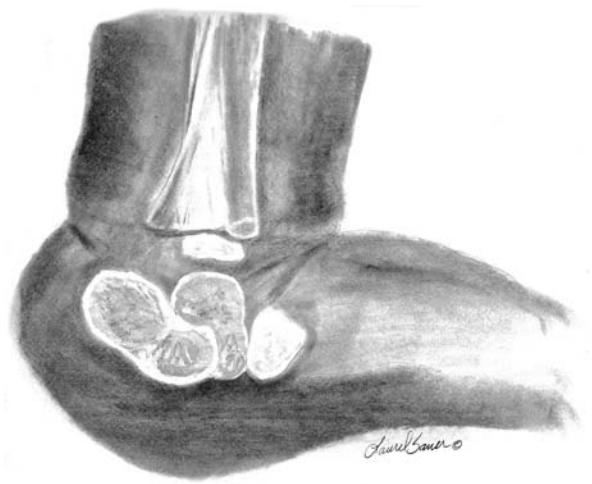

FIGURE 10.9 Vertical talus.

Arthrogryposis multiplex congenita refers to a symptom complex characterized by multiple joint contractures that are present at birth. The clinical literature has delineated as many as 150 entities under this term (8,36). The incidence of arthrogryposis as a whole is approximately 1 per 3,000 live births. *Amyoplasia* (which literally means no muscle growth) affecting all four limbs is less common, at approximately 1 in 10,000 live births (8). There are many different ways to classify the arthrogrypotic conditions (5,37). A simple way is to divide the contracture syndromes into three different groups (5). Group number 1 involves arthrogryposis multiplex congenita, Larsen syndrome, and more or less total body involvement.

Larsen syndrome is a rare condition involving multiple congenital dislocations of large joints, a flat facies, and significant ligamentous laxity (38,39). Patients commonly have abnormal cervical spine segmentation with instability and can be associated with myelopathy. Group number 2 would include the distal arthrogryposis predominantly involving hands and feet.

Distal arthrogryposis type II involves the presence of facial findings, whereas type I does not. *Freeman–Sheldon syndrome* is an example of distal arthrogryposis type II, with a characteristic "whistling face" appearance (40,41). Group number 3 involves the *pterygium syndromes*. Pterygium comes from the Greek word meaning "little wing." Pterygiums can be isolated or multiple. Multiple pterygium syndrome is characterized by webbing across every flexion crease in the extremities, most prominently across the popliteal space, elbow, and axilla (42). Popliteal pterygium syndrome has features involving the face, genitals, and knees (43). A popliteal web is usually present bilaterally running from the ischium to calcaneus, resulting in severe knee flexion deformities (Figure 10.10). The diagnosis of arthrogryposis can be suspected with prenatal ultrasound. Absence of fetal movements of distal or proximal joints in combination with polyhydramnios is suggestive (44). The *birthing process* can be complicated by joint contractures, with neonatal fractures resulting. Perinatal fractures are common and believed to be secondary to hypotonia and rigid joints (45). Therapy should not be initiated in a newborn until such fractures are ruled out (46). Children who survive infantile arthrogryposis often have upper and lower extremity involvement in typical patterns. *Common deformities* of the upper extremities include adduction; internal rotation contractures of the shoulders; fixed flexion or extension contractures of the elbows, either wrist flexion and ulnar deviation or extension and radial deviation; and thumb-in-palm deformities. In the lower extremities, flexion, abduction, and external hip rotation contractures with unilateral or bilateral dislocations are noted. Bilateral dislocations of the hip are more often left alone, whereas unilateral dislocations, because of scoliosis risk, are more often surgically treated (5). Fixed extension or flexion contractures of the knees are also seen along with severe rigid bilateral clubfeet. In the most severe rigid clubfeet, not

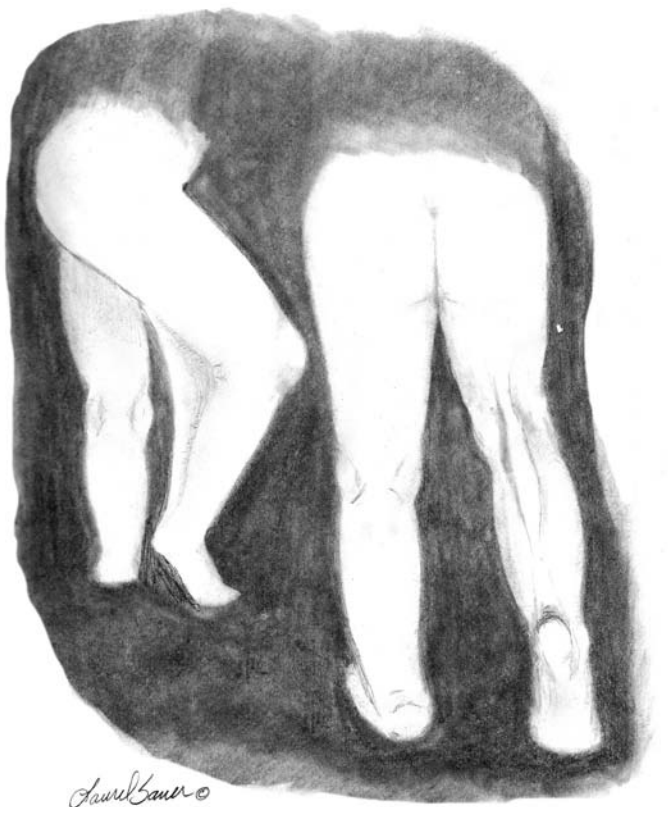

FIGURE 10.10 Isolated popliteal pterygium.

correctable with casting and conservative care, a *talectomy* may be necessary or talar enucleation in association with the posterior medial releases. Extension wedge osteotomies of the distal femur may be necessary to correct flexion contractures of the knee. There is always a well-recognized risk of neurovascular damage, with operative correction of knee flexion contractures needing careful consideration to avoid *overstretching* of the *neurovascular bundle*. Shortening osteotomies completed at the same time as the extension wedge osteotomy may minimize these risks. In the absence of degenerative neurologic conditions, individuals with arthrogryposis maintain their strength and ROM over time (5). *Surgical and rehabilitation goals* are generally centered on self-help skills, such as feeding, toileting, and mobility skills such as standing, walking, and transfers using assistive devices as needed. *Surgical procedures* of the upper extremity are usually delayed until the child is old enough for a more definitive functional assessment to be completed. If both elbows are involved with extension, surgery to increase flexion may be best done on only one side.

Outcomes appear better if joint surgery is completed prior to the age of 6 to avoid adaptive intra-articular changes (8). *Osteotomies for realignment* are usually performed closer to skeletal maturity. Early mobility and avoidance of prolonged casting may result in improved ROM and function postsurgery. *Most individuals do not have intellectual impairment* or sensory deficits. The children often have a keen natural ability to learn substitution techniques. A strong association between initial feeding difficulties and subsequent language development is known, which should not be misidentified as intellectual deficiency (47).

BRACHIAL PLEXUS PALSY

Birth brachial plexus injury occurs in between 1 and 2 per 1,000 live births in the United States. Babies with increased birth weights, multiparous mothers, and shoulder dystocia are at the highest risk for brachial plexus palsy (48,49). The most widely described mechanism of action for this is lateral stretch, which is logical secondary to the location of the brachial plexus, the high correlation with shoulder dystocia, and the positioning of the mother and infant (49). It has been described that between 50% and 95% of these infants will recover spontaneously. The overarching goal of treatment of brachial plexus injuries is maximizing arm and hand function. Goals are normalization of limb function, with optimization of nerve regeneration and mechanical increase of elbow flexion and shoulder stabilization. This can be achieved through aggressive rehabilitation and surgical intervention (48).

For any nerve that is injured, classification makes evaluation and comparison clearer. The Seddon Classification of Nerve Injury is commonly used. Neurapraxia occurs with no lasting anatomic changes, with fibers preserved. This is exemplified by a football "stinger" injury. Complete resolution is expected. In axonotmesis, there is an interruption of neural continuity to some degree. There is an extremely variable level of deficit that is difficult to evaluate and predict the degree of recovery. Neurotmesis is the most severe injury, with total disruption of the elements of the nerve, and this will not independently recover. If it is preganglionic, or proximal to the dorsal root ganglion, it is called an avulsion. If it is postganglionic, or distal to the dorsal root ganglion, it is called a rupture (50). Both of these require surgical intervention for recovery.

There are also descriptors for the levels of brachial plexus palsy. Injury at C5 and C6 is called Erb's palsy, sometimes called Erb–Duchenne palsy. This is the most common level of involvement, present in approximately three-fourths of those with birth brachial plexus palsy (BBPP). Involvement of C8 and T1 is Klumpke's palsy. It is debated whether Klumpke's can occur in a birth brachial plexus injury, though it definitely occurs in other types of brachial plexus injury. The reason for this question is whether it is anatomically possible to have a C8 and T1 lesion alone without the involvement of C5 and C7. It appears that if there is an anatomic variation—for example, a rib, tendon, bony, or another anomaly that leads to the compromise of C8 and T1—this can occur in

a birth brachial plexus injury. Otherwise, it appears that it cannot. Therefore, if a child presents with a C8 and T1 birth brachial plexus injury, it may be from anatomic anomaly, but there are two other options to consider. Most likely, it was initially a complete brachial plexus involvement but there was quick recovery of C5 and C7. This is likely, since the upper cervical root levels are relatively protected anatomically, so C8 and T1 may end up with the most severe injury. It is also possible that a spinal cord injury has been mistaken for brachial plexus palsy. All of these are important to consider during evaluation. There also may be complete brachial plexus palsy, including C5 and T1, with total motor and sensory loss. There also can be a variety of levels involved between upper plexus and total plexus palsy.

EVALUATION

Evaluation of patients with brachial plexus palsy includes clinical findings, electrodiagnosis, and MRI. There is debate about which of these is the most effective. MRI is expensive and requires sedation to perform on infants. It has been found to correlate with surgical findings 70% of the time, electromyography (EMG) 87% of the time, and clinical findings 60% of the time. The correlation was highest when all three of these were combined. MRI was effective only in those with C5, C6 root involvement (51).

Clinical exam consists of a history and physical examination. The history includes the birth number of the child, the birth weight, and presence of maternal diabetes during the pregnancy, along with the size of previous infants and the birth size of the parents. The motor and sensory findings at birth, along with any change up to the time of evaluation, are important. The use of vacuum or forceps may be indicative of any difficulty with delivery. The most common association is shoulder dystocia. Other useful information is whether there were signs of bruising or other injuries, or whether there was involvement of the contralateral arm or the legs at delivery.

Physical examination begins with visualization of the arm to include the size and bulk. A cool temperature may be noted in those with severe involvement. Sensory evaluation is critical to determine the extent and levels of involvement. Muscle stretch reflexes will be decreased or absent in the distribution of a brachial plexus injury.

The primitive reflexes are also important. Since the upper plexus has more frequent involvement, the Moro reflex, which shows shoulder abduction and elbow flexion, is valuable in assessing those active movements. Torticollis is frequently seen, and usually this is with the face turned away from the involved arm. ROM is an important part of the evaluation since contractures are commonly seen in shoulder adduction and internal rotation, wrist flexion, forearm pronation, and even at the elbow into flexion commonly in later months and years.

A key goal of the electrodiagnostic evaluation is to find subclinical nerve and muscle responses. The study must be specific depending on the clinical deficits noted, with studies performed that are pertinent to each individual's examination. Sensory nerve conduction studies, motor nerve conduction studies, and EMG are performed. Diagnostic evaluation should include nontraditional nerve conduction studies, and frequently not the classic median and ulnar nerves, due to frequent involvement of only the upper brachial plexus. Axillary, musculocutaneous, and radial nerves are among those useful for electrodiagnostic study. Sensory nerve action potentials (SNAPs) are important, as these are most sensitive to axonal loss (52). The presence of SNAP responses in an insensate area is indicative of a preganglionic lesion, due to the location of the sensory cell bodies in the dorsal root ganglion. EMG may show activation of motor unit potentials in muscles with no clinical motor activity. Electromyographic evaluation is reported as being of some benefit, but it underestimates the severity of lesions (53). It has been recommended to be performed early in the first few days, then with a repeat evaluation after several months to more accurately identify cases where there is reinnervation occurring and therefore providing earlier determination of the need for surgical intervention (54). EMG at 1 month has been shown to have the best prediction for recovery in babies (55,56).

Plain x-rays are useful in some clinical circumstances. Some abnormalities may mimic a brachial plexus palsy, including a fracture of the clavicle or humerus. Osteomyelitis may also mimic this, and has actually been reported as inciting temporary brachial plexus palsy (57). Neurofibromatosis or other tumors may also damage the brachial plexus.

TREATMENT

Education is initiated when a family is first seen. Therapy should be started as soon as possible after diagnosis. Positioning instruction begins immediately, and ROM exercises are generally initiated after 2 weeks. The wait is due to the fact that there is commonly noted pain with changing position of the shoulder for bathing or dressing in the first 2 weeks, so it appears that there is some early, short-term tenderness after the initial brachial plexus injury. It is also important to position the arm so that the baby will have maximal awareness of it. One way to accomplish this is with the use of a wrist rattle on the affected arm so that the baby's attention can be drawn to that arm by sound or vision, because the weakness of that arm usually limits it from being moved in front of the face spontaneously. It is also recommended to have the family replicate movements with the affected arm that the baby spontaneously does with the unaffected arm, such as bringing the hand to the mouth. It is important that the family be taught to

perform the exercise program several times a day. It is also important not to have such aggressive ROM in shoulder abduction or forearm supination that there is dislocation of the humeral head or radial head, respectively. Splinting is also commonly done by occupational therapy or physical therapy. Initially, there is frequently wrist drop, so splints may be made to provide optimal position of the wrist and fingers. Later on frequently there is an elbow contracture, so splinting is done to minimize that. Therapists also may do taping to help promote optimal positioning of the arm, particularly at the shoulder.

Electrical stimulation is sometimes done for brachial plexus palsy, though this is frequently not tolerated at a very young age. Over time it does become accepted by many young children. Most commonly, it is performed with surface electrodes to increase muscle bulk by use of sufficient stimulation to get a local muscle twitch for approximately 20 minutes twice daily. It has been shown that continuous electrical stimulation to denervated muscles with implantable electrodes will lead to improved muscle outcome after nerve regeneration (58). Implantable electrodes have not been widely utilized and are not currently available on the U.S. market.

It has been proposed that the adverse effects of prolonged denervation leave intramuscular axons deteriorated to such low numbers such that even with successful nerve regeneration, it is impossible to reinnervate enough muscle fibers for sufficient force (59). There are also proposals that low doses of brain-derived neurotrophic factor (BDNF) may protect against this decrease in those who have late nerve grafts, though high doses are inhibitory (60).

COMPLICATIONS

It is important to monitor for secondary complications. These commonly include muscle atrophy and joint contractures. Serial casting has been shown to decrease elbow flexion contractures (61). The affected arm frequently is shorter and has decreased circumference compared to the contralateral side. Joints may become dislocated, most commonly at the shoulder. Botulinum toxin injections decrease shoulder subluxation and decrease the need for surgery (62). Scapular winging is frequently seen. There may be torticollis, most commonly with the face turning away from the involved arm. General child development may be affected, including lack of awareness of the arm. Similarly, body image may be affected. There can be ulcerations from relatively minor trauma, particularly in insensate areas. Pain is infrequent after BBPP but common after brachial plexus palsy occurring later in life. Scoliosis has sometimes been linked to BBPP but two studies examining this question have found no correlation (61,63).

SURGICAL INDICATIONS

Indications for timing of brachial plexus surgery for infants have been controversial. It has been shown that a longer time for recovery leads to a worse shoulder function and that those who regain elbow flexion after 6 months of age have worse function than those who regain it between 3 and 6 months (64). Those with recovery by 3 months have normal function. Those who had microsurgery at 6 months did better than those who spontaneously recovered elbow flexion at 5 months (65). Surgical intervention is commonly recommended for those having less-than-antigravity strength in elbow flexion at 6 months of age (66). Estimates vary that from 4% to 34% of those with BBPP will require surgery for clinical improvement (67).

Later brachial plexus injuries are divided into supraclavicular and infraclavicular injuries, supraclavicular being 75% and infraclavicular 25%. Supraclavicular injuries are generally felt to be due to traction of the plexus (classically in a motorcycle crash), and these have a worse prognosis than infraclavicular injuries (68). There may be a fracture of the clavicle or cervical transverse process, and supraclavicular fossa swelling may be seen. Dorsal scapular nerve or long thoracic nerve injury may be present. Supraclavicular lesions may also be due to falls; large objects falling on a shoulder, such as a tree limb; skiing or climbing; or contact sports, including football (52). Other etiologies are backpacks that are too heavy, tumors and gunshot wounds, or lacerations or animal bites. Those who have ipsilateral Horner's syndrome and persistent pain have a worse prognosis (52).

Infraclavicular brachial plexus injuries are more commonly associated with fractures and dislocations about the shoulder or humerus, occurring more often in older adults. The posterior cord, axillary nerve, or musculocutaneous nerve are classically involved. Infraclavicular injuries are less severe and have better outcomes (69). Infraclavicular plexus injuries may also be due to falls, motor vehicle collision, or tumors (52). Gunshot wounds, stab wounds, and failed attempts at shoulder reductions may cause infraclavicular injuries as well (70). Brachial plexus palsy has been reported after axillary crutch use, anesthesia positioning (particularly with table tilt), and after bony fracture with malunion (71). For severe injuries later in life, recommendations are for surgical exploration and nerve grafting, most commonly at 3 to 4 months post-injury (70,72).

SURGERY

Surgical interventions for brachial plexus palsy are varied. There may be electrical testing, including evoked potentials, and nerve conduction studies done to assess the nerves in the operating room to be as specific as

possible with the procedures undertaken. Microsurgical repair yields results months later. Recovery is generally felt to proceed at the rate of approximately a millimeter a day or an inch a month. It is also believed that there is more nerve growth factor available in babies than older people so that both size and age have an impact in outcome. It is critical to have therapy postsurgery and to continue a faithful daily home program as well.

There are a variety of options for surgical procedures for brachial plexus injury. Neurosurgery may include neurolysis in which scar and fibrotic tissue are removed from nerve tissue. Direct nerve transfers have the advantage of quick recovery time due to short regeneration distance versus neurotization, which requires interposition of a nerve graft. The sural nerve and great auricular nerve are commonly used as donor nerve fibers for these grafts (73). More recently, end-to-side neurorrhaphy is performed for those who have some intact fibers for augmentation. The advantage of this is not requiring a sacrifice of any other nerves. Not uncommonly, synkinesis of newly innervated muscles with contraction of muscles innervated by the donor nerve may be seen, and is treated with therapy (74). Synthetic nerve conduits are now available for nerve grafting.

Some classic nerve procedures involve transfer from a functionally less important nerve to a distal denervated nerve. Common examples include taking intercostal nerves to the upper trunk or to the suprascapular nerve. Another classic surgery is the Oberlin procedure, which transfers one or several ulnar nerve fascicles to the musculocutaneous nerve as it enters the biceps muscle (75). Transfer of the spinal accessory nerve to the suprascapular nerve is also commonly used for shoulder abduction. Contralateral C7 transfers have been performed both in adults and infants for those with multiple severe avulsions. This procedure has been shown to provide adequate elbow flexion as a result, and most patients have had only temporary sensory deficits on the ipsilateral C7 side (76). This procedure clearly illustrates the point that nerve grafts are not required to have their original source but can have function coming from a variety of intact neurologic structures. This allows for greater flexibility and creativity in the surgeon performing the procedure, aiming for recovery of function.

Glenoid dysplasia with posterior shoulder subluxation is frequently a complication of children after BBPP. It was commonly thought to be the result of a slowly progressive glenohumeral deformation due to muscle imbalance and possible physeal trauma, but it was found that posterior shoulder dislocation happened at a mean age of 6 months, with rapid loss of passive external rotation. There was no correlation between the initial neurologic deficit and the presence or absence of dislocation (77).

Many musculotendinous surgical procedures are performed for children with BBPP. It has been shown that latissimus dorsi and teres major tendon transfer to the rotator cuff, along with musculotendinous lengthening, will provide improved shoulder function but no significant change in the bony position of the shoulder or humerus. This procedure does not decrease glenohumeral dysplasia (78).

With internal rotational contracture and glenohumeral joint deformity, along with significant abnormality of glenohumeral joint, a derotational osteotomy can result in improved shoulder function, along with improved internal rotation contracture (79).

Some children with BBPP have been described to have arthroscopic release of shoulder deformity alone before 3 years, and for those over 3 years of age, arthroscopic release with latissimus dorsi transfer. They all show improved shoulder position, but they do have loss of internal rotation. Some of the children under 3 years of age do have a recurrence and require a second procedure with a latissimus dorsi transfer (80).

In adults, performing a glenohumeral arthrodesis, in patients with upper plexus palsy with functional distal arm, as well as in those with total plexus palsy, has been shown to increase functional capabilities. The strength of the pectoralis major is a significant prognostic factor for outcome (81).

Performing wrist arthrodesis in adults with brachial plexus injury is done for improved function as well as pain relief. There will be limitations after having this procedure, and potential patients need to have full information in order to know what to expect prior to the procedure. There also remains some controversy regarding the ideal position to place the hand, which is generally placed in slight wrist extension and ulnar deviation in order to have the most powerful grip (71,82). A dramatic surgical procedure sometimes performed for children and adults with brachial plexus palsy is a free muscle transfer, most commonly performed with the gracilis muscle. The muscle is transferred with its vascular and nerve supply and attached to these in the arm. This procedure has been described as having reliable results for elbow flexion and wrist extension (71).

PAIN

Pain has not been reported as a severe problem in birth brachial plexus injury, although with one study reporting biting of the limbs in less than 5% of the cases, it is possible that this is a manifestation of pain. Self-mutilation has been reported in youngsters after a birth brachial plexus injury. A study of 280 patients with a birth brachial plexus injury found that 11 of these children had self-mutilating behavior by biting or mouthing the affected arm. The age of onset was between 11 and 21 months, and the duration of the behavior was 4 to 7 months. This was more frequent in children who underwent surgery, with 6.8% of these children, and 1.4% of children who did not have surgery. It is unclear if this is due to surgery or the

severity of the injury or a combination of these (83). It is also possible that this is a response to the unusual sensation of the recovering nerve, possibly a manifestation of what we see on examination as a Tinel's sign. It has been felt, however, that it is more likely biting with the resumption of nerve growth with sensation of tingling as there is recovery occurring, but this is not proven.

In those who have later traumatic or nontraumatic brachial plexus injuries, pain can be a significant problem. It has been described most commonly with avulsions as severe burning and crushing pain most commonly in the hand. This may develop days to months after the injury and almost always within 3 months. It is most commonly resolved within several years, but approximately 20% of those with pain have severe, long-lasting disruptive pain (84). This can be treated with transcutaneous nerve stimulation classically from C3-T2. Medications, including antidepressants and anticonvulsant agents, have been affective. Topical treatments, including topical lidocaine 5% pain patches, are sometimes useful. Nerve surgery is commonly effective in resolving pain (85,86). The author has seen children with traumatic brachial plexus injuries and severe pain complaints prior to their nerve procedure wake up postoperatively in the recovery room excited that the pain is gone. Amputation is not effective for resolving the pain (87).

REHABILITATION OF THE CHILD WITH RHEUMATIC DISEASE

Rehabilitation of the child with rheumatic disease requires an interdisciplinary approach that includes the child and family. Although most often the physiatrist is not the treating physician in rheumatologic disease, they can play a key role in the comprehensive management of these conditions, along with other members of the rehabilitation team, to maintain or restore age-appropriate function and development, prevent deformity and contractures, and help manage pain.

JUVENILE IDIOPATHIC ARTHRITIS

Juvenile idiopathic arthritis (JIA), formerly known as juvenile rheumatic arthritis (88), is the most common rheumatic disease of childhood, affecting approximately 16 to 150 in 100,000 (89). In 1995, the International League Against Rheumatism (ILAR), together with the World Health Organization, reclassified chronic childhood arthritis (90); the second revision occurred in 2001 (91). Chronic childhood arthritis is now known as JIA and is divided into the following seven subtypes: systemic arthritis, oligoarthritis, rheumatic factor (RF)-negative polyarthritis, RF-positive arthritis, psoriatic arthritis, enthesitis-related arthritis, and undifferentiated arthritis. JIA occurs in children before the age of 16 years, persists at least 6 weeks, and has had other known conditions excluded; etiology is unknown, but seems to include genetic and environmental components (89,92). In 2011, the American College of Rheumatology (ACR) published recommendations for the treatment of JIA, organized into five treatment groups; namely, arthritis of four or fewer joints, arthritis of five or more joints, active sacroiliac arthritis, systemic arthritis with active systemic features (and without active arthritis), and systemic arthritis with active arthritis (and without active systemic features) (93). In October 2013, these recommendations were further reviewed and updated, with emphasis on systemic JIA (94). Both recommendations will be discussed further in the treatment section.

Early arthritis may be manifested by swelling, warmth, and joint stiffness, typically worse at the beginning of the day then improving with activity. Symptoms usually fluctuate; uncontrolled inflammation leads to joint damage. Younger children rarely complain of joint pain, but may instead become irritable, stop walking or using an extremity, or regress in their behavior (95). Other symptoms include decreased appetite, malaise, inactivity, morning stiffness, nighttime joint pains, and failure to thrive (95). Enuresis may occur in a recently toilet-trained child (96). Later disease presents with reduced ROM, contractures, overgrowth or undergrowth of affected limbs, and resultant disability.

A characteristic feature of chronic arthritis in children is the effect the disease has on bone and joint development (97,98). Local growth disturbances at inflammation sites can lead to overgrowth (secondary to possible inflammatory-mediated increased vascularization and growth factor release) or undergrowth (secondary to growth center damage or premature fusion of epiphyseal plates). Irregular traction on growing structures secondary to muscle spasms and periarticular fibrosis can also cause aberrant growth (97,98). Micrognathia, leg-length inequalities, and developmental hip anomalies are all possible results from these processes. Steroids can also contribute to severe growth effects, as well as osteoporosis (99).

The differential diagnosis of JIA is large (Table 10.1 provides a full differential diagnosis). The assumption that JIA will universally resolve by adulthood is incorrect (100). Radiological joint damage occurs in children with systemic arthritis and polyarticular arthritis within 2 years, and in oligoarthritis within 5 years (101,102). Despite long-term persistence of disease activity in JIA, much improvement in functional outcomes has been made in the past decade (103,104). Indicators of poor outcome include greater severity or extension of arthritis at onset, symmetrical disease, early wrist or hip involvement, presence of RF, persistent active disease, and early radiographic changes (105). In the 2011 ACR recommendations, features of poor prognosis are further specified by treatment group. Features of poor prognosis for three

TABLE 10.1 DIFFERENTIAL DIAGNOSIS OF JUVENILE IDIOPATHIC ARTHRITIS

Pediatric Rheumatic Diseases
Systemic lupus erythematosus
Juvenile dermatomyositis
Scleroderma
  Localized (linear, morphea, etc.)
  Generalized (systemic sclerosis, CREST, etc.)
Mixed connective tissue disease (overlap syndrome)
Juvenile ankylosing spondylitis
Acute rheumatic fever
Reactive or postinfectious arthritis
Vasculitis
  Kawasaki disease
  Henoch–Schoenlein purpura
  Behçet's disease
  Wegener granulomatosis
  Polyarteritis nodosa
Autoinflammatory disorders
  Tumor necrosis factor receptor-alpha associated periodic syndromes
  Familial cold autoinflammatory syndrome
  Neonatal-onset multisystem inflammatory disease
  Chronic infantile neurologic, cutaneous, and articular syndrome
Periodic fever, adenitis, pharyngitis, and aphthous ulcer syndrome
Fibromyalgia
Complex regional pain syndrome, type II

Infectious Diseases
Bacterial arthritis
Viral arthritis
Fungal arthritis
Osteomyelitis
Fasciitis/myositis

Neoplastic Diseases
Leukemia
Lymphoma
Neuroblastoma
Primary bone neoplasms

Hematologic Diseases
Hemophilia
Sickle cell disease

Noninflammatory Disorders
Trauma
Overuse syndromes
Osteonecrosis syndromes
Avascular necrosis syndromes
Slipped capital femoral epiphysis
Toxic synovitis of the hip
Patellofemoral dysfunction (chondromalacia patellae)
Diskitis

Miscellaneous Disorders
Inflammatory bowel disease
Sarcoidosis
Collagen disorders
Chronic recurrent multifocal osteomyelitis
Growing pains
Hypermobility syndromes
Foreign-body arthritis
Psychogenic arthralgias/arthritis (conversion reactions)

Abbreviation: CREST, Calcinosis, Raynaud phenomenon, Esophageal dysmotility, Sclerodactyly, and Telangiectasia.

of the five categories, arthritis of four or fewer joints, arthritis in five or more joints, and systemic arthritis with active arthritis (and without systemic features), include arthritis of the hip or cervical spine. Four of the five treatment groups (all except for systemic arthritis with active systemic features [and without active arthritis]) show poor prognosis with radiographic damage (erosions or joint space narrowing by radiograph) (93).

CLINICAL FEATURES OF JIA SUBTYPES

Systemic JIA

Systemic-onset JIA presents with many extra-articular features and represents 10% to 20% of all JIA (92). Diagnosis requires arthritis in more than one joint for at least 6 weeks' duration in a child aged below 16 years accompanied or preceded by quotidian fever (spikes >39 degree Celsius once a day with return to normal between peaks) of 3 days' duration, plus one or more of the following: evanescent salmon-colored rash, generalized lymphadenopathy, hepatomegaly, splenomegaly, or serositis (91,94).

The goal of treatment for systemic JIA is similar to other categories of JIA, and focuses on the prompt control of active inflammation and symptoms, and the prevention of disease- and/or treatment- related morbidities such as growth disturbances, joint damage, and functional limitations (106). About 5% to 8% of children with systemic JIA develop a life-threatening complication known as Macrophage Activation Syndrome (107) with persistent fever, lymphadenopathy, and splenomegaly, and there is profound depression in one or more of the blood cell lines (often initially platelets) with raised liver function enzymes and clotting abnormalities. Definitive bone marrow examination shows numerous well-differentiated macrophages actively phagocytizing hematopoietic elements (108).

In one-half of children with systemic JIA, the course follows a relapsing–remitting course, with arthritis accompanying febrile episodes, then remission once systemic features are controlled. Long-term outlook for these children is usually good. In the other half, the disease is unremitting, with resultant severe joint destruction, and is probably the most severe JIA subtype (89,109). Poor prognostic signs include the continued presence of systemic features and a platelet count exceeding 600,000/mm^3,

6 months after onset (95). At least one-third of children will develop severe arthritis (110).

Oligoarthritis

Oligoarthritis is classified into two subtypes: persistent (affecting not more than four joints throughout the disease course) and extended (affecting more than four joints after the first 6 months of disease). Characteristically, there is an early onset before 6 years of age of an asymmetric arthritis, usually in the lower limbs, and predominantly in females. Antinuclear antibodies (ANAs) are detected in substantial titers in about 70% to 80%, and they represent a risk factor for iridocyclitis. Children with the oligoarthritis subtype generally have the best outcome (89); however, sight-threatening, clinically silent uveitis develops in the first 4 years from diagnosis. Regular ophthalmology follow-up is essential (111).

Polyarthritis

Polyarthritis must affect five or more joints in the first 6 months of the disease. RF-positive polyarthritis mainly affects adolescent girls, with a symmetrical pattern, and is the same as adult RF-positive disease (97). By 5 years from onset, severe deforming arthritis is generally present (98). RF-negative polyarthritis is a more heterogeneous group with more variable outcome. Approximately 20% to 40% of those affected are ANA-positive, and chronic uveitis is found in 5% to 20% (97); it is believed by some authors that this entity represents a later stage of early-onset oligoarthritis (112). Future versions of the ILAR classification of JIA may explore this more fully.

Psoriatic Arthritis

Psoriatic arthritis accounts for about 5% of JIA and requires the simultaneous presence of arthritis and the typical psoriatic rash, or if the rash is absent, arthritis plus two of the following: positive family history of psoriasis in a first-degree relative, dactylitis, and nail pitting. Psoriatic disease in children before the age of 5 years appears to be more difficult to control than in an older subset of children, with a median of 9.5 years (92).

Enthesitis-Related Arthritis

Enthesitis-related arthritis affects males after the age of 6 years (97,98) and most children are human leukocyte antigen (HLA)-B27-positive. The most common sites of enthesitis are the calcaneal insertion of the Achilles tendon, plantar fascia, and tarsal area. Arthritis commonly affects the joints of the lower extremities. Unlike other JIA subsets, hip involvement is common at disease presentation.

These children may progress to fulfill criteria for ankylosing spondylitis, reactive arthritis, or arthritis associated with inflammatory bowel disease. Uveitis is also a clinical problem in this subset, but it is usually sudden in onset, symptomatic, and more unilateral than in children with other JIA subsets (92).

Juvenile ankylosing spondylitis, not considered part of the JIA subclassification; mainly affects adolescent boys; is strongly associated with HLA-B27; and manifests as an asymmetric, often episodic, oligoarthritis in the lower limbs. Later on, bilateral sacroiliac joints become involved, and progression of the disease can lead to the characteristic "bamboo" spine on radiographic images secondary to ankylosis of spinal joints. In children, peripheral arthritis and enthesitis present early in the disease, but sacroiliac and spine joints are not involved until many years later (113). Rehabilitation involves maintaining spinal ROM through extension exercises, strengthening hip extensors and quadriceps muscles, custom shoe inserts to relieve pain, and deep breathing exercises to maximize chest expansion. Because of the chronic course of the disease, the child and parents should not restrict age-appropriate social and recreational activities (113).

Inflammatory bowel associated arthritis occurs in approximately 10% to 20% of children with ulcerative colitis and Crohn's disease. The arthritis usually affects a few joints and may be associated with spondylitis; erythema nodosum and growth failure may occur.

Undifferentiated Arthritis

This subset is not a separate entity, but is more of a catch-all category for those children who do not satisfy inclusion criteria for any category, or who meet criteria in more than one category.

REHABILITATION OF THE CHILD WITH JIA

Goals of treatment include controlling symptoms, preventing joint damage, achieving normal growth and development, and maintaining function and normal activity levels.

Treatment goals may vary during maintenance and acute flare-ups of the disease.

Resting a joint may be necessary during an acute flare-up to prevent aggravation of the disease process; activities that affect or excessively stress joints should be discouraged during acute flare-ups. Resting a joint may also be useful during the maintenance phase for joint protection. Rest periods may be necessary to reduce fatigue; resting in the prone position will help reduce hip and knee flexion contractures.

Splinting is used during a flare-up to provide alignment during a rest period. Functional splints may be used during flare-ups and maintenance phases if they provide

joint relief and allow functional activities without stressing inflamed joints. Splinting can be used during the maintenance phase to promote local joint rest, support weakened structures, and assist function. To prevent flexion contractures, the upper extremity is splinted in a functional position as follows: wrist 15 to 20 degrees of extension, some finger flexion, 25 degrees at the metacarpophalangeal (MCP) joint, and 5 to 10 degrees at the PIP joint, with the thumb in opposition. Ring splints can be used for finger deformities. Knee immobilizers may be used to maintain knee extension at night; rotate on alternate legs for better compliance. Dynamic splints or serial casts can increase ROM. Foot orthoses can promote arch support and reduce pain in weight-bearing.

Gentle ROM with passive extension greater than flexion two to three times a day is used to preserve joint ROM. Incorporating pain medication, progressive muscle relaxation, breathing exercises, biofeedback, massage, or doing the exercises in a nice, warm tub can greatly facilitate ROM exercises. Gentle ROM exercises should be done as tolerated during acute flare-ups to prevent flexion contractures.

Heat is an excellent modality in the maintenance phase to decrease stiffness, increase tissue elasticity, and decrease pain and muscle spasm. Hydrotherapy with temperatures 90 to 100 degrees Fahrenheit, fluidotherapy, paraffin, or moist heat can be used. Most children prefer heat to cold. Taking a hot bath or shower, sleeping in a sleeping bag, or using a hot pack (along with ROM exercises) may help relieve morning stiffness. Caution must be exercised in insensate areas to avoid burns. Ultrasound is contraindicated in children with open growth plates. Heat should not be used during an acute flare-up, as it increases the inflammatory response and causes further joint destruction.

Cold can be used during an acute flare-up for pain relief and to decrease swelling. It may also be beneficial during the maintenance phase for the same reasons. Cold should not be used over insensate areas or in those with Raynaud's phenomenon.

Adaptive strengthening exercises can be incorporated into play and recreational activities. Some examples include throwing a ball (strengthens elbow and shoulder), riding a bike (promotes knee and hip extension), and swimming (decreases weight-bearing on painful joints). Incorporating general aerobic conditioning is also important and may include activities such as swimming, dancing, noncontact karate, and tai chi. Isometric strengthening exercises are fine during an acute flare-up, but vigorous exercise should be avoided until the acute process is over. Hydrotherapy can be combined with land-based physiotherapy in treating JIA (114).

Adaptive equipment can be used for joint protection, rest, and to minimize further joint destruction during both phases. Examples include adaptive utensils, adaptive pens and computer access, table and desk modifications (to prevent excessive trunk and neck flexion), zipper pulls, dressing sticks, long-handled brushes, elastic waistbands, Velcro closures, and larger buttons. Children should actively participate in functional activities of daily living (ADLs) training in order to choose acceptable devices and improve their use.

Activity and ambulation should be encouraged as much as possible. A posterior walker for upright posture (with decreased flexion) and a standing program may be useful for functional mobility training if wheelchair use cannot be avoided. In children with JIA, custom-made semirigid foot orthotics with shock-absorbing posts have been found to significantly improve pain, ambulation speed, self-rated activity, and functional ability levels compared to prefabricated off-the-shelf shoe inserts or supportive athletic shoes alone (115).

A presurgical joint rehabilitation program aims to strengthen the muscles needed for mobility in the postoperative period, train for future ambulation aids, and identify other joint involvement that may affect the rehabilitation process. Postsurgical rehabilitation fulfills those goals set in the presurgical rehabilitation program. Ambulation aids such as the platform walker may be used to better distribute weight-bearing pressure on affected upper extremity joints after knee or hip surgery. In children, post hip prosthesis, the acetabular component should be checked for loosening (as opposed to the femoral component in adults), especially if children are active.

Growth retardation can occur during periods of active disease; it may also be compounded by corticosteroid use. Maximize growth by promoting optimal nutrition. Children with JIA should eat a balanced diet with supplemental multivitamins, calcium, vitamin D, and sunshine secondary to the high risk of osteopenia. Plenty of (nonimpact) activity again should be encouraged.

Counseling for both the child with JIA and his or her family should be provided to maximize psychosocial and emotional well-being. Treatment goals also include addressing family, school, and vocation. Assisting in the preparation of a 504 plan for school accommodations enables a child with joint disease opportunity for more complete participation in his or her school life and academic career. Summer camps are a practical way of addressing peer support within adolescent rheumatology services; positive effects include increased control, self-esteem, physical fitness, independence from parents, self-management of health care, and an opportunity to meet others with a similar condition (116).

SPECIFIC JOINTS IN JIA

Cervical Spine

Cervical spine involvement occurs more often in children with JIA than adults. Restriction of ROM, pain, and muscle spasms, which may present as torticollis, may be seen.

A soft cervical collar to serve as a reminder for proper alignment and provide warmth may be helpful in acute pain with muscle spasm. Minimizing time in flexion is important. If the transverse ligament becomes weakened, atlantoaxial subluxation can occur. If subluxation occurs, a firm cervical collar should be worn during automotive transport.

Temporomandibular Joint (TMJ)

This joint is affected in almost two-thirds of children with JIA (117) by causing pain in chewing and opening the mouth, stiffness, and micrognathia. Younger children will not complain of jaw pain, but will instead choose to modify their diet to avoid pain. Progressive jaw ROM exercises and modalities may help treat pain and stiffness. If the lower jaw does not develop properly, it may create an overbite, requiring orthodontist intervention and/or oral surgery. Mandibular and facial growth disturbances are more common in polyarticular types of JIA.

Upper Extremities

The shoulder is not commonly involved at the onset of disease. Approximately one-third of children with polyarticular or psoriatic disease may eventually develop shoulder involvement and loss of adduction and internal rotation affecting midline ADLs, such as grooming and toileting. The elbow requires at least 90 degrees of flexion range to perform ADLs such as eating, grooming, and reaching. Loss of more than 45 degrees of elbow extension limits the use of arms as levers to rise from a seated position and makes toileting and lower extremity dressing difficult. Wrist involvement is common in children; there is early loss of wrist extension with progressive flexion contracture. A nighttime resting wrist splint can maintain the wrist in 15 to 20 degrees of extension with the fingers in a few degrees of flexion; ulnar deviation can also be built in as necessary. Strengthening of wrist extensors and radial deviators is necessary to reduce wrist flexion and ulnar deviation contractures. Moist heat to reduce spasm and improve tissue elasticity followed by serial casting for 48 to 72 hours as tolerated may help reduce contractures by slowly increasing wrist extension while controlling ulnar deviation and subluxation; commercially available dynamic splinting may also facilitate stretching. Should ankylosis be inevitable, the hand should be splinted in a neutral position for optimal function in self-care.

Functional grasp may become limited as fingers lose both flexion and extension range. Flexion contractures of the metacarpal and PIP joints are often seen. The use of ring splints in metal or plastic can help control PIP flexion and extension seen in boutonniere and swan neck deformities, respectively. Fingers can be strengthened through play with clay and various adaptive putties.

Lower Extremities

In the lower extremities, flexion contractures occur at the knee and hip. Painful ambulation can lead to increased sitting, which in turn leads to increased flexion contracture, deconditioning, weakness, atrophy, and osteoporosis. Hip flexion contractures in children occur with internal rotation and adduction, compared with adults who tend to develop external rotation and abduction. Prone lying greater than 20 minutes per day with the hips and knees extended and feet off the edge of the bed can help prevent these contractures. Other strategies include strengthening of the hip extensors, external rotators, abductors, and quadriceps, along with ROM exercises to stretch the hip flexors, internal rotators, adductors, and hamstrings. Hip extensors can be strengthened through swimming, aquatic therapy, and bicycling. Encouraging upright posture and ambulation, using a stander as necessary, is also helpful. Hip development may be assisted by the use of a stander; a prone stander can strengthen neck and hip extensors, while a supine stander maintains the knees in extension and allows upright weight-bearing.

The knee is the most commonly affected joint in JIA; early involvement of the knee can cause quadriceps weakness that may not resolve. Knee contractures can lead to other joint contractures and further gait abnormalities. Bony overgrowth with resultant leg-length discrepancies (LLDs) is often seen. The knee can be maintained in extension using resting splints such as knee immobilizers and alternating legs every night as needed to increase comfort and compliance. Dynamic splinting using an adjustable knee joint can be used to improve ROM and limit excessive flexion and valgus tendency. Because forced extension of the knee with a contracture can exacerbate posterior subluxation, caution must be exercised in using bracing and splinting. Active quadriceps strengthening should be done post brace removal and also maintained with knee extension exercise or isometric exercises if too painful. Kicking, bicycling, and walking can also strengthen weak quadriceps muscles.

Multiple foot deformities can occur in JIA, including claw toe, valgus or varus hindfoot, and ankle plantarflexion contracture deformities. The mid-foot is frequently affected, and can be quite painful and difficult to treat. Tenosynovitis that is difficult to discern from joint disease may occur. Molded foot orthoses can be used to reduce pain at the metatarsal heads and heels with weight-bearing. A University of California at Berkeley orthosis can prevent or control varus and valgus deformities. A posterior leaf-spring ankle foot orthosis (AFO) or nighttime resting splint may be helpful to reduce loss of ankle dorsiflexion range and control varus and valgus. Ankle rotation exercises, balancing exercises, and raising the heel on a step can strengthen ankle muscles. Footwear should be comfortable and accommodate any foot deformities. High heels should generally be avoided, as they

can help develop plantarflexion contractures and add to foot deformities. Flip-flops should also be avoided secondary to their lack of adequate support.

Inflammation causing bony overgrowth at the distal femur can cause a true LLD, leading to pelvic asymmetry and scoliosis. The increased blood flow from inflammation may alternatively cause early epiphyseal closure and overall limb shortening.

MEDICAL AND SURGICAL TREATMENTS OF JIA

Children with JIA are treated with more of an induction and maintenance approach, taking advantage of windows of opportunity to modify the disease course, usually under the guidance of a pediatric rheumatologist (118). Treatment is aimed at inducing remission with the least medication toxicity. In 2011 and 2013, the ACR published recommendations for the treatment of JIA organized into five treatment groups as previously discussed (93,94). Within the five treatment groups, treatment is stratified into three disease activity levels (low, moderate, and high). Choice of treatment is guided by disease severity and the presence or absence of poor prognostic features.

In the first treatment group, history of arthritis in four or fewer joints, escalation of therapy typically proceeds from nonsteroidal anti-inflammatory drugs (NSAIDs) to intra-articular glucocorticoid injections to methotrexate to TNF-α inhibitors. In children with low disease activity (single joint, normal inflammatory markers, etc.), monotherapy with NSAIDs may be sufficient. Intra-articular injections of triamcinolone should provide relief for at least 4 months, and can be repeated if helpful. Methotrexate should be started as initial treatment in children with high disease activity and features of poor prognosis, or in those with lower disease activity who fail to show adequate response with NSAIDS and triamcinolone injections. In enthesitis-related JIA, sulfasalazine rather than methotrexate is recommended at a similar stage. After 3 to 6 months (depending on disease activity) of methotrexate with inadequate response, TNF-α inhibitor treatment should be considered.

In children with a history of arthritis in five or more joints (does not require five currently active joints), treatment with initial NSAIDs is more quickly escalated to methotrexate, with joint injection as needed. In moderate disease activity and poor prognostic features, or in children with high disease activity, treatment may start with methotrexate. Leflunomide is sometimes used as an alternative to methotrexate. The IL-6 inhibitor, tocilizumab, is now FDA-approved in children above 2 years of age and older with active polyarticular JIA (119). Tocilizumab can be used alone or in combination with methotrexate (120). Escalation to a TNF-α inhibitor follows in 3 to 6 months dependent on disease characteristics and severity if there is inadequate response to methotrexate or leflunomide. If there is still no response with a TNF-α inhibitor after 3 to 4 months, another TNF-α inhibitor can be tried, or abatacept (a T cell inhibitor). Rituximab may be used for nonresponders to the previously mentioned regimen in those children with RF-positive polyarticular JIA.

In children with the third treatment group, active sacroiliac arthritis, use of a TNF-α inhibitor is recommended more readily than in any other treatment group. A TNF-α inhibitor may be started after failure of an adequate trial of NSAIDs or methotrexate or sulfasalazine for 3 to 6 months (depending on disease characteristics and severity).

In the 2013 recommendations, systemic JIA was broken down into three phenotypes: significant systemic features and varying degrees of synovitis, significant arthritis and no significant systemic features, and features concerning MAS (94,121). In children with significant systemic features and varying degrees of synovitis, initial treatment with anakinra (an interleukin 1 receptor inhibitor) or corticosteroids was recommended in most cases. For children without systemic features and with varying degrees of active synovitis, initial treatment recommendations were methotrexate or leflunomide for an active joint count higher than 4, with change to abatacept, anakinra, a tumor necrosis factor α inhibitor, or tocilizumab (120,106) if there is inadequate response. The interleukin 1 β inhibitor, canakinumab, has also been FDA-approved for systemic JIA (122). NSAIDS or intra-articular triamcinolone joint injections were recommended as initial treatment for children with four or less active joints (106).

For children with MAS, which carries an estimated 6% or more mortality rate, initial treatment recommendations include anakinra, a calcineurin inhibitor, or systemic glucocorticoid monotherapy for up to 2 weeks. The use of abatacept, intravenous immunoglobulin, or tumor necrosis factor α inhibitors is inappropriate for children with MAS (94,121).

It is stressed that systemic JIA is a very different disease from polyarticular JIA and that children need to be treated aggressively with biologics right away. Moving up to the next step after 1 month of inadequate response is recommended (121). The reader is encouraged to check the ACR website, www.rheumatology.org, for the most current, detailed recommendations for treatment of JIA.

Early and aggressive treatment of JIA with newer agents holds unlimited promise for even better outcomes for children with JIA. Steroids are used as sparingly as possible to control inflammation in order to avoid long-term side effects such as weight gain, poor growth, and risk of infection. There is no systemic evidence that steroids are disease-modifying (100).

Children with JIA are at high risk of developing osteopenia secondary to the disease itself, to steroid treatment of the primary disease, lack of physical activity and weight-bearing, limited sunshine exposure, and inadequate vitamin D and calcium. Calcium and vitamin D supplementation, sunshine, and encouragement of physical activity should be incorporated into the treatment plan.

Surgery is rarely used in the early course of the disease; however, surgery can be used later in the course to relieve pain, release joint contractures, and replace a damaged joint. Older children whose growth is complete or almost complete and whose joints are badly damaged by arthritis may need joint replacement surgery to reduce pain and improve function. Soft tissue releases may be needed to reposition malaligned joints or release contractures.

INFECTIOUS DISEASE WITH ARTHRITIS

Infectious causes of arthritis include bacterial, viral or postviral, and fungal. Osteomyelitis and reactive arthritis can also be confused with JIA.

SEPTIC ARTHRITIS

Joint involvement in septic arthritis may be by hematogenous spread, direct extension from local tissues, or as a reactive arthritis.

Bacterial septic arthritis is usually monoarticular in children, but multiple joints can be involved. Children may present with fever, joint pain, and decreased joint mobility, especially in the knees, hips, ankles, and elbows. A child may not allow the affected joint to be touched and, sometimes, may not even allow the affected joint to be seen. An ambulatory child will refuse to bear weight on the affected extremity. Premature infants presenting with irritability, fever, and hips positioned in abduction, flexion, and external rotation should be checked for septic arthritis of the hip. Boys 3 to 10 years who present with hip or referred knee pain should be checked for transient synovitis. Ear infections are the most common source of bacteria leading to septic arthritis in children (123). Osteomyelitis or diskitis can develop in children with septic or reactive arthritis.

In all age groups, 80% of cases are caused by gram-positive aerobes (60% *S. aureus;* 15% beta-hemolytic streptococci; 5% *Streptococcus pneumoniae*), and approximately 20% of cases are caused by gram-negative anaerobes. In neonates and infants younger than 6 months, *S. aureus* and gram-negative anaerobes comprise the majority of infections.

Clinically affected joints require emergent aspiration and treatment. Aspiration of joint fluid is necessary for possibly identifying the agent and relieving pain.

Joint fluid reveals increased white blood cells (WBCs), protein, and low-to-normal glucose. Radiographic findings progress from soft tissue swelling to juxta-articular osteoporosis, joint space narrowing, and erosion. Treatment consists of appropriate antibiotic therapy, joint aspiration to relieve pressure and pain, and physical therapy to maintain ROM.

REACTIVE ARTHRITIS

Reactive arthritis is different from septic arthritis in that it is an autoimmune response triggered by antigen deposit in the joint spaces; synovial fluid cultures are negative. It is set off by a preceding infection, the most common of which would be a genital infection with *Chlamydia trachomatis* in the United States, usually in adult males (124). Reactive arthritis after *Yersinia* and *Campylobacter* can be associated with HLA-B27. *Yersinia enterocolitica* infection can show persistence of the organism in joint fluid, especially the knee. The main goal of treatment is to identify and eradicate the underlying infectious source with appropriate antibiotics, if still present. Analgesics, steroids, and immunosuppressants may be needed for patients with severe reactive symptoms that do not respond to any other treatment.

LYME DISEASE

Lyme disease is caused by the spirochete, *Borrelia burgdorferi,* with transmission to humans via the deer tick, *Ixodes dammini.* Lyme disease is the most common tick-borne disease in North America and Europe. The initial phase of Lyme disease (lasting about 4 weeks) consists of fever, fatigue, headache, arthralgias, myalgias, stiff neck, and erythema migrans. Erythema migrans looks like a reverse target skin lesion, as it is a large, red lesion with a central clearing area; it occurs 1 to 30 days after the tick bite. The late phase, lasting months to years, is characterized by arthritis, cardiac disease, and neurologic disease. Intermittent episodes of unilateral arthritis involve the knee most often; hip, shoulder, elbow, wrist, and ankle may also be involved. In 85% of children, the arthritis resolves before the end of the initial treatment; in 10%, a chronic inflammatory phase develops.

OTHER RHEUMATIC DISEASES OF CHILDHOOD

Systemic Lupus Erythematosus

Systemic lupus erythematosus (SLE) is a multisystem autoimmune disease with widespread immune complex deposition that results in episodic inflammation, vasculitis, and serositis. Children are more likely than adults to present with this systemic disease; 20% of cases begin in childhood. Females are affected 4.5 times more than males. One-third of children have the erythematous butterfly rash over the bridge of the nose and cheeks; this rash may occur after exposure to sunlight. Most children develop a transient, migratory arthritis of the extremities; radiographic evidence of joint deformity and erosion is not common. Pain may be out of proportion to joint findings on examination. Proximal muscle weakness

may be a result of acute illness, myositis, or the result of steroid-induced myopathy. Long-term steroids also increase the risk of avascular necrosis of the femoral head.

Systemic features of SLE may include pericarditis or endocarditis; proliferative glomerulonephritis or other renal disease; seizures, psychosis, memory deficits, headaches, or behavior changes; pulmonary hypertension and/or hypertension. Nephritis occurs in approximately 75% of children with SLE and is the main factor for determining outcome. Hematuria, proteinuria, persistent hypertension, chronic active disease, and biopsy proven diffuse proliferative glomerulonephritis are associated with a poor outcome. Ten-year survival is approximately 80%, although this number is lower in lower socioeconomic populations.

Management of SLE is symptomatic. Maintaining physical activity as much as possible, avoiding excess sunlight exposure, optimizing nutrition, and providing adequate social supports are key. For some children with open discoid lupus rash lesions, dressing changes and wound cares may be best facilitated with individualized whirlpool therapy, much like is used for burn wound cares.

NSAIDs are mainly used for arthritis and musculoskeletal conditions. Fever, dermatitis, arthritis, and serositis usually resolve quickly with low-dose steroids, whereas serologic findings may require weeks of steroid therapy. Hydroxychloroquine may be used for skin manifestations or in concert with steroids to lower the steroid dose. High-dose steroids, immunosuppressive agents, and biologic agents may be necessary for more severe disease manifestations.

Scleroderma

Systemic sclerosis is uncommon in children; linear and focal cutaneous involvement is most common in children. Girls between ages 8 and 10 years are more often affected; duration can last 7 to 9 years. Linear scleroderma presents with atrophic, erythematous skin areas, which later become fibrotic. This skin then binds to underlying subcutaneous tissues, and underlying muscle and bone also become involved. Children may have pain from these skin changes. Soft tissues can atrophy, leaving areas of asymmetry. Scleroderma en coup de sabre is a unilateral linear involvement of the face and scalp, often with loss of hair on the involved side, with loss of facial asymmetry. Systemic disease in children is uncommon. Physical therapy is necessary to prevent loss of ROM and contractures because of the cutaneous involvement. Soft tissue massage, moist heat, stretching, and ROM exercises help maximize joint mobility. Topical corticosteroids may be helpful in treating localized skin disease; systemic steroids, methotrexate, and physical therapy may alter the course of progressive disease.

HEMATOLOGIC DISORDERS

Hemophilia

Hemophilia is a bleeding disorder that affects about 18,000 Americans; each year, about 400 babies are born with the disease, and it occurs in 1 out of every 7,500 males. Of these, about 85% of cases are Factor VIII (hemophilia A) and 14% are Factor IX (hemophilia B).

In hemophilia, bleeding occurs without any recognizable trauma; spontaneous bleeding happens most often in the knees, ankles, elbows, and shoulders. Bleeding into the joints usually begins after a child begins to walk. As bleeding begins, the child may experience warmth or tingling in the joint. As bleeding progresses, there is usually a feeling of stiffness, fullness, and pain. The joint swells and may be warm and tender, causing synovial membrane thickening. Without treatment, hypertrophy of the synovium with its increased vascular supply, creates a cycle of more bleeding and destruction. Without intervention, fibrosis and arthritis set in, making joint replacement at an early age the only option. Pain and swelling can also lead to decreased active joint ROM, further leading to contractures. Other complications include muscle atrophy, osteopenia, peripheral neuropathy, and compartment syndrome.

The main treatment for hemophilia is injections of cryoprecipitate. Acute hemarthrosis requires joint immobilization for 48 hours to prevent further bleeding. Once pain and swelling subside, passive ROM should be started to prevent fibrosis and contracture development. Analgesics, anti-inflammatory medications, and aspiration of blood from the joint if overlying skin is tense are important in pain management. Joint function may be regained in 12 to 24 hours with early factor replacement, but may take up to 2 weeks for more blood reabsorption (125). ROM exercises can be done in the water to reduce stress on the joint while providing resistance; strengthening of specific muscle groups to maximize joint stability should be prescribed. Contact sports are generally contraindicated. Joint replacement is used in end-stage arthropathy; loosening occurs more often, especially in younger children.

Sickle Cell Disease

Joint involvement occurs in infancy in sickle cell disease. Bones and joints are often the site of vaso-occlusive episodes, and chronic infarcts may result. One of the earliest manifestations of sickling in young children is dactylitis or "hand–foot syndrome." An episode of painful swelling of the bones of the hand or foot may predict severe disease (126). Abnormalities of the vertebrae ("fish mouthing") are characteristic of sickle cell disease. Hyperplasia of the bone marrow may cause growth disturbances and osteopenia. Osteomyelitis is also more common and may

be difficult to distinguish from infarction; radionucleotide imaging and bone aspiration are often necessary to diagnose bone infection. Multiple joints can be involved in septic arthritis caused by *S. aureus, E. coli, Enterobacter,* and *Salmonella.* More often, noninflammatory joint effusions of the knee, ankle, or elbow occur during crises. Chronic synovitis in wrists, metacarpal heads, and calcanei with resultant erosive joint destruction has been reported in children with sickle cell disease.

Avascular necrosis of the femur and, less often, the humeral head and TMJ can occur in sickle cell anemia (127,128). Avascular necrosis of the weight-bearing joints (hip and shoulders) causes chronic pain and may require surgical intervention. Plain x-ray films may not detect early disease, and MRI may be necessary. Early disease may improve with coring and osteotomy (129). Late disease requires joint replacement. Patients with sickle cell disease have an increased incidence of infection and failure of prosthesis.

Ischemic stroke is one of the most devastating problems in children. The optimal setting for the care of patients with sickle cell disease is a comprehensive center, with a multidisciplinary team to provide ongoing support.

Summary

The management of children and adolescents with chronic rheumatic disease is broad and multidisciplinary (119). Pediatric physiatrists can help provide supportive treatment to children with rheumatic disease by prescribing appropriate pain medications, exercise, bracing, and equipment to maintain or restore age-appropriate function and development. Such treatment can help prevent deformity and contractures; promote normal growth; and maximize physical, psychosocial, and cognitive development in children with rheumatic disease.

BURN INJURIES IN CHILDREN

EPIDEMIOLOGY

Burns can be a devastating cause of morbidity and mortality in children. The American Burn Association estimates that 450,000 burned adults and children require medical attention each year in the United States and 40,000 individuals require hospitalization (130). Twenty to twenty-five percent of those hospitalized for burns are between 0 and 14 years of age (131). Males are more than twice as likely to become burned as females (130). Disabled children have a higher incidence of burns than their nondisabled counterparts (132). Almost three-quarters of all burns (130) and 80% of contact burns (133) occur in the home. Burns are the third leading cause of unintentional injury death in children 1 to 9 years of age (134). Mortality is highest in those who are very young, have larger burn size, develop sepsis, and/or experience inhalational injury (135).

BURN ASSESSMENT

Burn assessment includes an evaluation of the type, severity, and extent of the burn. Burns can result from thermal, chemical, and electrical exposure. Most burns occur as a result of fire or flame (43%). Burn injury due to scald occurs in 34%, contact in 9%, electrical in 3%, and chemical in 7% (130). However, predominant burn etiology varies with age. For example, scald injuries are most common in children under 4 years of age (135,136), and contact burns disproportionately affect children under 5 years of age (133,136). In older children, burns are more likely to occur as a result of playing with matches or other flammable material (136,137). Children, especially boys aged 10 to 19 years, have a high injury rate related to fireworks (138). About 10% of burn admissions in children are related to child abuse, and about 10% of all abuse cases include burn injuries (139). Features that should raise suspicion for child abuse include symmetric "dip" injuries of the limbs or buttocks, round cigarette or "dropped ash" burns, and prior history of repeated trauma, report of the child or sibling causing the burn, and accompaniment of the child by someone other than the parent (140). An investigator's checklist is available for use in suspected cases of deliberate burn injuries of children (139).

A burn evaluation must include an assessment of the severity of the burn (Figure 10.11). One may observe different burn severities within a single injury. Often, the center of the burn is more severely injured than the periphery. Superficial burns affect only the top layer of the epidermis. They do not blister but are warm, dry, and blanch with pressure. A minor sunburn is the most common example of superficial burn. Superficial burns typically heal without scarring within a few days. Partial-thickness burns affect the epidermis and portions of the dermis and are characterized as either superficial or deep. Superficial partial-thickness burns involve the entire epidermis and upper portions of the dermis. They are associated with blister formation within 24 hours and can be painful and weeping. Like superficial burns, they blanch with pressure and typically heal within 1 to 3 weeks without scarring.

Deep partial-thickness burns include injury through a substantial portion of the dermis and are characteristically different than superficial or superficial partial burns. They involve the hair follicles and glandular tissue, are painful only with pressure, and can vary from waxy white to blistered, red, and weeping. Healing takes several weeks and is almost always associated with scarring.

Full-thickness burns involve injury through the entire dermis and possibly into the subcutaneous tissue. These burns are not typically painful and can range from

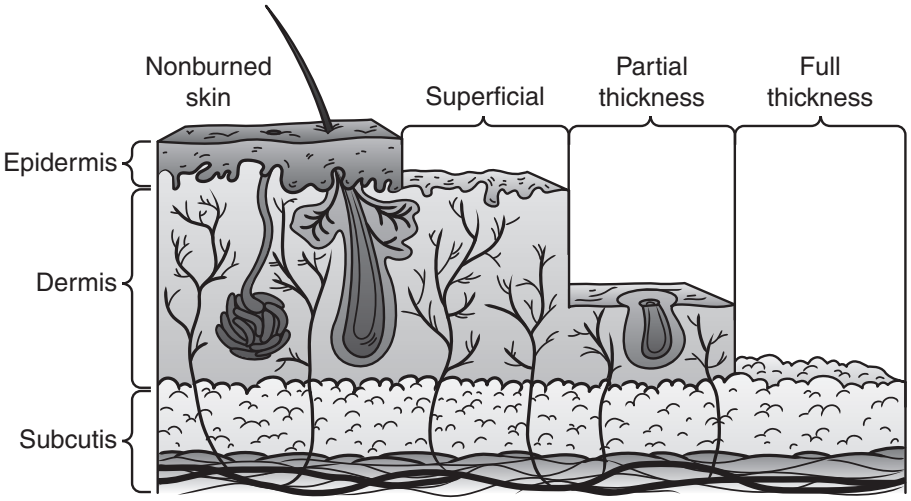

FIGURE 10.11 Classification of burns by severity.

waxy white to gray or black. Healing without medical and/or surgical intervention is unlikely and the risk of hypertrophic scar and contracture development is high. Laser Doppler imaging is increasingly used to accurately determine burn depth (141).

Also critical in evaluating burn severity is an assessment of the amount of body area involved, or percentage of total body surface area (TBSA) burned. Increased mortality is associated with a larger TBSA burned, though survival rates have improved considerably over recent decades. In a study of 952 hospitalized children from 1998 to 2008, those with burn size greater than 60% TBSA had an increased risk of mortality largely due to sepsis and multiorgan failure. It is notable, however, that children with greater than 90% TBSA burns, who might previously have had nearly 100% mortality, more recently had a 55% mortality (142). Standard charts, such as the Lund and Browder charts, are available for estimating burned surface areas for children of various ages (Figure 10.12). The standard adult "rule of 9s" (9% for each upper extremity and head, 18% for lower extremities and anterior and posterior trunk) applies to adolescents but does not apply to small children, who have relatively larger heads and smaller limbs. Another option is to estimate the palm size of affected children as 1% of their TBSA, although this approach may be less accurate (143).

ACUTE BURN MANAGEMENT

The decrease in burn morbidity and mortality seen in recent decades is believed to be related to improved acute care and management of individuals with burns during all phases of care. Adequate response to acute burn injury, including assessment of burns, resuscitation, and admission to a burn center when indicated, have all contributed. Burns have increasingly been managed in outpatient settings with frequent wound checks or dressing changes (130,140). More significant burns are best managed by a burn center. Criteria for admission to a burn center are summarized in Table 10.2.

During the next phase, management of hypermetabolism and organ function, burn surgeries and infection control are critical. As a result of a burn, increases in glucocorticoids, glucagon, catecholamines, and dopamine contribute to the development of the hypermetabolic response. Alterations in glucose, lipid, and amino acid metabolism can lead to organ dysfunction, infection, sepsis, and poor outcome (144). Early appropriate nutritional support is critical to meeting increased energy requirements. Mathematical formulas are available but indirect calorimetry is considered the most reliable method of assessment of energy expenditure (145). Also important in reducing effects of the hypermetabolic response is early burn wound excision and grafting of the excised area with temporary materials or autologous skin. Growth hormone, oxandrolone, propranolol, insulin, metformin, glucagon-like peptide, and fenofibrate are among the medications studied to treat the hypermetabolic response (144,146).

Pain control has been a major area of emphasis in the care of children with burns in the past decade (147). Immediate pain management includes using cool saline-soaked gauze or sheets over burned areas. Medications such as acetaminophen or ibuprofen may be used for smaller burns. Children with larger burns may require opioids such as morphine or methadone for pain control and benzodiazepines for sedation. With protracted, painful treatment in severe burns, tolerance to these medications develops, and very high doses may be required (148). More recently, protocols using ketamine or dexmedetomidine for sedation, amnesia, and analgesia have been described as safe and effective (149,150).

AGE vs. AREA

Initial Evaluation

UMC.519a, Rev 3.99

Area	Birth–1 year	1–4 year	5–9 year	10–14 year	15 year	Adult	2"	3"	Total	Donor Area
Head	19	17	13	11	9	7				
Neck	2	2	2	2	2	2				
Ant. Trunk	13	13	13	13	13	13				
Post. Trunk	13	13	13	13	13	13				
R. Buttock	2½	2½	2½	2½	2½	2½				
L. Buttock	2½	2½	2½	2½	2½	2½				
Genitalia	1	1	1	1	1	1				
R. U. Arm	4	4	4	4	4	4				
L. U. Arm	4	4	4	4	4	4				
R. L. Arm	3	3	3	3	3	3				
L. L. Arm	3	3	3	3	3	3				
R. Hand	2½	2½	2½	2½	2½	2½				
L. Hand	2½	2½	2½	2½	2½	2½				
R. Thigh	5½	6½	8	8½	9	9½				
L. Thigh	5½	6½	8	8½	9	9½				
R. Leg	5	5	5½	6	6½	7				
L. Leg	5	5	5½	6	6½	7				
R. Foot	3½	3½	3½	3½	3½	3½				
L. Foot	3½	3½	3½	3½	3½	3½				
						TOTAL				

Cause of Burn _____

Date of Burn _____

Time of Burn _____

Age _____

Sex _____

Weight _____

Height _____

Date of Admission _____

Signature _____

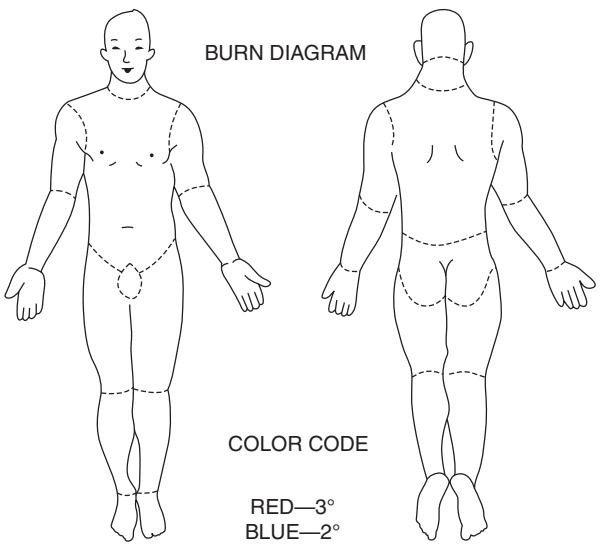

FIGURE 10.12 Lund and Browder burn chart used for estimation of total body surface area burned based on age and body proportions.

TABLE 10.2 CRITERIA FOR REFERRAL TO A BURN CENTER

Burn injuries that should be referred to a burn center include:

1. Partial-thickness burns greater than 10% total body surface area (TBSA).
2. Burns that involve the face, hands, feet, genitalia, perineum, or major joints.
3. Third-degree burns in any age group.
4. Electrical burns, including lightning injury.
5. Chemical burns.
6. Inhalation injury.
7. Burn injury in patients with pre-existing medical disorders that could complicate management, prolong recovery, or affect mortality.
8. Any patient with burns and concomitant trauma (such as fractures) in which the burn injury poses the greatest risk of morbidity or mortality. In such cases, if the trauma poses the greater immediate risk, the patient may be initially stabilized in a trauma center before being transferred to a burn unit. Physician judgment will be necessary in such situations and should be in concert with the regional medical control plan and triage protocols.
9. Burned children in hospitals without qualified personnel or equipment for the care of children.
10. Burn injury in patients who will require special social, emotional, or rehabilitative intervention.

Source: American Burn Association.

Nonpharmacologic adjuvants for pain control include distraction, music and art therapy, relaxation, massage, hypnosis, and imagery (147).

Acute burns are cleaned with a mild soap and water. Ice or very hot or cold water should be avoided. Superficial burns require moisturizer only because intact dermis will protect against infection (143). There is no evidence to support the use of vitamin E cream or topical onion extract ointment in improving scar appearance (151,152). For deeper burns, loose skin should be gently debrided. Controversy exists regarding whether blisters should be unroofed, though there is general agreement that needle aspiration should not be performed (143). Tetanus immunization is provided as needed. Intravenous antibiotics are reserved for those with wound infection and sepsis (153). Current burn care practice utilizes topical creams, ointments, and/or semiocclusive dressings in order to promote moist healing and rapid epithelialization. Scar symptoms such as pain, itching, and tightening may be reduced with the use of these products, although the effect on ultimate scar appearance is unclear (151). A 2013 Cochrane review of treatments for superficial and partial-thickness burns emphasized the relative paucity of high-quality evidence supporting the use of specific topical medications or dressings. However, they found that silver sulfadiazine was consistently associated with poorer healing outcomes than biosynthetic dressings, silver containing dressings, and silicone-coated dressings and that burns treated with hydrogel dressings heal more quickly (154). There is also evidence to suggest that infection rates may be higher with topical silver sulfadiazine (153).

Superficial partial-thickness burns typically heal within a week or two whereas deeper partial-thickness burns may take 4 to 6 weeks. Recently, a systematic review was undertaken regarding the treatment of partial-thickness burns in children (155). Daily to twice daily dressing using silver sulfadiazine or tulle gauze with or without an antiseptic is considered the standard of care in many burn centers. However, concern has been raised regarding disruption of newly formed epithelium, wound infection, and deepening of the wound bed using these products. Semisynthetic or synthetic membranous dressings such as Biobrane (reg.), Mepitel (reg.), and Aquacel Ag (reg.) have been demonstrated to reduce the number of dressing changes, pain, and frequency of wound infections compared to standard care. Amnion membrane as a biologic dressing has shown similar promise and is widely available. Human allograft skin treatment resulted in reduced hypertrophic scarring and need for subsequent surgery than topical creams.

Full-thickness burn management often includes excision and primary closure or coverage with allograft (skin grafts) or biosynthetic skin substitute. Definitive reconstructive surgeries for burns are usually delayed over a year or until scar tissue has matured.

CHRONIC BURN MANAGEMENT

Whether undergoing ambulatory or inpatient management, rehabilitation is critical in achieving improved outcomes. Gait and mobility training with gait aids may

be necessary. Other equipment and adaptive aids may help children and adolescents achieve increased independence in self-care skills. ROM and stretching exercises of areas affected by burns must begin in the acute care phase in order to help prevent contracture formation (Figure 10.13).

Positioning in the direction opposite of expected contracture formation is also important. Splints and other custom-molded orthotics are occasionally necessary to further advance this goal (156,157). Pillows, pads, and other bed-based apparatus may be helpful as well. Optimal positioning based on area burned is summarized (Table 10.3).

Children with severe burns may develop low bone density and an increased risk of long-bone fracture due to prolonged immobilization, nutritional deficits, and an alteration of the hormonal milieu (158). Intervention to improve bone density includes mobilization and improved nutritional intake of calcium and vitamin D (159).

Strength and endurance exercises are important and are facilitated by a physical or occupational therapist. Studies reveal that children with burns who participated in a resistance exercise program over 12 weeks had improved muscle strength, power, lean body mass (160), and pulmonary function (161) compared to those who participated in a standard rehabilitation program without exercise. There was also a decreased need for surgical release of burn contractures (162).

Because scarring is the most common long-term complication of burns, management requires careful attention (Figure 10.14).

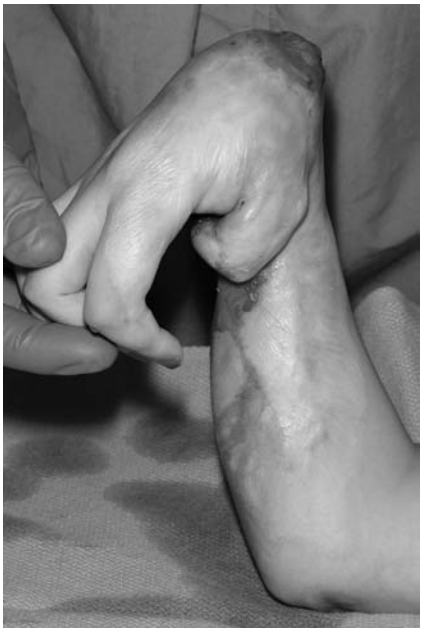

FIGURE 10.13 Girl with burn, upper extremity contractures, and skin wound.

Hypertrophic scarring and keloids are clinically distinct from one another (163). Hypertrophic scars stay generally within the boundaries of the initial injury, are raised, red or pink, and pruritic, and comprise organized type 3 collagen. Keloids, on the other hand, continue to grow and evolve over time, are comprised of disorganized

TABLE 10.3 POSITIONING OF THE PEDIATRIC BURN PATIENT

AREA INVOLVED	CONTRACTURE PREDISPOSITION	CONTRACTURE PREVENTING POSITION
Anterior neck	Flexion	Extension, no pillows
Anterior axilla	Shoulder adduction	90° abduction, neutral rotation
Posterior axilla	Shoulder extension	Shoulder flexion
Elbow/forearm	Flexion/pronation	Elbows extended, forearm supination
Wrists	Flexion	15–20° extension
Hands	Hyperextension	70–90° flexion
MCPs	Flexion	Full extension
IPs Palmar burn	Finger flexion, thumb opposition	All joints full extension, thumb radially abducted
Chest	Lateral/anterior Flexion	Straight, no lateral or anterior flexion
Hips	Flexion, adduction, external rotation	Extension, 10° abduction, neutral rotation
Knees	Flexion	Extension
Ankles	Plantarflexion	90° dorsiflexion

Abbreviations: IPs, interphalangeals; MCPs, metacarpophalangeals.

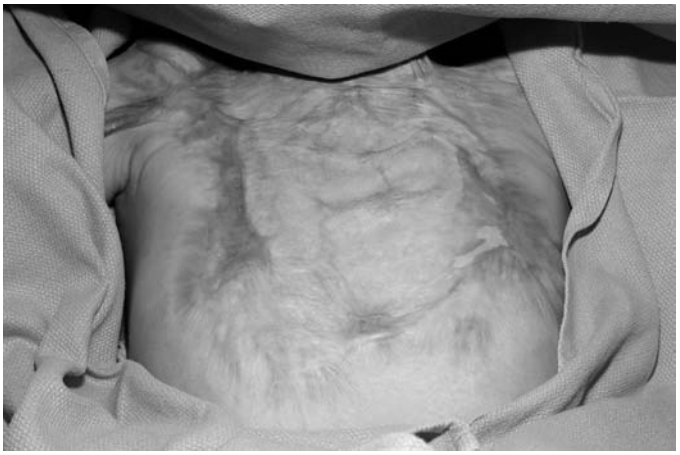

FIGURE 10.14 Girl with burn from improvised explosive device (IED) explosion and scarring of her chest.

types 1 and 3 collagen bundles, and do not enter the remodeling phase. They are more difficult to treat.

Numerous treatment strategies for hypertrophic scars and keloids have been developed. Surgical excision remains a mainstay of scar correction though not always with satisfactory results because of scar recurrence (163). Mature scars require surgical intervention when functional loss or cosmesis can be restored or improved. Later, when a hypertrophic scar, keloid, or contracture has developed, surgical interventions include scar excision, skin grafts, reorientation of scars using flaps, W- or Z-plasties, contracture release, and use of skin substitutes or tissue expansion. Surgical excision of hypertrophic scars and keloids without additional treatment results in a high rate of recurrence. Similarly, surgical correction of equinus contractures using the Ilizarov method resulted in recurrence rates of approximately 70% in younger children (164).

Nonsurgical management techniques include pressure garments, silicone gel or sheeting, and injections (165). These techniques can also be used as surgical adjuncts. Pressure garments have been the mainstay of scar management for decades. Proposed mechanisms of action include decreasing collagen synthesis by decreasing blood flow and realigning collagen bundles already present. A decrease in hypertrophic scar formation reduces the incidence of contractures as well as pain and itchiness (166). Pressure over scars may be achieved through traditional off-the-shelf or custom pressure garments. Care should be taken to avoid wound irritation from seams or zippers. Pressure is usually initiated after wound closure, and garments are worn 23 hours per day for 6 to 24 months (140), or until scar maturation. Garments should be replaced every 6 to 12 weeks in order to maintain compression. Over time, garment alterations may be necessary due to limb size fluctuation and growth of the child. The pressure required for effective treatment has not been scientifically established (166). Some have suggested 24 to 35 mmHg (163), while others have described improvement with pressures as low as 15 mmHg (167). Complications such as discomfort and skin breakdown occasionally result from the use of pressure dressings. High pressures may cause harm such as obstructive sleep apnea (168) or skeletal and dental deformity in children (169).

It is important to note that although pressure dressings are routinely used to prevent and treat burn scars, little scientific evidence exists to support their use (145,163,170). In fact, one prospective, randomized trial of the efficacy of pressure garment therapy showed no significant differences in burn outcome parameters using the Vancouver Burn Scar Assessment between one group that used pressure garments and another that did not (171).

Silicone gel sheeting has been proposed to both help prevent the development of scar tissue as well as reduce the appearance of hypertrophic scar once it has occurred (145). It is recommended to be used 12 hours per day for 2 months. A recent systematic review suggested only weak evidence of a benefit of silicone gel sheeting as prevention for abnormal scarring in high-risk individuals and some possible improvements in already existing scar thickness and scar color. The combined use of silicone plus pressure may be applied (165).

Vigorous scar massage may help to keep scars supple. Massage techniques are performed by a skilled therapist and are taught to the patient or family. Exercise and scar modification techniques should continue over the 12 to 24 months necessary for scar maturation (140) but benefit is based on experience rather than scientific evidence.

Additional management options for keloids and recalcitrant hypertrophic scars with some success include the injection of triamcinolone, antiproliferative agents (5-fluorouracil and bleomycin), calcium channel blockers, and interferon (163,165). Topical steroids and vitamin E creams have not been successfully used for scar treatment (172). Radiotherapy, laser therapy, and cryotherapy are most often used as adjuncts to surgical excision and have demonstrated mixed results (163).

OUTCOME

The most common complication for burn survivors is abnormal or hypertrophic scarring, though the actual prevalence is unknown (173). Abnormal scarring may cause contractures and impaired function. Scar contraction may lead to growth restriction in a child, with resultant distortion of anatomic features, limb length differences, and disfigurement. Based on a Medline review of 50 studies related to functional outcomes after burn injury, limited ROM was reported in 0% to 5% of children with minor burns (mean TBSA 6%) and 47% with massive burns (>80% TBSA). One-third of the children with massive burns were dependent on others for assistance for ADLs, years after injury. It was felt that insufficient data

exist to fully describe the burden of burn injuries (174). In a study of adult survivors of massive burns, quality of life was comparable to the general population. The strongest independent predictors of physical quality of life were size of full-thickness injury and hand function. Mental quality of life was best predicted by age at the time of injury (with younger age predicting a better quality of life) and perceived social support (175).

Following a burn injury, children have an increased incidence of psychological disturbance (176) and difficulty with behavior (175). Significantly higher levels of anxiety, phobias, and enuresis have been noted in this population, and 30% met criteria for posttraumatic stress disorder (PTSD) within 6 months of their burn (177). Pain, separation anxiety, and acute dissociation have been found to contribute to the development of PTSD (169). Feelings of depression and misery were reported by 79% of children with burns in another study (176). Self-esteem and confidence may decline. Fourteen percent to forty-three percent of individuals report dissatisfaction with appearance after a burn (175). Social reintegration may be difficult and social isolation prevalent, with a third having symptoms of antisocial disorder (176). One-fourth to one-third of children with burns experienced interference in playing with other children or seeing friends (175). Return to school and community activities may be eased by education provided to the child's peers and teachers prior to their return.

PREVENTION OF PEDIATRIC BURNS

Public education campaigns have played a key role in reducing the incidence of childhood burns in recent years. An example of this is children's familiarity with the "Stop, drop, and roll" practice taught by firefighters around the country. Other tactics related to reducing burn incidence include reducing hot water heater temperatures to 120 degrees, turning pot handles to the back of the stove, using back burners on the stove, keeping irons off the floor, using smoke detectors and outlet covers, and proper storage of chemicals. Public educational materials related to fire safety are available through the Centers for Disease Control and Prevention (CDC) and American Burn Association (130,178).

DEVELOPMENTAL CONDITIONS

Legg–Calvé–Perthes disease (Figure 10.15) is osteonecrosis of the capital femoral epiphysis in children (8). The condition usually presents between the ages of 4 and 10 years, with a peak incidence of 5 to 7 years. Presentation has been seen as early as 2 years and as late as the late teens. There is a definite male preponderance, with a 4:1 ratio. The incidence of bilateralism has been reported as 10% to 12% (179,180), rarely simultaneous. There is no

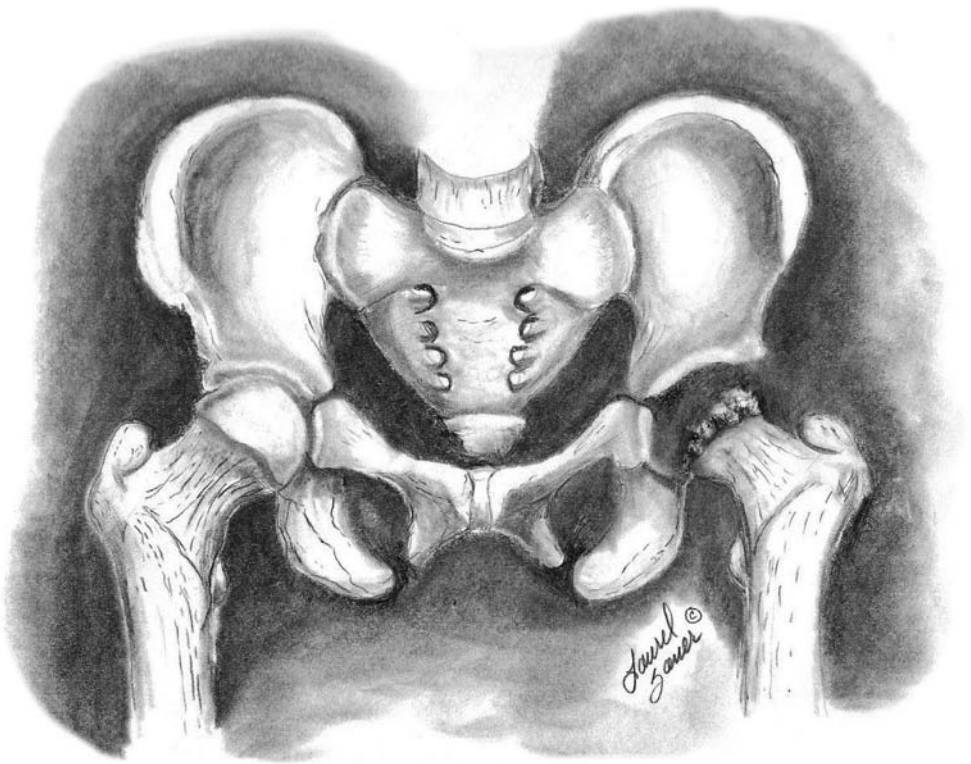

FIGURE 10.15 Legg–Calvé–Perthes disease: left hip.

evidence that the condition is inherited. Limitations in internal rotation, extension, and abduction of the affected hip, with slight shortening of the leg, are common physical findings. Children presenting with knee pain always require a thorough examination of the hip, as this is a common referral pattern.

Catterall classification (181) is graded over four degrees of involvement, depending upon the extent of necrosis across the capital femoral epiphysis. Catterall classification I involves up to 25% of capital femoral epiphysis involvement; classification II, 25% to 50%; classification III, 51% to 99%; and classification IV, 100% involvement. Bone scanning and MRI may provide the diagnosis before radiographs. Arthrography can be helpful in defining classification grades and extent of involvement of the capital femoral epiphysis (5). Laboratory tests include erythrocyte sedimentation rate (ESR), C-reactive protein, and WBC counts, often normal unless concurrent illness is present. Controversy exists about whether treatment of any type affects the natural history of the disorder, particularly when onset occurs under the age of 6. The short-term goal is reduction of pain and stiffness of the hip. The disease process is *self-limited* and may last for 2 to 4 years. NSAIDs are effective in reducing synovitis. Restriction of activity helps relieve pain, which at times may include non-weight-bearing with crutches. Abduction orthosis and casting may be helpful at some point, with improved abduction and femoral head containment the goal. For abduction orthosis to function satisfactorily, the affected hip must be able to be abducted in extension to 40 or 45 degrees. After the orthosis is applied, an anterior–posterior (AP) radiograph of the pelvis is obtained to ensure that the affected femoral head is contained within the acetabulum. To be effective, the orthosis must be worn full time. The use of the orthosis is continued until subchondral reossification is demonstrated on the AP radiograph (182,183,184). The important principal of treatment is based on femoral head containment within a spherical acetabulum so that at least theoretically, reossification is also spherical. Generally, the active phase of the disease that requires an orthosis is 9 to 18 months. Patients with Catterall classification I or II can be treated conservatively, while level III or IV often requires more surgical intervention. *Surgery*, including proximal femoral varus osteotomy, may eliminate longer-term bracing and allow earlier resumption of activities in some children. The prognosis again is better with earlier detection under the age of 8 years and with less than 50% involvement of the femoral head. With increased involvement of the lateral femoral head, more extensive surgical options may be chosen. Femoral osteotomy can result in elevation of the greater trochanter accentuating the abductor dysfunction. Patients older than 9 years of age at onset with Catterall groups III and IV have unpredictable success rates, regardless of treatment methodology. Return to high-impact athletics is restricted until a pain-free status is found during clinical examination and radiographs show healing. Osteoarthritis later in life is often seen, with 50% of untreated patients showing severe changes by the age of 50 years (185,186,187). Other causes of avascular necrosis always need to be considered, including sickle cell anemia, femoral neck fracture, Gaucher's disease, slipped epiphysis, congenital hip dislocations, rheumatoid arthritis, and other collagen disorders, not to mention steroid therapy. Bilateral involvement may be confused with multiple epiphyseal dysplasia or spondyloepiphyseal dysplasia and can be differentiated by doing a skeletal survey. *Acute transient synovitis* (ATS) of the hip is the most common cause of hip pain in children and can present in a fashion similar to Legg–Calvé–Perthes disease (5). In fact, a transient ischemia may occur during acute synovitis of the hip with some rare reports of progression to Legg–Calvé–Perthes disease at a later date. The condition may develop at any time from toddler age onward, with a peak age between 3 and 6 years, and is slightly more common in boys. At least half of the children with ATS have or recently have had an upper respiratory illness, including pharyngitis or otitis media. Trauma of a mild nature is frequently present. Annual hospital admissions for the diagnosis of ATS are reported between 0.4% and 0.9% (5,8,188). The actual incidence of ATS is likely higher, however, as many patients never seek medical attention, and a minority of patients are hospitalized once the diagnosis is made. A lower incidence in African Americans has been noted (189). Ninety-five percent of the cases are unilateral. Annual risk of recurrence for a child with an affected hip is 4% (190,191). Viral etiologies are suspected. *Common presentations* include rapid onset of limping, unilateral pain involving the hip or groin with referral to the knee, and refusal to bear weight on the involved extremity in an otherwise healthy child. A low-grade fever may be present related somewhat to an associated upper respiratory infection. Septic arthritis needs to be excluded, as this is a much more serious joint- and limb-threatening condition. Radiographs are frequently reported as normal when compared to the opposite side, but may show some slight intracapsular effusion. Ultrasonography remains most helpful in detecting effusion (192,193) and may correlate with MRI and a positive radionuclear bone scan. ATS remains a diagnosis only after other conditions have been excluded. *Laboratory evaluation* may show normal to mild elevation of the WBC count and ESR along with the C-reactive protein. Other laboratory parameters are generally within normal limits. Upon hospital admission, aspiration of the hip joint should be considered if septic arthritis is a possibility. Long-term follow-up of children with ATS has demonstrated some lifelong abnormalities, including increased coxa magna (defined as an enlargement of 2 mm or more of the proximal femoral epiphyses) in one-third of patients (194,195). A reactive increase in the blood supply to the femur with increased growth of the articular cartilage secondary to the transient inflammation may be associated with this finding (5,196).

Long-term changes of degenerative arthritis in the hip have been reported in some individuals as well. The fundamental treatment consists of rest and age-appropriate NSAIDs. Partial weight-bearing with crutches can begin with improvement in pain and full ROM through the hip. Most patients' symptoms are resolved within 5 to 7 days, and recurrence is uncommon unless premature activity occurs. Persistent symptoms should be re-evaluated, realizing that low-grade symptomatology can last, in some, up to several weeks. Full, unrestricted activity should be avoided until the hip is completely pain-free and there is no evidence of limping.

Femoral acetabular impingement (FAI) is a recently described concept of significant interest (8). The condition appears mostly in adolescents and young adults associated with decreased anterior head–neck offset in the proximal femur, acetabular overcoverage, or retroversion (197). Mechanical impingement causes anterior labral and posterior contrecoup articular cartilage damage. Clinical characteristics include anterior groin pain exacerbated by flexion activities. A positive impingement test is often present associated with limited hip flexion and internal rotation. The deformity causing impingement is often subtle and frequently missed on radiography. MRI is often required for diagnosis (198). The deformity causing FAI is often bilateral but not unusual to be symptomatic in just one hip. With advanced hip arthroscopy techniques, treatment is available for many through a minimally invasive approach. The goal of surgery is to create impingement free motion. Labrial debridement might be required along with osteochondroplasty (199). Treatment is effective in providing short- and intermediate-term symptom relief. Treatment effect on the natural history and risk of hip osteoarthritis later in life is unknown (8).

Slipped capital femoral epiphysis (SCFE) usually involves posterior inferior displacement of the epiphysis on the proximal femoral metaphysis (Figure 10.16). The term slipped capital femoral epiphysis is actually a misnomer. More accurately, the epiphysis remains in normal position within the acetabulum while the proximal femoral neck and shaft move anteriorly and rotate externally relative to the femoral head (200). The incidence of SCFE in the literature can vary between 1 and 61 per 100,000 persons (8). SCFE is approximately twice as common in boys than in girls and may be bilateral in up to 25% of cases, 5% of which occur simultaneously (5,201,202). More than 90% who develop late SCFE on the contralateral side are asymptomatic. Affected children are often large and overweight, and an association with endocrine factors such as hypothyroidism, hypopituitarism, hypogonadism, and excessive growth hormone (GH) has been reported. *Body mass index (BMI)* may be an accurate tool for assessment of risk for SCFE (203). Findings show that patients with SCFE had a statistically higher BMI during growth than normal developing children. It is more common in African American boys with accumulated risk

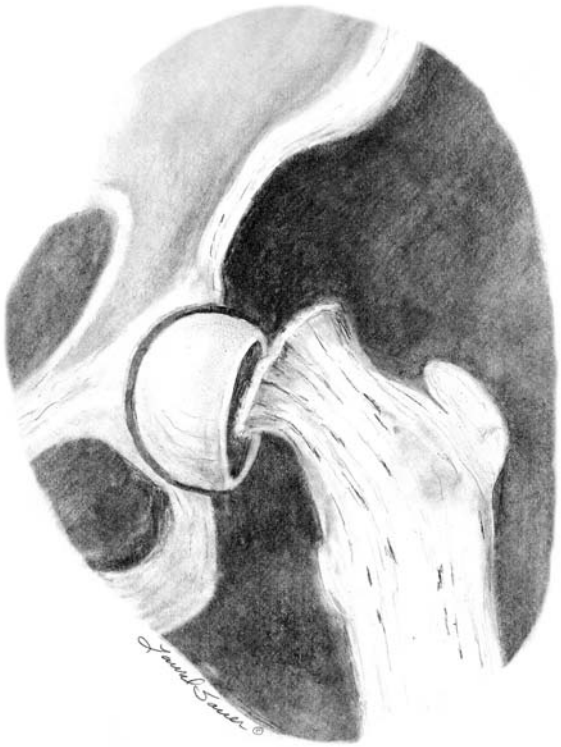

FIGURE 10.16 Slipped capital femoral epiphysis.

that may be as high as 1 in 400 (8). It is also more common in northern environments, possibly related to less sun exposure and relative vitamin D deficiency. Laboratory studies have demonstrated that estrogen strengthens and testosterone weakens the physes (204,205). SCFE may be thought of as occurring because of physiologic loads across an abnormally weak physis (more common in peripubertal children) or abnormally high loads across a normal physis (more common in obese children). The increased prevalence of hypothyroidism in children with Down syndrome is a likely explanation for the increased risk of this condition in these children (206,207,208). *Mechanical factors* appear important with an association of SCFE to decreased femoral anteversion and femoral neck shaft angle (209,210). Age at presentation is typically between 12 and 16 years for boys and 10 and 14 years for girls. Presentations of SCFE outside of these age ranges should alert physicians to potential endocrinopathy or alternative conditions such as renal osteodystrophy. The two most common features of presentation are pain and altered gait. The pain may come on acutely (unstable SCFE), but more commonly builds up over a number of weeks or months. As usual with hip pathology in children, pain occurs in the groin region and radiates to the knee and medial thigh.

It is aggravated by walking and other high-impact activities. External rotation of the leg is common with some shortening and antalgic Trendelenburg's gait. Physical examination demonstrates a loss of internal

rotation, diminished flexion, shortening of the leg, and atrophy of the thigh if the symptoms have been long-standing. Mild slips show displacement of the epiphysis up to one-third of the width of the metaphysis, moderate slips up to two-thirds, and severe slips greater than two-thirds displacement. The displacement is best quantified on lateral radiographs, which should not be lacking in the workup of a child with hip pain. *Klein's line or Trethowan's sign* is a line drawn along the superior surface of the femoral neck, which normally should pass through the lateral portion of the capital femoral epiphysis. If this line does not touch or passes above the epiphysis, at least minimal slippage is present and further intervention required (211,212,213). Slip angle is another good way of measuring degree of severity on a true lateral radiography (214,215). A perpendicular line drawn from the base of the capital femoral epiphysis bisecting a line drawn through the midshaft of the proximal femur is measured. An angle less than 30 degrees is mild slippage, 30 to 60 degrees moderate, and 60 to 90 degrees severe. When SCFE is suspected, ambulation should not be allowed until an orthopedic surgeon sees the child. Other radiographic features include widening of the epiphyseal line (Salter I fracture-type appearance) with metaphyseal changes including rarefaction and cysts. Bone scan and MRI can be helpful in determining the preslip stage as compared to the opposite uninvolved side (8). The *current standard of treatment* for SCFE is in situ pinning with cannulated screw fixation done on an urgent basis. The goal of treatment is to arrest further progression of the slip and to gain closure of the capital femoral epiphysis. Management of patients with unstable SCFE can involve minimal repositioning by an experienced orthopedist and two-screw fixation instead of one. Generally, the epiphysis is left in its displaced position because avascular necrosis is a 10% to 25% risk if manipulation is attempted. Spontaneous reduction of the slippage or controlled reduction by an experienced orthopedist under fluoroscopic guidance has not been associated with an increased rate of osteonecrosis in patients with unstable SCFE (8). Cortical bone grafts have also been used, crossing from the metaphysis to the epiphysis and resulting in epiphysiodesis. Spica casting is becoming a less common practice because of secondary complications in obese children and immobility for up to 3 months. After successful physeal closure, the proximal femur can remodel, particularly in children under the age of 10 years. Bony osteotomies can be indicated if further femoral head coverage is required despite more conservative care. Chondrolysis or acute cartilage necrosis may occur postoperatively in severe cases. If chondrolysis is present, most individuals go on to develop narrowing of the joint space with some degree of ankylosis, degenerative arthrosis, and pain. Total hip arthroplasty can be a consideration for older individuals. Weight-bearing is generally avoided for at least 6 weeks after surgery followed by active assistive exercises and strengthening to restore lengthening, adduction, and internal rotation. Full identification of this condition while only minimal displacement is present and immediate surgery generally allow rapid mobilization and return to full activity with no sequelae. *Prophylactic pinning of the contralateral hip* is an area of ongoing discussion. In one recent study of 94 hips treated with prophylactic pinning, there were no significant complications (216,217). The risk of osteonecrosis and chondrolysis was felt to be virtually negligible when using in situ two-screw fixation with improved imaging technology and radiolucent tables. Opponents of prophylactic pinning cite the complications and potential risks of pinning numerous hips that will never slip. They also point out that with appropriate patient counseling and close follow-up, most subsequent slips will be detected early while they are still mild and treatable. Currently, prophylactic contralateral hip fixation is recommended for patients with established metabolic or endocrine disorders, those with increased risk from radiotherapy or chemotherapy, and for children with SCFE who are younger than 10 years of age. Once the triradiate cartilage is closed (around the age of 14 to 16 years) and when Risser lines appear, the risk of contralateral slip is felt to be negligible (8). *SCFE differs from other pediatric disorders* of the hip such as Legg–Calvé–Perthes disease and developmental dysplasia of the hip (DDH), in that SCFE occurs at an age when the majority of the acetabulum has been developed and thus less acetabular adaptation to deformity of the femoral head can occur. All of this speaks to early detection and early treatment, particularly in those children of elevated risk.

Developmental dysplasia of the hip (DDH) is the most common disorder of the hip in children and the musculoskeletal condition, causing the highest level of concern for the pediatric practitioner (8). Dysplasia of the hip (mostly involving the acetabulum) occurs in approximately 1 in 100 births, with frank dislocation in approximately 1.5 births per 1,000. DDH is classified as either typical (90%) or atypical (10%) (Table 10.4). DDH is not always detectable at birth, and thus the term "developmental" rather than "congenital" has been chosen by the Pediatric Orthopedic Society of North America. The term DDH is felt to more accurately reflect the variable presentation of this complex disorder. Dysplasia refers to an underdeveloped acetabulum, subluxation of hip still in partial contact with the acetabulum, or dislocation of the femoral head not contained in the acetabulum. The dislocated hip should be detectable clinically in the newborn period by 4 to 6 weeks. Atypical teratologic hip dislocations (atypical) occur in utero and are not reducible on neonatal examination. Atypical dislocations are present about 10% of the time and are more commonly associated with other chromosomal or neuromuscular conditions, such as myelomeningocele, arthrogryposis, or Ehlers–Danlos syndrome. Typical DDH occurs in an otherwise normal infant and may take place in utero, perinatally, or postnatally (Table 10.4). Risk factors associated with DDH are listed in Table 10.5.

TABLE 10.4 CLASSIFICATION OF DEVELOPMENTAL DYSPLASIA OF THE HIP

CLASSIFICATION	DESCRIPTION
Atypical (10%) or teratologic	Primarily malformed acetabulum or femoral head in utero associated with myelomeningocele, arthrogryposis, Ehlers–Danlos syndrome
Typical (90%)	Otherwise normal infant but varying degrees of hip morphology and placement
Subluxed	The femoral head and the acetabular cartilage are in contact, but not correctly centered
Dislocatable	The femoral head can be dislocated with maneuvering
Dislocated	The femoral head is completely out of the acetabulum

DDH classification is based on the Graf method (218) with varying severity having type I, a normal hip, to type IV, a dislocated hip. DDH predominates in the left hip (60%), but often bilateral involvement can be discovered. Bilateralism can be most difficult to diagnose with the absence of asymmetry as a helper. Beware of bilateral DDH when thigh skin folds extend past the anus and decreased absolute abduction is present on both sides (5). In the older child, bilateral involvement may be detected only by hyperlordosis and a waddling gait. First-born females presenting with breech have the highest risk for DDH at 8% (8). Risks for DDH in subsequent pregnancies are 6% when neither parent has a positive history and 12% when one parent is with a positive history. The presence of idiopathic clubfeet does not obligate special screening (219,220), but this may be helpful in a small percentage.

TABLE 10.5 RISK FACTORS ASSOCIATED WITH DEVELOPMENTAL DYSPLASIA OF THE HIP

Caucasian
Hip swaddling in extension (Native American, Lapland)
Female–male ratio 6:1
Breech birth
Positive family history
Primiparity
Ligamentous laxity
High birth weight (>4,000 gm)
Congenital muscular torticollis
Metatarsus adductus
Oligohydramnios
Hip asymmetry (limited abduction of one or both hips)
Congenital knee dislocation/recurvatum

Neonatal hips with mild dysplasia that have no instability do not benefit from early treatment, as more than 95% of such hips spontaneously normalize (220,221,222).

Certainly infants with risk factors (see Table 10.5) need to be screened in ultrasound followed by careful clinical examinations until they reach walking age. Ultrasonography is an established method of diagnosing and following dysplasia and has been found to be more sensitive than clinical exam in infants (221,223,224).

The alpha angle is measured from the vertical reference through the iliac bone and tangential to the osseous roof of the acetabulum. This angle represents the hard bony roof and reflects acetabular depth (8). The beta angle is created by the vertical reference through the iliac bone, intersecting with a line drawn through the labrum representing the cartilaginous roof of the acetabulum. The beta angle indirectly reflects the lateral position of the femoral head. A normal alpha angle is greater than or equal to 60 degrees and a beta angle is less than 55 degrees (225).

After 4 to 5 months of age, when the ossific nucleus of the femoral head has generally appeared, radiographic screening replaces ultrasound in evaluation of infants with DDH. An acetabular index of greater than 30 degrees, a break in Shenton's line, or displacement of the femoral head from the socket are all indicative of dysplasia. Additionally, femoral head placement outside the lower medial quadrant formed by the intersection of Hilgenreiner's and Perkin's lines is abnormal. Parameters for monitoring hip dysplasia in this age group are represented in Figure 10.17. Measurement of center-edge angle

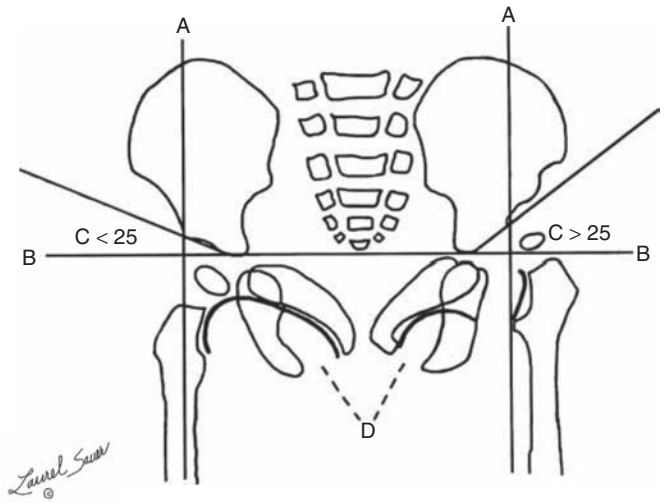

FIGURE 10.17 Radiographic evaluation in developmental hip dislocation. (A) Perkin's vertical line: perpendicular dropped from the lateral acetabular margin. (B) Hilgenreiner's line through the Y cartilages. The femoral head should lie in the lower medial quadrant formed by the intersection of the two lines. (C) Acetabular index: the angle formed by a line through the acetabular roof and Hilgenreiner's line; normal below 25 degrees. (D) Shenton's line: the arc appears broken in the presence of dislocation. The abnormal hip appears on the right.

becomes useful in the patient who is more than 5 years of age and particularly useful in the adult patient (226). Center-edge angles less than 20 degrees (angle between two lines drawn through the center of the femoral head, one vertical and the other tangential passing through the lateral acetabular edge) are concerning for unwanted lateralization of the femoral head.

Clinical examination with repetitive follow-up continues to be the mainstay of diagnosis for DDH (5). The infant should be quiet and comfortable so that the muscles about the hip are relaxed and supple to exam. In early infancy, instability is the most reliable sign (214). Instability declines rapidly with age, over 50% within the first week. Stiffness, shortening, and limited abduction become much more prominent by 2 to 3 months of age. Initial instability may be the result of maternal or fetal hormonal laxity, genetics, and intra- or extrauterine malpositioning. The longer the femoral head remains in a subluxed or dislocated position, the more likely progressive change in acetabular anatomy will occur. A hip that is reduced at rest but subluxed or dislocated by adduction, flexion, and posterior pressure has a positive Barlow's maneuver. Concurrent acetabular dysplasia may or may not be present (220,221). Barlow's tests often become negative by 2 to 3 weeks of age as maternal or fetal hormonal influences diminish. Hips that are dislocated can be reduced back into the acetabulum by abduction and forward lifting of the thigh producing a palpable "clunk." A hip that is reduced this way has a positive Ortolani's sign and is often accompanied by acetabular maldevelopment. Hip "clicks" are short-duration, high-pitched sounds that are common, benign, and need to be distinguished clearly from "clunks." "Clicks" and asymmetrical thigh folds are common in normal infants and generally benign (214). A positive Galeazzi's sign may be seen in infants with DDH, noting a decrease in height of the involved knee with the hips flexed supine to 90 degrees. In infants older than 3 months, limitation of motion and apparent limb shortening predominate. The dislocated or subluxed hip develops tightness in the adductor muscle groups with limited asymmetric abduction. Again, this is much easier to detect when unilateral than bilateral. Parental or family reports of an infant with unusual positioning of legs or crawling warrant investigation. In older ambulatory patients, Trendelenburg's limp, waddling gait, and hyperlordotic posture require evaluation. Fatigue, pain, and instability can still occur in adolescence.

For the infant with a positive Barlow's sign and normal ultrasound at 4 to 6 weeks (no evidence of instability on stress maneuvers) with clinical stability returned, no treatment or radiograph follow-up is recommended (8). Serial clinical examinations of the hip should continue by the primary care physician until the child reaches walking age.

For children with dysplasia and an abnormal ultrasound or persistent subluxation, treatment is with the Pavlik harness (Figure 10.18). The *Pavlik harness* is not appropriate for teratologic dislocation. Follow-up clinical examinations in the Pavlik harness should be completed at least every 2 weeks, with serial ultrasound studies at least monthly. The harness needs to be adjusted at least every 2 weeks for the rapid growth evident in this young infantile population. Failure to adjust the Pavlik harness can cause additional acetabular pathology, including a now-dislocated hip that was previously reduced. The anterior adjustable straps for the Pavlik harness are set to keep the hips flexed at approximately 100 degrees (Figure 10.18). Excessive flexion and tightening need to be avoided, as additional problems can be caused such as femoral nerve palsy. The posterior straps are meant to encourage gentle abduction of approximately 45 degrees. They should be loose enough to allow two to three fingerbreadths between the knees when the knees are held flexed and adducted. Forced abduction should be avoided to minimize any complication of osteonecrosis. The child can be weaned from the Pavlik harness over a 3- to 4-week period once ultrasound parameters become normal along with stability on clinical examination. *Treatment* with the Pavlik harness for neonatal acetabular dysplasia is more than 90% successful. Follow-up is still required with AP radiographs through the growing years, with a 10% risk of abnormality necessitating clinical follow-up into adolescence. Fixed hip abduction orthosis replaces the Pavlik harness in children over 6 months of age, generally because of strength and size (227). *Early screening* and repetitive clinical examinations have been shown to significantly reduce surgical procedures and hospitalizations, including late presentation

FIGURE 10.18 Pavlik harness.

of DDH in this population (221,228,229,230,231). Persisting with the Pavlik harness when reduction is not achieved by 3 to 4 weeks may cause additional femoral head deformity or posterior fixation, and make a closed reduction difficult or impossible (214). If the hip is not reduced, traction, adductor tenotomy, or closed or open reduction including arthrogram and spica cast needs to be considered (226).

For *Pavlik failures* in children under 18 months of age, closed reduction may be tried next, provided it can be achieved without undue force (8). The preliminary use of traction for 3 to 4 weeks before attempting closed reduction is becoming less common (232,233). The hips are generally maintained in about 0 to 100 degrees of flexion, with abduction less than 60 degrees. Reduction of the hip or hips in the spica cast must be confirmed, usually by CT scan or arthrography (5). Immobilization in the spica is continued for approximately 3 to 4 months with cast changes about every 6 weeks. Attention to the difficulties presented in hygiene, mobility, transport, and general development by the presence of the cast is important, and positioning and mobility devices such as scooters, adapted car seats, and accessible toys should be provided. With clinical stability achieved and visualized on radiographs, abduction bracing can be used subsequently until a normal acetabulum is achieved.

When treatment with the Pavlik harness and/or closed reduction fails, *surgical reduction* is required, more commonly after the age of 18 months (231). Often, the decision to perform open reduction is made in the operating room following arthrography and failed closed reduction. The purpose of open reduction is to remove obstacles to reduction and achieve increased stability and clinical outcome. Intraoperative arthrography can be helpful in defining specific anatomic blocks to reduction and choosing the best surgical approach. Obstacles to reduction include the iliopsoas tendon, joint capsule, transverse acetabulum ligament, fibrous fatty tissue, and the ligament teres. Numerous pelvic and femoral osteotomies are available, with choice based on the pathology and the experience in surgical preference (231). Femoral shortening can relieve pressure on the femoral head and acetabulum, reducing cartilaginous pressure and the risk of osteonecrosis (5,234,235,236). Derotational femoral osteotomy can be helpful if excessive anteversion is present requiring extreme internal rotation to maintain reduction. Secondary procedures, including redirectional femoral and pelvic osteotomies, are more common after the age of 2 in an effort to maintain concentric reduction and minimize the risk of osteonecrosis. Remodeling of the hip and acetabulum is most predictable under the age of 4, less predictable between the ages of 4 and 8, and unpredictable after the age of 8. *Secondary procedures* should be performed if at all possible prior to the age of 8 for best outcome (231). Failure to achieve reduction in the older child results in a permanently subluxed hip with marked gait deviation and susceptibility to osteoarthritis and pain syndromes. *Long-term outcomes* can include joint arthrodesis and the need for total hip arthroplasty in the younger adult (5). The importance of early diagnosis and treatment of developmental hip dysplasia cannot be overemphasized: The results are generally good with appropriate intervention and disastrous if neglected (237).

Traumatic hip dislocations in children are relatively rare, and when they occur, they are usually posterior (5,212,238). The mechanism is usually traumatic, with a direct blow to the knee with hip and knee flexed, as occurs with a fall during ground impact or dashboard contact during a car accident. Some dislocations have occurred while playing mini rugby, in which players kneeling on the ground have had someone fall on top of them. Avascular necrosis may occur in up to 10% of cases. Sciatic nerve palsy is rare, but needs to be ruled out. Only 5% of all traumatic hip dislocations occur in patients younger than 14 years. Males account for approximately two-thirds of these dislocations, with more than 99% being unilateral. Posterior hip dislocation is an emergency that requires immediate referral to an orthopedic specialist (5).

Overuse syndromes are generally conditions caused by unresolved submaximal stress in previously normal tissues. They involve microtrauma resulting from chronic repetitive insults to the musculoskeletal system. Growth cartilage seems to be more susceptible to stress and overuse than adult cartilage. Growth cartilage is present at three different sites: the physes, the joint surface, and the major muscle–tendon insertions or apophyses. With focus on single sports early in life, overuse injuries have become more prevalent in the pediatric athlete (5,214). Treatment of overuse syndromes generally involves conservative modalities and rest, followed by strengthening and stretching of muscle imbalances and gradual return to activity as tolerated. Please see the sports musculoskeletal section for further discussion of these and other conditions as helpful.

The elbow continues to be the most commonly injured joint in children (5). Acquired dislocations account for about 8% of elbow injuries and are most frequent in children under the age of 10 years (8,239,240). Typically, the injury involves the nondominant extremity with a fall onto the outstretched hand (241). *Nursemaid's elbow* consists of radial head subluxation from a sharp upward pull on the extended pronated arm in preschoolers. A generalized ligamentous laxity in children with large cartilaginous components of the distal humerus and proximal ulna, in addition to osseous instability, with numerous secondary ossification centers and epiphyses all contribute to the tendency for the pediatric elbow to dislocate. Posterior or posterior-lateral dislocations account for up to 90% of the injuries (5,8). *Closed reduction* with the patient under sedation is the treatment of choice (11,241,242). With nursemaid's elbow, typically the child will not move the arm and holds it in a slightly flexed and pronated position. Radiographs are usually not indicated,

as the injury is more subluxation of the annular ligament rather than true joint subluxation. Longitudinal traction and additional pronation followed by flexing the elbow above 90 degrees and then fully supinating the forearm produces reduction in most cases. A click or snap is often felt as the annular ligament repositions (5). Ulnar and median nerve entrapment needs to be ruled out clinically postreduction.

Shoulder injuries remain relatively uncommon in the overall picture of injuries to the pediatric musculoskeletal system (8). When they occur, they include separation of the acromioclavicular joint from direct trauma, osteolysis of the distal clavicle (mostly in weight lifters), and cervical clavicular injuries in the young thrower (5,11,243). Rotator cuff injuries remain less common in the younger athlete. Conservative treatment for musculoskeletal injury in children includes rest, ice, compression, and elevation (RICE) in addition to NSAIDs such as Tolectin, naproxen (Naprosyn), and ibuprofen (Children's Motrin, Children's Advil). Appropriate equipment, coaching, recreation environments, and training often prevent sports injury, with safety remaining the primary consideration.

Osteochondritis dissecans is a condition resulting in partial or complete separation of a segment of normal hyaline cartilage from its supporting bone (Figure 10.19). Depending on the separation, cartilaginous or osteochondral intra-articular fragments may form (214). Mechanical symptoms may arise within the joint such as catching or locking. Although it has been more than 100 years since Konig (243) coined the term osteochondritis dissecans, the cause remains unclear. Five theories commonly suggested are ischemia, genetic predisposition, abnormal ossification, trauma, and cyclical strain (5). The condition most commonly affects the knee (lateral aspect of the medial femoral condyle in 70% of patients, lateral femoral condyle in 20%, and the patella in 10%) or can be seen in the elbow (8). Treatment of osteochondritis dissecans remains controversial. Intact lesions can often be treated symptomatically, with or without activity modification or immobilization (5). Free fragments often require surgical removal. Drilling techniques are commonly utilized and can help stimulate new bone formation healing and return of mobile bodies to their original donor sites (8). Long-term sequelae can develop in up to 25% with atypical lesions, older age, effusion, and larger lesions.

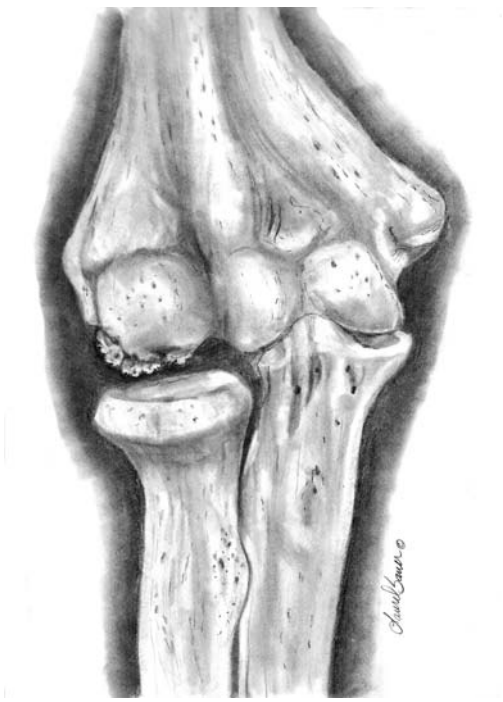

FIGURE 10.19 Osteochondritis dissecans: right elbow (capitulum).

Chondromalacia of the knee needs to be distinguished from the more serious osteochondritis dissecans. Chondromalacia is a term used to describe anterior knee pain of undetermined cause in the younger athlete associated with softening of the articular cartilage beneath the patellar surface. The pain is frequently worse with squatting and climbing stairs, and is associated with a high-riding patella or malalignment. Patellar dislocations can occur in association and are usually lateral and associated with genu valgum, external tibial torsion, and general ligamentous laxity. The subluxation of the patella is usually reducible, but can be painful. Exercises to strengthen the quadriceps, particularly the vastus medialis and the use of patella tracking braces, may be helpful. Surgical stabilization of the medial patellar tissues and lateral retinacular release can be helpful in more difficult cases.

Osteochondrosis is characterized by a disturbance in endochondral ossification, including both chondrogenesis and osteogenesis, in a previously normal endochondral growth region (5). The term osteochondrosis is preferred, as not all conditions are inflammatory, making the term osteochondritis inappropriate (214). Osteochondrosis is *idiopathic* and has been reported in nearly every growth center of the body, including apophyses, epiphyses, and physes. Their eponyms are generally named according to the region of the body and growth center involved (214). Most osteochondroses have well-defined natural histories and generally predictable outcomes (244). *Freiberg's disease* involves collapse of the articular surface in subchondral bone, usually of the second metatarsal (11,245,246). *Kohler's disease* involves irregular ossification of the tarsal navicular joint with localized pain and increased density. Freiberg's disease is more common in girls between the ages of 12 and 15 years, whereas Kohler's disease occurs in younger individuals aged 2 to 9 years and is frequently reversible with conservative care including orthoses and casting. *Apophysitis* is relatively common at the knee, foot, and ankle, all secondary to traction overuse and microtrauma. Apophysitis at the inferior pole of the patella is called *Sinding-Larsen–Johansson* syndrome. *Osgood–Schlatter*

disease involves apophysitis at the tibial tuberosity, and *Sever's disease* (Figure 10.20) involves apophysitis at the posterior calcaneus. These conditions generally occur around the age of 10 to 15 years of age, a few years earlier in girls, and are generally treated conservatively with the *RICE protocol*. Care should be taken not to overgeneralize treatment, however, as each condition can be different and require special attention. *LaNec disease* or ischial pubic synchondrosis, for instance, can be confused with a bone tumor if not careful and subsequently overtreated (214). *Heel cups* may be helpful with Sever's disease in addition to short periods of casting and/or splinting. Stretching of the quadriceps and hamstrings can be helpful with Osgood–Schlatter disease in addition to knee sleeves or knee straps. NSAIDs are often prescribed as well. Pain-free strengthening of weight-bearing soft tissues using more closed kinetic chain techniques may be best. Chondromalacia needs to be differentiated also from the osteochondroses in the young person with anterior knee pain.

Osteochondrosis of the vertebral endplate is known as *Scheuermann's disease* (Figure 10.21). The incidence of Scheuermann's deformities in the general population ranges between 0.5% and 8%, with an increased prevalence in males (5,247,248). It is distinguished from postural roundback by its more rigid structural characteristics. Symptoms are common during the early teenage years and in most instances decrease in late adolescence (8). When *three or more consecutive vertebrae* are wedged more than 5 degrees, radiographic criteria for Scheuermann's disease are met (249). The radiographic picture includes irregular vertebral endplates, protrusion of disk material into the spongiosum of the vertebral body, Schmorl nodes, narrowed disk spaces, and anterior wedging of the vertebral bodies. The *cause of Scheuermann's disease* again is unknown, but thought by some to fall within the spectrum of repetitive microtrauma and fatigue failure of the immature thoracic vertebral bodies. An increase in the incidence of disabling back pain in adults has been reported and may lead to surgery in this older age group (5,8,247). More severe pain is reported in patients with kyphotic deformities greater than 75 degrees.

FIGURE 10.20 Calcaneal apophysitis—Sever's disease.

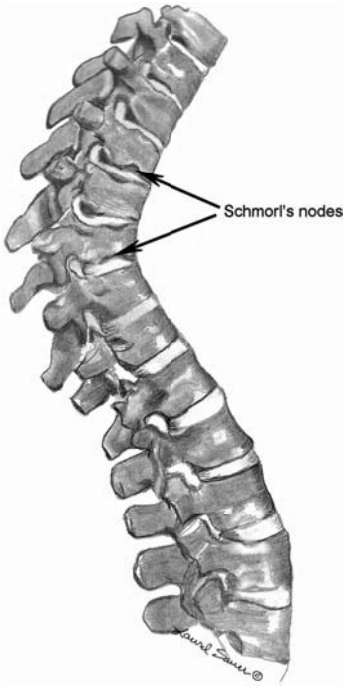

FIGURE 10.21 Scheuermann's disease.

Cardiorespiratory conditions may occur in patients with severe deformities (kyphosis greater than 100 degrees). *Atypical Scheuermann's disease* (248,250) or thoracolumbar apophysitis is named because it does not meet the usual radiographic criteria for Scheuermann's disease established by Sorenson (251). This phenomenon is usually seen at the thoracolumbar junction and may be the pediatric equivalent of an adult compression fracture. There is a 2:1 male-to-female predominance, with a peak age of incidence between 15 and 17 years. When Scheuermann's disease is associated with pain in the presence of one or more irregular vertebral bodies, physical exercise is prohibited. A *thoracic–lumbosacral orthosis (TLSO)* or Milwaukee brace is used for more severe involvement. Sometimes bracing is required for 3 months to achieve pain control. *Conservative care*, including traditional RICE protocol, gentle flexibility routines, and NSAIDs, can be helpful. For correction of spinal deformity with bracing, a mobile kyphotic deformity is required in addition to at least a year of growth remaining in the spine (8). In most cases, brace treatment must be continued for a minimum of 18 months to have an effect on vertebral wedging. Severe involvement progressing to more rigid kyphosis, greater than 75 degrees, may require spinal fusion, both posterior and anterior (5,8).

Intervertebral disk injuries in children and the young athlete are uncommon (8). In contrast to the selective motor and sensory deficits often observed in adults with

disk herniation, athletes under 20 years of age have pain and tenderness localized generally to the midline and, to a lesser extent, over the course of the sciatic nerve (11,252). Of surgically treated disk herniations, only 1% to 2% occur in the pediatric population. Many of these children have underlying congenital anomalies, including transitional vertebrae, spondylolisthesis, and congenital spinal stenosis.

Spondylolysis has never been found in the newborn. Its occurrence increases between the ages of 5.5 and 6.5 years to a rate of 5%, close to the frequency of 5.8% in the Caucasian population (5,253). The condition involves a fracture to the *pars interarticularis* and is more common in athletes involved with repetitive flexion–extension and hyperextension activities of the lumbar spine. Oblique radiographs of the lumbar spine show the classic *"scotty dog" sign* (254). Common sports associated with this condition are: gymnastics, weight lifting, and figure skating. Please see the sports musculoskeletal injury section for further discussion as helpful.

The incidence of *back pain* in *backpack users* of school age has been noted in up to 74% of individuals (252,255). Heavy backpack use, female gender, and larger BMI were all associated with back pain. Back pain from backpacks needs to be considered readily in all individuals, particularly those with spondylitic conditions and regular daily use (11,256). *Spondylolisthesis*, or slipping forward of the vertebral body, may occur during childhood, with a prepubertal peak incidence and promoted by hyperlordosis. Grading of spondylolisthesis (Figure 10.22) is according to the classification developed by Meyerding (257).

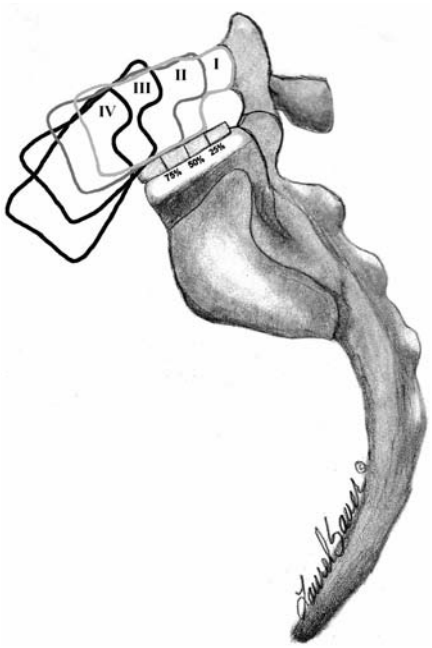

FIGURE 10.22 Meyerding grading classification for spondylolisthesis.

The superior border of the inferior vertebrae is divided into four equal quadrants, with slips in each quadrant accounting for one grade. Surgical treatment is necessary in the presence of neurologic signs or forward slipping of the vertebral body beyond 50% of its width. Other apophyseal injuries in the spine include *slipped vertebral apophysis* or endplate fracture (248,258). This condition may mimic a herniated lumbar disk and is often associated with heavy lifting. Commonly, the inferior apophysis of L4 is displaced into the vertebral canal along with some attached disk material (8). Radiographs reveal a small bony fragment pulled off the inferior edge of the vertebral endplate. A CT scan or MRI reveals an extradural mass. Surgical excision can provide excellent relief of symptoms in those in whom conservative care has failed. Epidural steroids may be used for individuals in whom initial nonsurgical treatment is unsuccessful. Strains of the lower back are less common in children in view of the open iliac apophysis. Children with *iliac apophysitis* usually have a belt-like pain along the muscular attachments of the superior iliac crest (248,259). Lumbar interspinous process bursitis, or *"kissing spines,"* also needs consideration in the young patient, especially those participating in gymnastics or other activities involving hyperextension of the thoracolumbar spine.

Diskitis is a rare condition (occurring in less than 1%) that also causes back pain in children (231). It can be divided into *septic and aseptic* types. Between the vertebrae, the notochord expands to form a gelatinous center of the intervertebral disk called the nucleus pulposus. This nucleus is later surrounded by circularly arranged fibers from sclerotome-derived mesodermal cells called the annulus fibrosis. The nucleus pulposis and the annulus fibrosis together constitute the intervertebral disk. The *intervertebral disk* is *vascular* in children up to 7 years. Around the age of 7, the disk begins to develop some of the end arteries common to the adolescent and adult. From the age of approximately 13 years, all end arteries are thought to be formed and thus, the disk becomes avascular. It may well be that the more vascular nature of the disk is a major reason why diskitis occurs almost solely in children (5,260,261,262). Positive cultures are generally more common in younger children, with *Staphylococcus aureus* by far the most common finding (231). A slower, indolent form of diskitis may develop in a child from brucellosis or tuberculosis. A skin test for tuberculosis may be helpful. Trauma might cause release from the disk tissue enzymes such as *phospholipase A2*, known to be a potent inflammatory stimulator, which could, in theory, cause inflammation. Viral causes are also thought to be present and likely make up a substantial component of the aseptic variety. High fever, toxemia, elevated WBC counts, positive blood cultures, and bone scans in a child under the age of 3 who refuses to sit or stand is a common history. The *diagnosis* must be considered in a child with just mild illness who has abdominal pain or refuses to walk for unclear reasons. Pain frequently

occurs at night, and children are usually not systemically ill (5). An MRI scan shows involvement of the disk space and vertebral bodies one level above and below. The two most serious diseases in the differential diagnosis include *vertebral osteomyelitis*, rare in children, and spinal tumors. Biopsy of the disk space may be necessary, particularly in an adolescent suspected of abusing drugs. Vancomycin may be the treatment of choice or other staphylococcal antimicrobials. When there is no response to early antibiotic therapy, aspiration or biopsy should be performed, followed by culture-specific antibiotic treatment (8). Immobilization of the child may or may not be helpful. *Hematogenous spread* is the most common cause of vertebral osteomyelitis, with *S aureus* the most common organism. Vertebral osteomyelitis generally involves the more anterior aspects of the spine and may be associated with paravertebral collections. Tuberculous spondylitis or *Pott's disease* remains common worldwide and is still seen in some neglected areas of the United States (263).

Gait abnormalities, although frequently benign, can be a great source of parental concern. The child's whole posture needs to be looked at carefully, particularly from the waist downward, because malalignment of any lower extremity joint may stem from another. Figure 10.23 shows anteversion of the femoral head and neck on the femoral diaphysis in addition to coxa valga and coxa vara. The normal angle of the femoral neck and shaft at birth is approximately 160 degrees and decreases to approximately 140 degrees at 5 years and 120 degrees at adulthood. At birth, the normal anterior femoral neck angle relative to the transcondylar line of the distal femur is approximately 40 degrees. This angle decreases to approximately 25 degrees by age 5 and 15 degrees in adulthood (264,265). An increase in the anteversion angle is frequently associated with in-toeing and increased internal rotation best assessed with the child lying prone. Figures 10.24 and 10.25 show normal degrees of internal and external rotation throughout the lifetime within two standard deviations. The degree of femoral neck anteversion is generally thought to be about 20 degrees less than full internal rotation of the hip (5). An estimate of anteversion can be measured by trochanteric palpation with the child prone on the examination table. The degree of internal rotation measured at the point where the greater trochanter is most prominent on the lateral surface of the hip is the estimate of anteversion. In-toeing may persist into adulthood, but often improves with time in the physically normal child by the age of 8. Exercises to strengthen the external rotators of the hip with physical and verbal cues to out-toe and compensate may, at times, offer benefit. This benefit is achieved through facilitating motor memory and improved compensatory strategy to increase out-toeing and not the result of any change in the bony anteversion angle. Severe in-toeing in the able-bodied child, not correcting over time, associated with falls and significantly limited external rotation, can be corrected surgically. Surgery is uncommon, deferred at least beyond the age of 6 years and frequently after 10 years, when there is less chance of postoperative derotation of the corrected torsion. Surgery should not be taken lightly, and good indication should be present along with well-educated parents and child to justify the risk.

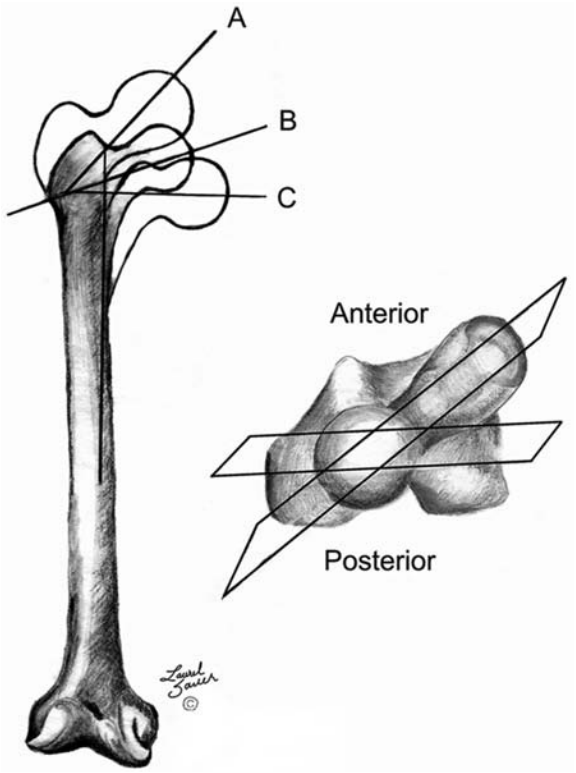

FIGURE 10.23 Angle of neck shaft and anteversion of the femur: (A) increased coxa valga, (B) normal, (C) decreased coxa vara. The smaller diagram shows a top view relating a plane from left to right through the greater trochanter and femoral head referenced to the transcondylar femoral axis distally.

Excessive hip external rotation with minimal internal rotation, often tested with the child lying prone with hips extended (Figure 10.25), is associated with femoral retroversion (opposite of anteversion). This condition can be seen more common in children with low tone and increased joint laxity, such as those with Down syndrome and Ehlers–Danlos syndrome. Gait is with excessive out-toeing, and familial traits may be present. Most rotational variations in children resolve spontaneously with time and minimal intervention (266,267). Careful examination is required to exclude more serious disorders.

Tibial torsion, both internal and external, may occur as compensation for the femoral version or by themselves, causing in-toeing and out-toeing respectively. The transmalleolar axis may be palpated in prone and knee-flexed positions. The lateral malleolus is approximately 5 to 10 degrees posterior to the medial malleolus in the toddler and increases to approximately 15 degrees by adolescence (268). Figure 10.26 (269) shows the normal degree

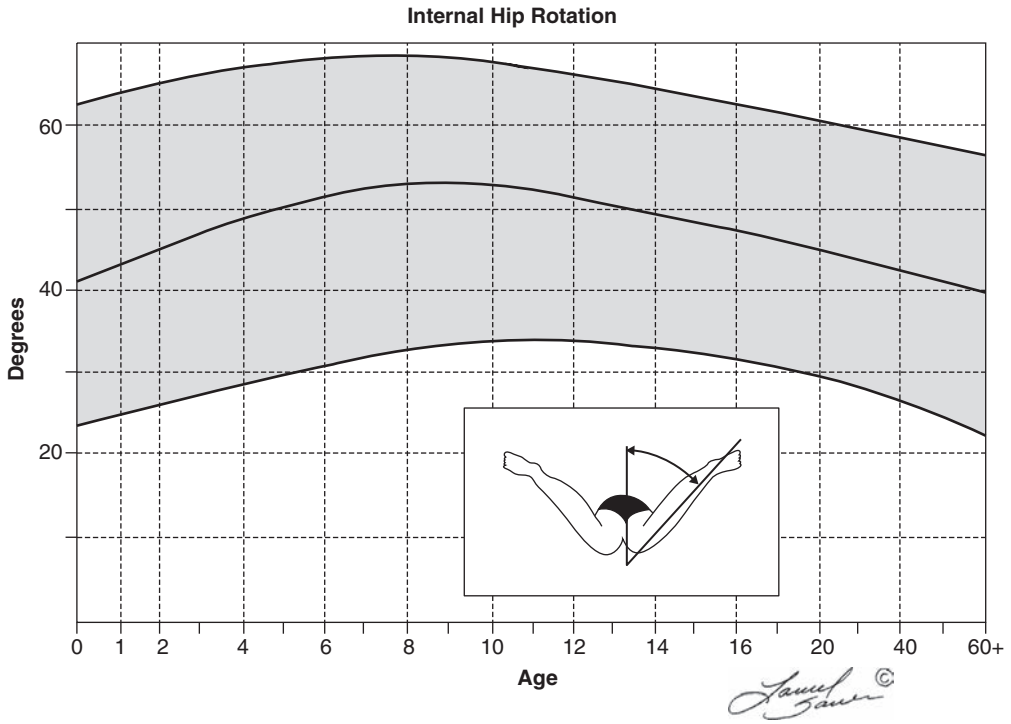

FIGURE 10.24 Hip internal rotation assessed with the child prone. Normal ranges are shaded.

Source: Adapted with permission from Ref. (269). Heath CH, Staheli LT. Normal limits of knee angle in white children genu varum and genu valgum. *J Pediatr Orthop*. 1993;13(2):259–262.

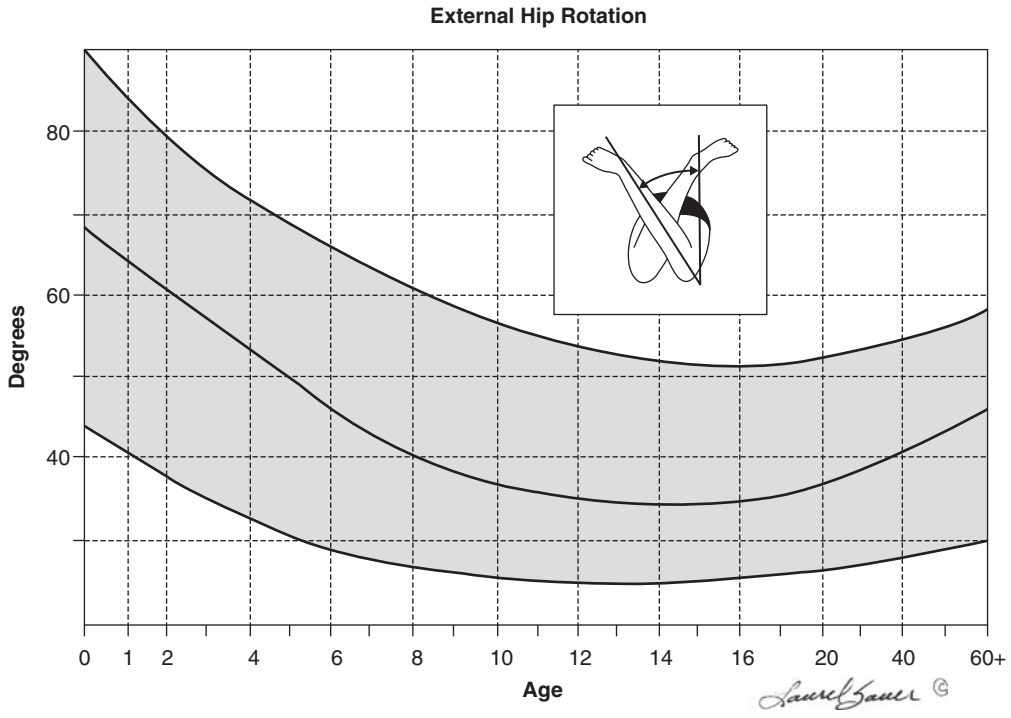

FIGURE 10.25 Hip external rotation assessed with the child prone. Normal ranges are shaded.

Source: Adapted with permission from Ref. (269). Heath CH, Staheli LT. Normal limits of knee angle in white children genu varum and genu valgum. *J Pediatr Orthop*. 1993;13(2):259–262.

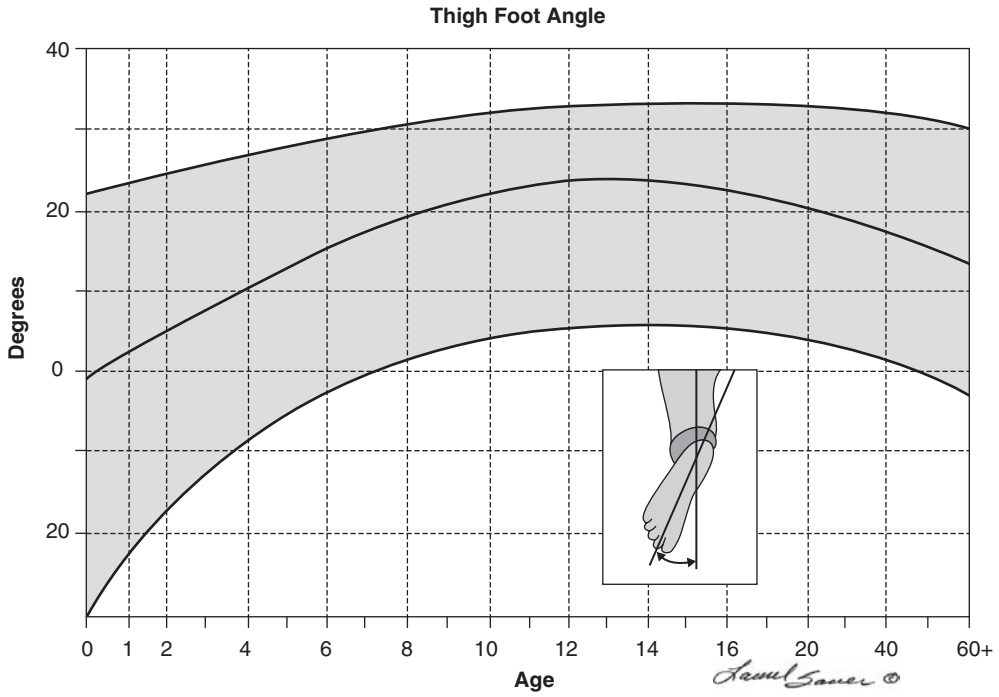

FIGURE 10.26 Rotational status of the tibia assessed by evaluating the child in the prone position. Foot placed in plantigrade neutral position.

Source: Adapted with permission from Ref. (269). Heath CH, Staheli LT. Normal limits of knee angle in white children genu varum and genu valgum. *J Pediatr Orthop*. 1993;13(2):259–262.

of thigh-foot angle over the lifetime within two standard deviations. Denis Browne bars have been found to have essentially no effect in altering tibial torsion and have generally fallen into disuse for this condition (5). In measurement of the thigh-foot angle, the foot is placed into the plantigrade and hindfoot neutral position palpitating talonavicular alignment. This helps eliminate other, more intrinsic foot conditions, such as metatarsus varus and adductus, that can otherwise confound the thigh-foot angle measurement. Figure 10.27 shows normal foot

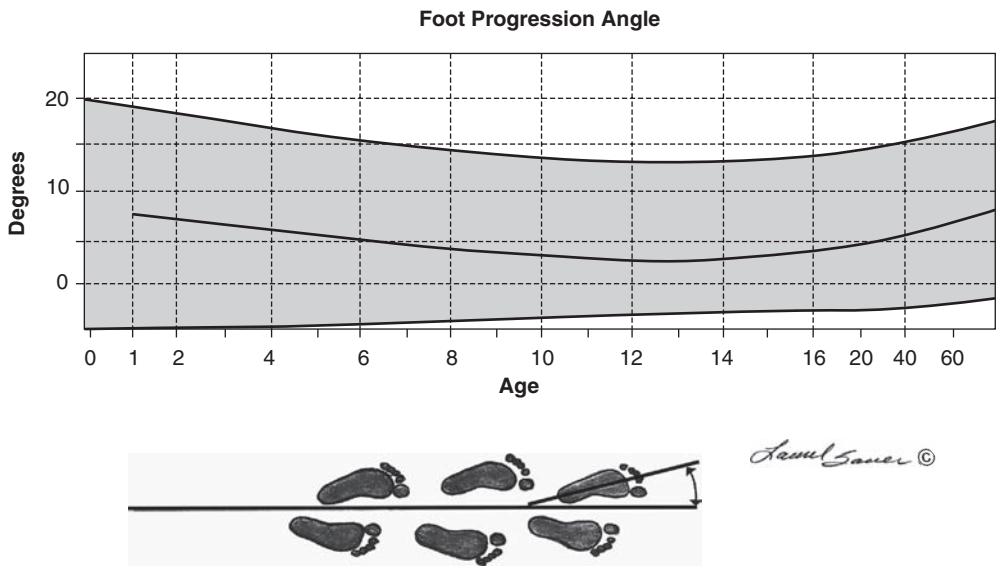

FIGURE 10.27 Foot progression angle. Normal ranges shaded.

Source: Adapted with permission from Ref. (269). Heath CH, Staheli LT. Normal limits of knee angle in white children genu varum and genu valgum. *J Pediatr Orthop*. 1993;13(2):259–262.

progression angle over the lifetime (269). All rotational abnormalities of the lower extremities have influence on the foot progression angle. Flat feet, or pes planus, is no exception. Flexible pes planus is usually asymptomatic, at least in earlier years, and more common than its rigid counterpart in children. Inexpensive scaphoid pads or medial inserts may help to create more plantigrade weight-bearing and improve foot progression angle, but they do not correct the deformity. Extreme cases such as in children with hypotonia may require surgery after the age of 5 years in the form of calcaneal lengthening once bony cortices are more solid. Untreated progression with increased external foot progression angle may occur along with compensatory hallux valgus, planovalgus, and secondary bunion and toe deformities. Pes planovalgus is associated with more active or shortened peroneal musculature, progressing over time, with the development of pain particularly in later years. Rigid pes planus is a congenital deformity associated with other anomalies in 50% of cases, as discussed earlier in this chapter.

Angular deformities of the femoral–tibial alignment are also a source of frequent concern for parents and families. At birth, the infant has a bowlegged posture with a genu varum of 10 to 15 degrees (270,271). The bowing gradually straightens so that the femoral–tibial alignment is neutral or zero degrees by 12 to 18 months of age (5). Continued growth results in a peak valgus angulation of 12 to 15 degrees by the age of 3 to 4 years (8). Subsequent growth reduces the genu valgum to normal adult values of approximately 5 to 7 degrees by the age of 12 years. At any age there is a fairly wide standard deviation of normal (269). Figure 10.28 shows the normal variation of valgus and varus at the knee from 6 months of age through adulthood values (269). Measurements between the medial femoral condyles or intermalleolar distance help to quantitate the deformity (269). The most common cause of genu varum in children is physiologic bowlegs. Children with this condition have genu varum that persists after the age of 18 months, usually resolving before the age of 3 years. X-rays show symmetric growth plate anatomy and medial bowing that involves the proximal tibia as well as the distal femur. Measurement of the metaphyseal–diaphyseal angle in addition to the tibiofemoral angles is helpful in the differential diagnosis (Figure 10.29) (11,272,273,274). The differential diagnosis includes infantile tibia vara or Blount's disease, hypophosphatemic rickets, metaphyseal chondrodysplasia, focal fibrocartilaginous dysplasia, and trauma to the epiphysis. *Blount's disease* occurs in children with no apparent abnormality at birth, having a typical history of genu varum worsening with gait before the age of 2 years. The less frequent juvenile onset may occur between 4 and 10 years and the adolescent form over 11 years. The condition is more frequent in African Americans and girls, and is seen with obesity and in children walking at an early age (275). The condition is also more common in certain geographic locations such as the southeastern part of the United States (214). Classic radiographic changes associated with Blount's disease and tibial varum are seen in the *Langenskiold classification* (276). Blount's disease is believed to result from abnormal compression of the medial aspect of the proximal tibial physes, causing retardation of growth in that area or increased growth laterally of the proximal tibia or fibula (5,277). In juvenile Blount's disease, the etiology is less clear and may relate more to malalignment, leading to the characteristic changes visible on radiographs. Patients with metaphyseal–diaphyseal angles greater than 16 degrees have been reported to experience progression of the angular deformity (5,278). Early and continuous bracing in Langenskiold stage I and stage II disease (276) can achieve good results (8). Bracing should not be initiated after 3 years of age, nor should brace treatment be continued if Langenskiold stage III changes develop (5,279,280). The authors' preference is

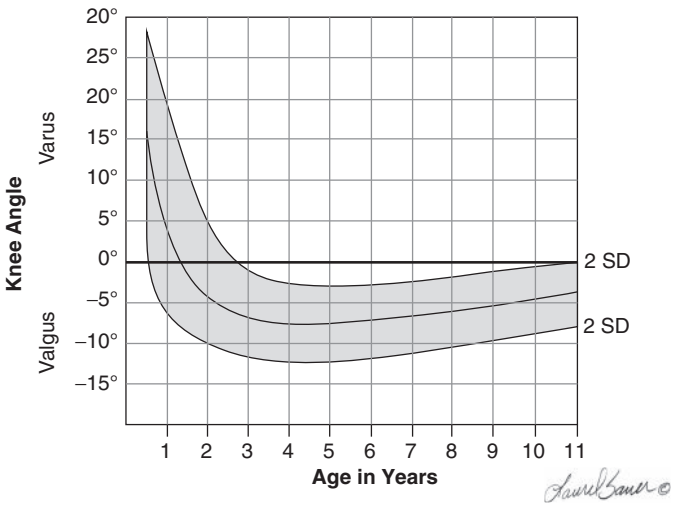

FIGURE 10.28 Normal values for knee angle measured in valgus and varus.

Source: Adapted with permission from Ref. (269). Heath CH, Staheli LT. Normal limits of knee angle in white children genu varum and genu valgum. *J Pediatr Orthop.* 1993;13(2):259–262.

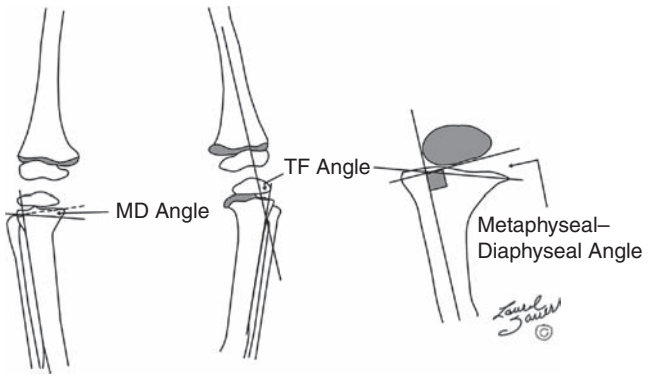

FIGURE 10.29 Measurement of metaphyseal–diaphyseal angle and tibiofemoral angle.

a medial upright knee ankle foot orthosis (KAFO) with valgus padding pulling lateral to the medial through the center of the knee axis with free-swinging knee and ankle. Proximal valgus osteotomies may be required for severe persistent angular deformity after the age of 3 years, along with consideration of *Ilizarov techniques* (8). Stapling of the lateral physis (often both tibia and femur) are also considerations, particularly in the adolescent prior to cessation of growth. Increased fragmentation, declination, and beaking of the medial-proximal tibial epiphysis generally indicate the need for surgery. Surgical complications can include compartment syndrome with persistent neurovascular compromise. Careful postsurgical follow-up of the child is required to prevent unnecessary over- or undercorrection. Graphs for timing of hemiepiphysiodesis are available and can be helpful in experienced hands (281).

Genu valgum, or "knock-knees," is a concern in children who are developing peak valgus alignment around the ages of 3 to 4 years. Almost 99% of the time, this valgus is benign in nature, correcting toward adult values by early adolescence. X-rays show symmetric growth plates with no particular abnormalities. Observation is the treatment of choice in these individuals. Children who have genu valgum with a tibiofemoral angle greater than 20 degrees require follow-up, but generally the problem resolves spontaneously. If abnormal genu valgum persists into the teens, correction by hemiepiphysiodesis or stapling of the medial physis may be effective (8,270). Staples that are placed extraperiosteal for varus or valgus deformity allow for growth to resume once removed. Rebound phenomena can be anticipated, undoing some of the corrected valgus or varus. Overcorrection slightly in anticipation of this problem, especially in children under the age of 12, needs to be considered (214).

Idiopathic toe walking is a common condition in children under 3 years of age. By 3 years of age, children should walk with a heel strike (282,283,284). Persistent toe walking beyond this age is abnormal (5). Little is known about the natural history of idiopathic toe walking, with most individuals improving or showing resolution prior to the age of 6. Persistent toe walking in the older child and young adult can result in leg pains, more activity-related, and frequently in the anterior tibial or knee regions. Toe walking can diminish or cease with time, as body mass becomes too large to be supported by the triceps surae or as a result of secondary development of external tibial torsion (285,286). Toe walking developing sometime after birth can be associated more with problematic conditions, such as muscular dystrophy, dystonia, tethered cord syndrome, central nervous system neoplastic processes, or autism (11,287). A family history is often positive along with that of prematurity and a slight male predominance (5,287).

Leg pains in children are generally benign, but need to be followed carefully for signs of progression or persistence despite conservative care. Conservative care, generally involving the RICE protocol, NSAIDs, and warm baths and massage, often relieves most of the discomforts. A pattern of increased pain with activity or recreational pursuits is common. If improvements are not noted within a few weeks of conservative care, additional workup is required to rule out other, more concerning entities. Workup should include radiographs, hematology and metabolic parameters, ESR, possible nuclear medicine scan, and Lyme disease titer along with other inflammatory and rheumatologic markers.

In children who toe walk, walking is generally not delayed as a developmental milestone, and when this occurs, conditions like *spastic diplegia* should be *considered*. A few beats of clonus at the ankle can be helpful in differentiating associated mild diplegia from idiopathic toe walking. Nonoperative treatment, including heel-cord-stretching routines with the calcaneus midline or inverted, can be helpful when performed on a regular basis along with dorsiflexion-strengthening exercises. Stretching a tight heel cord with the hindfoot in valgus can contribute to mid-foot breakage while being ineffective in lengthening plantarflexion soft tissues. Articulating AFOs with plantarflexion blocks or posterior leaf-spring types can be helpful in maintaining position both day and night. *Serial casting* can be an option for resistant equinus deformity not felt to be surgical at the time. Casting should occur with progressive dorsiflexion as tolerated, again with the heel in a neutral or slightly inverted position. Two or three sets of short-leg casts of a walking nature, lengthening the heel cord, can result in greater passive dorsiflexion. Short-term weakness of the anterior tibialis and dorsiflexors can be anticipated postcasting requiring additional strengthening intervention. *Clostridium botulinum* toxin A injections can be helpful also in weakening partially the plantarflexors, facilitating improved stretch into dorsiflexion along with relative strengthening of active dorsiflexion. Orthotics can be weaned over 3 to 6 months once toe walking has resolved and improvements obvious. *Nighttime splinting* can be discontinued in the absence of recurrent toe walking. Surgical intervention, including heel cord lengthening and/or gastrocsoleus recession, is reserved for those who have failed conservative trial. Toe walking after the age of 6 years often does not improve, and heel cord contractures can worsen (5). External tibial torsion can progress further, developing as compensation for lack of foot-flat contact. The torsion may become severe enough with excessive external foot progression angle to warrant corrective osteotomy. Surgical lengthening is performed sufficiently to obtain 10 degrees of dorsiflexion with the knee extended (5,8). *Overlengthening of the heel cord* can be disastrous, resulting in persistent crouched gait and associated pain syndromes and limitations. In more severe and chronic equinus deformities, posterior ankle capsular release may be required. Short-leg casting postoperatively is

common up to 6 weeks followed by custom-molded AFOs for up to 2 months thereafter. Home exercise, along with physical therapy for gentle heel cord stretching and strengthening ankle dorsiflexion, is mandatory or recurrent equinus deformity can be anticipated. Long-term outcomes of surgical lengthening in skilled hands are generally positive when recommendations are followed with satisfactory heel–toe walking over the lifetime (286,288,289).

SCOLIOSIS

OVERVIEW

Scoliosis is a complex three-dimensional (3D) deformity of the spine defined by a frontal plane deformity greater than 10 degrees. It is classified into congenital, idiopathic, neuromuscular, and functional types (Table 10.6). While the etiology, onset, prognosis, and treatments vary between classifications, the possible outcomes of severe untreated scoliosis are the same: respiratory compromise, seating compromise, pain, gait impairment, difficulty with ADLs, and psychological distress (290,291). Understanding the natural history and available interventions is important in helping patients achieve long-term comfort and functionality.

EMBRYOLOGY, GROWTH, AND CURVE PROGRESSION

Spinal development is a complex process, which begins in the first month of gestation when mesoderm cells surrounding the notochord begin to differentiate into sclerotomes. These will ultimately form vertebral bodies and arches. Injury in early gestation often impacts other nearby organs, primarily the cardiac, renal, and gastrointestinal systems. Approximately 60% of those with spinal anomalies have other congenital malformations, so it is essential to screen for these (292).

The thoracic spine accounts for 63% of thoracolumbar growth, but because the lumbar vertebras grow more segmentally, early fusion of the lumbar spine will result in a greater loss of total height (293). Fortunately, newer surgical techniques that allow curve correction with continued growth and avoid early fusion are widely available (294).

TABLE 10.6 TYPE OF SCOLIOSIS WITH CATEGORICAL DESCRIPTION

TYPE OF SCOLIOSIS	CAUSE	SEX/AGE OF ONSET (YR)	COMMON ASSOCIATED CURVES	COMMON CHARACTERISTICS
Functional	Nonstructural, secondary to leg-length discrepancy, herniated disk, trauma, arthritis	Any	None	Resolves with correction of underlying cause
Congenital	Failure of somite formation or segmentation 60% have other anomalies	Birth, but delayed diagnosis possible	None	Progressive tendency, surgery more likely
Idiopathic				
Infantile (<1%)	Positional contributions	Male predominance, <3 years	Left thoracic	May resolve spontaneously
Juvenile (19%)	Etiology unknown	Male = Female, 3 to 10 years	Any	Aggressive traits, surgery more likely
Adolescent (80%)	Multifactorial, polygenetic	Female > Male Females progress more often, >10 years	Right thoracic	10% require treatment (bracing > surgery)
Neuromuscular	Upper or lower motor lesions, myopathic processes	Any age	Long sweeping typical	Aggressive, less responsive to bracing, progress after maturity

The relationship of scoliosis to growth has been well established, and screening programs and surgical interventions are best planned with these in mind. Unlike limb growth, vertebral growth is nonlinear. Two major growth spurts typically occur: the first before the age of three, and the second during puberty.

The *Tanner* 5-stage classification is based on physical signs of pubertal development and can help predict growth spurts (295). Growth typically accelerates in girls at Tanner stage 2, and in boys at Tanner stage 3, although race, heredity, physical activity, physical disability, and nutrition may impact this.

Risser lines, seen in posterior–anterior (PA) radiographs of the iliac crest, are used in staging skeletal maturity and predicting growth as well (Figure 10.30). Ossification of the iliac crest is tracked as it proceeds from lateral to medial, ranging from Risser grade 0 (no ossification) to grade 5 (complete fusion to iliac apophysis) (296). Risser 1 represents the period of most rapid skeletal growth, and correlation of the Risser sign with the degree of a scoliotic curve can be predictive of curve progression.

The *Sanders digital maturity stage*, based on hand and wrist radiographs, also correlates highly with progression of idiopathic scoliosis, and may be more accurate than Risser staging (297,298). The *Lonstein–Carlson nomogram* uses initial curve magnitude, Risser sign, and chronological age to predict curve progression (299). Guidelines to inform treatment options in idiopathic scoliosis were developed by Society on Scoliosis Orthopedic and Rehabilitation Treatment (SOSORT) based on this information (300).

CURVE CLASSIFICATION AND NAMING

Scoliosis curves are named by their direction, location, and magnitude. The curve's convex apex indicates its named direction and location, and measurement by the Cobb angle provides its most reliable magnitude (Figure 10.31) (301). If more than one curve exists, the largest degree curve is designated as major and the others minor (Figure 10.32). Curves over 100 degrees are associated with restrictive lung disease.

Rotational deformities complicate bracing and surgical correction. Rotation of the spine is measured using a scoliometer when the child is bending forward (Adams test), and radiographically by pedicle visualization (Nash–Moe x-ray method) or by CT scan (302,303).

HISTORY, PHYSICAL EXAM, AND TREATMENT OVERVIEW

The history and exam for scoliosis patients vary depending upon age and associated diagnosis. Reflexes, strength, sensation, ROM, general posture, muscle bulk, and gait must be examined. Seating systems and assistive devices should be assessed, as improper walker or crutch height, and truncal weakness with poor seating support can impact spinal position in children with disabilities.

A positive family history is particularly pertinent in congenital and idiopathic scoliosis. The presence of back

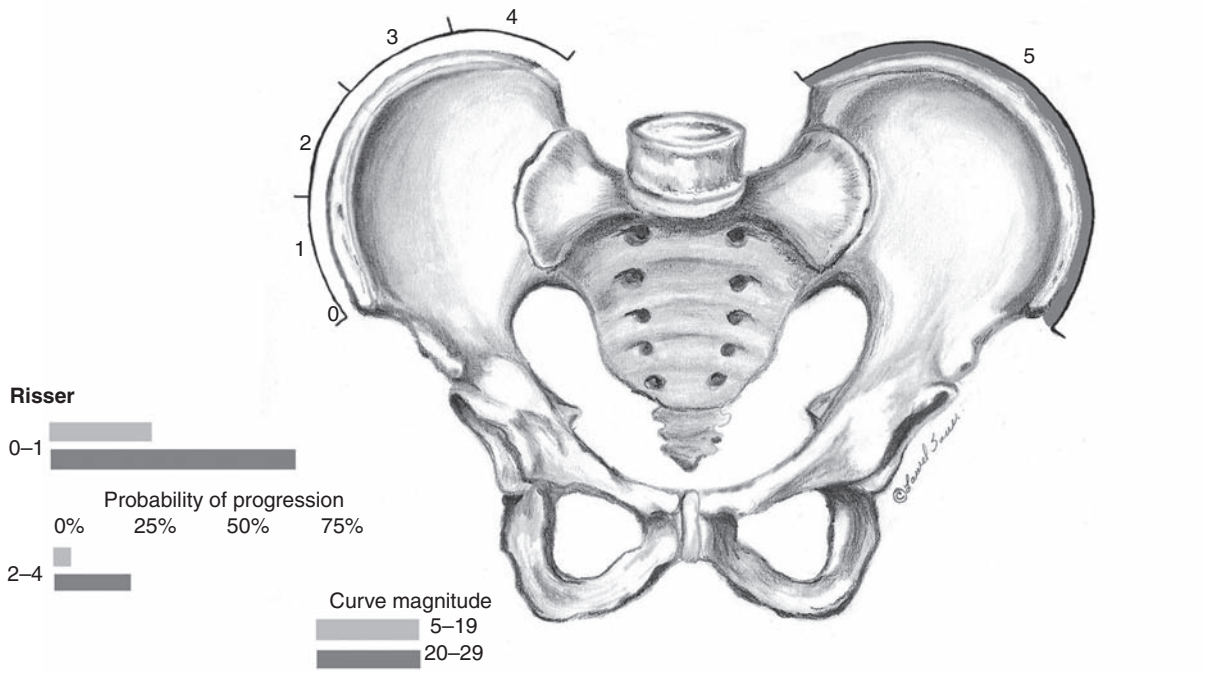

FIGURE 10.30 Risser sign. Likelihood of progression is based on the Risser sign and curve magnitude.

Source: Adapted from Ref. (299). Lonstein JE, Carlson JM. The prediction of curve progression in untreated idiopathic scoliosis during growth. *J Bone Joint Surg Am*. 1984;66(7):1061–1071.

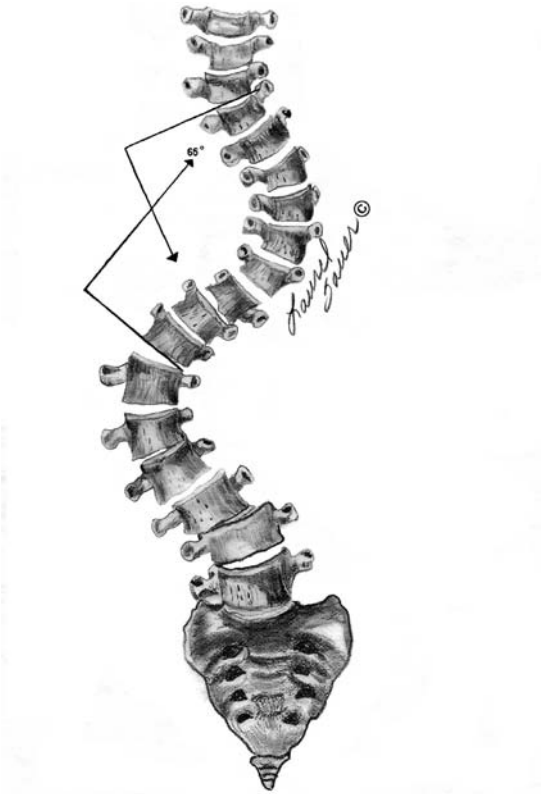

FIGURE 10.31 The Cobb method of measuring curvature in scoliosis. The angle measured is formed by perpendicular lines drawn through the superior border of the upper vertebra and the inferior border of the lowest vertebra of a given curve.

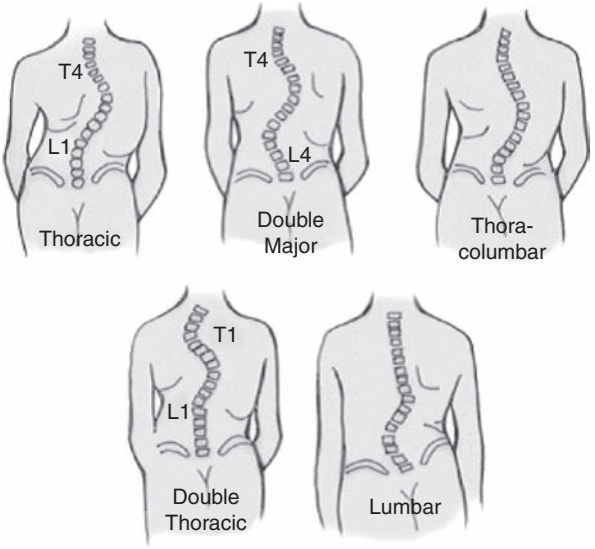

FIGURE 10.32 Classification of scoliosis. Scoliosis is classified into general categories by level.

Source: Adapted with permission from Ref. (296). Staheli L. Practice of Pediatric Orthopedics. 2nd ed. Philadelphia, PA: Lippincott Williams & Wilkins; 2006.

pain may indicate a serious diskitis or tumor, while rapid curve progression, bowel and bladder changes, recent trauma, associated weight loss, muscle weakness, or joint pain can point to other serious primary processes. These might include spinal cord syrinx or tethered cord, spinal fracture, rheumatologic disease, and osteoblastoma. Presence of café au lait spots, webbed neck or low hairline, and hairy patches or skin dimples may lead to recognition of disorders such as Klippel–Feil, spina bifida occulta, or neurofibromatosis.

Excessive height, arm span, or joint hyperextensibility may signal a connective tissue disorder of which scoliosis is only a presenting symptom. Excessive lordosis or kyphosis, LLDs, and limited hip or hamstring ROM could indicate hip dysplasia or an underlying muscle or neurologic disorder such as hemiplegia, herniated disk, spondylolisthesis, or dystrophy.

Pelvic obliquity, asymmetry of the scapula or shoulder girdle, or asymmetry at the waist triangle as the arms hang down should be noted. Asymmetrical prominence of the rib cage of over 7 degrees by scoliometer, seen on forward bending with the feet and palms together (Adam's test), warrants further investigation.

Side bending may help assess the flexibility or rigidity of a curve, which is important when considering treatment options. Decompensation, another sign of scoliosis, can be measured by dropping a plumb line from the C7 spinous process and noting if it passes through the gluteal cleft (normal) or deviates laterally (292).

Curve Documentation

A full spine PA radiograph (less radiation to sensitive tissues than AP radiograph) is usually appropriate for screening purposes, although certain curves (ie, congenital, infantile) may require CT or MRI evaluation. Curves with significant rotational components or kyphosis may require lateral views. Radiographs should be taken standing if possible, with Cobb angle and Risser noted, and if wearing an orthosis, done both *in and 24 hours out of any brace*. Neuromuscular curves may progress after maturity, so continued screening may be warranted (296).

Radiation Exposure

A typical child with scoliosis incurs an average of 23 radiographs over his or her treatment, inciting a significantly higher risk of breast, lung, and ovarian cancer (304). Low-dose radiographs can minimize exposure, but four-dimensional (4D) surface topography (ie, Diers Formetric) is gaining popularity and availability in the United States. Used extensively in Europe, topography methods

utilize no radiation and can monitor curves and their progression reliably, limiting routine x-rays (305,306).

Orthotic Management

There is now definitive evidence that brace treatment can be extremely effective in reducing curve progression in some types of scoliosis (298,307). The purpose of orthotic wear is to stop progression during high-risk growth periods, typically beginning when curves reach 20 degrees, although age and other risk factors may indicate earlier bracing. When recommended, orthotics should be as low profile and comfortable as possible to encourage compliance. Curve location impacts choice of brace and materials used. Wearing times of 18 hours a day are needed to best delay curve progression, and compliance "chips" can be inexpensively added to orthosis to document wearing time for family, patient, and physician feedback. Brace wear continues until maturity, or surgical intervention is required. Bracing curves over 45 degrees can still be effective (307).

Many types of orthosis exist and an experienced orthotist is essential (Figure 10.33). The Charleston

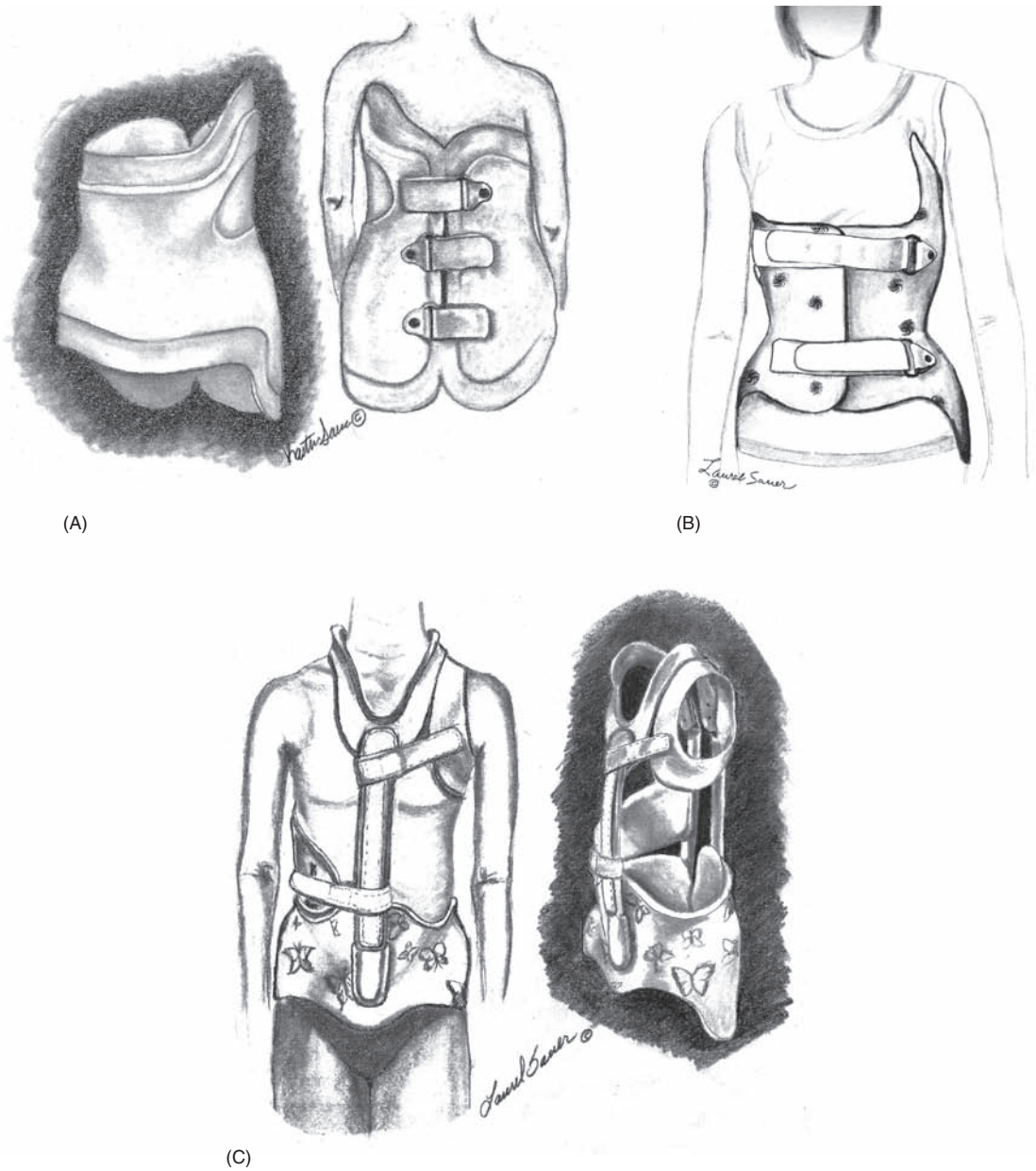

FIGURE 10.33 Types of braces. These are common braces and generalizations about their use. (A) Charleston: least obtrusive, nighttime only most curves. (B) TLSO (Cheneau): moderate, 16–22/24 hours, most curves. (C) Milwaukee: most difficult to accept, 16–22/24 hours, high curves.

Source: Adapted from Ref. (296). Staheli L. Practice of Pediatric Orthopedics. 2nd ed. Philadelphia, PA: Lippincott Williams & Wilkins; 2006.

braces, worn only at nighttime, overcorrect away from a low-magnitude, low-risk curve. The Boston brace, used most commonly in the United States, is easy to fabricate, but typically has a back closure making it difficult to independently don and doff. The Lyon, Rigo, Cheneau, and SPoRT braces, all used commonly in Europe, vary in their weight, difficulty in fabrication, and style, but have shown good benefit for scoliosis correction. The Milwaukee brace cervical thoracic lumbar sacral orthosis (CTLSO) can still be effective for curves above the T7 apex (308). Compliance in wearing braces can be low and thus their utilization is decreasing in certain centers. All brace styles should be considered to achieve best results.

It is up to the family, physiatrist, and orthotist to help optimize a child's function within a brace, and provide support and encouragement. Studies show that while there may be short-term distress at the thought of brace wear, there is not a significant negative psychological impact in the long run (309). Peer led support groups such as "Curvy Girls - Scoliosis" can help adolescents understand their diagnoses, make healthy lifestyle choices, improve self-esteem, and assist in finding practical answers to questions such as brace compatible clothing.

Scoliosis-Specific Exercise (SSE)

For patients with the desire and ability to cooperate with a therapy program, *scoliosis-specific exercises* (SSEs) can be beneficial in improving awareness of body alignment, reducing pain, improving pulmonary dysfunction, and addressing psychological stress, while also attempting to slow down curve progression. Many certification programs and training schools exist (Shroth, SEAS, FITS), all predominantly of European origin. In the United States, there is limited exposure to SSE by physical therapy training programs (310). Patient exercises are done in an outpatient setting with home follow-up. Flatback and rotational deformities are targeted, with axial elongation, derotation and stabilization emphasized (308). While there is a lack of high-quality research regarding SSE, patient satisfaction is typically high, and guidelines for the appropriate use of exercise have been published (300).

Surgical Interventions

Surgical techniques vary by type and severity of scoliosis, but continuous intraoperative spinal monitoring (somatosensory evoked potentials [SEPs], motor evoked potentials) are important for preventing neurologic injury during all surgeries (303). Achieving a balanced spine, a solid arthrodesis, and a reduction in the deformity is the primary goal. The most common complications of surgery are infection, pseudarthrosis, anemia, hypotension, and hardware failure.

Due to concerns over the loss of spinal height and the impact that early fusion of the thoracic spine may have on long-term pulmonary function, numerous "non-fusion" surgical technologies have been developed. The use of "growing rods" that are anchored proximally and distally to the spine, allows curve correction, but can be lengthened twice a year to allow growth. Vertical expandable prosthetic titanium rib (VEPTR) placement involves uni- or bilateral rods, anchored proximally on a rib and distally on the pelvis. Biannual adjustments allow curve correction and growth, and can be used in neuromuscular curves as well as children with congenital rib anomalies (294,311,312).

If a definitive fusion is needed, posterior fusions are preferred, but AP procedures may be done in young children to prevent the risk of crankshaft deformity (overgrowth of posterior elements if only anterior fusion is done). Instrumentation compatible with MRI technology is essential given the comorbidities many children have.

TYPES OF SCOLIOSIS

Congenital Scoliosis

Congenital scoliosis accounts for approximately 20% of all scoliosis and is due to prenatal disruption of vertebral formation (hemivertebra, wedge vertebra) or vertebral segmentation (block vertebra, unilateral bar). A single hemivertebra is the most common anomaly. A positive family history may be present, with 5% to 10% of siblings having a similar disorder (293).

While congenital scoliosis may not be clinically evident until later in life, problems related to defective organogenesis may lead to its early detection. Abnormalities of the trachea, esophagus, renal tract, gastrointestinal tract, lungs, heart, radius, ear, lip, and palate often accompany congenital scoliosis. Up to 25% of children may have renal disorders, 10% may have cardiac problems, and 30% may have spinal dysraphism (292,313). Scoliosis is a primary symptom in vertebral defects, anal atresia, tracheoesophageal fistula, radial and renal dysplasia (VATER) syndrome, and thoracic insufficiency syndrome.

Unilateral, unsegmented bars that restrict growth on one side of the spine while the other grows normally, especially in the thoracic area, produce curves that are the most likely to progress. If in the cervical area, torticollis may be a presenting symptom (303). Unsegmented block vertebra, as seen in Klippel–Feil syndrome, generally do not produce a progressive scoliosis, but restrict ROM (296). Avoiding activities that place these patients at risk (diving, contact sports) is important.

MRI of the brainstem and entire spine provides the best evaluative tool to visualize not only bony abnormalities, but spinal cord dysraphism (diastematomyelia, lipoma, hydromyelia) that may coexist, yet not be evident

on routine radiographs (313). Myelography is rarely used. Close monitoring every 3 to 6 months until age 4 and again in the adolescent years is essential (292).

TREATMENT. Typically, orthoses are ineffective, except perhaps in small-degree, long, flexible curves. If an orthosis is used, the family, physiatrist, and therapist must work to encourage the child's acquisition of developmental skills through adaptive activities that accommodate their reduced spinal range. Maintaining cardiovascular health and endurance is especially important prior to surgery.

Approximately 50% of children with congenital scoliosis require surgical intervention at an early age, before spinal rigidity or secondary pulmonary deficiencies occur (296). Surgical options are aimed at prevention of deformity. They include hemivertebra excision, convex growth arrest (hemifusion), fusion with instrumentation and allograft, or instrumentation without fusion (sparing growth). Due to concerns over the loss of spinal height, and the impact that fusion of the thoracic spine may have on long-term pulmonary function, numerous nonfusion technologies have been developed.

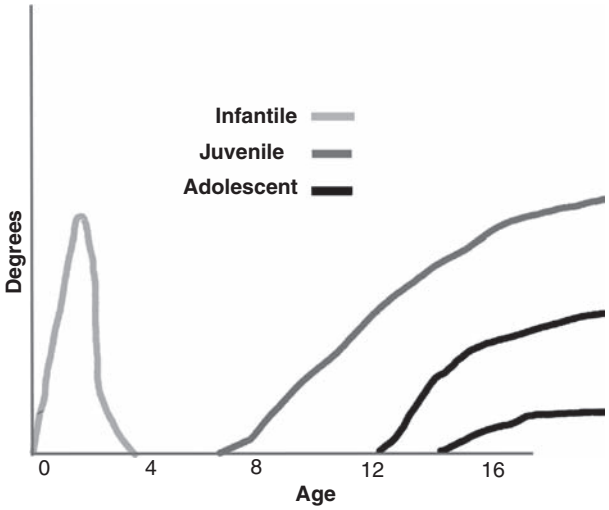

FIGURE 10.34 Natural history of idiopathic scoliosis. Progression is related to the age of onset of the scoliosis.

Source: Adapted from Ref. (296). Staheli L. Practice of Pediatric Orthopedics. 2nd ed. Philadelphia, PA: Lippincott Williams & Wilkins; 2006.

Congenital Kyphosis

Congenital kyphosis is most common at T10-L1 and due to a failure of vertebral segmentation and/or formation. It may accompany myelomeningoceole or spinal dysraphism, and progressive deformity may lead to paralysis. In surgical correction of curves less than 50 degrees, a posterior fusion is done, whereas larger curves may require AP fusion and/or kyphectomy (292).

Idiopathic Scoliosis

More than 80% of scoliosis cases belong to the idiopathic category, which is subgrouped into three types defined by age of onset (Figure 10.34). All differ significantly in demographic distribution, progression, and treatment type.

INFANTILE. Infantile scoliosis is rare in North America and has an unknown etiology. It occurs within the first 3 years of life and often spontaneously resolves, as there is no anatomic anomaly. It has significant geographic variance, with a higher incidence in European subpopulations known to keep their infants in a supine or lateral decubitus position (292). Left thoracic curves are common in infantile scoliosis, and boys are predominantly affected. Plagiocephaly, DDH, and congenital muscular torticollis are often associated, so radiographs of the spine and hips and MRI of the brainstem and spinal cord should be obtained (303). Neuromuscular disease, congenital scoliosis, and intraspinal pathology (Chiari malformation, tumor) must be ruled out.

JUVENILE. Juvenile scoliosis appears equally in males and females between the ages of 3 and 9 years, and is unfortunately very aggressive with about 70% of children requiring treatment—either bracing (50%) or surgical intervention (50%) (292). Tumors or spinal abnormalities may be causative and an MRI of the spine and brainstem, along with radiographs, is necessary for a thorough evaluation.

Scoliosis-*specific* exercises can be beneficial, and bracing may be effective if started early. Surgical intervention with "growing rod" systems is often recommended once the curve reaches 50 degrees. Physiatry emphasis is on social, academic, and physical integration both pre- and postoperatively.

ADOLESCENT. Adolescent idiopathic scoliosis (AIS) develops at or after age 10, and is the most common form of scoliosis, occurring in about 3% adolescents. Fortunately, most curves are fairly benign and only about 10% require treatment other than observation (303). Females are more commonly affected, and their curves, often right thoracic, progress more quickly. A left thoracic curve, especially in an adolescent male is suspicious and causality should be thoroughly investigated. The exact etiology of AIS is unknown, but genetics play a role as about 30% of patients have a positive family history, and there is 50% concordance among twins (293,314).

SCREENING. With recent confirmation of the effectiveness of early bracing, there has been a renewed call for scoliosis screenings: two for females at ages 10 and 12, and one for males at ages 13 and 14 (315,316). Currently,

adolescent scoliosis screening is mandated by fewer than half of U.S. states due to concerns over cost, low incidence, risk of radiation exposure, and the anxiety involved in what many have termed "schooliosis." While a diagnosis of scoliosis creates enormous anxiety for adolescents, it is entirely treatable, so early detection by pediatricians, sports trainers, physiatrists, and so on is essential.

CURVE PROGRESSION. Curve progression depends on several factors including age of onset, curve size, and level of skeletal maturity. Young (<12 years), premenarchal, skeletally immature (Risser < 2) females with large curves (>20 degrees) are at the most risk.

Benefit from early bracing and scoliosis-specific exercises has been observed, both in working to delay curve progression, and to psychologically allow the patient some active participation in his or her treatment. Bracing is recommended for curves over 20 degrees in a skeletally immature patient in whom 5 to 10 degrees of progression have been noted over a 6-month period. Braces should be worn 18 to 24 hours a day until skeletal maturity is reached (298). This is typically at either Risser stage 4 in girls or stage 5 in boys, or 2 years postmenarche (317). Brace wear is discontinued in lieu of surgical intervention if the curve reaches 50 degrees or more rapid progression is noted.

Treatment in adolescence is important because psychological distress and social limitations have been noted in adult females who have disfiguring curves greater than 40 degrees (291). Curves less than 30 degrees at skeletal maturity do not tend to progress in adulthood, but curves over 50 degrees, especially in the thoracic area, do. Progression of just one degree per year in adulthood can lead to degenerative changes that may become painful, and rigid curves in osteopenic adults are difficult to treat surgically.

SURGICAL INTERVENTION. Curve classification systems such as the King–Moe and Lenke may be utilized for surgical planning in idiopathic scoliosis (303). Operative interventions vary with the number, location, and size of curves, and the child's skeletal maturity. For the typical right thoracic curve of 40 to 50 degrees in a skeletally immature female, a posterior spine fusion is recommended. Harrington rods have largely been replaced by more image guided or computer assisted spinal instrumentation, which may provide better 3D correction. For thoracolumbar or lumbar curves, anterior fusion may offer an advantage of reducing the number of levels fused.

AP fusions are often needed for severe curves over 60 to 70 degrees, for rigid curves (do not improve to less than 50 degrees in bending), and for skeletally immature children who are at risk for crankshaft deformity (292). However, the use of new and stronger pedicle screws may allow the anterior portion of the fusion to be deferred. In adults with untreated idiopathic scoliosis, AP fusions are often needed for correction, with possible spinal cord decompression taking place as well.

Neuromuscular Scoliosis

CURVE TYPES. Scoliosis in children with cerebral palsy, muscular dystrophy, or spinal cord injury is common, with long sweeping curves that begin early and progress quickly often seen. Young, nonambulatory patients with thoracolumbar curves are at greatest risk for curve progression. In children with myelomeningocele, rapid progression of scoliosis may be indicative of a tethered cord, worsening hydrocephalus, or hydromyelia. In children with neurofibromatosis, intraspinal tumors may develop. In both instances, MRIs should be obtained.

Intrathecal baclofen (ITB) therapy is commonly used for spasticity in children with cerebral palsy and neurologic impairment, who may have concomitant scoliosis. ITB therapy has not been noted to have a significant impact either positively or negatively on curve progression (318). Due to the entrance of the intrathecal catheter at the thoracolumbar junction, care needs to be taken when fusing the spine or placing a pump after fusion.

In Duchenne muscular dystrophy, scoliosis is often relentless and progresses at up to 8 degrees per year. The use of oral steroids such as deflazacort to slow the decline in muscle strength and delay nonambulatory status can significantly attenuate the development of scoliosis and need for spinal surgery (319).

As neuromuscular curves over 50 degrees may continue to progress at a rate of 1.5 degrees per year even after maturity, the long-term advantages of early surgical intervention need to be discussed so that valid anticipatory guidance can be given (292). Advancing age, reduced bone quality, a more rigid curve, limited respiratory reserve, and impaired skin integrity can adversely affect the outcome.

NONSURGICAL MANAGEMENT. Orthoses are often utilized in idiopathic curves of less than 40 degrees, although they are often not as effective in slowing curve progression (320). Often neurologic, pulmonary, cardiac, or gastrointestinal comorbidities impact the child's ability to wear spinal orthosis, so careful surveillance of curve progression is important. However, orthosis may improve trunk control, sitting, and head position, and serve a functional purpose. Often, a softer foam orthosis, rather than one of rigid orthoplast, will be more tolerable to the patient and have less adverse impact on pulmonary function (321).

SURGICAL INTERVENTION. Neuromuscular scoliosis differs from idiopathic scoliosis due to the likelihood of concurrent pelvic obliquity, long sweeping curves, and the presence of osteopenic bone, which all complicate surgical intervention. A comparison of the surgical hospitalizations of children with neuromuscular scoliosis to those with idiopathic curves shows their stays to be longer, more complicated, and more costly (322).

Lengthier fusions extending from T2 to the pelvis are common in nonambulatory patients. Pelvic stabilization is avoided if possible in ambulators to reduce problems related to limiting lordosis.

Posterior fusions are preferred, as this bone is more stable and there is less difficulty accessing the spine. Anterior fusions produce sympathectomies and are associated with superior mesenteric artery syndrome (293). AP fusions are often done for severe curves (>60 degrees), although this may involve a two-stage procedure and may not improve correction that significantly. While safe, effective, and at times necessary due to a patient's medical stability or surgeon's skill, staging can increase cost and length of hospital stay (323).

Partial or complete vertebral body resections or fusions may be necessary to achieve stability in children with spina bifida (292).

Surgical intervention needs to be timed to maximize pulmonary status (forced vital capacity [FVC] > 35%) and curves of 20 to 30 degrees are not uncommonly corrected in order to improve seating and respiratory function early on (292,324). The increased risk of anesthesia-induced malignant hyperthermia in patients with Duchenne muscular dystrophy and mitochondrial disease needs to be recognized.

PHYSICAL MEDICINE & REHABILITATION (PM&R) ROLE. Functional goals of maintaining sitting tolerance, cosmesis, transfer capabilities, pulmonary and gastrointestinal function, and skin integrity are typical long-term concerns that should be considered (325). Preoperative nutritional and health optimization, and perioperative infection and pain control are important for successful spinal surgery (326). Postoperative nutritional supplementation, pressure sore vigilance, pulmonary toilet, gastrointestinal motility, aspiration prevention, and rapid upright sitting posture and mobility to prevent deep venous thrombosis are needed.

Revision of seating systems, assessment of equipment needs, and physical therapy needs should be addressed proactively by both the physiatrist and the surgeon in order to achieve best outcomes.

Despite the challenges of surgical correction in children with neuromuscular scoliosis, studies show that curve degree, lung function, seating position, and ADLs may all improve postoperatively, potentially improving the quality of life and caregiving abilities (290).

Functional Scoliosis

Functional or "secondary" scoliosis is a flexible, nonbony curve secondary to LLD, herniated disk, spondylolisthesis, diskitis, muscle spasm, trauma, arthritis, or hip disease. Treatment of the underlying problem typically resolves the curve.

LEG-LENGTH INEQUALITY

Leg-length inequality is common, with estimates of up to one-third having a 2-cm or less discrepancy measured between the length of their legs (5,8,327). There are two basic types of LLDs: true and apparent. True LLD is present when bilateral leg-length measurements between the greater trochanter and the medial malleolus demonstrate shortening on one side. Apparent LLDs are present when bony lengths are the same but joint alignment or pelvic femoral asymmetry is present (eg, adductor spasticity, pelvic obliquity). Apparent discrepancies can best be measured using a tape from the umbilicus to the medial malleolus of either side.

Radiographic measurement is the most reliable. The *scanogram technique* avoids magnification by taking separate exposures of the hip, knee, and ankle so that the central x-ray beam passes through the joints, giving true readings from scale (Figure 10.35) (328,329). CT scanogram is still the standard, reducing errors from angular deformity (330). If the examination is done specifically for this purpose, economic cost can be competitive (multiple sections unnecessary) and radiation exposure less with the microdose technique (331,332). Causes of true LLD are many and can be classified by growth retardation versus growth stimulation (5,8). Growth retardation has included conditions such as congenital hemiatrophy, developmental hip dysplasia, Legg–Calvé–Perthes disease, slipped capital femoral epiphyses (SCFEs), polio, achondroplasia, dyschondroplasia, and severe burns. Causes by growth stimulation include congenital giantism, Wilms' tumor vascular abnormalities such as Klippel–Trénaunay–Weber, thrombosis of femoral or iliac veins, and traumatic arterial venous aneurysms. Tumors such as giant cell,

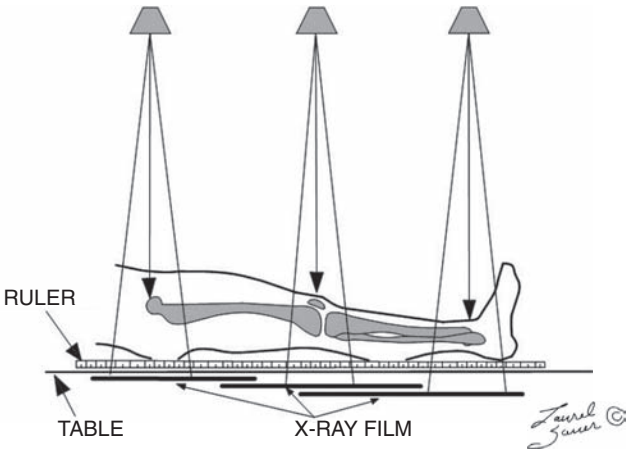

FIGURE 10.35 The scanogram technique avoids errors of magnification and is preferred for children who can remain still for three exposures.

neurofibromatosis, and bony fractures can cause growth retardation or growth stimulation also. The child with *true hemihypertrophy* needs to be screened every 4 months for the possibility of *Wilms' tumor* (5,333) up through the age of 8 and every 6 to 12 months through the age of 10. Screening involves obtaining a renal ultrasound and often a laboratory draw for alpha fetoprotein level. Eighty percent of Wilms' tumors present prior to the age of 8, with an average age at presentation of 3 years. The tumor may be associated also with aniridia (lack of an iris in the eye) and secondary metastases to the skeleton. A firm, nontender abdominal mass may be palpated. Damage to the growth plate with trauma and epiphysiodesis, including fractures with marked overriding of fragments, tends to cause more growth retardation.

Treatment objectives include obtaining leg-length equality, producing a level pelvis, and improving function. LLD of less than 1.5 cm is usually just observed. Shoe lifts can be utilized for differences up to 3 cm. Horizontal alignment of the iliac crest or sacral base in the standing position should also be witnessed with appropriate shoe lifts in place. Early attention should be given by the age of 7 or 8 to observe and record the pattern of growth and appropriately time any growth plate arrest.

The *Greulich–Pyle norms* for skeletal maturation of the hand (334) and the charts of Green–Anderson (334) are used for prediction of future growth and the timing of surgery when stapling epiphysiodesis of the longer side is considered for true discrepancies between 3 and 6 cm. Stapling techniques across the physis produce a tethering effect and can be removed later once equalization has been achieved (335,336,337). *Surgical epiphysiodesis* is an all-or-nothing procedure that completely and permanently arrests physeal growth. The principle is to produce a symmetrical bony bridge that tethers the physes and prevents future growth (338). *Epiphysiodesis* is most commonly performed 2 to 3 years prior to maturity (girls aged 11–12 years; boys aged 12–13 years).

Shortening procedures can also include removal of a section of bone for limb equalization performed in adults or adolescents who are no longer growing (8). Charts of Green and Anderson are displayed in Figures 10.36 and 10.37, respectively. Total leg length versus skeletal age for boys and girls are shown, respectively. Plotting of leg length versus skeletal age is critical in the timing of any surgical procedure projecting limb length equalization into the future (339,340). The *Green and Anderson studies* provide good documentation for the general population studied, but no guarantees for children of other races or genetic stock. Additional and more specific determination of LLD can be obtained through three additional methodologies (5,8). These include the arithmetic method, the growth remaining method, and the straight line graph method—not described further, as such is beyond the scope of this text. Growth discrepancies beyond 6 cm are best treated by *limb lengthening* through

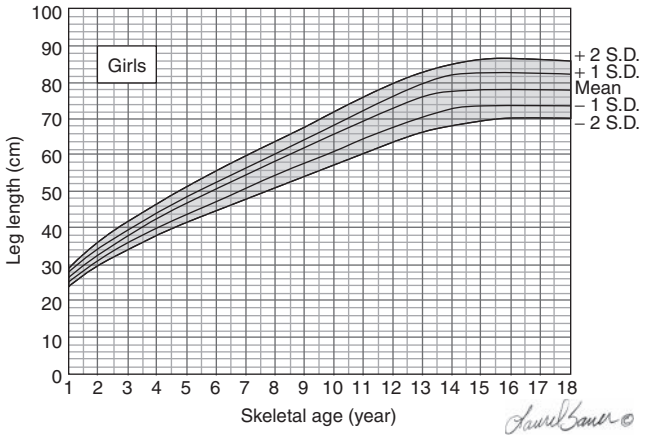

FIGURE 10.36 Grafts showing total leg length vs. skeletal age for girls. It provides useful analysis of leg-length data, allowing a projection into the future on the basis of present status.

such methods as *Wagner or Ilizarov* (5,8). Unlike epiphysiodesis, leg-lengthening procedures can be performed at almost any skeletal or chronological age. Discrepancy greater than 15 to 20 cm should consider combined shortening and lengthening procedures in addition to amputation. Codivilla first reported mechanical bone lengthening in 1905 (341). Subsequent advancement in limb lengthening has been by the method of Ilizarov (342), whose biologic principle of distraction osteogenesis has revolutionized the surgery. Ilizarov's circular external fixation system is complex, but provides for multilevel correction, including angular deformities and lengthening simultaneously (Figure 10.38). *Corticotomy technique* is utilized with care so as not to disturb the

FIGURE 10.37 Grafts showing total leg length vs. skeletal age for boys. It provides useful analysis of leg-length data, allowing a projection into the future on the basis of present status.

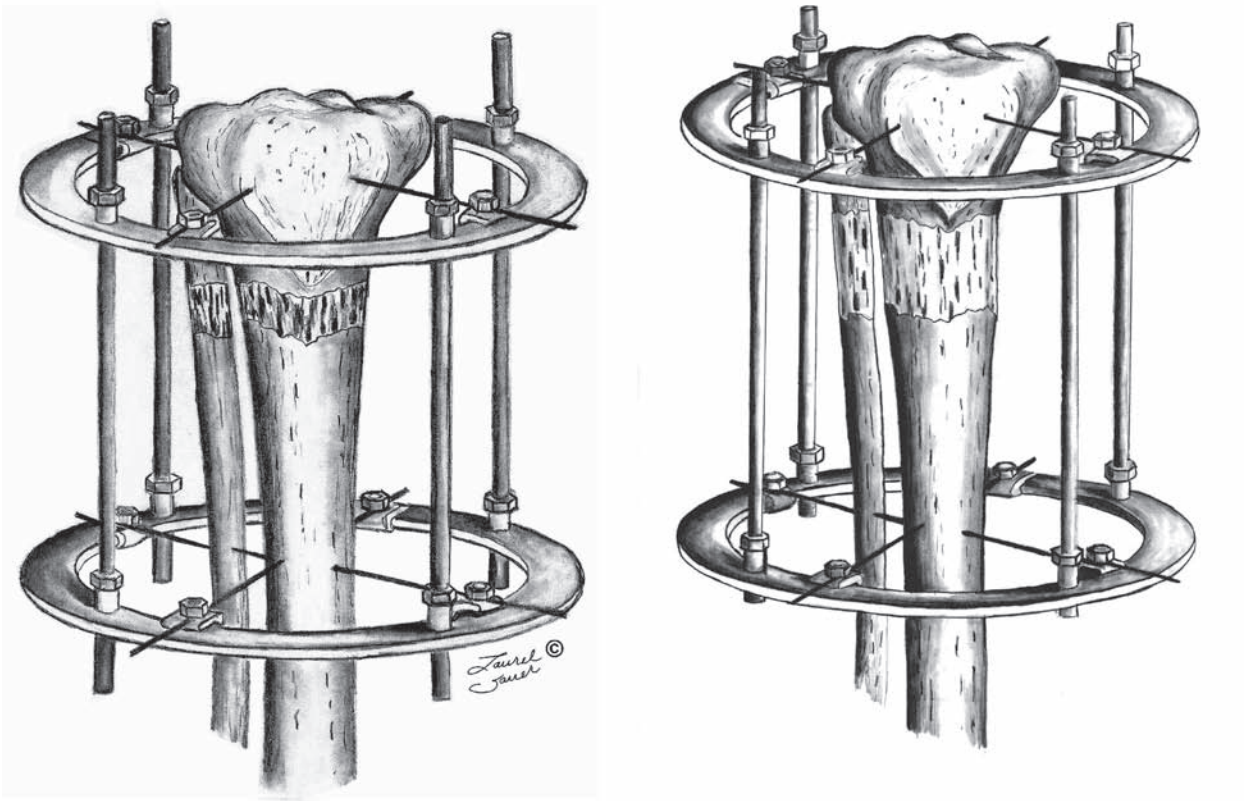

FIGURE 10.38 (A) Sequential metaphyseal lengthening. (B) Elongation through the active metaphysis promotes osteogenesis and strength by the large cross-sectional area across the lengthening gap.

medullary cavity contents such that they may make their greatest contribution to osteogenesis during lengthening (5,8).

Elongation through the metaphyses promotes osteogenesis because metaphyseal bone is so active and promotes strength by the large cross-sectional area. The lengthening process begins approximately 5 to 10 days after surgery. Lengthening of 1 mm per day or approximately 1 inch per month is recommended (5,8). External fixators are worn until the bone is strong enough to support the patient safely. This usually takes about 3 months for each inch. A normal lifestyle during treatment is encouraged. Some children even go swimming with the external fixator in place. *Complications* include pin tract infections (most common), fracture, axis deviation, delayed union, and soft tissue contractures. A child whose family is not capable of sustained follow-up may be a poor candidate for limb lengthening. Significant patient and family education needs to occur, including preoperative and postoperative phases, preparing the child and family both physically and emotionally for the long treatment. *Counseling services* may prove helpful. Rehabilitation services are quite valuable, including frequent physical therapy visits for any successful long-term outcome.

CONSTITUTIONAL OR INTRINSIC BONE CONDITIONS

Constitutional conditions of bone may be divided into five categories:

- Defects of tubular bone or spinal growth
- Disorganized cartilage and fibrous components
- Abnormal bony density or structure
- Metabolic conditions usually affecting calcium or phosphorus metabolism
- Extraskeletal disorders

DEFECTS OF TUBULAR BONE OR SPINAL GROWTH PRESENT AT BIRTH

Achondroplasia

Achondroplasia is an autosomal-dominant genetic skeletal disorder; with approximately 85% new mutations, caused by a mutation in the fibroblast growth factor receptor-3 gene (FGFR3). It is the most common of the skeletal dysplasias (343,344).

CLINICAL FEATURES. The diagnosis of achondroplasia is made clinically with characteristic features on a radiograph. These conditions are often associated with shortened trunk, narrow thorax, and variant body proportions, including enlarged head size with frontal bossing, hypoplasia of the midface, short limbs and fingers, lordotic lumbar spine, and bowed legs. Although typically normal in intelligence, secondary to their size, these individuals are often looked upon and treated as younger than their stated age. Secondary to transitory muscular hypotonia, early motor milestones are frequently delayed in infancy. Visual-spatial learning issues or deficits similar to other children with compensated hydrocephalus may be observed. Physical therapy that includes exercises with these babies prone is also important in order to minimize thoracolumbar gibbus (344).

PREVENTION: OBESITY. There are specific weight charts for those with achondroplasia (345). Obesity is also correlated with the increase in cardiovascular-related deaths (346). Lumbar-region symptomatic spinal stenosis may be seen in achondroplasia; it is aggravated by obesity. Signs of this include lower back and leg pain, and may be observed in 50% of those with this condition. There may be weakness, altered deep tendon reflexes, paresthesia, and later, claudications. Early treatment includes anti-inflammatory medication and corticosteroid injections to treat lumbar radiculopathy, with one-third eventually needing lumbar laminectomy (347). Kyphoscoliosis is common.

PREVENTION: EAR, NOSE, AND THROAT. Tonsillectomy and ventilation ear tubes may help prevent conductive hearing loss. Otitis media may be recurrent secondary to shortened eustachian tubes secondary to midface hypoplasia. This is a significant problem in approximately 40% of those with achondroplasia. Often, there are too many teeth than can be accommodated and teeth need to be pulled or the jaw needs to be expanded (348). This is necessary for dental alignment.

PRECAUTIONS, MONITORING, AND SURGICAL INTERVENTION. This group requires precautions with regard to atlantoaxial instability. The instability may be from maldevelopment of the odontoid, transverse ligament laxity, or longitudinal ligament abnormalities. MRI of the brain, including the cervical junction and the spinal cord, is recommended between the ages of 6 and 12 months. Signs of cervical cord compression (myelopathy) are increased reflexes of the lower extremities, clonus, severe hypotonia, central sleep apnea, and sudden death. Polysomnography is used to demonstrate the central sleep apnea (347). Referral to the appropriate specialist is necessary for evaluation and treatment.

Hydrocephalus, if present in achondroplasia, must be carefully evaluated and may need surgical intervention (348). Head circumference must be monitored every 6 months while growing (especially in the first 2 years of life), and symptoms of increased cranial pressure must be evaluated (344,347). MRI of the cervicomedullary junction, as well as cerebrospinal fluid (CSF) flow studies, may be normal with a neutral neck position. With flexion and extension of the cervical spine, complete blockage of CSF flow in the former and posterior cervicomedullary compression in the latter may be demonstrated (348). Flexion and extension imaging is warranted if there are mild to severe symptoms and signs present, as surgical options such as ventriculoperitoneal (VP) shunt or decompressive surgery can be corrective (348).

TREATMENT. This may include human GH therapy (349). The long-term sequelae of this are unknown. Parathyroid hormone has been shown to improve bone growth and mitigate the effects of FGFR3 mutations found in achondroplasia (350). Limb lengthening is a possibility, but has many risks involved, including infections as well as soft tissue, nerve, and joint damage. This remains controversial (347). There is an increased number, compared to the general population, of sudden deaths thought to be caused by foramen magnum stenosis in children under 5 years of age. There is also an increase in mortality from cardiac and neurologic diseases, and from drug overdose and suicide in older patients (346). Reports of depression, low self-esteem, poor body image, and chronic pain need to be addressed. The key to successfully treating this condition is a multidisciplinary team that includes the rehabilitation specialist, occupation and physical therapists, social worker and/or psychologist, neurosurgeon, cardiologist, gastroenterologist, endocrinologist, and orthopedist.

DISORGANIZED CARTILAGE AND FIBROUS COMPONENTS

Fibrous Dysplasia

Fibrous dysplasia is a condition characterized by the presence of expanding fibro-osseous tissue in the interior of affected bones. It is characterized by cancellous bone being replaced by fibrous tissue. Primarily this is a lesion of the growing skeleton (5).

CLINICAL CHARACTERISTICS. Fibrous dysplasia may cause pain or limping gait, extremity length discrepancy, bowing, or fractures. This may be associated with endocrine abnormalities such as Albright's syndrome, which consists of the triad of multifocal bone involvement, precocious puberty, and cutaneous pigmentation.

DIAGNOSTICS. Radiographic lesions typically are sharply marginated with sclerotic bone and appear as ground glass or lytic expansile lesions of the diaphysis or the metaphysis.

TREATMENT. Treatment typically includes observation. Surgery may be necessary for those lesions causing progressive deformity, pain, fracture, or impending fracture.

ABNORMAL BONY DENSITY OR STRUCTURE

Osteogenesis Imperfecta

Osteogenesis imperfecta (OI) is a heritable bone disorder with abnormal bone quality or quantity (351).

CHARACTERISTICS. Fractures are the hallmark of OI. The number of fractures in a lifetime varies from a few to several hundred. There are numerous associated clinical findings, but phenotype can vary greatly even within families with the same genotype (352). Short stature is common, as are blue sclera, relative macrocephaly and triangular facies (353). Cognition is normal. People with OI tend to have a high-pitched voice. In addition to fractures, musculoskeletal findings can include scoliosis, muscle spasms, and hypermobility. Multiple bone microfractures can lead to bowing and increased fracture risk (354). Respiratory failure is the leading cause of death in OI (355). Abnormalities of the skull base can cause brainstem compression and neurologic compromise (356). Skin tends to be fragile, leading to increased bruising (357). With fractures and bruising, some children with mild OI may be difficult to distinguish from those sustaining nonaccidental trauma (351,353). Hypercalciuria and renal calculi can occur, as can aortic dissection and mitral valve prolapse (353). Hearing loss may require amplification or surgery. Dentinogenesis imperfecta can be present. Mobility ranges from no limitations to inability to sit, though most people with even severe OI can sit and use a power wheelchair.

Sillence described four types of OI (351,352,357, 358,359,360). Type I is relatively mild. Type II is fatal in the perinatal period. Type III is severe with progressive bony deformities. Type IV tends to be moderate. There is overlap in the clinical presentation, particularly in Sillence types III and IV. Types I–IV, are associated with abnormal quantity or quality of type I collagen and are dominantly-inherited. In recent years, many new, rarer types of OI have been described based on unique structure found on bone biopsies as well as clinically distinguishing features (351). The newly described types are usually recessively inherited and associated with normal collagen. A new classification has been proposed, keeping the Sillence types I to IV of mild, lethal, severe, and moderate but removing the molecular aspects of the classification (361).

REHABILITATION. Infants can be positioned to encourage active movement while decreasing fracture risk (351,352,361). Improvement of head control can be encouraged by prone lying or lying on a reclining parent's chest or shoulder. Towel rolls can be used to avoid excessive hip abduction while supine or support the infant's back in side-lying. Diaper changes should be done by rolling the infant, not by lifting the legs (354). Lifting the infant should be done with a wide base (eg, hands spread apart), not under the arms.

ROM exercises should be active (352). Aquatic therapy has been recommended to increase strength and mobility (352,362). Weight-bearing can improve bone strength. Clamshell bracing may be used to provide support for weight-bearing (351,353,363). Long leg bracing may be shortened later (351). There has been a trend for less bracing recently, as infants have been starting early intervention programs and sit in their first year (351).

Sports and recreational activities may be added in the school-age or teen years (364). However, high-impact activities such as gymnastics, aerobics, martial arts, hiking, and contact sports are not recommended (365). The Osteogenesis Imperfecta Foundation (OIF) published a book that provides detailed therapy recommendations and rates the relative risks and benefits of various sports (366). The OIF also provides excellent resources for patients, families, and health care providers. Children should stand or walk daily (365). They may benefit from playing a wind instrument or singing to improve pulmonary health (365). Independence with ADLs can be gradually increased. Children should avoid staying in their wheelchairs for the entire day. Armrests can be removed from manual wheelchairs to decrease forearm bowing (353). Adolescents can learn to manage their medical care, drive, and transition to college or work (367).

MEDICAL INTERVENTIONS. Multiple drugs have been tried in OI without success until bisphosphonates were shown to increase bone density, decrease risk of fractures, increase mobility in some patients, and decrease pain (368,369). Side effects such as transient fevers and discomfort were relatively mild for most children. Markers of bone turnover decreased. Some studies have shown benefit for infants and toddlers (370,371). There are concerns that prolonged use could cause decreased bone healing (369). Long-term risks are unknown. Some studies (372,373), but not all (374), have shown improved function with bisphosphonate treatment. The optimal drug and dosing has not been determined (375). Adequate calcium and vitamin D intake should be ensured or supplemented. Additional treatments such as GH therapy in combination with bisphosphonates are being studied (376).

SURGICAL INTERVENTIONS. The risk of fracture has been found to increase significantly when long-bone angulation is 40 degrees (363,377). Intramedullary rods can improve fracture risk but can migrate into joints (378). As a child grows, the bone "unprotected" by the now too-short rod can break. Telescoping rods have been used, but still have risks. Some surgeons have found fewer surgical complications in children treated with bisphosphonates (379).

OUTCOMES. Despite the fractures, surgeries, and mobility impairments common in OI, people with OI rate their quality of life well (380,381). A recent study showed that children with OI rated higher than the reference norms on the psychosocial summary of the Child Health Questionnaire (381). Adults with OI often attend college and have employment similar to the general population without disabilities (380).

METABOLIC CONDITIONS USUALLY AFFECTING CALCIUM OR PHOSPHORUS METABOLISM

Rickets

Rickets is caused by vitamin D deficiency that results in osteomalacia, the delayed or inadequate mineralization of osteoid in mature cortical and spongy bone (5). Rickets is a rare condition in the United States. However, it may be found in higher numbers in dark-skinned breastfed babies who are not supplemented and all breastfed babies who themselves and/or their mothers have little to no exposure to the sun on a daily basis. Typically, this becomes problematic after 6 months of age.

CLINICAL CHARACTERISTICS. The clinical features of nutritional rickets include early-onset craniotabes, rachitic rosary (costochondral junction enlargement), and thickening of the wrists and ankles. As rickets continues, clinical findings include progressive bowing of the legs; poor linear growth; and abnormal serum calcium (ionized calcium is the most accurate test), phosphate, and alkaline phosphatase levels. In severe cases, the baby may have seizures.

DIAGNOSTICS. The diagnosis of rickets is made with radiographic demonstration of metaphyseal flaring, cupping, and decreased mineralization of the distal metaphysic, as well as laboratory evidence of elevated alkaline phosphatase.

TREATMENT. The treatment includes supplementation of vitamin D and/or formula. If left untreated, permanent deformities may ensue.

Mucopolysaccharidoses

Mucopolysaccharidoses (MPS) are hereditary progressive conditions secondary to mutations in genes that code for enzymes responsible for lysosomal degradation of glycosaminoglycans (GAGs) (formerly called mucopolysaccharides) (382). The underlying problem is a defect in the degradation of GAGs leading to the accumulation of lysosomes (vacuoles found in almost all cells) (383). All seven types of MPS are a form of lysosomal storage disease. The accumulation of GAGs results in clinical sequelae in respiratory, cardiac, gastrointestinal, neurologic, and musculoskeletal systems. For MPS I, II, IVA, and VI there is recombinant human, enzyme replacement therapy (ERT) available (384–390). ERT was first available in 2001 and approved in 2003 for MPS type I (537). Therefore, ERT in MPS is a young and vibrant field of vast research worldwide. Hematopoietic stem cell transplantation (HSCT) has shown improvement in MPS I (382,391), particularly in regard to neurologic deterioration. Gene therapy holds promise and risk (391).

Various parameters to assess walking and endurance such as the 6-minute walk test, have been used in studies at multiple sites nationally and internationally. ERT in the MPS VI population has demonstrated improvement in endurance, ambulation, pulmonary function, and survival (390). Similar benefits from ERT have been shown in MPS I, MPS II, and MPS IVA. ERT has variable clinical outcomes depending on the age that it is initiated and the organ system studied. Following ERT, there is still significant morbidity with regard to cardiac valve, skeletal, and joint disease. Therefore there is rationale to start ERT early before disease sequelae progress. Marked heterogeneity exists within each of the seven groups, and life expectancy for some can be before the first decade and can reach into the fifth decade or even more (382). Many of these individuals appear normal at birth. In general, the later the clinical onset, the slower the clinical picture (383,392).

CLINICAL CHARACTERISTICS. The cranium is typically disproportionately large and the facial features are coarse and may include bushy eyebrows, corneal clouding, prominent forehead, a depressed nasal bridge, thick lips, and enlarged tongue. Short stature is present to some degree in all seven types. Odontoid hypoplasia and resulting atlantoaxial instability along with accumulation of GAGs in the atlantoaxial area compressing the spinal cord frequently require decompression and fusion. This is a major complication of this condition, potentially causing spinal cord compression with resulting respiratory compromise and tetraplegia. The time of surgical intervention is critical. Contractures may present at the shoulders, elbows, forearms with decreased supination, fingers, hips, knees, and ankles. Fingers are short and stubby, and hands are wide. Fingers typically have flexion contractures along the distal interphalangeal (DIP) joints. Carpal tunnel syndrome (CTS) is common. Typically, the presenting complaint for CTS is difficulty with fine motor tasks, not pain (393). Release of the carpal tunnel at our institution has improved the DIP contractures presumably because there is more room for the tendons to glide once released. Cognitive decline is difficult to predict in young patients. Mental deterioration eventually occurs in most type III individuals and those with severe types I, II and VII. Kyphosis can appear early and is usually marked. Blindness may result from optic atrophy or

corneal clouding. Deafness may occur as well. Dysostosis multiplex is a constellation of radiological findings: thickened diaphyses, bullet-shaped metacarpals, hypoplastic epiphyses, spatulate ribs, abnormally shaped vertebrae, and enlarged skull (394). Individuals with MPS require regular imaging of the spine and hips, at least until skeletal maturity.

SPECIFIC TYPES. MPS are typically divided into seven groups and subgroups as MPS III A, B, C, and D or MPS IVA and IVB, each with one or more deficient enzymes. In MPS I, there are three clinical subtypes all with mutations in the same gene: the most severe form is Hurler's syndrome and the mildest form is Scheie syndrome (MPS I-S, formerly MPS V). The mode of inheritance is autosomal recessive for all groups except MPS II Hunter's disease, which is X-linked recessive. Type III is Sanfilippo A, B, C, D. Type III is the most difficult to diagnose because it does not have the clinical features prominent in the other types and is primarily a syndrome involving the nervous system. Falling, hyperactivity, attention deficit, or speech and sleep problems may be the first signs of this condition.

Type IVA and IVB Morquio syndrome: Type IVA, the more common of the two types, is characterized by normal intelligence and gross motor milestones early in life. Over time, gait may progressively worsen with severe genu valgum, ligamentous laxity, pes planus, hip dysplasia, and increased sternal protrusion. The chest deformity can be restrictive and cause cardiorespiratory symptoms (392). Many of these individuals do not reach normal body length.

Type VI Maroteaux–Lamy has the facial features typical of MPS; intellect generally remains normal; and obstructive sleep apnea, corneal clouding, and deafness are common. VII Sly type can present as hydrops fetalis, or life expectancy can be into the second decade. There can be a wide variability of cognitive ability. Type IX, Natowicz syndrome, is characterized by normal intelligence, short stature, and painful masses around joints and is extremely rare, with four known individuals with this type of MPS.

REHABILITATION. A multidisciplinary approach is essential for the management of individuals with MPS. Specific rehabilitation issues may include hand and wrist bracing to help avoid progression or formation of contractures of the wrist and fingers and help with the management of CTS. Lower extremity bracing, walkers, and gait trainers can help with ambulation and postural alignment. A TLSO for the back may help prevent progression of scoliosis. Aids for functional independence are essential. Quality of life was increased with decreased dependency of wheelchair use in those with Morquio IVA syndrome (395). Power mobility for community independence is essential for many with MPS.

MEDICAL AND SURGICAL INTERVENTIONS. SEPs can be helpful in determining compression of the cervical spine (396). SEPs can be done at 6- to 12-month intervals to look at changes in latency of the waveforms. Yearly MRIs for monitoring cord compression and potential surgery are typically used in the United States. There is an increased risk with anesthesia, and a medical center with an anesthesia team that specializes in this condition is recommended. Intraoperative monitoring for surgical procedures is important to make sure that the spinal cord is stable and the neck is in good position during anesthesia. The use of an ultrathin fiberoptic bronchoscope may be necessary during intubation.

Carpal tunnel release can lead to improved function and can decrease contractures of the DIPs. Cervical spine decompression and stabilizing is important to prevent tetraplegia and ventilator dependency. Hip surgery is also important for transfers and ambulation. The ERT does not cross the blood–brain barrier or enter the joint space/cartilage or cornea. There are trials as this chapter goes to press on intrathecal ERT for MPS I and MPS II (ClinicalTrials.GOV). Earlier HSCT in MPS I is thought to have the best possibility of good results. VP shunt can help manage hydrocephalus (392). Genetic consultation is important secondary to prolonged life expectancy with ERT (397,398).

EXTRASKELETAL DISORDERS

Sickle Cell Anemia

Sickle cell anemia has been discussed earlier in this chapter. The reader is referred to external references for additional discussion as needed.

Chronic Kidney Disease

Children with chronic kidney disease (CKD) are at great risk for short stature. Adequate nutrition may be problematic. With failing kidneys, erythropoietin production is inadequate and anemia may result. These children may be resistant to their own elevated GH and may require recombinant human GH subcutaneously (399). Renal osteodystrophy is a term that describes the bone disorder spectrum in CKD. It is most commonly associated with a high turnover bone disease secondary to hyperparathyroidism (399). Osteitis fibrosa cystica is the pathologic skeletal finding in this condition. In CKD there are calcium and phosphate metabolism changes that lead to hyperparathyroidism, hyperphosphatemia, calcitriol deficiency, and hypocalcemia. The osteocyte produces a circulating peptide called FGF23 that is instrumental in regulating the excretion of phosphate. There is an increase in FGF23 for all age groups with CKD, and this is inversely proportional to the glomerular filtration rate. In addition, there is defective hydroxylation of vitamin D.

Vitamin D supplementation (both inactive and active forms) may reduce cardiovascular morbidity and proteinuria by improving hyperparathyroidism. Additionally, phosphate binders (that do not contain calcium) decrease mortality of adults with CKD and show promise in the treatment of the pediatric population as well (400). The excessive parathyroid hormone is a response to correct the hypocalcemia by increasing the bone resorption (399). Clinically, these patients have muscle weakness, bone pain, and fractures from minor trauma. Rachitic changes as well as varus and valgus deformities of the long bones and SCFEs may be seen in growing children. The x-rays demonstrate subperiosteal resorption and widening of the metaphyses in the hands, wrists, and knees (399). Medical management for this condition is by a nephrologist. Diets include low phosphorus, phosphate binders, vitamin D, and noncalcium-based diets for those who are prone to hypercalcemia. Recombinant human erythropoietin subcutaneously and iron orally or intravenously are important treatments for anemia (399).

SUMMARY

As mentioned at the onset of this constitutional bone condition section, typically, these individuals have normal intelligence. They may, however, be perceived differently, especially if they are smaller than their chronological age. Age-appropriate interventions are key for this group. It is important to know the key features in these groups as well as serious complications. As these conditions generally have increased risk of atlantoaxial instability, these individuals should be restricted from contact sports and other high-risk activities (11).

MUSCULOSKELETAL PAIN AND CHILD ABUSE

Musculoskeletal pain in children is variable. Depending on age, and verbal and cognitive abilities, assessing pain in the pediatric patient may present additional challenges. Children under 5 years may have difficulty describing pain. A scale with faces illustrating different emotions may help children describe how they feel (401). The faces scale, has been in use for over a decade, continues to be used during assessment, and was recently used in a study published on pain in the pediatric population (402). At the age of 6 years, children can usually score their pain on a level between 0 and 10 by increasing severity (11).

COMPLEX REGIONAL PAIN SYNDROME

Complex regional pain syndrome (CRPS), also known as reflex sympathetic dystrophy, is a condition that usually involves one limb and more commonly the lower extremity in children. There is a scarcity of data in the pediatric population. There are two types of CRPS. Type 1 follows an injury or illness to the region affected and type 2 occurs after a distinct nerve injury. CRPS is a complex condition characterized by dysesthetic sensory component. Motor and sensory cortexes are reportedly reorganized and there are inflammatory and neurologic changes peripherally (403).

Clinical Characteristics

CRPS is characterized by pain, hyperesthesia, edema, cold or warm extremity, cyanosis, mottling of skin, limited ROM, and patchy bone demineralization. Adults with CRPS complain of hot and cold extremities, but kids more than likely have a cold cyanotic extremity (404).

Acute Phase Lasts up to 3 Months

Burning pain and increased sensitivity to touch are the common findings during this time. Joint stiffness, swelling, redness, and increased warmth follow typically (405). Other findings may include an increase in the rate of nail and hair growth as well as sweating. *Stage II*: dystrophic, 3 to 12 months; swelling more constant. *Stage III:* after 1 year.

Diagnostics

Unlike adults, who usually have an inciting event such as a fracture, surgery, prolonged immobilization, or vascular insult, children usually do not have a clear event that precipitates the condition (406). There appears to be a sympathetic nervous system reflex arc mechanism of action. The majority of children with this condition are teenage girls around 12 to 13 years of age. Radiographs are useful to rule out a fracture or osteomyelitis (407). Regional nerve blocks may be both diagnostic and therapeutic. The diagnosis should be considered with trauma and pain that is out of proportion to the stimulus and worsened with use.

Treatment

A multidisciplinary approach is useful. The earlier the recognition and treatment, the more rapidly recovery is possible. Once contractures and atrophy set in, this is a much more difficult entity to treat. Some advocate medications such as calcium channel blockers, beta-blockers (propranolol), and tricyclic antidepressants such as amitriptyline. Relapse rates are high (408).

FIBROMYALGIA

The etiology of fibromyalgia in children and adolescents is unclear.

Clinical Characteristics

Diffuse musculoskeletal pain involving the neck, back, and upper and lower extremities is common in fibromyalgia in children and adolescents. Sleep disturbance, headaches, fatigue, and problems with peer relationships (408) are common among those diagnosed. The diagnosis of juvenile primary fibromyalgia syndrome (JPFS) is given when the child or adolescent has muscle and join pain, and/or pain "all over" without any other illness.

Diagnostics

Polysomnography is frequently positive while other tests are negative (409). There has been an increase in children whose mothers have the condition; this may be cultural rather than genetic. Females are more affected, and the onset ranges from around 11.5 to 15 years. Children with fibromyalgia can have fewer trigger points than adults, although the exact number is uncertain.

Treatment

Education and psychological interventions are the first line of treatment. Those with JPFS are often treated by rheumatologists, as they may present similar to a child or adolescent who has lupus or arthritis (410).

Outcomes

There is a high prevalence of fibromyalgia in those adults who clinically had onset of fibromyalgia in adolescence. Furthermore, these individuals have been found to be the most impaired both emotionally and physically in adulthood. In one study, greater than 80% of patients who presented as juvenile-onset fibromyalgia continued to express symptoms of fibromyalgia in adulthood and approximately half of those met the criteria of the ACR. Additionally, the fibromyalgia group was more likely to marry at an earlier age and have children rather than move out of their homes and attend college compared to age-matched healthy peers (411).

BACK PAIN

Back pain in children, once thought to be relatively uncommon, was in a recent review article from the United Kingdom reported as a common symptom in this group. This article reports that this is more common and is increasing with prevalence rates between 7% and 58% (412). Usually, the child with back pain presents with a muscular-strain-type pattern related to poor posture, activities at school or home, or other recreational or sporting pursuits (5). Up to 30% of pediatric individuals will experience back pain by the time they are teenagers (412). When carrying backpacks of greater than 10% to 20% the body weight of the child or adolescent, musculoskeletal strain is common. Children and adolescents generally do well with strategies such as decreasing backpack weight, making sure the backpack is level to their shoulders, carrying the backpack on both shoulders, and using proper body mechanics when picking up items from the ground. With prompt adherence to these guidelines, only a small percentage of children and adolescents go on to have chronic symptomatology. Conservative intervention with physical therapy, correction of biomechanics, postures, and proper equipment, are often all that is required for resolution of symptoms. NSAIDs with food along with the RICE protocol are utilized as well. Back pain that is not improving within 2 to 4 weeks of conservative care needs further investigation. Less likely in the adult, chronic back pain in children can be associated with serious pathologic entities, including neoplasm, infection, and inflammatory disease (8).

Spondylolysis and Spondylolisthesis

In those over 10 years of age, spondylolysis and spondylolisthesis are the most common etiologies of back pain. Caused by a defect in the pars interarticularis affecting most commonly the fourth and fifth lumbar vertebrae, spondylolisthesis is typically caused by a stress fracture that facilitates the vertebral segment to be placed forward on the inferior one. The seriousness of the spondylolisthesis determines intervention. The first line for diagnosis is a stork leg test with extension of the individual's back. If there is pain in the lower back elicited with this maneuver, then typically oblique views of the spine are obtained. CT scan can be the most sensitive (412). *Scheuermann's kyphosis* may present as back pain. This affects boys and girls equally and is seen in up to 8% in this group (413). There can also be pain in up to 23% with AIS (414).

Osteoid osteoma is a benign tumor that is seen in approximately 11% of those from ages 10 to 25 years. Scoliosis develops because of the muscle spasm secondary to the asymmetric vertebral involvement. A radiolucent nidus with surrounding sclerosis is apparent on an x-ray. Treatment is variable (415). *Aneurysmal bone cysts* appear on x-ray as a cystic area in the bone (common in the pelvis and posterior spinal elements). Treatment is often surgical (248). A full discussion of back pain in children is

beyond the scope of this text, and the reader is referred to other sources (5,8,232,412). Additional discussion of back pain in children is provided in Chapter 11.

Referred Back Pain

Many conditions can produce referred back pain. These include pyelonephritis, pneumonia, endocarditis, cholecystitis, pancreatitis, osteomyelitis, pelvic inflammatory disease, and other more general conditions affecting the muscles, as well as conditions such as inflammatory arthritis. Sickle cell pain crisis can cause back pain. Conditions that usually have the presenting complaint of nighttime pain are osteoid osteoma (the most common benign bone tumor) and ankylosing spondylitis (415). For malignant neoplasms of the spine, 90% are secondary sites and not primary tumors. Leukemia may present as back pain (412). Functional pain issues also present the clinical challenge of ruling out an underlying, more serious disease. A good history and physical exam often point out inconsistencies.

CHILD ABUSE

A recent publication of an evaluation of 16,897 child abuse visits to a 20% stratified sample of nationwide emergency departments from 2008 to 2010 found that 30.7% required hospitalization. Hospitalization was more prevalent in the younger group, in those on average around 2 years as opposed to those who were older. Perpetrators were male or female partners of the child's parent/guardian, more than 45% of the time. Fractures were most common at 63.5% and intracranial injuries were seen in 32.3%. Of these individuals who presented for treatment, death occurred in 246 (416).

The incidence of abuse or neglect in the United States annually is estimated up to 1.25 million children (417).

Initial action depends on whether the suspicion is great enough to warrant making a report to Child Protective Services (CPS) (418). It is essential to obtain a detailed history, including the mechanism of injury, and to look for inconsistencies. Knowledge of child development is essential. Suspicion is increased if the injuries are inconsistent with the child's developmental level or mechanism of injury, blamed on the victim's siblings, or not witnessed. Children, for example, generally cannot roll over until the age of 4 months. Most children who fall off a piece of furniture have a fracture risk of less than 2%. Therefore, a history of a 3-month-old infant rolling off a piece of furniture and sustaining a severe injury should raise suspicion of child abuse. Multiple injuries in various stages of healing should increase suspicion.

With suspected child abuse, physical exam includes an ophthalmologic examination for retinal hemorrhages as well as a head-to-toe examination that also looks for skin bruising, swelling or deformity of extremities, malnourishment, and poor hygiene. Photos are useful for clinical documentation of any abnormalities, and frequently skin marks, bruises, welts, or burns should be recorded. Radiographs or a bone survey is necessary for any extremity that is tender, has swelling, or has limited range. A radionucleotide study can be of added help to the skeletal survey. There are no pathognomonic fracture patterns, but high suspicion fractures include posterior rib fractures; metaphyseal corner fractures; sternum, scapula, or spinous process fractures; bilateral acute long-bone fractures; complex skull fractures; fingers in nonambulatory children; and multiple fractures in various stages of healing. The most common type of fractures involved with child abuse is transverse, followed by spiral fractures, then avulsion fractures, and later oblique fractures. Those fractures with low specificity include clavicle fractures, simple skull fractures, and isolated long-bone fractures (419). Spine fractures constitute only about 3% of these fractures (420). When child abuse is suspected, the physician is legally obligated to file a report with the appropriate child protection agency. Adequate supportive measures and counseling should be in place before returning any abused child to the home. When in doubt, temporary foster placement should be seriously considered.

TUMORS OF THE BONE

The prevalence of bone tumors in the United States is approximately 7 children per million under the age of 15 years. Although rare, with approximately 400 cases diagnosed per year, osteosarcoma is the most common primary malignancy of bone during the adolescent growth spurt (rapid bone growth) (5,11). There is a slight preference for boys. It is followed by Ewing's sarcoma, with approximately 200 cases diagnosed per year. Ewing's sarcoma is more common in those less than 10 years of age (5,421). However, both tumors present more commonly in the second decade of life. Osteosarcoma may develop from irradiation treatment of Ewing's or other malignancies. Tumors may mimic various pain syndromes throughout the body. Primary bone tumors common to the upper extremities include Ewing's sarcoma of the scapula, osteogenic sarcoma of the proximal humerus, and osteoblastomas and chondroblastomas common in the diaphyses and epiphyses of long bones (5,422). Spine neoplasms are rare (412). The most common presenting manifestations of osteosarcoma are pain, limp, and swelling. Similar presentation may be found in Ewing's, as well as weight loss and fever.

Diagnostics

The timing of the presentation complicates the differential diagnosis. The symptoms may be attributed to a growth spurt, sprain, or sports injury. Those presenting with osteosarcoma are usually taller than their peers. A complaint of pain that awakens a child or adolescent from sleep is suggestive of malignancy. The most common location of the osteosarcoma is the distal femur, followed by the proximal tibia and proximal humerus. Symptoms not responding to conservative treatment require further investigation, specifically with a radiograph. A sunburst pattern or Codman's triangle (lifting of the cortex by new bone formation) are classic radiographic findings found in two-thirds of those presenting with osteosarcoma. With Ewing's, a permeating "moth-eaten appearance" is demonstrated on x-ray. If suspicion of tumor is high, and radiograph is negative, seen with medullary osteogenic sarcoma, MRI should be obtained of the entire long bone, as no pattern on x-ray is pathognomonic (423). Laboratory tests, including a complete blood count (CBC), will usually be normal. Elevated sedimentation rate, alkaline phosphatase, or lactic dehydrogenase levels may be found. Early diagnosis is key, as the prognosis is better if there is less spread of the disease. Metastasis to the lungs remains the most likely cause of death. Additional primary bone tumors to the lower extremities include those of the long bones. These include histiocytosis X in the diaphysis and eosinophilic granuloma in the epiphysis. Tumors more common in the area of the pelvis include osteoblastoma, aneurysmal bone cyst, and fibrous dysplasia. Additional metastatic tumors to the lower extremities include neuroblastoma and lymphomas of various types.

Treatment

This may requires wide resections of the long bone and adjuvant chemotherapy. Once diagnosed, further workup and treatment is necessary at a center with expertise in managing these tumors. The age of presentation of Ewing's sarcoma and osteosarcoma and the complex care needed to treat these bone sarcomas present a challenge, as these individuals are transitioned from pediatric to adult care. In addition, decreased survival in teenagers and young adults with bone sarcomas has been related to the lack of comprehensive transitional services as these younger patients move into the adult health care system (424).

Rehabilitation

Physical activity and contracture management are important rehabilitation issues during acute treatment. Chronically, residual limb skin integrity, prosthesis management, and contracture management are important issues when managing this patient population. Team management is critical, led by the pediatric rehabilitation medicine specialist in the comprehensive care of patient, family, and loved ones (11).

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11

ACUTE MANAGEMENT AND REHABILITATION OF SPORT-SPECIFIC MUSCULOSKELETAL INJURIES

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Participation by children under the age of 12 in athletics and related recreational activities has increased greatly over the past decade (1). A casual notice of the growing number of children playing organized sports such as soccer, baseball, and hockey—or other non-organized athletic activities including skateboarding and pick-up basketball—after school or on a weekend illustrates this point. Because of this, the pediatric physiatrist needs to be well trained in basic sport-specific and musculoskeletal injuries related to these children and their activities. This chapter attempts to provide a basic discussion in this regard.

FOOTBALL

By far and away, there are a greater number and wider array of injuries incurred playing the sport of football than any other commonly played sport in the United States. This is due partly to football's large number of participants as compared to other sports. This is especially true at the high school level where over 1 million of the 7.5 million high school athletes participating in competitive interscholastic sports annually are doing so by playing football. As a result, an increased frequency of exposures to contact/collision during games and practices alike contributes to football having one of the highest rates of injury of all competitive sports. Fortunately, injuries such as *contusions*, *muscle strains*, and minor *joint sprains* predominate. The gross majority of these injuries involve the lower extremities and most result in relatively few days lost from participation due to injury in comparison to more significant macrotrauma.

In the lower extremity of the football player, the knee is particularly vulnerable to injury—whether due to direct trauma from being tackled or colliding awkwardly with another player while the foot is planted in the ground bearing the athlete's weight. An *anterior cruciate ligament (ACL) injury* frequently occurs with the latter of these two mechanisms. With the player's foot planted, a forced twisting moment about the knee joint—often medial rotation—results in a partial or complete tear of the ACL. This can occur as an isolated injury without contact or, as is more frequently the case in football, resulting from direct contact to the knee. Oftentimes, this twisting force is coupled with a valgus force at the knee joint originating from an impact along the lateral aspect of the knee—either from the injured player being tackled at the knee or having the knee landed upon by another player with his or her foot planted. The injury may involve avulsion of the anterior tibial spine. It is not uncommon for the athlete to also incur a concomitant *medial collateral ligament (MCL) sprain* and/or *meniscus injury*. The valgus force at the knee effectively opens the medial aspect of the joint placing stress upon the MCL resulting in a partial tear or complete rupture. An associated *medial meniscus tear* may also occur due to its substance being contiguous with the fibers of the MCL. Concomitant injury to these three structures is commonly referred to as the "Unhappy Triad." Commonly upon injury, the athlete describes a "popping" sensation about the knee with immediate onset of pain and swelling—the latter due to quick development of a hemarthrosis. On examination, the knee generally presents with instability noted by positive Lachman's and anterior drawer testing. Initial treatment includes rest, ice, compression, and elevation (RICE) principles and a period of immobilization followed next by range of motion exercises.

Anterior cruciate ligament reconstructions in children, when performed, need to consider early closure of the distal, femoral, or proximal tibial physes or other growth disturbances with grafts that might cross the growth plate (2). Potential consequences of these growth disturbances include limb length discrepancy and angular deformities. Autogenous patellar tendon grafting appears to be the

surgical choice, not to exclude other surgical considerations of autologous iliotibial graft or hamstring autograft or allograft. Over-the-top femoral graft placements (graft passed through the inter-condylar notch of the femur) have been reported by some authors with good success and efforts to spare excessive physeal penetration (3–5). Additional physeal-sparing reconstructions with minimal risk of growth arrest have been reported, with good success in the younger child (under 12) and adolescent (6).

An isolated *MCL sprain,* commonly the result of a valgus impact on the lateral aspect of the knee, is frequently able to be treated nonsurgically incorporating physical rehabilitation and, on occasion, bracing. An MCL injury is extra-articular in its location and therefore generally should not result in the development of a knee effusion. MCL sprains, as with all other ligament sprains, are graded on a scale of 1 to 3 with Grade 1 being a stretch injury to the ligament fibers without underlying instability; Grade 2 being a partial tear of the ligament fibers resulting in instability but with an end point remaining on exam; and Grade 3 being a full-thickness ligament tear with more marked instability and no end point felt on exam.

Meniscus tears can be isolated or associated with injury to other structures (eg, an MCL sprain). The mechanism of injury is similar to that seen with ACL injury and is associated with a twisting, shearing force at the knee with the foot planted. A small to moderate knee effusion is often present on exam. Meniscus provocative testing on exam, such as McMurray's testing, is frequently positive. MRI of the knee is the best imaging modality for diagnosis of a meniscal tear. Treatment is generally conservative utilizing appropriate rehabilitation and, on occasion, bracing. Surgical intervention via arthroscopy and repair of the torn meniscus is reserved for cases not responding to conservative management or more severe tears. The report of a mechanical locking sensation about the knee by the athlete may represent a loose body within the joint resulting from the meniscal tear and would be another indication for surgery in order to facilitate its removal.

Patellar dislocation/subluxation is commonly seen resulting from a twisting movement about an extended knee joint with the foot planted, coupled with contraction of the quadriceps musculature. More than 90% of these dislocations occur laterally and frequently result in the rapid development of a knee swelling due to a hemarthrosis from tearing of the medial patellofemoral ligament and medial patellar retinaculum. Athletes at risk include those with anatomy contributing to excessive lateral patellar drive such as increased Q-angle, genu valgum, external tibial torsion, weak vastus medialis musculature, tight lateral retinaculum, and generalized ligamentous laxity. Acute management involves incorporation of RICE principles, anti-inflammatories, and a short period of knee immobilization with the joint in extension. Rehabilitation focuses upon strengthening of the patellar-stabilizing quadriceps musculature and is frequently augmented with the use of a patellar-tracking brace. Surgical intervention should be considered in those individuals with poor response to conservative rehabilitation or recurrent dislocation/subluxation.

Posterior cruciate ligament injury is relatively uncommon—especially in the pediatric and adolescent population. The mechanism of injury is a force directed anterior to posterior at the proximal tibia, which can be seen resulting from a fall directly onto one's knees or as a classic "dashboard injury" associated with a motor vehicle collision. Conservative, nonsurgical management is the predominating care for this injury.

Another lower extremity injury common in football is the *quadriceps contusion*. While most contusions incurred playing football tend to be minor and short-lived in their duration, a contusion of the quadriceps muscle has the potential to be more severe. Hematoma formation about the anterior thigh is not uncommon and requires swift treatment in the acute stage including compression, ice, and positioning of the knee in the flexed position to encourage tamponade immediately after the injury. If left unchecked, the development of a large hematoma can lead to significant pain, restriction of the athlete's range of motion, and weakness resulting in prolonged time away from sport. An additional risk, over time, is the development of *myositis ossificans,* calcification of a portion of the hematoma, which may require surgical intervention for removal. Therapeutic stretching and strengthening, as well as the use of ultrasound during physical therapy sessions can be helpful with reducing time away from sport.

A contusion of the iliac crest, known as a *hip pointer*, results from a direct blow to the bony pelvis. This can result from direct impact from a helmet when tackled or secondary to a hard fall to the ground. The affected area is often exquisitely tender due to underlying bony contusion as well as injury to the surrounding hip abductor and abdominal musculature. Initially, RICE principles and anti-inflammatories are utilized along with gentle range of motion stretches about the hip girdle. In more severe cases, avulsion injury to the iliac crest can occur resulting in longer time to return to play. Prevention is best achieved with the use of properly placed hip pads when playing.

In the foot, a *turf toe* injury results from a hyperextension injury to the metatarsophalangeal (MTP) joint of the great toe. More commonly seen in athletes playing on artificial turf, the hyperextension causes tearing of the joint capsule of the first MTP joint at the metatarsal neck. Radiographs are performed to rule out fracture of one of the sesamoid bones and stress views can demonstrate instability of the joint confirming the diagnosis. Early range of motion at the first MTP joint is extremely important though the use of a stiff shoe for pain relief may be necessary with ambulation. Turf toe injuries can be particularly troublesome and result in time away from sport ranging from 2 weeks to more than 2 months. Surgical referral is appropriate in those cases not responsive to conservative management.

While injuries to the upper extremities in football are less common than those affecting the lower extremities, many can result in significant time away from sports participation. *Shoulder dislocation/subluxation* is one of these injuries. This is generally unidirectional, occurring most commonly in the anterior direction, when the humerus is forced into extreme abduction (at or greater than 90 degrees), extension, and external rotation. This is often seen as the result of a defensive player reaching out laterally with a single arm to tackle an opposing player running past. The athlete is usually quite aware of the sensation of their shoulder "coming out of the socket." Acutely, the player is unable to move the affected arm, prompting the need for immediate medical evaluation. Expedient reduction of the dislocated shoulder should be performed as long as there is no concern for fracture (eg, crepitus), and a number of safe methods for closed reduction are commonly utilized. Neurologic exam of the affected arm after reduction—to rule out injury to the musculocutaneous and axillary nerves among others—as well as radiographs to evaluate for the presence of bony injury are important. A *Hill–Sacs deformity*—an impaction fracture of the humeral head—and a bony *Bankart lesion*—fracture of the anterior–inferior glenoid—are two concerning bony injuries that may be associated with anterior glenohumeral dislocations and warrant orthopedic consultation. MRI of the injured shoulder is helpful for evaluation of injury to the surrounding soft tissue structures including the rotator cuff and the glenoid labrum. Initial management postreduction includes a period of immobilization—the duration of which is currently debated but can be up to 6 weeks. Rehabilitation of the dislocated/subluxed shoulder focuses upon strengthening of the rotator cuff musculature as well as scapular stabilizers with the aim of preventing recurrence. As with most other musculoskeletal injuries, athletes will begin a progressive return to the play program when they have shown full, pain-free range of motion about the shoulder as well as strength equal to the opposite uninjured joint. Surgical intervention is generally noted to be with good results in anterior, unilateral dislocations but is generally considered only after the athlete fails conservative management, has developed a history of frequent recurrence, or participates in a sport with a high risk for recurrence.

Other upper extremity joint dislocations/subluxations associated with football may occur at the elbow. *Elbow dislocation* involves complete slippage of the ulna posterior to the humerus and is usually associated with disruption of that elbow's ulnar collateral ligament (UCL), radial collateral ligament, anterior capsule, and brachialis musculature. *Elbow subluxation* involves the trochlea being perched on the coronoid process. The gross majority of both of these injuries occur posteriorly and result from a fall onto an outstretched hand (FOOSH) injury. Associated elbow fractures—most commonly at the radial head—are not uncommon. Reduction should be done expediently, with care to monitor the neurovascular status of the distal extremity. After confirmation by radiograph of successful reduction, the arm is generally splinted in a position of 90 to 100 degrees of flexion with early protected range of motion initiated. Return to full athletics is generally at 8 to 10 weeks postinjury.

Additional shoulder region injuries commonly seen in football include *acromioclavicular (AC) joint separations/sprains.* These commonly occur resulting from direct trauma to the shoulder from tackling/blocking or from a fall with the arm adducted across the body. The force of this impact can disrupt the AC and/or coracoclavicular (CC) joint(s) while the more stable sternoclavicular (SC) joint remains intact. The athlete presents with focal tenderness to palpation overlying the AC joint as well as pain with terminal 20 to 30 degrees of glenohumeral abduction and cross-body adduction. This injury includes various presentations and severities with Type I (AC joint ligament sprain alone) and Type II (AC joint tear and CC joint sprain) being the most common. No step-off is usually palpated in either Type I or Type II. A Type III sprain involves complete tears of both the AC and CC joint ligaments, a step-off is usually palpated, and radiographs will show varying degrees of clavicular displacement superiorly. Type IV to VI injuries are associated with varying degrees and directions of displacement of the clavicle with associated injury to surrounding musculature, brachial plexus, or vasculature. Type I and II AC joint injuries are treated conservatively with RICE principles, anti-inflammatories, and sling immobilization initially. Progression to protected range of motion and strengthening of the rotator cuff and shoulder girdle musculature is next. Treatment of Type III injury is debated with respect to nonsurgical versus surgical approaches, with the latter being the default if the athlete fails conservative treatment after 3 to 6 months, the injury is to the dominant arm of a high-level throwing athlete (ie, a quarterback or pitcher), or the athelete desires to return to a contact/collision sport with a high risk for reinjury. Type IV to VI injuries warrant orthopedic consultation for operative management.

Fracture of the clavicle is one of the most common fractures of childhood usually resulting from a direct blow or a fall onto the clavicle or lateral aspect of the shoulder, or FOOSH. More than 80% of these fractures involve the middle third of the clavicle. These are generally treated with RICE principles, anti-inflammatory medications, and sling or figure-of-eight immobilization. Healing time varies dependent upon age, ranging from 3 to 4 weeks in young children to 6 weeks in adolescents and young adults. Both clinical healing—no pain with palpation and full, pain-free range of motion—and radiographic healing—bridging callous on x-ray—are required for consideration for return to athletics. An additional 2 weeks after this point is often incorporated when the athlete is returning to a contact/collision sport. Treatment of clavicle fractures involving the proximal and distal third of the bone is commonly referred to orthopedics for definitive management.

Neck injuries in football are not uncommon with *cervical strain/sprain* predominating. These often occur as the result of poor mechanics in tackling such as spearing and can put the athlete at risk for cervical cord injury. Icing the injured cervical paraspinal musculature and judicious use of anti-inflammatories are often adequate in the short term, with gentle range of motion exercises incorporated as well. Radiographs are necessitated when there is midline cervical spine tenderness or significant active range of motion restriction. A *stinger* or *burner* is the name given to the most common nerve injury in football and describes a brachial plexus neuropraxia. Compression or traction injury to the upper nerve roots of the brachial plexus results in this injury—often occurring after making a tackle associated with strong, lateral displacement of the neck. Painful, unilateral dysesthesias, radiating down into the upper extremity, are frequently coupled with weakness in the C5/C6 innervated musculature, most notably in the deltoid. This injury is commonly temporary—with return to play requiring return of pain-free range of motion and strength that is equal to the unaffected limb—though resolution of symptoms may vary from minutes to days. In more severe cases lasting weeks, electromyographic evaluation is warranted after symptoms have lasted greater than 3 weeks to assess for active healing via reinnervation. *Transient quadriplegia* is an uncommon presentation of injury to the cervical region and primarily associated with the sport of football. The player presents on the field with paralysis of the extremities that is self-resolving, frequently within minutes. Computed tomography (CT) scan or MRI of the neck frequently shows evidence of cervical canal stenosis. Future return to contact and collision sports for those with this injury is controversial owing to a perceived increased risk of more severe cervical cord injury (7–20).

BASEBALL AND SOFTBALL

Injuries to the upper extremities are the predominate causes of lost time from athletic participation in baseball and softball. These are most frequently overuse injuries related to the high amount of overhead throwing inherent in each of these sports—especially in baseball pitchers. The muscles of the rotator cuff—the supraspinatus, infraspinatus, subscapularis, and teres minor—are particularly vulnerable to injury due to the shoulder's reliance upon them to maintain glenohumeral stability. *Rotator cuff injury* most commonly involves the supraspinatus muscle and may present as an acute strain or more subacute/chronic tendinosis. This frequently presents with complaint of pain with movement of the arm overhead such as throwing, lifting, or reaching for objects. Location of pain is variable about the shoulder and may be complicated by the presence of other concomitant shoulder pathology. Scapulothoracic dyskinesis—abnormal shifting in the typical position or motion of the scapula during coupled scapulohumeral movements—weakness in the scapular stabilizers and/or rotator cuff muscles, core weakness, and poor throwing mechanics are common contributing factors to the development of a rotator cuff injury. Pain persists with continued throwing or overhead activities and may be associated with reduced velocity or accuracy of throws. The shoulder exam reveals pain and, on occasion, weakness of the affected arm with isolated testing of injured rotator cuff muscle(s). Glenohumeral internal rotation deficit (GIRD) is also often present on exam—evidenced by increased glenohumeral external rotation range of motion coupled with decreased internal rotation range of motion in the painful throwing arm that is asymmetric in comparison to the opposite arm. Initial treatment often requires a period of relative rest from throwing and other overhead activities, icing of the shoulder, and anti-inflammatory use. This is followed by a rehabilitative protocol focusing upon strengthening of the rotator cuff and scapular stabilizers, addressing GIRD, if present, with specific stretching exercises, as well as evaluation of throwing mechanics and core strengthening. In the pediatric and adolescent athlete, rotator cuff impingement plays a less significant role in rotator cuff injury than in adults as *anterior glenohumeral instability* is the primary contributor. This occurs over time as the anterior shoulder capsule becomes repetitively stretched due to the mechanics of the overhead throwing motion allowing for increasing amounts of anterior–posterior translation of the humeral head within the glenohumeral joint. On exam, the athlete will commonly have discomfort with the anterior glenohumeral apprehension test followed by resolution of pain with a relocation maneuver preventing anterior translation of the humerus. The contribution of glenohumeral instability to rotator cuff injury underscores the need for glenohumeral stabilization exercises more than those focused upon stretching of the shoulder capsule and rotator cuff musculature. Final steps in return to play include completion of an interval throwing program that is commonly initiated once the athlete exhibits pain-free range of motion and strength equal to the opposite arm.

Instability of the glenohumeral joint may lend itself to the development of a *glenoid labrum tear.* This can be chronic in its presentation—or acute such as occurring secondary to a traumatic dislocation. Pain is frequently described as being located deep within the shoulder and may be associated with complaint of a clicking or popping sensation. The most common of these labral lesions in the throwing athlete is the *superior labrum anterior and posterior lesion* (SLAP). Tearing or fraying of the superior glenoid labrum at the attachment of the long head of the biceps is felt to occur due to excessive traction force applied to the site with the mechanics of overhead throwing—particularly deceleration of the arm. On exam, provocative tests of the labrum, such as O'Brien's test, is commonly positive. Radiographs are generally normal and MRI with arthrogram is the imaging modality of choice to evaluate for glenoid labrum injury.

In the evaluation of the pediatric and adolescent athlete with shoulder pain, special care needs to be taken to keep in mind the presence of open physes—especially at the proximal humerus. *Little leaguer's shoulder (proximal humeral stress fracture)* is an uncommon but potentially serious cause of shoulder pain in this population—as 80% of longitudinal growth of the humerus comes from its proximal physis. Presentation is often insidious and may last months before presentation to the physician. Tenderness to palpation over the proximal humerus is often present as can be pain at rest. Plain radiographs may show asymmetric widening of the proximal humeral physis in comparison to the opposite arm. Further evaluation with MRI of the shoulder can confirm the diagnosis, showing physeal edema. Treatment of little leaguer's shoulder includes discontinuation of all overhead throwing for a minimum of 6 to 8 weeks followed by a rehabilitative protocol similar to that utilized with rotator cuff injury, and then initiation of an interval throwing program.

Elbow pain in the young throwing athlete is not uncommon with 20% of pitchers aged 10 to 14 complaining of elbow pain. The majority of injuries at the elbow, like those at the shoulder, are overuse injuries. *Little leaguer's elbow* is a common term used to describe one of many chronic, overuse injuries about the elbow. Medial elbow pain is most common and related to muscular strain of the wrist flexor/pronator complex arising at the medial epicondyle. Valgus stress at the elbow during the throwing cycle results in opening of the medial joint line and places excessive stress upon these muscles. In this scenario, the skeletally immature athlete is at particular risk for development of *traction apophysitis of the medial epicondyle*. Pain is noted overlying the medial epicondyle and is worsened with resisted wrist flexion, pronation, and valgus stress of the elbow. Widening of the medial epicondylar apophysis as compared to the opposite arm may be noted on plain radiograph. MRI of the elbow will show edema at the apophysis. Initial treatment includes cessation of throwing upwards for 4 to 6 weeks—to prevent nonunion of the apophysis—as well as rehabilitation focusing upon strengthening and stretching of the wrist flexor–pronator musculature. Subsequent completion of a short-interval throwing program allows for return to play.

The repetitive valgus stress placed upon the elbow during the throwing cycle may result in a *UCL sprain*. The complaint of pain is generally greatest during the acceleration phase of throwing as valgus stress is highest. Over time, stretching or tearing of the UCL may develop, sometimes progressing to the point where the athlete describes feeling a sensation of the elbow joint opening when throwing. Tenderness over the UCL on exam is noted. Laxity of the UCL with valgus stress placed upon the elbow joint is not unusual. Plain radiographs generally are normal. MRI is most commonly utilized to assess for UCL injury. While partial tears can be treated with a period of rest from throwing and then a progressive rehabilitation program similar to other causes of medial elbow pain, full-thickness tears require surgical intervention if return to a throwing sport is desired. An uncommon complication of UCL sprain, *ulnar neuritis*, may occur due to the repetitive valgus stress at the elbow during the throwing cycle causing stretching of the ulnar nerve. Less frequently, subluxation of the ulnar nerve out of the cubital tunnel may occur. Dysesthesias along the lateral forearm into the fourth and fifth digits of the hand (ulnar nerve distribution) are often noted. Ulnar neuritis may be treated conservatively in most cases. If associated with a complete UCL tear, it is commonly repaired surgically as part of what is termed "Tommy John" surgery, fixing both injuries.

Lateral elbow pain in the throwing athlete results from lateral compressive forces to the elbow joint during the throwing cycle—primarily at the radiocapitellar joint. *Osteochondritis dissecans (OCD)* of the capitellum and *Panner's disease* are two causes of lateral elbow pain in the young throwing athlete. Both are chronic in onset of pain, with throwing commonly worsening slowly over weeks to months. OCD tends to occur in early adolescence, at ages 13 to 16, and is frequently associated with loss of range of motion with decreased elbow extension as well as complaints such as joint locking or popping. Plain radiographs may show focal lesion development within the capitellum that may have progressed to becoming fragmented and displaced from the distal humerus. MRI of the elbow may show evidence of OCD of the capitellum before it is noted on radiographs. Orthopedic consultation is recommended, with treatment varying dependent upon the progression of the lesion and presence of displaced fragmentation of the capitellum. Contrasting with this, Panner's disease does not tend to result in range of motion reduction or mechanical symptoms. Treatment consists of relative rest from throwing for a period of time, possible short-term immobilization, and serial radiographs to assess for bone healing over 6 to 12 weeks. Outcomes in athletes with Panner's disease are generally good (15–17,21–27).

BASKETBALL AND VOLLEYBALL

Both acute traumatic and chronic overuse injuries to the knees and ankles are common to these sports owing to the frequent movements inherent in each sport including running, jumping, and sudden changes in direction on hard surfaces. *Patellar tendinosis*, also known as *jumper's knee*, is the most common of the overuse injuries. It is caused by repetitive contraction of the knee extensor complex with the majority of the force overloading the patellar tendon. Activities such as jumping, stair climbing, running on an incline, and repetitive knee extensor resistive strength training can contribute to its onset. Lack of hamstring and quadriceps flexibility is a risk factor along with "doing too much too soon." Pain on exam is often relegated to the patellar tendon—but may also involve the inferior pole of the patella—and can be marked, limiting the athlete's

ability to participate in sport. Treatment includes a period of relative rest from the inciting sport, icing, and anti-inflammatory use. This is then followed by stretching and protected strengthening of the lower extremity musculature. The use of a patellar strap to modify the fulcrum of force over the patellar tendon may be helpful.

Osgood–Schlatter disease is another common cause of knee pain in the pediatric and adolescent athlete. This traction apophysitis presents as point tenderness overlying the tibial tuberosity at the site of the insertion of the patellar tendon. Pain may be to a level of severity where athletic participation is limited—especially if associated with concomitant patellar tendinosis—though it often tends to be most painful whenever the inflamed tibial tuberosity is met with a direct impact. Conservative management incorporating relative rest, icing, anti-inflammatories, and protective padding is most common. Strengthening of the core and patellar-stabilizing musculature is helpful as well. Although the overall prognosis for Osgood–Schlatter is quite good, the course of resolution is frequently prolonged, taking months to years during adolescence. *Sinding-Larsen–Johansen syndrome* is similar to Osgood–Schlatter though occurring at the inferior patellar pole in contrast to the tibial tuberosity. Pain is located overlying this site which, on plain radiographs, gives the appearance of small avulsion-like fragmentation. Treatment is similar to Osgood–Schlatter—although the use of a patellar strap, as with patellar tendinosis, may provide additional benefit.

Similar in its presentation as a cause of anterior knee pain, *plica syndrome* involves inflammation in one of four synovial tissues about the knee joint originating from embryonic development. Commonly, these tissues are fully resorbed during fetal development but if this occurred incompletely they will remain. Inflammation of the plica is thought to result from direct trauma, repetitive stress/overuse, meniscal tears, or other knee pathology. Pain occurs with repetitive flexion of the knee and the plica rubbing against one of the bony structures about the knee—with the medial plica abrading the medial femoral condyle as the most commonly proposed site—though this explanation is controversial. Tenderness on exam over the area of the inflamed plica as well as palpation of the structure itself is common. RICE principles, anti-inflammatory use, and stretching/strengthening of the knee extensor/flexor mechanism may be helpful. In reticent cases, surgical removal of the painful plica is performed.

Finger sprains and fractures are not uncommon in basketball and volleyball owing to the frequent passing, catching, handling, and attempted blocking of the ball to varying degrees in each sport. These can generally be treated with minimal periods of immobilization or simply buddy taping dependent upon the location and severity of the injury. Contrasting with this, a *mallet finger* involves injury to the extensor digitorum tendon of the finger usually resulting from a hyperflexion injury at the distal interphalangeal (DIP) joint. This commonly occurs when the athlete catches a basketball awkwardly and an extended finger becomes jammed, forced into hyperflexion disrupting the distal extensor tendon. The athlete will usually present with an inability to fully extend at the DIP joint of the injured finger and tenderness over the dorsum of the DIP joint. Plain radiographs are performed to assess for the presence of an avulsion fracture which, if substantial, may require surgical fixation. Otherwise, conservative treatment with continuous splinting of the injured finger in hyperextension at the DIP joint for a minimum of 6 weeks is commonly performed (13,14,20,28).

SWIMMING

The most commonly injured joint in swimmers is the shoulder. More often than not these injuries are due to chronic overuse and not acute trauma. *Swimmer's shoulder* is the term used to describe the common constellation of shoulder pathology including tendinosis of the rotator cuff and long head of the biceps, subacromial bursitis, and glenoid labrum injury seen frequently in this athletic population. *Multiaxial shoulder instability* is a primary contributing factor to the development of these other diagnoses and results from the development of increased laxity about the shoulder capsule over time, resulting from the wide arcs of movement inherent in numerous swimming strokes—most notably freestyle, backstroke, and butterfly. Scapulothoracic dyskinesis and weakness of the scapular stabilizers also play a role. The athlete not only complains of pain during swimming but also frequently describes pain with activities of daily living such as reaching for objects above the level of the shoulder and dressing. Marked glenohumeral laxity may have the athlete describe a sensation of instability with these movements. On exam, positive provocative testing such as the Hawkins impingement test (arm abducted to 90 degrees, elbow flexed to 90 degrees, and the humerus internally rotated by the examiner) and the "empty can" test (resisted shoulder abduction in the scapular plane—with the arms abducted to 90 degrees and forward flexed 30 degrees, elbows fully extended, humerus fully internally rotated) are frequently noted. Glenohumeral instability is assessed on exam as well in order to document multiaxial instability. Treatment incorporates rest from swimming or modification of swimming technique as well as a rehabilitation program focused upon strengthening of the muscles of the rotator cuff and the scapular stabilizers (15,16,29).

RUNNING SPORTS

The gross majority of injuries seen in runners are of the chronic, overuse variety affecting the lower extremities. Due to the repetitive nature of the lower extremity movements and associated impacts inherent in training for their

sports, running athletes can be placed at particular risk for development of injury due to a wide variety of factors. These can include training errors such as increasing one's distance or duration of running too quickly, changes in running surfaces, improper shoe wear, and anatomic considerations (ie, overpronation, increased Q-angle, minor skeletal asymmetry, limited muscular flexibility, or poor core stability).

Knee pain in the runner is most commonly located anteriorly with *runner's knee (patellofemoral stress syndrome—PFSS)* being the most common diagnosis. This usually involves a component of *chondromalacia patellae* with softening of the retropatellar articular cartilage in addition to simple inflammation. The etiology of the athlete's pain is felt to be related to excessive movement of the patella within the femoral intracondylar groove during running. This most commonly occurs in a lateral direction and contributing factors include one or more components of *miserable malalignment syndrome* (widened Q-angle, femoral anteversion, genu valgum, external tibial torsion, and overpronation) with weakness of the vastus medialis and/or core musculature. Patients report diffuse, retropatellar pain that increases with activities such as running, stair climbing, or squatting, as well as prolonged periods of sitting. Frequently a recent increase in training intensity, frequency, or duration is noted. Physical examination includes evaluation for those anatomic risk factors noted previously as well as overall patellar laxity and lower extremity strength and flexibility. A positive patellar grind test (compression of the patella into the intracondylar groove with the knee extended and the quadriceps actively contracting) is confirmatory. Imaging is not usually necessary to make the diagnosis of PFSS. Treatment focuses upon strengthening the patellar stabilizers and core musculature as well as improving lower extremity flexibility. Addressing anatomic issues contributing to the athlete's symptoms with orthotics, kinesiotaping, or making changes in running biomechanics may also be beneficial.

Medial tibial stress syndrome (MTSS), or *shin splints,* is also commonly seen in running athletes and presents with the gradual onset of diffuse pain overlying the medial aspect of the middle or distal third of the tibia. MTSS is the most common overuse injury of the lower leg and is frequently seen in individuals with overpronation or those who have increased the duration of their training regimen. Pain worsens during impact-loading activities. Radiographs of the lower leg are normal. Treatment typically includes RICE principles, anti-inflammatory use, and may also incorporate cross-training, changing running surfaces to those more forgiving, recommendations on proper shoe wear, and orthotic prescription. In contrast to MTSS, a *tibial stress fracture* tends to present with pain at a focal site on the tibia and oftentimes is of a greater severity with pain at rest being a common complaint. Tibial stress fracture is on the continuum of lower leg stress injury and may develop after having initially presented as MTSS, progressing to stress fracture due to continued impact loading by the athlete. Radiographs may show periosteal thickening or cortical defects. Bone scan will show focal tracer uptake in contrast to a more diffuse picture seen with MTSS. MRI may be beneficial in cases where the diagnosis is unclear. Treatment for tibial stress fracture requires removal from impact loading and, in some cases, a period of protected weight-bearing via boot immobilization for up to 4 to 6 weeks. For both MTSS and tibial stress fracture, a progressive weight-bearing program is utilized for return to play, carried out over a varied time course based upon the athlete's recurrence of pain, if any, during rehabilitation.

Sever's disease (calcaneal apophysitis) is seen not only in running athletes but generally across the board in almost all sports that require pediatric and adolescent athletes to run. The underlying pathology is not dissimilar to Osgood–Schlatter disease at the tibial tuberosity as it is a traction apophysitis occurring at the attachment of the Achilles tendon to the calcaneal apophysis. Pain is related to the level of activity exacerbated by running and, in particular, jumping. It is most commonly seen in boys between the ages of 8 and 13 during periods of rapid growth with loss of heel cord flexibility increasing the pull upon the calcaneal apophysis. Physical exam is significant for tenderness to palpation or squeezing of the calcaneus—most notable posteriorly and inferiorly. Radiographs are usually not necessary for making this clinical diagnosis. Treatment includes RICE principles, heel cord stretching, anti-inflammatories, and strengthening the dynamic stabilizers of the ankle. In some cases, use of a cushioned heel lift reduces force through the Achilles tendon and pads the heel. Activity participation is generally allowed based upon the child's tolerance of symptoms. *Achilles tendinopathy* may occur concomitantly with Sever's disease or be an isolated diagnosis. Pain is located within the distal fibers of the heel cord, frequently 3 to 6 cm superior to its calcaneal attachment. On exam, focal tenderness over the Achilles tendon is noted as is pain with resisted ankle plantarflexion, heel raises, and/or jumping. Treatment is similar to that of Sever's disease as already reviewed, though it may require a longer period of relative rest away from sports before initiating a progressive return to play.

In the foot, *plantar fasciitis* occurs infrequently in the pediatric and adolescent population. It involves inflammation and degeneration of the supporting structures of the longitudinal arch. The affected athlete frequently reports pain along the medial aspect of the heel that is at its greatest with the first step(s) in the morning or after long periods of non-weight-bearing. Common contributing factors include pes planus, overpronation, and use of shoe wear with inadequate arch support. Treatment involving relative rest from inciting activities or weight-bearing in general (in more severe cases) is often coupled with stretching of the heel cord and plantar fascia. Therapeutic ultrasound and/or use of night splints

that provide a passive heel cord and plantar fascia stretch may provide additional benefit in more persistent cases. Return to sport is allowed, provided the athlete does not show signs of antalgia when participating.

Sesamoiditis is another potential source of foot pain in the running athlete. The sesamoids do not fully ossify until 7 to 10 years of age and as such this is seen more commonly in adolescent runners. Their primary role is to absorb the majority of impact forces along the medial aspect of the forefoot. In addition to running sports, sesamoiditis can be seen in a variety of other sports such as tennis, gymnastics, dance, and soccer. Discomfort is commonly noted at the plantar aspect of the first MTP joint of the foot occurring during the toe-off stage of gait and with most jumping or running activities. Examination may show swelling at the first MTP joint with the athlete experiencing tenderness overlying the affected sesamoid(s). Radiographs may show evidence of a bipartite sesamoid, which can be differentiated from sesamoid fracture by bone scan. Treatment focuses on limiting movement of the great toe via taping, use of a stiff soled shoe or footplate, or even short-term boot/cast immobilization in more difficult cases.

Pain over the medial aspect of the ankle or foot in the runner may represent *posterior tibialis tendinopathy*. This overuse injury results from overloading of the posterior tibialis musculotendinous unit during running often in response to overpronation of the foot with pes planus as the foot tries to maintain itself in a talar neutral position at impact. Pain on exam is commonly noted along the course of the posterior tibialis tendon posterior to the medial malleolus, extending along the medial aspect of the foot to its insertion in the navicular and medial cuneiform bones. Weakness with resisted inversion is not uncommon and frequently associated with pain. If there is notable tenderness and swelling at the navicular bone in a skeletally immature athlete, consideration for a diagnosis of an inflamed *accessory navicular bone* should be made. These patients typically also complain of pain with rubbing of the accessory navicular within their shoes, and callous overlying the navicular is not uncommon. Plain radiographs can be helpful in confirming this diagnosis. Treatment for both posterior tibialis tendinopathy and inflamed accessory navicular bone includes relative rest from impact-loading activities, ice, anti-inflammatories, and strengthening the posterior tibialis and the other dynamic stabilizers of the ankle joint and arch of the foot. In cases felt to have a strong contribution from the athlete's overpronation and pes planus, prescription of a custom-molded orthotic can prevent recurrence (14,30–35).

WRESTLING

Many injuries common to the sports of wrestling are seen in other sports as well. The joints of the wrestler are at particular risk for injury due to subluxation/dislocation due to the amount of torque and force placed upon them related to the variety of holds utilized in the sport. At the shoulder, glenohumeral subluxation/dislocation (see Football) is not uncommon and primarily unidirectional (anterior). Injury to the fingers also occurs with sprains of the MCP and proximal interphalangeal (PIP) joints predominating. For the most part, these can be treated with a short period of splint immobilization followed by buddy taping with early, protected range of motion exercises. Prolonged immobilization via splinting is frequently avoided to prevent stiffness.

Apart from the shoulder, the most common joint injured in the wrestler is the knee. *Prepatellar bursitis* occurs secondary to repeated impacts to the knees. The wrestler notes swelling over the anterior aspect of the knee, which may be associated with range of motion restriction with knee flexion. Without the presence of marked erythema, tenderness, or fever, the likelihood of the bursitis being secondary to infection is low—though aspiration can be diagnostic. Treatment includes the use of protective padding over the knee as well as a compressive wrap, icing, and anti-inflammatories. Aspiration and drainage may be helpful—especially if range of motion is compromised—though recurrence after the procedure is not uncommon. Surgical intervention via bursectomy in reticent cases is rare. Also at the knee, *lateral collateral ligament (LCL) sprain* is seen in this athletic population. This results from a varus force about the knee joint—which can occur during a single-leg takedown maneuver. Swelling is primarily localized and a knee effusion is not present due to the nature of this injury being extra-articular in location. These injuries tend to be fairly mild, and conservative treatment, including short-term use of a hinged brace for support during return to athletics, tends to be adequate. Rarely, surgical intervention is necessary with full-thickness LCL tears (10,36).

GYMNASTICS

Gymnasts experience a wide variety of both acute and chronic injuries to the extremities as well as the axial skeleton owing to the intense amount of training needed to compete at a high level in their sport. It is not uncommon for gymnasts to begin their training at the young age of 4 to 5 years old with their most competitive years being in the middle to end of adolescence for girls and young adulthood for boys. Wrist injuries are more common in gymnastics than most other sports due to the frequent weight-bearing through the upper extremities on events such as floor exercise, pommel horse, and vault. Most common are wrist sprains involving one or more of the many ligaments about the wrist running between the radius, ulna, and carpals. Activity modification, RICE principles, anti-inflammatory use, and

limited immobilization followed by range of motion, stretches, and wrist strengthening exercises make up the rehabilitation protocol.

Gymnast's wrist (*distal radius physis stress injury*) occurs due to repetitive compression force on a dorsiflexed, radially deviated wrist—a position commonly utilized during floor routine and vault (37,38). The majority of those affected are female and in early adolescence during periods of rapid growth. Focal tenderness over the distal radius limiting athletic participation is the common presenting picture. Plain radiographs frequently show widening of the distal radial physis of the affected wrist. Initial treatment includes removal from weight-bearing activities through the upper extremities until resolution of pain, which may be 2 to 4 weeks. Short-term immobilization is needed in more severe cases. Potential for untreated injury to cause premature physeal fusion, growth arrest, and radial shortening is an important consideration. Also at the wrist, *triangular fibrocartilage complex (TFCC) tear* is an infrequent but potentially debilitating injury. The TFCC is the primary stabilizer of the distal radioulnar joint (DRUJ) and is made up of various ligamentous and cartilaginous structures that allow for smooth translation of the carpals over the distal radius and ulna in flexion and extension as well as limited rotation of the radiocarpal unit along the ulnar axis. Injury typically occurs secondary to a fall onto a pronated, hyperextended wrist. This can occur with or without a rotational force. Athletes frequently complain of clicking at the wrist with movement and pain with ulnar deviation. Radiographs frequently show ulnar negative variance. MRI is helpful to confirm the diagnosis. Conservative treatment includes prolonged immobilization ranging from 4 to 10 weeks followed by supervised rehabilitation. Surgical intervention via arthroscopy or an open approach is generally indicated in cases of persistent instability of the DRUJ despite conservative management or with more severe tears.

Gymnasts are at particular risk for stress injury to the lumbar spine including *spondylolysis* and *spondylolisthesis*. *Spondylolysis*, a stress fracture of the pars interarticularis, most frequently occurs at the L4/L5 and L5/S1 levels. Mechanism for injury is felt to be related to hyperextension injury, most commonly chronic in nature though acute spondylolysis secondary to direct injury has been described. Most commonly, repetitive hyperextension of the lumbar spine, such as that seen frequently in many of the movements in gymnastics, results in the development of stress reaction at the pars, which can progress to stress fracture. There is some thought that individuals with these injuries may have had pre-existing hypoplastic pars placing them at increased risk for development of spondylolysis. Pain is exacerbated with extension, with rare radiation into the gluteal muscle groups. On exam, pain is noted with deep palpation lateral to the affected lumbar level. Lumbar extension elicits pain and is most marked with stressing of the pars such as coupling extension with single-leg stance. Plain radiographs may show the pars defect on oblique films and spondylolisthesis on lateral films. Bone scan with single-photon emitted computerized tomography (SPECT) will show increased tracer uptake in the area of the spondylolytic lesion often used in the case of normal radiographs or to help decide acuity of injury. CT scan and MRI are also becoming more increasingly used as diagnostic tools. Treatment initially focuses upon activity restriction and comfort measures. Initiation of a rehabilitative program emphasizing lumbar stabilization—without extension—hamstring flexibility, and core strengthening is subsequent. The efficacy of using a thoracic–lumbosacral orthosis (TLSO) in the early phase of healing is debated.

Spondylolisthesis involves slippage of a vertebral body upon the next inferior vertebral body and requires bilateral spondylolysis. This is generally a gradual occurrence and not the result of an acute trauma. Spondylolisthesis is graded from 1 to 4 based upon the degree of slippage of one vertebral body upon the other. Grades 1 and 2 (<50% slippage) are generally treated conservatively in a fashion similar to that of spondylolysis. Serial radiographs are taken every 3 to 6 months to assess for progression of slippage until skeletal maturity. Grades 3 and 4 (>50% slippage) spondylolisthesis warrant orthopedic consultation and often surgical fixation (15,33,39–44).

SOCCER

Soccer-related injuries predominantly involve the lower extremities and include many of those previously described as occurring in running sports. Apart from contusions and skin abrasions due to contact between players, the ball, or the ground, the most common injury seen in soccer—and in all of athletics—is injury to the ankle.

The *lateral ankle sprain* is the most common of these injuries comprising 85% of all ankle sprains. Lateral ankle sprains result from an inversion injury with the foot planted. These ankle sprains involve injury to one or more of the lateral ligaments of the ankle—the anterior talofibular ligament (ATFL), the calcaneofibular ligament (CFL), and the posterior talofibular ligament (PTFL). With all ankle injuries, initial evaluation on the field should focus upon whether there is evidence of a more serious trauma such as a fracture or dislocation that requires immediate neurovascular evaluation, immobilization, and orthopedic consultation. Barring these findings on the field, or in the clinic evaluation, the examiner should pay special attention to the location and severity of swelling, ecchymosis, and any deformity, if present. Palpation of the medial and lateral ankle ligaments should be performed along with other important anatomic structures including the ankle joint lines, medial and lateral malleoli, Achilles tendon, syndesmosis, anterior tibiofibular ligament, the navicular, and base of the fifth metatarsal. The "squeeze test" of the proximal tibia and fibula may elicit pain inferiorly just superior to the ankle joint in the setting of a high

ankle, or syndesmotic sprain. Range of motion assessment and strength testing, if not significantly limited by pain, is also performed. Next, focused evaluation of the laxity/stability of the ankle ligaments is extremely important. The anterior drawer test evaluates the ATFL while the talar tilt test is used to assess the CFL primarily. The external rotational stress test is used in conjunction with the aforementioned squeeze test to evaluate for high ankle sprains. Grading of ankle sprains is based upon the degree of instability noted within the injured ligament(s). Grade I ankle sprains tend to have tenderness to palpation at the injured ligament but no instability noted in comparison to the uninjured ankle. Grade II ankle sprains are tender as well but also exhibit increased ligamentous laxity of the partially torn ligament(s) in comparison to the opposite side but maintain a definite end point with stress. Grade III ankle sprains are the most severe, exhibiting more marked laxity on exam that is without a discernible end point due to complete rupture of the ligament(s) injured.

Radiographs are indicated when criteria for Ottawa ankle rules are met—tenderness to palpation overlying the malleoli, bony tenderness over the distal tibia or fibula, and inability to bear weight for more than four steps at the time of injury and in the office. Radiographs of the foot should be performed if there is tenderness over the base of the fifth metatarsal to rule out possible avulsion fracture (see Dancer's fracture and Jones fracture in the section titled Dance). Additionally, when a *medial ankle sprain* is suspected based upon mechanism of injury (eversion) and exam findings (swelling, ecchymosis, and/or tenderness over the deltoid ligament), radiographs of the ankle and lower leg are indicated—the latter to rule out possible proximal fibular fracture. In the case of the suspected high ankle sprain, stress radiographs of the ankle mortise, looking for widening of the mortise with compression of the proximal tibia and fibula suggestive of ankle joint instability, aid decision making.

Initial treatment of most ankle sprains incorporates RICE principles, anti-inflammatories, protected weight-bearing, and range of motion exercises. Rehabilitation of Grade I or II lateral ankle sprains can generally begin as soon as tolerated so as not to let the ankle joint become stiff. Exercises include strengthening of the dynamic stabilizing musculature of the ankle and proprioceptive balance training. Grade III lateral ankle sprains may require a period of immobilization in a short-leg cast or walking boot before progressing to active rehabilitation. When the injured ankle exhibits pain-free range of motion and strength that is equal to the opposite, uninjured side, progressive dynamic activities and sport-specific drills, in preparation for full return to athletics, are initiated. For Grade II and III lateral ankle sprains it is commonly recommended to utilize a functional support such as a lace-up brace, stirrup brace, or taping during athletic participation upwards of 3 to 6 months after the injury for additional support in preventing reinjury.

Rehabilitation of medial ankle sprains is typically more prolonged than that of lateral ankle sprains typically taking a minimum of 3 to 6 weeks for even mild sprain. Even mild Grade I medial ankle sprains are treated with stirrup bracing with some Grade II medial ankle sprains requiring a short period of immobilization. Grade III medial ankle sprains are commonly casted for 6 to 8 weeks and may require surgical repair. Special consideration should also be made in the case of the skeletally immature child with lateral ankle pain—specifically focal point tenderness overlying the lateral malleolus at the distal fibular physis—and normal radiographs. The diagnosis of a Salter–Harris I fracture must be considered as the open physis is weaker than the surrounding lateral ligaments, resulting in injury with widening of the growth plate requiring treatment as an ankle fracture with short-leg casting or boot immobilization.

Due to the significant amount of continuous running, directional changes, and kicking inherent in the sport of soccer, muscle strains affecting the lower extremities are quite common and include: *hamstring strains*, *groin strains*, and *hip flexor strains*. Muscle strains are graded based upon the severity of the underlying muscle injury. First-degree strains are mild, partial tears of the underlying muscle tissue, are minimally painful, and do not generally result in loss of range of motion or strength. Second-degree muscle strains are more severe, partial tears involving a greater number of muscle fibers, are commonly associated with significant pain and visible ecchymosis, and usually result in loss of range of motion and strength of the affected muscle. Third-degree muscle strains are the most severe involving complete rupture of the injured muscle resulting in a palpable defect, as well as significant pain, range of motion loss, and weakness. In some instances, third-degree muscle strain may be associated with an avulsion injury at the origin or insertion of the affected muscle. Examples of this include *avulsion fractures of the ischial tuberosity* occurring with a hamstring strain and *avulsion fracture of the anterior–superior iliac spine or anterior–inferior iliac spine* with sartorius and rectus femoris strains, respectively (both are hip flexors). Muscular flexibility lags behind longitudinal bone growth in children, and with associated growth spurts and limited stretching or warm-up activities, epiphyseal avulsions are more common (45). Sometimes, late diagnosis of the avulsion of ischial tuberosity is mistaken for an osteosarcoma.

Acute management of these muscle strains emphasizes control/reduction in pain, edema, and hemorrhage into the muscle through the use of RICE principles. Immobilization with the muscle in the lengthened position, to prevent contracture and further range of motion loss, is also important. If tolerated, early, pain-free passive or active-assisted range of motion stretches are added near the end of the first week after injury. During the subacute phase of healing and rehabilitation, continued range of motion stretching is coupled

with low-intensity strengthening such as submaximal isometric exercises, stationary bike use, or pain-free pool running. Later, during the remodeling phase of rehabilitation, isotonic and isometric strength training may be added within pain-free guidelines as well as low- to moderate-intensity plyometrics to increase eccentric loading of the musculature. Finally, during the functional phase of rehabilitation, resistance strength training intensifies and sport-specific skills and drills are introduced with progressively increasing intensity and dynamics. After completion of this regimen and having range of motion and strength of the injured muscle equal to the uninjured opposite limb, the athlete is then able to resume full athletic participation (14,34,46–50).

CYCLING

As would be expected, injury to the lower extremities—particularly that related to overuse—is most common in those participating in cycling. *Iliotibial band (ITB) friction syndrome* is one such injury and is also commonly seen in runners. Discomfort commonly results from impingement of the posterior aspect of the fibers of the ITB at one of two sites along the lateral aspect of the femur: the greater trochanter and lateral femoral epicondyle. In addition to inflammation of the IT band itself, irritation of the bursae that underlie it at these sites may develop, causing painful bursitis. Flexion of the hip and/or knee to 30 degrees during pedaling or the gait cycle typically causes pain. Risk factors for development of ITB syndrome include improper training with too steep an increase in mileage, lean body habitus (reduced subcutaneous tissue between the ITB and underlying bone), genu varum, and weak hip abductors—the latter of these being especially problematic in runners, causing increased Trendelenburg and increased pressure at the greater trochanter. Symptoms typically present increasingly over time with pain at the greater trochanter or lateral femoral epicondyle most notable with running, prolonged cycling, and stair climbing. In addition to focal tenderness at these sites, the physical exam frequently reveals tightness of the ITB itself as indicated by a positive Ober's test (patient is side-lying with affected side up, knee flexed at 90 degrees, hip extended, the leg initially abducted at the hip and then allowed back into adduction while only the ankle is held—lack of return to adducted position is positive and suggests tightness of the ITB band). Treatment includes initial rest and icing along with anti-inflammatory use. Rehabilitation focuses upon stretching of the ITB, quadriceps, and hamstrings as well as a component of strengthening the core and thigh musculature (especially gluteal muscles). Iontophoresis or even corticosteroid injection may provide benefit in more resistant cases. Surgical release of a portion of the ITB is rarely indicated (51).

TENNIS

Injuries in tennis run the gamut involving both the upper and lower extremities and include both acute and chronic injury types. Sprains of the ankles and knees as well as muscle strains of the trunk and extremities are common and similar to those mentioned in other sports previously. The spine, particularly the low back, is put at risk for injury due to the frequent directional change of movement on the court by the tennis athlete and repetitive loading and torquing associated with serving and hitting groundstrokes. The overhead motion associated with serving also lends itself to rotator cuff and glenohumeral pathology. *Tennis elbow (lateral epicondylitis)* occurs due to repeated overloading of the wrist extensor–supinator mechanism with activities that require prolonged grip. Less common in the pediatric and adolescent population than in adults, Tennis elbow results in discomfort at the lateral elbow at the epicondyle and proximal wrist extensor musculature. Passive wrist flexion or active wrist extension elicits pain. Poor mechanics with the backhand stroke using "too much wrist," incorrect grip size, and use of a racquet strung too tight are common culprits in this sport. Treatment focuses upon RICE principles, stretching/strengthening of the forearm musculature, and proper mechanics. Forearm straps and counterforce bracing may provide benefit as well (52).

GOLF

Golfer's elbow (medial epicondylitis) is a relatively uncommon diagnosis in children with medial elbow pain. This is due mainly to the presence of open physes and apophyses at the elbow of the growing child, which are more prone to injury due to the weakness inherent in their growth cartilage in comparison to the adjacent bone and musculotendinous attachments (see Little Leaguer's elbow). In the skeletally mature adolescent though, golfer's elbow may occur, due to repetitive overloading of the wrist flexor–pronator musculature of the forearm at their origin on the medial humeral epicondyle. Pain on exam is elicited with passive wrist extension or resisted wrist flexion. Treatment is similar to that of tennis elbow and consists of RICE principles, use of anti-inflammatories, strengthening of forearm muscles, focusing on proper mechanics, forearm straps, and counterforce bracing.

DANCE

Dance injuries typically involve the lower extremities and include both acute and chronic types. Despite the variety of genres of dance performed—including jazz, tap, hip hop, modern, and ballet among others—all require

intense practice focusing upon learning and perfecting the performance of specific body and extremity movements through repetition. It is the frequent, repetitive nature of dance practice that lends itself to the development of overuse injury. *Metatarsal stress fracture* is not uncommon in dancers and is the second most common stress fracture seen in athletics overall. Overloading of the metatarsal bone due to recurring impact (ie, repetitive jumping/landing in dance, distance running) results in the development of fatigue fracture. Prolonged periods of training with repetitive impact loading, poor gait or dance mechanics, hard training surfaces, lack of proper footwear, and reduced bone health due to nutritional concerns are common risk factors. The athlete typically presents with complaint of gradual onset of pain over the forefoot at the affected metatarsal site that worsens with activity. On exam there is generally minimal swelling, if at all, though there is commonly marked tenderness to palpation at the fracture site as well as with axial loading of the metatarsal. Plain radiographs are usually normal though may show periosteal thickening over time. Bone scan is quite sensitive and can be helpful in confirming the diagnosis, as can an MRI. Treatment includes a period of rest from impact-loading activities, up to 6 weeks. The athlete can continue to maintain aerobic fitness by swimming or running in a pool during this period. A progressive return to dance or athletics, with increasing amounts of weight-bearing, is incorporated thereafter.

Snapping hip syndrome is another injury common in dancers. While this most frequently presents as an overuse injury, it can occur acutely as well. The athlete frequently describes a sensation of "snapping" or "popping" of the hip, which is oftentimes audible. This may or may not be painful and is generally felt by the athlete to be located laterally or anteriorly and deep within the groin. Sources of this can be considered internal—such as the iliopsoas tendon sliding over the pectineal eminence of the pelvis or an acetabular labral tear—or external—movement of the ITB or gluteus medius over the greater trochanter. Internal causes can often be reproduced on exam with extension of the flexed (30 degrees), abducted, and externally rotated hip. Radiographs of the hip are generally not helpful. Treatment focuses upon rest from inciting activities, analgesia, stretching of the tight musculature felt to be causative, and core strengthening.

A *dancer's fracture* is an avulsion fracture involving the base of the fifth metatarsal. This injury commonly results from landing awkwardly from a jump onto the lateral aspect of the foot when in an inverted position. This is treated with immobilization for 4 to 6 weeks followed by rehabilitation focusing upon strengthening the dynamic stabilizers of the ankle and foot. Progressive return to dance is initiated thereafter. Care must be taken not to overlook the possibility of a fracture at the base of the fifth metatarsal being a *Jones fracture,* which occurs slightly more distal along the fifth metatarsal at the metaphyseal–diaphyseal junction. A Jones fracture is at particular risk for poor healing due to the tenuous blood supply at the fracture site. If acute and nondisplaced, these fractures are generally treated with nonweight-bearing and casting for 6 to 8 weeks. Displaced Jones fractures, or those that show evidence of nonunion on plain radiographs (sclerosis with a persisting fracture line), will commonly require surgical fixation. In contrast to the predominance of lower extremity injuries seen in ballet and other, more common, dance genres such as jazz, tap, hip hop, or modern dance, individuals participating in breakdancing also commonly incur injuries to the spine and upper extremities—with the shoulders and wrists most frequently involved in the latter.

Breakdancing injuries are most frequently sprain/strain injuries and are due to the increased amount of weight-bearing through the upper extremities as well as significant stress placed upon the lumbar spine with many of the twisting movements. Wrist sprains may require a short period of time off from weight-bearing "tricks." Splinting may be beneficial in some cases but rarely results in prolonged time away from breakdancing. The chronic lumbar sprain/strain picture frequently seen in this population has been termed "breakdance back syndrome" and is not generally associated with bony pathology such as spondylolysis or facet joint arthropathy. The cervical spine is also placed at particular risk from both compressive and torsional stresses when a head spin is performed, which can lead to cervical sprain/strain. Maintenance of excellent core strength and flexibility can reduce the risk of injury in breakdancing (14,33,35,48,49,53,54).

SKIING/SNOWBOARDING

Injuries in skiing and snowboarding are most commonly acute and secondary to falls such as contusions and lacerations. Advances in equipment design have resulted in decreased numbers of lower extremity injuries though they continue to represent two-thirds of all injuries in these sports. Serious injuries such as a *boot top fracture* can occur when the binding holding the boot to the ski or snowboard does not release. This is a high-impact injury that results in fracture of the tibia and, on occasion, the fibula just above the level of the rigid boot worn by the skier/snowboarder. When both bones are broken, the injured leg is unstable generally requiring surgical fixation followed by prolonged cast immobilization to ensure full fracture healing.

In the upper extremity, *Skier's thumb* is commonly associated with a FOOSH mechanism of injury, with the thumb hyperextended and abducted. This results in a sprain of the UCL of the first metacarpophalangeal joint. Pain and swelling is generally relegated to the medial joint line of the first MCP joint. On exam, joint stability must be assessed with injuries involving complete tears of the UCL, and greater than 45 degrees of joint opening

commonly requires surgical fixation. Less severe sprains can usually be treated conservatively with placement into a thumb spica splint for 4 to 6 weeks' duration. Treatment of Skier's thumb injuries may be complicated by a Salter–Harris III avulsion fracture of the base of the proximal phalanx or by the development of a Stener lesion—entrapment of the proximal aspect of the UCL at the adductor pollicis aponeurosis—both of which generally require surgical fixation for correction (55).

ICE HOCKEY

As a result of being a contact/collision sport, the most common ice hockey injuries include contusions, muscle strains, ligament sprains, and lacerations. Concussions, fractures, and dislocations (primarily the shoulder) are also seen. Although not specific to ice hockey, *osteitis pubis* is seen more frequently in this sport in comparison to most others due to the frequent medial/lateral movement of the pelvis and lower extremities associated with skating and potential for overuse of the hip adductor musculature. The affected athlete commonly complains of severe groin pain with activities such as skating, running, or kicking felt due to tendinosis of the hip adductor musculature. On exam, pain with palpation over the adductor tendons and pubic symphysis is noted. Weakness with resisted hip adduction due to pain is not uncommon. Widening of the pubic symphysis, sclerosis, and osteolysis of the os pubis may be seen on plain radiographs though this may not be noted early on in its course. MRI or bone scan though can be helpful during this period of time. Treatment includes relative rest from inciting activities, anti-inflammatories, and rehabilitation focusing upon hip range of motion, and core strengthening. Corticosteroid injection into the pubic symphysis may also be helpful. Time frame for return to play is often prolonged, upwards of 6 to 12 weeks dependent upon the duration of symptoms at the time of treatment initiation (10,49).

SKATEBOARDING

A wide variety of injuries are seen in those individuals participating in skateboarding, the majority being acute, macrotrauma injuries secondary to loss of balance resulting in a fall. These equally involve both the upper extremities—commonly resulting from a FOOSH injury—and lower extremities—frequently secondary to missing an aerial maneuver and landing awkwardly on the ground. With many skateboarding "tricks" being performed at a significant rate of speed (some upwards of 40 mph with high elevation), there is an inherent risk of serious injury.

A distal radius fracture can result from a FOOSH injury and is one of the most common types of fractures seen in the pediatric population and most common fracture of the upper extremity. When weight-bearing through the upper extremity, the majority of force is transmitted through the radius. In the pediatric population, the distal metaphyseal–diaphyseal region of the radius is particularly susceptible to fracture during the growing years due to weakness in the bone's relatively thinned cortex at that site related to longitudinal growth. These fractures most commonly do not involve the physis but will frequently have some degree of angulation and, in some cases, displacement, which may necessitate closed reduction. A distal radius fracture is generally treated with placement into a short-arm cast for 4 to 6 weeks duration based upon the athlete's age. This is followed, on occasion, by a short period of use in a wrist immobilizer during initial return to play. Though remodeling potential for angulated radius fractures in the pediatric and adolescent population is extremely good, care must be taken to closely follow healing with repeat radiographs and serial examination of range of motion including forearm supination and movements about the wrist. The use of protective wrist guards when skateboarding can prevent this type of injury.

Cervical injuries, concussions—and more severe traumatic brain injuries—are also not uncommon when skateboarding athletes are unable to complete inversion maneuvers correctly, landing upon their head and neck from high elevation. The incidence of these injuries increases with age and is more common in males than females. Reduced use of helmets in older children and adolescents, as well as their increased speed of skating contribute to this fact. An increasingly more common intervention within urban areas aimed at reducing the risk of injury to skateboarders as well as those people around them when skating in public spaces has been the construction of skateboard parks. These parks effectively isolate the skateboarders from potential collisions with pedestrians, bicyclists, and traffic, while also encouraging adult supervision (42,56,57).

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12

CONCUSSION MANAGEMENT AND REHABILITATION

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INTRODUCTION

A 16-year-old male football player has just been tackled and is lying still on the ground. He is unconscious but awakens after 2 minutes complaining of headache. He is unsure what happened and does not recall the events of the day leading up to his hit. He does not recognize his coach or teammates and keeps repeating himself.

Another 16-year-old male football player has just tackled another player. He falls to the ground, but gets up immediately. He appears to be running normally and does not seem to have any symptoms. He notes a mild headache to himself but continues to play in the game. The following morning, he complains of severe headache, nausea, and dizziness.

While most people would recognize that the first football player sustained a concussion, many, including coaches, athletic trainers (ATs), and physicians, would fail to recognize the second player's concussion without an appropriate sideline evaluation. This is partially due to failure of the athlete to report symptoms and to recognize postconcussive signs and symptoms. Both of the athletes have concussions despite their very different presentations, and both require proper medical evaluation and treatment. The purpose of this chapter is to describe the diagnosis, recognition, evaluation, and treatment of all pediatric patients with concussion.

WHAT IS A CONCUSSION?

A concussion is a type of mild traumatic brain injury (TBI), which may occur with or without an associated loss of consciousness. A concussion occurs by a direct blow to the head or a jolt to the body with forces transmitted to the head and leads to functional changes within the brain. A concussion is diagnosed and recognized based on the presence of signs and symptoms following an injury and not by radiographic findings or documented loss of consciousness. Generally, concussion symptoms are completely reversible without any long-term neurologic or cognitive sequelae.

PATHOPHYSIOLOGY OF CONCUSSION

The mechanical insult, which occurs during a concussion, initiates a complex cascade of metabolic events leading to alteration of the neurons. At the time of impact, excitatory neurotransmitters are released, reaching high extracellular levels. This neurotransmitter release places the brain into a level of hyperalertness. The injury also causes calcium and sodium influx into neurons and surrounding glial cells. Acute axonal stretch releases potassium and leads to further increase in intracellular calcium levels. This increased intracellular calcium causes mitochondrial dysfunction and exhausts stores of adenosine triphosphate (ATP), leading to an energy metabolism disturbance and leaving the cell more susceptible to further injury (1,2,3). This occurs at a time when the brain needs this energy for healing.

SYMPTOMATOLOGY

Symptoms of concussion can be broken into four main categories; physical, cognitive, emotional, and sleep (see Table 12.1 for further information). Symptoms of concussion may appear immediately or may develop over several hours. It may take up to 72 hours for concussion symptoms to fully develop.

Because symptoms may evolve over time, it is imperative that children and adolescents receive proper evaluation by trained professionals in a timely manner. This includes sideline evaluations by an AT or coach who has sufficient concussion knowledge and removal from sports and other physical activities following a possible concussion.

TABLE 12.1 SYMPTOMS OF CONCUSSION

THINKING/REMEMBERING	PHYSICAL	EMOTIONAL/MOOD	SLEEP
Difficulty thinking clearly	Headache Fuzzy or blurry vision	Irritability	Sleeping more than usual
Feeling slowed down	Nausea or vomiting (early on) Dizziness	Sadness	Sleep less than usual
Difficulty concentrating	Sensitivity to noise or light Balance problems	More emotional	Trouble falling asleep
Difficulty remembering new information	Feeling tired, having no energy	Nervousness or anxiety	

Source: Adapted from CDC website (www.cdc.gov).

Concussion symptoms can differ for each patient, depending on the areas of the brain that are affected by the injury; however, certain symptoms of concussion are more common than others. The most common symptoms noted within the first week of injury are as follows:

- Headache
- Concentration difficulties
- Fatigue, drowsiness
- Dizziness
- Mental fogginess
- Feeling slowed down
- Light sensitivity
- Balance difficulties
- Memory difficulties

EPIDEMIOLOGY OF CONCUSSION

Recent data from the Centers for Disease Control and Prevention (CDC) suggest that there are 1.6 to 3.8 million TBIs per year in the United States. Of these, the majority (75%–90%), can be classified as a mild TBI or concussion. Over 50% of TBIs occur in individuals less than 24 years of age. The highest peak incidence of TBI seen in emergency departments (EDs) across the country occurs in individuals aged 0 to 4 years, followed by those 15 to 19 years of age. Children aged 0 to 14 account for almost 500,000 ED visits for TBI across the country each year.

In the pediatric population, the majority of concussions occur during sports and recreational activities, making up almost 50% of all injuries. While sports-related injuries account for a large number of concussions, they are not the sole etiology. Other common etiologies include falls (25%), striking an object/person (10%), and motor vehicle accidents (10%).

In the past 10 years, the number of high school students who have been diagnosed with a concussion has risen significantly. This is likely due to more stringent monitoring and sideline assessments and greater community knowledge and education. Approximately 25% of high school students report having experienced one concussion and 20% have experienced two or more concussions. While the majority of research, discussion, and media buzz surround sports-related concussion, it is imperative that even nonathletes be properly diagnosed, evaluated, and treated.

SPORTS-RELATED CONCUSSION

The overall incidence of concussion in high school athletes is 24 per 100,000. Football consistently causes the highest number and percentage of concussions at the high school and college levels, ranging as high as nearly 75%. Ice hockey injuries are also quite common. In female athletes, soccer is the most common cause of concussion and is implicated in nearly 50% of cases (4,5). Injuries in lacrosse and field hockey are also fairly common, and the number of concussions diagnosed in cheerleaders is also on the rise.

In gender-comparative sports, such as soccer, females are more susceptible to concussion than their male counterparts. It is suspected that this is due to a number of factors including anatomic, psychosocial, and sociocultural differences. There are higher rotational velocity forces of the head on the neck in females. Due to the smaller neck size and lesser neck muscle mass, the head accelerates more quickly leading to increased forces at the gray–white matter junction. This axonal injury may contribute to the increased recovery time demonstrated in some female athletes compared to male athletes.

In a 2012 study, the overall rate for sports-related concussion was found to be 2.5/10,000 athletic exposures. (Athletic exposure is defined as one practice/game for one player.) The most common mechanism of injury in sport is player-to-player contact, followed by player-to-playing-surface contact. In their study, greater than 40%

of all athletes, except for girls participating in swimming and track events, had symptom recovery by 3 days post-injury. Approximately 80% of the athletes with concussions were able to return to play within 3 weeks of injury.

CONCUSSION EVALUATION

Acute Evaluation

All concussions require evaluation by a medical provider. The initial evaluation may be performed on the field by a coach or AT; however, if there is concern for a possible concussion, further evaluation should be conducted by a physician or another practitioner with concussion training. Whenever a player shows any sign of concussion, onsite evaluation is warranted. If sideline evaluation is not possible, the player should be removed from play and urgent evaluation by a licensed health care provider should be arranged.

There are several sideline assessment tools that are validated for use following concussion. These assessments are performed in an athlete with concern for possible concussion. Although developed as sideline tools, they are frequently used in the physician's office as well. The most common sideline tool is the Sport Concussion Assessment Tool, edition 3 (SCAT-3), used for athletes aged 13 years and older, and the Child-SCAT-3, for those aged 5 to 12 years. This tool asks the athlete to report current symptoms from a checklist, and evaluates cognitive performance and balance. The cognitive assessment includes evaluation of orientation, memory and concentration.

For concussions that do not occur during sports or those that occur without an AT present, initial evaluations are typically conducted by a primary care provider or in the ED. There are several concussion red flags that warrant immediate attention in an ED setting to evaluate for more severe injury. These are listed in the following section, Red Flags.

Evaluation for concussion begins with history regarding the injury and documenting the presence of any red flags, which warrant a higher level of care. A full examination should be performed, paying particular attention to the neurologic examination. A fundoscopic examination is warranted as well. Balance assessment is performed during the initial and subsequent office visits. This includes Romberg, tandem gait, and tandem and single-leg stance, with eyes open and closed. Visual tracking observation is important in the setting of concussion. This includes smooth pursuits, saccades, vestibulo-ocular reflex (VOR), and optokinetic responses. Following injury to the vestibular system, impairments in VOR and report of dizziness particularly with motion are readily observed and noted.

If there are any red flags present, the child or adolescent should be evaluated in the ED setting and may require admission to the hospital to allow for frequent monitoring, neurologic checks, and treatment of symptoms. This may include intravenous (IV) medications for headache abortion or IV fluids and antiemetics for persistent emesis.

Red Flags

Worsening headache
Weakness, numbness, decreased coordination, slurred speech
Repeated vomiting
Appear very drowsy or cannot be awakened
Unequal pupil size
Post-traumatic seizure
Severe confusion
Increasing confusion, restlessness or agitation, unusual behavior
Loss of consciousness

Neuroimaging

Concussions are functional brain injuries and cannot be seen on computed tomography (CT) scan or MRI. Thus, these studies are not routinely recommended for all patients with concussion. Acute CT scan should be considered if there are any abnormalities on neurologic examination or if there is a loss of consciousness (see the list of red flags). The purpose of the CT scan is to evaluate for any underlying intracranial bleeding such as an epidural, subdural, or intraparenchymal hemorrhage.

During the recovery phase of concussion, MRI of the brain may be warranted, especially if the patient is not following the anticipated recovery pattern. MRI may reveal axonal injury or underlying individual factors, such as a Chiari malformation, which may predispose the patient to protracted symptoms.

Several recent studies have evaluated various other neuroimaging techniques, such as diffusion tensor imaging, functional MRI, and positron emission tomography (PET) scan, to determine whether there is any benefit to using these newer techniques earlier in the course of concussion. It is unclear at this time whether any of these techniques will affect treatment interventions or outcomes (6).

SECOND IMPACT SYNDROME

As described previously, the metabolic cascade that occurs following a concussion includes abrupt neuronal depolarization, release of excitatory neurotransmitters, ionic shifts, changes in glucose metabolism, altered cerebral blood flow, and impaired axonal function. These alterations are associated with periods of postconcussion vulnerability and neurobehavioral abnormalities. The concept of second impact syndrome largely rests on the interpretation of anecdotal case reports. Second impact syndrome was initially thought to occur due to

two separate injuries closely spaced together; however, review of autopsy reports of those who died from "second impact syndrome" demonstrates that this is not the case. The majority of cases of second impact syndrome demonstrate an association with intracranial hemorrhage, typically a subdural hemorrhage, in the setting of cerebral swelling and do not coincide with a second impact or injury. There are also reports of delayed onset of cerebral swelling, which is also occasionally referred to as "second impact syndrome." This appears to be more so due to a genetic mutation in calcium-gated channels rather than a second impact to the brain. Frequent monitoring for worsening symptoms in the setting of concussion helps to identify this condition so that appropriate medical treatment can be started timely.

CONCUSSION MANAGEMENT

Physical activity, physiologic stress, and cognitive loads can exacerbate concussion symptoms and prolong recovery. Thus, the key to recovery after a concussion is rest. While most health care providers recognize the importance of physical rest following concussion, the concept of cognitive rest is only recently becoming part of the prescription for concussion treatment. A recent article, published in the *Journal of Pediatrics*, evaluated the effect of various degrees of cognitive rest on the length of concussion recovery (7). This study confirmed that full cognitive rest and relative cognitive rest following injury lead to shorter recovery times. Of note, this study does not mention full bed rest. In fact, other studies (8) refute the idea of full bed rest quoting prior studies, which demonstrate that bed rest in normal individuals can lead to postconcussive-like symptoms. Also after 3 days of bed rest, deconditioning begins and mood and sleep patterns may also begin to deteriorate especially in those individuals with pre-existing conditions. Full rest may lead to avoidance and fear of environments, which may trigger symptoms, which will only make the conditions harder to treat long term.

Physical and cognitive rest allows the brain to use most of its energy to focus on healing. By not requiring brain energy for cognitive and physical tasks, the brain is thought to be set up for quicker recovery, which is demonstrated in some, but not all, recent studies. During the initial phase of recovery, abortive headache medications may be utilized to make the patient more comfortable and allow for easier rest and a quicker return to and tolerance of normal physical and cognitive activities. Frequently, medications such as melatonin may be introduced if there are persistent sleep difficulties.

Although it is important to address all postconcussive symptoms, three main symptom areas should be focused upon to help improve speed of recovery. These include headaches, sleep disturbances, and mood difficulties. Effectively addressing these three areas seems to shorten the duration of the postconcussive period based on anecdotal evidence alone; however, no studies to date support this.

In our concussion clinic, we have found that a short course of a scheduled nonsteroidal anti-inflammatory drug (NSAID) may be beneficial to attain better control of headaches when rest alone does not work. Typically, after at least 1 week of constant daily headache, a trial of Naprosyn twice daily for 5 days will improve the headache intensity and/or frequency of headaches. This improvement of headaches is beneficial with transition back to school. After a 5-day course, the scheduled use of these medications should be stopped to prevent the occurrence of rebound headaches.

The use of neurocognitive testing during concussion recovery is becoming the standard of care. This testing helps demonstrate, along with baseline school performance, when cognitive recovery has occurred. This testing is more sensitive to subtle cognitive difficulties that may persist even after reaching full school demands. This will be discussed in more detail later in this chapter.

In addition to neurocognitive testing, several other tests and monitoring are utilized in the office. These include self-reported symptoms, objective balance assessments such as single-leg and tandem stance or more in-depth testing with standardized balance assessments, and assessment of visual tracking such as the King–Devick test, which looks at saccadic eye movements, smooth pursuits, and the vestibulo-ocular reflex. As each of these tests measures a separate area of concussion recovery, one test alone should not be depended upon to make all decisions regarding return to school and return to sports activities.

RETURN TO SCHOOL

Once acute concussion symptoms are under better control, gradual return to school should begin. Evidence seems to suggests that several days to 1 week of relative cognitive rest results in decreased symptoms and improved cognitive performance, and that early cognitive activity during the postconcussive period increases symptom duration (9). Recent evidence suggests that there is no difference in concussion recovery between full cognitive rest and relative cognitive rest, but that both are superior to no cognitive limitations (10). Return to school should be individualized. Some children and adolescents may be able to resume full days of school at the normal workload; however, others require a gradual return to school or school-based accommodations. If needed, school days may start at half-time attendance with workload accommodations. In addition to reduction in class work and homework, the student should be given extra time to complete assignments. Use of computer and smartboard should be restricted, as often, vestibular symptoms are triggered by eye and head movements. The student may

require frequent rest breaks during the school day to help with tolerance.

Once tolerating half-time attendance, the progression to full days should begin. Typically accommodations will remain in place until the student is tolerating the day with accommodations. Accommodations may remain in place longer if the student continues to demonstrate active cognitive recovery on neurocognitive testing; however, this is determined on an individual basis.

RETURN TO SPORT

Return to play (RTP) after a concussion follows the guidelines established originally in Vienna in 2001, at the International Conference in Concussion in Sport (ICCS). This conference has taken place an additional three times to provide updates based on current evidence. These subsequent conferences focused on improving the evaluation, management, and RTP of concussed athletes. The most recent ICCS took place in Zurich in 2012 and, thus, the current RTP guidelines are sometimes referred to as the Zurich guidelines. In 2010, the American Academy of Pediatrics (AAP) published basic concussion management guidelines for children and adolescents, adapted from the ICCS recommendations that emphasized a graduated RTP protocol and the importance of having an athlete follow a stepwise progression in their return to sport (11).

A child or adolescent suspected of having a concussion should not be allowed to return to play on the day of injury. Prior to return, the child or adolescent should be completely symptom-free (or with baseline preinjury symptoms) with a normal examination and have successfully completed full return to school. When conducted, neurocognitive testing should also demonstrate that full cognitive recovery has occurred.

CONCUSSION RECOVERY

Evidence suggests that children and adolescents, who are still actively undergoing brain development, take longer to recover than adults. Several factors are postulated to contribute to this longer recovery pattern as demonstrated in recent studies. These include a larger head-to-body ratio, reduced neck and shoulder musculature, which allows greater forces to be transmitted to the head, a larger subarachnoid space within which the brain can shift, reduced myelination of the central nervous system (CNS) subjecting children to greater shearing forces, and differences in cerebral blood volume. Despite this prolonged recovery pattern compared to adults, the majority of children and adolescents will recover quickly and completely. Approximately 80% of patients with the first concussion will recover within the first 2 weeks following their insult, usually 7 to 10 days. An additional 10% to 15% of patients will recover within the next 2 weeks. There is a notable difference in recovery patterns for sports-related versus non-sports-related concussions. About 10% of athletes with sports-related concussions will have symptoms that persist beyond 2 weeks. In all concussions, persistent symptoms beyond 3 months warrant evaluation for other conditions, which may be contributing to a protracted recovery or which mimic postconcussive symptoms.

There are a number of risk factors that may increase the likelihood a child/adolescent will have symptoms lasting longer than this anticipated time frame. The strongest risk factors in meta-analyses have been prior concussion and female sex. Evidence suggests that underlying mood disturbance may be a strong predictive risk factor of prolonged concussion recovery. Underlying mood disturbance may be exacerbated as a direct result of a concussion or changes may occur as a result of alterations in daily activity. Therefore, mood must be addressed and monitored early since it may lead to prolonged postconcussive symptoms. Other risk factors include history of frequent or migraine headaches, learning disabilities including attention deficit hyperactivity disorder (ADHD), history of speech therapy, premorbid sleep difficulties, and a family history of migraine headaches or Alzheimer dementia.

Initial symptoms experienced within the first few days may correlate with length of recovery. Dizziness following concussion is a marker of poorer prognosis and prolonged recovery, especially if present on the field or at the time of injury. Dizziness at 2 weeks postinjury is the single predictor of persistent postconcussive syndrome 6 months after mild TBI (12); therefore, careful assessment and treatment of dizziness should begin early. Other contributors to early persistent dizziness include migraine headaches, cervical spine dysfunction, visual-perceptual dysfunction, and autonomic dysfunction, all of which can be addressed with therapies.

Other factors that predict slower recovery are the presence of retrograde amnesia, post-traumatic amnesia, higher initial symptom score, and mental status change lasting longer than 5 minutes. Very poor scores on neurocognitive testing, such as Immediate Post-Concussion Assessment and Cognitive Testing (ImPACT) testing, in at least three of four areas within the first 72 hours of injury are also predictive of prolonged symptoms. The presence of premorbid anxiety or depression or post-traumatic stress can increase the likelihood that recovery will be slow.

THERAPIES FOR CONCUSSION

In most uncomplicated concussions, rest alone is enough to achieve full recovery within a short period of time. When children and adolescents do not recover after a period of rest, the initiation of therapy services can help expedite recovery. Experimental animal data demonstrate

that voluntary exercise within the first week after concussion impairs recovery while aerobic exercise performed 3 to 4 weeks after concussion improves recovery. This improvement is hypothesized to be due to upregulation of brain-derived neurotrophic factor and restoration of normal cerebral homeostasis, which helps with neuronal recovery. Prolonged rest, especially in athletes, can also lead to secondary symptoms such as fatigue, depression, and sleep disturbances, which slow recovery. Thus, the onset of therapies should begin by 14 to 21 days postinjury. There is evidence that starting therapies sooner may be warranted in some individuals; however, this should be submaximal as there is evidence based on animal studies that early vigorous exercise may slow down recovery (8). The referral to particular therapies is dependent on the signs and symptoms that the child or adolescent is still experiencing.

Most commonly the referral to physical therapy is made for a submaximal aerobic program with a gradual progressive increase in intensity as tolerated by the patient. In addition to helping with recovery, it also helps to build the child's or adolescent's confidence and hopefully prevent the development of depressive or psychosomatic symptoms, which further slow recovery. In addition to an aerobic program, physical therapy should also address any vestibular deficits. This includes balance work and visual training exercises to improve these deficits. The goal of these exercises is to restore postural stability and dynamic gaze stability. Vestibular rehabilitation is most effective in patients whose headaches are adequately controlled.

If postconcussive symptoms are persistent beyond 3 weeks, however are not exacerbated by any degree of physical activity, then an alternate diagnosis should be explored. The most common alternate diagnoses that may mimic postconcussive syndrome include post-traumatic stress disorder, depression, migraine headache, or cervicogenic headaches/vestibular dysfunction (13). Physical therapy may also be warranted in these cases; however, the therapy prescription may be slightly different. A gross screen for cervicogenic dizziness is to demonstrate relief of this symptom with gentle cervical traction. Neck pain must also be present if this is the underlying cause of the dizziness. If cervicogenic dizziness is present, physical therapy should include cervical range of motion, manual therapy for segmental hypomobility, manual cervical traction, and deep cervical flexor isometric stabilization exercises.

In the setting of significant visual deficits, referral to occupational therapy should be made to further concentrate this effort on recovery. Occupational therapy also provides some cognitive rehabilitation to work on deficits as they impact school and daily activities. For more significant cognitive deficits, referral to speech and language pathology should be incorporated into the therapy program. Prior to referral to all therapies, the patient should have evaluation and screening for any noninjury factors such as mood changes or school avoidance, which may be leading to continued reporting of symptoms. Also, if not already started, referral for neuropsychological evaluation is recommended for all patients with persistent symptoms.

NEUROCOGNITIVE TESTING

Cognitive assessment is generally regarded as essential in the diagnosis and management of concussion, as delineated by the ICCS held in Zurich in 2008. The increased vulnerability of the adolescent athlete relative to adults is well recognized as to duration of symptoms and differential recovery pattern. Furthermore, the effect of repeat concussion, treatment options, school demands, restriction of exposure to risk (continued sports participation)—both during recovery and subsequently—and the potential effect on a developing brain (14) are all factors that argue for the role of neuropsychological assessment in the care of such patients. The nature of this type of testing will now be explored.

There are two primary approaches to assessment in concussion. These include traditional paper and pencil testing and computerized assessment. Traditional neuropsychological testing via pencil and paper typically carries acceptable reliability and validity and have demonstrated sensitivity in some areas to concussion effects (4). These types of tests, however, lack the sensitivity of computer-based assessment and do not lend themselves as easily to the type of repeated assessment as required in the monitoring of concussion (15). Further, they often lack alternate forms and therefore are subject to practice effects. Further, traditional tests often lack the ability to detect subtle changes in reaction time and speed of mental processing.

Thus, the relatively recent computerized testing assesses abilities typically disrupted by concussion. There are several advantages of the use of computerized testing. One is that the tests have alternate forms, which allows for serial testing at different points in time. The second is the relatively short testing time duration, which is typically under 30 minutes. Third, and perhaps most useful, is the ability to baseline individuals' preinjury. This is particularly relevant to child athletes, who are at substantially higher risk for concussion.

The abilities that these tests tap include speed of processing and reaction time, and are done with varying stimuli. An inherent limitation is the lack of auditory presentation in these instruments, where all stimuli are visual in presentation, even though language stimuli are used in conjunction with nonverbal stimuli (spatial location, line drawings) in one test listed. The repeatability and ease of administration is an advantage of these tests and so they can be used for the serial monitoring recommended for complex concussion recovery. Scores on these devices

serve as guidelines of functional capacity that determine return to activities, whether that is around cognitive demand (school) or physical demand (gym class, sports, bike riding, etc.). Balance assessment can also be used as a specific monitor representing a high-level dynamic function of the brain's motor control and ability required for competent physical activity participation. There is ongoing debate about the sensitivity of cognitive versus balance deficits as the most sensitive indicator of concussion sensitivity.

There are several widely used computerized batteries that have norms within the pediatric population. The HeadMinder Concussion Resolution Index (CRI) (16) has norms for ages 18 to 22 and "under 18." The latter refers to a normative sample down to age 13, with analysis yielding no difference in the scoring of adolescents from ages 13 to 18 (16). The CRI is an Internet-based platform with six subtests, taking 25 minutes to administer. It yields three scores: processing speed index, simple reaction time index, and complex reaction time index. Verbal (written) stimuli were specifically avoided, with all stimuli in a visual icon format to minimize error due to language disability or English-as-second-language issues.

ImPACT (1972) is available in Windows and Macintosh applications as well as through an online version. An on-field palm-based version is also available and includes a brief on-field mental status evaluation. It does use verbal stimuli, and there is reading involved in testing instructions, with a sixth grade required reading level (17). It has eight subtests in its current version and registers demographic/history data, current concussion details (including information about anterograde and retrograde amnesia), as well as somatic and cognitive symptoms. There are four scores from ImPACT: verbal memory, visual memory, reaction time, and visuomotor speed. ImPACT has norms for ages 10 and above. Adolescent norms on this battery are extensive, and there is extensive literature on its use. Though developed primarily for sports concussion management, it has recently been used to characterize concussions presenting to an emergency room (5). There also exists a Pediatric ImPACT, which is an adaptation of the ImPACT battery for children. This is normed for children aged 5 to 12 years old. It consists of seven subtests that mirror the traditional ImPACT subtests, but present the stimulus material such that it is more "kid friendly" and more appealing to children. Psychometric analyses have yielded strong developmental age effects, test–retest reliability, and reasonable internal consistency (18), according to the authors of the test.

The use of computerized assessment in concussion diagnosis and management is not without its weaknesses. First and foremost, the majority of the literature that publishes effectiveness of these tests often comes from the institutions or authors who developed the instrument. Therefore, it is important to look for authors who do not have connection with the instruction or company that developed the test to attempt to reduce bias. Examination of the literature in these specific areas can be difficult due to lack of peer-reviewed studies in this area; however, there are some studies that show the potential downfalls of using these tests.

An important consideration in the use of computerized testing is test–retest reliability. Test–retest reliability is estimated by performing the same test to the same subjects, under the same conditions, over different time intervals. One study examining computerized testing found that test–retest reliability was reported to be 0.82 for processing speed, 0.70 for simple reaction time, and 0.68 for complex reaction time during a 2-week test–retest period for the HeadMinder Concussion Resolution Index (19). Another study conducted by Register-Mihalik and colleagues (20) administered the ImPACT test along with several other traditional paper and pencil tests to college-age and high-school-age subjects. The subjects were assessed at three separate time intervals with approximately 1 day (24 hours) in between sessions. The weakest value was found for verbal memory, while the highest (0.71) was found for processing speed (20). Both of these studies demonstrate that while test–retest reliability is moderate, there is concern for this particular psychometric property among computerized testing when it is repeated to the same group of subjects. Both of these studies, however, tested subjects in very limited time intervals (24 hours to 2 weeks). The literature is sparse for time intervals longer than this; thus this continues to question the reliability of both ImPACT and HeadMinder. This also questions the effects and utility of serial testing in concussion assessment and management, as practice effects in these close time intervals raise questions.

Validity is also a major concern when it comes to neuropsychological testing, especially computer-based. Validity refers to whether a test is measuring what it is intended to measure. Most research to date has focused on the sensitivity of ImPACT results to a suspected postconcussive injury. The majority of the studies have administered ImPACT to concussed athletes and have compared the scores to baseline scores, control group, or age and gender norms. Symptomatic athletes indeed often produce postinjury scores that are lower than their baseline, yielding construct validity, as the test is sensitive to "history of recent concussion." Most studies have also found, however, that measuring recovery is a complex matter that is often complicated by practice effects as well as other psychometric confounds that may result from serial testing (15), as mentioned previously.

It is important to note in this section that a major gap in this literature is due to the majority of these studies being focused on groups of college or adult athletes, with little attention paid to middle-school-age athletes or children and adolescents who sustain concussions from non-sports-related injuries. This makes interpretation less useful and interpretation of these tests questionable for younger children. This is an area for more development,

especially for those practitioners who work with children and adolescents under the age of 18 who sustain concussion from mechanisms that are different from sports, such as motor vehicle accidents and falls, as the majority of the literature focuses specifically on sports-related concussions.

There are other criticisms to the use of this testing outside of psychometric properties. First pre-existing cognitive issues, often deemed "exceptionalities," such as a diagnosis of ADHD, a learning disability, psychiatric diagnoses, and/or a variety of adjustment issues can skew testing results. Baseline tests can also present with difficulties with interpretation due to situations where children are put in a group to test and are distracted by friends or not taking the test "seriously." In other situations, children or adolescents may attempt to sandbag, or "trick the test" into yielding lowered scores, so that recovery may be judged sooner and that they may return to play (21). Given the variety of factors that may impair the interpretation of testing results, it is very important to include an experienced neuropsychologist as part of the multidisciplinary team when it comes to concussion management. Further, the need may arise for the neuropsychologist to conduct a more in-depth evaluation when assessing chronic symptoms, rather than strictly relying on screening tools (15).

Overall, there is a depth of literature that discusses both the benefits and downfalls of computerized neurocognitive testing as well as traditional neuropsychological testing in the assessment and management of concussion. It is important to note that, despite more advanced methods of testing cognition, no RTP decisions should ever be made on the use of neurocognitive testing alone. These judgments should be reached by the collaboration of a multidisciplinary team, which provides a more comprehensive picture of the patient's functioning after an injury. There is not yet a gold standard for RTP decisions, and therefore all points of data, including physical examination, neurocognitive testing, self-reported symptoms, and assessment of environmental functioning should be considered in order to make appropriate decisions.

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PEDIATRIC LIMB DEFICIENCIES

Ann Modrcin, Matthew McLaughlin, and Matthew Luetke

INTRODUCTION

One of the more rewarding experiences in medicine comes from engaging children with limb deficiency in a clinical context. Patients and families experience varied outcomes and challenges, which allow providers the unique opportunity to positively intervene at various points in the patients' lives with technology, innovation, and reassurance. Despite the challenges resulting from limb deficiency, the majority of these patients are socially well-adjusted, independent-minded, and able to interact well with providers, peers, and society.

Treatment strategies in the pediatric limb deficiency population differ significantly from those of adults with amputations. In pediatrics, health care providers must consider the lifelong implications of choices made during childhood and examine the expected longitudinal timeline to achieve the best possible outcomes. Surgical considerations vary even among the pediatric age groups and may require expected revisions or in-depth, multidisciplinary discussion of surgical options. Rapid growth results in more frequent prosthetic adjustment and replacement to keep up with growth and developmental changes of a child. Prosthetic intervention should always augment rather than hinder a child's function, whether for daily activities or for specific tasks, and sometimes the best device is none at all.

CONGENITAL DEFICIENCIES

INCIDENCE

Epidemiologically, limb deficiencies occur in 5 to 9.7 births for every 10,000 live births, with a ratio of 3:1 upper to lower extremity anomalies (1). The United States does not have a formal complete registry of birth defects, so the precise number is unknown. The National Birth Defects Prevention Study has reported that 6% of all types of birth defects are limb deficiencies and the majority of limb deficiencies involve the upper extremity (2,3).

Early identification of limb anomalies occurs with routine ultrasound. A detailed level 3 ultrasound, as well as echo three-dimensional (3D), amniocentesis, and cordocentesis to anticipate syndromes, is recommended if limb deficiencies are detected (4). Parents may experience a variety of emotions after learning of the diagnosis of limb deficiency. Prenatal counseling provides resources and psychological support for parents, and educates families about the process of expected functional level and potential prosthetic utilization.

ETIOLOGY

Although the presence of a limb deficiency evokes salient questions about etiology, frequently there is no identifiable cause of the limb deficiency (5). Embryologically, the first trimester is the most crucial trimester for the genesis of limb production. Congenital limb deficiency occurs as a result of failure of formation of part or all of the limb bud. The mesodermal formation of the limb occurs at 26 days' gestation and continues with differentiation until 8 weeks' gestation. The various limb segments develop in a proximal-to-distal order so that the arm and forearm appear before the hand, and the thigh and leg before the foot (6). Limb development is a complex process that involves orchestration of a number of genes, some of which are well known and studied. Known abnormalities account for various identified syndromes (7). A relatively small set of genes and gene families appear to control the early stages of limb development. More than 80% of heritable limb deficiencies are associated with anomalies outside the musculoskeletal system (7). When other anomalies are suspected, it is crucial to take an expanded history regarding feeding difficulties, respiratory distress, bowel dysfunction, and cardiac abnormalities (8).

Compared to lower extremity limb deficiencies, upper limb deficiencies are more commonly associated with other anomalies, particularly craniofacial, cardiac, and hematologic disorders. This is due to the chronology of development in the first trimester (9). Bilateral deficiencies are more common with craniofacial abnormalities,

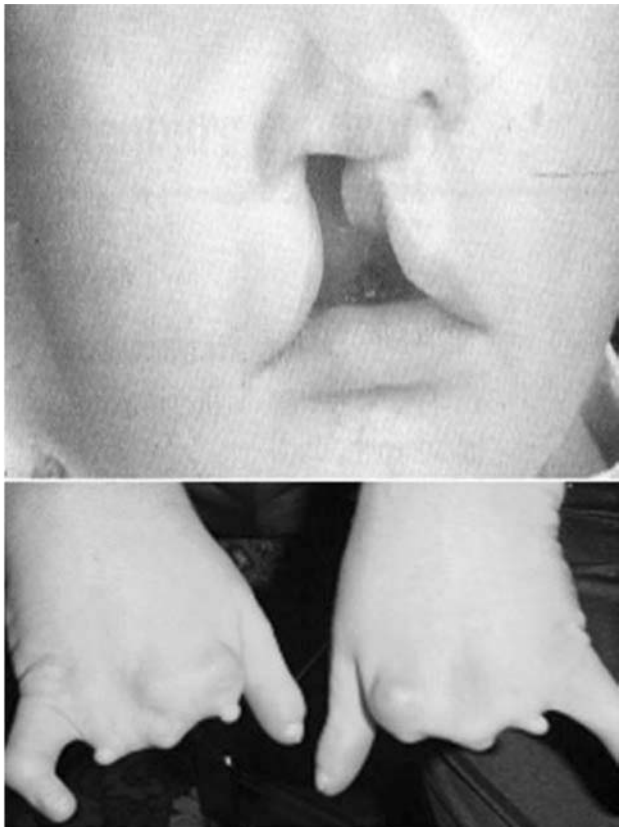

FIGURE 13.1 Two key components of clinical presentation in Moebius syndrome: craniofacial (A) and upper limb (B) deformities.

whereas left–right asymmetry of organogenesis is more commonly associated with unilateral and left axial deficiency (10,11). Vascular pathology is not inherited, so the risk of recurrence is small (5,7). Despite this low risk of recurrence, the frequency of vascular issues contributing to limb deficiency is 34% (5). Conditions with implied vascular disruption include Adams–Oliver syndrome, gastroschisis, Klippel–Feil syndrome, Moebius syndrome, Poland syndrome/sequence, and terminal transverse limb deficiency (12–14). Two key features of Moebius are demonstrated in Figure 13.1: craniofacial anomalies and upper-limb deformity.

Other factors increase the risk for limb deficiency, including maternal diabetes, and gestational diabetes (15,16). Although alcohol, heroin, and cocaine have not been found to be related to limb deficiency, all maternal ingestions and first-trimester abnormalities should be documented during the initial visit (17,18). Smoking increases the risk of digit anomalies (19). Thalidomide historically presents a clear association with limb reductions (20). Case reports implicate valproic acid and calcium channel blockers (21,22). Maternal occupation may play a role, with exposure to chemicals, as in the agricultural setting (23). Uterine abnormalities have been reported in several cases of limb deficiencies, theoretically due to compression of the fetus (6).

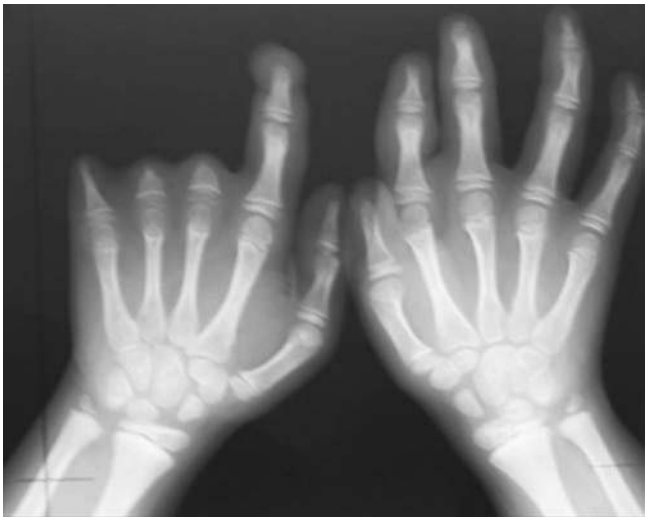

FIGURE 13.2 X-ray of hands affected by amniotic band syndrome.

In addition, disturbances to the uterine environment, such as chorionic villus sampling, are implicated in deficiencies (24). Amniotic band syndrome is associated with fibrous bands that may constrict the limbs (25,26). Radiological findings of amniotic band are illustrated in Figure 13.2. Prenatal vitamins reduce the risk of limb deficiencies (27). Postnatal problems, such as gangrene from vascular emboli and neonatal injury from vascular compromise secondary to umbilical catheters, may necessitate immediate amputation (28–30). Although the perinatal causes for amputation may be different from congenital disorders, the clinical issues for the child and the rehabilitation team are more similar to congenital disorders than acquired disorders of later childhood.

CLASSIFICATION

The International Society for Prosthetics and Orthotics (ISPO) has adopted a definitive system for congenital deficiencies. This system describes limb deficiency on anatomic and radiological basis only (31). The utilization of this system aims to decrease ambiguity by more precisely classifying the type and location of the limb deficiency. First, the limb is classified as having either a transverse or longitudinal deficiency. No longer is it necessary to learn ancient language roots to describe limb deficiency (32–34). However, like an old language and culture, the terminology once used in clinics is difficult to change. Clinical teams often use a fusion of terms.

Many clinics still describe deficiencies by the Frantz classification system (Table 13.1). In this system, deficiencies are either terminal, representing the complete loss of the distal extremity, or intercalary, denoting the absence of intermediate parts with preserved proximal and distal parts of the limb. Those deficits are then divided into horizontal and longitudinal deficits.

TABLE 13.1 CLASSIFICATION OF LIMB DEFICIENCY

ISPO/ISO	FRANTZ–O'RAHILLY	CLASSIC
Longitudinal radius deficiency	Intercalary radial deficiency	Radial hemimelia/club hand
Terminal transverse humerus deficiency	Terminal horizontal humerus deficiency	Above-elbow amputation

ACQUIRED AMPUTATIONS

TERMINOLOGY

The terminology utilized for acquired amputations follows the convention for adult limb loss. Upper extremity amputations include intrascapulothoracic, shoulder disarticulation, transhumeral (above-elbow amputation), elbow disarticulation, transradial (below-elbow amputation), wrist disarticulation, and partial hand amputations. The types of lower extremity amputations are translumbar (hemicorpectomy), transpelvic (hemipelvectomy), hip disarticulation, transfemoral (above-knee amputation), knee disarticulation (through-knee), transtibial (below-knee amputation), ankle disarticulations (ie, Syme, Boyd, and Pirigoff), and partial foot (ie, Chopart and Lisfranc) (35). Figure 13.3 illustrates present classifications of acquired amputations.

TRAUMATIC INJURIES

In the pediatric age group, the most common causes of acquired amputations are trauma and disease (36). Trauma causes limb loss twice as often as disease (37). The most common traumatic injuries result from automobile and motorcycle collisions and train accidents. Causes for traumatic injuries vary with region. In rural areas, farm accidents, lawn mower accidents, and high-tension wire injuries occur more frequently (38–41). For the older child, vehicular accidents, burns, gunshot wounds, and power tools are the most frequent causes of limb loss. Boating accidents can produce amputations by propeller injury. Sadly, in the 1- to 4-year-old age range, power tools such as lawn mowers and household accidents are frequent mechanisms of amputation (42,43). Fortunately, in those traumatic events where digit or even hand replantation is possible, pediatric patients experienced improved functional and cosmetic outcomes with less frequent complications than their adult counterparts. However, patients identified as African American or Hispanic and those without insurance were less frequently noted in another study to have attempted replantation (44,45). For those injuries that require inpatient hospitalization, a multidisciplinary approach produces the best outcome at the time of discharge. Older adolescents and patients with traumatic leg amputations have longer stay during initial hospitalization and higher hospitalization rates (46).

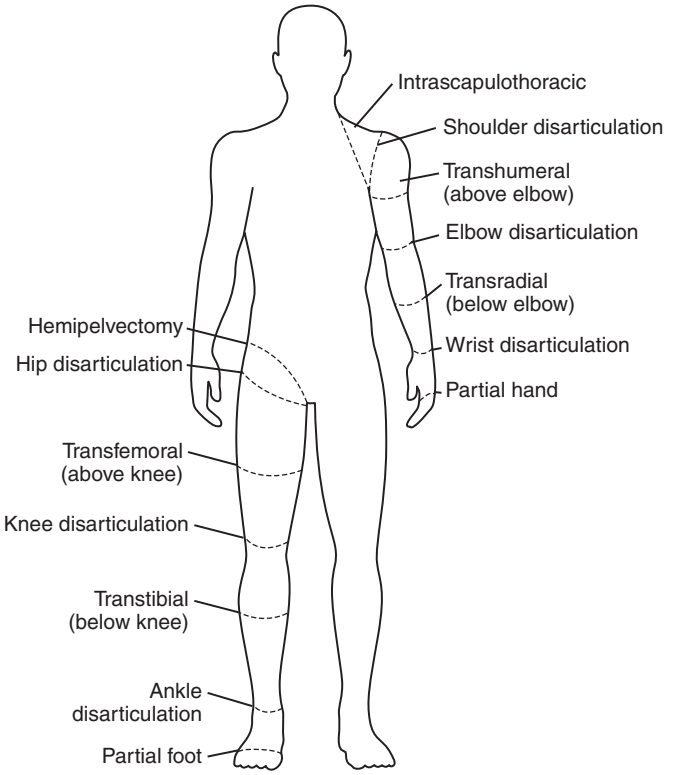

FIGURE 13.3 Classifications of acquired amputations.

A single limb is involved in more than 90% of acquired amputations, of which 60% involves the leg (see Table 13.2). The male-to-female ratio of acquired amputation is 3:1.

TABLE 13.2 LONGITUDINAL LIMB DEFICIENCIES

UPPER LIMB	"DESCRIBED AS"	LOWER LIMB	"DESCRIBED AS"
Humerus	Complete	Femur	Complete
	Partial		Partial
Radius	Complete	Leg	Complete
	Partial		Partial
Carpals	Complete	Tarsals	Complete
	Partial		Partial
Metacarpals	Complete	Metatarsals	Complete
	(1–5)		(1–5)
	Partial		Partial
Phalanges	Complete	Phalanges	Complete
	(1–5)		(1–5)
	Partial		Partial

TUMORS

Tumors are the most frequent cause of amputations due to disease. Tumors represent the most common cause of amputations in the European Surveillance of Congenital Anomalies (EUROCAT) data system (1). The highest incidence of malignancy is in the 12- to 21-year-old age group. Osteogenic sarcoma, Ewing's sarcoma, and the rare rhabdomyosarcoma are responsible for the majority of tumors resulting in amputation (47,48). Unprecedented improvement in survival has occurred with earlier detection and combined therapy (49). Definitive surgery for osteosarcoma depends upon the site of the primary tumor and the extent of invasion or metastasis (50). Surgical removal of the affected bone and the surrounding soft tissue remains the treatment of choice, whether by amputation or limb-salvage procedure. Limb salvage with an endoprosthesis can be offered to 90% of children with osteosarcoma (49–51). This procedure, which involves replacing the affected bone with a metal endoprosthesis, is accompanied by orders to prohibit contact sports. With the advent of extendable endoprostheses, it has been suggested that children who have undergone this treatment have results that are superior to those who have undergone amputation surgery (52–54). Families may wish to pursue this due to the improved cosmesis, to prevent the loss of a limb in a growing child, and ultimately, to achieve the best functional outcome.

The surgical procedure of choice is that which obtains a tumor-free margin of 5 to 8 cm above the proximal limit of the medullary tumor. The decision to proceed with limb salvage or amputation is dependent on the aggressiveness of the tumor, the stage, the responsiveness to neoadjuvant therapy, and the likelihood of obtaining tumor-free margins (55–57). The knee poses a challenge for soft tissue sarcomas. Despite complications, the knee may be reconstructed with allografts (58,59). Rehabilitation physicians discuss the likely functional outcomes of each choice and provide continued support and maintenance of functional status during the chosen course of treatment. During the limb salvage, amputation, and recovery process, rehabilitation physicians and therapists promote strategies to prevent any decline in function, treat pain, and anticipate progression of potential treatments.

Chemotherapy has now proven to be an effective adjunct to surgery. Prior to 1972, only 15% of the children were disease-free and survived with surgery, compared to the 60% to 70% who now survive with surgery and the addition of chemotherapy (60,61). Rehabilitation may be confounded by factors of fluctuation in limb-volume status, fatigue, and the psychological aspects of combined treatments. Physical therapy emphasizing range of motion (ROM), strengthening, and functional activities is important for children with lower extremity sarcoma after limb-salvage surgery (62). Outcomes were similar for ambulation, stair climbing, employment, and psychological adjustment when comparing amputation to limb salvage as surgical management of sarcomas. Patients benefit functionally from gait training with prosthetic devices following amputation (63,64). Additionally, osteosarcoma patients who underwent amputation compared to limb salvage experienced similar socioeconomic outcomes, such as education, employment, and marital status at a 20 year follow-up; however, the most important factor for determining the quality of life in these patients was the functionality of the limb regardless of amputation or limb-salvage procedure (65,66). Conflicting studies argue that limb-salvage procedures appear to improve quality of life over amputation; however, this study did not directly evaluate functional status (67).

INFECTIONS

Infectious emboli from meningococcemia may autoamputate limbs or digits (68). The process frequently involves all four limbs. Growth plates may be affected, resulting in angular deformity and the need for surgical epiphysiodesis (69). Frequently, the skin is affected as well as the limb (70). Multiple surgical skin grafts limit the prosthetic fitting; a coordinated burn team is often best prepared to handle initial management (71). Over the past few decades, the incidence of invasive meningococcal disease in the United States has remained relatively stable (72–74). Pneumococcal septicemia also can produce purpura fulminans, characterized by acute onset of rapidly progressive hemorrhagic necrosis of the skin and thrombosis (29). An example of the multiple distal amputations and angular deformities caused by emboli from infections is seen in Figure 13.4.

SURGICAL APPROACH: GENERAL PRINCIPLES

Adherence to the general principles of childhood amputation surgery promotes optimal function. The principles are: (a) preserve length, (b) preserve growth plates, (c) perform disarticulation rather than transosseous amputation, (d) preserve the knee joint whenever possible, and (e) stabilize and normalize proximal portions of the limb (75).

The cardinal surgical dictum to conserve the whole limb length if possible is true for children as well as adults. In growing children who require amputation, disarticulation rather than a transdiaphyseal amputation may be preferred (76). Disarticulation preserves the epiphyseal growth plates and ensures longitudinal growth (77). Disarticulation also avoids the development of terminal or appositional overgrowth of new bone.

Frequently, children with limb differences experience problems with the length of their affected limbs. Because of known growth patterns, limb-length differences tend to vary with patient age and involved bone,

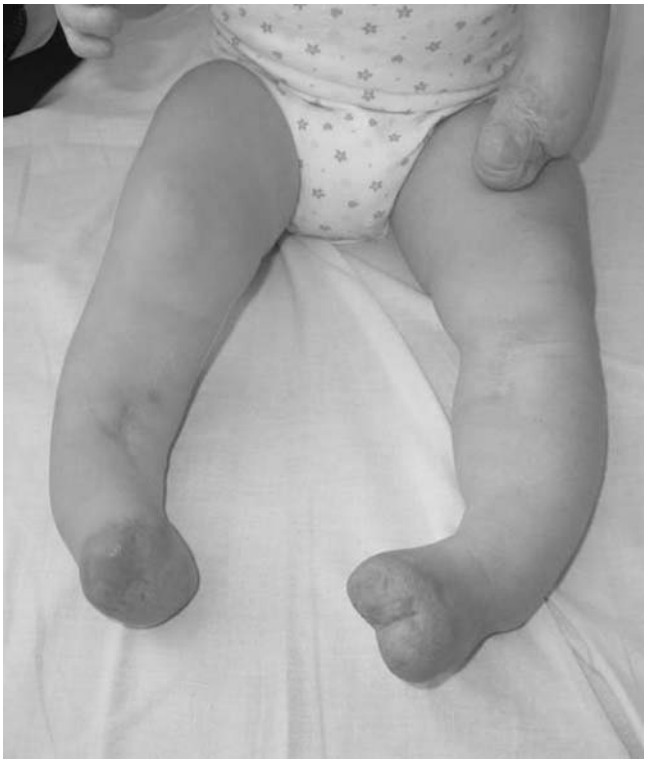

FIGURE 13.4 Angulated amputations as a result of meningococcemia and subsequent purpura fulminans.

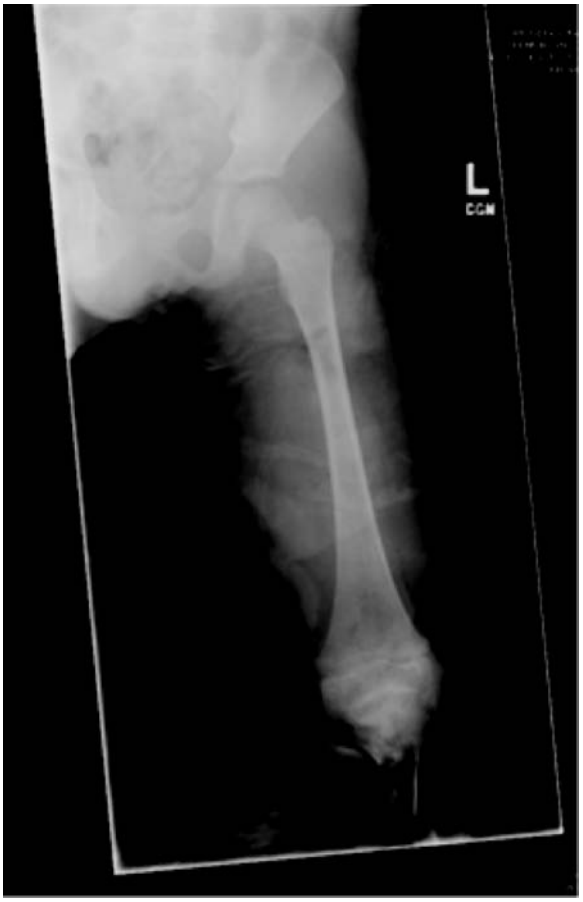

FIGURE 13.5 Surgical differences between pediatrics and adults enable this very short salvaged tibial remnant to be successfully managed with below-knee prosthetic intervention.

and growth projections over time are crucial for surgical decision making. For example, 70% of femoral growth occurs at the distal physis (78), while the contribution of the proximal tibial growth plate is about 57% of the total length. So saving a short tibial remnant in a 4-year old reaps greater biomechanical benefit than for a 17-year old, despite the risk of bony overgrowth. In the younger child, saving the knee is more meaningful, with potential for better prosthetic function (79). Figure 13.5 demonstrates a very short salvaged tibial remnant successfully managed with below-knee prosthetic intervention. Fortunately in the pediatric population, tissue viability is enhanced, so strategies such as split thickness skin grafting, skin traction with delayed closure, and wounds closed over more tension heal better compared to the same techniques in adults, providing viable wound coverage and incision closure. Figure 13.6 demonstrates extensive skin and tissue loss, with successful subsequent healing and prosthetic tolerance.

Terminal overgrowth, often referred to as *spiking*, at the transected end of a long bone is the most common complication following amputation in the immature child (80,81). Diaphyseal overgrowth may also occur in children with acquired intrauterine anomalies, such as amniotic band syndrome, in which the epiphysis is no longer present. It occurs most frequently in the humerus, fibula, tibia, and femur. During appositional growth, the distal bone begins to form in the shape of an icicle. Figure 13.7

FIGURE 13.6 The photos for Figures 13.5 and 13.6 show the surgical difference between pediatric and adult acquired limb deficiency. Skin grafting and the ability for healing allows surgeons to be more creative with wound coverage. Despite extensive scarring, prosthetic fitting can still occur.

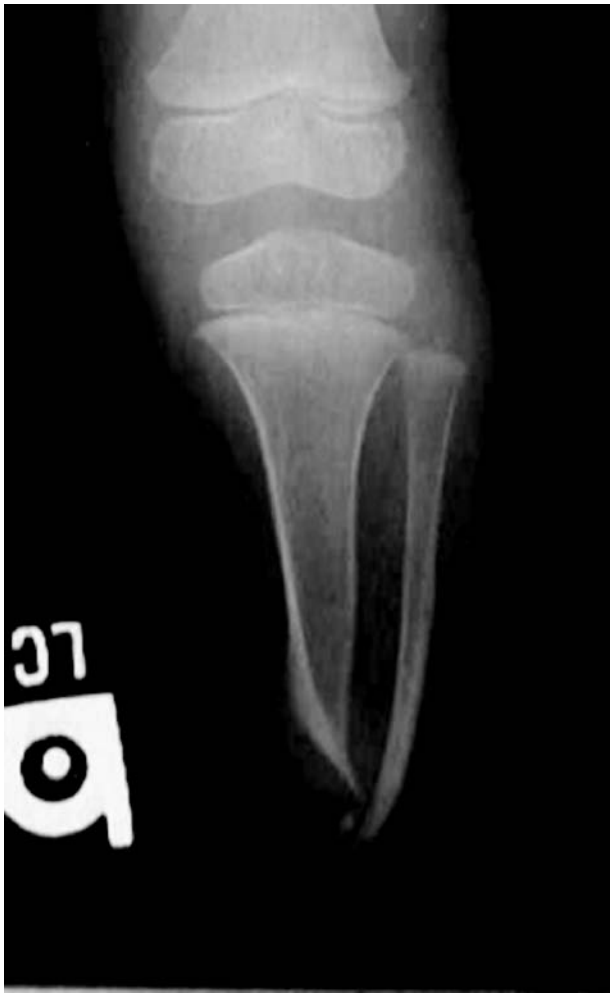

FIGURE 13.7 Terminal overgrowth represents a common problem in the limb deficient child. The fibula continued to grow in length in this radiograph representing problems with skin integrity and prosthetic fitting. (Photo courtesy of the personal collection of Dr. Vincent Mosca, MD.)

demonstrates a typical radiographic finding of terminal overgrowth. As the pointed segment creates insult to the adjacent soft tissue, a bursa may form to protect the distal residuum. During this time, the child may experience significant pain with reduced tolerance of prosthetic use. Frequent socket modifications are necessary to accommodate these anatomic changes. Treatments such as aspiration, steroid injections, and stump wrapping are usually ineffective. Unfortunately, the rate of growth may be so vigorous that the bone pierces the skin; at this stage, the treatment of choice is surgical revision. Distal resection and stump capping with the use of autografts or plastic polymers are surgical options (82). Once surgery becomes necessary, the problem is likely to recur until skeletal maturity. Each time that bone is resected, the overall length of the bone is reduced, thereby reducing its mechanical advantage for control of the prosthesis. Bone spurs may form at the periphery of the transected bone,

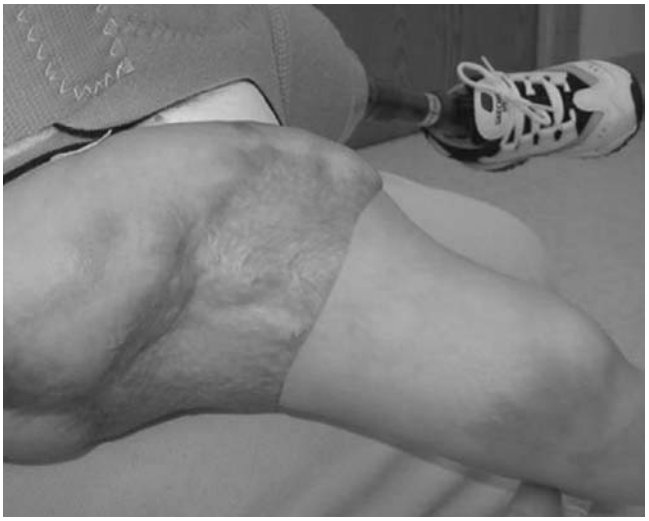

FIGURE 13.8 Residual limb with reconstructed skin grafts and custom liner.

and resection may be necessary. The resulting stump scarring, which interferes with weight-bearing, requires prosthetic modifications and exploration of custom interfaces. Plastic surgeons are involved with reconstruction of skin flaps or with complicated repairs of residual limbs (51,83). In Figure 13.8 an example of complicated residual scarring is shown.

PHANTOM SENSATION

Phantom sensation is an individual's sensation of feeling a missing limb like it were actually present. These sensations are rarely reported as painful or unpleasant in the pediatric population. Since phantom sensation is not painful, no treatment is necessary; however, if phantom limb pain becomes an issue in older children and adolescents, it can interfere with the rehabilitation process. Phantom limb pain rarely occurs in children under 10 or during growth, but is reported in teenagers. In addition, children with congenital limb deficiencies are less likely to experience phantom sensations than those with acquired amputations, but it is important to recognize that it can occur (84,85). Phantom sensations in children with limb deficiency are explainable if we recognize the brain as a generator of sensory information (86).

UPPER LIMB

There are differences in the approach, acceptance, and management of the upper limb amputee versus the lower extremity amputee. Despite significant and ongoing improvements, upper limb prosthetic devices do not yet replace the sensory function of the hand, and are best

considered as a mechanical tool (87). The hand is used to explore the environment and to manipulate objects within it. The hand needs to reach the body and precisely approach an object, grasp, and then release it. Acceptance of the prosthesis is variable (88). Frequently, the exposed skin of a residual limb is preferable to an encased limb. Stump sensation may even be enhanced to compensate for the loss of prehensile area (89). In ipsilateral congenital limb deficiencies, issues with organogenesis can occur and may require further evaluation; however, bilateral limb deficiencies may be associated with craniofacial abnormalities (8). Scoliosis occurs frequently in this population, but rarely requires surgical correction (90).

COMMON UPPER LIMB DEFICIENCIES

Digital Deficiencies

Digital deficiencies are common but rarely present in isolation. Removal of additional digits and intervention with Z-plasty procedures produce acceptable results for children with polydactyly and syndactyly, respectively. Amniotic band syndrome or Streeter's dysplasia commonly presents with digital constriction banding, though the etiology of this condition remains controversial. In addition, other anomalies may be present, ranging from evidence of other banding, to craniofacial clefting (in 78%) or abdominal to one or more lower limb amputations (in 70%), and abdominal wall, spine, or thorax (in 52%) that have occurred in utero (91). Hand impairments can be attended to if they affect the child's ability to perform activities of daily living (ADLs) or don and doff a lower extremity prosthesis (92).

Etiologies such as Moebius syndrome and Poland syndrome (sequence) result in digital deformities associated with a more serious underlying condition. Moebius syndrome often affects the sixth and seventh cranial nerves, which compromises the child's ability to visually follow objects, swallow, and communicate. In addition to hand anomalies, Poland syndrome involves a partial absence of the ipsilateral pectoralis muscle and hypoplastic chest; therefore, a more proximal evaluation of any distal absence of limb is indicated.

Absence of individual digits creates a multitude of surgical and nonsurgical options. These include no intervention, therapy to enhance hand function, pollicization, or toe transfers. Due to the physiologic function of the normal thumb, hand impairments can vary widely, depending upon which digit(s) is/are absent. There is often greater consideration for surgery and function if the thumb is absent. Pollicization to the most radial digit can provide oppositional grasp (93). Through this microsurgery procedure, children can achieve remarkable functional improvement with improved cosmesis (94). Toe transfers can be transplanted from the second or third ray and minimize effects on gait mechanics (95,96).

Partial Hand and Wrist Disarticulation Deficiencies

Partial hand deficiencies are quite common and are often treated as wrist disarticulation level limbs. Very small underdeveloped vestigial digits, sometimes referred to as nubbins, are present in a majority of these cases, as is shortening of the ipsilateral radius and ulna. These vestigial remnants are rarely problematic, nor do they need to be surgically removed. The child can be quite functional with no intervention, and these remnants may enhance tactile input. The major functional drawback of this particular limb length is the inability to perform prehensile tasks with the involved limb, though the adaptability of the child can be remarkable. Plastic surgeons may be consulted for digit- and hand-level deformity.

Transverse Deficiencies of the Forearm

Transverse deficiency of the upper third of the forearm is the most common (major) upper limb deficiency (97). The clinical presentation of these children is similar to that of children with longer, transradial residual limbs with ipsilateral humeral shortening and small nubbins. The proximal radius in these shorter residua is often unstable, subluxing anteriorly during full extension, giving the appearance of hyperextension of the elbow. This creates a challenge to prosthetic fitting because the amount of length available for contact with the prosthesis is limited. Residual limbs spanning to the middle third or longer, tend to be more easily fit with a prosthesis, as they have more surface area over which to distribute the forces of the socket interface. They also have longer lever arms with which the patient can control the prosthesis. The ease of use of a prosthesis contributes to more functional tasks and prosthetic training. Traditionally, surgical intervention at this level remains rare. However, more recently, some pediatric hospitals have implemented pediatric hand transplant programs. These are generally in the research stage and remain limited to older pediatric patients due to size and logistics issues (98). If prosthetic intervention is not attempted or accepted, bimanual tasks will be performed via grasping of objects in the cubital fold, between one's legs, in the axilla region, or under the chin.

Both with and without prosthetic intervention, children with this level of congenital amputation can expect full functional independence. While this revelation casts some skepticism on the objective value of prosthetic intervention, technology continues to improve the functional enhancement afforded by prosthetic intervention.

Elbow Disarticulation and Transhumeral Deficiencies

The more articulations that are absent, the greater the functional deficit. When the elbow joint is compromised or absent, the child has fewer options to assist in

pre-positioning the distal limb in space and experience more limited pronation and supination for functional tasks of the prosthesis. In this case, the child relies solely on the muscles and ROM of the shoulder complex. The true elbow disarticulation limb has the distal epiphysis present, which is important for the overall growth of the residuum. A drawback of any disarticulation is the lack of room to fit prosthetic components and maintain humeral length equality by implementing elbow joints at the natural level.Transverse deficiencies of the humerus are analogous to acquired transhumeral amputations in children. The residual limbs are often medium to short in length compared to their contralateral limbs. This level of deficiency has been previously noted as the most common to experience diaphyseal overgrowth. This leads to a shorter limb that potentially is less functional and amendable to prosthetic fitting.

Shoulder Disarticulation and Intrascapulothoracic Deficiencies

It becomes increasingly difficult to restore the functions of the anatomic arm as the level of deficiency reaches the shoulder and higher with current technology, though promising innovations for prosthetic restoration are on the horizon. Children with remnant humeri can use these segments to assist in their activities. Often, the axilla will be used to assist these individuals to grasp and manipulate objects. If the child has unilateral limb deficiency, the contralateral noninvolved limb will be or become the dominant side for grasping, with manipulation taking place between the knees, in the mouth, or trapped between chin and chest or chin and shoulder. When the child has bilateral deficiencies at the shoulder level, the latter method is all that is possible to grasp objects. In these cases, the child will be strongly encouraged to use the feet to grasp and manipulate objects, often with remarkable facility.

Many designs of upper extremity prostheses require a degree of body movement (excursion) to operate the mechanical components. Most of this excursion is not present in the shoulder disarticulation level, as glenohumeral flexion no longer exists as a source of control input. This is further magnified when children have an intrascapulothoracic (forequarter) level of involvement, as they only have uniscapular motion to capture for prosthetic limb control. These two issues will be discussed at length in the following sections.

UNCOMMON UPPER LIMB DEFICIENCIES

Longitudinal Deficiencies of the Forearm

Radial deficiencies are approximately three times as common as ulnar deficiencies, occurring in 1 in 30,000 and 1 in 100,000 live births, respectively (99). Fanconi anemia; thrombocytopenia and absent radius (TAR); Holt–Oram syndrome; vertebral defects, anal atresia, tracheoesophageal fistula, radial and renal dysplasia (VATER); and Robert's syndrome are just a few examples of etiologies with associated radial involvement with variable level of limb deficiency, contractures of joints, and syndactyly (100,101). Figure 13.9 illustrates the complex issues with Robert's syndrome. The clinical presentation of radial deficiencies usually involves the radial-side digits of the hand as well. Depending upon the classification of the radial deficiency, prehensile capabilities may be compromised by a hypoplastic or absent thumb. In these situations, pollicization or toe-transfer procedures are considered. Treatment for radial deficiencies is focused on reconstructing the thumb and, in both the radial and ulnar deficiencies, is directed at centralization of the hand (102).

Ulnar deficiencies are associated more with musculoskeletal conditions than systemic conditions, and isolated genetic predispositions have been discovered (13). Cornelia de Lange syndrome, ulnar mammary syndrome, and ulnar fibula dysplasia are examples of syndromes that involve ulnar deficiencies. With ulnar-side involvement, the thumb and another digit are usually present.

Central ray syndrome, a form of ectrodactyly, had been described previously as having genetic predisposition. This is commonly referred to as "lobster claw," as the central component of the hand and/or feet are absent. This can present as a mild condition, with the more ulnar and radial digits still present, or it can present as two longer and thicker digits. Functional abilities with this condition will vary, depending upon the degree at which the syndrome affects the limb. Many of these individuals will not need prosthetic restoration, as the limbs are at full length and have prehensile and tactile capabilities. Surgical reconstruction may be recommended if the child lacks the oppositional capabilities that the thumb usually offers.

FIGURE 13.9 Child with Robert's syndrome. Note flexion contractures of all limbs.

Longitudinal Deficiencies of the Humerus

When a longitudinal deficiency of the humerus is present, it is often associated with deficiencies in the radius and ulna and with phocomelic digits. The length of the arm is compromised, which reduces the envelope of space needed to perform some bimanual tasks. For this reason, creative prosthetic fitting is more likely a consideration compared with isolated longitudinal deficiencies of the forearm. The shoulder complex may be compromised as well. Therefore, with prosthetic fitting, the child would most likely require some externally powered components. Frequently, the phocomelic digits will be used to provide manipulation of these components.

INTERVENTION, PROSTHETIC TREATMENT, AND ADAPTIVE EQUIPMENT

Although prosthetic treatment may seem indisputable for an individual with a limb absence or acquired amputation, the decision is not as straightforward as one might imagine. The inability to provide or restore the function of the human arm and hand poses great challenges to individuals with partial or complete limb loss (103). These fittings are generally limb-level-dependent as well and vary among oppositional, body-powered, and externally powered options. Discussing what the patient and family value as well as goals for a prosthesis prior to ordering may help with expectations and limit rejection. Acceptance of prosthesis is a complex issue; factors that influence acceptance include level of limb loss, presence of other complicating medical conditions, comfort and usefulness of the prosthesis, and acceptance of the limb deficiency by the family. In general, the higher the limb absence, the less likely it is that a child will find a prosthesis useful enough to wear it regularly. For example, transradial patients will tend to wear their prostheses more than transhumeral patients, and transhumeral patients will tend to wear their limb more than shoulder disarticulation patients (90). In general, for upper extremity loss, prostheses can be considered tools to accomplish certain tasks rather than being a necessity for routine day-to-day activities.

Goals of early intervention and training revolve around achieving age-appropriate milestones. Children with upper limb differences frequently achieve developmental milestones at or around the same age as children without limb anomalies. Prostheses are generally considered around 3 to 6 months of age (104). Until recently, 6 months of age used to be the time at which fitting was initiated (105,106). This was the age chosen because it was the time the child was expected to have achieved sitting balance and to begin to engage in bimanual tasks. Clinical experience versus evidence-based study guides fitting timetables (107). Although there are general guidelines for fittings, the initial fitting is something that is discussed in the clinic between the team members and family. Many children will be fitted with prostheses prior to 1 year of age. Early prosthetic fitting is designed to encourage bimanual tasks, establish a wearing pattern, increase overall independence, provide for symmetrical crawling, and reduce "stump dependence"—sensory dependence on the end of the residual limb (107). Early fitting does not guarantee acceptance (108, 109). The prostheses needs to fit comfortably, which can be challenging to assess in an infant, be relatively easily donned, equalize lengths with the noninvolved limb, allow for growth, and provide restoration acceptable to family (82).

Several different terminal devices may be considered for the first prosthesis. Age-appropriate prostheses are fitted to children; oppositional prostheses are generally the first design utilized. Options include hands, hooks of various shapes, mitts, and other nonhand designs. The vast majority of parents prefer a terminal device that looks like a hand. For this reason, it is recommended that a passive hand be provided rather than a hook or other nonhand device. The two basic passive hand options for infants are the closed, "crawling hand" design and the open hand design, or a hybrid between the two. The parents should be involved in the decision-making process—this involves providing information about the pros and cons of each style and, more importantly, letting the parents decide which design is most acceptable in their eyes. If parents view prosthetic use as a benefit, as the child ages both the patient and family may be more inclined to evaluate prosthetic components based on functional qualities in addition to appearance. If parents are involved in the decision and accept the device, they are more likely to encourage the youngster to wear the prosthesis. Figure 13.10 shows an infant passive hand.

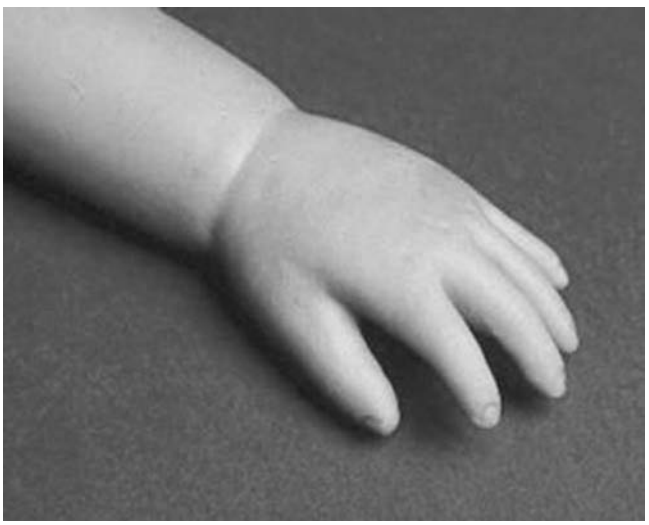

FIGURE 13.10 RSL Steeper infant foam-filled passive hand.

FIGURE 13.11 Transcarpal limb deficiency with adequate length for bimanual tasks.

It is questionable whether it is appropriate to fit children with partial hand deficiencies and wrist disarticulations at a very young age. They have long residua and can use them for bimanual tasks. Figure 13.11 illustrates a transcarpal limb deficiency with adequate length for function. The prosthesis would serve the purpose of providing a wearing pattern and also reducing dependence on the sensation of the limb. The latter can arguably be considered as a positive rather than a negative. Opposition posts are sometimes considered for the child with carpals and wrist motion. These devices can be rigidly fixed or placed in several different positions to accommodate for grasping different-sized objects.

For the child with a limb that extends distal to the elbow, the initial prosthesis is usually self-suspending, using a supracondylar design, with or without a suspension sleeve. If this is not achievable, a narrow Dacron harness may be designed in a figure-eight or figure-nine configuration. This harness should be easy to put on the child, have elastic as part of the straps for increased shoulder motion, and have snaps or fasteners that make it easy to put on and take off, ideally by the child without the need for assistance by a parent.

The same oppositional terminal device options are appropriate for the child with a limb deficiency proximal to the elbow. The major difference between these levels is that the absence of the elbow joint makes it more difficult to pre-position the terminal device for bimanual tasks. The child is not cognitively ready for an articulating elbow; therefore, a curve-shaped "banana" arm is often provided in order for this child to engage the prosthesis with the contralateral hand as well as reach levels that are closer to the midline and face (110). Figure 13.12 displays the passive "banana arm" prosthesis.

The next developmental milestone is walking, which usually occurs at 11 to 13 months of age. This will indicate that the child is ready for a more sophisticated upper extremity prosthesis. At this time, the child is ready to perform simple grasp-and-release activities using the prosthesis. It is imperative that the family be involved in the clinical decision making about their child's prosthesis. The prosthetist should design the prosthesis in a manner to accommodate growth. It is best to keep the control system as simple as possible at this early age in order to ensure early success. Other developmental factors to be considered are understanding of holding function, attention span longer than 5 minutes, and willingness to be handled by an occupational therapist to go through terminal device opening motion. The prosthetic training at this stage relies on a child's ability to grasp a cause-and-effect-type relationship when participating in therapy.

When the child is developmentally ready for terminal device activation, options include body-powered hooks or hands as well as myoelectrically controlled

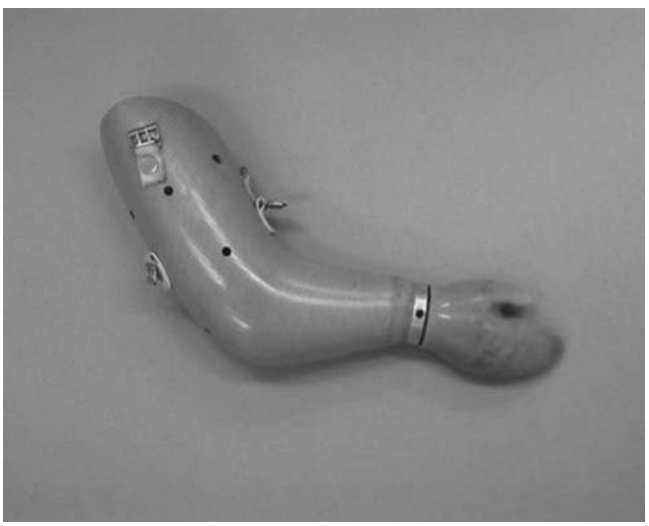

FIGURE 13.12 Transhumeral passive "banana arm" prosthesis.

hands. The majority of parents prefer hands over hooks due to the cosmetic improvement gained; the hands that provide optimal function at this age are myoelectrically controlled, though the weight of the prosthesis must be considered relative to the length of the residual limb. At this age, the simplicity of control is of paramount importance. An electric hand that is controlled by one electrode in a voluntary-opening control scheme has proven effective and natural. This electronic scheme permits the child to activate the hand opening with a contraction (usually on the side of the wrist extensors) and relaxation that enables the hand to automatically close, or "cookie crusher" design. This electromechanical design is analogous to a split hook, voluntary-opening prosthesis. Designing such an electronic control scheme eliminates the need for the child to maintain muscle contraction in order to continue grasping the object, which proves tiresome and may limit function. As the child grows older, another electrode can be added to the flexor side of the forearm, enabling the child to have volitional control opening and closing the myoelectric hand in a more physiologic manner (111).

Myoelectric hands of the past were too large and difficult for a 1- or 2-year-old child to use successfully. Therefore, it was recommended that these hands not be fitted on children until 4 to 5 years of age. Currently because of improvements in prosthetic design, it is common for these hands to be fitted successfully on 1-year-old children. Prosthetic technology has improved dramatically as a result of miniaturization, improved materials, battery weight, and simpler control to better meet the needs of very young children. Figure 13.13 shows a transradial myoelectric prosthesis with myoelectric hand terminal device.

Body-powered devices may not work well for this age group because they lack the requisite force and excursion, as well as the cognitive ability, to relate shoulder motions to terminal device operation. The voluntary-opening-style terminal devices permit the user to grasp an object and allow the force of the elastic bands or springs to keep the object in the terminal device. As the number of bands increases, the amount of force required to open the terminal device increases as well; thus, the child may not be able to overcome the force required to activate the terminal device. The designs of voluntary-opening terminal devices for children are not very aesthetically pleasing, with the exception of the mechanical hands. The hands, however, have the drawback of providing minimal efficiency. Once a cosmetic glove is applied to the mechanical hand, it can lose up to 40% of its efficiency, compared to the function of the hand without the glove. Voluntary-closing terminal devices have gained in popularity, although the child must maintain force and excursion through the harness to maintain grasp on an object. The amount of grasping force is directly proportional to the force that the child puts into the harness (112). The prosthetic team can predict the ability of the child to control the myoelectric components when using evaluative tools such as the Capacity of Myoelectric Control (113,114), by means of developmentally engaging devices triggered by the muscles targeted for prosthetic activation.

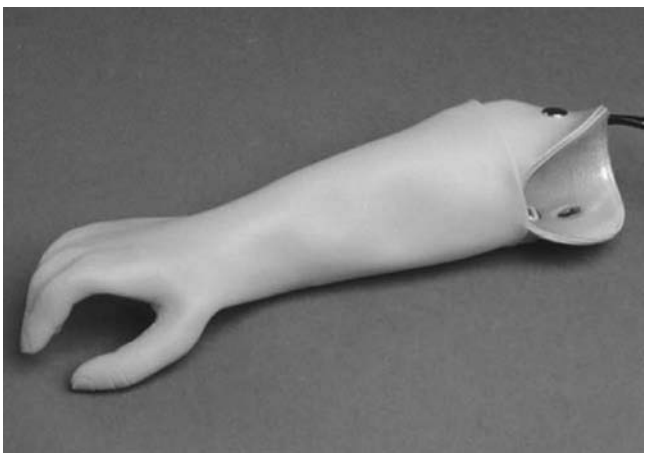

FIGURE 13.13 Transradial myoelectric prosthesis.

By the time children are 4 or 5 years old, they are able to operate virtually all types of prosthetic components and control schemes presently available (112).

The developmental milestones described previously should guide the fitting schedule of the child with transhumeral limb involvement. Because of the nature of a transhumeral prosthesis, it can be more of an encumbrance than the transradial design. This can cause difficulty in rolling over, and may impede the child's development if fitted too early. The terminal device should be activated shortly after the child begins to walk. Terminal devices for the transhumeral level are the same as for the transradial. The addition of a prosthetic elbow is the key difference. The first prehensile prosthesis will employ a friction elbow to allow positioning of the terminal device. It is useful to limit the ROM at the elbow by producing flexion and extension stops to prevent the elbow from flexing excessively during weight-bearing activities (eg, crawling). The initial prosthesis may be suspended by a harness or by silicone suction suspension. The silicone suction socket (3S) has proven effective because it allows free ROM at the shoulder and provides excellent suspension. The child with a transhumeral deficit should be fitted with an activated terminal device once he or she begins to walk.

Considerations for terminal device selection include appearance, weight, ease of operation, and cost. With the initial prosthesis, the myoelectric hand offers reasonable appearance and ease of operation when controlled by a single-site voluntary-opening circuit; however, it is a heavier and more expensive prosthesis compared to body-powered. By starting with a passive hand device, a child may demonstrate ability to tolerate a prosthesis prior to incorporating more expensive myoelectric components.

Either voluntary-opening or voluntary-closing designs can be used successfully by the child with transhumeral limb involvement once the child has sufficient strength and the cognitive ability to understand how to operate the device. This usually is possible at 2 to 3 years of age. When the child is strong enough to operate an active elbow, usually at ages 4 to 5, a conventional body-powered elbow may be provided; however, locking of the elbow by conventional methods may prove challenging. Depending on the length of the residual limb, the child may have insufficient strength/excursion to operate the body-powered elbow. In this case, an electric elbow may be considered, although the increased weight may preclude this option. The terminal device illustrated in Figure 13.14 is a voluntary-opening split hook and can be utilized on both transhumeral and transradial deficiencies.

The shoulder disarticulation level is treated differently due to the challenge in positioning the shoulder, elbow, and terminal device. With bilateral involvement, different components may be selected for each side, with function and gadget tolerance guiding prosthetic decision making. The child may be fitted with a passive endoskeletal shoulder and elbow with an active terminal device. Externally powered hands controlled by either electromechanical rocker switches or force-sensing resistors may be used. The child is encouraged to maintain good ROM for shoulder elevation/depression and protraction/retraction in order to make contact with these input devices. The return or enhancement of function using these devices is quite limited. Therefore, there are no "right" philosophies for the fitting of these complicated cases. The team should recognize that prostheses need to be useful to the child, or they will be rejected despite careful decision making. A task-specific prosthetic terminal device is illustrated in Figure 13.15.

The patient with bilateral total upper extremity transverse deficiency and phocomelic residua rarely requires amputation revision; indeed, the terminal digits can activate switches or myoelectric sensors (103). In the case of higher level bilateral deficiencies, it is wise to start as simple as possible, recognizing that each child has a certain level of tolerance for "gadgets." With the vast array of prosthetic components now available, it would be easy for the well-intentioned clinic team to recommend components that would overwhelm the user and lead to prosthetic rejection. Critical factors in the success of the high-level bilateral amputees are prosthetic weight, complexity of control, proprioceptive feedback, wearing comfort components, and motivation and attitude of child and family.

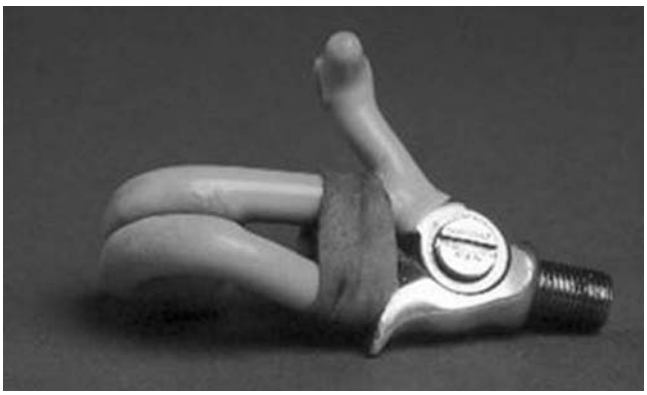

FIGURE 13.14 Hosmer voluntary-opening, split hook terminal device.

THERAPY AND TRAINING

The preprosthetic period is mainly focused on the needs of the parents (115). The family level of distress or stress related to the child's limb deficiency will vary (116). It is important during this initial contact for the clinic team to present an honest forecast of the prosthetic plan. The team members must walk a fine line between presenting the prosthetic options in an honest manner without sounding negative or disheartening. Despite the sophistication of prosthetic technology, it remains far from the ideal of replacing a physiologic arm.

The parents should be encouraged to treat the child as they would a child with normal limbs. Introducing age-appropriate ways for children to explain their limb deficiencies to peers allows for improved social interactions. Additionally, allowing parents the initial time to grieve over the absence of their child's limb is helpful. Many parents benefit from being introduced to other parents and children with similar limb deficiencies (117). Having coordinated clinic scheduling often allows for spontaneous interactions between families and encourages a more open, welcome environment for discussion of challenges or problems.

FIGURE 13.15 Adaptive prosthesis allows for modified activities to enhance a prothetic user's quality of life. Discussion about a patient's desires for learning a new skill prompts creative solutions. (Photo courtesy of TRS website.)

Typically, children younger than 3 years of age have therapy provided in the home through an early intervention program. Prone positioning is important for encouraging trunk extension and mobility. Gross motor milestones are generally not delayed, but may be affected by asymmetry imposed by unilateral upper limb deficiencies. Children compensate and substitute for the missing action of limbs. With preprosthetic and prosthetic training, therapists bridge the delivery of the prosthesis to the initiation of function, and create a comfortable environment for children to explore options, both with and without the prosthesis. The goal is to increase the child's awareness of the affected side, including the prosthetic device. The child should also be encouraged to use the prosthesis for transitional movements, such as sitting to crawling, and leaning on the prosthesis for weight-bearing while reaching with the dominant hand. The parent is encouraged to maintain contact with the therapist to answer questions regarding follow-through with prosthetic usage. A recheck through the clinic should be scheduled within a month after delivery of the prosthesis and then every 3 to 4 months. Frequent evaluations in the clinic allow for prevention of complications and continued open dialogue about prosthetic interventions and modifications.

As the child grows and the prosthesis increases in complexity, the therapist will again provide initial instruction to the parents and child. A structured approach to the use of the terminal device assists parents, child, and therapist in gaining confidence and competence; however, children may be different in tolerance and function with a prosthesis, even if they have the same deficiency (118). Initially, the therapist will work with the child and parents using toys that encourage bimanual use, like Legos, pop beads, and other age-appropriate, simple activities. It is useful to concentrate on activities that require the prosthetic side to hold while the dominant hand manipulates (ie, to use the prosthesis as a helping device). When training a child in the use of a myoelectric hand using this control scheme, the therapist should encourage activities that cause the hand to open. Because of the placement of the electrode over the forearm extensors, activities that elicit an extensor activity are appropriate. Once the hand is open, the therapist can quickly place a toy in the hand and encourage the child to release it. The child will learn through repetition.

It is unrealistic and inappropriate to teach the child to use the prosthesis for dominant hand activities, even in the case of traumatic amputation to the previously dominant side. Children with high-level bilateral upper extremity limb deficiencies will utilize their feet in a natural manner. The child with an isolated limb deficiency or amputation is capable of achieving age-level academic skills. Few studies have been done to define achievement academically. Good social adjustment is reported for children with myoelectric prosthetic use (119). School placement is almost entirely within the regular school system, with an Individualized Education Program (IEP) to address educationally related function. Occupational therapists will assist with issues of grasp and fine motor control for paper, computer, and ADL tasks needed in school. Informational pamphlets have been developed for the teacher to prepare the able-bodied students for integration of children with physical disabilities into the regular education classroom.

Adapted physical education may be necessary, but regular physical education is often encouraged. The philosophy promoted for children with physical disability is that of "participation, not observation." Participation in athletic endeavors such as skiing, tennis, and other physical exercises improves the self-concept of the child or adult with limb deficiency. Specialized, adaptive prosthetic components that enable children with unilateral or bilateral limb deficiency to participate in sports such as golf, shooting, and ball sports have escalated since the 1980s.

Functional assessments recently developed to determine the use of upper extremity prosthetics and function have included Assisting Hand Assessment, the Prosthetic Upper Limb Functional Index, The University New Brunswick Test of Prosthetic Function, Child Amputee Prosthetic Project—Functional Status Index, Child Amputee Prosthetic Project—Functional Status Index Preschool, Shriners Hospital Upper Extremity Evaluation (SHUEE), Capacity for Myoelectric Control (113), and Unilateral Below Elbow Test (120–126). It is typical for children to perform ADLs with their prosthesis, but often choose not to utilize them (127). In addition, prosthetics are often utilized for specific tasks versus everyday tasks. Children typically utilize nonprosthetic options from 3.5 through 13 years of age (115). The emphasis of prosthetic training should be on independence both with and without the use of a prosthesis.

Outcomes related to patient satisfaction are increasingly important to evaluate for prosthetics (128,129).

Recent studies have indicated that children with unilateral, below-elbow deficiencies who do not wear prostheses perform as well as or better than their counterparts who wear prostheses (88).

ADVANCEMENTS IN UPPER EXTREMITY PROSTHETICS

Many additional components are now offered for children with upper limb involvement; however, one of the most exciting advancements comes in the form of a new application to nerve transfers. Although historically only performed in adults, targeted muscle reinnervation (TMR) is an effective means of creating additional, physiologically appropriate myoelectric sites for individuals with high-level, upper extremity amputations (130–132).

Following amputation, the remaining peripheral nerves (ie, median and distal radial nerves) are grafted to denervated muscle sites in order to create additional,

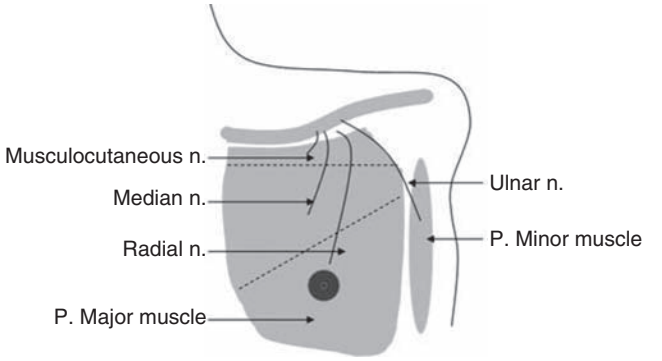

FIGURE 13.16 Proposed nerve transfer for targeted muscle reinnervation of an individual with shoulder disarticulation amputation.

distinct myoelectric sites for the user that are physiologically appropriate, as illustrated in Figure 13.16. For example, on the transhumeral limb, the medial head of the biceps and the lateral head of the triceps are denervated and reinnervated by the median and distal radial nerves, respectively. When the reinnervation is complete (after approximately 4–6 months), these two additional myoelectric sites are available for physiologic control of closing and opening a myoelectric terminal device. The prosthetic socket then incorporates four independent myoelectric sites for control of elbow flexion and extension, via native lateral biceps and medial triceps, and control of the terminal device by the aforementioned reinnervated muscles. Increased efficiency and ease of use have been positive outcomes from this surgical intervention.

Pattern recognition is beginning to be utilized as a new intuitive control strategy when incorporated into transradial sockets or higher level prostheses after TMR surgery. Pattern recognition software utilizes an increased number of electrodes within the socket versus conventional placement directly over the muscle belly. Users have shown the ability to complete tasks 0.290 to 1 second faster and with greater accuracy as compared to clinically localized electrode placement (133).

In the adult population in the past few years, hand transplantation surgery has increased in frequency; however, less is known about the ability to perform this surgery in the pediatric population. The issues of drug-related side effects, uncertain long-term outcomes, length of immunosuppression, and high cost of surgery appear to be the barriers at this point.

LOWER LIMB

Deficiencies of the lower limb are less frequent than deficiencies of the upper limb, but surgical and rehabilitation management may be more involved. Most of the common congenital lower limb anomalies are longitudinal deficiencies. Despite the complexity of the early intervention, lower limb prostheses generally have high acceptance rates due to the functional gain of ambulation. In addition, mobility demands less precision than the positioning and fine motor skills of the upper limb.

Surgical intervention is often required to correct the deformity or provide a functional lower limb. This is the most challenging aspect of the early management of these children. Parents are often faced with difficult decisions of choosing among such surgeries as foot ablation, angulation osteotomies, epiphysiodeses, limb lengthening, and rotationplasty. In addition to the usual risks of surgery and uncertain outcome, ethnic and religious barriers are important in family decisions. Parents may benefit by meeting other families who have faced similar situations. This may ease the discomfort of the decision making for the parents and child (117).

COMMON LOWER LIMB DEFICIENCIES

Longitudinal Deficiency of the Fibula

The most common, and possibly the most controversial, deficiency regarding management is the longitudinal deficiency of the fibula. Many classification schemes and levels of involvement exist. With partial deficiencies of the fibula, outcomes will vary.

There is no evidence that this anomaly, in isolation, is genetically transmitted (13). It has been suggested that the fibula is undergoing "regressive evolution" and that may be the reason for the prevalence of deficiency of the fibula and susceptibility to congenital absence (134).

The clinical presentation of longitudinal deficiency of the fibula, with a completely absent fibula, generally has a foreshortened tibial section, and frequently, ipsilateral femoral shortening. This tibial section appears shorter than it is as a result of torsional bowing. This anterior bowing of the tibia shortens the segment longitudinally and creates an anterior prominence of the tibia. This anterior prominence is indicated by a subcutaneous dimple, which can range from superficial to invaginated. Figure 13.17 shows a child with a fibular deficiency. Proximally, the limb is often in genu valgum or drifts into genu valgum as the child grows. The distal involvement is usually an equinus position and a valgus posture during weight-bearing due to lack of lateral support. Lateral tarsal and ray absences are often associated with this lateral long bone absence. As the child grows, the popliteal area becomes convex, with the medial hamstrings descending much lower than the lateral hamstrings. On physical examination, the degree of internal hip rotation is often less than that of external hip rotation.

Many surgical options are available for the treatment of longitudinal deficiencies of the lower limb. Historically, the most common treatment of a complete fibular absence

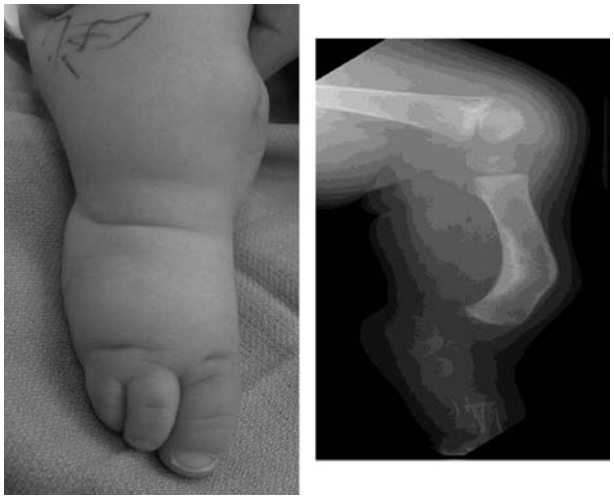

FIGURE 13.17 This child with fibular deficiency and an element of angular deformity subsequently required a transtibial, or below the knee, amputation to improve functional mobility and facilitate ambulation with a prosthesis. (Photo courtesy of the personal collection of Dr. Vincent Mosca, MD.)

has been with a Syme amputation, which is successful in providing an end-bearing surface for ambulation, with or without a prosthesis (135,136). Amputation takes place when the child is beginning to pull to stand and cruise with the assist of furniture or toys to time the surgery to assist with developmental milestones. Migration of the heel pad posterolaterally has been noted in the follow-up of many Syme amputations, as shown in Figure 13.18. This migration may be due to the use of the posterior calf musculature during active ambulation in the prosthesis. The Boyd amputation serves to centralize the heel pad more effectively and is the surgery of choice in many clinics. In addition to the ankle disarticulation procedures, it may be necessary for the child to undergo unilateral epiphysiodesis or angulation osteotomies if the genu valgum becomes a prosthetic challenge to fit. Outcomes from Syme amputations have shown that these children are able to perform very well in their communities, have a good self-image, and are rarely limited in activities (137).

External fixator applications and advancements have provided options that challenge the team and orthopedists to reconsider amputation. Saving the foot would be the first choice if the procedure were proven to be as successful as the Syme amputation (137). Considerations for these procedures include level of involvement or "grade," risks and psychological effects of multiple surgeries, potential (and probable) infections around pin sites, and physical effects of "downtime" the child will experience during and after wearing the external fixator (7). Some centers advocate multiple, sequential surgeries in addition to lengthening, and it will be interesting to learn long-term outcomes in comparison with the more

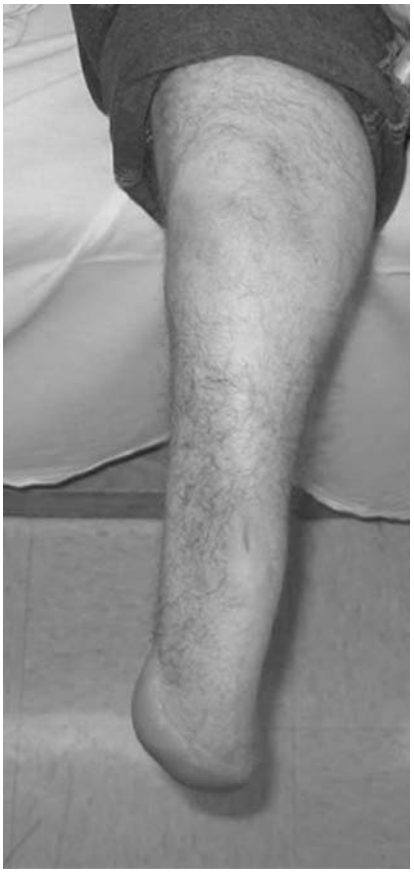

FIGURE 13.18 Limb with longitudinal deficiency of the fibula following Syme amputation. Note posterior lateral migration of heel pad.

simple "one and done" approach with amputation. Factors for consideration will include stability of the residual ankle, foot (especially if three-toed or less), and knee, compared with solid prosthetic restoration. In the event that the foot ablation is not imminent, orthotic fitting combined with shoe modifications will allow the child to ambulate successfully.

Femoral Abnormalities

The term that has been used to define the most common deficiency of the femur is *proximal femoral focal deficiency* (PFFD); despite ongoing controversy regarding the descriptive utility of this name with varying levels described first by Aitken in the late 1960s, PFFD has been the acronym of choice for many femoral anomalies. Congenital short femur differs from PFFD by having an intact proximal femur and acetabulum.

Although the skeletal structures are quite variable, the clinical presentation for limbs with PFFD is similar. The femoral section is shorter, with a larger mass of soft tissue, which includes musculature, between the pelvis and the involved knee. A typical appearance is shown

FIGURE 13.19 The three photos represent a child with proximal femoral focal deficiency who underwent a fusion of the femoral remnant to the tibial remnant, effectively creating a residual limb that functions and has prosthetic needs of a knee disarticulation patient. A Van Ness rotationplasty is another surgical procedure frequently used in this population. (Photos courtesy of the personal collection of Dr. Vincent Mosca, MD.)

in Figure 13.19. These muscles are highly ineffective as a result of being slack and not stretched to their full potential. The hip posture and stability are quite variable. All of the limbs present with some degree of hip flexion, abduction, and external rotation. For the congenitally short femur and more subtle PFFD, the labrum is present, resisting proximal subluxation. More involved presentations have progressive subluxation to dislocation of the femur. Often, there is an associated fibular absence.

Surgical options for the congenitally short femur are numerous and each option should be shared with the family. Surgery may be a topic that needs to be addressed multiple times as the child ages and changes. The first option is "no surgery." Some clinics and families feel that no surgical intervention is the best option for this disorder. In these cases, various lengthening devices such as shoe lifts and extension prostheses are necessary. These are sometimes referred to as "prosthoses," because they often combine a proximal orthosis with distal prosthetic components. An example of the "prosthosis" is seen in Figure 13.20. Children with bilateral femoral involvement often need no surgeries and no prostheses. For children with an intact short femur and both proximal and distal growth plates, this option may be a consideration; the addition of an external fixator to lengthen the foreshortened femur may also be considered. If the amount of lengthening necessary is unattainable, the limb lengthening may be performed in conjunction with appropriately timed epiphysiodesis of the contralateral leg to equalize leg lengths at full maturity.

Ankle disarticulation along with knee arthrodesis is another option for PFFD. It is considered when the proximal femur is affected and when the length discrepancy is such that an external fixator could not achieve the desired lengthening. The ankle disarticulation amputation is either a Syme or Boyd. Figure 13.21 is an example of a Syme amputation. Fusion of the knee may be delayed, as arrest of the proximal tibial and distal femoral growth plates will occur at that time, leaving a shorter overall limb length to control a prosthesis. This delay may be unnecessary if the overall limb length and the attempt to provide adequate space for congruency of a prosthetic knee with the noninvolved knee may be achieved during one surgery.

Another surgical option is a rotationplasty procedure, such as the Van Ness procedure. An intact fibula is preferred for this surgical procedure. The procedure involves rotation of the foot 180 degrees through removal of the distal femoral and proximal tibial epiphyses, and rotation of the distal segment prior to internal fixation (75). The rotated foot can now act as a knee, utilizing ankle dorsiflexion as knee flexion and ankle plantar flexion as knee extension, as in Figure 13.22. This procedure has demonstrated effective outcomes; however, the aesthetic appearance of the limb following surgery has limited its popularity (138,139). It is crucial that the therapist and family aggressively work on maintaining the full ROM of the ankle, especially in the sagittal plane. If this does not occur, all that has been accomplished is turning a foot "backwards" on the leg. The prosthetist can make angular adjustments to the prosthesis to further increase ankle ROM as the patient begins to ambulate. Derotation of the foot has occurred on occasion, requiring additional surgery to again position the forefoot posteriorly. Patients who have undergone this procedure demonstrate fluid gait patterns in a prosthesis and maintain good contact

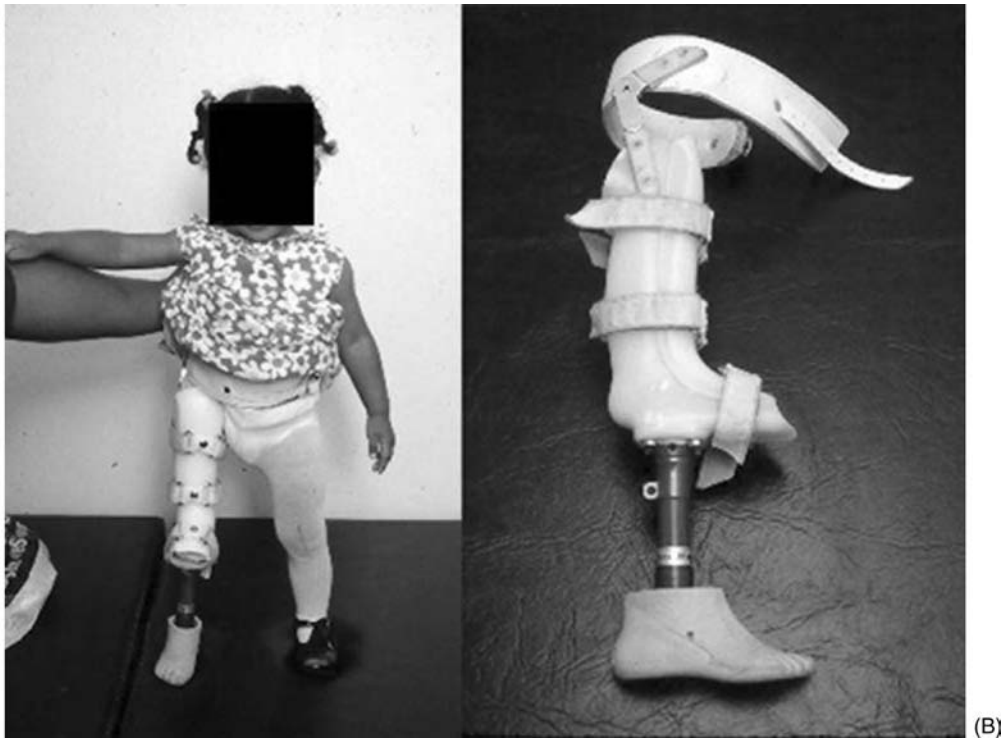

FIGURE 13.20 (A) Child with femoral abnormality wearing "prosthosis." Prosthosis is a combination bivalve knee ankle foot orthosis (KAFO) with pelvic band and hip joint (B), extended with a prosthetic pylon and foot.

FIGURE 13.21 Child with femoral abnormality following Syme amputation. Note bulbous distal end of limb and proximal thigh musculature.

with the socket over pressure tolerant areas. The Van Ness procedure has been utilized to restore mobility in other disorders such as burns and osteosarcomas (140,141).

In order to address the proximal subluxation of the femur and provide for a single articulation within the prosthesis, an iliofemoral fusion may be performed.

This may be in conjunction with a rotationplasty procedure or in isolation (142).

Longitudinal Deficiency of the Tibia

Longitudinal deficiency of the tibia is rare, occurring in 1 in 1 million births. Genetic transmission has been associated with these anomalies, particularly when a bifurcated distal femur exists; 30% of partial tibial deficiency occurs as an autosomal-dominant inherited pattern. The treatment is straightforward since the tibia is the major weight-bearing bone. Differences in treatment occur between complete and partial tibial absence. Figure 13.23 shows examples of tibial deficiency by means of a radiograph.

The clinical presentation of a longitudinal tibial deficiency may include a varus foot and lower leg, a short leg, and an unstable knee and ankle (or both). The foot may have medial tarsals, metatarsals, and rays missing as well, or may demonstrate fibular duplication and mirror-like polydactyly. On a radiograph, a distal femoral bifurcation may add to the challenge of prosthetic fitting as well as be an indicator for genetic influence (143).

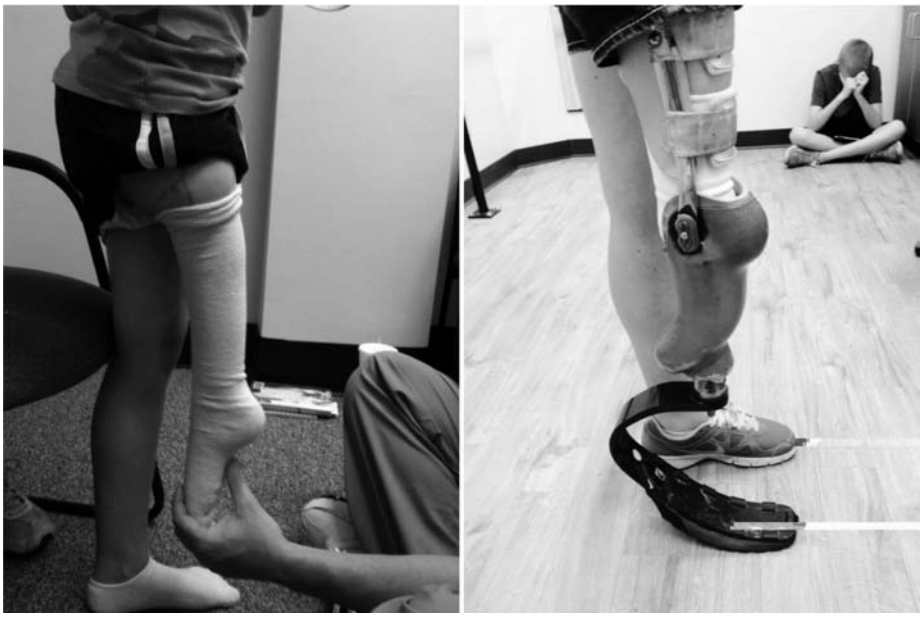

FIGURE 13.22 Limb post-rotationplasty with prosthetic fitting capable of high level gross motor activities.

When there is a complete absence of the tibia, the treatment of choice is disarticulation at the knee. The fibula cannot sustain weight-bearing of the individual at full maturity, and the instability of the knee and ankle is too great for corrective measures. For the child with a partial tibial deficiency, the segment length is important. If the tibial segment is short, the surgeon creates a synostosis with the intact fibula in conjunction with amputation of the foot. If the heel pad is retained, this procedure will create a walking surface for the child, providing stability without a prosthesis. However, this limb length is shorter than the aforementioned Syme amputation and may prove difficult for the child to walk without his or her prosthesis. Although treated as a "transtibial amputation," many of these residua grow in quite a different manner. Often, the distal tibia and fibula fuse, while the fibula continues to grow at a faster rate than the tibia. The resulting deformity is a laterally bowing lower leg (the distal end is pushed medially), a fibular head that becomes more prominent and continues to grow proximal-laterally, or both. These additional deformities may be challenging to accommodate within the socket of a prosthesis and may necessitate revision surgery, complex prosthetic care, or both.

UNCOMMON LOWER LIMB DEFICIENCIES

A few of the less common lower limb deficiencies include those resulting from amniotic band syndrome, central ray syndrome, Robert's syndrome, and sacral agenesis. The amniotic banding can occur at any level, but frequently causes autoamputation at the transtibial level (25). The

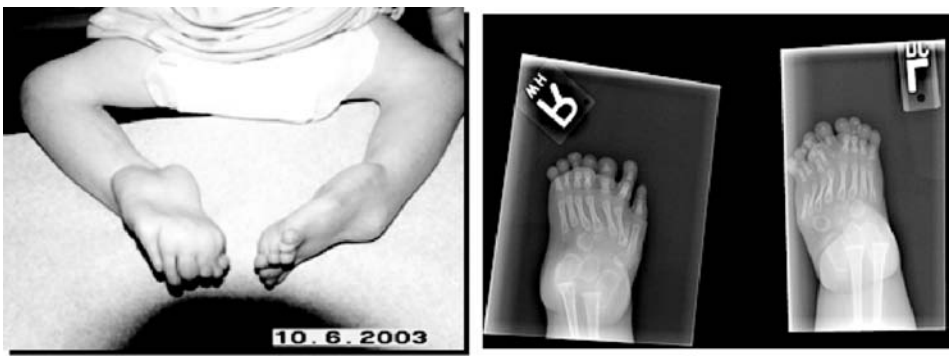

FIGURE 13.23 Absent tibia with duplication of the fibulas bilaterally with mirror duplication of the foot.

critical factor in working with and fitting these children is keeping in mind that the likelihood of bony overgrowth is great as a result of the banding occurring at the diaphyseal level. Central ray syndrome and Robert's syndrome have autosomal-dominant inheritance. They can present with a wide array of lower limb anomalies. The result of these presentations and subsequent surgeries will vary. It should be noted that in the case of Robert's syndrome, maintenance of ROM should be stressed to prevent severe limb contractures; these contractures can affect both fit and function in prostheses. Sacral agenesis is a frequent cause of hip disarticulation or hemipelvectomy; the higher the level of amputation, the more difficult to obtain satisfactory prosthetic fitting.

COMMONLY ACQUIRED LOWER LIMB AMPUTATIONS IN CHILDREN

Acquired lower limb amputations are the result of trauma, tumor, or infection. Traumatic lower limb amputations occur more frequently than traumatic amputations of the upper limb. These amputations occur secondary to lawn mower, train, motor vehicle, and farming accidents (38–40). Lawn mower accidents often result in partial foot amputations. Train accidents are generally a result of teenagers attempting to board slow-moving trains. The amputations are often high and/or bilateral in nature as a result of the current that draws the youngster under the wheels. Motor vehicle and farming accidents present with varying amputation levels. Because of the pathologic differences between adults and children who undergo acquired amputations, surgeons can be more creative with skin coverage. Additionally, the energy expenditure after amputation of age-matched controls appears similar for lower limb amputations that have an intact knee unit; however, patients with amputations at the knee disarticulation level or higher required a reduced walking speed and increased oxygen utilization (144). Involving a limb deficiency team prior to surgery helps families evaluate their options and may lead to less distress over the surgery. The success rate of limb-salvage attempts demonstrates the pediatric population's resiliency to trauma, with some reports of 84% success rate with limb salvage (145).

Meningococcemia and staphylococcal infections with the onset of purpura fulminans can be destructive. If the child is fortunate enough to survive, there are often multiple limb amputations. Lower limb amputations can range from partial foot to transfemoral levels; the most frequent are transtibial levels. Complications due to growth plate arrest, bony overgrowth, and fragile skin may necessitate revision to the knee disarticulation level (69,73).

Both osteogenic and Ewing's sarcoma are more prevalent in the lower limb than in the upper limb. Osteogenic sarcoma tends to have a better survival rate, in that it involves more skeletal than soft tissue structures. Limb-salvage techniques and endoprostheses in conjunction with chemical and radiation therapies have often averted the necessity of amputation (49,50, 52–54,58,59,63,140,146–148). Regardless of whether a child undergoes amputation or limb-salvage procedure, maintaining mobility and preventing loss of any functional status help overall outcomes.

Many lower limb amputations that occur resulting from osteogenic sarcoma are at the transfemoral level, while Ewing's sarcoma tends to migrate more proximally to the region of the upper thigh and pelvis.

INTERVENTION, PROSTHETIC TREATMENT, AND ADAPTIVE EQUIPMENT

Prosthetic fittings for the child with a fibular deficiency and subsequently managed by ankle disarticulation should be successful. At the time of the child's first prosthetic fitting, there is still significant soft tissue surrounding the lower leg and ankle region. As the child grows, the definition around his or her ankle becomes greater, resulting in a "bulbous-shaped" distal residuum. This is not as large as the typical Syme amputation because the lateral malleolus is not present. Therefore, using the bony prominences of the ankle as a sole means of suspension is inappropriate (13). A sleeve will act as auxiliary suspension with the purpose of keeping the prosthesis on the child's limb, but should be pliable enough to not restrict ROM during ambulation (52). As the child and limb mature, anatomic suspension can be utilized. Children with Boyd amputation may be able to take advantage of this sooner, as the distal residuum becomes more bulbous sooner. The prosthetists should be able to take advantage of the distal residuum and eliminate the need for auxiliary suspension. Many methods have been used to accommodate donning of bulbous residuum into prostheses. One method is to utilize a gel or silicone liner used in conjunction with a flexible inner liner and rigid frame with cutouts to allow for expansion. The major challenge is to permit the larger, distal end to pass through a narrower portion of the socket that should provide total contact with the limb when it is fully seated.

Intervention for children with femoral abnormalities varies from shoe inserts to transfemoral (PFFD style) prostheses. The child with a small femoral length discrepancy may need nothing more than a shoe insert or external shoe lift in order to equalize the length of the legs. This is only possible if hip flexion, abduction, and external rotation are addressed. Hips contracted in a flexed position often lead to a compensatory flexed knee. This posture is unstable in the early stance phase of gait and may, therefore, require further intervention. Ankle foot orthoses (AFOs) that accommodate for the angles of the leg and foot (usually equinus) can be used with shoe modifications. These orthoses may need to provide

an external extension moment (ie, floor-reaction AFO design) if contractures have not been resolved.

As leg length discrepancy increases, the orthoses begin to morph into prosthoses, combining elements of both prosthetic and orthotic design. The components used are no longer shoe lifts, but prosthetic feet inside regular shoes. The gait of the child can be asymmetrical because of different knee center heights. Transtibial prostheses have been used for some children with stable knees and knee centers that are higher than the contralateral side. Benefits of this include better gait mechanics and control of external knee flexion movement in early stance by quadriceps versus hip extensors. A drawback of this type of fitting is that when this individual sits, the top of the affected knee will be much higher than the nonaffected knee because the tibia is longer than the femur. This is generally acceptable to the user and preferred to lengthening or amputation. In the event that the entire leg length, including foot or ankle disarticulation limb, is equal to or more proximal than the contralateral knee, a PFFD-style, transfemoral prosthesis is indicated. Capturing the proximal contours of the limb, with or without a foot present, and determining the appropriate height of the prosthesis are just two problems for the prosthetist. Attempts are made to fit ischial containment sockets to block the motion of the pelvis with respect to the femur, thus preventing subluxation of the femoral head. The difficulty with this is that with the soft tissue mass in the proximal thigh, the socket often is so high that it contacts the perineum on the contralateral side. If an articulation is added to these longer limbs, many times, "outside hinges" are used in conjunction with an elastic extension assist for stability at initial contact and loading response. Single pivot, upper extremity hinges are frequently used because of the size of the child at the initial fitting. If there is room, a locking knee joint may be added initially to provide stability and can be unlocked during sitting. Frequently, polycentric knee joints are used for these children as they get older to address hip instability and control of the prosthesis, enable swing phase clearance because the linkages "shorten" the lower leg when the knee is flexed, and provide minimal femoral length discrepancy during sitting.

A myriad of prosthetic knees and feet can be used for these children, provided there is room for the components and that they are at an appropriate functional level to benefit from the components. Even with higher levels of involvement, children are frequently variable cadence ambulators and can take advantage of the high-technology components and components that can adapt to changes in speed and terrain (149,150). Figure 13.24 shows a foot with a shell that allows sandal wear.

When a knee disarticulation has been performed, as is typically the case with a complete absence of the tibia, prostheses similar to the "above-knee" prosthesis mentioned for the children with femoral absence exist. The knees and feet are used in a similar sequence and fashion. The main difference is the socket design. The child may need a relatively aggressive socket at a young age to capture the limb and provide maximum stability. When the child matures, the socket will be trimmed much lower because the presence of good hip musculature, including the hip adductors, will enable the child to control the prosthesis well and walk with only minor gait deviations. Care must be taken to not lose rotational control of the prosthesis when lowering the proximal trimlines of the socket or create a stress riser on the femur.

The child with a partial tibial deficiency is fitted with a prosthesis that resembles a standard transtibial design. Once the tibiofibular synostosis has healed, the child can utilize most of the options of transtibial prostheses, including pin-locking liners and multiaxial dynamic response feet. The angular deformities that follow surgical reconstruction may prove challenging to the prosthetists in terms of the socket design. They may need to provide a means of donning the device that is atypical of transtibial designs and more like that of ankle disarticulations. A socket with a removable panel (door) may need to be created to permit the limb to successfully enter the socket. Closure is often provided for by straps or Velcro.

Congenital lower extremity limb deficiency may present with odd combinations of absent portions of the extremity and deformities of the remaining segments. The deficiency may include proximal muscles, skin, nails, and parts of the joint. The child with PFFD, for instance, may also have upper extremity limb deficiencies, which create challenges for donning/doffing clothes, prosthesis, and the use of prosthesis.

Treatments of varying levels of deficiency of acquired amputation are individualized based on the level, number, and condition of the amputation(s).

Children with traumatic limb loss are treated in a fashion similar to adults, with the exception of considerations for potential growth and "overgrowth." Children with amputations secondary to sarcoma may be treated slightly different, as their limb volume will often fluctuate dramatically when they are undergoing chemotherapy and radiation therapy (59). The major concern for fitting the child with a septicemic cause of amputation is the resulting condition of skin and bone (71). The child will

FIGURE 13.24 College Park Industries TruPer Foot. Note that configuration of separated great toe on foot shell permits use of flip-flops or sandals.

most likely have experienced skin grafting procedures, and underlying bone will often progress at different rates than expected. These growth rates may be sporadic, delayed, or cause angulation deformities to occur (69).

FITTING TIMETABLE

The child with lower limb deficiency should be fitted with a prosthesis when he or she is ready to pull up to a standing position (52). This usually occurs between 9 and 10 months of age. The goals in fitting a prosthesis at this early age are to allow for normal two-legged standing, provide a means for reciprocating gait development, and provide a normal appearance. The prosthesis should be simple in design, allow growth adjustment, suspend securely, and be lightweight. Historically, at an early age, the transfemoral prosthesis should not utilize a knee joint due to the complexity of operating a free knee; however, this philosophy is being re-evaluated. Knee joints were usually added between 3 and 5 years of age, at times with a manual locking option (151). Knee units can be added initially if an extension assist on the knee is utilized to help bring the knee into full extension prior to loading. Either an endoskeletal or an exoskeletal construction may be employed; each has advantages and disadvantages (149). Endoskeletal construction is good for growth consideration and is generally durable enough in most settings. The foam cover of the endoskeletal design requires more maintenance than an exoskeletal finish. Many children and families choose to customize the socket with a design or character instead of using a standard traditional cover. Endoskeletal design allows for ease of height adjustments, greater patient acceptance, and decreased cost, as a cover replacement is not needed. The exoskeletal construction is robust and should be considered for those individuals who will test the limits of durability.

The child who acquires an amputation will be treated much the same as the congenital limb-deficient child, with a few exceptions. A child who undergoes an amputation will likely require a preparatory prosthesis while postoperative swelling subsides. The preparatory limb will probably be worn for approximately 3 months. If the eventual functional level is known, thought may be given to utilizing definitive componentry initially, and replacing the socket when the limb stabilizes. In the case of the child amputated at a transfemoral level who is undergoing chemotherapy treatment, a volume-adjustable socket incorporating a gel liner and lanyard suspension may accommodate for changes in size.

TRAINING

The preprosthetic period for the lower limb is mainly focused on addressing the information needs of the parents. In addition, an assessment of strength, coordination, joint ROM, skin condition, and sensation should be performed.

Each child must be assessed as an individual, with consideration given to the child's age (both developmentally and chronologically), physical abilities, interests, and activities. The goal of physical therapy is to develop a normal pattern of gait, including stride length, step length, and velocity. The normal child does not establish heel-to-toe gait until about 2 years of age. At about 20 months, the normal child can stand on one foot with help; at 3 years, on one foot momentarily; at 4 years, for several seconds; and at 5 years, for longer periods. Toddlers tend to stand and ambulate with a wide-based gait, with their lower extremities externally rotated, abducted, and flexed. As their gait matures, these characteristics change to a more narrow-based, upright fashion (152). The prosthesis should incorporate these same features in order to allow for normal gait development. Because the goal of physical therapy is symmetry of posture and movements during developmental activities, proper alignment, controlled weight shifting, and balance activities are emphasized for children with lower limb prostheses. Use of a polycentric knee unit (Figure 13.25) allows a more normal cadence. Kinematic studies demonstrate that cocontractions of the limb are reduced and may result in joint instability, so strengthening both agonists and antagonist muscles about the joint is important (153).

Functional goals for the child with bilateral lower extremity amputations should be optimistic. Functional outcomes measured for the child with a lower extremity prosthesis with the Pediatrics Outcomes Data Collection Instrument (PODCI) reflect excellent acceptance and use for both congenital and acquired amputations (154). As long as children have arms with which to balance, they should be expected to walk independently (155).

FIGURE 13.25 Polycentric knee units from Seattle Limb Systems and Hosmer-Fillauer.

FIGURE 13.26 This Cheetah series foot helps to fill in the gap for patients when they have reached adolescence and have a higher activity level and requirements but still do not fit into the size and weight categories of adult feet.

Step-in-place training is appropriate pregait training for children (151). Weight control is a concern for the child with lower extremity amputations. Dietary instruction should be emphasized early and often. Gait analysis has been performed on adults with amputations. Crutch walking, with or without a prosthesis, increases energy expenditure during gait. In groups of traumatic amputations, the oxygen cost progressively increases with each higher level amputation. Amputees preserve their energy expenditure by decreasing their chosen walking speed. Children's effort levels have been reported for transtibial amputations among crutch walking, Solid Ankle Cushion Heel (SACH) foot, and the Flex-Foot. The chosen walking speed was higher for the children using the Flex-Foot, approaching normal. This study only involved five children, so statistical significance could not be determined. A slightly higher oxygen consumption occurred for children using SACH feet (127). The Carbon Copy II prosthetic foot and Seattle Foot are energy-saving designs that permit the athlete a more natural gait. The energy-storing feet are available for children (Figure 13.26).

TRAINING FOLLOWING AMPUTATIONS

Following surgery, the remaining leg assumes the dominant role in all transfer and locomotor activities. Therefore, the sound leg should be evaluated for strength and, if necessary, an appropriate exercise program developed. Education regarding proper utilization of prosthetic devices decreases inappropriate stresses on the foot/ankle complex. It is difficult to instruct the young active child in specific exercises and positioning due to limited comprehension and attention span. If specific exercises are indicated, a therapist often needs to be creative with games and use of equipment to get the desired responses, such as using a prone scooter to maintain or work on hip extension (103). Edema control using Ace bandage wrapping, elastic shrinker socks, layers of elastic stockinettes, rigid dressings, or removable rigid dressings improves initial prosthetic fit tolerance and wear. Parental and child understanding of the proper utilization of these devices for edema control improves outcomes. With the exception of wound care and hygiene, the edema control system works maximally if worn for 24 hours of the day (156). Parents and children should be trained in desensitization and scar mobilization of the residual limb.

To avoid increasing the patient's anxiety level, the therapist should not dwell on phantom pain, but the patient should be made aware of the normal postoperative discomfort that is to be expected. The adaptation to prosthetic ambulation is dependent on the fit and comfort of the residual limb and socket/suspension. The therapist, working closely with the prosthetist at this point, can identify the fixable problems and ensure continued use for the child to gain confidence and competency.

Play is the primary motivation for desired movements and activities. Parents should be instructed on how to care for the prosthesis and encouraged to maintain contact with the prosthetist for routine adjustments and follow-up. Often, a change in wearing time and gait pattern, and limp are among the first signs that signify an evaluation of the prosthesis is warranted. Children and adolescents place high mechanical stresses on their prosthesis, which can result in catastrophic failure of the prosthesis. For children with an acquired limb difference, this prosthetic failure can rekindle stressful memories and cause feelings from the amputation trauma to resurface, so it is paramount to replace or repair the prosthesis as soon as possible.

Adolescents widen their sphere of mobility to include the community by using public transportation or by driving. The site of the amputation or limb loss will determine the degree of difficulty an amputee will have driving standard vehicles. In most cases, the person with a partial or full amputation of an upper limb requires adaptive driving equipment to compensate for the loss of ability to reach and operate standard driving controls. Most amputees are able to independently get into and out of a standard-size sedan. Driving aids are available for the driver who has normal strength and mobility of upper extremities. Control systems used include push–pull control, push–right angle pull control, and push twist. Each has the acceleration and braking system connected to usable upper extremity function. While most drivers with transtibial, and some with transfemoral amputation can drive without adaptations, providers and patients should be aware of different state requirements and liability issues related to licensure for driving, and equipment requirements vary. Physicians should be aware of their responsibility in certifying the capabilities of a potential driver. The evaluation for driving potential as well as specific equipment modifications should be discussed and made available for the individuals with multiple limb or complex limb deficiencies.

For children with amputations secondary to tumors, returning to school remains a difficulty. In a study concerning the adjustment post-tumor amputation, 67% could not keep up in their coursework (148,157). In addition to direct intervention for psychological support, support groups exist to aid families and children with limb deficiency. Many clinics provide opportunity for the interaction and peer support of their population. Frequently, the parent-to-parent or child-to-child interactions surpass the effect of professional input for education, information, and resources (117,157). Resource guides exist to provide pragmatic information for the child and parents (118,119). The Internet serves as a common place for information and allows parents and children to connect through the Amputee Coalition of America (www.amputee-coalition.org) and Amputee Empowerment Partners online community (www.EmpoweringAmputees.org).

Children with complex limb deficiency, such as tetraphocomelia, benefit from the early introduction of power mobility. Movement provides a sense of independence and competence derived from exploring one's environment. When unable to explore their environment, long-lasting negative impacts can exist. Motorized wheelchairs traditionally have been used when a child is 5 to 6 years of age; however, innovative seating systems have been developed for the 1- to 3-year-old child, consistent with developmentally appropriate expectations regarding independence with mobility. Salient features include the following:

- A powered device
- Proportional control drive with an adjustable joystick used with the head, chin, or lower or upper extremity buds
- Adjustable positioning seating in an upright frame into which inserts can be attached with growth potential
- Compactness, durability, portability, reliability, and safety
- Low profile with mounting potential for children to interact on a peer level, or height adjustable to accommodate floor play, facilitate transfers, and peer interaction

In addition to power mobility, other adapted mobility devices are available that are child- and environment-friendly. Ultimately, the child will gravitate to the device that provides the best function, despite the best ideas and intentions of the clinicians.

ADVANCEMENTS IN LOWER EXTREMITY PROSTHETICS

The improvement in a variety of prosthetic components designed for children augments the ability of a prosthetic team to improve outcomes. For years, few options existed for the pediatric population. SACH feet and friction knees came standard; however, increased awareness on the part of the parents of children with limb deficiencies and amputations, along with pressure from the rehabilitation community, influenced the manufacturers to recognize the need for improvements in this market. Children test the limits of many components by competing in recreational activities that range from neighborhood skateboarding to extremely competitive sports against their "able-bodied" peers. Hybrid dynamic response feet that are functional for everyday walking and running allow children to redefine what it means to have a limb difference. Children have benefited from the introduction of smaller components that provide improved responsiveness and control, and variable cadence, while also compensating for variable terrain.

Professionals involved with the population of children with loss of limb appreciate the possibilities that exist for an individual to compensate and accomplish as much as anyone else. Improved materials, technology, and greater availability of resources contribute to increasingly versatile prosthetic options. Involvement of a child and family with a comprehensive amputee clinic team provides therapeutic choices throughout the child's life, laying the groundwork to enter adulthood as well-adjusted, confident, and capable individuals. Close collaboration of physicians, family, and all professionals remains essential for a cohesive and practical rehabilitation program. As in all pediatric conditions, the process of decision making, implementing treatment options, and delivering care varies and should be discussed with the child and family, who are an integral part of the rehabilitation team.

THE AGING LIMB-DEFICIENT CHILD

As children with limb deficiencies age and mature physically, beneficial and detrimental changes can occur to the body in relation to the limb deficiency. For example, a child with transradial amputation who was not strong enough to use a voluntary-opening terminal device may eventually gain enough strength to use one in adolescence. The same transradial child who was successful with an externally powered prosthesis as a child during grade school may not be able to tolerate adult-size components secondary to increased weight and length of lever arm created by the prosthesis. As children grow longitudinally in affected limbs, occasionally the shortened limbs may result in the need for revision amputation more proximally, such as in the case of a bilateral below the knee amputee with shortened tibial segments that were able to be contained in a socket previously. After growth occurs in this instance, it may be too difficult to contain a patient in the traditional ways and prosthetic teams may need to be more creative with suspension or visit the idea of more proximal repeat amputation or disarticulation.

PEARLS AND PERILS

UPPER EXTREMITIES

1. The younger the child is at the time of acquired/traumatic amputation, the easier the transformation of hand dominance.
2. Children with high-level, bilateral upper limb loss may benefit from a prosthesis for ADLs. Since limited body movements are available, the child may benefit from at least one hybrid or completely externally powered prosthesis. Prior to the consideration of prosthetic fitting, it is paramount that the child and family begin exploring the use of the child's lower limbs, as independence can also be achieved with feet.
3. At birth, the severely deformed upper extremity often detracts from the identification of a more important systemic workup. Although there may not be any other underlying etiologies or comorbidities, it is essential for the clinic team to explore these possibilities (Figure 13.27).

FIGURE 13.27 This child with bilateral longitudinal radial limb deficiencies represents a visible portion of a possible syndrome of findings that could occur. Evaluation of this child should include screening for cardiac, hematologic, and renal abnormalities.

4. It is important to teach parents about the loss of surface area corresponding to the absent limb. Active children with multiple limb loss have a reduced surface to radiate heat loss, so they may have an increase in sweating and flushing about the head and neck.

LOWER EXTREMITIES

1. Children tend to do well with lower limb prostheses, often requiring little or no formal gait training but will still benefit from therapy to maximize balance and address any muscle imbalances.
2. Limb-volume changes occur following amputation and can be controlled by rigid dressings in the postoperative period (156). Although the postoperative edema is not as great as that for the adult dysvascular patient, children will benefit from these rigid dressings for the control of edema as well as to initiate earlier ambulation and prosthetic fitting. A rigid removable dressing is illustrated in Figure 13.28. This is particularly important for children who have had a remnant or dysfunctional limb segment for which they have some psychological attachment. Early ambulation may serve as a distraction to the surgery and provide a new focus on skills of ambulation.

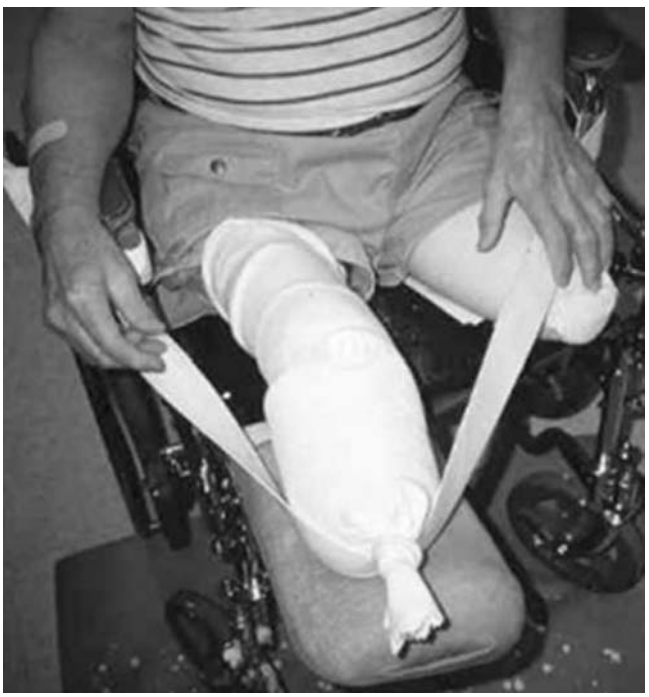

FIGURE 13.28 Individual with transtibial amputation and removable, rigid dressing. Note strap used to maintain compression on limb.

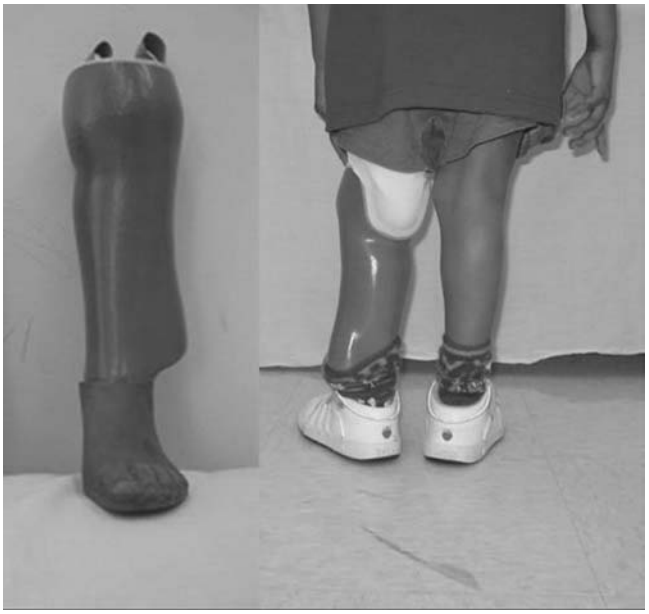

FIGURE 13.29 Angulation deformity accommodated by prosthesis.

3. When fitting an ankle disarticulation prosthesis, the prosthetists should strive to create a prosthesis that permits near full weight-bearing on the distal end of the child's residuum. This will ensure that the distal end of the residuum and the heel pad remain toughened. An end-bearing residuum of this length is beneficial for limited ambulation without the prosthesis (eg, using the bathroom in the middle of the night), as well as long-term fitting.
4. The surface area of the distal residuum will remain fairly consistent throughout the child's life; therefore, the child is encouraged to maintain a reasonable weight, so as not to lose the ability for distal end bearing.
5. Prosthetic fittings may be affected by angular deformities as the alignment of the device must be biomechanically appropriate and not necessarily the most cosmetic. Most of the angulation deformities can be accommodated in a prosthesis; however, it is not possible to provide a device that is advantageous to appropriate gait mechanics and satisfies the cosmetic expectations. The accommodation of angular deformity is illustrated in Figure 13.29.
6. The utilization of flexible inner liners in upper and lower extremity prosthesis can allow for increased life expectancy of a prosthesis by accommodating growth through removal or remolding.

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14

CEREBRAL PALSY

Mary McMahon, David Pruitt, and Jilda Vargus-Adams

Cerebral palsy (CP) is defined as "a group of permanent disorders of the development of movement and posture, causing activity limitations that are attributed to nonprogressive disturbances that occurred in the developing fetal or infant brain." (1). There are three major criteria for diagnosis of CP: a neuromotor control deficit that alters movement or posture, a static brain lesion, and acquisition of the brain injury either before birth or in the first years of life. Due to the breadth of these criteria, CP is an extremely heterogeneous diagnosis in terms of clinical presentation, etiology, and pathology. Although the brain lesions that result in CP are not progressive, the clinical picture of CP may change with time as the affected individual grows and develops.

EPIDEMIOLOGY AND RISK FACTORS

CP is the most common motor disability of childhood, affecting approximately 3.1 per 1,000 school-age children (2) with at least 8,000 new cases each year in the United States (3). The population of children with CP may be increasing due to premature infants who are surviving in greater numbers (4), higher incidence in normal-weight term infants (3), and longer survival overall. The proportion of CP that is most severe is also increasing, with as much as a third of all children with CP having both severe motor impairments and intellectual disability (5).

The etiology of CP is often not well understood. The majority of cases in term infants do not have a clear etiology, although some risk factors have been identified including placental abruption, birth asphyxia, and neonatal medical problems (6). Factors that may contribute to brain injury and CP include prematurity, infection, inflammation, and coagulopathy (7). There is also considerable interest in the contributory roles of various biomolecules and cytokines that accompany infectious or inflammatory processes (8). Risks such as consanguinity, Rhesus incompatibility, vaccine-preventable infections, and iodine insufficiency are more common in the developing world (9).

The greatest risk factor for the development of CP is prematurity. Premature infants (born earlier than 37 weeks' gestation) are much more likely to develop the condition than term infants, and incidence rates are highest in the very earliest infants (9,10). Rates of CP in premature and low birth-weight infants vary from 40 to 150 per 1,000 live births (11), with some reports suggesting increasing (4) or decreasing rates (11,12) in the past two decades. Figure 14.1 (10) demonstrates the roles of prematurity and birth weight in CP. The vertical axis shows the incidence rates of CP for groups of varying gestational age. Higher rates, approaching 1 in 10 live births, are evident for premature birth of less than 32 weeks' gestation age whereas term births show rates closer to 1 in 1,000 live births. The horizontal axis shows variation from standard or preferred birth weights as determined by gestation age and represented by z scores. The effect of low (or high) birth weight is demonstrated with the "U"-shaped curves of CP incidence peaks across most gestational ages. This figure demonstrates the profound effect of prematurity as a risk factor for CP.

Prenatal risk factors for CP include being small for gestational age (13), being of low or very low birth weight (14), multiple gestation (15), developing infection (especially chorioamnionitis and cytomegalovirus) (16), having evidence of stroke (17), or having neonatal encephalopathy (18). Maternal risk factors for CP include chorioamnionitis (16) or fever during labor, coagulopathy or bleeding (19), placental infarction, and thyroid disease (20). Postnatal risk factors for CP are often related to social disadvantage, and include trauma in developed nations (21) and infection in developing nations (22). Additional risk factors for CP include kernicterus (23), methyl mercury exposure (24), and genetic causes (25).

Severe birth asphyxia in term infants is not a major cause of CP. A small minority of children with the condition had asphyxia (6,26), in contrast to prematurity, which is associated with up to half of all cases of CP. Nonetheless, for children who have true birth asphyxia, the risk of CP is increased (27). Fetal monitoring in the United States has probably increased the rate of cesarean section deliveries, but has not been associated with any decline

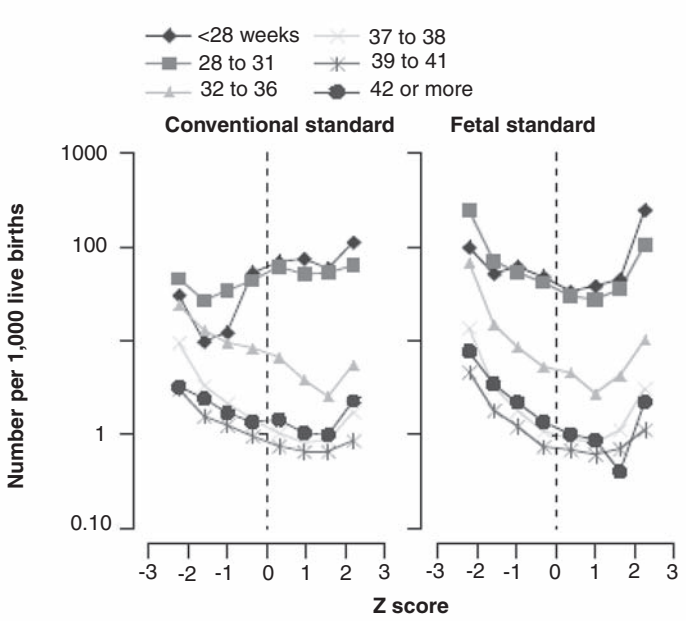

FIGURE 14.1 Prevalence of CP by z score of weight for gestation.

Source: Reprinted with permission from Ref. (10). Jarvis S, Glinianaia SV, Torrioli MG, et al. Cerebral palsy and intrauterine growth in single births: European collaborative study. *Lancet*. 2003;62:1106–1111.

in rates of CP (28). Term infants described as having birth asphyxia often manifest certain signs, including acidosis, bradycardia, or neonatal encephalopathy. Intrauterine exposure to infection or a coagulation disorder can cause a similar clinical picture at birth and may be mistaken for complications of birth asphyxia. Neonatal encephalopathy generally is diagnosed in neonates with significant neurologic dysfunction, including respiratory difficulties, altered tone, low consciousness, and/or seizure activity. It is the best predictor of CP in term infants, regardless of the cause of the encephalopathy.

CLASSIFICATION

CP has traditionally been classified by type of movement disorder and anatomic distribution; see Figure 14.2 (29).

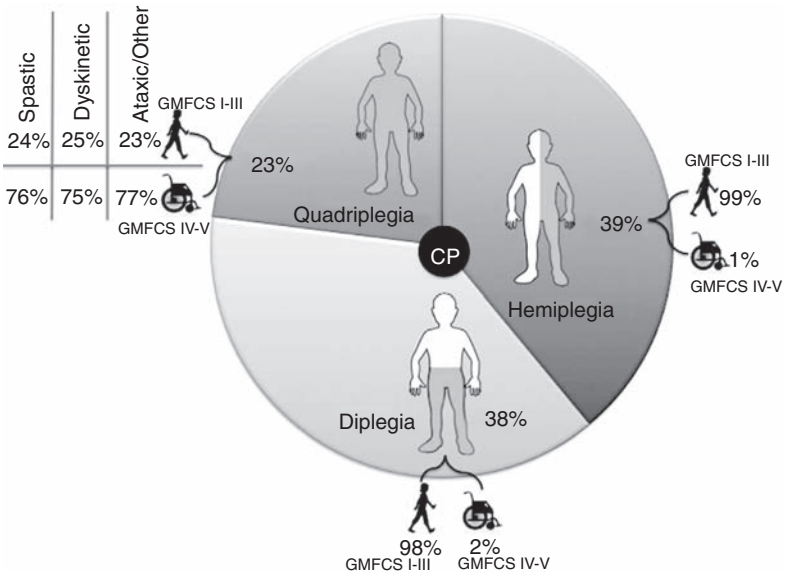

FIGURE 14.2 Proportion of CP by topography and severity.

Source: Reprinted with permission from Ref. (29). Novak I. Evidence-based diagnosis, health care, and rehabilitation for children with cerebral palsy. *J Child Neurol*. 2014;29:1141–1156.

FIGURE 14.3 A child with dystonic CP.

Movement patterns include spastic, dyskinetic, hypotonic, ataxic, and mixed forms. The most common movement pattern is spastic including 77% in the United States (2) with a minority of cases being primarily dyskinetic, ataxic, or hypotonic (30). The distinction between spasticity and dystonia is not always clear. An interdisciplinary group developed a consensus statement on the definition of each term. Spasticity was defined as hypertonia in which one or both of the following signs are present: (a) resistance to externally imposed movement increases with increasing speed of stretch and varies with the direction of joint movement, and/or (b) resistance to externally imposed movement rising rapidly above a threshold speed or joint angle (31). Dystonia was defined as a movement disorder in which involuntary sustained or intermittent muscle contractions cause twisting and repetitive movements, abnormal postures, or both (Figure 14.3) (31). Hypotonic and ataxic forms of CP are rare and, therefore, any child suspected of having either of these diagnoses should receive a thorough diagnostic evaluation for other neurologic conditions.

The anatomic distribution of motor problems in CP is the primary means of classification. The three categories of hemiparesis, diparesis, and quadriparesis occur with fairly equal frequency (5,30). Hemiparetic CP affects only one side of the body and typically demonstrates greater impairments in the upper extremity (UE; Figure 14.4). Diparetic CP affects the lower extremities more than the upper extremities (Figure 14.5). Quadriparetic CP affects the entire body, including the axial as well as appendicular skeleton (Figure 14.6).

An interest in classifying children with CP based on function in addition to the distribution of motor impairment resulted in the development of the Gross Motor Function Classification System (GMFCS). The GMFCS

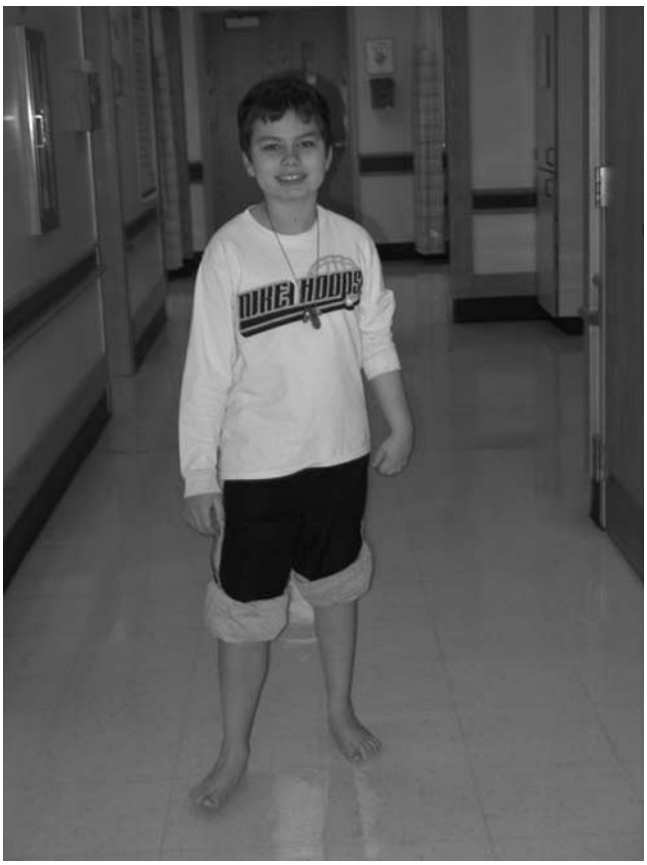

FIGURE 14.4 A child with hemiparetic CP.

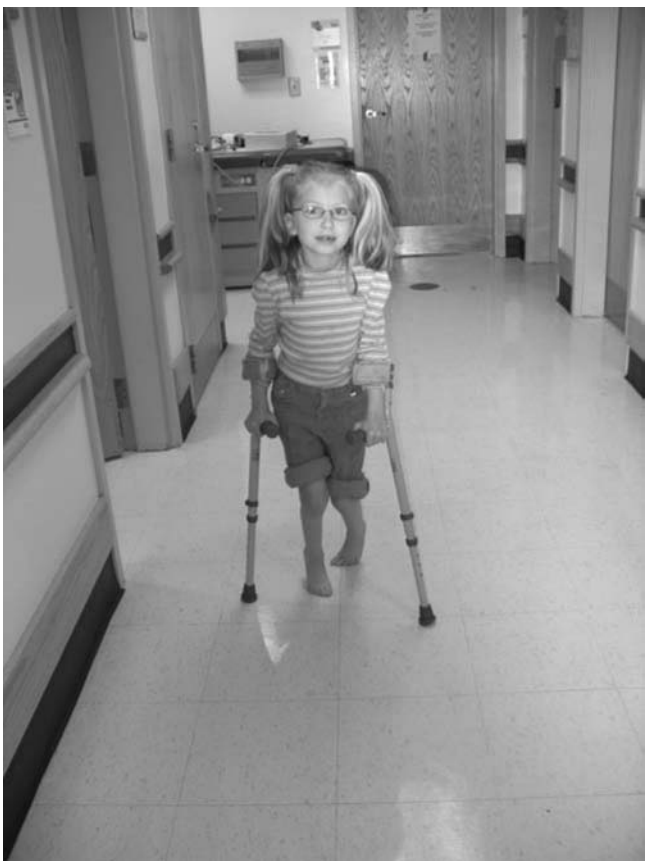

FIGURE 14.5 A child with diparetic CP.

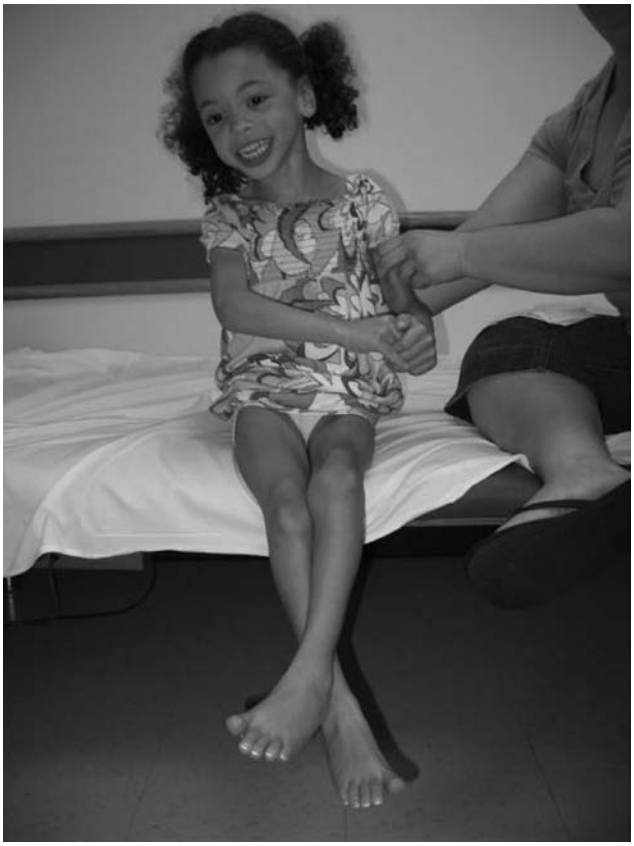

FIGURE 14.6 A child with quadriparetic CP.

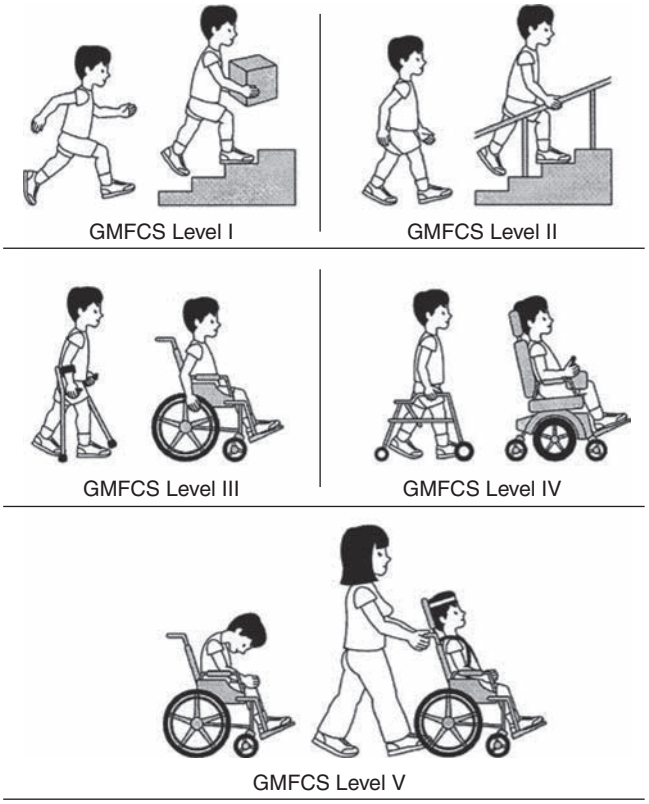

FIGURE 14.7 The Gross Motor Classification System for children aged 6 to 12 years.

Source: Reprinted with permission from Graham HK. Classifying cerebral palsy. *J Pediatric Orthop*. 2005;25:128.

stratifies children with CP into five groups based on gross motor skills (32); see Figure 14.7. In this system, specific descriptions of mobility functions, based on age, allow each child with CP to be categorized. In GMFCS I, children walk indoors and outdoors and climb stairs without limitation. Children who are GMFCS II walk indoors and outdoors and climb stairs holding onto a railing but experience limitations walking on uneven surfaces and inclines. Children who are GMFCS III walk indoors or outdoors on a level surface with an assistive mobility device. Children may climb stairs with a railing or propel a manual wheelchair. Children who are GMFCS IV may walk short distances with a device, but rely more on wheeled mobility at home and in the community. Children at GMFCS V have no means of independent mobility. A related classification system for UE function, the Manual Abilities Classification System, permits categorization by fine motor performance (33), and the Communication Function Classification System applies to language and communication, including cognitive and motor performance (34).

PATHOLOGY

More than 80% of children with CP will have abnormal findings on neuroimaging (35–37). These abnormal findings can provide valuable clues to pathogenesis.

The most common abnormality on neuroimaging is found in the white matter near the lateral ventricles and is called white matter disease of prematurity. Findings may include various lesions, the most significant of which is periventricular leukomalacia (PVL) (38), with reports of up to 56% of all cases of CP demonstrating abnormalities in this location (37) (Figure 14.8). PVL occurs much more commonly in premature infants than in term infants (90% vs 20%), often consistent with an insult in the second trimester, and is a common outcome of intraventricular hemorrhage in premature infants (37). Because the corticospinal tract fibers to the lower extremities are medial to those of the upper extremities in the periventricular white matter, children with PVL typically have spastic diparesis. One large study found that PVL was present in 71% of the children with diparesis, 34% of those with hemiparesis, and 35% of those with quadriparesis (35).

Deep gray matter lesions to the basal ganglia and thalamic region are mainly associated with dystonic CP, and have been found in approximately 12% of children with the condition (35). Historically, large numbers of children acquired athetoid CP following a diagnosis of kernicterus, due to concentrated damage to the basal ganglia with bilirubin encephalopathy. These cases are

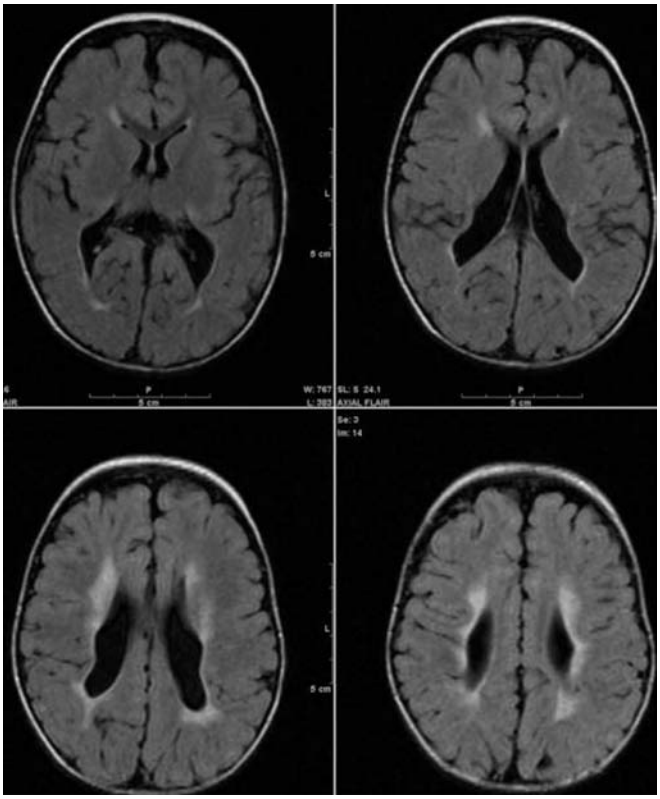

FIGURE 14.8 Periventricular leukomalacia.

far less common with advancements in the treatment of neonatal jaundice.

Focal cortical infarcts involving both the gray and white matter are found almost exclusively in patients with hemiparesis, and are typically related to middle cerebral artery strokes. In a group of children with hemiparetic CP, 27% were found to have a focal infarct on imaging (35).

Brain malformations can be found on neuroimaging in approximately 10% of children with CP (35–37,39). Neuronal migration disorders or insults early in pregnancy can result in lissencephaly, polymicrogyria, schizencephaly, or holoprosencephaly. Some in utero infections, such as those caused by cytomegalovirus, can also cause distinctive brain malformations (35). Brain malformations are more commonly found in cases of term infants and hemiparesis (36).

Children who sustain diffuse brain insults demonstrate more extensive injury on neuroimaging. Infection and ischemia are two of the more common causes of generalized encephalomalacia. A wide range of findings may be present on MRI, including multiple cysts, cortical thinning, white and gray matter damage or loss, and microcephaly. Children with diffuse brain lesions or anomalies typically demonstrate spastic quadriparesis and are at high risk for additional medical and cognitive problems.

INITIAL EVALUATION AND CLINICAL FINDINGS

SIGNS AND SYMPTOMS

Early identification of children who have CP allows for early therapeutic intervention and screening for associated conditions. Because CP is a descriptive term that does not infer a single etiology, pathology, or prognosis, there is no specific diagnostic test. It is a diagnosis of exclusion based on a careful history and physical exam. It can be difficult to make a definitive diagnosis in infants less than 6 months old. Prior to this time, the infant has a limited repertoire of volitional movements, which makes milder delays in motor development difficult to detect. In addition, abnormalities in tone and reflexes are often subtle in early infancy. As the cortex matures in the second half of the first year, the diagnosis typically becomes more apparent.

The first step in the evaluation for suspected CP is a comprehensive history, including a detailed account of potential risk factors and family history. A thorough history of developmental milestones is also important. Often the parent's initial concern is a significant delay in attaining motor milestones. Prematurity must be considered when evaluating development because milestones are generally corrected for the degree of prematurity. A discrepancy between motor and cognitive milestones should always raise suspicion for CP. Certain deviations in developmental milestones are associated with CP. For example, early hand preference or asymmetric use of the extremities may be the first indication of hemiparesis. Early head control, rolling, or rigid standing are all associated with abnormally increased tone and/or exaggerated primitive reflexes. The parent may also describe unusual means of mobility, such as bunny hopping, combat crawling, or bottom scooting. The most important aspect of the developmental history is to confirm that the child has not lost any skills or milestones, as this would suggest a neurodegenerative disorder.

Following a detailed history, a thorough physical examination should be performed. A careful neurologic exam is an essential piece of the evaluation. In infancy, the neurologic exam focuses on tone and infantile developmental reflexes. Deep tendon reflexes, plantar responses, and the presence of clonus are more informative in the older child. Tone should be assessed by gently moving the infant's joints through their appropriate range of motion (ROM) and evaluating the amount of resistance. Careful observation will also provide information about an infant's tone. Infants with severe hypotonia will lay in a frog-leg position with their hips abducted, flexed, and externally rotated. Their arms will lie limply at their sides. Persistent fisting or scissoring may be observed with increased tone. Most infants will undergo an early stage of mild or moderate hypotonia prior to more traditional

signs of CP. A prolonged period of hypotonia or fluctuating tone is more typical of dyskinetic CP. In general, however, longer periods of hypotonia and severe hypotonia are associated with more severe motor deficits, regardless of the type of CP.

The earliest indication of CP may be a delay in the disappearance of primitive infantile reflexes. Commonly examined primitive reflexes include the Moro reflex, palmar grasp reflex, asymmetric tonic neck reflex, and tonic labyrinthine reflex. During the first 6 months of life, maturation of the cortex gradually overrides these primitive responses, and voluntary motor activity should increase. Persistence of these primitive reflexes past 6 months of age, asymmetry of the response, or an obligatory response at any age should be considered highly suspicious for a significant motor impairment. As the primitive reflexes become suppressed, postural or protective reactions such as the parachute and the equilibrium or tilting reactions should emerge. In children with CP, postural reactions may be less effective, appear later than usual, or fail to develop.

A general movement assessment (GMA) can also be useful in evaluating a child for CP. A systematic review of tests, including GMA, neurologic examination, MRI, and cranial ultrasound, found that the GMA had the best evidence and strength for predictive accuracy in high-risk infants (40). GMA involves observation of the infant's spontaneous movements. Abnormal general movements in a young infant are described in the following three categories: (a) poor repertoire: monotonous movement sequences, (b) cramped synchronized: muscles contract and relax almost simultaneously, resulting in the movements being rigid, (c) chaotic: large-amplitude movements of all limbs that lack fluency or smoothness (41). At 3 months' corrected age, general movements of a fidgety nature are typical. Normal fidgety movements included small movements of moderate speed and variable acceleration of neck, trunk, and limbs in all directions that are continual in the awake infant except during fussing and crying. Abnormal fidgety movements are of larger amplitude with moderately exaggerated speed and jerkiness (41). A systemic review found that the pooled sensitivity and specificity for GMA in predicting CP were 98% and 91% respectively (40). Formal assessment tools, such as the Developmental Assessment of Young Children (42) and the Hammersmith Infant Neurological Evaluation (43) have also been shown to be highly predictive of CP in infants less than 12 months of age.

IMAGING

Neuroimaging can be helpful in determining the etiology of CP and the timing of the insult. The Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society published two practice parameters that address the use of neuroimaging in the neonate and the child with suspected CP (44,45) (Figure 14.9). Recommendations for imaging in the preterm neonate include a screening cranial ultrasonography on all infants with less than 30 weeks' gestation, between 7 and 14 days of age, and again between 36 and 40 weeks' postmenstrual age (44). This recommendation was based, in part, on the fact that a 10-fold elevation in the risk of adverse outcome for the very low birth-weight infant was identified with ultrasound evidence of grade 3 or 4 intraventricular hemorrhage, periventricular cystic lesions, or moderate to severe ventriculomegaly (44).

In term infants with neonatal encephalopathy, the practice parameter recommends a noncontrast computed tomography (CT) to detect hemorrhagic lesions when there is a history of birth trauma, low hematocrit, or coagulopathy (44). If the CT is inconclusive, MRI should be performed between days of life 2 and 8 in order to assess the location and extent of injury. Abnormalities of the thalamus and basal ganglia were associated with increased neurodevelopmental disability at 1 to 2 years of age (44).

Neuroimaging can also be useful in determining an etiology in children suspected of having CP outside of the neonatal period. The practice parameter on the diagnostic assessment of the child with CP found an abnormal MRI scan in the majority of children with CP (average 89%) and that MRI was more likely to show an abnormality when compared to CT (average 77%) (45). The practice parameter, therefore, recommends neuroimaging in the evaluation of a child with CP if the etiology has not been established, and MRI is preferred to CT.

LABORATORY FINDINGS

Metabolic or genetic causes for CP are unusual, and laboratory studies to investigate these conditions are not routinely recommended. Metabolic or genetic testing is recommended in the following conditions: if neuroimaging does not determine a specific structural abnormality or if it reveals a developmental malformation, if there is evidence of developmental deterioration, or if there is a family history of a childhood neurologic disorder associated with a diagnosis of "cerebral palsy" (45). The practice parameter also recommends consideration of diagnostic testing for a coagulation disorder in children with an unexplained cerebral infarction on neuroimaging (see Figure 14.9).

DIFFERENTIAL DIAGNOSIS

Young infants with CP often present with hypotonia. The differential diagnosis for the floppy infant is vast. The most common etiologies include central nervous system disorders such as CP, neuromuscular disorders, genetic

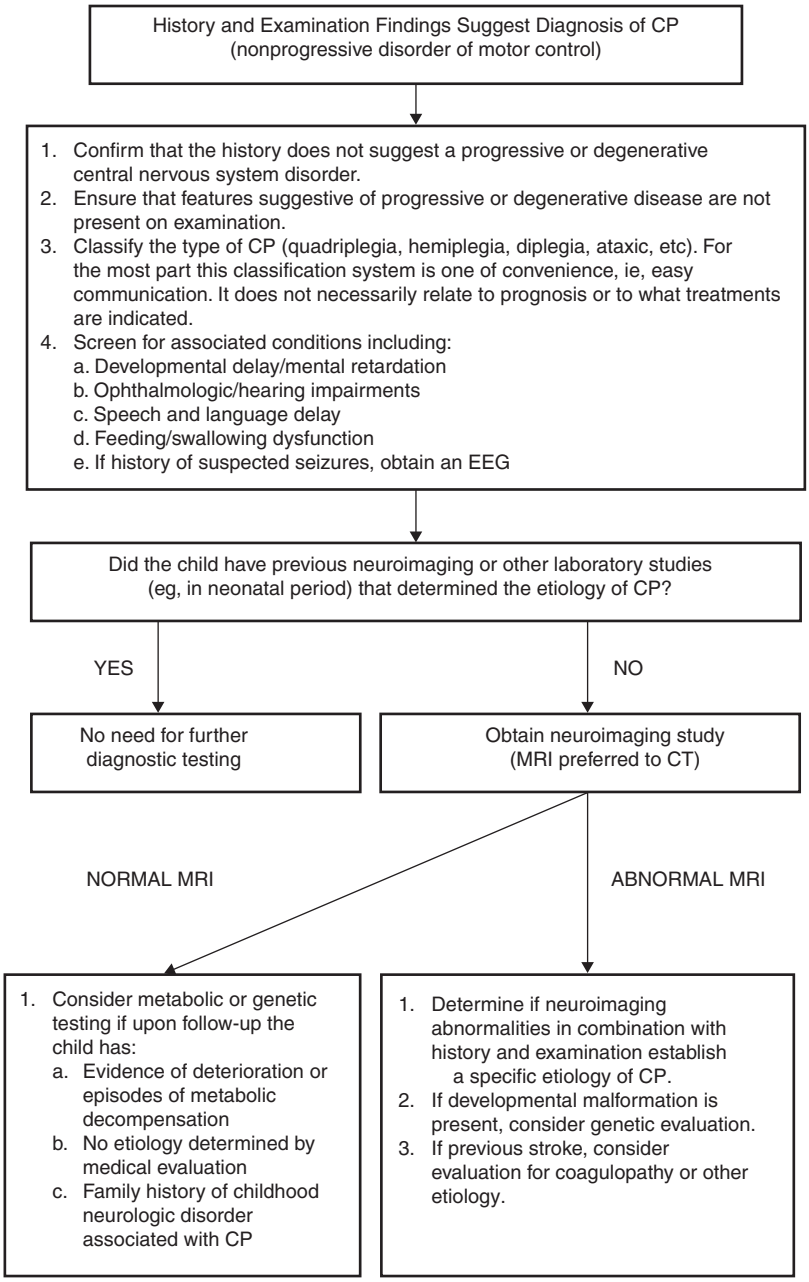

FIGURE 14.9 Algorithm for the evaluation of the child with CP.

Source: Reprinted with permission from Aswal S, et al. Practice parameter: Diagnostic assessment of the child with cerebral palsy. Report of the Quality Standards Subcommittee of the American Academy of Neurology and the Practice Committee of the Child Neurology Society. *Neurology*. 2004;62:851–863.

disorders, and metabolic disorders. Clues to a neuromuscular disorder include diminished deep tendon reflexes, weakness (which may result in absent infantile reflexes), or a positive family history. Dysmorphic features may suggest a genetic cause for hypotonia, such as Down syndrome, Prader–Willi syndrome, or Angelman syndrome. Metabolic disorders may present at any age, but are most likely to present in infancy. Metabolic disorders should be considered if a previously healthy child presents with an acute encephalopathy without an adequate explanation. Metabolic acidosis, hypoglycemia, hepatic involvement, or cardiac involvement should also prompt consideration of a metabolic disease. Dystonia and spasticity are present in a number of metabolic disorders, including mitochondrial disorders, glutaric aciduria type I, Lesch–Nyhan syndrome, and homocystinuria. A diagnosis other than CP should always be sought in children who have evidence of progressive disease or loss of previously obtained milestones.

A definitive diagnosis of CP should be made cautiously, especially in the first 6 months of life. Infants who are suspected of having CP should be followed closely

with serial developmental evaluations and physical exams until the diagnosis is clear. An early diagnosis is considered best practice, however, because it will allow access to diagnosis-specific early intervention and parental support (29).

ASSOCIATED DISORDERS

SENSORY IMPAIRMENTS

CP is defined as a disorder of movement control and posture, and therefore sensory impairments are easily overlooked. Deficits in two-point discrimination, proprioception, and stereognosis have been described (46–48). Sensory deficits are believed to be most common in children with hemiparesis. A study of children with spastic hemiparesis found that 97% of the spastic limbs had a stereognosis deficit, 90% had a two-point discrimination deficit, and 46% had a proprioception deficit, and these sensory deficits were more commonly present in limbs with a greater size discrepancy (48). Sensory deficits can also be found in the limbs that do not appear to be affected by CP (49). Bilateral sensory deficits were found in 88.8% of children with hemiparesis in one study (46). Stereognosis and proprioception deficits were the most common bilateral abnormalities, and the extent of sensory loss did not mirror the motor deficit. Another study identified abnormalities of tactile spatial discrimination in the hands of children with spastic diparesis with apparent normal motor function in their upper extremities (47). Little is known about the significance of right- or left-sided brain injury on sensory deficits in CP. A trend toward children with left hemiparesis to perform worse on spatial-tactile tests has been reported (49). Beyond the brain injury itself, it has been hypothesized that a reduced number of perceptual motor experiences throughout development may contribute to the reorganization of the sensorimotor cortex (50). Sensory deficits are important to recognize because they can significantly affect functional use of the extremity.

VISUAL IMPAIRMENTS

Visual impairments are common in children with CP, with a reported prevalence of 39% to 100% (51). The inherent difficulty in doing an ophthalmologic exam on children with varying degrees of cognitive and motor impairments makes it difficult to determine the precise incidence of visual disorders. Strabismus is the most commonly reported visual disorder, but a wide variety of other disorders have been described. There is emerging evidence that each clinical type of CP may be associated with a distinct neuro-ophthalmologic profile. In a study of 129 patients with CP who underwent an extensive evaluation of the visual system, investigators found that visual dysfunction in diplegia was characterized mainly by refractive errors, strabismus, abnormal saccadic movements, and reduced visual acuity (52). The participants with hemiplegia showed strabismus and refractive errors while oculomotor involvement was less frequent. Altered visual field was most common in subjects with hemiplegia. Children with tetraplegia had the highest rate of visual impairment, characterized by ocular abnormalities, oculomotor dysfunction, and reduced visual acuity (52). Another study demonstrated a relationship between visual deficits and severity of CP as measured by the GMFCS (53). In this study, children in each level of the GMFCS had visual deficits 10- to 70-fold higher than those reported in the general age-matched pediatric population. Children with milder CP, GMFCS levels I to II, had visual deficits that resembled neurologically normal children with strabismus and amblyopia. Children with the most severe CP were at greatest risk for high myopia, absence of binocular fusion, dyskinetic strabismus, severe gaze dysfunction, and optic neuropathy or cortical visual impairment (53). Visual impairments likely play a pivotal role in motor and cognitive function in children with CP (52). Early assessment and accurate detection of specific disorders is an important aspect of the rehabilitation care and overall quality of life (QOL) of these children.

HEARING IMPAIRMENTS

Considerable variation exists in estimates of hearing loss in CP, in part due to the difficulty in assessing hearing, as well as the population studied. A systematic review of population-based data on children with CP revealed a range of reported hearing loss of 4% to 13% and a range of severe hearing loss of 2% to 12% (54). The proportion of severe/profound hearing loss appears to be increasing in CP and this is felt to be related to the increasing number of survivors of extremely preterm birth or better detection (54). Sensorineural hearing loss is most commonly associated with congenital toxoplasmosis, rubella, cytomegalovirus, and herpes (TORCH) infections, bacterial meningitis, hypoxia, and ototoxic drugs. In the past, kernicterus was a relatively common cause of sensorineural hearing loss in athetoid CP. Hearing loss can significantly impact the development of communication and cognition and therefore early assessment and accurate detection are recommended.

COGNITIVE IMPAIRMENTS

Cognitive impairments are common in CP. It is difficult to make generalizations about the specific relationship of CP and cognitive function because CP is a heterogeneous disorder and the available literature often does not differentiate between the various types. In addition,

assessment of intellectual functioning can be difficult in patients with severe motor and communication difficulties, which may lead to an underestimation of cognitive function. An overestimation of cognitive function can occur in patients who are socially responsive. The overall frequency of an IQ score of 69 or below is reported to be 50% to 70% (55). In general, patients with more severe neuromuscular impairments are at greater risk for cognitive impairments, but some patients with severe motor impairments can have normal cognition. For example, a patient with athetosis secondary to a discrete lesion in the basal ganglion is likely to have normal intelligence.

Recent research has suggested that disorders of higher level cognitive tasks and executive functioning (EF) are also common in CP (56–58). Even children with mild unilateral CP can have significant EF impairments in comparison to typically developing children (56). A systematic review of brain structure and EF in CP found that parieto-occipital periventricular hemorrhage, intraventricular hemorrhage, ventricular dilatation, white matter reduction and bilateral lesions were associated with greater EF impairments (59), but in general there is a paucity of brain imaging studies focused on EF in children with CP. It is important to identify EF dysfunction early on due to its effect on academic and vocational success, as well as social integration. Therefore a systematic neuropsychological examination with particular attention to EF should be obtained in children with CP (60).

EMOTIONAL AND BEHAVIORAL IMPAIRMENTS

The prevalence of emotional and behavioral problems in different populations of children with CP is reportedly 30% to 80% (61) but in general, it has not been well defined in the literature. A wide variety of behavioral and emotional disorders are possible, including attention deficit disorder, passivity, immaturity, anger, sadness, impulsivity, emotional lability, low self-esteem, and anxiety. A population-based analysis of behavior problems in children with CP identified problem behaviors in 25% of the children as assessed by parent reports (61). Specific behaviors that were most common in this population included dependency, being headstrong, and hyperactivity. An additional population-based study in Europe found a similar prevalence of significant emotional and behavioral symptoms in 26% of children with CP (62). The most common problems identified were in peer relationships (32%), hyperactivity (31%), and emotion (29%). Difficulty with peer relationships has been found even in children with milder CP (GMFCS I). Compared to their classmates, children with mild CP were found to have fewer reciprocated friendships, fewer sociable and leadership behaviors, and were more isolated and victimized by their classmates (63). A potentially important contributing factor to impaired behavioral and social functioning is decreased cognitive EF (64).

Although, there is widespread agreement that emotional and behavioral dysfunction is common in children with CP, there is very limited research on effective interventions to address the issue. There is emerging evidence that parenting interventions and cognitive behavioral therapy, such as the Stepping Stones Triple P and Acceptance and Commitment Therapy, may be beneficial (65). An early awareness of the risk for emotional and behavioral disorders by professionals and parents is important, so that they can be recognized early. Referral to a mental health specialist should be considered, as well as a neuropsychological evaluation, including EF.

EPILEPSY

The overall occurrence of epilepsy is reported to be between 15% and 55% in a mixed population of children and adults with CP (66). A wide variety of types of seizures are possible, and a clear correlation between various risk factors and seizure frequency or type has yet to be established. Seizures are more common in children with more severe CP and in children with quadriparesis and hemiparesis versus diparesis (67).

OROMOTOR IMPAIRMENTS

Oromotor impairments are associated with more severe CP. A weak suck, poor coordination of the swallowing mechanism, tongue thrusting, and a tonic bite reflex may all lead to feeding difficulties and increased risk for aspiration. Speech disorders range from mild articulation disorders to anarthria, and are most commonly seen in children with spastic quadriparesis or athetosis. Oromotor dysfunction may also lead to difficulty controlling oral secretions and drooling, which may negatively affect social interactions. A wide range of interventions exist to address drooling, but a recent Cochrane review concluded that there is insufficient evidence to inform clinical practice (68). Oromotor impairments are also associated with dental malocclusion and difficulty with oral hygiene, leading to an increased risk of periodontal disease.

NUTRITIONAL DISORDERS

The assessment of growth and nutrition in children with CP can be difficult due to the lack of a reliable means of measuring stature in children with contractures and scoliosis and the lack of appropriate reference data or growth curves specific to CP (69). Population-based growth patterns of CP have been published (70), but they probably include many children with conditions affecting growth and feeding, and therefore should not be considered prescriptive of how children with CP should grow (69).

Poor oromotor skills, gastroesophageal reflux, and the inability to self-feed or communicate hunger can all increase the risk for malnutrition in children with CP. The North American Growth in Cerebral Palsy Project (NAGCPP) is a population-based study that identified the presence of feeding problems in 58% of children with moderate to severe CP (71). In addition, children with a pattern of severe feeding dysfunction were described as having the greatest risk for poor nutritional status and health, but even those with only mild feeding dysfunction were identified as being at risk for poor nutritional status. Subjects who were enterally fed were taller and had greater body fat stores when compared to subjects with similar motor impairments who were exclusively fed by mouth (72). Data from the NAGCPP also revealed that children with the best growth had better health and social participation (71).

For children who are unable to obtain adequate nutrition by mouth, gastrostomy or jejunostomy tubes are often utilized. There is evidence that this results in improved weight gain, but there is the potential for adverse events including overfeeding, site infection, stomach ulcer and reflux (73). A recent Cochrane review of gastrostomy feeding versus oral feeding alone in children with CP did not reveal a single randomized controlled trial (RCT) on this topic (74). Given the lack of evidence to guide clinical practice, the decision on the most appropriate means of nutritional support should be made in the context of each individual patient and family.

Although malnutrition is a primary concern, children with CP are also at risk for overfeeding and obesity. Children with more severe CP have lower total energy expenditure and higher body fat content than age- and sex-matched children without disabilities, placing them at risk for overfeeding with energy-dense enteral feeds (75). A study of ambulatory children with CP showed an increase in the prevalence of obesity from 7.7% to 16.5% over a 10-year period, an increase similar to that seen in the general pediatric population in the United States (76).

GENITOURINARY DISORDERS

The development of urinary continence is typically delayed in children with CP. A study of 601 children with CP found that by the age of 6, 54% of children with spastic quadriparesis and 80% with spastic hemiparesis or diparesis had gained urinary continence spontaneously (77). The most important factors associated with urinary incontinence were quadriparesis and impaired cognition. Incontinence was the most common complaint, but frequency, urgency, hesitancy, and urinary retention may also be present (77). Frequency and urgency are often associated with spasticity of the detrusor muscle, causing small, frequent voids. Detrusor over activity and a small bladder capacity were the most common findings on urodynamic studies in children referred for voiding dysfunction, but a minority of children were also found to have detrusor sphincter dyssynergia (78,79).

The association between lower urinary tract dysfunction and upper urinary tract dysfunction in CP is unclear. In general, it is reported to be uncommon (80). Symptoms of detrusor sphincter dyssynergia (interrupted voiding, urinary retention, and hesitancy) and culture proven febrile urinary tract infections were shown to correlate with upper tract dysfunction in one study (81). Urodynamic studies should be considered to evaluate the physiology of the bladder in patients with lower tract symptoms. The results can aid in promoting continence, but it is also important to assess basic communication, mobility, equipment and environmental supports, which have also been shown to be important components to obtaining continence (82).

RESPIRATORY DISORDERS

Children with CP are at increased risk for respiratory illnesses. Impaired control of respiratory muscles, ineffective cough, aspiration due to impaired swallowing, gastroesophageal reflux, and seizures all increase the risk for chronically increased airway secretions. Increased airway secretions may lead to wheezing, atelectasis, recurrent aspiration pneumonia, restrictive lung disease, or bronchiectasis. Bronchopulmonary dysplasia, in an infant born prematurely, will also increase the risk for respiratory disorders.

BONE MINERAL DENSITY DISORDERS

Decreased bone mineral density (BMD) and increased risk of fracture are present in patients with moderate to severe CP, especially those who are nonambulatory (83). A population-based study revealed that children in GMFCS levels I to III had a similar incidence and pattern for fractures as normally developing children (83). By the age of 10 years, most nonambulatory children have osteopenia, as defined by a BMD *z* score of less than -2.0 in their femurs (84). Data from the NAGCPP revealed that increasing severity of neurologic impairment, increasing difficulty feeding the child, use of anticonvulsants, and lower triceps skin fold *z* scores all independently contribute to lower BMD z scores in the femur (84). A published systematic review of available research, used to inform evidence-based clinical practice, concluded that there was probable evidence for bisphosphonates, possible evidence for vitamin D and calcium, and insufficient evidence for weight-bearing activities as effective interventions to improve low BMD. The evidence addressing fragility fractures was sparse. There was possible evidence that bisphosphonates prevent fragility fractures and inadequate evidence to support the use of weight-bearing or

vitamin D or calcium to decrease fragility fractures (85). The current evidence for weight-bearing activities is conflicting and consists of studies with small sample sizes, which may result in inadequate power, and studies of relatively short duration and significant variability in how much weight-bearing actually occurred (85). An observational, population study showed a fourfold reduction of nontrauma-related fractures in children who used standers versus those who did not, but this outcome may be attributed to reasons the children were not using standers, such as severe contractures (83). A recent small study demonstrated greater increases in BMD in dynamic loading interventions versus passive, suggesting this method of weight-bearing deserves further study (86). Given the possible effectiveness of vitamin D and calcium supplementation, and the relative safety of this intervention, vitamin D monitoring and supplementation, as well as ensuring adequate calcium intake is recommended for children with CP at risk for osteoporosis (85).

MUSCULOSKELETAL DISORDERS

Foot/Ankle

Foot deformities in CP are typically caused by an imbalance of the muscles that control segmental foot and ankle alignment. This imbalance may be a consequence of spasticity, disrupted motor control, or impaired balance function (87). Equinus deformity, due to increased tone or contractures of the gastrocsoleus complex, is the most common musculoskeletal deformity in CP. Equinovarus foot deformity is primarily due to a combination of spasticity of the posterior tibialis muscle and the gastrocsoleus complex, resulting in inversion and supination of the foot and a tight heel cord (Figure 14.10). This deformity is most common in a child with hemiparesis. In children with diplegia, the varus will often overcorrect into valgus (88). Equinovalgus foot deformity is due to spasticity of the gastrocsoleus complex and the peroneal muscles, as well as weakness in the posterior tibialis muscle. This deformity is most common in older children with spastic diparesis and quadriparesis. Planovalgus foot deformities begin with the lateral displacement of the navicular, causing the talar head to become uncovered and prominent in the mid-foot (88). Hallux valgus deformities are associated with valgus deformities of the foot, which may lead to a painful bunion at the head of the first metatarsal.

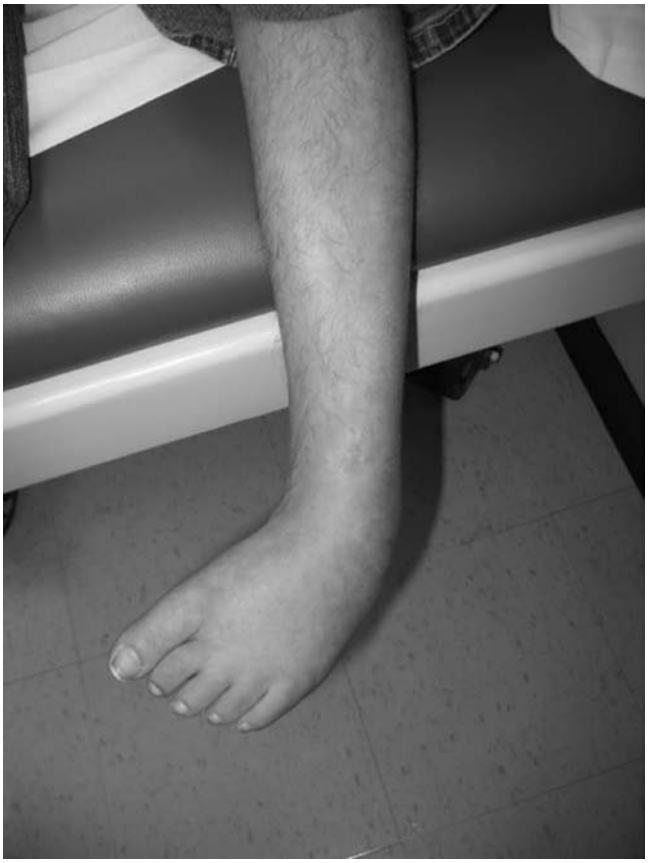

FIGURE 14.10 Equinovarus foot in a child with CP.

Knee

Knee flexion contractures are common due to spasticity in the hamstring muscles and static positioning in a seated position. If a severe knee flexion is present, hip flexion will be limited, resulting in lumbar kyphosis in the seated position. Flexion contractures at the knee are associated with hip and ankle flexion contractures and patella alta. Genu valgus may also occur, and is most commonly associated with excess femoral anteversion.

Hip

Acquired hip dysplasia is common in CP and often leads to progressive subluxation and possible dislocation. Hip subluxation can begin as early as age 2 years (89) and should be monitored closely by exam and serial radiographs. On exam, passive hip abduction of less than 35 degrees and a hip flexion contracture of more than 20 degrees are concerning signs of hip instability (90). On x-ray, hip subluxation is typically defined as a migration percentage greater than 30%. Close surveillance of hip migration with intermittent serial hip radiographs is recommended once hips have subluxed (91). Details of consensus-based hip surveillance programs have been published and include a baseline anterior–posterior (AP) pelvic radiograph at ages 12 to 24 months for all children with CP (92).

The reported incidence of dislocation in untreated hips varies, but 25% to 35% is the average estimate from most large series (91,93). Causative factors include persistent excessive femoral anteversion, a dysplastic

acetabulum, and muscle imbalance from overactive hip adductors and flexors. These factors cause the hip to be adducted, flexed, and internally rotated, placing it at risk for posterior dislocation. A large population-based sample of children revealed a linear relationship between the incidence of hip displacement and level of gross motor function on the GMFCS (91). The incidence of hip displacement for each GMFCS level was as follows: I 0%, II 15%, III 41%, IV 69%, and V 90%. The natural history of hip dislocation has not been well described. Early osteoarthritis and difficulty with positioning and hygiene are not uncommon. The reported incidence of pain associated with a dislocated hip varies, but is commonly felt to be present in at least 50% of patients with dislocations (93).

Children with CP may also develop a "windswept deformity" of their hips, described as an adduction deformity of the elevated hip and an abduction deformity of the opposite hip, which also tends to be externally rotated and commonly results in pelvic obliquity (Figure 14.11). The hip on the elevated side is at significant risk for dislocation, and positioning can be challenging. Hip dislocation with pelvic obliquity is often associated with scoliosis, but any potential causative relationship remains unproven.

FIGURE 14.11 Windswept hip deformity in a child with CP.

Spine

Spinal deformities, including kyphosis, lordosis, or scoliosis, are common in children with CP. Kyphosis is often seen in conjunction with significant weakness of the spinal extensor muscles and tightness in the hamstrings, leading to a posterior pelvic tilt. Lordosis is frequently associated with hip flexion contractures. The likelihood of scoliosis increases with the severity of CP. An overall incidence of approximately 20% (94) has been reported, with an incidence as high as 68% in children with spastic quadriparesis (95). Curves greater than 40 degrees tend to progress, regardless of the patient's skeletal maturity (95). The risk of progression is greatest for patients with quadriparesis, increased spasticity, a larger curve, a younger age, poor sitting balance, or pelvic obliquity (90).

Upper Extremity

Spasticity and muscle imbalances can often lead to joint deformities in the UE. The shoulder is often positioned in an adducted and internally rotated position. Spasticity in the biceps, brachioradialis, and the brachialis frequently result in elbow flexion contractures. Elbow flexion contractures less than 30 degrees rarely have functional significance. Forearm pronation deformities are common and can significantly affect functional use of the hand. The most common deformity of the wrist is flexion, typically with ulnar deviation (Figure 14.12). The most common finger deformities are flexion and swan neck deformities due to hand intrinsic muscle spasticity. A thumb-in-palm deformity is commonly seen with adduction at the carpometacarpal joint, which may be associated with hyperextension of the metacarpophalangeal and interphalangeal joints.

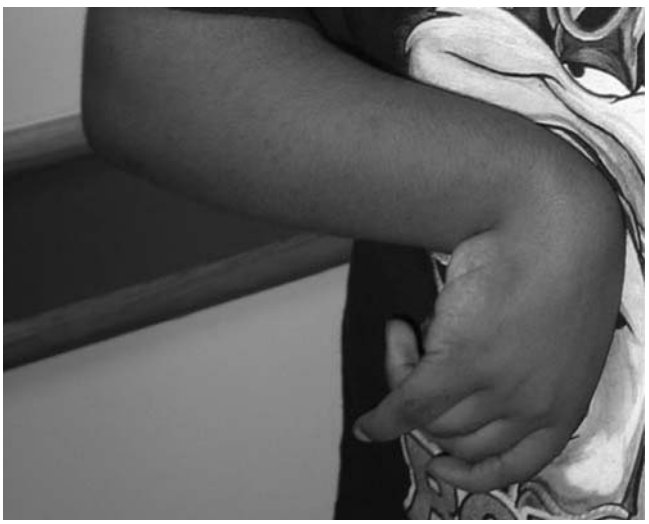

FIGURE 14.12 Wrist and finger flexion and ulnar deviation in a child with CP.

GAIT IMPAIRMENTS

A wide variety of gait classification systems have been developed to assist in diagnosis and clinical decision making, and to facilitate communication among health care providers. A systematic review of the literature, however, concluded that no single classification system appeared to reliably and validly describe the full magnitude or range of gait deviations in CP (96).

The following is a description of the more common gait deviations associated with CP (Table 14.1). At the hip, increased hip adduction tone can cause scissoring and difficulty advancing the limb in swing phase. Increased tone in the iliopsoas can lead to increased hip flexion, resulting in an anterior pelvic tilt and a crouched gait. Increased femoral anteversion can contribute to in-toeing. At the knee, tight hamstrings can inhibit the knee from extending during stance phase, further contributing to a crouched gait. Spasticity of the rectus femoris may limit knee flexion during the swing phase, causing a stiff-knee gait pattern. At the ankle, spasticity of the plantarflexors can lead to toe walking, difficulty clearing the foot during swing phase, or genu recurvatum (due to limited dorsiflexion in stance phase creating an extension moment at the knee). Spasticity of the ankle invertors, most commonly seen in spastic hemiparesis, can lead to supination of the foot and weight-bearing on the lateral border of the foot. Weight-bearing on the talar head is more common in spastic diparesis or quadriparesis, and is associated with an equinovalgus deformity. Malrotation of the leg can interfere with stability during stance phase and effective pushoff. Internal rotation is more common with a varus deformity and external rotation with a valgus deformity.

TREATMENT

GENERAL PRINCIPLES

The treatment of a child with CP requires a multidisciplinary approach. Once the diagnosis is made, the infant or child should be evaluated by a comprehensive rehabilitation team. The members of this team will vary, depending upon site and availability. Potential team members may include a physiatrist, developmental pediatrician, orthopedist, neurologist, physical therapist, occupational therapist, speech and language pathologist, therapeutic recreation specialist, orthotist, psychologist, social worker, and a nutritionist. The team should work with the child's caregivers to develop short- and long-term goals that address neuromuscular concerns such as maintaining ROM and tone control, as well as functional goals related to self-care skills, mobility, and communication. Goals related to increased societal participation should also be included. Goals should be routinely reassessed to ensure that they continue to be valid as the child grows older, and the child should be encouraged to take an active role in goal setting when appropriate.

Once the goals are determined, the family and the team must determine the most appropriate therapeutic approach. Although there are many treatment options to

TABLE 14.1 COMMON GAIT DEVIATIONS IN CP

LOCATION	IMPAIRMENT	POTENTIAL EFFECTS
Hip	Increased adductor tone	Scissoring; difficulty advancing leg in swing phase
	Increased iliopsoas tone	Anterior pelvic tilt; increased lumbar lordosis; crouched gait
	Increased femoral anteversion	In-toeing; false genu valgus; compensatory external tibial torsion
	Abductor weakness	Trendelenburg's gait
Knee	Decreased hamstring range of motion	Crouched gait
	Hamstring/quadriceps cocontraction	Stiff-knee gait
Ankle	Increased gastrocsoleus tone or contracture	Toe walking; genu recurvatum; difficulty clearing foot during swing
	Internal tibial torsion	In-toeing; ineffective pushoff
	External tibial torsion	Out-toeing; ineffective pushoff
	Varus	Increased ankle supination in stance or swing
	Valgus	Increased pronation in stance or swing; mid-foot break

choose from, little scientific evidence exists on which to base one's treatment decisions. The heterogeneity of CP, in addition to the lack of controls and disease-specific outcome measures, all contribute to this lack of evidence. In general, treatment should always start with the least invasive means with consideration of the cost-effectiveness of treatment options.

PHYSICAL AND OCCUPATIONAL THERAPY

Therapy Methods

Physical therapists and occupational therapists working with children with CP may choose from a variety of therapy methods, including neurodevelopmental therapy, Vojta, and Rood. There is, however, no clear scientific evidence to support the superior effectiveness of any one particular approach. These methods are sometimes referred to as the neurophysiologic approach, which elicit and attempt to establish normal patterns of movement through controlled sensorimotor experiences (97). This approach focuses on changing factors within the child at the domain of body function and structure, with the assumption that these changes will positively impact the domains of activity and participation. In recent years, the effectiveness of this approach has been questioned (97–99). Some have questioned whether facilitated automatic movement will improve voluntary, active movement (97). This has led to child-active or functional therapy approaches. This therapeutic approach involves actively practicing real-life tasks, typically in real-life environments, for the purpose of gaining skills that are important to the child and the family (29). Proponents of this type of therapy suggest that this approach is most likely to induce maximal neuroplasticity (29). Scientific evaluation of this method is still quite limited, but the level of evidence is highest for this approach in improving hand function (98). An RCT of child-focused therapy (therapy focused on changing identified constraints in the task or environment) versus context-focused therapy (therapy focused on minimizing identified impairments in the child), involving 128 young children with CP, found significant improvements in both groups and no difference in outcomes between groups (100). This suggests that both approaches can be successful. Often, therapists will use a combination of these therapeutic methods. The ideal duration and frequency of therapeutic programs remain unclear.

Stretching

Children with CP are at significant risk for contracture formation due to muscle imbalances and static positioning. Contractures can interfere with comfortable positioning, functional activities, and care needs, such as dressing, bathing, and toileting. After an initial assessment of baseline ROM, institution of a daily home exercise program with repetitive stretching exercises is usually recommended, although there is no clear evidence to support its efficacy or provide guidance in regard to the ideal frequency or duration (101). There is some evidence to suggest that a sustained stretch is preferable to manual stretching (102). Positioning techniques, orthotic devices, splints, and casting are often recommended to provide a more prolonged stretch. Serial casting is a technique where a series of successive casts are applied in the hope of progressively increasing the ROM with each cast. It is used most frequently at the ankle joint, often in conjunction with botulinum toxin serotype A (BoNT-A), in order to improve dorsiflexion ROM. A systematic review of the literature supports the use of casting at the ankle (98). The evidence suggests a short-term effect on improved ROM (103) and stride length during ambulation (104). Although a number of small RCTs have compared BoNT-A and casting, there is no strong and consistent evidence that casting, BoNT-A, or the combination of the two is superior to the others (105). Lack of evidence was primarily attributed to methodological limitations of the available studies.

Strengthening

Formalized strength testing in ambulatory children with spastic diparesis or hemiparesis has confirmed greater weakness in all muscles tested using age-matched controls (106). Weakness was more pronounced distally, as expected, and hip flexors and plantar flexors were relatively stronger than their antagonists when compared to the strength ratios of the control group. Strength of the uninvolved side in children with hemiparesis was also weaker than age-matched controls (106). Deficits in voluntary muscle contraction in CP are felt to be due to decreased central nervous system motor unit recruitment, increased antagonist coactivation, and changes in muscle morphology, including muscle fiber atrophy and increased fat and connective tissue (107). This weakness is thought to be a large contributor to functional deficits in children with CP, but historically, strengthening programs were not recommended due to concerns of increasing spasticity. A number of studies have shown, however, that strengthening programs can increase strength without adverse effects such as increased spasticity, resulting in an increased interest in strengthening programs for children with CP (108,109).

Strength or resistance training has been the subject of several systemic reviews and has been included in reviews of different therapy approaches to improve gait or other aspects of motor function (109–111). The general consensus across studies is that strength can be predictably increased through a properly designed short-term program, but training likely needs to be continued to retain benefits (109). Strengthening in functional positions may transfer more readily to improvement in motor

tasks; however, it is uncertain whether it is the loading or the functional practice that makes the difference (110).

Although strengthening has the potential to positively affect children with CP in many areas of the International Classification of Functioning, Disability and Health (ICF) model, most studies have focused on changes in strength alone. Some activity level improvements have been reported with strength training, as measured by the Gross Motor Function Measure (GMFM) (112–114). Other studies have reported an improvement in strength, but no associated improvement in function (115–117). The lack of functional improvement has been attributed to a number of potential causes including: insufficient dose or density to drive neuroplasticity (118), the lack of context-specific training (118), or inadvertently strengthening nondesired muscles, leading to further muscle imbalance (119). Walking is a complex task and there are a number of other impairments besides weakness that can make ambulation difficult including impaired motor planning, impaired balance and postural control, abnormal tone, and decreased ROM (116). Although not typically measured, increased participation, self-esteem, and participant-rated functional mobility have been associated with participation in a strengthening program (116,120,121). Strengthening appears to be a promising intervention for children with CP, but future studies are needed to determine the effect of individual patient factors on a wide variety of potential outcomes, including societal participation.

TREADMILL TRAINING (TT) AND PARTIAL BODY WEIGHT SUPPORT TREADMILL TRAINING (PBWSTT)

TT is utilized to provide repetitive task-specific walking practice. The goals are to increase lower extremity (LE) strength, increase speed, improve symmetry of gait patterns, or to increase endurance (110). A systematic review and recent studies provide support for improvement in temporal–spatial characteristics of gait, 10-m walk test, and GMFM scores with the use of TT (122–124). Little evidence is available on the effects of TT on participation.

To facilitate step training, weight support systems were developed to be used in conjunction with treadmills to reduce postural requirements and thus the amount of physical assistance needed to safely participate in ambulation training (110). A systematic review of PBWSTT also revealed significant improvements in temporal–spatial characteristics of gait and GMFM scores (122). An RCT comparing PBWSTT to overground gait training found no additional benefit with the PBWSTT (125).

Robotic-assisted treadmill therapy utilizes two mechanically driven leg orthoses, resulting in a kinematic pattern resembling normal walking. This allows for an intensification of locomotor training by increasing the amount of stepping practice, as well as altering the amount of body weight support being provided while decreasing the therapist's manual assistance. To date, few studies have reported on the effects of robotic-assisted TT in children with CP. A small noncontrolled study showed improvements in walking and standing performance on the GMFM (126) and a prospective controlled cohort study revealed improvements in activity and participation (127).

Larger RCTs will be needed to increase confidence in the positive effects of TT and PBWSTT and to determine practice guidelines.

CONSTRAINT-INDUCED MOVEMENT THERAPY (CIMT)/INTENSIVE BIMANUAL TRAINING

CIMT and intensive bimanual training are motor-learning-based approaches that focus on upper limb (UL) function in children with hemiplegic CP. CIMT was developed for treating adults with hemiparesis or "learned nonuse" following a stroke (128). The therapy includes intensive motor practice or shaping of the paretic UE combined with restraint of the uninvolved extremity. Classic CIMT is defined as restraint of the unaffected limb in conjunction with at least 3 hours per day of therapy for at least two consecutive weeks, whereas modified CIMT (mCIMT) uses a more child-friendly approach with the use of a mitt, splint, or bandage and varying frequencies/duration of restraint.

CIMT focuses on training unilateral dexterity, but children with hemiplegic CP have impairments with coordination of two-handed activities (129). CIMT does not address this impairment directly and thus generalization of the training may not apply (130). This led to the development of intensive bimanual training, which engages the child in activities that require the use of both hands as a means to increase use of the affected extremity. One highly structured form of bimanual training is hand arm bimanual training (HABIT) (131). Sequential use of CIMT followed by intensive bimanual therapy (hybrid therapy) has also been tried in order to obtain the unique benefits of both individual therapies (132).

CIMT and intensive bimanual therapy are some of the more thoroughly studied interventions for CP and there is strong evidence for their effectiveness (98). A meta-analysis published in 2013 included 5 classic CIMT studies, 20 mCIMT studies, and 2 hybrid studies (133). The results revealed that individually these studies predominately reported improved UE outcomes, compared with usual care, at substantially lower dosages. The meta-analysis revealed modest to large effect sizes of mCIMT on improving efficiency and quality of movement of the impaired arm compared with usual care. The comparison of unimanual therapy (eg, mCIMT) with an equivalent dose of bimanual therapy yielded similar results. This same study reviewed the success of implementing these therapies in the home setting and found that for mCIMT, between 50% and 80% of the anticipated dose

was achieved and 85% for bimanual training suggesting that this form of therapy may be easier to implement in the home setting (133).

The preferred frequency, duration, or method of CIMT or bimanual therapy has yet to be determined. An expert consensus panel concluded that future research should be focused on the following three key questions: (a) the effect of age on treatment effect, (b) benefits of repeated treatment sessions, and (c) whether the amount of structured training matters (134).

ELECTRICAL STIMULATION

Proponents of electrical stimulation suggest that it increases strength and motor function, and it is an attractive alternative for strengthening in children with poor selective motor control (135).

Neuromuscular Electrical Stimulation (NMES)

NMES utilizes electrical current to produce a visible muscle contraction. The results of two small case series found increased active and passive ROM at the ankle after stimulation of the anterior tibialis (136) and improved sitting balance following stimulation of the abdominal and posterior back muscles (137). Two RCTs failed to identify any statistically significant improvement in strength or function following NMES of the quadriceps (138) or gluteus maximus (139), but both of these studies were underpowered.

Functional Electrical Stimulation (FES)

If NMES is used to make a muscle contract during a functional activity, it is termed FES. FES is commonly used at the anterior tibialis muscle to increase dorsiflexion during ambulation. A small case series documented improvement in heel strike and ankle dorsiflexion following FES in children (140). Another study identified clinically significant improvements in gait in only 3/8 subjects with CP, as measured by a three-dimensional gait analysis (141). One proposed reason for lack of response was spasticity of the antagonist muscles limiting range and speed of movement.

Commercially available devices, commonly referred to as neuroprostheses, are available to deliver asymmetrical biphasic surface electrical stimulation to the common fibular nerve, triggered by a tilt sensor, to improve foot clearance during swing phase. FES may have potential benefits versus traditional orthotic use in foot drop. It does not restrict motion, produces muscle contraction, and thus has the potential to increase strength and motor control (142). It also is likely to be more acceptable from a cosmetic point of view. The results of small studies support its effectiveness and tolerability in managing foot drop in children with CP (142,143). One study reported use-dependent muscle plasticity in CP, but permanent improvements in voluntary ankle control after repetitive stimulation were not demonstrated (144). Some participants had a decrease in active ankle dorsiflexion after using the device and this was felt to be because they had "learned" to let the device take over ankle control (144). A recent review of the five available RCTs to date on FES in CP found that FES was more effective than no FES intervention, but had a similar effect to activity training alone (145).

Threshold Electrical Stimulation (TES)

TES is a low-level electrical stimulus, often applied during sleep that does not result in a visible muscle contraction. The proposed mechanism of TES is that increasing blood flow during a time of heightened trophic hormone secretion results in increased muscle bulk (146). There have been four RCTs evaluating TES to date, and three of them failed to show any improvement in strength or function (138,147,148). The parents, however, reported a perceived positive effect of treatment in two of the studies (147,148), and a decreased impact on disability as measured by the Lifestyle Assessment Questionnaire was found in the third (138).

A systematic review of electrical stimulation in CP concluded that the scarcity of well-controlled trials makes it difficult to support definitively or discard the use of this therapy (135). Another systematic review revealed low quality of evidence to support improved gait and moderate-quality evidence to support improved strength with electrical stimulation (98). Further studies with more rigorous designs, longer follow-up, larger sample sizes of more homogeneous subjects, and clarity in the reporting of stimulation parameters are recommended to clarify the age and type of patient most likely to benefit from this intervention (135).

SPEECH THERAPY

Involvement of a speech and language pathologist is useful in the assessment of children prior to early intervention or early childhood educational planning. Many children with CP have oromotor deficits, dysphagia, dysphonia, and/or articulation and language deficits. It is essential to recognize these deficits promptly and enroll these children into speech therapy services to address treatment strategies in an effort to correct or improve these concerns.

HYPERTONIA MANAGEMENT

Hypertonicity affects the majority of children with CP (149,150). It may occur focally in distinct muscle groups, as is often the case in diparesis or hemiparesis, or more

globally, affecting the majority of axial and appendicular skeletal muscles. Hypertonicity can result in a number of negative effects. It can interfere with positioning, contribute to the formation of contractures and musculoskeletal deformities, and be a source of discomfort. It can also negatively affect function and make caregiver tasks, such as transfers and dressing, more difficult. Increased tone can sometimes assist with function. For example, increased extensor tone in the lower extremities may assist with standing and transfers.

A wide variety of treatment options for hypertonicity are available, including oral medications, nerve blocks, and surgery. Determining whether abnormal tone is present globally or focally and the magnitude of its effect on an individual's musculoskeletal system, function, and comfort should guide one's treatment plan. The specific goals of tone reduction should always be determined prior to any intervention. Evaluating for medical causes of increased tone, such as pain or constipation, is very important. The first-line approach should include stretching, splinting, and positioning as appropriate. Other medical or surgical interventions can be used in conjunction with these when further reduction in abnormal tone is desired.

CHEMICAL DENERVATION

Chemical denervation should be considered for the treatment of significant focal increases in tone.

Alcohol Blocks

Alcohol nerve and motor point blocks have been used for many years to reduce focal increases in tone. Phenol injections, at 3% to 5% solutions, either at motor points of selected muscles or perineurally, denature proteins and disrupt efferent signals from hyperexcitable anterior horn cells by inducing necrosis of axons (151–153). Alcohol blocks have the potential to cause painful dysesthesias (153). Nerves that are more commonly treated with phenol include the musculocutaneous and obturator nerves, given the reduced sensory function of these nerves and the lower risk for dysesthesias. The low cost of phenol, coupled with reports of duration of action exceeding 12 months (154), renders phenol injections an attractive treatment option in selected patients with focal spasticity (151). They are frequently done under general anesthesia, however, adding additional risks and costs. There is no published evidence to support or refute the use of these blocks in children with CP (98,155).

Botulinum Neurotoxin (BoNT)

BoNT is a protein composed of a heavy chain, which binds nerve terminals at the neuromuscular junction, and a light chain, which is transported into the nerve terminal blocking the release of acetylcholine presynaptically and thereby weakening the force of muscle contraction produced by the hyperexcitable motor neurons. BoNT exists in seven serotypes, designated A through G. The *Food and Drug Administration* (FDA) has not approved BoNT for the treatment of spasticity in children.

Muscles commonly treated with BoNT include the gastrocsoleus complex, hamstrings, hip adductors, and flexor synergy muscles of the UE. Intramuscular injections can be localized by surface landmarks, electromyographic guidance, and/or ultrasound. Following injection, muscle relaxation is evident within 48 to 72 hours and persists for a period of 3 to 6 months (156). Dosing is based on units derived from the mouse lethality assay and is not equivalent among the various brands. It is dependent upon both body weight and size of the target muscle(s). Universally accepted dosing guidelines do not exist, but consensus statements of dosing and injection techniques are available for guidance (155,157–159). Injections are typically spaced a minimum of 3 months apart due to concerns of antibody formation (151,160).

Side effects are rare with BoNT, but may include pain during injection, infection, bleeding, a cool feeling in injected limbs, rash, allergic reaction, flu-like symptoms, excessive weakness, and fatigue (161–163). Although reports of life-threatening side effects are extremely rare following treatment with BoNT, the risk of systemic spread is possible. The FDA sent out an early communication in 2008 about an ongoing safety review of BoNT A, B following reports of reactions that were suggestive of botulism. The most serious cases resulted in hospitalization and death, and occurred most commonly in children with CP treated for limb spasticity. At the time of writing, the FDA has not completed its review, but recommends that all health care professionals who use BoNT should: (a) understand the potency determinations expressed in "units" or "U" are different among the botulinum toxin products; clinical doses expressed in units are not comparable from one botulinum product to the next; (b) be alert to the potential for systemic effects, following administration of botulinum toxins, such as: dysphagia, dysphonia, weakness, dyspnea, or respiratory distress; (c) understand that these effects have been reported as early as 1 day and as late as several weeks after treatment; (d) provide patients and caregivers with the information they need to be able to identify the signs and symptoms of systemic effects after receiving an injection of botulinum toxin; (e) tell patients that they should receive immediate medical attention if they have worsening or unexpected difficulty swallowing or talking, trouble breathing, or muscle weakness (164).

Many studies in the literature describe the effects of BoNT-A in children with CP. A number of evidence-based reviews have been published summarizing this vast literature. There is strong evidence that BoNT-A is an effective treatment to reduce spasticity in

the upper and lower extremities in children with CP who have localized/segmental spasticity (98,155). There is conflicting evidence regarding functional improvement in children with CP for both the upper and lower extremities (155,157,158). In spite of the lack of clear evidence, published expert opinion and reviews support the use of BoNT-A in the lower extremities in conjunction with physical therapy to improve walking function (98,165). The use of BoNT-A in the UE in combination with occupational therapy to improve function is also supported by expert opinion and reviews in the literature (98,157,166). The use of BoNT-A to improve comfort and ease of care in children with CP who are nonambulant has not been well studied. There has been only one RCT, to date, in this population, which revealed children with CP who received BoNT-A with therapy, versus therapy alone, had better results on the Canadian Occupational Performance Measure, decreased pain, and a corresponding improvement in health status (167).

ORAL MEDICATIONS

Oral medications are often used as an early treatment strategy for global spasticity. Medications that are most frequently used include baclofen (Lioresal), dantrolene sodium (Dantrium), clonidine, diazepam (Valium), and tizanidine (Zanaflex). All of these medications work through the central nervous system, with the exception of dantrolene sodium and, therefore, have the potential for sedation (Table 14.2). None of these medications have been found to be universally effective in relieving spasticity (168), and evidence related to functional improvement is extremely sparse. The choice of medications is, therefore, often based on the impact of potential side effects on the individual patient. Widely accepted dosing guidelines do not exist for any of these medications for CP and therefore most clinicians utilize a low starting dose followed by a gradual dose escalation to find the best clinical response with tolerable side effects.

Benzodiazepines

Benzodiazepines have an inhibitory effect on both the spinal cord and supraspinal levels mediated through binding near but not at the gamma-aminobutyric acid (GABA) receptors and increasing the affinity of GABA for GABAA receptors (169). Diazepam is the most frequently used benzodiazepine and oldest antispasticity medication that is still in use (170), but like other oral medications in CP, its effectiveness has not been well evaluated. It is rapidly absorbed, reaching peak drug levels an hour after drug administration. The positive effect of diazepam may be related to general relaxation that permits improvements, especially in those individuals with athetosis and spasticity (171,172).

Baclofen

Baclofen is a GABA analogue that acts at the spinal cord level to impede the release of excitatory neurotransmitters implicated in causing spasticity (173). Low lipid solubility impedes passage through the blood–brain barrier with more than 90% of the absorbed drug remaining in the systemic circulation (174). As a result, large doses may be necessary to achieve an effect. A study of pharmacokinetics (PK) in children with CP taking oral baclofen found that the PK was largely proportional to dose/kg, making dose adjustments uncomplicated by PK issues (175). Very few studies have been published regarding the use of oral baclofen in CP. Two small double-blind, placebo-controlled, crossover trials produced differing conclusions regarding the effectiveness of baclofen in reducing spasticity, but neither employed validated outcome measures (176,177). Additional studies assessed the effect of oral baclofen for reduction of spasticity and improved function in small numbers of subjects with moderate to severe spasticity. One study showed possible deleterious effects on motor function (178), while the other demonstrated no difference with placebo except in goal attainment (179).

Dantrolene Sodium

Dantrolene sodium is unique in that it works primarily through actions on the skeletal muscle and not through central nervous system pathways. It inhibits the release of calcium from the sarcoplasmic reticulum, thereby uncoupling electrical excitation from muscle contraction and reducing contraction intensity. It is well absorbed within 3 to 6 hours after ingestion and is metabolized in the liver to 5-hydroxydantrolene, with peak effect in 4 to 8 hours (180). Doses in children range up to 12 mg/kg/day (172). It is often suggested that dantrolene be considered for the treatment of spasticity of cerebral origin because its mode of action is not central nervous system mediated and it is less likely to be sedating (170,172,181), although an expert panel noted that it is rarely used in clinical practice to reduce spasticity in children with CP (155). Their conclusion is that this is most likely due to the lack of evidence in the literature to support its efficacy along with the potential for frequent and serious side effects. Side effects from treatment can include sedation, nausea, vomiting, and diarrhea. Use of dantrolene is also associated with hepatotoxicity (180,182). Liver function studies should be done prior to instituting treatment and periodically while on maintenance therapy (170). There are a few published trials of Dantrium in CP. One report of long-term use of dantrolene in children with spastic diparesis indicated that young children achieved greater levels of function than predicted prior to dantrolene administration and older children were able to move more easily and maintain their highest levels of function (183).

Additional oral medications used to treat spasticity in children with CP include alpha 2 agonists, such as

TABLE 14.2 MEDICATIONS USED TO TREAT SPASTICITY IN CHILDREN

DRUG	MECHANISM OF ACTION	SIDE EFFECTS AND PRECAUTIONS	PHARMACOLOGY AND DOSING
Baclofen	Binds to receptors (GABA) in the spinal cord to inhibit reflexes that lead to increased tone Also binds to receptors in the brain leading to sedation	Sedation, confusion, nausea, dizziness, muscle weakness, hypotonia, ataxia, and paresthesias Can cause loss of seizure control Withdrawal can produce seizures, rebound hypertonia, fever, and death	Rapidly absorbed after oral dosing, mean half-life of 3.5 hours Excreted mainly through the kidney Dosing: in children start with 2.5–5 mg/d, increase to 30 mg/d (in children 2–7 years of age) or 60 mg/d (in children 8 years of age and older)
Diazepam	Facilitates postsynaptic binding of a neurotransmitter (GABA) in the brainstem, reticular formation, and spinal cord to inhibit reflexes that lead to increased tone	Central nervous system depression causing sedation, decreased motor coordination, impaired attention and memory Overdoses and withdrawal both occur The sedative effect generally limits use to severely involved children	Well absorbed after oral dosing, mean half-life 20–80 hours Metabolized mainly in the liver In children, doses range from 0.12–0.8 mg/kg/d in divided doses
Clonidine	Alpha 2 agonist Acts in both the brain and spinal cord to enhance presynaptic inhibition of reflexes that lead to increased tone	Bradycardia, hypotension, dry mouth, drowsiness, dizziness, constipation, and depression These side effects are common and cause half of the patients to discontinue the medication	Well absorbed after oral dosing, mean half-life is 5–19 hours Half is metabolized in liver and half is excreted by kidney Start with 0.05 mg bid, titrate up until side effects limit tolerance May use patch
Tizanidine	Alpha 2 agonist Acts in both the brain and spinal cord to enhance presynaptic inhibition of reflexes that lead to increased tone	Dry mouth, sedation, dizziness, visual hallucinations, elevated liver enzymes, insomnia, and muscle weakness	Well absorbed after oral dosing, half-life 2.5 hours Extensive first pass metabolism in liver Start with 2 mg at bedtime and increase until side effects limit tolerance, maximum 36 mg/d
Dantrolene sodium	Works directly on the muscle to decrease muscle force produced during contraction Little effect on smooth and cardiac muscles	Most important side effect is hepatotoxicity (2%), which may be severe Liver function tests must be monitored monthly, initially, and then several times per year Other side effects are mild sedation, dizziness, diarrhea, and paresthesias	Oral dose is approximately 70% absorbed in small intestine, half-life is 15 hours Mostly metabolized in the liver Pediatric doses range from 0.5 mg/kg bid, up to a maximum of 3 mg/kg qid

Source: Reprinted from Ref. (188). Green LB, Hurvitz EA. Cerebral palsy. *Phys Med Rehabil Clin N Am*. 2007;18(4):866–867, with permission from Elsevier.

clonidine and tizanidine, as well as certain anticonvulsants, including gabapentin (Neurontin). The alpha 2 agonists result in decreased motoneuron excitability by decreasing the release of excitatory amino acids (182). The side effects associated with these agents are frequently the cause of their more limited use and include nausea, vomiting, hypotension, sedation, dry mouth, hallucinations, and hepatotoxicity. In addition, reversible liver enzyme elevations have been noted in 2% to 5% of patients (170). There is little literature to guide the effective use of tizanidine for the management of spasticity in children with CP (155). Gabapentin is structurally similar to GABA, readily crosses the blood–brain barrier, and is not protein-bound. It does not activate GABA, but results in increased brain levels of it (170). Reports of its use in children with spasticity are not available as of yet.

INTRATHECAL BACLOFEN (ITB)

ITB was first described by Penn and associates in 1984 and was FDA-approved for the treatment of spasticity of cerebral origin in 1996. Baclofen is delivered directly to the cerebrospinal fluid (CSF) via a catheter connected to an implanted device in the abdomen. The device contains

FIGURE 14.13 SynchroMed II programmable pump.

a peristaltic pump, a reservoir for baclofen, and electronic controls that permit regulation of the pump by telemetry. This feature allows baclofen infusion rates to be either continuous throughout the day or at varied dosages in order to accommodate the patient's specific needs (173). The newer versions of the pump, such as the SynchroMed II (Figure 14.13), have an expected battery life of 5 to 7 years. By infusing baclofen directly into the subarachnoid space around the spinal cord, potentiation of GABA-mediated inhibition of spasticity can be achieved while minimizing side effects related to high levels of baclofen in the brain (151). Administration of intrathecal baclofen produces levels of baclofen in the lumbar CSF that are 30-fold higher than those attained with oral administration (151). The half-life of intrathecal baclofen in the CSF is 5 hours (184).

Patient selection for ITB therapy is very important. A European panel of experts published a consensus on the appropriate use of ITB therapy in children and included the following guidelines: (a) confirmed spasticity that interferes with patients' abilities and QOL and cannot be adequately controlled by managing aggravating factors, therapy, oral medications, or BoNT; (b) family has the ability and motivation to attend regular follow-up and monitoring; (c) ITB therapy is licensed for children older than 4 years old, but has been used in younger children; (d) sufficient body size to accept pump bulk and weight (185). Prior to surgical implantation, a test dose of 50 to 100 mcg of intrathecal baclofen is often given, via lumbar puncture, to verify a reduction in tone. Some centers are opting to not use a test dose because it is believed the results of a single bolus do not reliably predict the results of continuous dosing over time and may also increase the risk of complications after implantation (185).

Once implanted, the intrathecal pump is typically programmed to deliver baclofen at a continuous rate, typically at a total daily dose similar to the bolus dose given during the trial. The dose is not related to age or weight (184). The dose is then slowly titrated over the first few months (185). Bolus dosing can be considered if spasticity is particularly troublesome during a specific time of the day. Ongoing monitoring for side effects of ITB including hypotonia, somnolence, headache, convulsions, dizziness, urinary retention, nausea, and paresthesias is important. Refills of intrathecal baclofen are generally needed every 2 to 6 months, depending on baclofen infusion dosage, the size of the pump, and the concentration of the baclofen being used.

Complications from ITB therapy are common and can result from many causes including: programming error, pump failure, pump flip, catheter failure, granuloma formation at the catheter tip, skin erosion from pump or catheter, CSF leak, and infection. The most common postoperative complications are pump pocket fluid collections and infections (151). Infection may remain isolated to the pump pocket or may track along the catheter, resulting in meningitis (184,186). A review of 430 consecutive pediatric patients receiving ITB therapy at one center found a 25% complication rate. The most common complication was related to the catheter (15.1%). Infection was also common (9.3%) and was more likely if the pump was placed subcutaneously versus subfascially or if it was a pump replacement surgery versus a first pump implantation. CSF leaks were less common (4.9%) and pump failures were rare (1%) (187). Because complications are common with ITB therapy, and can be life-threatening, any center providing this therapy must have clinicians readily available who are knowledgeable about the evaluation and treatment of such complications.

Catheter or pump dysfunction can result in decreased baclofen delivery and baclofen withdrawal. Intrathecal baclofen withdrawal can also be seen in cases of battery failure without low battery alarm warning (170). Early symptoms of withdrawal include pruritus, dysphoria, irritability, increased spasticity, tachycardia, fever, and changes in blood pressure (188). If not recognized and managed optimally, baclofen withdrawal may progress to serious and life-threatening complications, including severe hyperthermia, seizures, rhabdomyolysis, disseminated intravascular coagulation, altered mental status, psychomotor agitation followed by multisystem failure, and death (189,190).

If acute withdrawal is suspected, referral for immediate medical care is recommended. Although high-dose oral baclofen is recommended, in some cases this will not alleviate the withdrawal symptoms. Treatment for withdrawal can include any combination of oral baclofen, intravenous diazepam, or infusion of intrathecal baclofen through the use of a lumbar drain (191). Cyproheptadine, a serotonin antagonist, has also been used as an adjunct to baclofen and diazepam for treatment of severe intrathecal baclofen withdrawal (192,193). Dantrolene sodium use should also be considered in patients with suspected rhabdomyolysis as a result of withdrawal.

Investigations into the causes for withdrawal should then ensue, including plain radiographs to assess pump and catheter placement in comparison to previous radiographs. Further studies may include dye or isotope studies to assess for catheter placement, leakage, and kinking.

Overdoses have been reported, typically as a result of human error in programming or refill procedure (170). Medtronic also released a product advisory in 2014 noting that pumps can rarely malfunction and be potentially life-threatening over infusion (194). Symptoms of overdose include drowsiness, lightheadedness, nausea, vomiting, seizures, respiratory depression, hypothermia, and loss of consciousness. In such cases, the pump should be stopped and respiratory support provided until the effects of baclofen have worn off. Intravenous physostigmine or withdrawal of 30 to 40 mL of CSF can be tried in severe overdoses (188).

A number of studies have reported on the outcomes of ITB therapy, especially in children with GMFCS levels IV and V. Reviews of the available literature have found that outcomes are based primarily on evidence for levels 3 to 4 and that the best established treatment effect of ITB therapy is in reducing spasticity of lower extremities in patients with severe spasticity, commonly found in GMFCS levels IV to V (155,185). Noncontrolled trials have demonstrated improvements in joint ROM, function, ease of care, and reduced pain (195–199). Treatment with intrathecal baclofen has also been associated with an increase in weight gain velocity (200). Retrospective studies in children with CP receiving ITB have documented varying effects on scoliosis, including rapid progression (201,202) and/or no significant effect on curve progression, pelvic obliquity, or the incidence of scoliosis when compared with matched controls (203). The methodology for the first RCT for ongoing ITB therapy in CP has been published (204). In spite of the lack of level 1 evidence, and significant risk for serious complications, ITB therapy is widely used for the treatment of severe spasticity that is refractory to other spasticity interventions in children with CP.

SELECTIVE DORSAL RHIZOTOMY (SDR)

SDR is a neurosurgical procedure that involves partial sensory deafferentation at the levels of L1 through S2 nerve rootlets (203). Operative technique involves the performance of single or multilevel osteoplastic laminectomies, exposing the L2 to S2 roots (151,205,206). Motor and sensory roots are separated to allow for electrical stimulation of individual sensory roots. The selection of rootlets for cutting is based on the LE muscular response to electrical stimulation of the rootlets. Although there is variability in percentages of rootlets cut, in general, a maximum of 50% of the sensory rootlets at any level are cut (207), thereby preserving sensory function (208). Following the procedure, the reduction in spasticity often unmasks a significant amount of LE weakness that could negatively impact functional activities, such as ambulation. As a result, an extensive amount of intensive therapy is necessary to guide the patient through appropriate motor patterns and strengthening programs.

Although immediate perioperative complications are common with SDR, long-term complications such as sensory dysfunction and bowel or bladder dysfunction are infrequent (209,210). The risk of subsequent spinal deformities may increase after laminectomies or laminoplasties done in conjunction with SDR, although this may be less of a problem in the lumbar or lumbosacral area than higher in the spinal column (211). Decreased spasticity and alterations in the balance of muscle tone in the trunk and hips may also influence the development of spinal deformities (211). In a review of six studies with a follow-up period of 5.8 to 21.4 years, 5% to 29% of the patients reported back pain at the long-term follow-up, and a variety of spinal abnormalities were common (210). Scoliosis (41%–56%) and lumbar lordosis (10%–50%) were the most common abnormalities reported at long-term follow-up. The authors of this review concluded that no conclusion can be drawn about the relationship of the spine abnormalities and SDR (210).

SDR is an irreversible procedure; therefore, appropriate patient selection is important, but it remains unclear and controversial. A widely cited meta-analysis of short-term gains following SDR in children with CP suggests that ideal candidates include children between the ages of 3 and 8 years who are GMFCS level III or IV (212). A more recent systematic review of published SDR studies was performed to summarize the selection criteria for SDR in children with CP (208). In order to be included in the review, the article needed to provide a detailed description of their SDR selection process, although only 21% of the 52 articles described the process in sufficient detail to be reproducible. Most of the selection criteria were based on clinical rationale versus scientific evidence and focused primarily on the ICF domain of body structure and function. Only 27% of the studies stated their specific treatment goals and a limited number of studies looked at the influence of preoperative patient characteristics and outcome following SDR (208). The results of this review revealed that no study has shown a correlation between outcomes after SDR and the degree of preoperative spasticity, muscle strength, motor control, ROM in the lower extremities, or truncal hypotonia (208). The authors of the review concluded that at present there is no international consensus on the selection of patients for SDR, but since these studies concluded, new and better methods of assessment have been developed and should impact future attempts to agree on selection criteria (208).

The results of short-term outcomes of SDR in children with CP are more readily available than long-term outcomes. A meta-analysis of three randomized controlled studies comparing short-term outcomes of SDR plus physical therapy with physical therapy alone has

been published (212). Findings included a clinically important decrease in spasticity, as well as a small but statistically significant advantage in function (GMFM-88) with SDR plus physical therapy up to 2 years following surgery. The subjects in these studies were primarily ambulatory children with spastic diparesis; those with dystonia, athetosis, and ataxia were excluded. Another early systematic review summarized the short-term positive effects of SDR on ICF body structure and functions and activity domains (213).

Studies evaluating outcomes at 5 years or greater were published more recently. A systemic review of this literature notes that the strength of the evidence in the available studies is very poor and the conduct of the studies was weak to moderate (210). The authors concluded that there is moderate evidence that SDR has a positive long-term influence on the ICF body structure and function domains, but there is no evidence that SDR has an influence on the ICF activity and participation domains (210). A study published following this review on long-term outcomes, evaluated a select group of patients in levels I, II, and III of the GMFCS (214). They looked at the GMFM-66 scores at 5 and 10 years relative to what might be expected on previously published reference centiles or motor curves. To be considered better or worse, they had to demonstrate a change of more than 20 centiles in comparison to the reference centiles. The results revealed that none of the children showed deterioration of gross motor function based on centile ranking. Five years after SDR, 10 out of 28 children showed improvement and 10 years after SDR 6 out of 20 children had improved (214).

Although SDR is performed routinely for the treatment of significant spasticity in lower extremities of children with CP, there is limited published data on long-term outcomes or a consensus on the most appropriate selection criteria. There is also no agreement on the optimal surgical technique. Future long-term studies are needed with larger patient populations. It has been suggested that matched controlled studies would be the most feasible and that the ICF domains of activity and participation should receive special attention (210).

ORTHOPEDIC SURGERY

Neurologic impairments are associated with progressive musculoskeletal pathology in the majority of children with CP (215). Orthopedic surgery is most often recommended in children with muscles that are dysphasic, firing out of phase, or those muscles that show excessive activity while working in phase, thereby overpowering their antagonist and inhibiting smooth joint motion (216). The combination of this muscular imbalance with the lack of stretching of the muscles in the relaxed state leads to contracture formation as the muscle–tendon unit fails to keep up with the skeletal growth of the child, and may lead to bony changes as well as fixed deformities (217). The usual goal of surgery is to weaken these dysphasic muscles and reduce potential contracture formation and muscle imbalance. This may be achieved through tendon lengthenings or transfers. The muscles that are most frequently addressed surgically in the LE are those that cross two joints, including the hip adductors, hip flexors, hamstrings, rectus femoris, and gastrocsoleus complex. Rotational osteotomies are occasionally done to correct femoral anteversion or tibial torsion that results in significant gait disturbances. Guided growth in the form of an anterior hemiepiphysiodesis of the distal femur may help correct fixed knee flexion deformities in the younger child (218). Common indications for UE surgery include elbow flexion contracture, excessive pronation, and thumb-in-palm deformity (219,220).

When improved function is the goal of surgery, multiple muscles and joints may be targeted because they are all interrelated in specific movement patterns; therefore, a single-event multilevel surgery (SEMLS) is more common than multiple staged surgeries (221–223). A common multilevel soft tissue surgical approach includes three procedures: the hamstring lengthening, rectus femoris transfer, and gastrocsoleus lengthening (224). There have been many studies published on the results of SEMLS in CP. The only systemic review of SEMLS in CP was published in 2012 (215). Thirty-one studies met the inclusion criteria for review. The majority of these studies were retrospective and only five had concurrent, nonrandomized controls. A meta-analysis of the results was not considered appropriate due to a number of factors, including the heterogeneity of the participants, outcome measures, surgical interventions, and study quality. For the ICF domain of body structure and function, the authors concluded that the balance of evidence suggests improvement in ROM, selected gait parameters, overall gait index, and energy efficiency. There was less evidence to make conclusions in the ICF activity and participation domains. The changes in GMFM after SEMLS were usually small, variable, and inconclusive (215). The authors noted widespread acknowledgment of the importance of postoperative care and rehabilitation, yet the information about the intensity, frequency, and type of rehabilitation therapy was often sparse in the published studies. When reported, the inpatient stays in America and Australia were generally much shorter (1 day–1 week) versus European studies (4–6 weeks) (215).

Following this review, one RCT has been published evaluating SEMLS in 19 children with spastic diplegia, mean age of 9 years and 8 months (225). The control group underwent a program of progressive resistance strength training and then surgery at 12 months. At 12 months, the surgical group had a 34% improvement on the Gait Profile Score and a 57% improvement on the Gillette Gait Index, while the control group had a small, nonsignificant decline on both measures (225). There was no difference in the groups on the GMFM-66, the Functional Mobility Scale, time spent in the upright position, and health-related QOL on the Child Health Questionnaire (CHQ; 225).

Orthopedic surgery is ideally delayed until the age of 4 to 7 years, due to the high risk of recurrence of tightness and contracture formation in younger children (217,221,226). In a retrospective study, a recurrence rate for Achilles tendon lengthening was found in 18% of children with diparesis and 41% with hemiparesis (226). Children older than 6 years of age at the time of initial operation do not typically have recurrence (226). This finding of increased risk of recurrence rate at younger age of surgery was confirmed by the results of a systemic review of surgical correction of equinus deformity in CP (227). The authors of this review also reported a greater incidence of calcaneal gait and deformity in children with diplegia versus hemiplegia (227).

Postoperative care should include aggressive pain management to minimize pain-related muscle spasms, which may further increase discomfort. Rapid mobilization with minimal casting is also recommended, usually with only a 2- to 3-day period of recumbency following surgery. Although not well studied, there is support for likelihood of increased weakness following SEMLS in children with CP (228). The need for physical therapy should be assessed and started as soon as possible to minimize postoperative weakness and disuse atrophy, as well as improve muscular reeducation and training in those muscles or tendons that were manipulated. Although strengthening exercises have been shown to increase strength and function in patients with CP during the first postoperative year, there still may be residual increased weakness and loss of function postoperatively, in spite of improved gait kinematics (228). It is possible that a more rigorous rehabilitation program following surgery could facilitate an improvement in function, a result that is uncommon in the current literature.

Scoliosis is common in CP. Indications for surgical management may vary between centers, but, in general, curves greater than 40 degrees in skeletally immature persons and greater than 50 degrees in skeletally mature persons are recommended for evaluation and consideration of possible fusion surgery (229). Goals of surgical intervention include alleviation of pelvic obliquity, achievement of a balanced spine, maintenance of seating ability, and minimization of pain (230). Before pursuing spinal fusion, the child should receive careful preoperative evaluation and preparation, including close monitoring of nutrition and respiratory status, in order to reduce postoperative complications. For young children, bracing may be used to slow the progression of the curve until optimal timing for a definitive surgical procedure (230) and to facilitate balanced sitting and functional use of the upper extremities.

ORTHOSES

Many children with CP utilize orthotic devices for maintaining or increasing ROM, protection, or stabilization of a joint, or promotion of functional activity. Orthoses can be expensive, and with a wide variety of designs to choose from, care should be taken to provide the appropriate design to meet the child's needs. Dialogue between the child and family, the physiatrist, the orthotist, and the therapist is essential when deciding on the treatment goals of the orthoses. When an orthotic is prescribed, it is important for the care team to communicate to the family the rationale for the orthotic, under what condition it should be worn, and for how long in each 24-hour period (231).

Upper Extremity Orthoses

Static wrist–hand orthoses (WHOs) are commonly used in CP to improve hand position for functional activities and to maintain ROM. Dynamic WHO are much less commonly used because children are often reluctant to use them for functional activities, in part due to the decreased sensory feedback caused by the orthosis. There is also concern that they will produce the unwanted effect of inhibiting joint movement and thus muscle activity, which in turn may limit the engagement of neural pathways and neuroplasticity (232). The use of either type is not well studied in CP, but a systematic review and meta-analysis of the available literature (six studies) concluded that the moderate-quality evidence indicated a small benefit of nonfunctional hand splints plus therapy on upper limb skills over therapy alone and results were diminished after splint wearing stopped (232).

Lower Extremity Orthoses

Many different types of LE orthoses are utilized in the management of CP, including supramalleolar orthoses (SMOs), solid ankle foot orthoses (AFOs), hinged AFOs, posterior leaf-spring AFOs, and ground-reactive AFOs. Knee-ankle-foot orthoses and hip-knee-ankle-foot orthoses are rarely used in CP. In spite of many published studies on the effectiveness of LE orthotics in CP, precise indications have yet to be established. A systematic review of 27 studies resulted in the following recommendations: (a) only orthoses that extend to the knee and have a rigid ankle, leaf-spring, or hinged design with a plantarflexion stop can prevent equinus deformities; (b) SMO designs with tone-reducing features (or dynamic AFOs) do not prevent equinus; (c) preventing plantarflexion or equinus has been shown to improve the temporal parameters of gait, such as walking speed and stride length for the majority of children, and thereby improved gait efficiency; (d) children with less severe impairments often performed better on stairs and moving from sitting to standing in less restrictive hinged, leaf-spring, or SMO designs (233).

Rotational-control orthoses, both twister cables and rotation straps, are also used occasionally in children with CP. Twister cables have a pelvic band with attached cables of twisted spring steel, with torque typically

applied to provide an external rotation force by attaching to the shoes or AFOs. Rotation straps are elastic and attach to buckles on AFOs or to an eyelet attachment on shoestrings, and can provide internal or external rotation forces depending on the application of wrapping the straps around the lower extremities. While these orthoses can help to control rotation, especially in younger children, families often complain that they are cumbersome and often prefer not to use them.

Spinal Orthoses

The role of spinal orthoses in children with CP and scoliosis has not been well studied. There are no RCTs, and there is no agreement as to whether spinal orthoses can prevent the progression of scoliosis. There is general agreement that if bracing controls the progression of scoliosis, it will not work in every patient (234) and it at best is only likely to slow progression, delaying surgery until a more ideal time (235). Regardless of its effect on curve progression, a positive effect on sitting stability and function has been reported by parents and caregivers (236), but this also has not been well studied.

ADAPTIVE EQUIPMENT

The goal for the use of adaptive equipment is to improve positioning either in the supine or sitting position, or to improve the level of function in self-care skills, including in the home, school, or community. These devices include, but are not limited to, seating or support systems, mobility devices, augmentative communication devices, computer or computer aids, and environmental control devices. A team, including physicians and therapists to assess physical capabilities, as well as to develop and refine appropriate goals, is essential to address and optimize adaptive equipment needs for children with CP.

COMPLEMENTARY AND ALTERNATIVE THERAPIES

The use of complementary and alternative medicine (CAM) in CP is not uncommon. CAM has been defined by The American Academy of Pediatrics as "strategies that have not met the standards of clinical effectiveness, either through randomized controlled clinical trials or through the consensus of the biomedical community" (237). It is not surprising that caregivers would be attracted to therapies that promise significant functional improvement when traditional medicine may appear to have little to offer. CAM is more commonly used in children with chronic diseases, such as CP, despite lack of substantiating evidence (238). CAM is often used in addition to orthodox medicine, but often its use is not discussed with the child's treating physician secondary to a feared negative response (238). Several studies have documented increased use of CAM in children placed in higher GMFCS categories (239,240). One study found that 56% of families surveyed had utilized at least one CAM therapy for their child with CP (239). The most commonly utilized therapies were massage therapy (25%) and aquatherapy (25%). The most significant predictors of use were the child's age (younger), lack of independent mobility, and parental use of CAM (239). Other CAM therapies utilized by children with CP include conductive education (CE), patterning, hyperbaric oxygen therapy (HBOT), Adeli suit therapy (AST), acupuncture, cranial osteopathy, and many others.

Hyperbaric Oxygen Therapy

Proponents of HBOT propose that "dormant areas" can be found surrounding injured areas in the brains of children with CP and that high levels of oxygen in the brain reactivate, or "wake up," the cells of this dormant area (241). Delivery of hyperbaric oxygen typically consists of treatments with pressures of 1.5 to 1.75 atmospheres for 1 hour per session, sometimes as often as five to six times per week, for up to 40 treatment sessions in a phase of treatment. A blind, randomized, controlled clinical trial of 111 children with CP compared treatment with hyperbaric oxygen at 1.75 atmospheres with a control group that received air at a pressure of 1.3 atmospheres (242). Both groups demonstrated significant functional improvements, but no differences were found between the groups. While some authors have argued that this demonstrates the value of elevated oxygen, even at minimal levels (243), others argue that the effect demonstrates a "powerful clinical trials effect" (244), with the effect primarily due to highly motivated parents spending many hours with the children in an intensive setting, knowing that developmental outcomes would be evaluated (241). A systematic review of the evidence revealed that there is inadequate evidence to establish a significant benefit of HBOT or for identifying potential adverse effects of HBOT in children with CP (245) and that adverse events are possible, and it is therefore not recommended (98).

Conductive Education

CE is a combined therapeutic and pedagogic program for children with CP, developed by the Hungarian child neurologist Andras Peto in the 1940s, which has been given increased attention in Western countries in recent years with the main elements being task-oriented learning within highly structured programs; facilitating and commenting on motor actions by rhythmic intending, for example, rhythmic speaking or singing; integration of manual abilities into the context of activities of daily living; and child-oriented group settings to facilitate psychosocial learning to increase the level of participation (246). In this program, the "conductor" is trained in

special education and therapy and administers the CE program. As CE has spread from Hungary to other countries, it has been packaged in an array of delivery models, making it difficult to ascertain specific criteria that define CE as a program (247). The use of adaptive equipment such as splints, walkers, and wheelchairs in the classroom is generally discouraged (241). An American Academy of Cerebral Palsy and Developmental Medicine (AACPDM) Treatment Outcomes Committee Evidence Report was conducted to evaluate the current state of evidence regarding CE and found that the present literature base does not provide conclusive evidence either in support of or against CE as an intervention strategy, primarily due to the limited number of studies and their weak quality (247).

Adeli Suit Therapy

AST was introduced in 1991 and incorporates a prototype of a device developed in Russia in the late 1960s to maintain neuromuscular fitness during weightlessness experienced by cosmonauts. The treatment is based upon three principles: the effect of the suit (working against resistance loads, increased proprioception, and realignment), intensive daily physical therapy for 1 month, and active motor participation by the patient (248). The suit consists of a vest, shorts, kneepads, and specially connected shoes; pieces of the suit are connected by hooks, rings, and elastic bands that are adjusted to optimally position limbs and joints. The bungee-like cords are adjusted by therapists to mimic normal flexor and extensor patterns of major muscle groups in an attempt to correct abnormal muscle alignment (249). The theory is that once the body is in proper alignment, aggressive movement therapy can be performed that will reeducate the brain to recognize correct movement of the muscles (249). It is also felt that deep pressure at the joints improves the sensory and proprioceptive information at that joint, enhances the vestibular system, and improves coordination (241). Treatment is typically given at a higher intensity, at 1 to 2 hours per day, multiple times per week, for a 4- to 6-week period. One randomized, controlled, clinical trial compared the efficacy of AST in children with CP to neurodevelopment treatment (NDT) (248). Both groups received the same intensity of treatment, totaling 20 sessions in 4 weeks, and were evaluated with the GMFM-66 at baseline, after 1 month of AST or NDT therapy and again 9 months later after they had returned to their baseline therapies. When administered with equal intensity, the AST did not show superior motor skills retention in comparison with NDT (248).

Cranial Osteopathy

Cranial osteopathy is otherwise known as craniosacral therapy. It said to stimulate healing by using gentle hand pressure to manipulate the skeleton and connective tissues, especially the skull and sacrum. It is based on a theory that the central nervous system, including the brain and spinal cord, has subtle, rhythmic pulsations that are important to health and can be detected and modified by a skilled practitioner. In a randomized, controlled trial 142 children with CP, aged 5 to 12 years, underwent either 6 months of cranial osteopathy or a waiting list with partial attention control. At 6 months there was no statistical difference in outcomes, which included a blinded assessment of the GMFM and the CHQ (250).

Additional Therapies

Patterning (Doman–Delacato method), hippotherapy, Feldenkrais, massage, and acupuncture are additional CAM therapies that are sought out by parents of children with CP. In regard to patterning, the American Academy of Pediatrics concluded that "patterning treatment continues to offer no special merit, [and] that the claims of its advocates remain unproved . . ." (251). There are a few uncontrolled and controlled studies revealing improvements in GMFM scores as well as other benefits in regard to decreasing muscle tone, improving head and trunk postural control, and developing equilibrium reactions in the trunk from hippotherapy (252–255). No published studies are available on the use of the Feldenkrais method in children with CP. There is conflicting evidence on the ability of massage to reduce pain, spasticity and improve function in children with CP and the quality of the evidence is low (98). Most studies published in English regarding acupuncture are uncontrolled and primarily case series.

The American Academy of Pediatrics Committee on Children with Disabilities published recommendations for counseling families on CAM, which include the following: maintaining a scientific perspective, providing balanced advice about therapeutic options, guarding against bias, and establishing and maintaining a trusting relationship with families (237). Ethically, families have the right to use alternative medicine therapies for their children as a matter of autonomy, but they also have the duty not to harm their children (256). The care of patients should be based, to the greatest extent possible, on existing sound evidence revealing that the therapy recommended is effective in reducing morbidity; the benefits outweigh the risks; the cost of the treatment is reasonable compared to its expected benefits; and the recommended therapy is practical, acceptable, and feasible (241).

COURSE AND PROGNOSIS

OUTCOME MEASURES

Children with CP often change over time, due either to growth and development or as a result of treatment.

Various means of determining change may be employed. Subjective evaluations that ask the child, parent, or therapist his or her opinion are most commonly used. Occasionally, more quantified techniques are employed, particularly in research settings, although clinical use also occurs.

Outcome measures may best be classified by the domains they seek to measure and the methods of assessment. Using the International Classification of Functioning, Disability and Health—Children and Youth Version (ICF-CY) (257), measures can be divided into those that define body structures, body function, activity and participation, and domains outside the ICF (Table 14.3), and this rubric is the focus of efforts to develop a core set of outcomes for CP (258). Few measures are pure assessments of only one domain; rather, it is common to see items spread across multiple ICF domains (259).

Body Structure and Function

When considering children with CP, few outcome measures directly relate to body structure. Imaging such as functional MRI or physiologic measures like transcranial magnetic stimulation or electromyography could be considered in this domain. Because very few interventions for CP are expected to alter body structures, such as brain tissue, these types of outcome measures are seldom employed. Many outcome measures for CP address body function. Body function is assessed with spasticity/movement disorder measurement (Ashworth, Modified Ashworth, and Tardieu scales, the Hypertonia Assessment Tool, or specialized measurement systems), strength measurement (muscle grading or dynamometry), or ROM.

Activity and Participation

Because many interventions for CP are intended to reduce activity limitation or promote participation, a wide range of outcome measures are specific for these ICF domains. Common assessments of gross motor function and walking include the GMFM and Gross Motor Performance Measure as well as gait analysis, ranging from observational scales (Physician Rating Scale) to instrumented digital kinematic analysis. Fine motor ability may be assessed with the Quality of Upper Extremity Skills Test, Assisting Hand Assessment, and Melbourne Assessment of Unilateral Upper Limb Function, among others. More global functional measures include the Functional Independence Measure for Children (WeeFIM), the Pediatric Evaluation of Disability Inventory, the Pediatric Outcomes Data Collection Instrument, and the Bruininks–Oseretsky Test of Motor Proficiency. Assessment of energy expenditure or efficiency, timed walking tests, and movement monitors are also used to assess the domain of activity in children with CP.

The Pediatric Evaluation of Disability Inventory (standard and computer-adapted test versions) and the Canadian Occupational Performance Measure assess both activity and participation realms. Participation for children with CP is most often assessed with the Children's Assessment of Participation and Enjoyment and the Preferences for Activities for Children. The Activities Scale for Kids and Assessment of Life Habits for Children are also employed in this domain. Some instruments address health status or QOL, and may be placed in the domain of participation, while other instruments assess environmental factors. Common outcome measures in this group include the CHQ, KIDSCREEN, or other generic pediatric measures, the Cerebral Palsy Quality of Life—Children, and Goal Attainment Scaling.

Developmental assessments are generally wide in scope and used more frequently in younger children. These include the Peabody Developmental Motor Scales, Battelle Developmental Inventory, Vineland Adapted Behavior Scales, Denver II, Bayley Scales of Infant Development, and Revised Gesell Developmental Schedule.

Three key arenas of assessment are discussed in the following.

Gross Motor Function Measure (GMFM)

The GMFM is a functional outcome tool that was developed specifically for use in CP (279). Widely used in research settings, the GMFM is also employed clinically for the evaluation of children with CP. The GMFM consists of a broad range of gross motor tasks, in which a trained evaluator observes a child attempting to complete over a 45- to 60-minute time interval. The item set and basal and ceiling approaches for the GMFM permit more rapid administration (296). Five dimensions of function (lying and rolling; sitting; crawling and kneeling; standing; and walking, running, and jumping) are examined. Specific scoring algorithms result in a score that can be used as an interval measure.

Gait Analysis

Instrumented gait analysis is another objective functional measure that is widely used in CP (283). Many centers do not use gait analysis; other centers rely upon it heavily, particularly in guiding treatment decisions such as orthopedic surgery. This technique can only be employed for children who have some ability to walk, even if they require gait aids. Gait analysis involves having a child walk in a specialized laboratory wearing markers and muscle activity sensors. Using sophisticated computers, cameras, and force plates implanted on the floor surface, the child's movement patterns can be analyzed in great detail. Information about movement patterns in all planes, kinetics, and kinematics is generated. Although some controversy exists as to the reproducibility of gait analysis results and the means by which gait analysis should be employed to guide surgical decision making (297), gait analysis remains a common tool for the evaluation of CP.

TABLE 14.3 OUTCOME MEASURES USED IN CP

	BODY STRUCTURE	BODY FUNCTION	ACTIVITY AND PARTICIPATION	QUALITY OF LIFE OR NON-ICF DOMAINS
Administered by questionnaire or self-report		Child Health Questionnaire (260) PedsQL (261)	Pediatric Outcomes Data Collection Instrument (262) PedsQL (261) Pediatric Evaluation of Disability—CAT (263) Children's Assessment of Participation and Enjoyment and Preferences for Activities for Children (264) Assessment of Life Habits for Children (265)	Cerebral Palsy Quality of Life—Child (266) PedsQL (261) Child Health Questionnaire (260)
Measured by a trained investigator or with specialized equipment	Functional MRI (267) MRI (36) Diffusion tensor imaging (268) Transcranial magnetic stimulation (269) Positron emission tomography (PET) scan (270)	Spasticity [Ashworth Scale (271), Modified Ashworth Scale (272), *Tardieu Scale* (273), specialized systems (274)] Strength [muscle *grading* or dynamometry (275)] *Hypertonia Assessment Tool* (276) Range of motion (277) Electromyography (278)	Gross Motor Function Measure (279) Gross Motor Performance Measure (280) Gait analysis [observational *scales* (281,282) *to instrumented digital analysis* (283)] Quality of Upper Extremity Skills Test (284) Assisting Hand Assessment (285) Melbourne Assessment of Unilateral Upper Limb Function (286) *Functional Independence Measure for Children (WeeFIM) (287)* Pediatric Evaluation of Disability Inventory (288) Bruininks–Oseretsky Test of Motor Proficiency (289) Energy expenditure/efficiency (290) Movement monitoring (291) Timed walking (292) Canadian Occupational Performance Measure (293)	*Goal Attainment Scaling* (294) KIDSCREEN (295)

Note: Italicized measures are commonly used in clinical settings and may also be used for research, whereas other measures are predominantly research tools.

QUALITY OF LIFE

Children with CP experience limitations in mobility and are at risk for lower participation in leisure and social activities, and therefore, there is a perception that they have a lower QOL. The World Health Organization (WHO) defines QOL as "an individual's perception of their position in life in the context of the culture and value systems in which they live, and in relation to their goals, expectations, standards and concerns" (298). QOL is, by definition, subjective, yet most of the literature to date looking at QOL in CP has used data from parents rather than the children themselves. The literature also has tended to focus on functional skills and their role in QOL, and little attention has been paid to other important contextual factors such as environment and family functioning, which are felt to be important determinants of QOL (299).

Particular interest focuses on child self-report of QOL and contextual factors outside of physical functioning. In a population-based study of 217 children with CP aged 6 to 12 years, the authors found that the QOL was highly variable, but about half experienced a QOL similar to typically developing children (299). Children were less likely to rate themselves low for psychosocial well-being when compared to their parents' report. In general, parent and professional assessments are often similar, but may vary from child reports (300). Functional limitations were good indicators for physical but not psychosocial well-being, and family functioning, behavioral difficulties, and motivation were all found to be important predictors of social-emotional adaptation.

Other studies of self-reported QOL found no difference in QOL between children with CP and typically developing children (301–303). One of these studies was a large population-based study of 1,174 children between 8 and 12 years in Europe (301), one looked at a convenience sample of 81 children 10 to 13 years with GMFCS levels I to III (302), and the third compared children with CP with a range of other children with and without chronic conditions (303). The finding that many children perceive their QOL as similar to their peers is encouraging and suggests that children who grow up with an impairment incorporate it into their sense of self from birth and it is possible for them to embrace growth, development, and living with the same excitement as typically developing children (301). Future large population-based and longitudinal studies would be helpful to validate these findings and to look more closely at the contextual factors that may affect QOL and determine potential changes in QOL over time.

PROGNOSIS FOR AMBULATION

Shortly after caregivers are given the diagnosis of CP, they will often want to know if their child will walk. Many studies have been published on this subject, and the best predictors of eventual ambulation appear to be persistence of primitive reflexes, gross motor development, and type of CP. The persistence of primitive reflexes or the absence of postural reactions at age 2 years is associated with a poor prognosis for ambulation (304). A longitudinal study of 233 children with mixed types of CP found that all of the children who were sitting by the age of 2 years eventually ambulated and that only 4% of the children who were not sitting by 4 years ever gained the ability to ambulate (305). Prognosis for eventual ambulation is also closely related to the type of CP. Children with spastic hemiparesis have the best prognosis for ambulation, with nearly 100% achievement. More than 85% of children with spastic diparesis will eventually ambulate. The likelihood for ambulation is much less with spastic quadriparesis, but the studies have revealed a wide range of eventual ambulation of 0% to 72% (304). This wide range is likely due to differences in the population of children studied and the definition of ambulation that was used. The presence of severe intellectual impairment also is a poor predictor for walking. A large population-based study in Europe found that a severe intellectual impairment increased the risk of being unable to walk 56 times in hemiplegic CP and 9 times in bilateral spastic CP (306). If one takes into account all of these potential predictors, it is possible to make a relatively accurate prognosis for ambulation by the age of 2 to 3 years. This will help the child's caregivers set realistic goals and guide appropriate therapeutic intervention.

AGING WITH CEREBRAL PALSY

The United Cerebral Palsy Association has estimated that there are approximately 400,000 adults with CP living in the United States (307). It is expected that this number will grow due to improvements in medical care. A number of studies have published data on the life expectancy of persons with CP. A population-based Health Surveillance Registry in British Columbia was utilized to study a cohort of 3,189 persons with CP born between 1952 and 1989 (308). The overall survival rate at 30 years was estimated to be at least 87%. Intellectual disability and epilepsy were determined to have a negative effect on survival. The projected life expectancy of children who currently have CP is unknown, as these surveillance studies are based on medical practices from previous decades.

Musculoskeletal symptoms are commonly identified complaints in adults with CP, even at a relatively young age. Issues that are commonly identified include cervical pain, back pain, and hand paresthesias (307). Other concerns include maintenance of mobility, availability of adaptive aids, incontinence, and lack of appropriate preventative medical care (307). Systematic reviews found that mobility declines in at least 25% of adults with CP (309) and muscular strength, muscular endurance, and

cardiorespiratory endurance were all significantly lower in adults with CP compared to able-bodied peers (310). A large number of adults with CP do not obtain regular general health evaluations or rehabilitative care. This is largely due to the lack of adult physicians with an interest and knowledge of medical issues in persons aging with CP and the lack of an organized system of care similar to what is currently available for children with CP.

Information on education and employment in adults with CP is limited. Reported competitive employment rates vary from 24% (311) to 53% (312). The only population-based study took place in Europe and found 33% of young adults participated in higher education (vs 77% of controls) and 29% were competitively employed (vs 82% of controls) (313). Proposed reasons for lower education and employment rates include impaired cognition, employment policies, inadequate accessibility, attitudes toward individuals with CP in the workplace, and impaired social functioning (313). Vocational rehabilitation services positively predict employment outcomes and should be recommended to maximize the employability of adults with CP (314).

It is clear that more attention needs to be paid to issues related to aging in CP. Adult medical care providers need to be identified and educated, and a routine means of transitioning care needs to be in place. An early emphasis should be placed on independent living skills. Adaptive equipment needs to be routinely reassessed for its appropriateness. Adults with CP need to be aware of the community support services available to them and learn to advocate for themselves. Active vocational counseling should begin in high school. Hopefully, the growing awareness of this population will lead to improved QOL and increased functional independence.

PEARLS AND PERILS

1. Although prematurity is a major risk factor for CP, most children with CP were not premature infants.
2. Hand preference prior to the age of 18 months may be an indication of hemiparetic CP.
3. Additional workup for an etiology other than CP should be undertaken in any child who has lost developmental milestones.
4. Sensory impairments, especially in hemiparesis, can be an important contributing factor to decreased functional hand use.
5. Children with severe motor impairments related to CP can have normal cognition.
6. Periods of rapid growth in children with CP may be associated with worsening contractures because spastic muscles fail to grow as quickly as bones.
7. Children who have a sudden increase in spasticity should be evaluated for constipation, urinary tract infection, esophagitis, musculoskeletal pain, or other potential sources of noxious stimulation.
8. Oral baclofen should be titrated up slowly to minimize sedation and titrated off slowly to minimize the likelihood of withdrawal symptoms, including increased tone and seizures.
9. When evaluating toe walking due to equinus, always evaluate and address spasticity and contractures of more proximal muscles, in particular, the iliopsoas and the hamstrings.
10. Not all children who walk on their toes have CP. Toe walking can also be idiopathic or due to proximal muscle weakness, as is the case with Duchenne muscular dystrophy.
11. Children with CP who sit independently by age 2 years are likely to be functional ambulators, while those who fail to walk by age 4 years are unlikely to be functional ambulators (305).

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15

SPINA BIFIDA

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Spina bifida is the second most common disability in children. The National Spina Bifida Association (SBA) documents more than 70,000 individuals in the United States living with spina bifida. This is a small fraction of all those affected worldwide. Spina bifida is a complex disorder that has physical, psychological, and social implications. Medical professionals treating these individuals should have a thorough understanding of the spectrum of the disability.

EPIDEMIOLOGY

According to estimates by the Centers for Disease Control and Prevention (CDC), spina bifida and anencephaly, the two most common neural tube defects (NTDs), affect approximately 3,000 pregnancies yearly in the United States. NTDs vary in prevalence, depending on race and ethnicity, with women of African American and Asian descent having the lowest, while women of Hispanic ethnicity having the highest.

It is notable that affected pregnancies in both Hispanic and non-Hispanic Whites have declined significantly since the mandatory fortification of grain products in the United States with folate (see the discussion in the section "Genetic Influences"). Folic acid fortification can substantially decrease a woman's risk of an NTD-affected birth. Although the impact of folic acid on NTDs does not appear to be influenced by race or ethnicity, the disparity between Hispanics and other races and ethnicities remains, and the causes are unknown.

It is hypothesized that differences in eating habits, supplement-taking practices, and in general an awareness of how nutrition affects pregnancy outcomes have a major impact on the developing fetus. Studies have demonstrated also that other risk factors such as maternal obesity, nutrient intake, and supplement use affect racial/ethnic groups differently. Genetic factors can have a major direct impact on NTD incidence and serve to alter susceptibility to numerous environmental influences. It is this interaction that requires continued study in order to discern how racial and ethnic factors change over time and affect NTDs.

A study of spinal defect repairs from 1988 to 2010 showed a decrease in surgical correction; however, more operations were performed per 10,000 births for Hispanic patients as compared to Caucasian or African American members of this cohort (1). It has been suggested that folate fortification of corn masa flour may improve the known higher rate of myelomeningocele in the Hispanic population due to the prevalence of this flour's use by this population (2).

The current American Academy of Pediatrics (AAP) guidelines for folic acid supplements are:

- All women of childbearing age: 400 µg or 0.4 mg/day.
- Women who have spina bifida, or a female with a previous NTD pregnancy: 4,000 µg or 4 mg/day 1 to 3 months prior to conception and through the first trimester. This reduces the risk by 70% as per the SBA 2014 recommendations (3).
- High-risk pregnancies (such as a mother who is taking valproic acid or has maternal diabetes): 4 mg/day (4).

Understanding the multifactorial etiology of this disorder is complex, and even though much is known about the genetic errors associated with folate metabolism, other underlying genetic risks are still to be elucidated. Although incidence and recurrence risk vary around the world, most children are born to families without a prior affected child (0.1%–0.2%). The risk for recurrence in a family with one child with NTD is 2% to 5% and increases to 10% to 15% if two siblings are affected. If one parent has spina bifida, the risk of having a child with a similar disorder is 4% (5).

ETIOLOGY

It is important for the clinician to understand the embryogenesis of NTDs. The two forms of defects, open and closed, can occur at any point along the neuraxis with varying prevalence. Closed forms (encephalocele, meningocele, iniencephaly, lipomeningocele, sacral agenesis, and occult spinal dysraphism) and open forms (anencephaly, and myelomeningocele) may result from differing yet overlapping factors (6). Spina bifida is a complex, heterogeneous disorder whose etiology in humans appears to be multifactorial. However, spina bifida, classically defined as myelomeningocele, is the consequence of neural tube closure failure during embryonic development. The following section discusses the normal central nervous system (CNS) embryogenesis and the pathologic differences associated with these neural tube defects (7–10).

NORMAL DEVELOPMENT

During the first 2 weeks, postfertilization embryonic development involves repeated cell division and organization, resulting in a blastocyst, an embryo with two layers: the epiblast and the hypoblast. At the end of this period, on days 13 to 16, a primitive streak forms, beginning caudally and progressing toward the rostrally located prochordal plate initiating the rostral-to-caudal orientation of the embryo.

The development of the primitive streak is followed by invagination of epiblast cells, which remodels the embryo (ie, gastrulation) into a three-layered structure comprising ectoderm, mesoderm, and endoderm, the precursors of all tissue types and body structures. As the primitive streak regresses, presumptive notochord cells migrate through Hensen's node aligning themselves along the midline of the embryo between the endoderm and the ectoderm.

In humans around day 16 the overlying ectoderm is induced by cell-to-cell contact with the notochord, thereby forming the neural plate. By about day 21, the plate bends as the groove deepens, and its walls and their adjacent cutaneous epithelium begin to oppose one another, beginning the formation of the brain and spinal cord.

The eventual closure of the neural tube proceeds over a period of 4 to 6 days and typically involves primary closure of the cutaneous ectoderm. This is followed by the neuroectoderm, which then separates from the overlying cutaneous ectoderm, resulting in a closed tube. Closure begins at a point just caudal to the developing rhombencephalon and proceeds via several waves rostrally and caudally rather than in the continuous "zipper-like" fashion previously envisioned. There is, however, an alternative view proposed by Van Allen and colleagues that describes several closure initiation sites over the same period of time (11,12). Regardless, closure of the primary neural tube is typically complete around embryo developmental day 27. This process, primary neurulation, completes the presumptive spinal cord down to the lower lumbar and/or upper sacral levels.

A secondary wave of neurulation begins around day 25 from a collection of remaining primitive streak cells and mesoderm located along the midline axis from the caudal end of the primary neural tube to the cloaca. Cavities form that coalesce into a tube, which eventually becomes continuous with the primary neural tube. This process completes the formation of the sacral levels of the spinal cord and terminal filum, and is species-specific. Understanding the process of primary and secondary neurulation is of paramount importance in comprehending the pathogenesis of spina bifida. The failure of primary neurulation is known to be associated with open spinal dysraphisms such as myelomeningocele, craniorachischisis, or anencephaly. Disruption of secondary neurulation, the mechanism for the development of the caudal spinal cord, is associated with closed spinal defects (eg, spina bifida occulta) and spinal cord tethering (13,14). The process of neurulation is completed by the end of the first month of embryonic development.

Expansion of cranial brain structures via the development of a primitive ventricular system is thought to be accomplished by temporary occlusion of the caudal (spinal) neural tube on days 23 to 27, which creates a rostral-enclosed fluid-filled space. This provides pressure to expand the cranial lumen and is the impetus for brain enlargement. Theory suggests that, in part, failure of this expansion pressure is a cause for Chiari malformation (15) seen in spina bifida.

PATHOLOGY

Spina bifida is typically considered to be the result of a primary failure of neurulation. Failure of neurulation and, thereby, loss of neural tube closure, prevents the mesoderm, adjacent to the notochord, from forming muscle and bone (ie, via somitic mesoderm). This normally forms around the tube to protect it. Therefore, the mechanisms involved in this process are suspected to be involved in the pathology of this disorder. Although this is the most popularly accepted theory, there are other proposed mechanisms. Several forms of myelomeningoceles are not failures of neurulation, but a failure of Henson's node to lay down the notochord correctly—in other words, a failure in gastrulation (16,17) that causes significant errors in the induction of the neural tube. Further research is necessary to verify current proposed theories.

The mechanisms by which the neural tube is formed and closed are varied. Morphogenic changes in cell populations such as wedging and planar cell polarity result

in the creation and shaping of the neural plate into a tube-like structure early on (18,19,20–22). Several other mechanisms are proposed for the closure of the neural tube, such as an interaction between various glycoproteins and cell adhesion molecules, various signaling protein/receptor interactions, the interlinking of numerous cell filopodia, and formation of intercellular junctions. The current view suggests the process likely involves all of these and perhaps others not yet visualized. Failure of any of these processes could therefore negatively impact CNS development.

GENETIC INFLUENCES

Genetic mutations have a significant impact on CNS development, as demonstrated experimentally in rodents and documented clinically in humans. Alterations in the genes that affect metabolism, nucleotide synthesis, cell programming, and cell-to-cell signaling can all affect aspects of neural development. This ranges from the signaling aspects of the induction of neural tube formation initiated by the notochord to alterations in apoptosis (programmed cell death).

Induction of the neural plate is controlled by a variety of genes. Sonic Hedgehog (*SHH*) is a vertebrate gene expressed by cells within the notochord that—in conjunction with the Patched (*PTC*) gene—produce proteins involved in the induction of the floor plate during embryogenesis (23–25). During embryogenesis neuronal subtypes such as motor neurons proliferate, and somite development begins. The expression of various signaling proteins on the surfaces of cells allows for the sequential transmission of signals regulating the cell's fate.

PTC is a gene that functions downstream of *SHH*. Its function is hypothesized to serve as negative feedback to *SHH*, thereby regulating the induction of numerous cell types in the developing neural tube. Failure of this system and its feedback loop and/or overexpression of one portion of the process may lead to neural tube development failure. Although not currently implicated by empirical data, research is in place to elucidate the impact of this system on human NTDs.

Alterations in genes that impact the movement of epithelial and mesenchymal cells that create and shape the body axis have also been suggested to be associated with NTDs. Planar cell polarity pathway genes such as *SCRIB* and *PRICKLE* may contribute to the multifactorial risk for spina bifida in humans (18,19).

Genes that regulate folate metabolism and methyltransferase reactions associated with methionine and homocysteine metabolism are both of major interest (26). Folate serves as a cofactor for enzymes that participate in nucleotide synthesis, and is important in the methylation processes. Evaluations of folate levels of mothers with NTDs shortly after birth have produced equivocal results, suggesting that either absolute folate deficiency is rare or that their diet and folate level may have been different at the beginning of the pregnancy. However, disturbances in the metabolic pathways that utilize folate may predispose to NTDs. This cannot be corrected with folate supplements. Metabolism of folate and homocysteine are interdependent, and the risks associated with alterations in their metabolism are thought to be connected. Elevated homocysteine levels in pregnant women are a known risk factor for NTDs. Mutations/polymorphisms in the enzyme 5,10-methylenetetrahydrofolate reductase (*MTHFR*) have been associated with diminished plasma folate levels, with commensurate elevated homocysteine levels. These alterations have been identified in patients with spina bifida as well as in their parents. In addition, using cultures of fibroblasts from NTD-affected patients, homozygosity for defects in the *MTHFR* gene have been shown to have a 7.2-fold increased risk for NTDs. The prevalence of these defects appears to vary with race. Homozygosity for the *C677T MTHFR* mutation is a known risk factor for upper-level spina bifida lesions in those of Hispanic descent. The *MTHFD1 1958G>A* polymorphism is also associated with NTDs in those of Irish descent. Although folate supplementation has proven to be very successful in attenuating the prevalence of NTDs in various populations worldwide there is a portion resistant to such supplementation. Animal models show folate resistant NTDs are sensitive to the intake of inositol. Limited human experience has also suggested folate resistant NTDs may be amenable to inositol supplementation (27,28).

In early stages of nervous system development, more cells are produced than needed and that the processes of apoptosis and autophagy are coordinated during development to yield a well-defined and functioning CNS. Apoptosis, the most studied of these processes, is modulated by various members of the *Bc12* gene family, the caspase family of cysteine proteases, and other genes that produce necessary intermediator proteins. The variation of genetic expression controls the development of cells within the CNS. Several mouse models have shown altering the expression of these genes (ie, knockout experiments) results in NTDs similar to that identified in humans. Such evidence strongly suggests their involvement in human neural tube pathology. Autophagy, an autodegenerative cell process, has a significant impact on the recycling of cellular components in the cytoplasm as a result of cellular organelle damage. This process can also be affected by nutritional stresses. A variety of genes that affect this process have been investigated using mouse models. Loss of Beclin 1 and Ambra 1 expression has been noted to result in overgrowth of the developing CNS. Therefore, identifying the human equivalent of these and other similar genes could yield information as to the cause of various NTDs and provide information for new therapeutic targets.

ENVIRONMENTAL INFLUENCES

The external environment has a significant impact on embryonic development and the incidence of NTDs (29). Hyperthermia during early pregnancy, the first 28 days during neurulation, has been shown to increase the incidence of NTDs. Specifically, maternal febrile events as well as sauna and hot tub use have increased the risk of NTDs (30–34).

Parental occupation has been demonstrated to have a definitive influence on the risk for NTDs. Increases in NTD risk have been noted for occupations involving exposure to solvents (eg, painters, industrial process workers, etc.). Health care, agricultural, and transportation workers have also been noted to have an increased NTD risk (35). The etiology of this occupational increase in NTD risk can only be hypothesized at this time.

Nutritional influences have a broad impact on NTD risk and interact in many ways with environmental as well as genetic influences. As indicated earlier in this chapter, a primary example is folate metabolism. Folate is a cofactor for the enzymatic process involved in purine and pyrimidine synthesis and facilitates the transfer of methyl groups during the metabolism of methionine and homocysteine. Taken together, alterations in these folate-sensitive processes can have an impact on cellular proliferation. Lowered intake of foods containing folate in the diet is associated with an increase in NTD risk. Disorders of absorption of folate in the intestine can alter folate levels and potentially increase NTD risk. However, studies of folate receptor carrier densities in the intestines of women with NTD offspring or their progeny do not have abnormally low receptor levels (36). Studies have shown that supplementation can decrease prevalence. Mandatory supplementation of folate in grain products in the United States has caused a steadily declining NTD incidence since initiated in 1996. Since the introduction of mandatory folate supplementation, the number of NTD affected pregnancies has declined from approximately 4,000 to 3,000 per year. Studies have shown the NTD recurrence risk can be decreased by approximately 50% by taking the recommended folate supplementation.

The risk for NTDs varies for couples, depending on whether there is a prior history of such defects. U.S. couples with a prior history of NTD births have an increased risk for recurrence (2%–5%) (5). Because of this, the U.S. Public Health Service and the CDC have two separate recommendations for supplementation based on prior NTD histories. Elevated supplementation is appropriate for couples with a prior NTD birth. Limited studies have also identified zinc deficiency as a nutritional entity that can also elevate NTD risk. It was discovered that women with the genetic disorder of zinc metabolism acrodermatitis enteropathica are at high risk for NTDs and that supplementation can lower those risks.

Maternal obesity and concomitant diabetes are associated with increased NTD risk. Specifically, women with a prepregnancy body mass index (BMI) suggestive of obesity (>29 kg/m) are more inclined to have offspring with NTDs. There appears to be a direct relationship between maternal BMI and spina bifida (37).

This holds true for women with diabetes, although the etiology of this association may be linked to alterations in glucose metabolism during organogenesis. It is notable that experimentally manipulated glycosylation in rodents results in birth defects not unlike those seen in children born to mothers with diabetes. Risks for NTD-affected births have been estimated at 2% in the United States and as high as 7% in England. These risks include spina bifida as well as other significant NTDs such as anencephaly. The NTD recurrence risk for mothers with diabetes in the U.S. is around 4%, which is similar to that found in mothers without diabetes.

Teratogenic influences from the environment, such as the consumption of prescribed drugs, have been associated with NTDs, particularly myelomeningocele (38). Valproic acid taken for seizures during pregnancy has been shown to increase the incidence of NTDs. Valproic acid appears to work by disrupting folate metabolism, thereby inhibiting neural tube closure. An alteration in folate-dependent methylation of regulatory proteins is theorized to be the cause. Regardless, administration of folate during pregnancy counteracts valproic acid associated NTDs.

Of note, the 2014 edition of the British HIV association guidelines indicate nonnucleoside reverse transcriptase inhibitors such as efavirenz should be acceptable for HIV infected women wishing to conceive (39). Earlier limited studies suggested against the use of antiretroviral agents such as efavirenz due to the apparent risk of NTDs; however, a meta-analysis by Ford and colleagues did not find an increased risk of overall birth defects associated with first trimester use (40). The authors caution due to small sample size and relative low NTD incidence, conclusions should receive further prospective evaluation. World Health Organization (WHO) and U.S. guidelines advise against first trimester use but can be considered later in pregnancy (41).

Drug-induced interference with DNA synthesis during embryonic development would likely have an impact on gastrulation and neurulation. Other drugs are also associated with NTDs, such as isotretinoin (Accutane), used for acne treatment; and etretinate (Tegison), a psoriasis treatment; and anticancer agents such as methotrexate. Consumption of alcohol and coffee and cigarette use have been suggested to increase the risk of spina bifida. However, a recent study has reported confounding data (42). Fetal alcohol syndrome has an association with increased risk for abnormal CNS development, including NTDs. Ethanol-induced impairment of polyamine homeostasis has been suggested as a potential cause of NTDs and intrauterine growth restriction in fetal alcohol syndrome (43).

Some chromosomal disorders that have multivariate etiologies and presentation are known to be associated with increased NTD risk. Trisomy 21 (Down syndrome) and trisomy 13 (Patau syndrome) are notable examples. Although the incidence is relatively small, studies have shown various NTDs, including spina bifida but not anencephaly, have been found on the autopsy of definitively karyotyped infants. Down syndrome is associated with genetic polymorphisms involved in homocysteine/methionine methylation (see the previous discussion on folate metabolism) and has a noted NTD familial clustering.

PRENATAL SCREENING

Prenatal screening is recommended for pregnant women to detect not only NTDs but also to screen for Down syndrome and other disorders. Prenatal diagnosis of spina bifida is important for families. It allows for decisions regarding continuation of the pregnancy, prenatal interventions, choice of appropriate birth facility, and preparation of parents and other family members for the birth to be made. Many parents choose to continue the pregnancy, although in a review of 17 studies Johnson and colleagues found that the overall frequency of termination of pregnancy ranged from 31% to 97%, and was more common for diagnosis confirmed before 24 weeks' gestation, with defects of greater severity, and in Europe versus the U.S. (44).

Prenatal diagnosis is generally carried out by one of several methods (45–48). A quad screen blood test is done in the second trimester. The test includes alpha fetoprotein (AFP), human chorionic gonadotropin (HCG), estriol, and inhibin A. Elevated levels of AFP suggest an NTD is present, and the need for further testing is indicated. The serum AFP test is not specific for spina bifida, however, and requires the gestational age to be correct. A study of open NTDs found maternal serum biochemical markers in the first trimester did not distinguish an open spina bifida. Further testing is required, using high-resolution ultrasounds (US) and amniocentesis (49). The fetal US helps identify characteristics both of the brain and the spine, such as an abnormal shape of the head, enlarged ventricles, or abnormal vertebrae. US can detect a splaying of the pedicles and the classic "lemon and banana signs." The lemon sign relates to the shape of the head, and the banana sign is related to herniation of the cerebellar vermis through the banana-shaped foramen magnum. Amniocentesis can also be performed, typically at 15 to 20 weeks of gestation, to identify abnormally high levels of AFP. Fetal MRI, after diagnosis, is made to determine the extent of the defect.

Prenatal diagnosis allows for the ability to plan ahead. Fetal surgery has been available for families on a research basis through the Management of Meningomyelocele Study (MOMS) program comparing surgery before (in utero) and the standard surgery after birth. A discussion of in utero closure is beyond the scope of this chapter, but the procedure is carried out only in specialized settings. The MOMS study is ongoing, but not recruiting participants at the time this goes to print. Please refer to the website: http://clinicaltrials.gov/show/NCT00060606. Differences of opinion exist as to the preferred delivery method, vaginal or cesarean section (previously preferred method of delivery). The delivery should occur at a high-risk center with access to a neonatal intensive care unit and a full range of specialists, in neurosurgery and urology.

CLINICAL TYPES OF NEURAL TUBE DEFECTS

SPINA BIFIDA OCCULTA

- Bony defect with no herniation of meninges or nervous elements
- Incidental finding in 5% to 36% of adults; a small percentage can develop clinical findings (50,51)
- Can be associated with pigmented nevus, angioma, hairy patch, dimple, and dermoid sinus
- Usually found in the lumbosacral/sacral segments
- Can have associated tethered cord with development
- May have bowel and bladder involvement
- No hydrocephalus or Chiari malformation

SPINA BIFIDA CYSTICA

- Bony defect with herniation of spinal canal elements
- Meningocele herniation of the meninges, but does not contain neural tissue
  - Usually normal neurologic exam
  - No association with hydrocephalus or Chiari malformation
  - Uncommon: occurs less than 10%
- Meningomyelocele herniation of meninges and neural elements
  - Most common
  - Associated with hydrocephalus and Chiari type 2 malformations
  - Abnormal motor and sensory exam
  - Neurogenic bowel and bladder
  - 75% in the lumbosacral segment

CAUDAL REGRESSION SYNDROME

- Absence of the sacrum and portions of the lumbar spine
- Associated with maternal diabetes
- Associated findings include syringomyelia, anorectal stenosis, renal abnormalities, external genital abnormalities, and cardiac problems
- Motor and sensory abnormalities

CLINICAL SIGNS AND COURSE

The spinal cord defect associated with spina bifida is often associated with other malformations. This results in a multisystemic process that leads to a variety of health problems and potentially life-threatening complications. Motor and sensory deficits vary according to the level and extent of spinal cord involvement (52–54).

In the care of individuals with spina bifida, two levels are often described: the anatomic level of the lesion and the neurologic level of functional involvement. In terms of the level, it is the neurologic or functional level that gives health care providers prognostic information with respect to long-term expectations and functional outcomes. Spinal cord involvement may result in asymmetric motor and sensory deficits. Sensory deficits usually follow a dermatomal pattern and may not affect all sensory modalities equally (52,53).

Neurogenic bladder and bowel dysfunction may be present in all patients because of the distal level of innervation of the bladder and bowel. This is true even if there is no apparent motor deficit in the legs.

In the following discussion, clinical signs of muscle weakness are described. These functional neurologic levels may not directly reflect the anatomic level of the malformation.

Musculoskeletal deformities related to muscle imbalance may present serious clinical concerns. Deformities may be static deformities present at birth or may develop over time. Figure 15.1 summarizes segmental innervation, preserved muscle function, and musculoskeletal complications typical of various levels of spinal cord malformation. Providers must keep in mind that the overall functional outcome for the individual is related in part to neurologic level, in addition to other associated CNS, psychosocial, and medical issues.

THORACIC LESIONS

Thoracic-level malformations spare the upper extremities, with the exception of decreased ability to abduct the fifth digit (thoracic level 1 = T1). There is usually partial innervation of the abdominal and intercostal musculature, which may result in respiratory dysfunction or insufficiency. Kyphosis and kyphoscoliosis may result from trunk weakness and be more prominent in individuals with vertebral anomalies (55). The legs may be flaccid or show signs of spasticity when some portion of the distal spinal cord is preserved (56). The lack of volitional movements combined with the effect of gravity leads to lower extremity deformities. The usual lower extremity posture in the supine position is partial hip external rotation, abduction, and ankle plantar flexion from gravity. Knee flexion contractures and equinus foot deformities develop from sitting. Hip flexion contractures with compensatory lumbar lordosis increase any pre-existing kyphosis or kyphoscoliosis (56).

L1–L3 SEGMENT

Hip flexors and hip adductors are innervated at the L1 to L2 levels. With L2 sparing, knee extensors have partial innervation but are not at full strength. The distal lower extremity muscle strength is absent. The distribution of muscle imbalance—hip flexion and hip adduction with absent hip extension and hip abduction—leads to the development of contractures and early paralytic hip dislocation. Pelvic obliquity seen in asymmetric hip pathology enhances scoliosis. Gravity-related foot equinus deformity may develop (56).

Ambulation during young childhood is typical with the use of bracing and assistive devices. Long-term ambulation through adulthood is less likely as priorities change and there are further increases in the already high-energy demands of walking (54). The extent of bracing necessary to achieve ambulation is usually related to the amount of active knee extension.

L4–L5 SEGMENTS

Innervation of the hip flexors, hip adductors, and knee extensors is usually complete; however, hip abductors and hip extensors remain weakened. Coxa valga and acetabular dysplasia are still a concern. Typically, hip dislocation occurs later at the L4 to L5 segmental levels. Newborns with a well-defined lesion sparing L4 lie in a typical position of hip flexion, hip adduction, and knee extension. When the L5 segment is spared, the gluteus medius, gluteus maximus, and hamstrings have partial strength and knee extensor contracture is less likely. Because the tibialis anterior is unopposed by its plantarflexion and everter antagonists, a calcaneovarus foot deformity develops. If the peroneus muscles are spared, the varus is eliminated. Although the plantar flexors are partially innervated, they are not strong enough to counter the strong force of the ankle dorsiflexors (56).

SACRAL SEGMENTS

Active plantarflexion is stronger and some toe movements are present. Intrinsic foot muscles remain weak and may result in a cavus foot deformity with clawing of the toes (56).

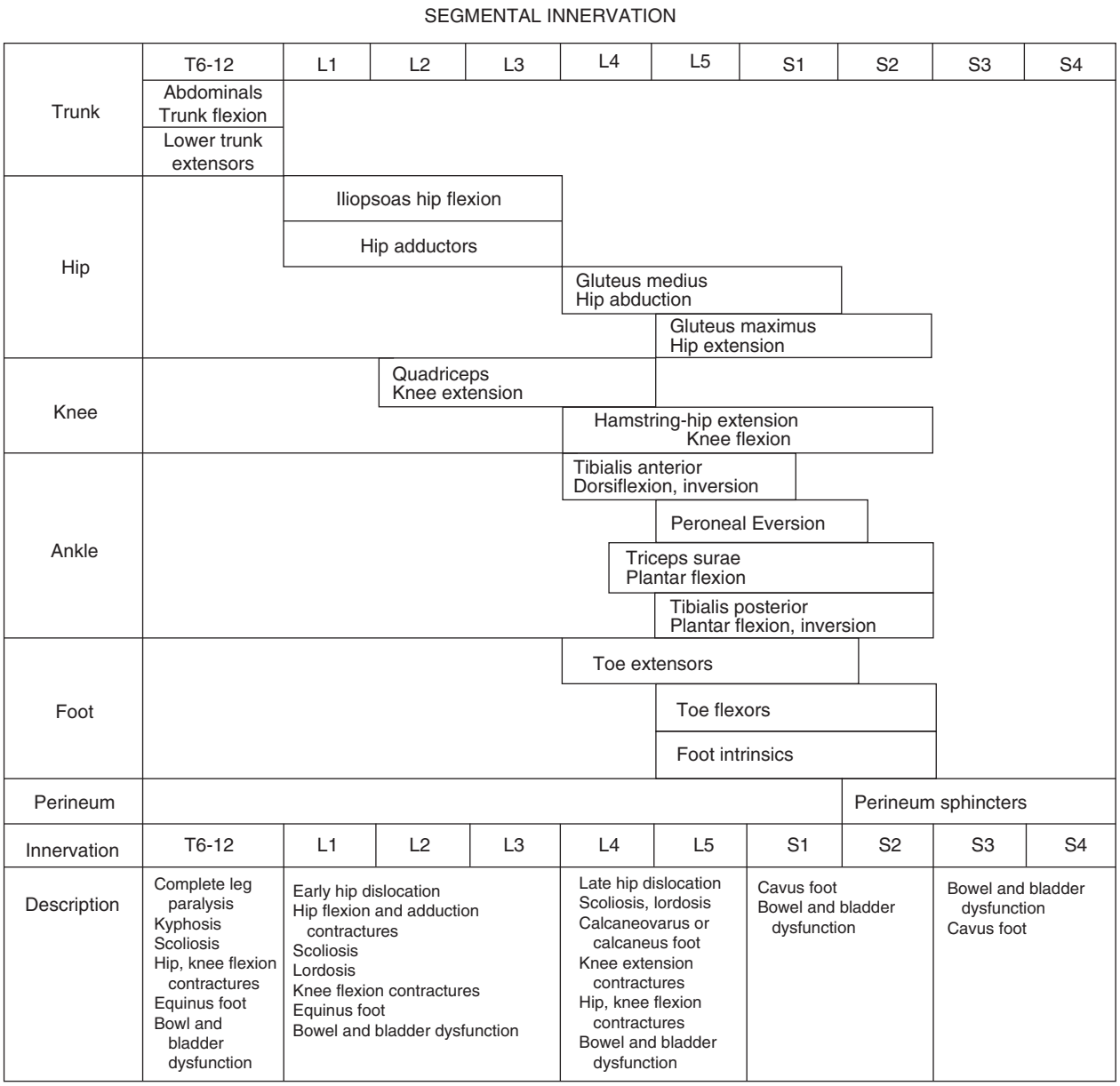


FIGURE 15.1 Musculoskeletal, sensory, and sphincter dysfunction by segmental level.

SENSORY DEFICIT

Partial or complete absence of sensation predisposes individuals with spina bifida to skin injuries because of decreased ability to perceive pressure, pain, trauma, or heat (52–54,57). Skin breakdown tends to occur over areas of prominence and weight-bearing. The lower back, intergluteal, perineum, feet, heels, and toes are the sites of predilection, but any area with sensory loss may be affected. Scoliotic and kyphotic prominences are areas prone to breakdown (55). Pressure ulcers often heal slowly, tend to get infected, and often recur. A refractory pressure ulcer may be a symptom of a tethered cord. Long-standing ulceration with deep tissue necrosis may spread to bone and lead to acute or chronic osteomyelitis. Weight-bearing is often on the anesthetic heel, and a deep ulcer can lead to osteomyelitis of the os calcis (56).

Other complications of denervation include vasomotor instability, neuropathic Charcot joints, and osteoporosis in individuals with extensive lower extremity weakness (54,55,57,58).

Sensation may be decreased with regard to sexual function thus causing different issues for males and females. Please see the section on Long Term Aging With a Neural Tube Defect where this is discussed in more detail.

ASSOCIATED CENTRAL NERVOUS SYSTEM MALFORMATIONS

Extensive neuropathologic studies have demonstrated that NTDs are associated with a high incidence of gross and microscopic malformations of the forebrain and hindbrain (59). Additional anomalies in the spinal cord may complicate the original local dysraphic defect (52,53,57,60,61). Table 15.1 lists associated anomalies and malformations by location.

SPINAL CORD

The spinal cord defect usually results in a lower motor neuron process. Spasticity is present in most individuals with spina bifida across their lifetime (62). The presence or gradual development of spasticity above the level of the spinal cord lesion may be related to tethering of the spinal cord, Chiari type II malformation exerting pressure on the cervical spinal cord, decompensating hydrocephalus, ventriculitis, syringohydromyelia, or coexistent encephalopathy sustained at birth (63–66).

Tethered cord refers to an abnormal attachment of the spinal cord at its distal end (57). Under normal circumstances, the conus medullaris ascends from its distal position to the L1 to L2 vertebral level during the first year of life (67). Focal abnormalities—including thickened and shortened filum terminale, supernumerary fibrous bands, persistent membrane reunions, dural sinus, diastematomyelia, entrapment by lumbosacral tumors, and adhesions in the scar tissue of the repaired myelomeningocele—interfere with this process (57). Almost all children born with spina bifida have a low-lying cord on MRI, and approximately one-third develop neurologic, urologic, or orthopedic complications or symptoms (68) (Figure 15.2).

Tethering of the spinal cord is the second most common cause of neurologic decline in children with myelomeningocele (68). The most common clinical signs or symptoms of a tethered cord include spasticity in the lower extremities, decline in lower extremity strength,

TABLE 15.1 ASSOCIATED CENTRAL NERVOUS SYSTEM MALFORMATIONS

Spinal cord
Tethering
Distal focal abnormalities
- Thick, short filum terminale
- Supernumerary fibrous bands
- Lumbosacral tumors (lipoma, fibrolipoma, fibroma dermoid, epidermoid cyst, teratoma)
- Bony vertebral ridge
- Diastematomyelia, diplomyelia, split cord

Brainstem
Arnold type II malformation
- Kinking, inferior displacement of medulla
- Herniation into cervical spinal canal

Abnormalities of nuclear structures
- Dysgenesis, hypoplasia, aplasia, defective myelination

Hemorrhage, ischemic necrosis
Syringobulbia

Cerebellum
Arnold–Chiari type II malformation
- Elongated vermis, inferior displacement
- Herniation into cervical spinal canal

Abnormal nuclear structures
- Dysplasia, heterotopia, heterotaxia

Ventricular system
Hydrocephalus
Aqueductal stenosis, forking, atresias

Forebrain
Polymicrogyria
Abnormal nuclear structures
- Heterotopia (subependymal nodules)
- Heterotaxia

Prominent massa intermedia
Thalamic fusion
Agenesis of olfactory bulbs and tracts
Attenuation/dysgenesis of corpus callosum

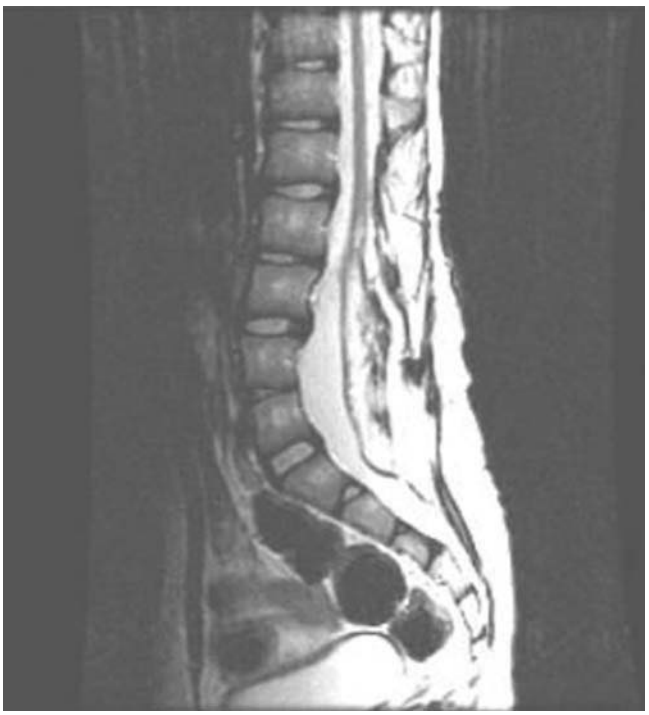


FIGURE 15.2 T2-weighted magnetic resonance image of tethered cord. There is tethering of the spinal cord with conus seen down to the L5 vertebral level, heterogeneous signal intensity characteristics, and areas of fibrofatty tissue.

Source: Molnar GE. Spina bifida: clinical correlations of associated central nervous system malformations. *Phys Med Rehabil State Art Rev*. 1991;5:289, with permission.

and worsening scoliosis. Other signs and symptoms that strongly suggest tethering of the spinal cord include back pain, changes in urologic function, changes in gait, and development of lower extremity contractures. In patients who are suspected of having a symptomatic tethered cord, the function of their shunt must be evaluated prior to proceeding forward with surgical management (69), as shunt malfunction can present similarly.

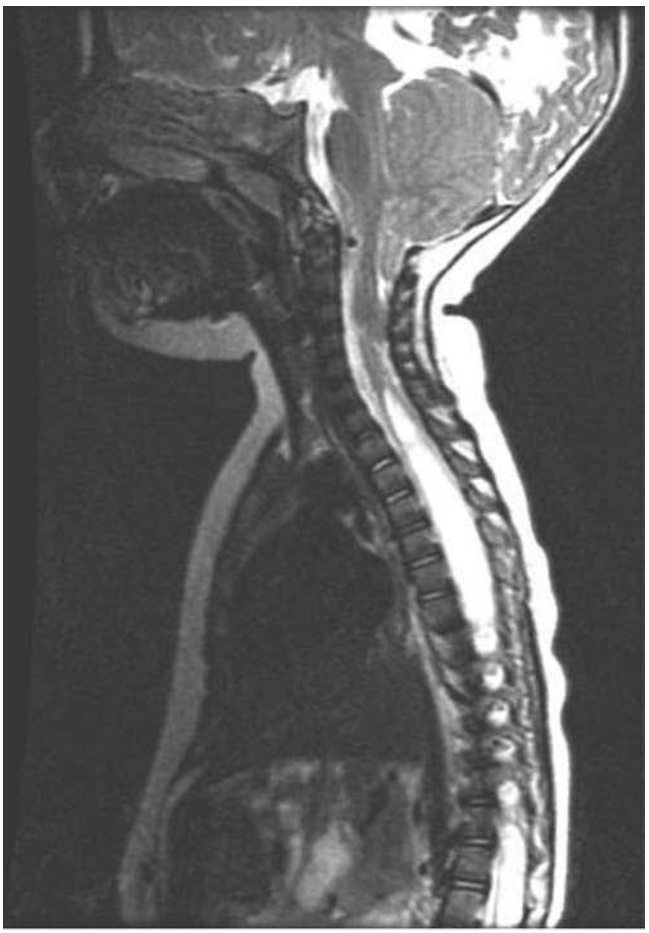
The reported functional outcome of surgical management of a tethered cord is variable. One study reported improvements in gait in almost 80% of patients following untethering, whereas another study reported improvement in as few as 7% (70). (Note: All cords tether to some extent following repair.) Less than 20% of children with a tethered cord experience back pain. However, this is the symptom most likely to improve with surgery (63,71). A recent article comparing prophylactic and symptomatic tethered cord surgery in those with lumbosacral lipomas found no change in long-term outcome. This suggests that delaying surgical intervention may decrease the number of tethered cord surgeries (72).

Diastematomyelia is a postneurulation defect that results in a sagittal cleavage of the spinal cord, most commonly affecting the lumbar and thoracolumbar levels of the spinal cord. It is more common in females (73,74). Diastematomyelia may have both neurologic and orthopedic presentations. Orthopedic symptoms include scoliosis, Sprengel's deformity (especially when associated with Klippel–Feil sequence), hip subluxation, and lower extremity limb-length discrepancies (74,75).

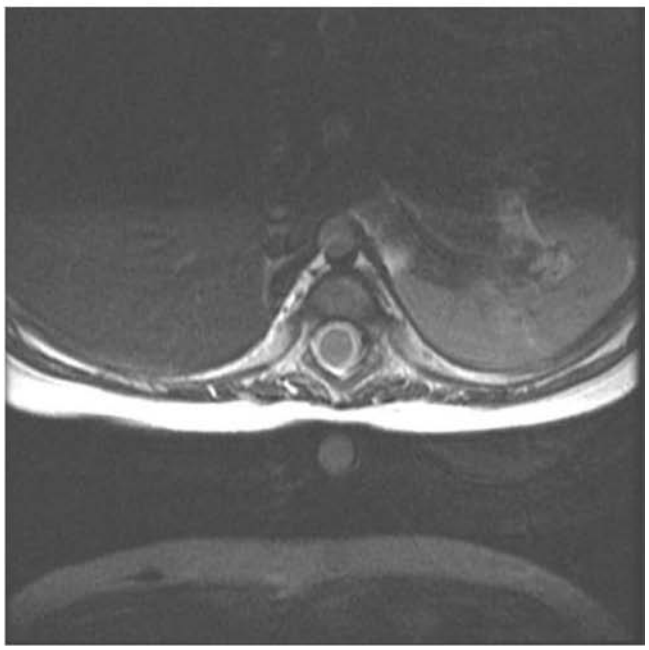
Neurologic symptoms include gait abnormalities, asymmetric motor and sensory deficits of the lower extremities, and neurogenic bladder and bowel (76). Symptoms of diastematomyelia may present in childhood or, less commonly, in adulthood (77).

It is not uncommon for individuals to develop syringomyelia—a tubular cavitation in the spinal cord parenchyma extending more than two spinal segments (78). Syringomyelia is present in up to 40% of individuals with myelomeningocele (79). The syrinx may be located anywhere along the spinal cord, medulla, or pons, but is most common in the cervical spinal region (52,54,80). MRI is used to detect syringomyelia (81) (Figure 15.3).

Often, a syrinx is of little clinical significance; however, if a patient develops decreasing function above the level of his or her lesion, syringomyelia must be considered in the differential diagnosis. Although shunt malfunction and spinal cord tethering are more common complications, symptomatic hydromyelia may explain a slower-than-expected progression through gross motor and fine motor developmental milestones or a decrease in strength or change in function. Early progression of scoliosis above the initial neurologic level may be the earliest sign of a syrinx. A shunt malfunction may contribute to a symptomatic syrinx, and shunt function should be evaluated. Placement of a syringopleural shunt may be necessary to decompress the syrinx.



(A)



(B)

FIGURE 15.3 T2-weighted magnetic resonance image showing sagittal (A) and axial (B) views. There is a large syrinx present, beginning at the mid portion of C6 and extending to L4.

CEREBELLUM AND HINDBRAIN

The most common hindbrain abnormality in NTDs is Chiari type II malformation, seen in 80% to 90% of individuals with myelomeningocele (52,53,57,82).

This malformation results in caudal displacement or herniation of the medulla, lower pons, elongated fourth ventricle, and cerebellar vermis into the cervical spinal cord (Figure 15.4). This often interferes with cerebrospinal fluid (CSF) outflow and is, therefore, almost always associated with hydrocephalus. Caudal displacement of the medulla may occur and result in traction neuropathies of the lower cranial nerves. Signs of bulbar compromise arise from compression of the herniated hindbrain.

A broad spectrum of clinical symptoms is seen in individuals with this malformation. However, only 20% will develop clinical signs of brainstem dysfunction, with most occurring in the neonatal period (83,84). Symptoms may be evident at birth or present within the first 2 to 3 months.

The most severe symptom is respiratory compromise, which may be both centrally and peripherally induced. Individuals may experience stridor, laryngeal nerve palsy with vocal cord paralysis, upper airway obstruction, periodic breathing, central or obstructive sleep apnea, or aspiration. Dysphagia and extraocular motion abnormalities, which are related to cranial neuropathies, may also be seen. Dysphagia may be severe enough that gastrostomy tube placement is required. Airway compromise may necessitate tracheostomy.

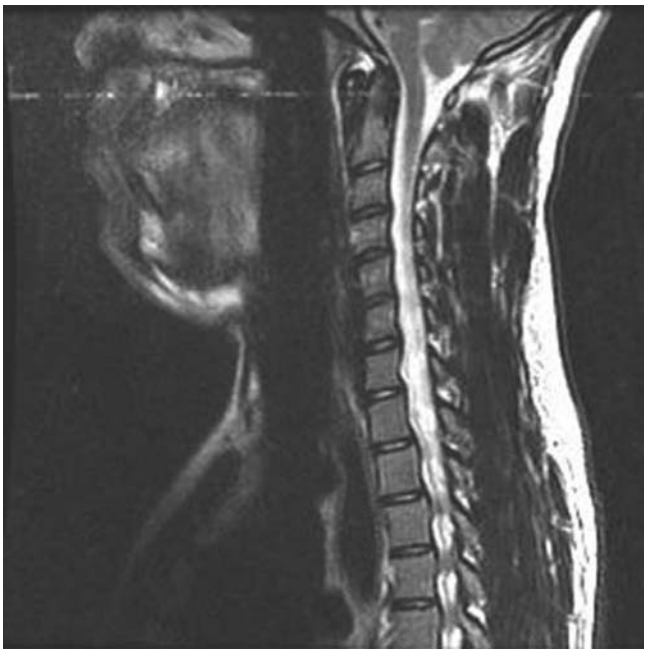


FIGURE 15.4 T2-weighted magnetic resonance image of the cervical spine with Chiari II malformation. The posterior fossa is crowded. There is cerebellar tonsillar herniation, with the cerebellar tonsils lying 9 mm below the foramen magnum.

In the presence of brainstem compromise, hemiparesis or tetraparesis may occur (this is more common in older children and adults than in infants). Impairment of fine motor hand function is well documented and is seen in more than half of individuals with thoracic-level lesions and approximately one-fourth of individuals with lumbosacral lesions.

Control of ocular motility (saccadic eye movements, visual fixation, and pursuit) is related to cerebellar function. Fewer than one-third of individuals with spina bifida have completely normal visual function (85,86).

Despite successful initial treatment with surgical decompression, problems may recur. Typically, vocal cord paresis in the first 2 months of life is a sign of irreversible damage, and surgical decompression is unlikely to result in clinical improvement (87).

VENTRICLES

Hydrocephalus is a significant problem in the majority of patients with myelomeningocele. The pathogenesis of hydrocephalus is multifactorial and is related to aqueductal stenosis, occlusion of the foramen of Luschka and Magendie, hindbrain herniation, obliteration of the subarachnoid spaces at the level of the posterior fossa, compression of the sigmoid sinuses with consequent venous hypertension, and fibrosis of the subarachnoid spaces (88,89). The prevalence of hydrocephalus in individuals with myelomeningocele is reported to be as high as 95%, with shunt rates ranging from 77% in the 1980s to 58% in more recent years (90). In a 23-year period from 1988 to 2010, the national trend was 56.6 % required surgical repair perinatally at the same time as shunt placement (91). Hydrocephalus rates are closely associated with the level of the spinal dysraphism. In one cohort, 100% thoracic, 87% lumbar, and 67% sacral myelomeningocele patients required shunting (92). In all cases of symptomatic hydrocephalus, surgical management is recommended.

Symptoms of hydrocephalus include those that are classic for increases in intracranial pressure. This varies based on the presence or absence of an open fontanelle. In an infant, signs of increased intracranial pressure include lethargy, decreased feeding, bulging fontanelle, increasing head circumference (greater than expected for age), poor developmental progress, and "sun downing." In patients with a closed fontanelle, signs of increased intracranial pressure include headache, vomiting, drowsiness, changes in behavior, changes in personality, irritability, diplopia, and papilledema. With the sudden onset of increased intracranial pressure, Cushing's triad may be seen. Cushing's triad consists of progressively increasing systolic blood pressure, bradycardia, and irregular respirations.

Presently, placement of a shunt is the standard of care for surgical management of hydrocephalus. Shunting has

many complications, including both mechanical and infectious. Up to 95% of adult patients with myelomeningocele have required at least one shunt revision. The rate of shunt infection is between 5% and 8% per procedure (68,93–97).

Endoscopic management of hydrocephalus is being increasingly presented as an alternative to shunting. Endoscopic third ventriculostomy (ETV) provides direct communication between the third ventricle and the subarachnoid space by way of interpeduncular and prepontine cisterns. The success rates for ETV, as the sole management for hydrocephalus in infants with myelomeningocele, range from 12% to 53% (98–102). More recently, ETV has been combined with choroid plexus cautery (CPC). This has resulted in an improved success rate of greater than 70% for treatment of hydrocephalus in infants. Failure of ETV combined with a CPC typically is within the first 3 months (103).

ETV may also be an option in the setting of a shunt malfunction in the older child. In one study the majority of ETV failures were during the first 6 weeks postoperatively. However, failures were seen as late as 5 years postoperatively (102). Longevity of the ETV/CPC for treatment of hydrocephalus beyond 2 or 3 years has yet to be determined. It is not known if there is a difference in neurocognitive outcomes in patients treated with an ETV/CPC (shunt-independent) as compared with individuals who are shunt-dependent. Although not yet considered the "standard of care," ETV in combination with CPC holds promise for surgical management of hydrocephalus without creating shunt dependency and the associated complications (103).

FOREBRAIN

Malformations of the forebrain are broad, and range from gross anatomic malformations to microscopic anomalies. Polymicrogyria are increased numbers of small-sized cerebral gyri with shallow disorganized sulci, and this is seen in up to 65% of individuals (104). Heterotopias are aberrant neural tissues in the form of subependymal nodules. They are present in approximately 40% of cases (59). Microscopic studies have demonstrated disordered cortical lamination, neuronal hypoplasias of the thalamus, and complete or partial agenesis of the olfactory bulbs and tracts. Dysgenesis or agenesis of the corpus callosum may be seen and may also be associated with a malformed cingulated gyrus and septum pellucidum (104). The contribution of these forebrain malformations to the development of cognitive and perceptual dysfunction remains unknown.

OTHER MALFORMATIONS

NTDs are also associated with an increased rate of malformations unrelated to the CNS. Vertebral anomalies are not uncommon and contribute to progressive kyphosis and scoliosis (56). Thoracic deformities may result from rib deformities, including absence, bifurcation, or reduction of the ribs. Malformations of the urinary system may be present and result in accelerated deterioration of renal function.

NTDs have been associated with genetic abnormalities, including trisomy 18, trisomy 13, Turner's syndrome, Waardenburg' s syndrome, renal aplasia and thrombocytopenia syndrome, nail-patella syndrome, 13q deletion syndrome, and others (105).

TREATMENT

TEAM APPROACH

A team approach is an important part of the care of the individual with congenital spinal dysfunction. The multidisciplinary team often includes neurosurgery, orthopedic surgery, urology, rehabilitation medicine, physical and occupational therapy, social work, nursing, nutrition, and neuropsychology. Coordination of all modes of treatment is important for a successful rehabilitation plan. Primary care for the usual childhood illnesses and health maintenance including immunizations should remain the responsibility of the pediatrician.

After birth, parents and families of individuals with spina bifida need to be informed about their child's diagnosis and its implications. A prenatal visit with the neurosurgeon and other medical specialists is recommended. Parents often ask questions regarding anticipated functional abilities and limitations, including self-care and ambulation. Cautious predictions based on the current functional level should be given. Medical providers should be factual in their discussion of the problems that the parents and child will face. Discussions and instructions about the child's care and handling at home may require several sessions so that the family members are not overwhelmed by the amount and complexity of the information. Families should be informed of the many issues involved and the need for seeing several medical specialists. Frequent follow-up after discharge from the neonatal unit is often necessary and typically involves visits every 3 to 4 months for a couple of years and then every 6 months thereafter (56).

NEUROSURGICAL TREATMENT

Involvement in the care of the individual with spina bifida begins with a prenatal visit to the neurosurgeon. Studies regarding prenatal surgical closure of a NTD are under way. To date, intrauterine repair has not been shown to decrease the motor deficits associated with myelomeningocele, but in some series it has been demonstrated to

decrease the degree of associated Chiari type II malformations and the need for shunting procedures for hydrocephalus in the first year of life (106–109).

Neurosurgical repair of an open NTD, such as a cystic lesion, is usually performed on the first day of life. If hydrocephalus is present at birth, surgical management may be necessary. Ninety-five percent of children with spina bifida are likely to have hydrocephalus, and 75% to 85% require surgical management. The average revision rate is 30% to 50% (110), and after 2 years of age there is a 10% per year risk of failure (96). Most neurosurgeons believe that a child with hydrocephalus that required shunting will remain shunt-dependent (97,111). These statistics may change as ETV with CPC is performed with increasing frequency.

Neurosurgical follow-up is required, even after the neonatal period, to monitor for symptomatic hydrocephalus, shunt malfunction, and other neurosurgical complications. Pediatric patients with myelomeningocele should be followed routinely, usually on an annual basis.

NEUROGENIC BLADDER

A neurogenic bladder is found in most individuals with spina bifida. Data from the CDC National Spina Bifida Registry presented at the American Association of Developmental Medicine 2014 annual conference, reported 94% occurrence of neurogenic bladder in individuals with myelomeningocele with an over 60% urinary incontinence prevalence in those above 5 years. Furthermore, most with myelomeningocele use a catheter to empty their bladders. From the newborn period, the successful treatment of a neurogenic bladder is accomplished by the team approach including the urologist, physiatrist, nursing, and family.

PHYSIOLOGY

The fundus is made up of three layers of crisscrossing smooth muscle, called the detrusor. These three smooth muscle layers extend down the proximal urethra and stop at the external sphincter, which comprises skeletal muscle. T10 to L1 supplies the sympathetic innervation for the bladder; this causes the detrusor to relax and the bladder neck and posterior urethra to contract, effectively allowing storage of urine. S2 to S4 provide the parasympathetic innervation to the bladder primarily at the fundus and the neurotransmitter is acetylcholine causing contraction to empty the bladder. The sympathetic innervation is active during bladder filling, and the parasympathetic innervation is active during urination. Somatic nerves via the pudendal nerve (from the sacral cord S2–S4) innervate the skeletal muscle component of the external urethral sphincter: contraction occurs in the resting state for continence, and relaxation should occur during urination (112).

BLADDER CAPACITY

The prediction of normal bladder capacity aids the diagnosis of abnormal voiding patterns. It is typically accepted that the bladder capacity of a baby during the first year equals the weight of the baby in kilograms times 7 to 10 mL. A study with 200 children (132 with normal voiding, 68 frequent and infrequent voiders) demonstrated that from approximately 1 to 12 years of age, the sum of age in years plus 2-equals the bladder capacity in ounces (30 mL = one ounce). Teenagers (if normal size) assume adult-size bladders, typically around 400 to 500 mL. Clinically infrequent voiding may cause an increase in bladder capacity over normal values. Clinically frequent voiding may cause a decrease in functional bladder capacity (113). Postvoid residual (PVR) is generally accepted as less than 20% of bladder capacity, taking into account the appropriate bladder capacity for age.

DIAGNOSTICS

Checklist for Diagnosing Neurogenic Bladder

- Is the current management preserving the kidneys and renal function?
- If present, does bacteriuria represent infection or colonization?
- Are the bladder and kidney studies up to date?
- Is urination true voiding or overflow incontinence?
- Are bladder capacity and bladder compliance appropriate for age?
- Is PVR appropriate?
- Is the sphincter mechanism competent?

DIAGNOSTIC TESTS

Urinalysis

The nitrate test indirectly detects urine bacteria with enzymes that reduce nitrate to nitrite in urine (eg, *Klebsiella*, *Enterobacteriaceae*, *Escherichia coli*, and *Proteus*).

THE LEUKOCYTE ESTERASE TEST. While leukocytes in the urine can disintegrate and disappear rapidly, leukocyte esterase persists.

Urine culture (UC): Culture of urine for suspected clinical infection.

Serum creatinine: Indicator of renal function, but typically lags renal function by several days.

FUNCTIONAL STUDIES

- US—Ultrasound of the kidneys and bladder to determine any structural abnormalities, including hydronephrosis,

trabeculation, stones, and PVR. If not on catheterization program, consider US to measure PVR.
- VCUG—Voiding cystourethrogram to detect vesicoureteral reflux (VUR), evaluate the bladder contour, and evaluate the urethra. For those without neurogenic bladders, the first study is a contrast VCUG for boys and girls. Subsequent VCUG studies, for boys and especially girls, can be radionuclear cystograms, as the radiation is reduced. However, for those with neurogenic bladders the fluoroscopic VCUG with contrast demonstrates greater detail of the bladder neck on all subsequent studies. Many centers perform cystograms as part of the urodynamic study, called videourodynamics. If videourodynamics are used, then a separate VCUG is not necessary.
- UDY—Urodynamics to determine detrusor leak point pressure, uninhibited bladder contractions now called detrusor over activity (114), detrusor sphincter dyssynergia, bladder capacity, PVR, and bladder compliance and sensation. The basic urodynamic formulas are:

Pressure detrusor = pressure vesical (bladder) – Pressure abdominal (rectum). On urodynamic testing, detrusor leak point pressure (p vesical – p abdominal) greater than 40 cm H_2O, with a bladder capacity less than 33% of expected, was associated with renal damage (115).

Bladder compliance = change in bladder volume/ change in pressure

It is recommended that these tests (US, UDY, and VCUG) be performed in the neonatal period, as baseline studies. The VCUG can be excluded if videourodynamics are utilized. As growth of the infant is rapid in the first 12 months, US should be repeated every 3 to 6 months during infancy. Greater frequency is encouraged if initial studies are abnormal. A common protocol incorporates US twice yearly the second year and then yearly. Abnormalities on US will likely lag those found on UDY. UDY should be repeated during the first year if the initial study shows a high detrusor leak point pressure of greater than 40 cm H_2O or detrusor sphincter dyssynergia. Many centers perform UDY yearly in all patients to aggressively detect potential bladder deterioration that could harm the kidneys, whereas others repeat only if renal changes are noted on US or continence is not obtained by school age (116). VCUG (or video UDY) are needed if VUR was noted or if there is a new onset of recurrent UTIs or hydronephrosis. UDY studies should be repeated with significant clinical changes in bowel or bladder incontinence, infections, or gait, all of which are potential signs of secondary tethered cord (117).

Other Studies

RENAL STONES. The excretory urogram (EXU) also known as intravenous pyelogram (IVP), once used to detect urinary tract (UT) stones, anatomic abnormalities, and obstruction, is rarely done anymore. Following diagnostic US for renal stones, the EXU has been replaced by CT without contrast for more precise imaging.

ANATOMIC VARIANTS. A magnetic resonance urogram with gadolinium is now used to demonstrate rare anatomic variants, particularly to delineate the ureters if ectopia is suspected. Diethylene triamine acetic acid (DTPA) and mercaptoacetyltriglycine (MAG3) are nuclear medicine studies used to evaluate differential function and excretion of each kidney; however, the MAG3 offers better imaging than DTPA to evaluate UT drainage/ obstruction. The DTPA and MAG3 radioactive tracers are filtered (DTPA) or excreted (MAG3) into the renal tubules. Dimercaptosuccinic acid (DMSA), a radiotracer that binds to the tubule, is not rapidly excreted, and it is the best test for imaging the functioning renal cortex to detect congenital hypoplasia or acquired scarring. This test should be done when there is abnormality on a renal US, a history of multiple urinary tract infections (UTIs), or pyelonephritis. Cystoscopy is recommended for persistent hematuria or bladder masses. There is controversy regarding the role of routine screening cystoscopy in patients with increased risk of malignancy due to either bladder augmentation surgery or long-term indwelling catheter use (118,119).

Figure 15.5 depicts a normal urodynamic study and Figure 15.6 is a tracing of a urodynamic study reflecting spastic bladder detrusor and sphincter dyssynergia.

RISK FACTORS FOR UPPER TRACT/KIDNEY DETERIORATION

- Detrusor leak point pressures greater than 40 cm H_2O determined by UDY
- Vesicoureteral reflux determined by VCUG/videourodynamics
- Detrusor sphincter dyssynergia determined by UDY

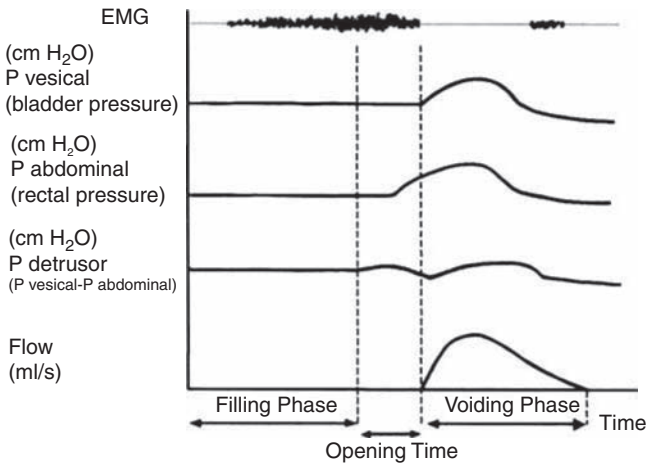


FIGURE 15.5 Normal urodynamic study.

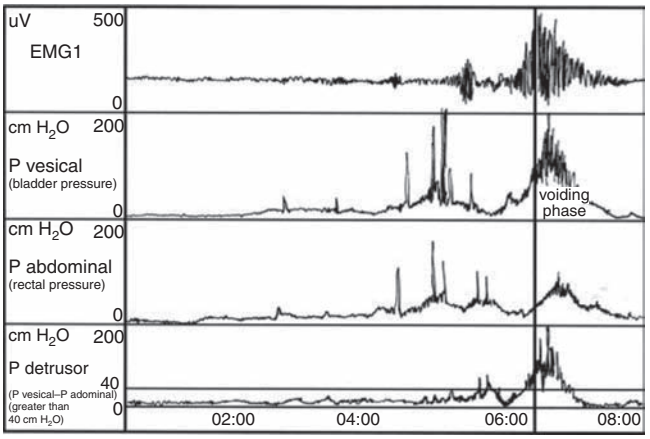


FIGURE 15.6 Urodynamic study reflecting spastic bladder detrusor and sphincter dyssynergia.

- Poor bladder compliance determined by UDY
- Detrusor overactivity previously known as bladder hyperreflexia
- Increased PVR greater than 20% of the total bladder capacity

TREATMENT

Goals

- Preservation of renal function
- Age—appropriate social continence
- No significant UTIs
- Normalized lifestyle

TREATMENT OF STORAGE DYSFUNCTIONS

Excessive storage pressures due to detrusor overactivity previously known as detrusor hyperreflexia, or decreased compliance, can be decreased with anticholinergic medications. Oxybutynin chloride is commonly used and is available as syrup, and in immediate-release and extended-release tablets [Note: The youngest child in the study was 6 years old (120)]. Oxybutynin has been used intravesically, but is an off-label use. There is a transdermal form of oxybutynin as well as other anticholinergic medications on the market, but these are not currently *Food and Drug Administration* (FDA) approved in children. Botulinum toxin A (off-label) can be injected into the detrusor muscle via a cystoscope for those whom anticholinergic medication is ineffective. Electrical stimulation of the bladder directly or of innervating nerves has shown some promise to improve storage characteristics but remains experimental.

Stress incontinence due to bladder outlet weakness caused by poor innervation of the internal and/or external urinary sphincters is more difficult to treat medically. Internal urethral sphincter resistance may be improved by the following alpha-sympathetic stimulation medications: phenylephrine, pseudoephedrine, and imipramine. External urethral sphincter weakness may improve with neuromuscular reeducation, but surgical treatment is typically required.

TREATMENT OF EMPTYING DYSFUNCTIONS

The typical day-to-day management of the neurogenic bladder in a majority of individuals with spina bifida is clean intermittent catheterization (CIC) every 4 hours while awake to keep bladder volumes within normal limits for age (113). In 1972, Lapides was the first to publish that the sterile single-use catheter is unnecessary in the management of persons with neurogenic bladders because it does not reduce bacteriuria (121). This continues to be substantiated in the pediatric population (122). If this intervention is unsuccessful, various pharmacologic and urologic surgical procedures may be explored. The Credé maneuver should be used with extreme caution. Using the Valsalva or Credé maneuver to empty the neurogenic bladder that has detrusor sphincter dyssynergia will likely raise the intravesicular pressure to greater than 40 cm H_2O, thus putting the kidneys at risk. Bethanechol is rarely used to treat weak expulsive force of the detrusor because emptying is typically incomplete and synergistic sphincter relaxation does not occur. Hyperactive internal sphincter mechanism may be treated with alpha-adrenergic blockers. Hyperactive external urethral sphincter may be treated with baclofen, neuromuscular reeducation of the pelvic floor, Botox injections (off label) (123), or surgery.

The Mitrofanoff procedure, first introduced in the 1980s, creates a continent catheterizable channel, between the bladder and the umbilicus/or lower abdomen. The appendix is most commonly utilized to create the channel, but other structures, such as a refashioned small bowel or ureter can be used. A tunnel between the bladder mucosa and detrusor muscle should provide sufficient compression to prevent external leakage; however, leakage may be problematic in a small percentage (124) but most should be continent. A Mitrofanoff procedure may be useful for patients unable or unwilling to cath per urethra, especially in females who may have more difficulty catheterizing than males. The procedure is commonly performed in the setting of bladder outlet surgery that usually prevents leakage per urethra but makes urethral catheterization more difficult. A vesicostomy may be a temporizing measure for younger children to avoid deleterious high bladder pressures. The ileal conduit, an incontinent urostomy to an external bag was commonly utilized in children with spina bifida prior to the 1970s before both CIC and oxybutynin were introduced. Follow-up studies showed a disappointingly high rate of renal deterioration, calculosis, hydronephrosis, and the need for reversal of the ileal

conduit procedure (56). Artificial urethral sphincters have been found to be helpful in some, but can cause infection, erosion, and mechanical problems.

PRIMARY CARE TREATMENT OF CHILDREN MANAGED WITH CLEAN INTERMITTENT CATHETERIZATION

Routine urinalysis (UA) and UC are not recommended during well-child check-ups if the child looks well and is asymptomatic. If bacteriuria is detected in the urine, it is important to determine whether it represents a clinical infection or colonization of the bladder. Only clinical UTIs with symptoms of fever, abdominal/back pain, painful urination/catheterization, new or worsening incontinence or urinary symptoms, or hematuria should be treated (122). Prophylaxis in the absence of VUR is not routinely recommended.

ANTIBIOTIC PROPHYLAXIS AND BACTERIURIA TREATMENT

A number of studies were done on antibiotic prophylaxis and bacteriuria treatment with individuals with neurogenic bladders. Kass found that if there is no VUR, bacteriuria is innocuous; in his study, 17 hydronephrotic kidneys showed significant radiographic improvement since starting CIC (125). Ottolini found that asymptomatic bacteriuria requires no antibiotic therapy in the absence of VUR (126). Van Hala found that there is no correlation among the number of UTIs, the type of catheter used, and the use of prophylactic antibiotics (127). Johnson and colleagues found that nitrofurantoin is an effective prophylactic agent during a 3-month period for bacteriuria (128). Schlager and colleagues found that asymptomatic bacteriuria persists for weeks in children with neurogenic bladders with normal upper UTs managed with CIC (129). The asymptomatic bacteriuria is different from the symptomatic bacteriuria. Jayawardena and colleagues found that patients with a spinal cord injury (SCI) frequently have asymptomatic bacteriuria without data to support treatment and that routine UCs should not be done at annual evaluations (130).

(Note: It may be appropriate for a pediatric patient without a neurogenic bladder and with frequent UTIs secondary to dysfunctional voiding to receive prophylactic antibiotics for a time. Patients with VUR and with or without a neurogenic bladder routinely receive prophylactic antibiotics.)

NEONATAL VERSUS CHILDHOOD TREATMENT

Early proper management is imperative for the preservation of renal function (131). A urologist should be involved from the newborn period as 20% are born with renal anomalies and kidney damage was found to be approximately 1 in 4 without proper management of the neurogenic bladder (115).

Treatment of neurogenic bladder dysfunction due to myelomeningocele in neonates is recommended. A study of 98 individuals (46 started CIC in the first year of life, 52 began CIC after 4 years of life) reviewed the charts of those using CIC who were believed to be at risk for renal deterioration. The mean follow-up of this study was 4.9 years, and the average age of the patient at the last follow-up was 11.9 years. The study found that neonatal treatment enabled UDY to identify those infants at risk for upper tract deterioration, which was prevented by the initiation of oxybutynin and CIC. There was a similar improvement in UTI rate, hydronephrosis, and reflux. The percentage of patients with worsening hydronephrosis and persistent high intravesical pressures who needed bladder augmentation was 11% in the earlier treatment group versus 27% in the later treatment group ($P < .05$) (132).

FURTHER SURGICAL MANAGEMENT FOR REFLUX AND SMALL NEUROGENIC BLADDERS

Lowering the bladder pressure below 40 cm H_2O is the goal. Ureteral reimplantation may be necessary for reflux; however, most people with neurogenic bladders have reflux from a high-pressure bladder and not from a ureterovesical junction that is dysfunctional. Medical management with CIC and anticholinergic medication frequently causes reflux to resolve, and surgical correction is rarely necessary. Bladder augmentation may be considered for a small or high-pressure bladder refractory to medical treatment. Bladder augmentation may increase the risk of bladder cancer, rupture, and stone, and mucus may be excessive in the urine, obstructing CIC (133). For cancer surveillance in those with augmented bladders, cystostomy was recommended annually starting 10 years following the bladder augmentation (118). Newer recommendations may be different and a urologist should be involved to determine adequate surveillance. Augmentation should be explored only after pharmacologic management has failed and the system continues to be a high-pressure system, thus putting the kidneys at risk. The complication rate for bladder augmentation in one study was approximately 1 in 3 (134).

In conclusion, those individuals with a normal neurologic exam with sacral-level spina bifida likely have a neurogenic bladder and need appropriate management. This point is demonstrated in a study of bladder dysfunction and neurologic disability at presentation in closed spina bifida. There were 51 individuals in the study, with a mean age of presentation of 3.3 years. Of these patients, 25 had UT disturbance, 12 had neurologic problems, 33 had a normal neurologic exam, 21 had a normal renal ultrasound

(RUS), and 31 had abnormal videourodynamics, despite a normal neurologic exam and RUS (135). The majority of individuals with spina bifida have a neurogenic bowel and bladder; even if there are no motor signs of weakness, infants found to have perineal sensation are likely to be continent as well as have decreased renal complications and improved survival long term (136). The management of neurogenic bladder starts in the NICU in the perinatal period by the urologist, physiatrist, nursing, and family.

SOCIAL ASPECTS

The bladder focus may put considerable strain on the family (137). Children and adolescents with neuropathic bladders using intermittent catheterization have worries about peers discovering catheters used to empty their bladder and regarding leakage of urine. Urinary incontinence does affect self-esteem, and it is important to aim medical management at continence for psychological (138) as well as physical well-being. Urinary continence is an important developmental milestone in individuals with and without spina bifida (138). As mentioned in the previous edition of this book, urinary continence, although important, should not occur at the expense of the kidneys.

NEUROGENIC BOWEL

NEUROGENIC BOWEL DYSFUNCTION

The colon, rectum, and internal anal sphincter are also controlled by autonomic nerves. Parasympathetic innervation is from S2 to S4, whereas sympathetic fibers arise from the lower thoracic and lumbar segments. Voluntary somatic motor and sensory nerve supply for the external anal sphincter is from S2 to S4 through the pudendal plexus. Coursing through the spinal cord, these nerves have direct connections with the integrating supraspinal centers in the pons and cerebral cortex (56).

Colon peristalsis propels feces into the rectum. The gastrocolic reflex increases peristalsis for about 30 minutes after food intake. Rectal fullness initiates an autonomic stretch reflex, with relaxation of the internal anal sphincter, and creates a sensation of predefecation urge. In contrast, the voluntarily controlled external sphincter remains contracted to retain feces. When the situation warrants, this action is further enhanced by voluntary contraction of the levator ani, and gluteal muscles. Defecation occurs when the external sphincter is voluntarily relaxed (56).

Many children with spina bifida have a patulous anus, absent cutaneous reflex response, and perianal sensory deficit. This indicates dysfunction of S2 to S4 segments and leads to fecal incontinence. With lesions above L2, there can be an intact reflex arc to maintain perineal sphincter tone, despite absent rectal sensation.

BOWEL MANAGEMENT

Fecal incontinence and constipation are common problems with spina bifida. The external anal sphincter can be weak and patulous. The reflex arc can be interrupted, causing an interruption of innervation to the anal sphincter. There are also motility issues affecting the entire colon.

Bowel management is essential for a successful bladder program in individuals with spina bifida. Not only are gastrointestinal (GI) symptoms and incontinence, with its self-esteem issues, reduced, successful treatment of constipation may improve bladder storage and reduce the frequency of UTIs. Daily bowel movements should be a goal. Aspects of daily bowel care include drinking enough fluids, using an osmotic laxative such as polyethylene glycol, a high-fiber diet to bulk up the stool, digital rectal stimulation or a glycerin suppository in infants and younger children, a bisacodyl suppository 5 mg rectally in younger children and 10 mg rectally in those at least older than 2 years (usually school age for the higher dose). The suppository will not work if placed directly on the stool in the rectum, so careful placement on the rectal mucosal wall is essential. In those with a patulous anus, suppositories may be ineffective because of inadequate ability to retain it. Although the use of the gastrocolic reflex is questionable in the spina bifida population, it is still advised to try having the bowel movement approximately 20 to 30 minutes after the nightly meal. (The morning or afternoon meal are both okay, too, but secondary to schedules, it may be difficult to embark on a bowel program just before or during school or work.)

If the bowels are void of stool, incontinence of stool and urine is less likely. With severe constipation seen on an abdominal film, or with palpable stool still in the abdomen, the previous procedure should be followed along with an enema. If the anus is patulous, a cone tip may be needed to administer the enema and keep the fluid within the rectum for a sufficient length of time. Anatomic bowel obstruction should be ruled out by abdominal x-ray in severe constipation before a colonic cleansing enema is performed. A surgical procedure may be necessary, such as a catheterizable appendicocecostomy through the abdominal wall to flush the large intestine from the proximal end with an enema (139). This is known as the ACE (antegrade colonic enema) procedure or MACE procedure (M for Malone who popularized the procedure). The placement of a cecostomy tube is another method to access the bowel for antegrade enema. A successful bowel program maintains bowel continence and can be performed independently or with minimal supervision.

Anorectal manometry and biofeedback in the presence of intact or partial rectal sensation anocutaneous

reflex offer encouraging results. Rectal sensation is considered normal when a rectal balloon inflated with 10 mL of water or less is perceived. The external sphincter activity can be recorded with surface electrodes. Repeated sessions of inflating and deflating the balloon comprise the biofeedback training (56). This can be done during a urodynamic procedure. For some patients, this will work to see the tracings and surface electromyography (EMG) electrode recordings when attempting to volitionally close the anal sphincter.

Social acceptance from peers is important and leakage of stool can be difficult in school and other social situations. Important goals include continence of stool by grade school entry as preparation for independence and increased career options.

ORTHOPEDICS

OVERVIEW

Children with spina bifida are prone to multiple orthopedic issues during the course of their lifetimes. Many of the problems such as scoliosis, hip dislocation, and the ability to walk can be predicted by understanding the effects the neurologic deficits will have on normal motor control and development. The goal of the multidisciplinary team is to anticipate these orthopedic issues and discuss realistic options and goals with the individual with spina bifida and his or her family. Surgeries designed to improve range of motion may be short-lived because of lack of compliance with the postoperative plan and/or unrealistic expectations.

SPINE

Spinal deformities are common in this population and can be grouped as congenital, or paralytic in nature (140,141). The common descriptions of spine deformities are classified as kyphosis, lordosis, and scoliosis. The probability of development of scoliosis tends to follow the neurologic level. Historically, those with thoracic lesions have an 80% to 100% chance of developing scoliosis, while lumbosacral levels have only a 5% to 10% risk. Stratification of spine risk defined by neurologic level makes intuitive sense. Glard and colleagues grouped children into four groups to predict spine deformities: Group 1 was L5 and below, Group 2 was L3 to L4, Group 3 was L1 to L2, and Group 4 was T12 and above (142). Based on these categories, Group 1 tended to have a low probability of developing spine deformities, Group 2 had a medium risk, and Groups 3 and 4 had a high probability of developing spinal deformities (143). Scoliosis tends to progress most rapidly during growth periods, especially during puberty. Scoliosis can affect sitting balance, create abnormal weight distribution, and increase the risk of pressure ulcers. Large curves can compromise respiratory capacity, cause functional changes, pain, alter body image, and impact ambulation. Realistic goals of the procedure, along with potential complications, should be discussed prior to surgery. These include improved sitting, reduced pelvic obliquity, impact on functional status, and ambulation. Most children with spina bifida and significant scoliosis are nonambulatory, so fusion to the pelvis is generally performed. Fusion to the pelvis is necessary to control pelvic obliquity. For the rare ambulatory child with significant scoliosis, discussions about fusion to the pelvis are important considerations (144,145). However, fusion to the pelvis can also interfere with sitting posture and transfers to level and other surfaces. The impact on function and self-perception after surgery remains controversial (146). Complications after surgery are common and include a high risk of infections, pseudorthrosis, and instrument failure. It has been noted that it takes several months to get back to presurgery ambulation baseline.

Treatment of the different spine deformities is grouped into observational, nonsurgical, and surgical. Curves less than 20 degrees may not progress but warrant observation (141). Nonsurgical options include bracing, seating, therapy, and complementary techniques. Spine braces are mainly thoracic lumbar sacral orthosis (TLSO) and use three points of pressure to maintain alignment of the spine. Wheelchair seating can be incorporated to optimize spine position using molded systems or lateral support. However, molded seating systems often encourage spinal curve and have to be redone more frequently than those that are noncontoured. Therefore, a TLSO is a good option to encourage the spine to be in as straight a posture as possible, especially during sitting when the pressure on the spine is the greatest (147). The pressure on the spine is increased in scoliosis (148). The pressure on the spine is greatest in sitting, followed by standing, and least in the supine position (147). One must be cognizant that bracing typically does not halt curve progression; however, bracing can be useful to improve sitting balance (149). Surgical options should be considered when spine curves are above 50 degrees and the child is at an appropriate developmental level. Different approaches for fusion include anterior and/or posterior fusion. Tethered cord, Chiari malformation, and syringomyelia must be ruled out prior to instrumentation and fusion (150). Tethered cord release can benefit curves less than 40 degrees. For curves greater than 40 degrees and/or in thoracic-level myelomeningocele, there is no reduction in degree of scoliosis after tethered cord release (151).

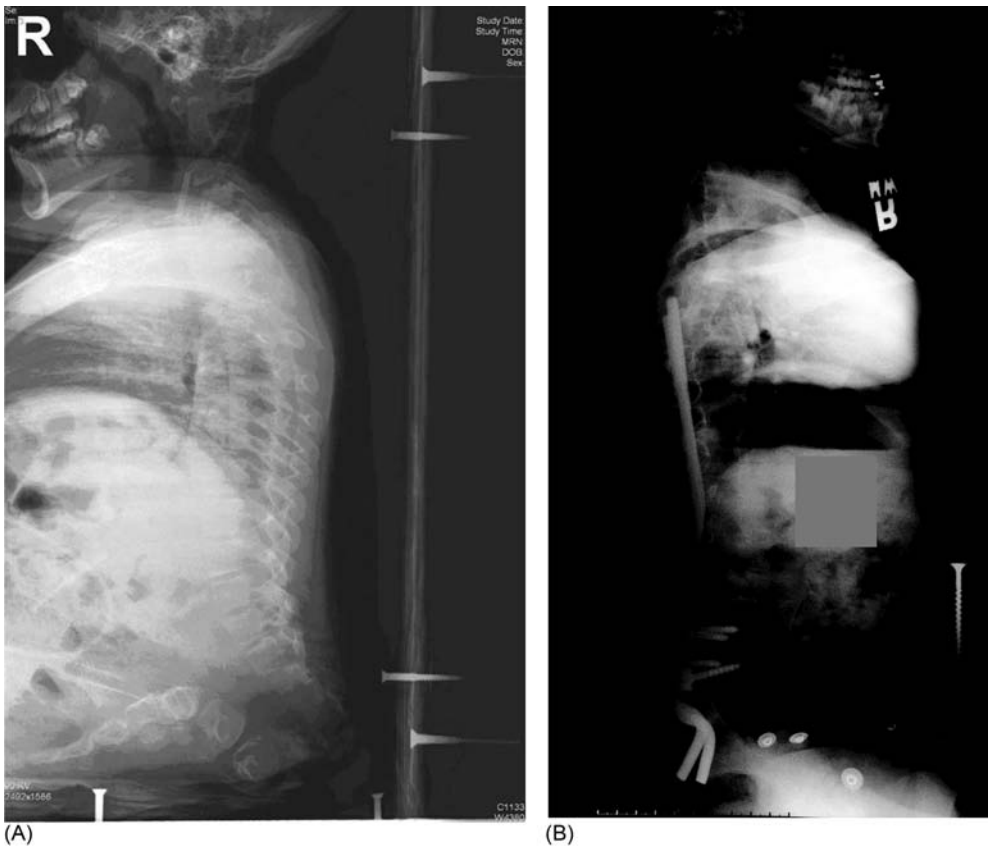
Kyphotic deformities in this population are especially challenging. The child with a high lesion in the thoracic area may be born with a congenital kyphosis and gibbus deformity (Figure 15.7). The x-ray is of a low-lying gibbus which is difficult to manage surgically; see Figure 15.7A, the x-ray after surgical intervention

for kyphosis (not the same patient as in Figure 15.7B). These structural abnormalities not only cause seating and mobility issues, but also present the clinician with challenges in maintaining skin integrity. The deformity can affect the development of the chest, and impair breathing and eating. Treatment options include conservative management with bracing and seating modification to much more aggressive approaches. Kyphectomy and limited posterior fusion done at an early age is one option. Older children with severe kyphosis can benefit from posterior spinal fusion. These surgeries are known for their high complication rate (89%) (152). In one series, 22% required shunt revision within 6 weeks due to surgically induced altered CSF dynamics (152) (Figure 15.7). Nolden and colleagues have described a kyphectomy technique with a decancellation vertebrectomy and preservation of the dural sac where morbidity is reduced while still achieving significant correction (153).

Newer surgical treatments for neuromuscular scoliosis kyphosis have evolved over the past decade. Techniques to deal with the growing child have encouraged the development of fusionless surgeries. Specific goals of these techniques are to delay definitive surgery until the child has reached a more optimal size, allow chest development, improve lung capacity, and sometimes avoid aggressive surgery. These techniques include growing rods, intervertebral stapling, and use of vertical expandable prosthetic titanium rib (VEPTR) (154,155,156,157) (Figure 15.8).

HIPS

The development of the hip and associated problems is related to the neurologic level. Muscle weakness and muscle imbalance likely contribute to the high rates of hip problems. For example, patients with an L3 level of involvement lack hip abductor strength and the unopposed hip flexors and adductors can lead to hip dislocation (158) (Figure 15.9). In a cohort study of myelomeningocele patients, aged 9 to 11 years, Broughton and colleagues found significantly higher rates of hip dislocations in patients with involvement at a thoracic, L1/2, or L3 levels (greater than 50%) as compared with patients with involvement at L4 (33%), L5 (20%), and sacral levels (2%) (159,160). In the same study, hip flexion contractures were significantly higher in patients with involvement at thoracic (22 degrees) and L1/2 levels (32 degrees) as compared with patients with involvement at L4 (9 degrees),



(A) (B)

FIGURE 15.7 (A) Congenital structural kyphosis with a sharply angled curve, or gibbus deformity, associated with thoracic-level spina bifida. (B) Corrected congenital structural kyphosis associated with thoracic-level spina bifida (not the same patient).

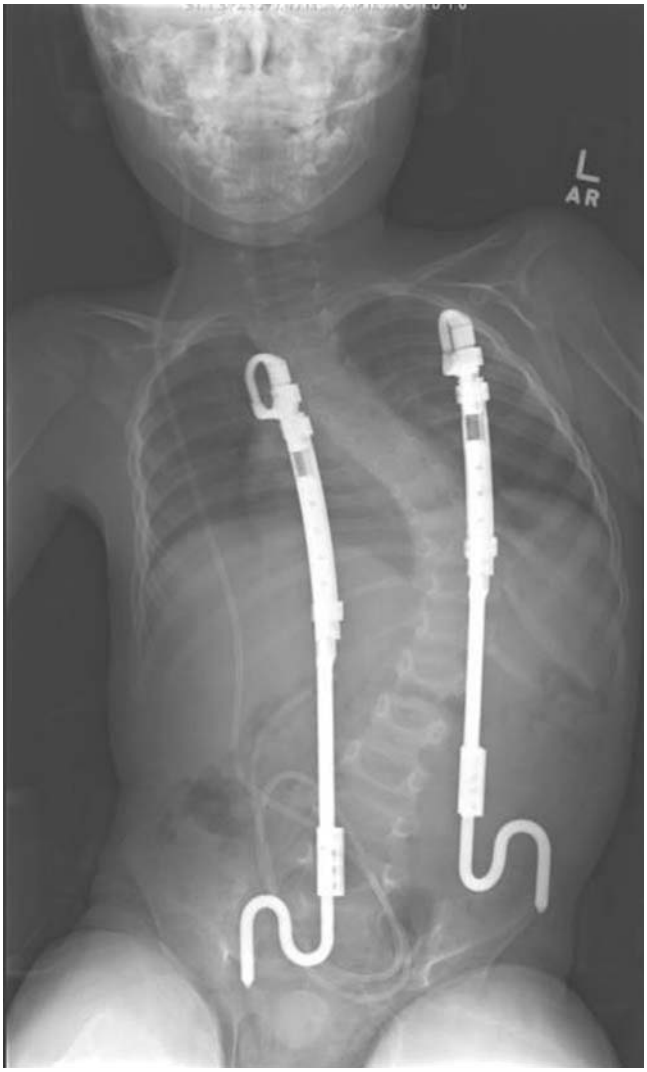


FIGURE 15.8 Child with spina bifida and scoliosis treated with vertical expandable prosthetic titanium rib (VEPTR). Note the spinal dysraphism with increased intrapedicle width and ventriculoperitoneal shunt.

These forces can result in hip subluxation and dislocation. This process occurs not only in the higher lumbar levels, but also in the mid- and lower lumbar segments. Weak or absent hip extension and abduction are directly related to hip dislocations. Unilateral hip dislocations tend to cause pelvic obliquity, and surgery has been advocated. Bilateral hip dislocations generally do not require surgical interventions. Heeg and colleagues found that it was more important to have a level pelvis and good range of motion for ambulation to have located hips (163).

Surgical treatment to reduce dislocated hips remains controversial. Multiple treatment methods have been developed to address hip dislocations, including tendon transfers to correct muscle imbalance (ie, iliopsoas transfer) (158,164,165), adductor and external oblique muscle transfers (166), osteotomies to provide bony stability [Chiari osteotomy (167,168), and varus osteotomy of the proximal femur] (158,164,169). However, results of these surgeries fail to show improvements in walking (169). A motion analysis study by Duffy and colleagues showed patients with successfully reduced hips following iliopsoas transfers had no improvement in pelvic obliquity and significantly worse pelvic rotation motion and worse hip abduction/adduction range during gait (170). Meanwhile, complication rates of surgical hip reduction are high. Postoperative hip instability is reported around 30% to 50% (161,164,167–169,171). At the same time, postoperative stiffness can result in limitations in sitting (171,172). Furthermore, pathologic fractures occur due to disuse osteopenia (161,167,171). The limited benefits and high complication rates raise questions about surgical reduction of dislocated hips in patients with myelomeningocele (172,173). One group that might benefit from surgical open reduction of a dislocated hip are sacral-level patients with intact hip abductor strength who may benefit from restoration of the fulcrum (173,174).

L5 (5 degrees), and sacral levels (3 degrees). The neurosegmental level of involvement also remains the most influential determinate of ambulatory ability, and not the reduction status of the hip (161,162).

The etiology of the hip dislocations can be explained as follows. Children with thoracic-level lesions have no muscle influence on hip stability and may or may not develop hip dislocation. These children tend to frog-leg (hip abduction and external rotation) when lying down and develop contractures of the hip flexors and external rotators. In addition, the tensor fascia lata becomes contracted and may need to be surgically lengthened if it affects positioning. Children with high lumbar lesions have an imbalance of muscle activity around the hip joint. The active hip flexors and adductors (L1–L2) with unopposed abduction and extension tend to result in persistent coxa valga and development of acetabular dysplasia.

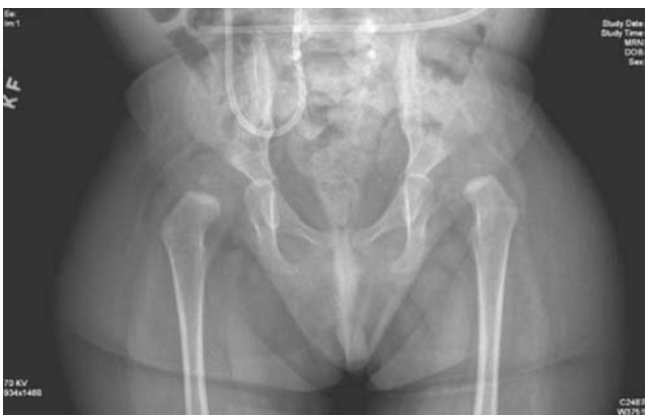


FIGURE 15.9 Bilateral hip dysplasia. Note the dysplastic acetabulum, femoral head migration, and broken Shenton's line. Ventriculoperitoneal shunt is in place.

The management of hip contractures typically takes greater priority over managing dislocations in the spina bifida population. Evaluating a group of low lumbar myelomeningocele patients with unilateral hip dislocations, Gabrieli and colleagues demonstrated that some patients walked with good hip symmetry (kinematic parameters of the hips were within 10 degrees for more than 50% of a gait cycle) (174). Hip range of motion, in particular symmetry of hip abduction/adduction, had the greatest influence on gait. Adductor magnus lengthening, and in severe cases proximal femoral osteotomies, might be considered to address hip contractures (10). Preventative and nonsurgical interventions include lying prone for 30 minutes daily with a flat level pelvis.

KNEES

The knee motion is influenced by the muscular control of the quadriceps and hamstrings. Knee flexion contractures are a common occurrence at all neurologic levels, but are seen in a higher frequency in thoracic and high lumbar lesions. Weak quadriceps and positional factors, along with fractures and spasticity, have been proposed as the etiology.

Treatment is geared toward preventive strategies of stretching, bracing, and standing. Surgery is indicated for contractures greater than 20 degrees that interfere with function. Aggressive posterior capsule release is used in thoracic and high lumbar lesions. For ambulatory patients, soft tissue lengthening and/or releases can be performed (175,176). If the knee flexion contracture is not severe, the "guided growth" approach developed by Klatt and Stevens can be used. This involves placing tension band plates; gradual correction of the deformity is achieved by the use of tension forces to guide bone development in growing children (177,178). Knee hyperextension can be seen in the L3 level from unopposed contraction of the quadriceps. Serial casting and capsule releases may be required. Ambulatory patients have exaggerated trunk motions, which results in abnormal sagittal plane forces across the knee joint (179). This can result in genu valgum and can cause knee pain and may require more aggressive bracing as a preventative strategy. Bracing such as knee ankle foot orthosis (KAFO) or forearm crutches to minimize further injury to the knee joint may be necessary (180).

TIBIA

Rotational deformities in the tibia are fairly common and can have a functional impact on ambulation. Internal and external tibial torsion can both affect gait patterns. In-toeing is often seen in L4/L5 neurologic levels and is related to muscle imbalances, particularly in the hamstrings. The unopposed medial hamstring may contribute to internal rotation of the leg. Derotational osteotomy can be of benefit. External tibial torsion is seen in older children and can exist with knee valgus, which can be a source of knee pain. External tibial torsion greater than 20 degrees may benefit from derotational osteotomy to align the foot and leg (181). Derotational surgeries should be used only in those who are ambulatory in the community (182).

FEET

Ankle valgus (Figure 15.10) is often seen in ambulatory patients and can manifest as problems with skin

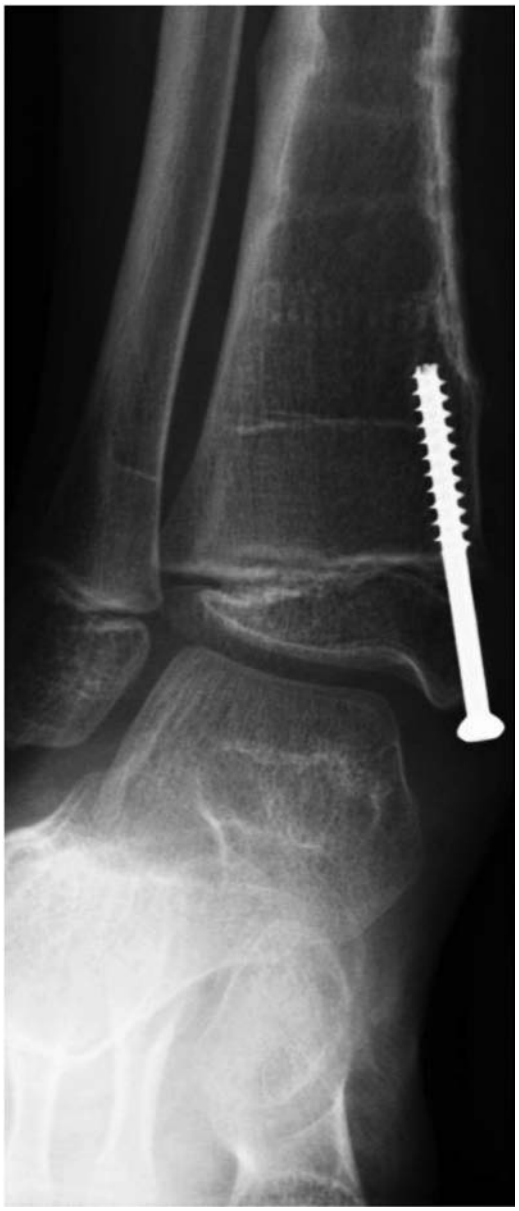


FIGURE 15.10 An anterior–posterior (AP) ankle film of an 8-year old with spina bifida and ankle valgus. A screw has been placed across the medial portion of the distal tibial growth plate.

breakdown over the medial foot. It can be mistaken as an AFO issue but one must remember to examine the frontal plane alignment of the ankle. The deformity is usually at the ankle joint with lateral tilting of the ankle plafond (ankle joint ceiling formed by the distal tibia). In a growing child, treatment consists of medial distal hemiepiphysiodesis with either an eight-plate or a medial malleolar screw (183); see Figure 15.11. Skeletally mature patients require an osteotomy. Ankle valgus can also be secondary to subtalar tilting, which requires a calcaneal osteotomy.

Foot deformities occur in the majority of patients with spina bifida. Foot management is based on developing a plantar-grade foot to protect vulnerable soft tissues. Clubfoot (talipes equinovarus) and congenital vertical talus (CVT) are found at birth. The acquired deformities of equinus, calcaneus foot, or cavovarus develop later and are related to the neurologic level. In general, soft tissue releases and tenotomies are more effective than tendon transfers. Also, foot fusion surgery is best avoided because it has been associated with an increase in foot ulcers and skin breakdown. Clubfoot deformity in these children can be more rigid than in other populations. The foot classically has hind foot varus and equinus; the forefoot is supinated and adducted and is rigid. The Ponseti method (casting and limited surgery) can be successful in patients with spina bifida but upwards of two-thirds of patients have a relapse. Relapse is treated with recasting or open surgery (184).

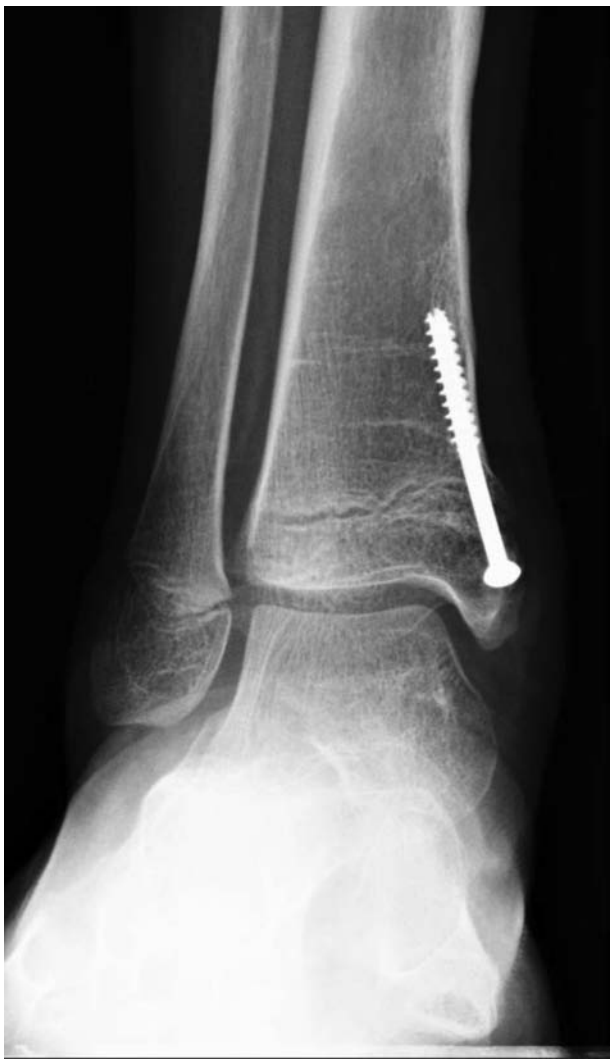


FIGURE 15.11 Three years later, the ankle valgus shown in Figure 15.10 is resolved secondary to the temporary hemiepiphysiodesis effect of the screw.

CVT deformity or rocker bottom foot is a nonreducible dislocation of the navicular on the talus. The talus is in equinus, and the Achilles tendon is short. The talus on radiographs is vertically positioned, and clinically the talus is medially located. Muscle imbalances are the implicated forces in this deformity. Serial casting and limited surgery can be effective but relapses are common (185). Surgical intervention is often required and best results are achieved before age 2 years (186). Calcaneus deformities occur when the anterior tibialis, toe extensors, and peroneal muscles are unopposed. This is seen in those with L4-level spina bifida. The calcaneal deformities affect the gait pattern and can cause the skin over the heel to break down. Stretching is not effective, and surgery is indicated. This includes tendon transfers of the anterior tibialis and anterior capsule release. Some plantar flexion power can be generated with anterior tibial tendon transfer to the calcaneus but this is generally not enough to walk without braces. Equinus deformities generally require an Achilles lengthening procedure. Cavus foot deformity is found in sacral-level injuries. Intrinsic muscle abnormalities lead to high arches and toe clawing. These deformities can cause areas of increased pressure and increase the risk for skin breakdown. Orthotics and extradepth shoes may reduce pressure points. Surgery is indicated if these measures fail. Plantar fascial release and multiple bony surgeries can be done. Toe deformities such as hammer toes often require tendon procedures and fascial release (187).

CLINICAL PEARLS

- Scoliosis associated with spina bifida can occur at any neurologic level, but is most common in the higher lesions. Thoracic and high lumbar levels almost always develop these spinal deformities.
- Hip dislocations are most common in the L3 level based on muscle imbalances of hip flexion and adduction being present while the opposing muscles are weak or absent.
- Foot deformity treatment is geared toward developing a plantar-grade foot and minimizing pressure areas.

Figure 15.12 shows scoliosis noted in a child with lumbar level meningomyelocele.

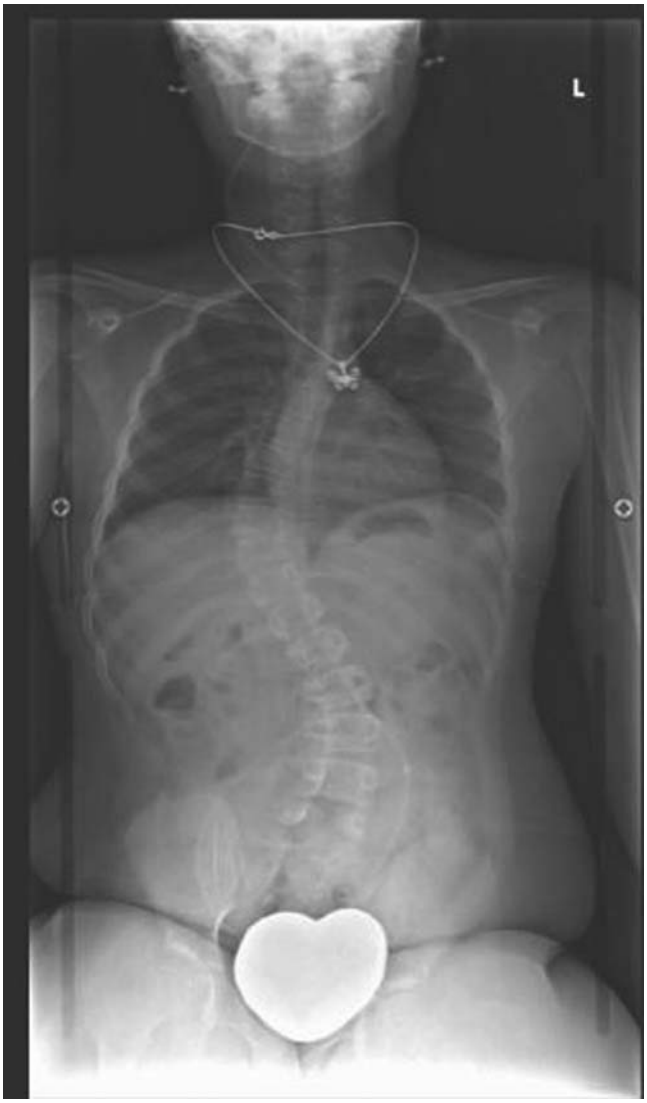


FIGURE 15.12 Scoliosis noted in child with lumbar-level meningomyelocele.

REHABILITATION

Children with spina bifida may require more specialized medical and rehabilitation care than is typical of many neurogenetic disorders (188,189). The pediatric/adolescent physiatrist works with a multispecialty interdisciplinary team to coordinate physical medicine and rehabilitation care for the individual with spina bifida. The coordinated plan of care will change based on the level of the lesion, the cognitive ability and developmental age of the individual, signs and symptoms including spasticity, impaired balance, central hypotonia, family resources, and community resources. This plan should be family-centered and include all pertinent disciplines. Communication with the child's local therapists and other providers is important to maximize their potential and the follow-through.

MANAGEMENT OF MUSCULOSKELETAL DEFORMITIES

Conservative management of potential or existing musculoskeletal deformities begins in the newborn and should continue as part of daily care thereafter. Passive range-of-motion (PROM) exercise is applied to all joints below the level of paralysis, with special emphasis on joints with evident muscle imbalance. The infant should not lie constantly in one position, but should be moved and turned frequently. This practice must be taught to parents, not only to mitigate contractures, including those related to gravity, but also to avoid breakdown of the anesthetic skin. For the same reason, splints must be used with great precaution, removed frequently to check for skin irritation, and adjusted or discontinued if such a problem occurs. PROM and splints are advisable after surgical correction of deformities to maintain joint mobility gained by the procedure. Strengthening exercises are sometimes beneficial for partially innervated muscles or after surgical muscle transfer for improving strength or function. Depending on the functional level of the lesion, strengthening is important for increasing cardiopulmonary endurance, ambulation training, and upper extremity strength for manual wheelchair use and assistive devices. The caregivers should be encouraged to facilitate functional independent mobility.

Examination of motor function in the neonate is based primarily on observation of spontaneous movements, presence or absence of deep tendon and infant-like reflexes, habitual postures, passive joint motion, and tone. For example, consistently maintained hip flexion, particularly when passive extension is incomplete, is a sign of hip extensor weakness. Palpation of muscle bulk is helpful because atrophy may be evident with severe or incomplete paralysis in particular muscles. In assessing motor or sensory function, the presence of spinal reflex withdrawal or triple flexion of hip, knee, and ankle should not be mistaken for voluntary motion and preserved sensation, particularly in high spinal lesions. A normal asymmetric tonic neck reflex elicited in the arms without response in the legs suggests lower extremity paralysis (87).

DEVELOPMENT

Development is the natural and predictable sequence that an individual progresses through to attain skills in multiple domains. Children with physical disabilities may not be able to accomplish these tasks, given their physical and cognitive limitations. The impairment will affect activities and participation. Motor acquisition can be predicted based on the level of the lesion, which affects normal balance, coordination, and postural control. However, this is a prediction, and each child will advance based on his or her own development, cognitive and physical strengths, and challenges.

FIRST SIX MONTHS OF LIFE

Most children with lipomeningocele do follow normal development, attaining head control, fine motor skills, and language. For infants with myelomeningocele, this can be disrupted in light of hydrocephalus, Chiari II malformation, medical complications, and neurodevelopmental issues (see Table 15.2).

SIX TO TWELVE MONTHS OF AGE

This is a critical time for gross motor development, where most typically developing children are sitting, crawling, and walking. Predictably, children with spina bifida can be expected to have delays in this domain. The individual's motor function will allow the medical team to discuss realistic expectations for family members. Early mobility mirroring normal development should be incorporated into the rehabilitation plan. Positioning should include supine, prone, and sitting upright. Lack of environmental experiences can lead to sensory/motor deprivation and affect developmental potential.

Head control is a crucial milestone and prerequisite for emerging skills. Most children achieve this skill irrespective of the level of lesion. Delays are mainly central in etiology. Children with high thoracic lesions lack adequate trunk and abdominal muscles to get and maintain sitting balance. Compensatory strategies include prop sitting,

TABLE 15.2 GROSS MOTOR SKILL ACQUISITION AND THERAPEUTIC INTERVENTION

	T 12 AND ABOVE	LUMBAR 1–2	LUMBAR 3	LUMBAR 4–5	SACRAL
Functional Level of lesion			Delayed, can sit independently. Therapy to work on transition skills.		
Rolling/Sitting	Delayed, related to lack of hip control, and trunk may be impaired. Can prop sit and uses upper extremities. Therapy to address head/neck/trunk development. Compensatory strategies.	Delayed, may have hip flexor/adductor weakness. Can sit but may use upper extremities. Therapy to address trunk and hip girdle strengthening. Compensatory strategies.		Generally on time.	Generally on time.
Floor mobility	Primary rolling but can combat-crawl and has limited bottom scooting, very energy demanding and may lead to skin trauma, early introduction of mobility devices.	Primary rolling, may army crawl, scoot on bottom along with some modified crawling, very energy demanding, early use of mobility devices.	Rolling may scoot on bottom, modified crawling, consider early mobility devices.	Rolling and four-point creeping, will transition to stand generally by 2.5 to 3 years.	Rolling, crawling/ creeping to walking generally on time.
Ambulation	Only with adapted equipment such as static and dynamic standers, or orthotics (HKAFO, KAFO, RGO) with walkers, mainly therapy and household mobility. Wheelchair mobility important to emphasize.	With adapted equipment static or dynamic standers, or orthotics KAFO, RGO, with walkers, mainly household ambulation. Wheelchair mobility important to emphasize.	With orthotics, KAFO, floor-reaction AFO, AFO, walkers, and crutches. Household and limited community ambulation. Antigravity quadriceps strength can walk with above equipment combinations; however, with increase in height and weight, most will lose ability to ambulate in teen years as this is too energy-consuming.	With orthotics AFO, GRAFO or possible KAFO, the child should be an ambulator unless significant contractures or orthopedic deformities prevent successful bracing or upright posture.	Excellent potential for community ambulation, may need orthotics including SMAFO, UCBL maximal control shoe insert.

Abbreviations: SMAFO, supra malleolar ankle foot orthosis; UCBL, University of California Berkeley Laboratories.

rolling to side, and pulling up. Sitting is necessary for play and hand skills, and appropriate equipment should be used. Sitting is also helpful for self-dressing. Sitting may be delayed in children with mid-lumbar and lower lesions, but they will likely achieve this skill. Rolling is typically delayed in children with thoracic and high lumbar lesions. To roll, a child uses their head, trunk, and legs. Thoracic and high lumbar muscle weakness delays this skill until the child can figure out adaptive motions, including using momentum to propel the legs. The therapist can help the parents incorporate activities in play to help the child learn to roll. Most will learn to roll by 18 months of age.

Floor mobility is a way for a child to move from place to place to obtain a toy or to explore their environment. Depending on the functional level of the lesion, the child will have different methods of mobility. Children with high-level lesions tend to roll, and in sitting, lean forward over the legs and combat-crawl dragging their lower extremities behind. Crawling uses hip flexor and knee extension strength and therefore will be unlikely in those with high-level lesions.

AMBULATION/MOBILITY

As stated, the job of a child is to explore his or her environment. The ability to ambulate and gait abnormalities have a direct relationship to the neurologic level of the spina bifida. Mobility can be achieved through various means, including self-propulsion of a wheelchair, power mobility, adapted equipment, and orthotics. Introduction of equipment should be developmentally appropriate. Introduction of dynamic standers can be done at the developmentally appropriate time to stand in thoracic and high lumbar levels. The choice is best made by evaluation of the child's need for additional support of head and upper thoracic support, ability of the child to tolerate standing fully upright, and potential to advance to walking with a walker. Devices include full support supine standers for children with poor head and trunk control, mobile prone stander, parapodium, and swivel walkers. The advantage of supportive standing and walking is not only mobility, but also passive stretch of the joints in the lower extremities, pulmonary and GI, bone health (190), and being at the upright level of age-matched peers.

Orthotics are used in all neurologic levels of spina bifida. Over the past 15 years with the use of gait analysis, the importance of hip abductor strength has become more appreciated (191). Weakness of the hip abductors has been correlated with the presence of abnormal knee valgus (179). Weak hip abductors have been identified as the most important muscle in changing gait kinetics (192). Individuals with myelomeningocele can have a decrease in the 6-minute walking distance and decreased aerobic capacity associated with weak hip abductors. Children and teens with spina bifida who are ambulatory and have associated weak hip abductors would benefit from endurance training and muscle strengthening (193).

The child with a thoracic and high-level myelomeningocele requires much more sophisticated bracing than the child with lower lumbar levels. Hip knee ankle foot orthosis (HKAFO) and KAFO stabilize the joints in the lower extremities to allow upright positioning. HKAFO is used when hip instability interferes with knee alignment. With the HKAFO, a child must use a walker or Lofstrand-type crutches and move the brace forward by either leaning or lifting to achieve ambulation. This is a difficult skill to acquire, plus the donning and doffing of the bracing is cumbersome, which is the reason why many children abandon this as they grow. Use of reciprocal gait systems includes a cross-linked hip orthosis, a reciprocal gait orthosis (RGO), or a free hinged gait orthosis such as a hip-guided orthosis (HGO). The isocentric RGO system uses a cabling system. The brace provides structural stability during the stance phase on one side while the opposite side advances. Simply putting hip flexion on one side causes hip extension on the opposite side through the cabling system.

In the Orlau swivel walker, used with high lumbar and thoracic level spina bifida, an attachment to the baseplate converts lateral trunk movements to forward propulsion. This adaptation lowers the energy requirement for walking compared with parapodium, although ambulatory function remains at household level. It is typically used with a walker and/or Lofstrand crutches. There are advantages to using a walking system in young children, as Mazur's study showed fewer fractures and pressure ulcers when comparing those who strictly used a wheelchair to those who used a walking system (194). Implementation of these types of braces is best employed when children are around 3 years of age. Earlier implementation may be possible with a trained physical therapist and a motivated preschooler and family. For an individual to be successful with a thoracic and high lumbar neurologic level, there must be a commitment to therapy to learn to use the extensive bracing and assistive device. There are positive physical and psychological benefits from ambulation. Thomas and colleagues evaluated oxygen consumption, energy cost, and velocity over time in children with T12-L3-L4 neurologic level spina bifida using HKAFO and RGOs over a 3-year period (195). All of the participants, except one who used a reverse walker, used forearm or Lofstrand crutches. Children using HKAFO had a faster velocity than those using RGOs, but had similar oxygen consumption. A lower oxygen cost for those adolescents using HKAFOs and a faster velocity were found compared to those who did not use these. This change was not found with RGOs. Over 50% continued to ambulate using HKAFOs compared to less than 15% of those using RGOs. The children who continued to walk into adolescence were lighter in weight than those who stopped walking, even if the ambulatory and nonambulatory had similar weight gains. The velocity of ambulation

was still less than 50% the speed of those typically developing children and the oxygen consumption was greater. Ambulation velocity and oxygen consumption were the major factors in continuation of ambulation over the course of this study. Those using HKAFOs did not have greater contractures at the hips and knees than those using RGOs. Ambulation for a young child with poor upper extremity strength may be better with an RGO and then as the child matures, an HKAFO may be more advantageous with improved upper extremity strength and the desire to keep up with one's peers.

Mid-lumbar lesions, L1– to L3, have knee extension muscles that have a great impact on ambulation. Hip and knee flexion contractures may be present at the first clinical visit or develop over time.

It is imperative that hip flexion and knee flexion contractures be addressed, as these affect upright position. Initially, twister cables attached to a pelvic band with an anatomically aligned free hip and knee joint, attached to solid AFOs, may give enough support to the knee extensors and allow the young child to develop his or her own inherent knee extensor strength. The child may need to use a reverse walker to assist. Adaptive aides such as reverse walkers for younger children and Lofstrand crutches in slightly older children (age 3 or above) can be very helpful for balance and stability especially for uneven surfaces and outdoor terrain. Wheelchair use should be considered early to facilitate independent community mobility as young as 15 to 18 months. The majority of these children can hope to have household ambulation with limited community ambulation. All children with spina bifida can be expected to have a delay in ambulation even at the lower sacral levels.

In children with the lower lumbar L4– to L5, parents can expect ambulation by age 4 years (196). Typical gait patterns include a Trendelenburg associated with weak hip abduction and steppage gait associated with weak dorsiflexors. Bracing includes AFOs and GRAFOs. GRAFO is used to assist knee extension and prevent the crouching patterns seen in stance phase. The foot must have some flexibility to accommodate this brace. Community ambulation is possible in these individuals. Knee flexion contractures remain common but hip dislocation is less likely. If the tibialis anterior is unopposed, the foot will be pulled into dorsiflexion with a calcaneus foot. Excessive pronation is common especially in weight-bearing, during standing. Initially when the child is young, an AFO may be used; however, with increase in height and weight a GRAFO may be needed to avoid crouch gait and to assist with knee extension. Bracing studies have shown that the use of ankle foot orthosis (AFO) in children with L4 sacral-level lesions had improved energy expenditure. Walking speed and stride length increased, while energy costs decreased, when using braces compared to not using braces. This is surmised to be related to the stability that the braces provide (197). If crutches are used, most children cannot learn this skill until at least 2 to 3 years of age. Walkers can be used at earlier ages, and dynamic standers can be used when children should be upright.

Children with sacral-level lesions often learn to walk by age 2 years. Children often walk well without any adaptive aides or braces, although distal weakness can lead to significant foot deformities including pes cavus, which may benefit from the use of orthotics as they grow in height and weight. Also spastic claw foot deformities may develop due to progressive spasticity and may benefit from botulinum toxin injections (off label under 18 years and in the lower extremity) management.

Ambulation is always one of the first questions parents will ask a health care provider: Will my child be able to walk? To address this question, one needs to look at the whole child and all the factors involved. Swank found that sitting balance and neurologic level were good predictors of ambulation potential (198). A study by Williams and colleagues tracked 173 children with spina bifida (196). Thoracic level was found in 35 children, and only 7 walked at 4.5 years. The study followed 10 children with L1/L2 lesion—and 5/10 walked by 5 years. They followed 15 children with L3, and 9/15 walked at 5 years. There were 45 children in the L4/L5 group, and 38/45 walked at almost 4 years. The 68 children with sacral level were able to achieve ambulation by 2 years. Walking was delayed in all groups, and the higher levels abandoned walking earlier than was previously documented. With development, those with a sacral level do not lose ambulation skills (196). Success in maintaining ambulation has been associated with muscle function of the hip abductors and ankle dorsiflexors (199).

Carbon fiber braces may be beneficial (200,201), as well as functional electrical stimulation, to assisting those ambulatory individuals with myelomeningocele. Several current studies with pediatric-age clients are in progress at this time.

Wheelchair mobility should be introduced to all children who will potentially use this as a primary or secondary option. This can be introduced at a fairly young age and children as young as 1 year can efficiently push a wheelchair. This allows them independence to explore the world around them. Wheelchairs should be appropriately configured to meet the needs of the child. A child is not a small adult and should not be placed in a wheelchair he or she can grow into. Seating will be adjusted based on the neurologic level, posture, and balance. Proper cushions, the seat back, seat, and foot rests should be positioned to prevent pressure areas from developing. This may be a paradigm shift toward earlier acceptance of wheelchair mobility as a viable option. The demands of walking increase as the person grows taller and requires more energy. Spine deformity has been well documented to have an impact on ambulation. Scoliosis surgery can change ambulation patterns.

Young children or adolescents who, because of their level of paralysis or due to associated medical complexities (eg, spinal cord syrinx), cannot use their upper

extremities, or do not have the cardiopulmonary capacity to effectively propel a manual wheelchair in the community or to conserve their endurance for their academic needs, may find a power wheelchair to be necessary to facilitate their independence. Children and adolescents who are always pushed by an adult develop a sense of dependency rather than independency. Having effective mobility in the home setting and community allows the child and adolescent with spina bifida to develop his or her own sense of individuality and independence similar to their able-bodied siblings or peers.

LATEX ALLERGY

Latex rubber gloves have been in common use over the past 30 years, leading to an increased incidence of latex allergy in health care professionals, as well as patients with early and/or frequent exposure to latex products, such as those with spina bifida (202). Today, latex allergy and latex precautions in the spina bifida population are well-known concerns in the medical community. Prior to the 1980s, latex allergy was a largely unknown entity. Allergy to latex and the potential for anaphylactic allergic reactions came to medical attention in the 1980s in increasing numbers, with the increased usage of latex gloves for barrier protection from hepatitis and HIV. In the early 1990s several children with spina bifida experienced anaphylaxis during surgery due to latex (203). It was theorized that the increased demand for latex gloves due to the HIV/AIDS epidemic led to poor quality control with production of latex gloves and more antigenic material in the gloves. The protein found on the outside and the inside of the latex glove are not the same and correlate to different latex allergies in children with spina bifida and health care workers. In children with spina bifida, the major latex allergens are Hcv b 1, 3, and 7 and for health care workers they include Hcv b 2, 5, 6.01, and 13 (204). Peixinho and colleagues in 2008 found that the concentration of Hcv b 1, and Hcv b 3 were significantly higher on the external surface of the gloves compared to the internal surface, which had significant high levels of Hcv b 5 and Hcv b 6.02 (205).

With increased awareness since that time, medical facilities typically take precautions with items containing latex and frequently do not allow products that have high loads of allergen, such as latex gloves (especially those with powder), latex balloons, rubber plungers, blood tourniquets, and rubber dams for dental procedures. It is also recommended that toys and other items with latex be avoided. Several prevention strategies have been adopted by professional organizations in the United States, Europe, and South America to protect both patients and those who are occupationally exposed, including implementing latex-free environments for patients at high risk, switching from powdered to powder-free and latex-free gloves, and providing information seminars about latex precautions to patients and professionals (206).

Various studies have been conducted to determine the percentage of children with spina bifida who are allergic to latex. The prevalence of latex sensitivity (ie, positive IgE skin testing) has been reported as high as 72% in the spina bifida population (207). Bueno de Sa′ and colleagues, in a study of 55 children and adolescents with spina bifida, found that 25% had latex sensitization and 20% had latex allergy, and further identified that current asthma, atopy, and having undergone four or more operations to be risk factors for latex sensitization (208). There are several types of reactions to latex including type I—immediate hypersensitivity manifested by urticaria, nausea, vomiting, faintness, rhinitis, conjunctivitis, bronchospasm, and anaphylaxis; type IV—delayed hypersensitivity, manifested by a papular, pruritic rash, vesicles and blisters; and irritant contact dermatitis, which is nonimmune and due to the chemical in the latex or handwashing (209).

Spina bifida patients with latex sensitivity are at high risk for anaphylactic response to latex-containing products. The propensity for latex allergy in the spina bifida population is increased by early exposure, specifically on the first day of life, and family of origin atopy. Neurosurgical procedures appear to be correlated with increased latex sensitization; intra-abdominal procedures are not (210,211). Studies continue to demonstrate that children with spina bifida have an increased propensity for latex sensitivity and allergy over those who have had multiple surgeries for other diagnoses, implicating that there is something inherent in the condition that predisposes them to this allergy (211–213).

In the 1990s, latex–fruit syndrome—most frequently involving the banana, avocado, kiwi, and chestnut cross-reactivity—was reported. Papaya, mango, bell pepper, fig, tomato, celery, and potato are other foods that are potentially problematic. This list, although comprehensive, may not include all problematic foods. The cross-reactivity is exhibited on radioallergosorbent test (RAST). There can be allergen cross-reactivity between latex and the proteins in these foods. Latex sensitivity and allergy develop over time; therefore, negative RAST tests are not definitive for future allergic reactions. Furthermore, negative skin tests may or may not be reliable and may depend on the source of the allergen. It is not always clear whether latex sensitization precedes or follows the onset of food allergy (214).

Both primary prevention from latex exposure and avoidance of latex for those with existing sensitivity are important for patients with spina bifida. Patients and families should be taught to avoid all products containing latex from birth. The SBA (www.spinabifidaassociation.org) has a number of informational pieces about latex allergy for patients and parents, as well as additional resources.

A detailed history regarding latex sensitivity and allergy is important. The management of this condition includes: (a) avoidance of latex-containing products and

cross-reactive foods, (b) wearing a MedicAlert bracelet, (c) ongoing latex avoidance education, and (d) EpiPens for those with severe allergies. Avoidance of latex even as early as day one of life and an anaphylaxis kit are recommended (215). Latex precautions should start as early as the first day of life and should be continued throughout the life span of the individual with spina bifida. For the adult with spina bifida, this includes attention to potential latex exposure in the workplace, as well avoidance of latex condoms. Potential risks must be discussed at each visit.

SELF-CARE

Children with spina bifida should be encouraged to acquire independence in age-appropriate activities of daily living (ADLs) at an early age, in order to optimize their opportunities for successful independence as adults. Fine, gross, and visual–motor skills are rarely significant enough to account for delays in ADLs. Delays are most likely related to difficulties with motor planning, balance, visual-perceptual, visual–motor, and organizational skills. Despite adequate intelligence and upper extremity function, delays in ADLs including dressing and bathing are often present. Anticipatory guidance regarding familial expectations of age-appropriate self-care activities and occupational therapy can be very beneficial to assist the family and child or adolescent with techniques to improve skills toward independence. The occupational or physical therapist can review necessary home equipment to assist with independence in self-care, such as bath/shower equipment or specialized toilet chairs (56).

OBESITY

Obesity can be a significant problem for individuals with spina bifida, resulting in further compromise in mobility, interference with self-care activities such as carrying out urinary catheterization, an increased risk for pressure sores, as well as risks that occur for individuals without spina bifida, such as type 2 diabetes, hypertension, and cardiovascular disease (216).

In addition, the risk of metabolic syndrome, characterized by obesity, low- or high-density lipoprotein, elevated triglycerides, elevated blood pressure, and insulin resistance, is very high, primarily due to obesity (217). Individuals with spina bifida are at risk for obesity due to several causes including deficits in cognition, and motor and sensory function, leading to the development of an inactive lifestyle and limiting participation in communal life (218–220).

Secondary to paralysis and wheelchair mobility, obesity increases the risk of decubiti. In addition, there is increased stress on the upper extremities, with physical activity. Self-image and poor social adaptation, and acceptance are also compounding factors with obesity and spina bifida; these are important in the populations as the development of positive self-image is greatly affected by social relationships (221).

In a review by Oliveira and colleagues, individuals with spina bifida had impaired cardiorespiratory endurance, muscle strength, body composition, and flexibility compared to healthy peers, and mobility restrictions were present in 26.3% to 61% of participants (222). In their review, Crytzer and colleagues found an obesity rate from 29% to 50%, with a lack of physical activity, decreased energy expenditure, and lower resting metabolic rate playing important roles (223). Van de Berg-Emons and colleagues found that adolescents and young adults with spina bifida are considerably hypoactive (224) and Littlewood and colleagues found that total energy expenditure and body cell mass were reduced for children with myelomeningocele (225). Calculating the BMI is not as useful for individuals with spina bifida, as height calculations may be difficult to accurately assess with acquired orthopedic deformities including neuromuscular scoliosis, lower extremity contractures, and leg-length differences. Shurtleff and colleagues found that using arm span and height are comparable for the calculation of BMI for those with lower level lesions, but not for those with higher level lesions who are more likely to have scoliosis or lower extremity deformity (226). A significant complexity to studying obesity in this population is finding a standard definition and measurement tool for this population with unique upper and lower body habitus (227). Subscapular skin-fold thickness is more reliable to assess for obesity.

Developing interventions for the prevention or treatment of obesity in children and adults for spina bifida can be challenging. For those with spina bifida, opportunities for physical exercise are fewer than those for age-matched peers without disabilities. Preventative anticipatory guidance regarding weight, exercise, and diet should be part of the comprehensive care and education for individuals with spina bifida from an early age, as weight loss may be difficult once the child/adolescent or adult is overweight. Although adolescents and young adults who do participate in physical activities do not report lower body fat, they do report improved health-related quality of life (228). Exercise training seems to improve both cardiorespiratory endurance and muscle strength in individuals with spina bifida (222), and regular exercise and daily physical activity is especially important for adults with spina bifida due to risk for further deterioration in function with increasing weight (223). It may be difficult for children and adolescents to grow into their weight secondary to shortened stature (see the following section). Recommended diet guidelines include decreased caloric intake by 10% to 20%, and a diet low in fat and carbohydrates and high in protein and fiber, with proper vitamin supplementation (56).

PRECOCIOUS PUBERTY

Precocious puberty traditionally is Tanner stage 2 breast development before age 8 years and testicular enlargement before age 9.5 years (229). Precocious puberty is associated with an accelerated growth velocity and early epiphyseal fusion (229).

Individuals with spina bifida and precocious puberty can have marked short stature if untreated. Their short stature results from abnormalities of the hypothalamic–pituitary axis, the Chiari II malformation, and hydrocephalus (230). These abnormalities are thought to cause premature pulsatile secretion of gonadotropin-releasing hormone (GnRH) (229).

Screening lab tests for girls include a luteinizing hormone and estradiol or testosterone level. For boys, morning testosterone values in the pubertal range is diagnostic with an elevated luteinizing hormone level. For both boys and girls, if the luteinizing hormone level is not clearly elevated, this should be retested following stimulation with a GnRH agonist before treatment is begun (229). Bone age should also be tested and will likely be advanced.

Treatment with growth hormone leads to desensitization of the pituitary gonadotrophs, decreasing the release of luteinizing hormone. Treatment may be associated with menopausal symptoms such as hot flushes and may be associated with headaches. In a recent study in the spina bifida population, near adult stature, improved BMI, better reported self-esteem, and better gross motor skills were reported after treatment with GnRH (230).

OSTEOPOROSIS

Osteoporosis is the pathologic reduction of bone matrix and minerals, whereas osteopenia is a reduced density of bone. Osteoporosis has been identified as a medical problem in the adult with myelomeningocele (231). Although typically considered an adult disease, osteoporosis is a disease that starts in childhood (232,233). The age at which abnormalities in bone mineral density (BMD) first present in the spina bifida population is not known. It has been reported that children with myelomeningocele have a higher fracture risk and that those individuals who fracture have a lower bone density than age-matched peers (234). In patients with myelomeningocele, fractures typically occur in the long bones of the lower extremity, most commonly in the femur and less so in the tibia (235,236). Recent data suggest that fractures are present at all levels of spina bifida, with an annual incidence of about 3%. The age for the first fracture was around 11 years, with the tibia and femur most involved. Of those with fractures, one out of four reported multiple fractures (237).

Most contemporary studies of osteoporosis utilize dual energy x-ray absorptiometry to assess bone density. Non-weight-bearing conditions such as cerebral palsy (CP), Duchenne muscular dystrophy (DMD), and SCI have been shown to be associated with decreased BMD that can result in fractures, even in the pediatric population (238–240). It has been shown that BMD of the lumbar spine and proximal femur in children often correlates poorly, particularly if BMD is low (241). Studies currently have found utility in assessing the BMD in the lateral distal femur, as the lower extremities are a common site of fracturing (242). Recently published studies in patients with myelomeningocele attempt to describe the effect of non-weight-bearing on BMD; however, these studies have been limited by small sample size, inclusion of a limited number of pediatric patients with myelomeningocele, and older technology (233,234,243–245).

There may be technical difficulties in obtaining adequate lumbar spine and proximal femur assessments due to vertebral abnormalities and hip deformities (246).

TREATMENT

Toddlers (age 1–3 years) require about 500 mg of calcium each day. Preschool and younger school-age children (age 4–8 years) require about 800 mg of calcium each day. Older school-age children and teens (age 9–18 years) require about 1,300 mg of calcium each day. This guideline is set by AAP to meet the needs of 95% of healthy children (247,248).

There are limited natural dietary sources of vitamin D, and adequate sunshine exposure for the cutaneous synthesis of vitamin D is not easily determined for a given individual. In addition, sunshine exposure increases the risk of skin cancer, and decreased sun exposure is not uncommon for individuals with disabilities. The recommendations from the AAP have been revised to ensure adequate vitamin D status. It is now recommended that all infants and children, including adolescents, have a minimum daily intake of 400 IU of vitamin D beginning soon after birth. The current recommendation replaces the previous recommendation of a minimum daily intake of 200 IU/day of vitamin D supplementation beginning in the first 2 months after birth and continuing through adolescence. These revised guidelines for vitamin D intake for healthy infants, children, and adolescents are based on evidence from new clinical trials and the historical precedence of safely giving 400 IU of vitamin D per day in the pediatric and adolescent population (249).

In the setting of osteopenia or osteoporosis, individuals' vitamin D status and dietary history should be evaluated. Any deficiencies should be treated. Weight-bearing activities should be encouraged; however, there has been little to no data in regard to stander use in the spina bifida population. Standing weight-bearing exercises or

activities can apparently increase BMD in the lumbar spine or femur in children with CP (250,251).

Treatment for pathologic fractures supports the use of medication such as bisphosphonates. Prevention is the key, and careful attention to daily calcium and vitamin D intake, as well as a standing or walking program for those who are nonambulatory, is essential to minimize the reduction in bone density and the fracture risk (252).

More aggressive pharmacologic therapies have been used in other pediatric patient groups for the treatment of osteoporosis. The current treatment garnering most interest is bisphosphonates. Bisphosphonate use has not been studied in the spina bifida population, but there has been increased use in pediatrics (253,254). A recent study of those with Rett syndrome demonstrated an increase in bone mineral density when they consistently used a vibration platform at a specific frequency (255).

COGNITIVE FUNCTION

NEUROPSYCHOLOGY AND LEARNING PROBLEMS ASSOCIATED WITH SPINA BIFIDA

Spina bifida is a heterogeneous congenital disorder associated with complex neuropsychological sequelae. This birth defect has long been associated with specific characteristic personality traits marked by deficits in nonverbal learning abilities, visual-spatial perception, spatial reasoning, time concepts, processing speed, organization, and attention (256). Verbal skills, thought to be a strength due to the precocious development of conversational speech in young children with spina bifida, typically lack complexity, organization, and abstract content (257).

The development of hydrocephalus is a key component in the spectrum of cognitive impairment. A study by Lindquist compared children with hydrocephalus to those with hydrocephalus and spina bifida. Both groups had impaired learning, memory, and executive function, suggesting that hydrocephalus is a major factor in these deficits (258). When individuals with spina bifida were stratified to those with and without hydrocephalus, those without hydrocephalus had relatively normal neuropsychological testing scores, while those with hydrocephalus showed impairments, especially in executive functioning (259). The combination of spina bifida and hydrocephalus has also been implicated in deficits of working memory and processing speed, along with retrieval problems (260,261). The level of the spina bifida in association with hydrocephalus has been implicated in additional difficulties with learning. Higher level lesions above T12 with hydrocephalus showed more severe structural brain anomalies and a poorer cognitive outcome (262). The structural abnormalities were noted in the midbrain, tectum, pons, and splenium. Imaging technology has identified structural differences in the brains of individuals with spina bifida and hydrocephalus, implicating myelination impairments and abnormal white matter tracts along with a decrease in the gray matter and caudate nucleus structure (263). The clinical implications are emphasized in a study by Matson, which tracked individuals through multiple shunt revisions with standardized neuropsychological testing. The testing revealed lasting cognitive effects after hydrocephalus in verbal IQ, processing speed, organization, and response inhibition (264). The Chiari II malformation has been implicated and associated with verbal memory and fluency deficits (265).

When analyzing the testing in children with spina bifida, there is often a discrepancy between verbal and performance IQ scores. Verbal scores tend to be higher than nonverbal. This strength in verbal skills may be reflected in the classic "cocktail party syndrome." The cocktail party syndrome describes a speech pattern characterized by repeating phrases, using multisyllable words, and talking about various unrelated topics. This pattern can create the impression of a higher IQ of an individual with spina bifida, to the untrained observer.

Nonverbal learning disorder (NVLD) is commonly identified on formal testing. There are three areas of difficulty for those with an NVLD: motoric, visual-spatial organization, and social (266). Neuropsychology testing identifies problem areas to be impulsivity, difficulty with staying on task, memory, sequencing, organization, higher reasoning, mental flexibility, and visual-perceptual skills. A typical child with spina bifida may have early success in preschool because of verbal skills, but begin to have difficulties once the academic demands become more challenging. Mathematics is a particularly difficult subject area whereas reading tends to be a strength (267). In areas of self-care, they often are not at the level of their same-age peers. These children and adults have problems with developing and maintaining bowel and bladder programs. The difficulties are not only in the sequencing to accomplish the task, but also in realizing the social implications. Social skills acquisition can be affected by such simple things as understanding complex conversations, plots of books, and social jokes.

Selective memory disorders have been identified as the inability to sort out information and prioritize it. This is an important skill in the classroom and in learning new tasks as to not get lost in the details (268). Attention and executive function impairments are well documented in the adolescent spina bifida population (269). The attention deficits tend to be in attention span, shifting from one topic to another, and control as opposed to hyperactivity (270).

Studies of adolescents who have spina bifida have revealed difficulties specifically in areas of initiation, mental flexibility, and organization (271). Abnormal executive control functions may be one of the major reasons why children and adolescents with spina bifida fail to achieve the typical functional adaptive competencies of their same-age peers, despite adequate intellectual abilities.

These problems with attention and NVLD tend to persist into adulthood, and may impact the development of social competencies such as dating, living independently, and employability (272). In addition, this contributes to reduced skills acquisition for functional daily living (self-catheterization, independent care of personal hygiene, execution of household chores, making and keeping appointments). Weak executive control capacities may also underlie the mental health problems so often seen in adolescents and young adults with spina bifida. Despite the availability of psychotherapy, they often have difficulty putting talked-about goals and plans into action. Early intervention programs utilizing cognitive rehabilitation and goal management training may help to ameliorate the previously noted deficits.

In summary, most individuals with spina bifida will have some type of neuropsychological sequelae and this may be related to structural changes within the brain along with hydrocephalus. Those who have hydrocephalus and multiple shunt revisions may have more impairments than those who do not. We know that deficits can be in multiple domains and include visual-spatial, perceptual motor, organization, executive function, sequencing, memory, attention, and learning challenges. Early identification and intervention programs are important in this population and suggestions are discussed in the following:

- Identification of the learning disorder is critical. Testing should be done early in the educational process.
- Modified program to address the specific needs of children and adolescents with spina bifida. Some children will need accommodations under section 504 of the Federal Rehabilitation Act of 1973 or Individualized Education Programs (IEPs).
- Provide structure and direction for education. Be specific and repetitive.
- Teach stepwise and subject matter sequentially (baby steps).
- Make sure to teach social education, as these children may not pick up nonverbal cues.
- Use multiple sources available on NVLD for guide in education, self-skills training, and social integration.

LONG TERM

AGING WITH A NEURAL TUBE DEFECT

Adults with spina bifida have the normal aging medical problems in addition to those associated with their disability. Age associated changes can have an impact on medical and functional systems. In treating the adult patient, one has to evaluate the usual age-related medical problems as well as those unique to this population. Medical complications and cardiovascular disease may present at an earlier age. Successful transition to adult-based clinics appears to be based on a few key factors and include preparation, flexible timing, care coordination, transition clinic visits, and interested adult-centered health care providers (273). Adults with chronic conditions generally require more medical visits yearly and have an admission rate nine times more than the nondisabled. Adults with spina bifida in general are satisfied with life, but the area of largest concern is in self-care ability and partner relationships (274,275).

Spina bifida is associated with abnormalities in the brain and spinal cord. Approximately 90% of adults will have ventricular-peritoneal shunts. Shunt malfunctions can occur at any age and present with the classic symptoms of chronic headaches, vomiting, personality changes, concentration difficulty, and other neurologic changes. Shunt malfunction can lead to significant morbidity, mortality, and sudden death (276,277). Treatment is geared toward reducing pressure within the ventricular system either by shunting or ventriculostomy. Adult-onset tethered cord should be considered in a deterioration of neurologic status, bowel or bladder changes, increasing orthopedic deformities, and gait deviations.

There are several age-related musculoskeletal and orthopedic complications. Spinal deformities, including scoliosis, kyphosis, and lordosis, can increase over time and cause back pain. Chronic lack of sensation and muscle imbalances can lead to Charcot joints. Overuse syndromes are common in wheelchair and crutch users. Wheelchair mobility tends to cause stress on the upper extremities, while community ambulators develop knee and hip pain. Carpal tunnel and rotator cuff disease are well documented in wheelchair users (278). Gait abnormalities from underlying muscle weakness can cause undue stress on joints in the lower extremities.

Neurogenic bowel and bladder function is an important component of adult medical care. It is a rare individual with spina bifida who has a completely normal urinary system. Despite these abnormalities, more than 80% of adults are able to develop social bladder continence (68). Methods to achieve this goal include all those previously discussed. In the past, renal damage leading to renal failure and death was a major contributor of morbidity and mortality in adults with spina bifida; although this is much improved, it still remains a problem (279,280). There is also an association between the presence of a neurogenic bladder and the development of bladder cancer (281). Recent data suggest that this occurs at a young age in the population that develops bladder cancer, with variable pathology and has a poor prognosis (282). Neurogenic bowel function can change over time. Gastric motility seems to decrease with age and affects bowel programs. Treatment needs to be adjusted for these changes and includes different medication, dietary modifications, and newer surgical interventions.

The development of chronic skin problems is inherent in those with insensate skin. Aging causes changes in fat and muscle distribution, which can affect pressure

ulcer formation. In the lower extremities, bracing can cause pressure and shear over bony prominences. Burns and abrasions can occur in unprotected skin. The wheelchair seated position results in pressure in the ischial and sacral areas. Prevention is imperative to avoid these secondary complications. The economic burden, along with psychological and functional impact, can be devastating (283,284).

Adults with latex allergies may have a higher rate than children for reactions, including anaphylaxis (285,286). This is probably related to repeated exposure to latex over the years, along with the increasing presence of latex in the environment.

Obesity is a health-related problem for both able-bodied and disabled adults. Nutritional studies indicate a decreased caloric expenditure with the disabled adult. Metabolic syndrome is more common in those with obesity and places these individuals at risk for coronary artery disease, diabetes, and hypertension. Interventions include nutritional counseling and healthy eating, exercise and fitness, and weight reduction. Interestingly, most researchers focus on obesity, although eating disorders also occur in the disabled population (284).

Sexuality and sexual function are often overlooked in the disabled population. It is a huge disservice not to address these issues. Current data shows that the majority of males and females with spina bifida have a desire for intimate relationships, including sexual contact (287). Recent data suggest 24% of adults have an active sex life, and gender and continence did not factor into this statistic (288). Men with spina bifida report ability to achieve erections in 72%, and 67% experience ejaculation, but only one-third are happy with the amount of rigidity (289,290). Sildenafil (Viagra) may improve erectile function in 80% of men (291). Fertility is impaired, as only 14% of men report fathering children, and neurologic level is an important factor (290). Women with spina bifida generally have normal menstruation, and 88% have adequate vaginal secretions during intercourse (289). Women with spina bifida are able to conceive and have children (284). Sexual counseling should inform individuals about the risk for pregnancy, sexually transmitted diseases, and contraception advice, including nonlatex condoms. Risk and benefits of Gardasil human papillomavirus vaccine should also be provided prior to sexual contact.

VOCATIONAL COUNSELING

Vocational counseling is an important aspect of transitional care of the individual with spina bifida, and current information in this area is limited. Recent data from the Netherlands reports a work rate of 62%, although 22% were in a sheltered environment. The definition of employment was based on at least 1 hour of paid wages per week. The best predictor of employment was level of education. This, along with gender and ability to care for self, was an important predicator of full-time employment (292).

Functional vocational planning should be started early in secondary school, assessing career interests, skills, and aptitude. The potential for success in a postsecondary school program should be explored along with vocational job training. A positive realistic approach may provide the best solution in planning for adult employment options.

RESOURCES FOR FAMILIES

Resources for families with spina bifida vary from state to state, often requiring some degree of research within the local community. On a national level, the SBA can be contacted at 4590 MacArthur Boulevard NW, suite 250, Washington, D.C. 20007-4226, by phone at 202-944-3285, or via the Internet at www.spinabifidaassociation.org. The SBA offers a plethora of information and resources including a national directory of local state SBA chapters where information on local resources, events, and contacts can be found.

The Health and Human Services (HHS) division is mandated by Congress to use appropriated funds to support University Centers of Excellence in Developmental Disabilities (UCEDDs). There are currently 67 UCEDDs in the United States, each state having at least one UCEDD. The state UCEDD can often serve as a resource for information not only for professionals but for families as well. A directory of UCEDDs can be found at www.aucd.org. Likewise, each state has its own Disability Rights Protection and Advocacy Center. These centers provide information and resources relative to the adherence of the Americans with Disabilities Act (ADA). A directory of states within this network can be found at www.ndrn.org. The National Association of Councils on Developmental Disabilities (NACDD; www.nacdd.org) also has a directory of state councils. These councils are family-centered and consumer and family driven in offering supports and assistance to enable individuals with disabilities to strengthen self-advocacy skills, independence skills, and to be integrated and included in their communities.

As the child with spina bifida matures and approaches transition age (typically 12–14 years), vocational rehabilitation can serve as a resource for educational training and vocational support. Due to the costs involved with raising a child with disabilities, families may also find support in Social Security and/or Supplemental Nutrition Assistance Programs. For information on either program, contact or visit the local branch within the community.

Individuals and families with spina bifida can also find support and resources at local nonprofits such as the Centers for Independent Living (CILs). These are

consumer-controlled community-based organizations that provide services. There are over 500 CILs in the United States. The directory of state CILs can be found at www.ilr.org. Aging and Disability Services can also be a resource, offering case worker coordination and assistance with social service applications. Contact your state office for details.

Recreational resources can be found in Special Olympics programs, Kiwanis Clubs, Shriners Hospital, and the local Parks and Recreation program in your community. Look for adaptive or inclusive programs that suit the child's interests.

Housing resources can be researched through the local Housing Authority branch office. They may be able to guide families to accessible homes or provide support to modify existing homes to meet the access needs of the family. Many nonprofit organizations donate services for home adaptations as needed.

One of the most effective methods for locating community resources is networking with other families with similar needs. With parental and family engagement in the process of researching resources, the art of advocacy can be keenly observed by the individual with disabilities and the seeds of self-advocacy are born.

CONCLUSION

The successful treatment of spina bifida requires a multidisciplinary team approach. Education of the child and family regarding lifelong expectations are a critical part of multidisciplinary management. Knowledge of all the different systems involved—including genetic propensity to latex and fruit allergy, neurologic, urologic, GI, orthopedic, endocrinological, skin, psychosocial, and rehabilitation issues—are essential for comprehensive care. Daily range of motion programs to avoid joint contractures, daily bowel and bladder programs to maintain bowel and bladder health and continence, and independent mobility promotes emotional and social well-being and aids toward educational and vocational advancement.

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16

SPINAL CORD INJURIES

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Children and adolescents with spinal cord injury (SCI) must deal with the multisystem involvement imposed by the injury that is compounded by physical and psychological growth and development, which cause complications not seen in the adult. Rehabilitation is a process that extends at least until the child is physically and psychosocially an adult. Involvement by a team that is expert in the management of children and adolescents with SCIs should continue throughout this period. This chapter discusses some of the main points to be considered by those who are involved in assisting this rehabilitation process. Advances have been made in the acute management of pediatric SCI, and there is new equipment and technology for rehabilitation, but the basics of rehabilitation in this area remain the same.

EPIDEMIOLOGY

INCIDENCE AND PREVALENCE

Compared to other disorders discussed in this text, SCI is a relatively rare disorder in the general population. The most comprehensive data on the epidemiology of SCI comes from the National SCI Database (NSCID), which is generated by the Model SCI Care Systems. The Model SCI Care Systems is a network of 28 past and current centers funded by the National Institute on Disability and Rehabilitation Research, an institute in the U.S. Department of Education. Since SCI is not a reportable condition, data collection is limited. The NSCID estimates that it collects data on 13% of the new SCI cases per year. From this data, the incidence of adult traumatic SCI is estimated to be approximately 40 cases per million in the United States, or roughly 12,000 new cases per year. This data does not include injuries that resulted in death prior to hospitalization (1). Selvarajah and colleagues used data taken from the Nationwide Emergency Department Sample from 2007 to 2009 and estimated the incidence of traumatic adult SCI to be higher at 56.4 cases per million (2). Using the same sample data set from 2007 to 2010, it was estimated the incidence of childhood and adolescent SCI presenting to emergency departments to be 17.5 cases per million, which represents 1,308 new cases per year (3). Acute SCI primarily occurs in young adults, though the average age has been rising. In the 1970s, the average age of injury was 28.7 years; since 2010, the average has increased to 42.6 years. The cause in this shift is unknown, though the incidence in SCI over the age of 60 has more than doubled since the 1980s. Other factors, such as prevention programs, advances in automobile safety, or referral patterns to Model Systems, may also be affecting the data. It was previously reported that 3% to 5% of all SCI occurred in children under age 15 and 20% of injuries occurred in those under 20. In their February 2013 report, the NSCID estimated that there were between 238,000 and 332,000 people living in the United States with SCI.

DEMOGRAPHICS

Publications have combined data from the Shriners Hospitals for Children and the NSCID. As in adults, males are four times more likely to have SCI than females overall, with the ratio being 1.5:1 in children under age 9 years. In children under 3 years, females have outnumbered males in some studies. The cumulative incidence of SCI in child and adolescent males was estimated to be between 2.4 to 2.8 times greater than females from the period of 2007 to 2010 (3). In younger children, there are no statistically significant racial trends. In those over age 15, there is an increased risk in African American and Hispanic American populations. These figures are all from specialized hospital data and may not represent those with milder injuries (eg, incomplete lesions and paraplegia) who are treated in smaller hospitals or in adult settings. Since the year 2010, the ethnic make-up for SCI treated in the Model Systems has been 67% White, 24.4% Black, 7.9% Hispanic, 2.1% Asian, and 0.8% Native American (1).

CAUSE OF INJURY

Trauma is the primary cause of SCI, accounting for at least 93% of all SCI. Since 2010, motor vehicle crashes (MVCs) account for 36.5% of SCI, falls 28.5%, violence 13.3%, and sports injuries 9.2%. The remaining 11.4% are other and unknown causes (1). In those under age 20, violence and sports injuries are more common than falls. The sports that are most commonly associated with SCI are American football, ice hockey, wrestling, diving, skiing, snowboarding, rugby, and cheerleading (4).

Hadley and colleagues (5) reviewed 122 cases of spine injury in children 16 years and younger. The median age was 15 years in males and 14 years in females. SCIs were due to MVC in 39% overall, with MVC the cause in 17% of children under 10 years of age, 26% of those 10 to 14 years, and 52% in those 15 to 16 years. Pedestrian versus MVC were 11% overall and 33%, 16%, and 3%, respectively, for the three age groups. Falls were the second leading cause under 10 years, with sports the second leading cause at ages 15 to 16 years. Fifty percent of those under the age of 10 had an occiput-C1 injury, with all levels of cervical injuries occurring in 72%, 60%, and 55% in the three age groups. Fifty percent of the subjects were neurologically intact, with bony or ligamentous injury only.

More recently in 2007, Bilston and Brown (6) reported similar data from Australia, looking at children 16 years and younger. MVC accounted for 30% of all spine injuries and 50% of serious injuries. Sports were the next most common cause of all spine injuries, though falls resulted in a higher (20% vs 16%) risk of serious injury. Gender plays a significant role in the cause of injury. Violence and sports-related injuries are more common in males, while MVC injuries are less gender-specific. The authors again demonstrate that children are at higher risk for cervical spine injury, especially under the age of 8, with higher injuries occurring in younger children.

Data for children and adolescents collected from a representative sample of U.S. Emergency Departments from 2007 to 2010 was reported by Selvarajah and colleagues (3). Their analysis of traumatic SCI showed a preponderance of males (72.5%) and a median age of 15 years. Those who required direct hospital admission were 62.4%, and 14.8% were transferred to another hospital for additional medical care. Traffic accidents were reported as the most common etiology of injury (37.0%) followed by falls (20.4%), struck by others/objects (16.3%), other (10.7%), firearms (8.7%), and sports (6.9%). Cervical SCI was the most common level and represented 40.5% of all injuries (19.8% high cervical and 20.7% low) and complete SCI represented less than 10% of all injuries. 14.3% of new cases of traumatic SCI in children and adolescents had a concurrent traumatic brain injury (TBI). Spinal cord injury without radiographic abnormality (SCIWORA) represented greater than 50% of all injuries. Children less than 5 years of age were more likely to be involved in a traffic accident, and have a cervical SCI that was more severe and a concurrent TBI.

The incidence of SCI in sports-related injuries since 2010 is 9.2%. SCIs in American football have decreased markedly since the mid-1970s, when "spearing" was made illegal. This now-banned tackling technique resulted in a high degree of axial cervical loading. Since institution of the ban, SCI in football has decreased by 80% (7). Since the 1980s, the incidence of spine injuries and SCI has been increasing in ice hockey. These injuries are most often the result of a player being checked from behind, with his or her head down, into the boards, again resulting in high axial compression loads (8). Over the past several years, cheerleading has evolved into a competitive sport. This often involves gymnastic moves, tosses, jumps, and pyramid formations. While the incidence of SCI is low, this is a risk category where females are more likely to be injured (9).

CLASSIFICATION OF SPINAL CORD INJURY

LEVEL OF INJURY—ASIA IMPAIRMENT SCALE

The most common method of classifying impairment from SCI is the American Spinal Injury Association (ASIA) impairment scale. The classification is based upon assessment of strength and sensation to light touch and pinprick in defined myotomes and dermatomes. Key muscle groups and sensory points are shown in Figure 16.1. The ASIA impairment scale has been modified over the years, originally based on the classification system defined by Frankel. For a child, completing the ASIA examination requires a certain level of maturity in being able to follow motor commands and respond appropriately to sensory stimulation. The examiner must take this into account when assessing children. Other factors that may limit examination (eg, long bone fractures or decreased level of consciousness) need to be taken into account as well. The motor examination scores strength on a six-point scale: 0 to 5. For each strength grade, the joint being assessed must be moved through a full available range of motion. A strength grade of 0 is given for total paralysis. A 1 is given for a visible or palpable contraction that cannot move the joint through its available range of motion with gravity removed. A grade 2 is given if the muscle group can move the joint through its range of motion with gravity removed. The joint is positioned parallel to the ground to limit the effect of gravity. A grade 3 is given when the patient can move the joint through a full available range of motion against gravity but cannot bear any additional resistance. If a patient can bear additional resistance, he or she is given a grade 4, and a grade 5 is given for normal strength. Motor scores are documented on the ASIA form and summed for a total motor score. A rectal exam must be performed to assess for voluntary contraction, and is scored as yes/no. As individual muscles are almost always

innervated by multiple spinal cord levels, a strength grade of 3 is considered normal for a muscle group if the level above has grade 5 strength. This implies that the grade 3 muscle group is only partially innervated and the more proximal innervation level is intact.

Sensory examination is performed using pinprick and light touch at key points, and it is graded as 0 for absent, 1 for impaired, and 2 for normal. These results are summed as well for total light touch and pinprick scores. Again, a rectal exam is necessary to assess anal sensation, also scored yes/no.

The ASIA neurologic level is the most caudal segment with intact motor and sensory exam. In addition to the level is whether the injury is complete or incomplete. With a complete injury, there is no motor or sensory function in the lowest sacral segment (ie, no anal sensation or voluntary anal contraction). A complete injury is classified as ASIA-A. Incomplete injuries are classified as B to E, as listed in Figure 16.1. While an "E" is described as normal sensory and motor function, this is in the context of a previously abnormal examination.

Paraplegia affects the lower extremities and, to varying degrees, the trunk. It does not affect the upper extremities; thus, T2 must be normal and any deficits are below that sensory and motor level. The preferred term from ASIA for involvement of all four extremities is tetraplegia, though quadriplegia is much more commonly used. Any injury that affects motor and/or sensation at or above the T2 level is tetraplegia. In addition, a number of syndromes have been described based upon the patterns seen after specific areas of the spinal cord have been injured.

CENTRAL CORD SYNDROME

The central cord syndrome was first described in 1954 (10). As its name implies, this is damage to the central area of the spinal cord. This most commonly happens in

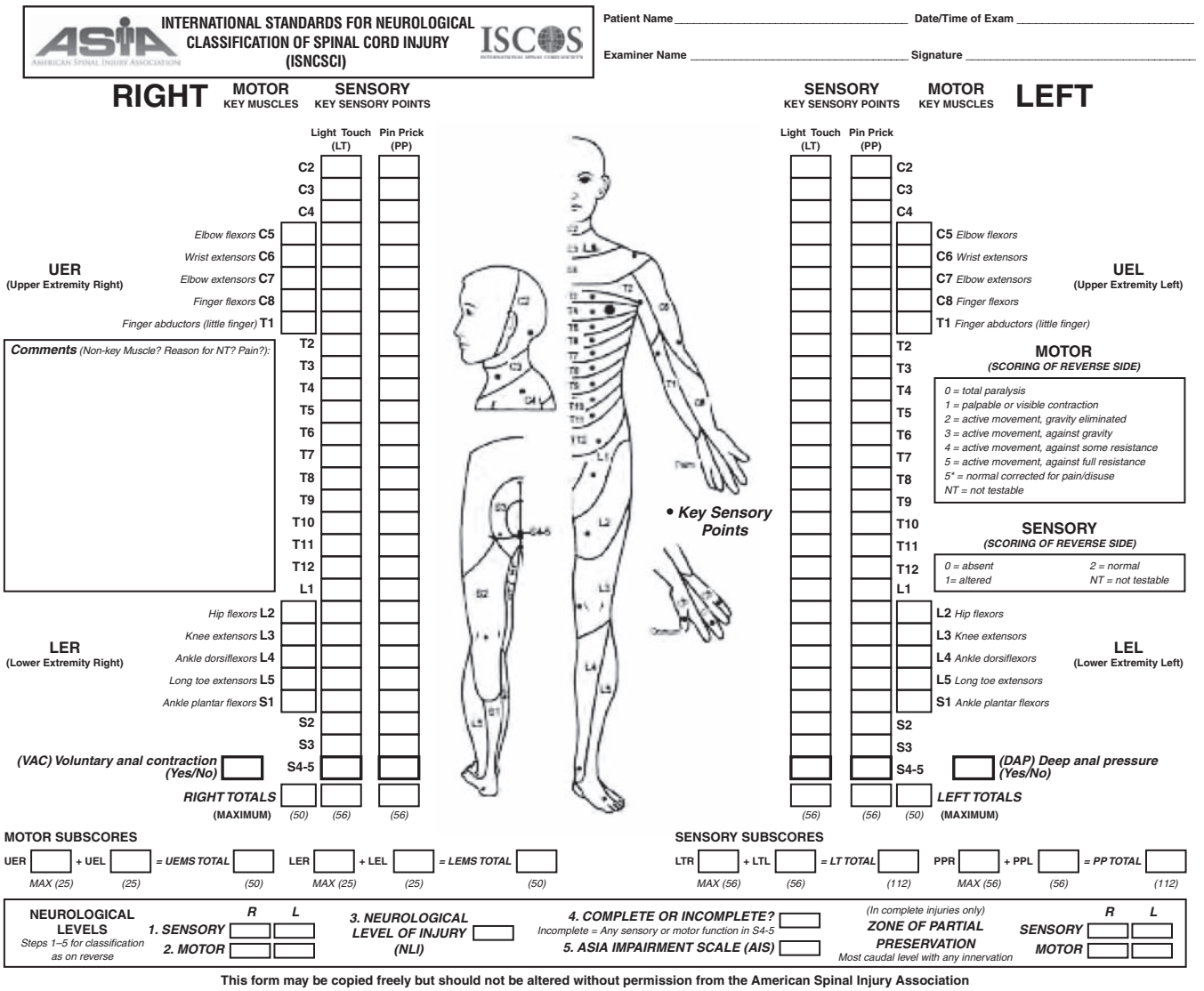

FIGURE 16.1 American Spinal Injury Association Exam Sheet.

the cervical region at the lower cervical levels because of the high amount of normal motion at these segments and is often due to a hyperextension injury. Disruption of decussating spinothalamic fibers at the site of the lesion results in impaired pain and temperature sensation at those dermatomes and may also be associated with the development of neuropathic pain. Dermatomes above and below the lesion may have normal sensation. As a lesion enlarges, damage may extend into the anterior horn cells and medial corticospinal tracts, causing weakness. Reflexes may be lost at the level of the lesion as well, with possible hyperreflexia at lower levels. As this is primarily a cervical syndrome, there are typically motor and sensory changes in the arms, with relative sparing of the legs, bowel, and bladder function.

BROWN-SEQUARD SYNDROME

Brown-Sequard syndrome results from a hemisection of the spinal cord. This is most commonly seen with low-speed penetration wounds, such as a stabbing. Corticospinal tracts and the dorsal columns cross in the brainstem, so their damage in this type of lesion leads to ipsilateral weakness and loss of vibration and position sense. The lateral spinothalamic tracts cross soon after entering the spinal cord, thus causing contralateral loss of pain and temperature sensation.

ANTERIOR CORD SYNDROME

The anterior (or ventral) cord syndrome is most commonly related to a vascular insult, causing infarction of the ventral spinal cord or a hyperflexion injury to the spinal cord. This injury includes damage to the corticospinal, spinothalamic, and descending autonomic tracts to the bladder. This syndrome results in paralysis, loss of pain and temperature sensation, and urinary incontinence. Vibratory and position sense, whose tracts are in the dorsal columns, are most often spared.

CAUDA EQUINA AND CONUS MEDULLARIS SYNDROME

Compressive injuries in the lower lumbar and sacral vertebral levels may result in damage to the cauda equina, as the spinal cord proper has terminated at a higher level. This results in scattered symptoms, depending upon which nerve roots are damaged. The cauda equina syndrome results in damage to the axon of lower motor neurons, leading to a flaccid paralysis. Conus medullaris syndrome shares many of the same features of cauda equina compression and represents damage to the bulbous, caudal portion of the spinal cord, which terminates near the L1 level. A large, nonselective lesion at this level may damage the most distal portions of the spinothalamic, corticospinal, and autonomic tracts as well as the descending nerve roots resulting in both upper and lower motor neuron findings.

SPINAL CORD CONCUSSION

Spinal cord concussions are an uncommon injury. These are transient injuries with full recovery. These are defined by four criteria:

1. Spinal injury with immediate neurological deficit of varying degree
2. Neurologic deficit corresponding to the level of spinal injury
3. Complete neurologic recovery within 72 hours (ASIA-E)
4. No evidence of injury on imaging

These injuries are most commonly at the cervical levels. While spinal cord concussion has been reported across the age span, it is more likely to occur in children. Long-term prognosis seems excellent, though data is very limited. The mechanism of the concussion is unknown (11).

SPINAL CORD INJURY WITHOUT OBVIOUS RADIOGRAPHIC ABNORMALITY

SCIWORA in children has been a known entity since at least the early 20th century (12), though the acronym did not come about until 1982 in an article by Pang and Wilberger (13). The initial definition focused on children with traumatic SCI, who did not have evidence of vertebral column injury on spine x-rays, conventional and computed tomographic studies, myelograms, or dynamic flexion/extension studies. This excluded injuries caused by penetrating trauma, electrical shock, obstetric complications, and congenital spinal anomalies. In 2004, Pang published a review on what is now known about SCIWORA (14). Incidence of SCIWORA ranges between 5% and 67% of cases of pediatric SCI, with an average of 34.8%. The incidence is much higher in children 9 years and younger. Pooled data indicated an incidence of SCIWORA of 63.1% in younger children and only 19.7% in older children. Most injuries occur in the cervical cord, most commonly with C5 to C8 lesions. Thirteen percent of injuries were in the thoracic cord. This injury is thought to be primarily present in children due to the unique physiology of the developing spine, being much more mobile, without resulting in bony fractures, but causing stretch injury to other tissues. This increased mobility was thought to result in damage to the soft tissue structure of the spine, including ligamentous

and neural structures, which cannot be demonstrated on radiographic studies. The advent of more advanced imaging techniques has demonstrated these soft tissue injuries. It should be noted that SCIWORA occurs in adults as well (15).

SCIWORA has been reported to cause complete and incomplete SCI, as well as central cord and Brown-Sequard syndromes. Pang classified ASIA-B/C as severe injuries and ASIA-D as mild. From this pooled data, SCIWORA results in ASIA-A 22.1%, ASIA-B/C 12.6%, ASIA-D 23.2%, central cord syndrome 29.4%, and Brown-Sequard 12.7% (14).

With the development of MRI, damage to the soft tissue structures in the spinal column is readily apparent. This information was recently reviewed by Yucesoy and Yuksel (16). These authors suggest that in the MRI era, SCIWORA may be an ambiguous term and that those with no lesions on neuroimaging be classified as "real SCIWORA" or spinal cord injury without neuroimaging abnormality (SCIWNA). In the strict sense, radiographs do not include MRI, yet in standard use, most practitioners would consider most imaging to fall into the category of radiograph, regardless of the nature of the physics involved in the imaging process.

PROGNOSIS FOR NEUROLOGIC RECOVERY

One of the most challenging aspects of rehabilitation medicine is talking with patients and families regarding prognosis for recovery. We must be able to present the best available information regarding prognosis (which is often not good) in a manner that people with varied levels of education and sophistication can understand and that can offer a reasonable and realistic degree of hope. SCI is truly devastating, and as rehabilitation specialists, we must take the impact of that into account when communicating with patients and families. We must also be aware that during these times of stress, communication may not always be effective. We must also be clear that neurologic recovery can be markedly different from functional recovery.

A complete SCI examination is necessary for any discussion of prognosis. Examination at least 72 hours after injury has been determined to be a better prognostic indicator than earlier examinations (17–19). (This does not mean earlier examinations are not necessary, only that they are of less prognostic value, as they may be limited for a variety of reasons.)

Most recovery from SCI occurs during the first 6 months, with a plateau reached around 9 months postinjury, though later recovery can occur. Neurologic recovery after a complete injury is poor. Ninety-six percent of those with complete paraplegia and 90% with complete tetraplegia at 1 month will remain ASIA-A. Muscle groups with a grade of 0 at initial examination, more than two neurologic levels below the level of injury, are unlikely to regain functional strength. Seventy percent of individuals with tetraplegia and a complete SCI can expect the myotome below the neurologic level to regain at least antigravity strength (20). Muscles with grades 1 to 2 have a 64% chance of increasing to functional strength in paraplegia and 97% in tetraplegia. Incomplete paraplegia has an average motor score increase of 12 points at 1 year post-injury. Seventy-six percent of those with incomplete paraplegia became community ambulators. For incomplete tetraplegia, 46% became community ambulators at 1 year (21).

PREVENTION

Prevention of injury is always more effective than treatment, and this is especially true in SCI. The hallmark of prevention is safety education beginning in early childhood. Use of safe equipment is the second tenet of prevention, and nowhere has this been more effective than in the use of infant and child auto restraints and adult lap and shoulder belts. This practice has also caused lap belt injuries, however, including SCIs, which are more common in children than in adults. Other prevention relating to motor vehicles is substance abuse education and laws relating to driving while impaired. Pedestrian safety is promoted almost exclusively through parent and child education.

Prevention of sports-related SCIs has improved because of education, rule changes noted previously (such as no spearing in football, no checking from behind in ice hockey), better coaching, and better conditioning of players.

The ThinkFirst National Injury Prevention Foundation promotes safety education. "ThinkFirst programs educate young people about their personal vulnerability and the importance of making safe choices. The message is: You can have a fun-filled, exciting life, without hurting yourself if you 'ThinkFirst'" (www.thinkfirst.org). There are separate programs for teens and children, which promote injury prevention through talks and publications.

EARLY TREATMENT

SPINAL STABILIZATION

Once it has been determined that the child has an SCI, the spine must be stabilized. The halo external skeletal fixation device was first described in 1968 for use in adults with cervical fractures by Nickel and colleagues (22). It has subsequently been adapted for use in children, with modifications required by the unique characteristics of the child's skull, which is thinner. Fixation pins must be carefully placed, with attention paid to both location and depth of insertion. For thoracolumbar and lumbar

fractures, nonsurgical management with a thoracic lumbar spine orthosis (TLSO) may be used either in place of or in addition to surgical stabilization (23).

USE OF STEROIDS

Various studies of the efficacy of the uses of methylprednisolone in acute SCI were conducted during the 1980s. The National Acute Spinal Cord Injury Study 2 (NASCIS 2) was published in 1990 (24), with the conclusion that patients with acute SCI treated with high-dose methylprednisolone in the first 8 hours after injury had better neurologic outcome than did those treated with placebo or naloxone. However, this was an adult study, with only 15% of patients being under 19 years of age and the youngest being 13 years old. Data are lacking in the pediatric population.

A 2012 Cochrane Collaborative Review noted only eight steroid trials for acute SCI, seven using methylprednisolone. The review concludes that methylprednisolone provides some benefit (and unlikely to change prognosis for ambulation), if given within 8 hours of injury, with an initial bolus of 30 mg/kg by intravenous (IV) for 15 minutes followed 45 minutes later by a continuous infusion of 5.4 mg/kg/hour for 24 hours (25).

RESPIRATORY FUNCTION

Most children with SCIs have impairment of normal respiratory function because of their injuries, even in the absence of other trauma causing pulmonary problems. The basic muscles of respiration are the diaphragm, intercostal muscles, abdominal muscles, and neck accessory muscles. Any SCI that weakens one or more of these muscles impairs respiration. The child with weak or absent diaphragm function needs ventilatory support. If the diaphragm is functional but intercostal and abdominal muscles are weak or nonfunctional, the child will need assistance with coughing and respiratory secretion management and may need ventilator support during respiratory illnesses or during sleep. If the child only has weakness of the abdominal muscles, assistance with coughing may be the only respiratory support needed.

All children with acute SCI should have respiratory function evaluated. At the very least, this evaluation should include chest radiographs and measurement of oxygen saturation and end-tidal carbon dioxide or arterial blood gases. If the child is able, forced vital capacity and inspiratory and expiratory forces should be measured on a daily basis until the child is medically stable. Because the child with SCI has restrictive respiratory dysfunction (so-called bellows failure), not lung disease, the earliest pulmonary abnormality will be hypercarbia and not hypoxia. End-tidal carbon dioxide measurement is a simple noninvasive way to follow hypercarbia over time, and may be useful for outpatients as well as for inpatients.

URINARY FUNCTION

Most children with acute SCIs develop a neurogenic bladder. During spinal shock the bladder is initially flaccid, and may subsequently become spastic or dyssynergic. Flaccid bladders need to be drained either continuously or intermittently. Because indwelling catheters are associated with infections, the child should be converted to a clean intermittent catheterization program as soon as there is no medical reason to have continuous monitoring of urine output. Spastic or dyssynergic bladder management is discussed in further detail later in this chapter.

GASTROINTESTINAL AND BOWEL FUNCTION

After acute SCI, the gastrointestinal tract usually stops functioning initially, thus requiring the use of nasogastric decompression. Once the ileus has resolved and the child is taking enteral feeding, a bowel program should be instituted, with the ultimate goal of continence without impaction. The consistency of the stool is normalized through the use of fluids, fiber, and enteral medications, as needed. Evacuation is assisted through the use of digital stimulation, enemas, and/or rectal medications.

FLUIDS AND NUTRITION

Careful attention must be paid to fluid balance and nutrition in the child with an acute SCI. A balance must be maintained between adequate hydration for intravascular volume/blood pressure, soft stool, and over hydration which may cause bladder distension, leakage and need for increased catherization.

To promote healing, the child must also receive adequate nutrition. A common standard is to start some form of nutrition within 24 hours of injury. Typically, this is parenteral nutrition initially, followed by either oral or tube feedings when the ileus is resolved. For some children who have the ability to eat orally, refusal to eat may be the only way they have of controlling their medical treatment, so nutritional intake should be closely monitored during the acute and rehabilitation hospitalizations.

REHABILITATION

Rehabilitation of the child with SCI is a lifelong process that starts soon after injury. It does not start and end with admission to and discharge from a rehabilitation unit, and

this must be made clear to the patient and family. Goals of rehabilitation will be dependent upon a number of factors, primarily the patient's age, level and completeness of injury, and amount of neurologic recovery. Rehabilitation of the child with SCI is comparable to rehabilitation of the child with any other acute change in function, usually with less need for cognitive rehabilitation. The entire rehabilitation process should focus on the whole child in the context of his or her family and community, and be performed by a rehabilitation team of professionals that focuses on the needs of children. It should be noted that on occasion, some older children may be more appropriate for an adult rehabilitation service, while some young adults or people with cognitive impairment may be better served on a pediatric rehabilitation service. When older children are treated on an adult service, it is important that the appropriate pediatric, social, and education services be available.

Goals for rehabilitation should include maintenance or attainment of good health and prevention of secondary complications, while promoting maximal and age-appropriate functional independence. The focus of rehabilitation can range from primarily family education (eg, C4 or higher complete tetraplegia with ventilator dependence) to primarily complete patient functional independence (eg, T10 complete paraplegia). The goals of rehabilitation will change as the child matures. Table 16.1 lists expected functional goals for levels of SCI.

MOBILITY

Mobility for the child with SCI begins with the process of learning to sit again. Sitting is compromised as a result of the lack of neurologic control of the trunk related to the SCI, impairments in the autonomic nervous system, and physiologic adaptations to deconditioning during the acute hospitalization. Hypotension and syncope can result. Support hose, wrapping of the lower extremities with elastic bandages, and abdominal binders may help maintain blood pressure. Progression to sitting is a gradual process and should be started as soon as possible to minimize deconditioning. Short periods of sitting as tolerated can be done multiple times during the day, gradually increasing the duration of the time up. Early use of a power or manual wheelchair (as appropriate) is encouraged. It is important to monitor insensate skin as the duration of time up increases to minimize the development of pressure ulcers. The use of pressure-relieving cushions or periodic pressure-relieving maneuvers is critical. Patients will begin working on bed mobility, rolling in bed, and transferring from the bed to the wheelchair. As the patient improves, more advanced transfers will be the main focus of physical therapy interventions.

Standing and walking, either with orthoses or independently, will be done as appropriate. Table 16.2 shows mobility guidelines from recommendations by the American Academy of Orthopaedic Surgeons and Shriners Hospitals for Children. It should be noted that often younger children with high lumbar or thoracic paraplegia may be ambulators with appropriate bracing. As with paraplegia from myelodysplasia, it is not unusual for these children to become more wheelchair-dependent as they reach adolescence. Ambulation at these levels is quite energy-inefficient, while the use of a wheelchair is very efficient. This later use of a wheelchair should be discussed with the patient and family well before it occurs so that it is seen as the expected path and not as a failure of the patient or family.

SELF-CARE AND ACTIVITIES OF DAILY LIVING

By the time neurologically intact children are 5 years old, they are independent in the majority of self-care activities with supervision. Regaining this independence after it has been lost because of SCI is of the utmost importance, especially to adolescents and preadolescents. The first step in this process is allowing some control over the environment in the rehabilitation unit. This may be as simple as a remote control for the television and an accessible call system to alert the nursing staff. A variety of systems are available, including "sip and puff" systems, head switches, mouth switches, and large buttons. Rehabilitation engineering, if available, can be vital in helping to provide options for accessible environmental controls. It is important that the family try to promote the child's independence as well, and they must be encouraged to give the patient a certain degree of freedom and independence despite increased effort, difficulty, or frustration with tasks.

Relearning self-care should follow an orderly pattern, but may begin with the activity in which the child is most interested, often self-feeding. Activities that must be relearned include dressing, bathing, hygiene, feeding, transfers, writing, computer skills, and leisure activities. For young children, teaching these activities may need to be incorporated into games and play activities. Children with high tetraplegia may not be expected to manage their own self-care needs, but should be taught how to direct caregivers to perform various activities.

COGNITION

It is important to assess cognition during the rehabilitation of the child with SCI. Just as MVCs are the primary cause of SCI, they are also the primary cause of TBI. Any force significant enough to cause an SCI can also cause a TBI. Any child who has had an SCI should also be at least screened for a TBI. These screenings may also be useful in assessing possible hypoxic injury in ventilator-dependent children (26).

TABLE 16.1 MOBILITY GUIDELINES

LEVEL OF INJURY	AGE	GOALS	ORTHOTIC OPTIONS
C1–4	Bracing available from age 1 year–prepuberty No standing after puberty	Standing	Prone and supine standers (stationary standers)
C4–7	Encourage from ages 1–5 years; available from age 5 years–prepuberty	Static standing and mobility	As above plus parapodiums/swivel walkers/mobile standers
T1–5	Encourage from ages 1–10 years after rehabilitation goals are met, increase upper extremity strength/endurance); if surgery is performed, intensive gait training available postoperatively Ages 11–21 years need to meet criteria: 6 parallel bar pushups; 25 wheelchair pushups; transfer level heights; <20 degrees of hip flexion contracture; <15 degrees of ankle plantarflexion contracture	Standing and household ambulation	As above plus RGO
T6–12 and L1	Strongly encourage in ages 1–10 years	Household and limited community ambulation	Same as above
L4	Strongly encourage for all ages	Community ambulation	Above plus HKAFOs, KAFOs, AFOs
L5–S1	Strongly encourage for all ages	Community ambulation	Include AFOs, GRAFOs; strongly encourage for joint protection

Abbreviations: AFO, ankle foot orthosis; GRAFO, ground reaction ankle foot orthosis; HKAFO, hip knee ankle foot orthosis; KAFO, knee ankle foot orthosis; RGO, reciprocating gait orthosis.

Source: Adapted from Betz RR, Mulcahey MJ, eds. *The Child with a Spinal Cord Injury*. Rosemont, IL: American Academy of Orthopaedic Surgeons; 1996:849.

BLADDER MANAGEMENT

After SCI, most patients develop a neurogenic bladder. While still in spinal shock, this tends to be a hypotonic bladder, but as spinal shock resolves, the bladder often transitions to a spastic bladder. Cauda equina syndrome and damage to the conus medullaris may result in a flaccid/hypotonic bladder. In the acute period, an indwelling catheter is typically placed to drain the bladder. This protects the bladder and kidneys from increased pressure and ureteral reflux, and allows for close management of fluid status. While easy to manage, long-term use of indwelling catheters leads to increased risk of urinary tract infection (UTI), shrinking of the bladder, stretching and incompetence of the sphincters, and breakdown of the urethra.

The goal of bladder management is to gain continence of the urinary bladder, promote independence, minimize UTIs, and protect the kidneys. Voiding pressures need to be maintained less than 40 cm H_2O to minimize the risk of ureteral reflux. During the acute phase, baseline evaluations of renal and urinary function need to be performed. These include blood urea nitrogen levels, serum creatinine, urinalysis, urine culture, and renal ultrasound or IV pyelogram. It should be noted that serum creatinine values are often unreliable measures of renal clearance due to the rapid loss of muscle mass and expected changes in serum creatinine during the acute phase of SCI. When the patient has emerged from spinal shock, they should undergo urodynamic testing.

Clean intermittent catheterization or intermittent self-catheterization is the method most commonly used today for bladder management after SCI. Numerous studies have shown its efficacy and safety for long-term management of the neurogenic bladder. Self-catheterization is dependent on upper extremity function, trunk strength and balance, and genital anatomy, and is generally easier for males and more problematic for females. Mirrors are frequently used by females to better visualize the urethral opening.

TABLE 16.2 FUNCTIONAL INDEPENDENCE AFTER SPINAL CORD INJURY

	LEVEL OF INJURY				
ACTIVITIES	**C1–4**	**C5**	**C6**	**C7**	**PARAPLEGIA**
Feeding	N	A	Y	Y	Y
Dressing UE	N	A	Y	Y	Y
Dressing LE	N	A	A	A	A
Bathing	N	N	N	Y*	Y
Bladder	N	Y*	A	Y	Y
Bowel	N	N	A	Y	Y
Rolling in bed	N	N	Y*	Y	Y
Transfer-level surface	N	N	Y*	Y	Y
Manual wheelchair	N	Y*	Y	Y	Y
Power wheelchair	Y	Y	Y	X	X
Driving	N	Y*	Y	Y	Y

N, not independent; Y, independent extremities; A, independent with assistive devices. Y*, may be independent, but not expected; X, not usually needed; UEs, upper extremities; LEs, lower extremities.

For those who cannot independently manage intermittent self-catheterization, external sphincterotomy may be considered for continuously draining the bladder, but this is rarely recommended in children because it destroys any chance for urinary continence when the child is older. It is also rarely recommended in females, as there is no good external collecting device. Condom catheters are commonly used in males after sphincterectomy. Complications of external sphincterectomy may include penile erosions from the condom catheter, need for reoperation, and erectile dysfunction. A suprapubic tube can be surgically placed through the abdominal/pelvic wall into the fundus of bladder to constantly drain the bladder. This eliminates the risk of sphincter incompetence or urethral breakdown; however, the other risks of indwelling catheter use remain.

A surgical procedure such as the Mitrofanoff procedure may be used to ease self-catheterization. This creates a stoma in the abdominal wall, typically through the umbilicus, which allows easier accessibility for catheterization. This is a major surgical procedure and should be performed by an experienced pediatric urologist. It should not be performed during the initial rehabilitation period, but later, after the child has had an opportunity to live at home. Outcomes of this procedure have come from Shriners Hospitals for Children (27,28) and report relatively high satisfaction with the procedure and improved level of independence. Bladder augmentation involves surgically expanding the bladder by using additional tissue (often intestinal) to increase the capacity of the bladder. This can be done concurrently with a Mitrofanoff procedure to improve bladder continence, reduce the frequency of catheterization, and reduce the risk of renal damage.

Various medications have been used in the management of the neurogenic bladder. In addition to treating UTIs, antibiotics are sometimes used for prophylaxis with a catheterization program or treating asymptomatic bacteriuria. Recently, Clarke and colleagues (29) completed a randomized trial of prophylactic antibiotics in 85 children with neurogenic bladder. They noted a six times higher incidence of UTI in subjects treated with antibiotic prophylaxis compared to those without antibiotics. This was thought to be a result of bacteria developing antibiotic resistance. Schlager's group (30) investigated the use of nitrofurantoin to clear asymptomatic bacteriuria. Approximately 70% of subjects had asymptomatic bacteriuria, which was not cleared by nitrofurantoin. While there was a change in the type of bacteria, it resulted in the growth of resistant organisms. At this time, it is not clear that antibiotics should routinely be used for neurogenic bladder, and use may increase the risk for resistant organisms. Cranberry juice is commonly recommended to prevent UTIs, though it has not been shown to be effective in children (31).

Antimuscarinic anticholinergic agents are commonly used to relax the urinary bladder by blocking the M2 acetylcholine receptors, which normally assist in detrusor contraction. The inhibition of this receptor results in a larger bladder capacity and reduced bladder

pressure. Commonly used oral agents are oxybutynin, tolterodine, darifenacin, and solifenacin. Other medications such as imipramine and hyoscyamine are also anticholinergic but are less selective for the M2 receptor and can be used as adjunct medications. Side effects include dry mouth, decreased sweating, blurred vision, heat intolerance, and constipation. As children with SCIs, especially at cervical levels, may have impaired thermal regulation, special caution must be used regarding anticholinergics and hot environments. Oxybutynin has been used intravesically to relax the bladder directly and avoid systemic side effects. The tablet is crushed, suspended in distilled water, and instilled in the bladder after catheterization. This practice is particularly useful where environmental temperatures are high and children wish to pursue outdoor activities. Selective alpha-1 receptor antagonists such as tamsulosin can be helpful in relaxing smooth muscles near the bladder neck and therefore reducing bladder outflow pressures. In recent years, onabotulinum toxin A has been used as an intravesicular injection to decrease bladder tone. This was initially evaluated in 2000 and has been increasingly used in Europe, less so in the United States (32,33). Injections seem to last, on average, 9 to 11 months and are effective with repeat injections (34). Onabotulinum toxin type A may also be used to relax the external urinary sphincter in a dyssynergic bladder (35).

NEUROGENIC BOWEL

With the loss of neural control, the gastrointestinal tract loses voluntary control, and peristalsis slows. Stiens and associates reviewed the anatomy, physiology, and management of the neurogenic bowel. A program to control incontinence while preventing impaction must fit into the child's daily life. Factors to consider are premorbid bowel function, timing, frequency, consistency, and volume of bowel movements. The new bowel regimen should duplicate, as closely as possible, the premorbid patterns. If possible, bowel movements should be timed shortly after a meal to take advantage of the gastrocolic reflex. It is often more practical to try to time this after the evening meal, as the child is likely to be home and have more time to manage the bowel movement. Factors to be considered in the new program are diet, physical activity, equipment, oral and rectal medications, and scheduling. The diet should contain adequate fluid and fiber to provide sufficient bulk to facilitate transit through the gastrointestinal tract. Table 16.3 summarizes commonly used medications for bowel programs in SCI. Young children may only need digital stimulation or no special program to evacuate completely. Older children may, likewise, need only digital rectal stimulation to evacuate completely, but, more commonly, one or more oral or rectal medications are necessary.

Sometimes, bowel continence cannot be attained just with medications, and surgical intervention may be necessary, especially for those prone to constipation or impaction. The Malone procedure or antegrade colonic enema (ACE) creates a stoma to allow antegrade use of enemas to improve bowel evacuation. This procedure has been shown to be effective in improving continence in SCI (36). Colostomy is another surgical option for neurogenic bowel management, although its use is far more common in adults with chronic SCI and the frequency and use in children are unknown. Often, individuals with neurogenic bowel dysfunction and colostomy must continue oral bowel medications to ensure full and regular emptying of stool.

RESPIRATORY FUNCTION

Although acute pulmonary problems may not be as frequent during rehabilitation as during the initial phase after SCI, close attention should be paid to pulmonary status, especially in children who are younger and less able to communicate and in those with tetraplegia, high paraplegia, or more complete lesions. Though children with lower cervical and thoracic lesions have full diaphragmatic innervation, complete or partial paralysis of the abdominal wall and accessory respiratory muscles will weaken the cough and clearance of pulmonary secretions. Clinical symptoms of respiratory problems often develop long before radiological or laboratory evidence is present. The child should be carefully watched for changes in secretions or cough, shortness of breath, headache, change in mental status, sleepiness, and snoring. Presence of morning headache should be assumed to be a sign of hypercarbia and promptly investigated. Routine monitoring of pulmonary status during rehabilitation should, at the least, include daily auscultation, measurement of end-tidal carbon dioxide tension and transcutaneous oxygen saturation, and measurement of vital capacity and maximal inspiratory and expiratory forces in all children with quadriplegia and infants and young children with high paraplegia. Consideration should be given to monitoring overnight transcutaneous oxygen and carbon dioxide saturation levels in children with complete quadriplegia because some studies have found that a high percentage of adults with complete quadriplegia have frequent nocturnal desaturations (37–39). Prevention of problems may include percussion and postural drainage, insufflation/exsufflation devices, percussion vests, and assisted cough techniques for atelectasis prevention and secretion management. TLSO can improve lung volumes in individuals with tetraplegia, and pneumococcal immunization and yearly influenza vaccines, adequate nutritional status, respiratory muscle training, and a cardiopulmonary fitness program may all further contribute to improved respiratory health.

TABLE 16.3 BOWEL MEDICATIONS

MEDICATION	EFFECTS	NEGATIVE EFFECTS
Bulk-forming agents Psyllium	Absorb water to keep stool formed and prevent dry, hard stool	Bloating, flatulence
Stool softeners Docusate Mineral oil	Allows water to enter stool Lubricant	Diarrhea, liquid form tastes bitter and is poorly tolerated Interferes with absorption of fat-soluble vitamins, causes lipid pneumonia after aspiration
Stimulants Senna Bisacodyl	Increases colonic motility, takes 6–12 hours to work Increases colonic motility	Diarrhea, cramping Diarrhea, cramping (less with rectal suppositories)
Lubiprostone	Activates chloride channels in the intestines to improve fluid secretion in the gut	Diarrhea, nausea, stomach cramping, or pain
Saline laxatives Milk of magnesia Magnesium citrate Saline enemas	Draws water into gut to stimulate colonic motility Stimulates colonic motility, used for complete bowel evacuation Acts to evacuate distal colon	Diarrhea Large volume, tastes bad, may cause electrolyte imbalance Cramping, may cause electrolyte disturbance
Hyperosmolar Lactulose, sorbitol Polyethylene glycol Glycerin suppositories	Draws fluid into intestine Draws fluid into intestine, used for complete bowel emptying Irritant	Diarrhea, cramping, flatulence Cramping, diarrhea
Prokinetic agents Metoclopramide	Affects neurotransmitters to increase gastrointestinal motility, including gastric emptying antiemetic Promotes gastric emptying	Interacts with many drugs, cardiac arrhythmia
Rectal agents Mini-enemas Carbon dioxide suppositories	Triggers colonic peristalsis Causes rectal distension	Behavior problems

NUTRITION

Adequate nutrition is necessary to promote healing of injuries and provide energy to participate in the rehabilitation process. For many children, refusal to eat may be present, either because of lack of appetite or because this may be the only activity over which they can exert control. Loss of the sense of smell due to olfactory nerve trauma might accompany some injuries, also contributing to anorexia. Dysphagia or odynophagia will negatively influence the ability to provide oral nutrition and can be a result of an associated brain injury, facial or neck trauma, cranial nerve or high cervical SCIs, the presence of a tracheostomy tube, or medications. If there is any suspicion for dysphagia, this should be assessed clinically and/or radiographically before instituting oral feedings. Nutrition must become a non-negotiable issue during rehabilitation. If the child is unable or unwilling to eat, short-term use of nasogastric tube feedings should be considered. If the inability to eat continues longer, the placement of a gastrostomy tube should be considered, regardless of the child's swallowing function. Once a child has finally begun to eat, care must be taken that he or she not overeat and thus become overweight. No calorie guidelines are available for children with SCI, but careful monitoring of weight can assist in determining the correct level of calories necessary for growth without promoting obesity.

SKIN

Pressure ulcers are a common complication of pediatric SCI and are caused by pressure, shear, and friction, with moisture being a complicating factor. Ulcers cause a huge burden in terms of time lost from school and other activities, cost, and psychological distress. Prevention is

clearly a better solution than any treatment. The basis of prevention is thorough education of the child and family about pressure relief, avoiding moisture, and treatment of ulcers in the earliest stage. Data from Model SCI Care Systems in 2006 show that 33.5% of patients developed ulcers while still hospitalized, including 53.4% of those with complete tetraplegia, 39% of those with complete paraplegia, 28.7% of those with incomplete tetraplegia, and 18.3% of those with incomplete paraplegia (40). Fifteen to twenty percent of those seen for annual examinations developed ulcers per year during the first 5 years after injury. Although these figures may be less in children, ulcers nonetheless are costly. Various systems of classification are used for pressure ulcers (Tables 16.4 and 16.5).

Large pressure ulcers may not heal with the relief of pressure for long periods, and surgery may be necessary. Various types of closures include linear closure and several types of flaps, which are well detailed by Apple and Murray (41).

AUTONOMIC DYSREFLEXIA

Autonomic dysreflexia (AD) is dysfunction of the autonomic nervous system after SCI at or above T6. As a result of noxious stimuli below the level of injury, there is increased sympathetic activity leading to vasoconstriction below the level of injury and hypertension. The central nervous system response is vasodilatation above the level of injury, with increased vagal tone and bradycardia. Symptoms of AD include pounding headache, sweating above the level of the lesion, red splotches on the face and neck, and nasal congestion. Inciting factors are bladder and bowel distention and rapid change in position from sitting to supine. UTI, renal or bladder stones, internal or external hemorrhoids, and suppository or enema insertion may also be inciting factors. AD can present as an acute emergency, more commonly in older adults than in children, who are better able to withstand extreme hypertension.

Treatment of AD consists of relief of inciting factors. The child is immediately placed in the sitting position, and should **not** be placed supine or in the Trendelenburg position to minimize the risk of hypertensive sequelae on the cerebral vascular supply. The child should be examined for potential noxious stimuli, such as tight clothing or pressure sores. Most episodes of AD resolve with these treatments. If a rectal examination must be done, this may exacerbate the AD and should be done with the use of local anesthetic on the glove. If AD persists, nifedipine should be administered sublingually. Transdermal nitroglycerin paste can be applied to the chest wall, which can be wiped off the skin, terminating its action once the hypertensive episode resolves. Prevention of AD consists of effective bowel and bladder management programs.

Wheelchair tetraplegic athletes have been known to induce AD ("boosting") to improve their athletic performance. Performance is improved by the increased sympathetic tone, shunting blood away from the viscera, thus improving cardiac output. AD can be induced by maintaining a full bladder or using a noxious stimulus (eg, a tack) below the level of injury. Boosting is dangerous, and thus is banned in wheelchair athletics.

TABLE 16.4 SHEA CLASSIFICATION OF PRESSURE ULCERS

GRADE	DESCRIPTION
1.	Red area or ulcer of epidermis or into epidermis
2.	Full dermis thickness to subcutaneous fat
3.	Fascia and muscle exposed
4.	Bone visible
5.	Large cavity through a small sinus

Source: Adapted from Bergman SB, Yarkony GM, Stiens SA. Spinal cord injury rehabilitation: medical complications. *Arch Phys Med Rehabil.* 1997;78:553.

TABLE 16.5 NATIONAL PRESSURE ULCER ADVISORY PANEL CLASSIFICATION

GRADE	DESCRIPTION
I.	Nonblanchable erythema
II.	Partial skin loss of epidermis, dermis
III.	Full-thickness skin loss
IV.	Damage through fascia, muscle, or bone

Source: Adapted from Yarkony GM. Pressure ulcers: classification and overview. In: Betz RR, Mulcahey MJ, eds. *The Child With a Spinal Cord Injury*. Rosemont, IL: American Academy of Orthopaedic Surgeons; 1996.

HYPERCALCEMIA

As discussed previously, hypercalcemia is most likely to occur in adolescent boys in the first 2 to 3 months after SCI. Serum calcium should be routinely followed throughout the rehabilitation inpatient course, and treatment with fluids, furosemide, and calcitonin, as described previously, should be instituted. There is very little data available on the safety and effectiveness of bisphosphonates in children; as such, use is based upon clinical judgment and is controversial (42).

DEEP VENOUS THROMBOSIS

Deep venous thrombosis (DVT) and pulmonary embolism (PE) are common, potentially life-threatening complications in SCI. Although DVT is somewhat less common in prepubertal children, it still does occur. The most common

time of occurrence is during the first few weeks after the SCI and can be associated with severe trauma or the presence of central venous catheters. Recommendations for prophylaxis against DVT in pubertal children include low-dose subcutaneous heparin or low-molecular-weight heparin and calf compression pumps during the rehabilitation hospitalization. Late-occurring DVT most commonly occurs with increased immobilization related to illness or surgery. Symptoms of DVT include a swollen, warm extremity, with or without fever. If the child has sensation, this may be accompanied by pain. It may also represent a trigger for AD. Differential diagnoses include cellulitis, fracture, reflex sympathetic dystrophy, and heterotopic ossification. Diagnosis is confirmed by Doppler ultrasound. If the ultrasound is negative and the index of suspicion for DVT is high, a venogram or MRI may be necessary. Plain radiographs should be obtained, especially in prepubertal children and in those whose SCI occurred more than 3 months previously to rule out fractures. Once a DVT is confirmed, treatment is bed rest until adequate heparinization is achieved to maintain the partial thromboplastin time 1.5 to 2.5 times control values. Treatment should continue for 3 to 6 months. Complications of heparin and warfarin include bleeding for both and heparin-induced thrombocytopenia. Warfarin may interact with many medications, and the patient and family should be fully educated about this if warfarin is to be continued after hospital discharge.

TEMPERATURE REGULATION

Children with SCI above T6 frequently have problems with temperature regulation because of the loss of central control of sympathetic and voluntary muscles (43). They must thus dress according to the environmental temperature. Before investigating the source of hyperthermia or hypothermia, investigation should be made into the temperature of the environment where the child has been. Often, undressing the child or putting a blanket over the child is all that is necessary to treat the hyperthermia or hypothermia. For children who reside in areas with cold weather, the use of a Mylar space blanket to maintain body heat is recommended for emergency situations. Baclofen withdrawal with resultant severe spasticity may cause extreme hyperthermia (44).

LATEX ALLERGY

Latex allergy is commonly seen in children with myelodysplasia and is now being recognized in children with SCI. A report states the incidence of latex allergy in children with SCI is 6% to 18% (45). Children and families should be educated about this potential problem and encouraged to avoid latex when possible. Latex allergy can lead to an anaphylactic reaction. Any child (and caregivers) with any type of latex allergy should be instructed on the use of an EpiPen for an emergency.

SPASTICITY

Approximately 50% of children with SCI have spasticity, which tends to be more common in those with incomplete lesions (46). Management of spasticity has the goals of promoting function and preventing contractures and pain because of the spasticity. Simple measures include ranging, positioning, and the use of orthoses. Some patients and families think that spasticity is reduced with a daily passive standing program. If spasticity still interferes with function, medications may be considered. See Table 16.6 for a summary of common antispasticity medications.

TABLE 16.6 COMMON SPASTICITY MEDICATIONS

MEDICATION	MECHANISM OF ACTION	SIDE EFFECTS
Baclofen	Direct GABA agonist	Sedation, nausea, seizures with abrupt withdrawal
Diazepam	Indirect GABA agonist	Sedation, potential for substance abuse
Dantrolene	SR calcium channel blocker	Liver dysfunction, weakness
Tizanidine	alpha 2 adrenergic agonist	Sedation, nausea
Clonidine	alpha 2 adrenergic agonist	Hypotension (less with transdermal than oral), dry mouth, constipation
Gabapentin	Unknown	Gastrointestinal, sedation, dizziness
Onabotulinum toxin	Blocks presynaptic release of Ach at NMJ	Weakness

Abbreviations: Ach, acetylcholine; GABA, γ-aminobutyric acid; NMJ, neuromuscular junction; SR, sarcoplasmic reticulum.

Local spasticity may be treated with splinting or casting or, if severe, with the use of intramuscular onabotulinum toxin. If spasticity continues to be severe and generalized after physical measures are employed and medications are maximized, surgical management of spasticity should be considered. Selective dorsal rhizotomy has been used in the United States since the mid-1980s. Although usually performed in children with spasticity of cerebral origin, the same technique may be used in children with SCI who are at least 6 months postinjury.

A newer surgical technique is the implantation of a subcutaneous pump for continuous administration of baclofen into the intrathecal space (47). Potential complications seen with intrathecal baclofen include infection, catheter disconnection or blockage, seroma around the pump, cerebrospinal fluid leak, seizures, failure to respond to increasing doses of baclofen, and pump failure. Some deaths have been reported after implantation of baclofen pump in children with SCI (48).

PSYCHOSOCIAL ISSUES

The primary psychosocial issue during rehabilitation is funding for care, equipment, therapies, and environmental modifications after discharge from inpatient rehabilitation. While parents are dealing with these issues, they must also adjust to the new needs of their child and assist their child in adjusting. The child must adjust to the new function of his or her body and learn to reenter home, community, and school. Recreation therapy can be of great help in assisting the child to learn to move about in the community, both from the physical and the psychosocial perspectives.

EDUCATION AND VOCATION

While the child is relearning mobility and self-care skills, he or she must also begin to resume schoolwork.

Adaptations necessary in the school environment need to be addressed, including architectural barriers, attitudinal barriers, and how to function with different physical skills. The child may need new ways to access computers for school or something as simple as two sets of schoolbooks—one for home and one in each classroom—to ease the physical challenges of returning to school. School staff and students need to be educated about SCIs to the extent the child and family wish this to be done. Often, it is helpful for several members of the rehabilitation team to visit the school to discuss SCI and present a video of the child engaged in some common activities. If this can be a question-and-answer session for the other students and school staff, many misconceptions can be eliminated and school reentry eased.

EQUIPMENT AND ENVIRONMENT

WHEELCHAIRS

Children with SCI are affected in many ways, and their equipment and environment can lessen the impact of their disabilities and enable them to participate in age-appropriate activities. Children can spend the majority of their day seated in a wheelchair or stroller and the prevention of poor trunk positioning and pressure ulcer formation is of paramount importance. The backrest should provide sufficient lateral and dorsal trunk support, and arm and head rests should be supportive. The ability to relieve pressure can be provided through appropriate chair sizing to maintain an anatomic sitting position, a pressure-relieving backrest and cushion, padding over pressure-sensitive areas, and the ability for periodic positional changes. Age and level of injury will dictate the extent of changes needed in the environment and the type of equipment needed (Table 16.7). Infants and toddlers may be well served by usual infant/toddler equipment, although those who require mechanical ventilation may be well served by a twin stroller to accommodate all of the necessary equipment. As children approach 2 to 3 years of age, they need to be provided with a mobility device to allow them to explore their environment. This may be a riding toy, such as a hand tricycle or powered riding toy, or an appropriately sized wheelchair. Transportation in a

TABLE 16.7 MOBILITY EQUIPMENT OPTIONS

AGE	LEVEL	MANUAL OR POWER	CONTROLLER	SPECIAL FEATURES
0–3 years	Paraplegia Tetraplegia	Stroller or riding toy Stroller	Hand	
3–10 years	Paraplegia Tetraplegia	Manual Manual and power	Hand, chin	Tilt, recline, vent tray, standing
>10 years	Paraplegia Tetraplegia	Manual Manual and power	Hand, chin	Tilt, recline, vent tray, standing

vehicle will still require the use of an appropriate toddler car seat. For those children with tetraplegia or with medical problems that preclude the use of a manually propelled wheelchair, a power wheelchair or power-assist wheels and manual wheelchair frame may be necessary. Prior to prescribing such a device, various control systems should be tried to see if the child can learn to drive a wheelchair and which system best suits their needs. Prerequisites to learning to drive a power wheelchair include:

1. At least one repeatable motor movement to drive the chair (eg, hand, head movement, or mouth sip/puff motion)
2. Understanding of cause and effect (knowing that an action causes something to happen)
3. Understanding of directionality
4. Ability to follow simple commands (49)

Children as young as 18 months have been shown to have the ability to drive power wheelchairs (50). However, if they require complex controllers, such as chin control rather than hand controllers, they may need to be closer to 4 or 5 years of age. Furthermore, you do not know if a child can use any specific controller until he or she has tried it and close collaboration with an assisted technology professional and physical therapist is very important. School-age children and adolescents need increasingly more independence and typically travel greater distances, so they may need power mobility to allow for this independence. All children who will be transported in their wheelchairs in vehicles should have transit-ready wheelchairs that meet WC19 standards (51).

ORTHOTICS

Orthotic management of the child with an SCI must consider the child's age, developmental status, and functional status as well as the physical features of his or her home and school environments. Orthotic options include orthoses for positioning as well as orthoses to enhance function in standing or ambulation. See Chapter 6 for a detailed discussion of orthotics. A recent study looked at ambulation in 169 children and youth with SCI. After a mean follow-up of 9 years, 56 of these patients were nonambulators, 17 were community ambulators, 42 were household ambulators, and 54 were therapeutic ambulators. Young age at injury and lower neurologic levels were positively associated with greater likelihood of ambulation (52).

SPECIAL CONSIDERATIONS IN HIGH TETRAPLEGIA

Children with high tetraplegia (C1–C4 levels) all have some type of partial or complete respiratory dysfunction. Whether they require full- or part-time mechanical ventilation depends on their levels and the completeness of their lesions. Some may be ventilated only at night via a face mask, while others require tracheostomies for full-time ventilation support. Issues unique to this population include increased risk of pulmonary infection, the developmental impact of being assisted by a machine for life support, and the impact of a tracheostomy on swallowing and communication. This group of patients also has more problems with maintaining blood pressure in the upright position and in accessing their environment. Wheelchairs should be "self-contained," with all necessary equipment carried and should have a means of mechanical pressure relief with a pressure-relieving backrest and cushion and a "tilt-in-space" function.

LONG-TERM FOLLOW-UP

Children with SCI can expect to live a relatively long time (53) and thus will most likely be affected by complications related to growth that adults do not experience. These complications include contractures, which are most likely to occur during periods of rapid growth, hip subluxation, and scoliosis. A study at Shriners Hospital for Children in Philadelphia found that 93% of their patients who sustained SCI under the age of 10 years had hip subluxation as compared to 9% of those over 10 years old at the time of injury (54). While the sample size was small (only 62 patients in total), this echoes the impression of clinicians. Researchers at the same institution also looked at prevention of scoliosis in children with SCI. They found that bracing with a TLSO before the scoliotic curve reached 20 degrees delayed the time to surgical correction of the deformity.

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TRAUMATIC BRAIN INJURY

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EPIDEMIOLOGY

Traumatic brain injury (TBI) is a major cause of death and disability in children. It is the leading cause of death in children over 1 year of age. In 2009 to 2010, the Centers for Disease Control (CDC) reported that TBI resulted in a rate of 2,193.8/100,000 emergency department (ED) visits, 57/100,000 hospitalizations, and 4.3/100,000 deaths in the 0 to 4 age group and a rate of 888.7/100,000 ED visits, 23.1/100,000 hospitalizations, and 1.9/10,000 deaths for those between the ages of 5 and 14 years of age (1). The incidence of pediatric TBI peaks at two separate periods: below age 5 and in mid-to-late adolescence. The incidence of hospitalization for TBI has been reported to be 125 per 100,000 children per year in the 15- to 17-year age group (2,3). Males are more likely to sustain TBI than females, at a ratio of approximately 60% to 40% (4). Falls are the leading cause of injury in all those aged 0 to 14, but motor-vehicle-related causes increase over age 4 (5). From 2001 to 2009, the rate of reported concussion among those under 19 years of age rose 5% and in 2009, an estimated 248,418 children (age 19 or younger) were treated in U.S. EDs for sports- and recreation-related injuries that included a diagnosis of concussion or TBI (5). The leading cause of TBI-related death for children under age 4 was nonaccidental trauma (5).

Children with a history of attention deficit hyperactivity disorder (ADHD) are at a greater risk to sustain TBI than those without it. ADHD affects approximately 6% of children, has a male predominance, and a hereditary tendency. Of children who sustain TBI, prevalence of preinjury ADHD is noted to be between 10% and 20% (6).

Some authors have also evaluated the incidence of TBI in the United States by race. Langlois and colleagues (7) evaluated information from the National Center for Health Statistics. They reported a significantly higher rate of both hospitalization and death due to traffic/motor-vehicle-related causes in children aged 0 to 9 in Blacks compared to Whites. Another group reported their experience in a regional trauma center and concurred that traffic/motor-vehicle-related accidents were more frequently seen in minority children; however, there was no difference in death rates or the severity of brain injury (7).

A recent report of the prevalence of TBI from a birth cohort of individuals between the ages of birth and 25 years in New Zealand indicated that the average incidence was from 1.10 to 2.36 per 100 per year with an overall prevalence of approximately 30% with 10% meeting criteria for moderate to severe injury (8).

COSTS OF INJURY

The costs associated with pediatric TBI are significant. In a study of hospital resource utilization for pediatric TBI in the year 2000, Schneier and colleagues (3) reported that more than $1 billion in hospital charges was generated for TBI patients less than 17 years of age. A survey study of needs after hospitalization reported that at 3 months after injury, 62% of children hospitalized for at least one night after TBI received at least one outpatient health care service during the interval since injury and 26% had unmet needs. At 12 months, 31% were reported to have unmet needs (9). The cost of TBI to families is something that is difficult to quantitate. However, Hawley and colleagues (10) published a report concerning parental stress after TBI in children and adolescents. The Parenting Stress Index and General Health Questionnaire results of parents of children with hospitalization of greater than 24 hours for TBI were compared to the same measures administered to a control group of parents that was identified by the subject parents. Loss of income due to the TBI was reported by 44.3% of families. For those with a child with a severe TBI, it was 69%. Also, parents of children with TBI were found to have significantly greater stress and poorer psychological health than the comparison parents. Parents of children with TBI were noted to have clinically significant levels of stress in 41% of the cases (10).

CAUSES OF INJURY

The cause of injury differs by age. Nonaccidental trauma is responsible for 17% of brain injuries in infants and 5% in those aged 1 to 4. It causes a disproportionate percentage of severe TBI, resulting in 56% and 90% of severe injury in these two age groups (11). Motor-vehicle-related injuries are more common in adolescents than young children, accounting for 66% and 20% of TBIs in the respective age ranges (2). Falls cause 39% of TBI in those under age 14, and are especially common in those under age 5 (4). Falls are the leading cause of injury in children under age 4 (5).

ASSOCIATION WITH OTHER INJURIES

It is common for TBI to occur in association with other injuries. Children with more severe injury are more likely to have been injured in a traffic-related accident and to have associated injuries (9). It has been reported that about 50% of children with TBI have other injuries as well (12). The presence of chest and abdominal injuries has been associated with decreased survival (13,14). In one study, undetected fractures during the acute care stay were found in 16 of 60 children with TBI, some having more than one fracture (13,14).

PATHOPHYSIOLOGY

PRIMARY INJURY AND SECONDARY INJURY

It is likely that the mechanism and consequences of TBI in children differ from those in adults for both primary and secondary injuries. Children have a relatively large head and weak neck musculature, higher brain water content, and lack of myelination (15). Primary injuries related to impact and deceleration and rotational forces can be influenced by these factors. It has been suggested that forces could be more easily transmitted to deeper brain structures as a result of lack of myelination and higher brain water content (15). Primary injury related to mechanical forces includes contusions on the surface of the brain, where the brain can impact against the inner surfaces of the skull (usually focal gray matter injury) and the shearing-type injury that is associated with deceleration and rotational forces (usually diffuse white matter injury or at gray–white interfaces). Primary injury results from mechanical disruption of membranes and axons (16,17).

Secondary injuries occur due to complications or other events after the initial trauma. Potential causes of secondary injury include hypotension, hypoxia, vasospasm, infarction, prolonged seizure activity, and diffuse edema, resulting in increased intracranial pressure and a decrease in cerebral perfusion pressure (16,18). Early management of TBI has a goal of preventing secondary injury. Unfortunately, there are no guidelines concerning cerebral perfusion pressure and intracranial pressure targets for children with TBI. Values are thought to be age-dependent (19).

Contributing to both primary and secondary injury in TBI are cascades of biochemical events. Injury evolves as the cascade is initiated and progresses. Mechanisms initiating these cascades include cellular power failure, acidosis, overstimulation of excitatory neurotransmitter receptors, lipid membrane peroxidation, increase in intracellular calcium, and cellular damage by free radicals (2,16). With increasing knowledge about the biochemical processes involved, researchers are attempting to identify biomarkers in serum and cerebrospinal fluid (CSF) that will assist in diagnosis and prognostication regarding the outcome of TBI (19–22). Likewise, additional information is being sought utilizing magnetic resonance spectroscopy (MRS). In Suskauer and Huisman's review of MRS evidence, H-MRS data obtained shortly after TBI has predictive value for long-term behavioral and cognitive outcomes. Several studies have shown decreases in N-acetyl aspartate (NAA) after TBI and neurometabolite abnormalities also are predictive of overall outcome in pediatric TBI (23). Babikian and colleagues (24) found that NAA on MRS scans acquired 2 to 10 days after TBI correlated moderately to strongly with cognitive testing at 1 to 4 years postinjury. Also, mean NAA/creatinine ratio explained more than 40% of the variance in cognitive scores. They hypothesize that these values might be of assistance in predicting long-term outcome soon after injury when length of unconsciousness is not as yet known.

DIFFUSE SWELLING AND SECOND IMPACT SYNDROME

It is more common for children to experience diffuse cerebral swelling than adults (19,24,25). This could be due to increased diffusion of excitotoxic neurotransmitters through the immature brain, an increased inflammatory response in the developing brain, or increased blood–brain barrier permeability after injury in the immature brain (25). When a lucid interval is noted in children prior to deterioration in neurologic functioning post-TBI, it is likely due to the development of cerebral edema, in contrast to this phenomenon in adults being most commonly related to a focal mass lesion (2,26). This diffuse cerebral swelling is associated with a poor outcome (27). Children may experience impaired cerebral autoregulation after severe TBI (28,29). Cerebral blood flow varies with age, being approximately 24 cm/s in healthy newborns, 97 cm/s in children aged 6 to 9 years, and then decreasing to the adult value of approximately 50 cm/s (28). Some studies have suggested that children with TBI

have a lower middle cerebral artery flow rate and therefore hypoperfusion is common (28).

Another phenomenon associated with cerebral swelling is called second impact syndrome, and is said to occur after repeated concussion in children and adolescents. Brain swelling can be severe, even fatal, and develops after seemingly minor head trauma in an athlete who is still symptomatic (though at times subclinically) from a previous concussion (30). Second impact syndrome is a theoretical condition with only a few case reports available. The theory describes an initial injury (the first concussion), which deranges the brain's autoregulatory and metabolic systems enough to produce vascular engorgement and poor brain compliance. This allows marked changes in intracranial pressure with small changes in intracranial volume (30). Second impact syndrome presumes that the brain cells are in a vulnerable state after the initial concussion. Minor changes in cerebral blood flow during the second concussion result in an increase in intracranial pressure and ultimately apnea due to herniation, cerebral ischemia, and brain death (31,32). Also, there have been reports of diffuse cerebral swelling after mild TBI in sports, usually occurring in male adolescents (32).

NONACCIDENTAL TRAUMA

Nonaccidental TBI is a special subset of TBI in children. It has been described as having a clinical triad of subdural hemorrhage, retinal hemorrhage, and encephalopathy, and is commonly associated with the history given, being incompatible with the severity of the injuries, and the injuries being unwitnessed and inflicted by a solitary care provider (33). Classically, this so-called shaken baby syndrome has been described as being due to shaking alone causing tearing of bridging veins and rotational forces causing diffuse brain injury. More recent studies have indicated that there is most likely an impact in addition to the shaking episode(s). Often, nonaccidental brain injury in young children is also accompanied by a delay in seeking medical attention, potentially resulting in a hypoxic component to the mechanism of injury (18,21,33).

IMPLICATIONS OF PLASTICITY

One must consider the effect of normal developmental activities of the immature brain on the mechanisms of developing damage after TBI. Apoptotic death of neurons is a part of plasticity and normal brain development. Does this result in the developing brain being more susceptible to activating the apoptotic cascade than the adult brain (15,25,34)? If so, this could help to explain the poorer prognosis for functional outcome for those injured at a very young age (34). In one animal study of post-trauma apoptosis, for a specific developmental age, the areas that had the highest density of programmed cell death were also noted to have high numbers of apoptotic cells in general (15). It may also be possible that excitatory neurotransmitter release could result in excessive stimulation of some pathways and stimulate the development of abnormal connections or that decreased excitatory activity could decrease connections (34). This implies that the relatively high plasticity of the developing brain could actually have a negative impact on the overall outcome after diffuse TBI and be at least partially responsible for the poorer outcomes seen in those injured at a very young age.

In a discussion of plasticity after early brain insult, Anderson and colleagues (35) conclude that neither plasticity nor vulnerability theories explain the range of functional outcomes seen and that multiple factors including the extent and severity of injury, age, and environmental influences such as family, sociodemographic factors, and interventions influence outcome as well. Evaluation at too young an age will prevent the identification of problems such as executive dysfunction as those skills do not emerge until later in life, supporting the need for long-term follow-up in children with TBI.

GROWING SKULL FRACTURE

A rare complication of skull fracture in children is a growing skull fracture. It is reported to occur when a linear skull fracture in a child under age 3 is accompanied by a dural tear and a leptomeningeal cyst develops. Fluid pulsations result in bone erosion and a palpable skull defect that requires surgical repair (36–38). A series of eight children with growing skull fractures had MRI evidence of a zone of signal intensity similar to brain contusion or CSF through the margins of the fracture, leading to the conclusion that MRI can be useful in diagnosing growing skull fracture early after injury (38).

NEUROIMAGING

Computerized tomography (CT) scans are typically obtained early after significant TBI. This relatively rapid imaging study is helpful in evaluating whether there is a condition that requires prompt neurosurgical evaluation and intervention (39–42). Specifically, it is helpful in detecting extra-axial hemorrhage, fractures, acute hydrocephalus, or parenchymal hemorrhages that are relatively large. However, the presence of a skull fracture is not indicative of intracranial pathology (40).

MRI is more sensitive for the detection of intraparenchymal lesions than CT scan, but takes longer than CT and often cannot be done early postinjury due to the child's medical instability and need for supportive

interventions. It is advisable, however, to obtain MRI when the child's condition allows it. Different MRI techniques can be used to evaluate for specific abnormalities (42). Susceptibility-weighted imaging was shown to identify a greater number of lesions than other techniques in one study comparing outcomes from pediatric TBI and imaging findings (39). Others have also reported association between the volume of lesion and severity of injury (43). The total amount of diffuse axonal injury (DAI) is better demonstrated using fluid attenuated inversion recovery (FLAIR) sequences and may be more important than localized injury for outcome prediction. Diffusion weighted imaging (DWI) is also helpful in detecting DAI in children. Galloway and colleagues (44) reported that the contrast between uninjured immature brain and injured tissue is improved with this technique.

Other authors have compared neuropsychological outcomes and imaging findings longer term after injury. One study of 14 children aged 10 to 18 years, 6 to 12 months after mild to moderate TBI and a matched comparison group used diffusion tensor imaging (DTI) to evaluate white matter. Authors reported that the groups had no difference in overall intelligence, but did demonstrate differences in processing speed, working memory, executive function, and behavioral problems. Also, the TBI group had lower fractional anisotropy (FA) in three white matter regions: inferior frontal, superior frontal, and supracallosal. FA in the frontal and supracallosal regions correlated with executive function. Supracallosal FA also correlated with motor speed and behavior problems (45). Another group reported DTI findings in an acutely injured child with normal CT imaging. DTI demonstrated temporary marked increase in anisotropy in large areas of the cortical and subcortical right hemisphere at 18 hours after injury. At 135 hours postinjury, subtle changes in anisotropy were present (46). Others have also noted that FA is related to injury severity including Glasgow Coma Scale (GCS) and Glasgow Outcome Scale (GOS) at 3 months postinjury (47); other investigators have found that increasing FA over time after TBI may be indicative of functional recovery (23).

Late after injury, several different imaging findings can be used to assess global change in the brain. These include cerebral diffusivity, corpus callosum volume, and volumes of brain and ventricles. Increased diffusivity is thought to be related to an increase in the extracellular space. In young children who experience TBI, late cerebral atrophy or decreased total brain volume could be related to tissue loss due to the injury itself or impaired brain growth. In typically developing individuals, white matter is reported to increase by 12.4% from age 4 to 22 (17). One study of children and adolescents at least 6 years after TBI found a correlation between corpus callosum volume and processing speed and visuospatial abilities. Ventricular volume did not correlate as well with results of neuropsychological testing. Corpus callosum is reported to continue to increase in size in typically developing individuals into early adulthood (48). It is imperative to evaluate scans over time to see the full extent of damage (41).

Functional magnetic resonance imaging (fMRI) has been reported to be useful in adults in the assessment of individuals with disorders of consciousness. There has been a recent case report indicating that this might also be of use in children in minimally conscious states. Nicholas and colleagues (2014) noted fMRI activation in a child in response to stimuli that were personally relevant, including the child's name and a familiar voice, suggesting that this demonstrated preserved cognitive function (49).

ELECTROENCEPHALOGRAPHY

EEGs are commonly obtained for children who have sustained TBI. In the practice parameter developed by the American Academy of Neurology concerning antiepileptic drug (AED) prophylaxis in severe TBI, the authors note that in their review of studies, they did not find sufficient data to be able to make a recommendation concerning the use of EEG (50). In one report of 22 children between the ages of 1 week to 14 years at the time of TBI, the degree of EEG abnormality (mild, moderate, or severe) combined with admission GCS were predictive of outcome in the young children. This was not the case for older children. The degree of EEG abnormality was statistically significantly correlated with full-scale IQ, attention and executive function, and memory (51). Additional evaluation of the usefulness of EEG in predicting outcome is needed.

INJURY SEVERITY

The main tools used for classification of brain injury severity are the GCS, length of posttraumatic amnesia (PTA), and duration of unconsciousness. Each has its merits and drawbacks.

GLASGOW COMA SCALE

The GCS has found wide clinical application since it was first published in 1974 (52). It rates a person's verbal, motor, and eye-opening responses on a scale of 3 to 15. It has the advantages of being simple, having a relatively high degree of interobserver reliability, and having the ability to be determined shortly after injury (53). A score of 8 or less is considered to be coma and classified as severe injury, 9 to 11 as moderate injury, and 12 to 15 as mild injury. There have been studies that indicate that a GCS score of 5 or lower instead of 8 or

lower should be considered as severe injury in children, as scores lower than 5 have been associated with a good outcome (13,54–56). Although the GCS was initially formulated to aid in acute triage and in neurosurgical management, many studies have correlated outcome with initial scores. There is, however, wide patient-to-patient variability. Some have noted that the GCS in the field is more predictive of survival (14,56), and GCS later in the postinjury course (particularly the motor component at 72 hours after injury) is a better predictor of disability (14,56). Adaptations of the GCS have been made to facilitate evaluation of children (57,58). Other refinements of the scale include the number of days until a patient returns to a GCS of 6 or 15.

POSTTRAUMATIC AMNESIA AND CHILDREN'S ORIENTATION AND AMNESIA TEST

The duration of PTA is another commonly used indicator of injury severity. There is general agreement that the duration of PTA is directly correlated with the severity of injury (59–61). Compared with GCS, PTA has the merit of a longer period of observation. However, there is no generally accepted and easily applied method for determining the duration of PTA, especially in children. Assessments must be adapted, as appropriate, according to an individual's age (61). The Children's Orientation and Amnesia Test (COAT) has been helpful in evaluating the length of PTA. It was designed to assess cognition serially during the early stage of recovery from TBI in children. The COAT is composed of 16 items evaluating general orientation, temporal orientation, and memory. The duration of PTA is indicated by the number of days COAT scores are in the impaired range (62). Although this test should be useful in prospective outcome studies of children without profound injury, it has a major disadvantage because it takes 5 to 10 minutes to administer and, therefore, has not become a routine assessment on most clinical services. It has also been shown to be sensitive to nontraumatic impairment. For example, children receiving special education services fall within the impaired range, and the COAT, therefore, should be interpreted with caution (63).

DURATION OF UNCONSCIOUSNESS

Duration of unconsciousness is another measure of severity and has the advantage of longer observation than GCS. It is also easier to recognize than the duration of amnesia in children and is more easily determined in retrospective chart reviews. Unconsciousness has been defined as the inability to respond to the environment in any adaptive, meaningful way. Children can have sleep–wake cycles and still be considered unconscious (64). This is the most appropriate measure in series of more severely injured children who are unconscious for many weeks, many of whom never regain recent memory. A study conducted by Massagli and colleagues (56) concluded that there was a strong correlation between the length of time to reach GCS of 15 and early and late outcomes.

Although most outcome studies have correlated outcome with only one index of brain injury severity (65,66), McDonald and colleagues (60) compared 10 measures. In their report, the number of days to reach age-adjusted 75% performance on the COAT, the number of days to GCS 15, and the initial GCS scores were most predictive of outcome across all neurobehavioral and functional measures when measured early and at 1 year postinjury. The intercorrelations of these brain injury indexes were also quite high. In general, these indexes could be used interchangeably and a single measure of severity predicted most outcomes almost as well as multiple measures. Severity ratings as determined by these alternative criteria are summarized in Table 17.1.

In summary, it is important to use these tools and correlate them with clinical findings to make an assessment of the severity of injury and therefore possible long-term outcome. Although useful, these assessment tools do have limitations in determining outcome, and a clinician's clinical impression is also important.

COMMON MOTOR DEFICITS

A wide spectrum of motor deficits is seen after TBI. This spectrum results from the variable nature of the injury and the combination of focal and diffuse damage.

TABLE 17.1 RATING OF BRAIN INJURY SEVERITY

	MILD	MODERATE	SEVERE	PROFOUND
Initial Glasgow Coma Scale	13–15 with no deterioration	9–12 with no deterioration	3–8	
Posttraumatic amnesia	<1 hour	1–24 hours	>24 hours	
Duration of unconsciousness	<15–30 minutes	15 min–24 hours	1–90 days	>90 days

FOCAL DAMAGE

Isolated focal brain injuries can occur from a variety of causes, including brain tumor resections, gunshot wounds, and other foreign-body penetrations. The cognitive and motor deficits may vary because of differences in brain injury loci. Obviously, if there is a unilateral penetrating or focal injury involving the motor area, a hemiparesis may result. Depending on the precise location of the damage, hemiparesis may be more pronounced in the upper or lower extremity. The long-term outcomes in motor, cognitive, and behavioral functioning may be better in focal injuries versus diffuse injuries given the isolated nature of the brain damage (67).

DIFFUSE DAMAGE

The diffuse nature of TBI has resulted in a constellation of motor impairments that is familiar to clinicians who work with these problems. These include difficulties with balance, coordination, and speed of response. Despite these impairments, however, a significant number of children achieve functional mobility. In a study by Boyer and Edwards (68), at 1 year after injury, 46% of the patients walked independently without assistive devices and 27% walked with an orthosis or an assistive device. Overall, 79% had independent mobility.

Swaine and Sullivan (69) have examined early motor recovery after TBI in 16 adolescents and adults who had a GCS score of 8 or lower for at least 6 hours. Assessments included evaluation of muscle tone, range of motion, abnormal and voluntary movement, primitive reflexes, equilibrium and protective responses, and specific motor skills. There were differential patterns of recovery and differential rates of recovery among the subjects, which is to be expected considering the heterogeneous nature of TBI.

Chaplin and colleagues (70) evaluated motor performance in children after TBI. Fourteen children with TBI who were unconscious for 24 hours or longer were compared with 14 age- and sex-matched children. The Bruininks–Oseretsky Test of Motor Proficiency was administered at least 16 months after injury. Children with TBI scored significantly poorer on the Gross Motor Composite, including all subsets: running speed, balance, bilateral coordination, and strength. Also, they scored lower on the fine motor subsets for upper limb speed and dexterity. Most of these subtests involve timed tasks. Chaplin and colleagues also found a correlation between the Gross Motor Composite score and the time since injury. They concluded that this correlation supports continuing long-term improvement in skills after TBI.

Kuhtz-Buschbeck and colleagues (71) looked at gait, gross motor proficiency, and hand function in 23 children after a TBI, severe in 17 and moderate in 6, during their 5 months of inpatient stay. They were compared with age- and sex-matched healthy controls. Children with TBI showed marked reduction in gait velocity, stride length, cadence, and balance. Deficits in fine motor skills, speed, and coordination were noted on hand function tests. Hand function skills improved less than gait; degree of impairment was noted to increase with severity of injury. Younger age at injury was not associated with better recovery. It has also been noted that the absence of spasticity is a good predictor of ambulation recovery by discharge (72,73).

Others have also noted impaired fine motor skills after TBI. Again, the speed component of the assessment on these tasks may account for some of the impairments that were observed. Long-term impairment of finger tapping has been described (74). Practice of activities requiring fine motor coordination improves skills, even long after injury (75).

BALANCE

Balance is frequently found to be abnormal after TBI, as it involves effective integration of the sensory, motor-programming, and musculoskeletal system (76). Cochlear and vestibular function may be impaired. True vertigo may be present. The clinical exam could be normal, despite children being symptomatic (76). Blocking visual input during quiet standing is a simple and sensitive test for postural instability (77). Gait analysis and vestibular testing may be necessary to evaluate subtle changes leading to imbalance (76). When postural instability is assessed quantitatively, long-term impairment of static and dynamic control of posture is often found after TBI (78,79). It may be related to latency of response and asymmetric stance (80). Treatment options include oral medications, visual therapy, vestibular balance rehabilitation therapy (VBRT), and surgery (81). Oral medications, including meclizine and scopolamine, should be used sparingly, as they could slow the natural compensatory process (81). Specific training with VBRT exercises that promote habituation and/or adaptation and/or substitution can be used (81).

TREMOR

Another motor impairment that is seen is tremor, which frequently is more pronounced proximally and increases with effort and movement. Lesions have been noted in varying areas. Treatment with medications typically used for tremor may be of benefit (82,83). Andrew and colleagues (84) report stereotactic surgery to be effective in management of tremors.

TONE ABNORMALITIES

Muscle tone abnormalities, including spasticity, dystonia, and rigidity, are common after TBI. The types of problems noted vary, depending on the time since injury as well as the severity of injury. The cause of acquired brain injury also influences the type of problem that is most commonly noted. Spasticity has been noted in 38% and combined spasticity and ataxia in 39% of children and adolescents 1 year after injury (66). Rigidity or dystonia is especially common when there has been secondary injury due to hypoxia or ischemia (85).

SPASTICITY

Spasticity results from an upper motor neuron injury and is manifested by increased deep tendon reflexes and velocity-dependent resistance to movement (86,87). Several different scales are available to evaluate spasticity, but they are all subjective (81), and available quantitative tests are time-consuming (88).

Physical Management

It is important to begin treating spasticity in the acute care setting to prevent contracture development (89). Treatment approaches include range of motion, stretching, casting and splinting, medications, and surgical interventions used alone or in combination to manage spasticity. Range of motion itself may be helpful to reduce tone temporarily (90). Also, one may begin with positioning options, including but not limited to, splinting and weight-bearing, if tolerated, as well as the use of neutral warmth, gentle shaking, and reflex inhibition (91). If a child has a tendency to assume a total extension posture, positioning in side-lying with hips flexed beyond 90 degrees and neck flexion may assist in interrupting the extension pattern. If active posturing is present, one must be careful in the use of splints and casts because constant pressure against the splint or cast may result in the development of an ischemic ulcer (91). Stretching should always be included in any treatment protocol for spasticity (81).

Pharmacologic Management

Medications for treatment of spasticity can be oral, intrathecal, or injectable. Enterally administered pharmacologic agents may be beneficial in decreasing abnormal muscle tone and posturing. Their potential side effects may limit their effectiveness in this population. This is especially true of the sedating effects of baclofen and benzodiazepines. Dantrolene sodium causes sedation, despite its action at the sarcolemma. Alpha-adrenergic agonists, such as clonidine and tizanidine, have also been reported to decrease tone (81). The effectiveness of all of these medications is variable.

Early after injury, when posturing may be a problem, chlorpromazine has been of assistance. It has significant potential to cause sedation (86). Bromocriptine has also been effective in reducing posturing early postinjury.

Injectable medications include botulinum toxin and phenol motor point blocks. They can be used in combination with positioning, splinting, and casting. Early after injury, with severe posturing and intolerance of splinting, botulinum toxin may be a helpful adjunct in attempting to maintain range of motion. It is reversible, so if there is significant motor recovery, there is no permanent effect of the injection. Functional gains have been noted with the use of botulinum toxin (92–95). Phenol blocks tend to be used later after injury when there is residual difficulty with increased tone. Phenol and botulinum toxin injections can be used concurrently to treat severe spasticity and to increase the number of muscles treated at one time. If severe deformity develops, surgical tendon or muscle lengthening may need to be considered (85).

Intrathecal baclofen (ITB) infusion using a programmable pump has been shown to be effective in the treatment of spasticity of cerebral origin, particularly cerebral palsy (96,97). Studies have also shown functional improvement in gait (94–96) with the use of ITB infusion in patients with acquired brain injury. Francisco (98) and colleagues also noted improvement in activities of daily living (ADLs) and decrease in pain. Two studies have shown caregiver and patient satisfaction in individuals treated with continuous infusion of ITB by an implanted programmable pump (99,100). ITB by an implanted programmable pump should be considered if severe systemic spasticity persists (101,102). Doses can be changed, depending on the patient's progress.

DYSTONIA

Dystonia is defined as a disorder in which involuntary sustained or intermittent muscle contractions cause twitching and repetitive movements, abnormal postures, or both (87). It has been reported as a rare motor impairment and is more commonly seen in those injured as children rather than as adults (103,104). The interval between injury and onset of dystonia varies. No consistent picture is seen on neuroimaging study. Medications such as trihexiphenidyl hydrochloride, carbidopa/levodopa, and bromocriptine are used in treating dystonia. ITB infusion has also been used effectively to treat dystonia (102,105).

RIGIDITY

Rigidity is the resistance to an externally imposed joint movement, with an immediate resistance to reversal of

the direction of the movement, and the limb therefore does not tend to return to a particularly fixed posture (87). Management of rigidity is similar to the management of spasticity and dystonia; however, it is often more refractory to intervention.

COMMON SENSORY DEFICITS

OLFACTORY DYSFUNCTION (ANOSMIA)

Olfactory dysfunction is a common consequence of TBI, most frequently associated with severe injury, and has also been seen with PTA of more than 5 minutes (106). Bakker and colleagues (107) report an association between severity of anosmia and executive function in children. The incidence of anosmia varies from 5% to 65%, depending on the type and severity of the brain injury (108). Olfactory dysfunction can be a partial loss of the sense of smell (microsomia) or a complete loss of sense of smell (anosmia) (109). In a study carried out by Yousem and colleagues (110) to locate and quantify the deficits using radiographic studies, most patients with impaired olfaction showed damage to the olfactory bulbs and tracts, followed by the inferior frontal lobes and volume loss in the olfactory bulbs and tracts. Both patients and their parents are seldom aware of their deficits (111) and therefore formal testing should be done in children with TBI. The three-screen test can be used for quick, gross identification, but the University of Pennsylvania Smell Identification Test (UPSIT) is more reliable in identifying all patients with deficits (112). There is usually poor recovery from anosmia in comparison to parosmia (108). Impairment in the sense of smell may have social and safety implications (109). Those with anosmia must be cautioned to use other senses to look for dangers, such as a gas burner left on, fire hazard, or similar problems. Teenagers and young adults may need to be advised about the use of fragrance when they cannot receive any feedback about its strength.

HEARING IMPAIRMENT

Hearing impairments and impairments of vestibular function are also commonly noted. Hearing impairment may occur secondary to several causes: central processing deficit, peripheral nerve damage, cochlear injury, or disruption of the middle ear structures. Cognitive impairments that are common after TBI often interfere with the child recognizing this difficulty. It is important for clinicians to have a high index of suspicion in children and initiate screening for hearing impairment.

Vestibular impairments have already been mentioned in the discussion on balance. Vertigo secondary to vestibular impairment commonly resolves within 6 months of injury (113,114), but electronystagmogram abnormalities can persist for years (115).

Central auditory processing impairment occurs with damage to tracts or cortical tissue. In such individuals, pure tone audiometry is normal, but other studies, such as speech discrimination, or late waveforms of brainstem auditory evoked potentials are abnormal (116). Central auditory impairment is difficult for most families to understand. Their intuitive conclusion is that hearing is related to the ear, so they frequently anticipate that interventions such as a hearing aid may be helpful.

Hearing loss may be conductive in nature because of disruption of the ossicles or CSF or blood in the middle ear. Both of these types of injuries are frequently associated with fractures of the temporal bone (117). Conductive hearing loss usually recovers spontaneously in about 3 weeks. If it persists for more than 3 weeks (particularly for >30 db), a repeat audiogram and exploration of the middle ear are recommended (118). Problems related to fluid in the middle ear usually resolve spontaneously.

Sensorineural hearing loss may also be seen, but less often than conductive hearing loss (117,118). Sensorineural hearing loss is commonly noted at higher frequencies (117) and is associated with inner ear pathology (113,118). Marked variation is seen in the recovery of sensorineural hearing loss (118).

There may be trauma to the eighth cranial nerve, or injury to the labyrinthine capsule, or labyrinthine concussion, which may result in hearing loss because of the transmission of high-energy vibrations and a pattern similar to the hearing loss after prolonged noise exposure (119). Injuries to the labyrinthine capsule and the eighth cranial nerve are frequently associated with basilar skull fracture.

VISUAL IMPAIRMENT

Because of the complexity of the visual system, a variety of visual impairments can be seen. Impairments may result from injury to cranial nerves, eyes, optic chiasm, tracts, radiations, or cortical structures (120,121). Early after injury, a child may appear to be functionally blind. Although vision is often assessed by looking at response to visual threat and visual tracking, these responses do not differentiate between peripheral and central impairments. One must assess cranial nerve function to make that differentiation.

Visual acuity reduction is the most frequently detected deficit in children, but the severity varies and is associated with the severity of injury (121). Visual acuity reduction is commonly associated with frontal lobe injuries (120,121). In children with greater visual acuity impairment, optic nerve atrophy, either complete or partial, is present (121). Usually, optic atrophy is seen within

1 month after injury (120), and is correlated with the site of impact and not necessarily with the overall severity of the brain injury. Chiasmatic injury results in bitemporal visual field impairment of varying degree and is found in 0.3% of TBI cases. It may be identified on MRI (122).

Homonymous hemianopsia is seen with injuries to the optic tracts and is often associated with hemorrhage and hemiparesis. Prism lenses may be of assistance, as well as learning compensatory techniques to increase scanning of the full environment (123). The presence of visual field impairments may be associated with more severe neuropsychological impairments (124).

Central visual dysfunction may be described as visual processing or visual-perceptual problems. Cortical injury is responsible for this type of impairment and may not be confined to the occipital lobes. For example, involvement of temporal lobes may produce visual memory impairment, and involvement of parietal lobes may produce impairment of spatial awareness (125).

Injury of the third, fourth, and sixth cranial nerves may lead to a variety of visual problems (126). Diplopia may result from extraocular muscle imbalance most commonly due to trochlear palsy (126) and may be present at all times or just in particular gazes. Patching is commonly used to eliminate diplopia but results in monocular vision and related disadvantages (127). In children under 11 years old, it is important to patch eyes in an alternating manner to avoid difficulty with amblyopia. Visual motor impairments due to unilateral abducens nerve palsy in children usually resolve spontaneously within 6 months (128). Deficits that persist longer than 6 months are more likely to be associated with bilateral or complete abducens nerve palsy and are unlikely to resolve spontaneously (128).

Difficulties with convergence may also result in diplopia, and are believed to be due to supranuclear impairment. Anatomic correlates of diplopia have not been well described (126). Accommodation may also be impaired (129).

COMMON COGNITIVE DEFICITS

Although TBI can result in both motor and cognitive impairments, it is generally the cognitive impairments that most profoundly affect the individual's ability to function. As noted previously, the full extent of the child's cognitive impairment may not be known until a significant time after injury, as deficits may not become apparent until the child is at a developmental stage when one would anticipate that he or she would have a particular cognitive ability, such as abstract thinking or metacognition. In general, when children have been followed long term after injury, those who were injured at a young age typically show more cognitive impairment than those injured later in childhood (130).

ATTENTION AND AROUSAL

Arousal is a precursor for attention. It has been defined as "the general state of readiness of an individual to process sensory information and/or organize a response" (131). Although there have not been systematic studies of pharmacologic interventions to improve arousal in children with TBI, a number of medications have been used and reported in case studies. One retrospective report of amantadine in children with TBI noted that compared to a group of children who had not been started on any neurostimulant medication, those on amantadine had a greater increase in their Rancho Los Amigos level during hospitalization. The amantadine group had lower initial Rancho scores and GCS (132). Dopaminergic medication use has also been reported, again in a retrospective review. In this report, the children's Western Neuro Sensory Stimulation Profile scores prior to and during medication were compared. Also, the rates of change in these scores before and after medication were compared. Significant differences were noted, suggesting that the medication could be contributing to the accelerated rate of improvement (133).

As noted previously, children with a prior history of ADHD are at an increased risk to sustain TBI. Likewise, attentional problems are common after TBI, affecting an additional approximately 20% (6). The severity of TBI is reported to be associated with the likelihood of developing attentional problems (134,135). The attentional problems seen after TBI in children are not the same as seen in developmental ADHD. It has been reported that skills that develop earlier in childhood are relatively spared compared to those that develop later. Therefore, sustained attention and divided attention are more significantly impaired than focused attention (136). Also, children with TBI tend to have slower response speeds than children with developmental ADHD (137). Both behavioral interventions and medications have been used as treatment for children with attentional problems after TBI. Case reports have noted improvements (138,139).

MEMORY IMPAIRMENT

Memory impairment is another common area of concern after pediatric TBI. Typically, the memory impairment that is seen is for the formation of new memories as opposed to long-term memory. This has significant implications for a child's ability to learn new information. As observed in other areas, severity of memory impairment appears to be related to the overall severity of injury. Impairment is seen in both immediate and delayed recall in severe TBI (140–143). When evaluating preschool children who had experienced TBI, Anderson and colleagues (141) found that over time, children did show developmental progress in their memory skills; however, children with more severe TBI did less well over time. They saw

this trend as well for both the learning and memory measures that they evaluated. It has been reported that verbal memory is more impaired than visual memory after TBI in children and that unstructured retrieval is the most impaired aspect of memory (144). Memory impairment is a challenging deficit to attempt to address during rehabilitation. Different approaches include trying to improve recall through memory practice, using organizational strategies or mnemonics, using teaching techniques to make learning more efficient (including backward chaining), and making use of compensatory techniques such as a memory notebook or electronic device (143,145,146). Avoiding purely verbal teaching, making use of structured activities in teaching, and increased repetition have been advocated as well (144).

BEHAVIORAL PROBLEMS

Behavioral sequelae are also common after TBI in children. These can include impulsivity, personality changes, depression, anxiety, becoming easily frustrated, aggression, and sleep problems (147). These problems persist long term and are reported in 10% to 50% of children with TBI (148,149). Some authors report an increase of emotional and behavioral symptoms over time (149). Also, a number of authors note that those who sustain TBI are more likely to have a preinjury history of behavioral or psychiatric concerns (148,150). Behavioral problems can be significantly disabling even in the absence of significant mobility or ADL impairment (148). Behavioral problems appear to be more significant and more common in those injured at a younger age (150). Approaches to address behavioral concerns include providing structured environments and daily routines but allowing the individual to make choices when possible, as well as assisting in breaking down tasks to their component parts, providing cues or aids for organization, creating situations in which the individual will be successful, and helping the individual to communicate the need to escape a task or situation. Positive reinforcement of desired behaviors has also been used. Involvement of family members in the process is important (150,151). Various medication interventions have been tried in the past, but none has been shown to be ultimately superior to others in addressing this variety of behavioral symptoms. It is imperative that those working with the individual understand that the behavioral problems are neurologically based. Behavioral symptoms are strong predictors of family burden over time (149).

COMMUNICATION DEFICITS

A variety of communication impairments can be seen after TBI in children. If there is focal injury in areas of the brain that control language, aphasia can be seen. Also, motoric impairment can contribute to dysarthria. In general, the communication impairments that are seen more commonly are due to other cognitive deficits, such as memory impairment and executive function concerns (150). Difficulties with response speed can contribute to a reduced rate of speech and, conversely, impulse control difficulties can result in a rapid rate of speech (152). Word finding and verbal learning deficits are common, potentially relating to memory impairment (150,153). Discourse, abstract language, and social interaction with language are all commonly impaired (150,154). Also, verbal working memory, which is commonly impaired, is important in acquiring language, reading, and arithmetic in children (155). Authors report that the ability to use language functionally is typically more impaired than one would expect from reported results of standardized intelligence testing (156).

EXECUTIVE FUNCTION

The area of executive function is one that is commonly affected, even in children who have experienced a mild TBI. It also is one in which the full effect of the injury may not be manifest until the child has matured to the point when one would expect him or her to demonstrate these particular skills. Executive function is defined as the ability to manage and direct more modular cognitive abilities in order to set, manage, and attain goals (6). This includes problem solving, organization, self-monitoring and self-regulation, self-appraisal, and self-management. It has been suggested that children are particularly susceptible to impairment in executive function if injured, as they are experiencing rapid development in this area (6). Impairments of executive function are noted to be more severe in children injured at a young age (157).

Working memory is one of the first executive function areas to develop, emerging between 7 and 12 months of age. It involves being able to temporarily store some information while concurrently processing and retrieving other data (158). It has been shown to be impaired after TBI, and the degree of impairment relates to the severity of injury (6,155,158). Other areas commonly affected by TBI include the ability to inhibit, shifting set, planning, self-monitoring and control, decision making, social cognition, and behavioral self-regulation. The Behavior Rating Inventory of Executive Function (BRIEF) is a tool that uses parent and teacher ratings to evaluate the impact of executive dysfunction on everyday life (6). Interventions for executive dysfunction have not been rigorously studied. Some have suggested using an approach that breaks tasks into problem-solving steps. Also, the provision of a structured environment and expectations is important. Incentives for progress toward a goal can be helpful. It is imperative that parents develop an effective working relationship with their child's school program providers to have open communication around the issues of executive dysfunction and its impact on school programming (6,157,159).

SOCIAL FUNCTIONING

It is not possible to totally separate social functioning from executive function; however, a separate comment on this important area will be undertaken here. A child's ability to effectively function within his or her social milieu is often significantly affected by TBI. Emotional lability is common (160). Often, children have difficulty interpreting social cues from others or recognizing the emotions being expressed (161,162). Janusz and colleagues (160) reported on social problem-solving skills in children with TBI. They found that although the children were able to articulate the social dilemmas, they chose less developmentally mature strategies as the best means to solve them and also used low-level reasoning to evaluate whether the strategies were effective. Social participation is also reported to be decreased in children with TBI compared to their typically developing peers. Bedell and Dumas (163) reported that 30% to 73% of the children with acquired brain injuries that they studied were restricted in at least one of the participation domains they evaluated. Family-reported institutional, social, and attitudinal barriers were more often contributing to this restriction than physical environmental barriers (163). One recent study reported that two-thirds of children and adolescents with TBI continued to experience difficulty in a variety of social settings at 2 years after injury. These challenges were associated with their behavioral symptoms (164).

MEDICAL CONDITIONS ASSOCIATED WITH TBI

Medical conditions associated with TBI can vary greatly from individual to individual. Essentially, all organ systems can be affected when a child sustains a TBI.

NEUROENDOCRINE DYSFUNCTION

Head trauma places the pituitary gland at risk for injury due to its encasement in the sella turcica, its delicate infundibular structures, and its tenuous vascular supply. The gland may be subject to edema, ischemia, transection of the pituitary stalk, or watershed injury (165). Dysfunction of the hypothalamic–pituitary axis can be categorized as either involving the anterior or the posterior pituitary. Posterior pituitary dysfunction results in syndromes including diabetes insipidus (DI) and the syndrome of inappropriate antidiuretic hormone (SIADH) secretion.

DI is commonly noted early after a moderate or severe TBI and can, therefore, be considered a potential marker for global hypothalamo-pituitary injury and dysfunction (166). SIADH also is a result of posterior pituitary dysfunction and needs to be distinguished from DI. The incidence of DI in children is poorly understood and poorly researched. One study (167) demonstrated incidence around 21.6% of DI in adults with moderate or severe brain injury. The study also found DI tended to be associated with a lower GCS and with the presence of cerebral edema. The fluid and sodium imbalance of DI results in a deficiency of antidiuretic hormone and excessive water loss. As antidiuretic hormone is produced in the hypothalamus, those patients who exhibit DI are felt to be predisposed to other hypothalamo-pituitary system dysfunctions. Patients with DI are hypernatremic and demonstrate polyuria and polydipsia. Although often DI is only a temporary problem for most people with TBI, it may persist. Treatment for DI is desmopressin acetate (DDAVP), which is a synthetic form of an antidiuretic hormone (168).

SIADH is another common fluid and electrolyte imbalance encountered in those with TBI, and needs to be distinguished from DI in order to provide appropriate treatment. In contrast to DI, these individuals exhibit decreased urine output, hyponatremia, and decreased serum osmolarity. SIADH is typically managed with fluid restriction and carefully reestablishing the serum sodium to a normal level in a cautious fashion. Rapid correction of the hyponatremia can cause pontine myelinolysis and possibly death (168).

CEREBRAL SALT WASTING

Cerebral salt wasting is the third cause of serum sodium imbalance in individuals with TBI. Like SIADH, cerebral salt wasting results in hyponatremia. It is essential that cerebral salt wasting be distinguished from SIADH. Unlike DI and SIADH, cerebral salt wasting does not involve the hypothalamo-pituitary system, but is believed to occur due to direct neural effects on renal tubular function. The low sodium levels seen are a direct result of abnormal renal tubular function, resulting in lost sodium along with lost fluid volume. These patients are dehydrated and, therefore, fluid restriction would cause their condition to further decline. The treatment for cerebral salt wasting involves fluid and sodium replacement (169).

ANTERIOR PITUITARY DYSFUNCTION

Literature suggests that approximately one-third to one-half of adults who have sustained a moderate or severe TBI have some hypothalamo-pituitary dysfunction (166). Children with TBI are at risk for hypothalamo-hypophyseal dysfunction, with one study identifying a rate of about 60% (170). Another group of 48 pediatric patients were found to have a 10% incidence of hypothalamo-hypophyseal dysfunction 6 months after

their brain injuries (171). The challenge in identifying which children to screen for anterior pituitary dysfunction is that many of the symptoms of anterior pituitary dysfunction mimic the effects of TBI. For instance, low levels of growth hormone are associated with symptoms such as fatigue, cognitive dysfunction, irritability, and DI (166). An individual who has sustained a TBI would commonly complain of these symptoms and have them dismissed as sequelae of the brain injury. In 2005, a consensus statement on screening for hypopituitarism after TBI recommended systematic screening for pituitary dysfunction for individuals with moderate to severe TBI who are at risk of developing pituitary dysfunction. They recommend screening for hypopituitarism if, while patients were hospitalized, they had DI or hyponatremia and hypotension. If they had SIADH or hypothyroidism identified, screening for anterior pituitary dysfunction would also be indicated. Reasons noted for foregoing anterior pituitary function screening include the individual being in a persistent vegetative state at a very low level of consciousness. Since little is known about the incidence of hypothalamo-pituitary dysfunction in children after TBI, the majority of the recommendations are extrapolated from adult literature. The treating physician should be knowledgeable, however, of the presenting features of hypothalamo-pituitary dysfunction in children, which may include growth failure, arrested or delayed puberty, amenorrhea, decreased libido, and short stature (172).

PRECOCIOUS PUBERTY

Precocious puberty is defined as the onset of puberty in girls before the age of 8 years and in boys before the age of 10 years. It can occur following TBI in children, with signs developing from 2 to 17 months after the initial injury. There is a positive correlation between increased ventricular size secondary to cerebral atrophy and the development of precocious puberty, and girls are affected much more frequently than boys (54.5% in girls to 4.5% in boys) (173). The signs of precocious puberty include onset of secondary sexual development prior to the predicted age and accelerated linear growth. These children demonstrate advanced bone age and premature closure of the epiphyseal plates. Because precocious puberty places a social and emotional burden on the patient and family, and because of the development of short stature secondary to premature epiphyseal plate closure, it is essential that the physician have a watchful eye for precocious puberty and be prepared to evaluate for it and treat it if indicated.

RESPIRATORY DYSFUNCTION

Recommendations for the treatment of children with TBI include transitioning from endotracheal intubation to tracheostomy for ventilatory support around the time the patient is 7 to 10 days postinjury. The tracheostomy allows for pulmonary support, easier secretion clearance, and better long-term airway management. The tracheostomy is not without complications, though, including, the potential for vocal cord paralysis, tracheal stenosis, subglottic and glottic stenosis, and tracheomalacia (174). The ultimate goal is to move toward decannulation once controlled ventilation is no longer needed and when the patient is able to manage his own secretions. Another reason to move toward decannulation is to avoid the increased nursing and respiratory care requirements when the tracheostomy tube is in place. These increased needs can complicate discharge, as some long-term care facilities are unwilling to provide care for patients with tracheostomies and family members may be anxious and apprehensive about caring for a child who has one (175). The stepwise fashion moving toward decannulation has been described by Klingbeil (176). The process begins with downsizing the tracheostomy tube sequentially until, ultimately, an uncuffed small tube is tolerated. Then capping of the tracheostomy tube is recommended as the clinician evaluates the patient's tolerance. If the patient is able to maintain oxygen saturations with a comfortable breathing effort and demonstrate effective cough with good management of secretions, the tube is removed and an occlusive dressing is placed to allow the site to heal. If the patient is demonstrating difficulty during the process of decannulation with worsening respiratory function or distress, it is recommended that the patient undergo direct laryngoscopy prior to decannulation in order to evaluate for concerns such as tracheal granuloma.

NUTRITIONAL MANAGEMENT

Very early after severe TBI, it is important for the primary team to place emphasis on the child's nutritional status. Guidelines have been established for achieving adequate nutritional management in this population (177). These guidelines are mostly from the adult TBI literature, as there is quite limited pediatric research regarding nutrition after TBI. Metabolism is reported to be increased after severe TBI in children, causing increased nutritional requirement. Phillips and colleagues (178) studied pediatric TBI survivors who had initial GCS between 3 and 8. Overall, the energy expenditure in those patients was 130% of their expected metabolism. Phillips also found that weight loss ranged between 2 and 26 pounds during their 2-week postinjury evaluation despite aggressive nutritional support. Moore and colleagues (179) identified metabolic profiles of pediatric TBI survivors who had initial GCS of less than 7. They found that the increased energy expenditure in that group averaged 180%. In adult literature, hypermetabolism in TBI survivors is well established. The guidelines for the adult population include the following recommendations: (a) full nutritional replacement should be initiated by day 7 postinjury;

(b) enteral nutrition should be started no later than 72 hours postinjury; and (c) tight control of serum glucose is necessary to avoid hyperglycemia, which is associated with worsening ischemic injury and worse outcome. Parenteral nutrition should be started if enteral nutrition is not full and complete by day 7.

TUBE FEEDINGS

Typically, enteral support of nutrition begins with nasal jejunal or nasal gastric feedings. Jejunal tube feedings are often tolerated better due to delayed gastric emptying (174), but the goal is to move to gastric feeds with boluses of nutritional formula for more typical meal feedings to decrease the complexity of equipment needs and to more approximate the typical physiology of enteral feedings. Percutaneous gastronomy (PEG) tubes are often placed at the time tracheostomies are placed, with the presumption that the patient who requires the tracheostomy will require tube feedings for longer than an acceptable time to leave a nasal tube in place. Nasal gastric and nasal jejunal tubes are associated with an increased risk of sinusitis, and the presence of the tubes in the posterior pharynx may be a source of irritation for the restless and the agitated child with TBI. If the child's cognitive status improves and he or she achieves full nutrition by mouth with no risk for aspiration, the PEG tube can be discontinued as early as 2 to 6 weeks after it was placed once the cutaneous-gastric fistula has matured (180). Janik and colleagues (181) found that gastrostomy tubes that remained in place in the pediatric patient for greater than 8 months required surgical closure of the fistula in 92% of the patients studied.

GASTROESOPHAGEAL REFLUX DISEASE

Prior to placing a gastrostomy tube, consideration should be given to the child's likelihood of having gastroesophageal reflux disease. This can be evaluated with an upper gastrointestinal radiological study, a pH probe study, or a milk scan. Occasionally, a Nissen fundoplication will be done in conjunction with the placement of a gastrostomy tube to avoid reflux and the risk for aspiration and its associated morbidity. Children with gastrostomy tubes in place should usually be placed on an H2 receptor blocker or proton pump inhibitor to decrease risk for acid reflux as well as gastrointestinal bleeding (182).

TRANSITION TO ORAL FEEDINGS

Evaluation of the child with TBI at the bedside by speech and language pathologists and/or occupational therapists is usually the first step in determining whether to begin transitioning to oral feeding. Studies show that dysphagia, oral motor impairment, and cognitive impairment are all highly correlated in the child with TBI; therefore, evaluation at the bedside of oral motor control as well as cognitive impairment helps to determine the degree to which dysphagia is present (183). The incidence of dysphagia in this population varies with the severity of the brain injury. The overall incidence is reported to be 5.3%. Children with mild brain injuries have an incidence of dysphagia of 1%, moderate brain injuries demonstrate a 15% incidence, and severe brain injuries 68% (184). The strongest factor predicting whether dysphagia will be present in a pediatric patient is the GCS. If the GCS is less than 9 (representing a severe TBI), the child is more likely to have dysphagia. These children tend to exhibit both oral and pharyngeal deficits with reduced lingual control and a delayed swallow reflex in the majority (183). Most dysphagia in this population resolves about 12 weeks postinjury and a normal diet is resumed. Once a child is evaluated by modified barium swallow and no silent aspiration is identified, the rehabilitation team can begin oral feeding in a stepwise fashion. Typically, the team will begin with tastes of pureed foods and thickened liquids, with progression over time, as tolerated, to solid foods and thin liquids.

BOWEL MANAGEMENT

It is important to maintain regular bowl movements early on in the critical care course of a child with a TBI. A bowel management program may involve stool softeners, suppositories, and/or laxatives in order to cause regular and routine bowel movements. The patient's bowel management program needs to be adjusted depending on his or her clinical response. Narcotic medications are constipating and antibiotic medications can cause loose stools, so close follow-up and regular adjustments are indicated. Once the child is medically stable and the routine for their bowel routine is better established, the team may choose to cause bowel movements at the same time of day with the use of a suppository. In this way, "functional continence" may be obtained, with the child's bowel movements being more predictable. The agents commonly used include senna, docusate sodium, polyethylene glycol, or glycerin suppositories.

BLADDER MANAGEMENT

During acute care of the child with severe TBI, it is common for a Foley catheter to be in place for measuring urine output. After children have been transferred from the critical care unit to the rehabilitation unit, they typically are placed in diapers for management of their incontinence. In children with cognitive impairment, the

majority of their incontinence is a result of a disinhibited bladder. In these children, the bladder is emptied completely and bladder volume is reduced. If their cognitive status allows, they may be able to participate in a timed voiding program to achieve functional continence or to learn continence. Children may also have a neurogenic-type bladder with uninhibited bladder contractions, which can be treated with anticholinergic medication. This will allow for increasing bladder volume. If the clinician is faced with bladder emptying that resembles an upper motor neuron or lower motor neuron voiding pattern, it is imperative that spinal cord injury be ruled out. This possibility can usually be eliminated by demonstrating low postvoid residual volumes (185).

CENTRAL AUTONOMIC DYSFUNCTION

Central autonomic dysfunction (CAD) is a clinical entity that is manifest by a myriad of symptoms, including hyperthermia, hypertension, diaphoresis, generalized rigidity, tachypnea, decerebrate posturing, tachycardia, and pupillary changes. It has many names, including, diencephalic seizures (186), autonomic storming, autonomic dysfunction syndrome (187), hypothalamic midbrain dysregulation syndrome (188), central seizures, central storming, central fevers (189), and posttraumatic hyperthermia (190). CAD is a result of an injury to the brain that interrupts the diencephalic–brainstem connection, leading to what is called "brainstem release phenomenon (188)." Signs and symptoms will often disappear as neurologic improvement is noted, but medical management may be necessary for 6 months or more after injury in a select group of patients (191).

Management of CAD is usually initiated due to concern about an elevated body temperature. It is imperative the clinician rule out an infectious etiology, as central temperature elevation is a diagnosis of exclusion. CAD responds poorly to antipyretic medication (187), such as nonsteroidal anti-inflammatory drugs. This may be helpful when ruling out infection. Initial management at the bedside usually consists of attempting to lower the temperature by providing cooling blankets and ice packs, turning down the temperature in the room, or providing a fan in the room to cool the patient. Often, the patient's hypertension is marked enough to warrant treatment with a beta-blocker such as propranolol, which will also help reduce the heart rate and can be used on an as-needed basis (189). Bromocriptine is used by some clinicians to reduce the symptoms of CAD and has ultimately resulted in a decreased need for antipyretics (189). Morphine in combination with bromocriptine has been useful in one study. ITB has also been reported to effectively treat CAD associated with TBI (101,192).

CAD is associated with a poor prognosis. In a retrospective review of a series of children with acquired brain injury, CAD correlated positively with more protracted periods of unconsciousness and overall worse cognitive and motor outcomes one or more years postinjury. Follow-up computed axial tomography (CAT) scans in these children revealed ventricular enlargement and marked brain atrophy (191).

HETEROTOPIC OSSIFICATION

Pediatric TBI survivors have about a 14% to 23% chance of developing heterotopic ossification (174). It is more common in children who are over 11 years of age and also more common in children who have two or more extremity fractures (193). Heterotopic ossification in children with TBI is most common at the hips and knees. Diagnosis is often made approximately 1 month after injury (193,194). Signs and symptoms of heterotopic ossification include pain, decreased range of motion, and occasionally swelling (194). Deep venous thrombosis may be present concomitantly with heterotopic ossification and warrants further evaluation (195).

Treatment of heterotopic ossification begins with regular and aggressive passive range of motion for these patients. Occasionally, splinting is necessary to prevent worsening contracture. Nonsteroidal anti-inflammatory drugs such as indomethacin, ibuprofen, and aspirin are often employed in an effort to halt progression once it is identified (193,194). Although in adult rehabilitation patients heterotopic ossification is often treated with high-dose disodium etidronate, it is avoided in pediatric patients due to concerns for development of rickets or rachitic syndrome (196). Rarely does pediatric heterotopic ossification require surgery (174,193,194).

POSTTRAUMATIC EPILEPSY

In recent years, whether pediatric TBI survivors should be treated with AEDs prophylactically has been discussed frequently in the literature. Seizures after TBI are separated into immediate, early, and late posttraumatic seizures. Immediate seizures happen within the first 24 hours of injury, and early seizures happen within the first 7 days. Late seizures occur anytime after the first week following the brain injury and may begin many years after injury (197).

In adults who have TBI, early seizures correlate with the development of late seizures. However, this correlation is not seen in the pediatric population after brain injury (198). The incidence of posttraumatic seizures is greater in children than in adults. Although the majority of posttraumatic seizures in children are immediate seizures, the incidence of early seizures ranges from 20% to 39% (57,198–200) and the incidence of late seizures ranges from 7% to 12% (198,201,202). It should also be noted that

lower GCS and younger age are associated with a higher risk of early posttraumatic seizure (57,198–200,203). Children less than 2 years of age have a threefold greater risk of early posttraumatic seizures compared with children who are 2 to 12 years of age (200). In one study of children who were 3 years of age and younger at injury, the risk of late posttraumatic seizures was greatest for those under 1 year of age at the time of injury (58).

The risk of epilepsy after TBI in children and young adults was studied based on a population study from Denmark (204). The relative risk of epilepsy was twice higher after mild brain injury and seven times higher after severe brain injury compared to no brain injury for all ages. Brain injury was associated with an increased risk of epilepsy in all age groups but was highest among those older than 15 at injury. Having a family history of epilepsy increased the risk of developing posttraumatic epilepsy.

Consensus guidelines established in 2003 state that currently there is insufficient data to support a standard guideline for the prevention of pediatric posttraumatic seizures (177). The guidelines recommend that prophylactic AED not be used to prevent the development of late seizures. They did note, however, the bulk of the evidence does suggest considering AED as a treatment option to prevent early seizures in high-risk patients. The American Academy of Physical Medicine and Rehabilitation agrees that "[a]ntiepileptic drugs are not recommended after one week for seizure prophylaxis in nonpenetrating traumatic brain injuries." Young and colleagues (202) conducted a randomized, double-blinded, placebo-controlled study to evaluate phenytoin in 41 children with TBI who were followed for 18 months postinjury for the development of seizures. No statistically significant difference was distinguished between the groups in the development of late posttraumatic seizures.

Posttraumatic epilepsy is diagnosed when the patient has two or more seizures in the late period after TBI. For the child who transfers to the pediatric rehabilitation medicine unit on phenytoin or another AED, the process of weaning the medication is fairly simple. If serum levels of the AED are subtherapeutic, it is safe to discontinue the medication without weaning. Otherwise, the dose can be reduced by approximately 50% the first week and can be discontinued thereafter. Since early seizures in children are not correlated with the development of late seizures, one can obtain an EEG in children who had early seizures and if no epileptiform activity is identified, consideration can be given to weaning the AED (168).

In children who develop posttraumatic epilepsy, AED therapy should use medications that have the least effect on cognitive function. This medication should then be used at the lowest clinically effective dose in order to maximize the cognitive recovery of these patients. The consulting pediatric neurologist considers which AED to use in a given child based on factors including the clinical seizure pattern, the EEG activity, and the side effect profile of the AED.

POSTTRAUMATIC HYDROCEPHALUS AND CEREBRAL ATROPHY

Ventriculomegaly is seen commonly after severe TBI in children (64). The enlargement of the ventricular system can be either from high-pressure hydrocephalus or from cerebral atrophy resulting in hydrocephalus ex vacuo. True hydrocephalus is a result of either an obstruction in the cerebral spinal fluid flow or impairment in the absorption of cerebral spinal fluid, ultimately resulting in an increase in cerebral spinal fluid volume and pressure. Hydrocephalus can be described, therefore, as either communicating (where there is abnormality in absorption) or noncommunicating (where there is an obstruction in the flow of the cerebral spinal fluid). The majority of hydrocephalus is caused by impaired cerebral spinal fluid absorption, secondary to inflammation or secondary to subarachnoid hemorrhage.

Hydrocephalus ex vacuo describes enlargement of the ventricular system that results after cerebral atrophy and loss of brain volume (Figure 17.1). To distinguish between clinically significant hydrocephalus and the expected consequence of cerebral atrophy after severe TBI, one must consider the patient's clinical status as well as the amount of time that has passed since the injury. Overall, if the patient is continuing to demonstrate ongoing and regular improvements in his or her clinical status, ventriculomegaly is more likely to be due to cerebral atrophy. The patient who has hydrocephalus typically continues with poor clinical improvement or clinical deterioration. The CT scan findings will yield clues as well, with cerebral atrophy demonstrating areas of encephalomalacia or enlargement of sulci, while hydrocephalus demonstrates more specific changes around the ventricular system outlined in Table 17.2 (205).

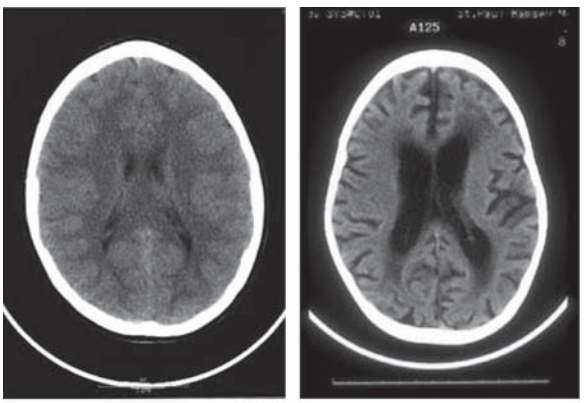

FIGURE 17.1 Cerebral atrophy. (A) Normal computed tomography (CT) scan. (B) CT scan showing posttraumatic brain injury cerebral atrophy with ventriculomegaly and increased sulci.

TABLE 17.2 COMPUTED TOMOGRAPHY CRITERIA FOR THE EVALUATION OF HYDROCEPHALUS

1. Increased size of the lateral ventricles at the anterior horns
2. Increased size of the temporal horns and the third ventricle
3. Increased size of the basilar cisterns and the fourth ventricle
4. Sulci appear normal or of decreased size
5. Periventricular hypodensity

Hydrocephalus should be suspected if clinical improvement is not noted in a patient status post-TBI or if the clinical picture includes functional decline, seizures, abnormal posturing, or increased tone. Consideration of hydrocephalus in these patients is paramount, as failure to identify hydrocephalus when it is present may delay recovery. CT scan of the brain allows for rapid detection of hydrocephalus. The treating team may then choose to have a ventricular-peritoneal shunt placed, which may improve the clinical status of the patient when normal ventricular pressures are reestablished (206).

REHABILITATION

Rehabilitation's goals are to reduce disability and help a child achieve the maximum degree of age-appropriate functional independence in physical, cognitive, social, and emotional areas after having sustained a TBI (207). In addition to the prevention of secondary impairment, facilitation of improved function, education in the use of compensatory techniques, and evaluating and potentially modifying the child's environment are also important considerations in minimizing handicap. Rehabilitation efforts include attempting to restore function or, when that is not possible, to teach adaptive techniques to compensate for areas of deficit (208). Context-sensitive rehabilitation with integration across many domains of functioning and providers should be practiced (209). For example, when a child is returning to school, in addition to appropriate special education, social reintegration, help with ADLs, and comfortable positioning should all be addressed. Parent and caregiver education are important as well. It is, therefore, imperative that children with TBI be involved with rehabilitation services (210). Also, it is important that these rehabilitation services be provided by individuals knowledgeable in child development (208). Rivera and colleagues (211) reported on the development of quality of care indicators for the rehabilitation of children with TBI using the RAND/UCLA Appropriateness Method and the Delphi technique. Eight domains of management including general management; family-centered care; cognitive communication, speech, language, and swallowing impairments; gross and fine motor skill impairments; neuropsychological, social, and behavioral impairments; school reentry; and community integration were identified.

They then used these indicators to examine the variations in the structure and organization of care among a sample of rehabilitation programs identified using data from the National Association of Children's Hospitals and Related Institutions, Uniform Data System of Medical Rehabilitation, and the Commission on Accreditation of Rehabilitation Facilities (CARF). Of the 74 inpatient units that treat children who were identified, 31 pediatric units and 28 all-age units responded to a 12-question survey that was developed from the indicators noted previously. Pediatric specialty units generally treated more than 20 children with TBI per year and had at least one child with TBI on the unit over 90% of the time and also had more pediatric specialty trained staff. Those centers with higher numbers of children with TBI also had more resources. Overall, being a pediatric-focused inpatient unit and admitting a large volume of pediatric TBI patients were associated with the total number of quality indicators met (212).

An additional study was done including chart abstraction related to seven domains of acute rehabilitation care at nine institutions. They studied 174 patient records. They found significant within and between institution variations in the percentage of individuals receiving recommended care for each of the domains. Institutions with pediatric specialty trained therapists had higher adherence to motor, neuropsychological, and community quality indicators. Those that only admitted children scored higher on cognitive, neuropsychological, and school reentry domains, and CARF accreditation was associated with adherence in the school reentry domain (213).

A recent retrospective review of children admitted to hospitals in the United States concluded that rates of discharge to inpatient rehabilitation after admission for TBI varied by as much as a factor of 3. There was also a variation noted regarding mortality. The authors concluded that an additional 1,981 children could experience inpatient rehabilitation after TBI each year if all states performed at the highest observed level (214).

EARLY REHABILITATION

Initiating rehabilitation services early shortens the overall hospital and rehabilitation stay (215,216). Rehabilitation efforts, therefore, should begin early while the child is in the intensive care unit (ICU). Early efforts should be aimed at reducing potential complications of immobility, including ischemic ulcers, compression neuropathies, and contractures (217). Complications due to excessive pressure can be prevented by frequent repositioning, special mattresses, and padding bony prominences.

Contractures can be prevented by initiating range-of-motion exercises and use of resting splints. Also, stimulation therapy is important during the ICU stay. Stimulation therapy involves presenting a brief structured stimulus for which one anticipates a response. It is a means of frequently assessing the child but does not cause awakening. Sometimes, rehabilitation interventions in the ICU must be limited because stimulation can increase intracranial pressure (91).

It is also helpful to have a social worker to meet with the family while the child is still in the ICU to begin education about brain injury and the rehabilitation process, as well as to provide support (91). Early transfer to a rehabilitation setting is indicated as soon as the patient is medically stable (210).

INPATIENT REHABILITATION

Inpatient rehabilitation requires the participation of an interdisciplinary, specialized team led by a rehabilitation physician to manage the multiple physical, cognitive, and social issues faced by the child (218,219). Central to this team is the injured child and his or her family.

SENSORY STIMULATION

Even before a child is following commands, rehabilitation may be initiated. In addition to providing structured stimulation and assessing responses on a frequent basis, physical and occupational therapy may work with positioning, including specialized equipment, and activities. Head and trunk control are facilitated. Also, localized responses are channeled into more purposeful activity using hand-over-hand techniques. Oral stimulation is started to help with evaluating oral motor function, and may facilitate more control and begin the process of evaluating for attempt to transition to oral feeding (91).

Computer-assisted rehabilitation can be used at many times in the rehabilitation continuum. Even when a child is not yet consistently following commands, computer programs may be useful to elicit auditory or visual attention. As responses increase, various types of switches can be used to assess the understanding of causality. Obviously, with children who are cognitively able, a wealth of software is available to work on various cognitive areas and provide structure and immediate feedback in reference to performance (91). The use of computers in rehabilitation activities can continue after discharge from the inpatient service. Although commonly used, there is no certainty whether computer-assisted therapy is more effective than more traditional neurorehabilitation intervention. Computers are only one facet of the overall rehabilitation approach (220).

INTERVENTIONS BASED ON THE COGNITIVE LEVEL

As children become more responsive and interactive, therapy can become more cognitively based, addressing specific areas of identified deficits that have been previously noted. An eclectic therapeutic approach should be used (91). Classic neurorehabilitative therapy approaches, adaptive equipment, the use of technology, and environmental modification all have the ultimate goal of increasing the child's independence and ability to function, and continuing to facilitate ongoing development and acquisition of skills. Cognitively based rehabilitation should continue even after discharge from the inpatient rehabilitation setting, as improvement in this area has been noted as far as 2 years postinjury (217).

Speech can also be impaired after a TBI. Children therefore should be assessed by a speech pathologist who can provide them with directed therapy or communication aids as appropriate (217).

PSYCHOSOCIAL SERVICES

An acquired brain injury of a child changes the entire family. Roles and responsibilities change, and the degree of disability affects the family's future activities and opportunities (91,221). Supportive services are essential not only for the injured child, but also for the entire family. It is also important to assess preinjury family functioning because this factor has been shown to have an impact on long-term outcome, especially with regard to behavioral problems (222). The injured child participates in supportive counseling in addition to cognitive rehabilitation activities. Counseling is imperative to assist in preparing for community reentry and in the recognition of the differences seen after a return to the community, in contrast to the artificial environment of the inpatient rehabilitation unit.

Providing supportive counseling and education for the patient's siblings is also important. Medical play can be an effective technique for both injured children and their siblings. Siblings may also benefit from peer support (91).

Counseling and education about TBI and its consequences can be helpful to parents. Proper training enables them to become advocates for their children and to help their children deal with the challenges they face because of the injury (222). These counseling and education needs may be long term because the parents initially may be in denial concerning the severity of injury and permanence of impairment (91,223,224). The injury results in the need to negotiate systems with which parents were previously unfamiliar. These include special education, medical and rehabilitation services, and publicly supported programs (222). Also, for families of children with severe injury and those who had difficulties before injury, stressors

continue long term, and families may need additional attention and resources to assist them in coping with the consequences of their children's injuries (225). One of the areas most severely affected after a TBI is social and peer reintegration. The inpatient rehabilitation process should also address this issue (163).

Another issue that requires attention is the potential impact of a child's TBI on family finances. Osberg and colleagues (226) found that parents of children who required transfer to a rehabilitation unit experienced difficulty with work and finances. Proactive planning, contact with employers, and the exploration of alternative funding sources can be of substantial benefit.

DISCHARGE PLANNING

Rehabilitation has become a continuum of care, being provided at many different sites and intensities of service. It is important to begin discharge planning early in rehabilitation hospitalization. The costs of caring for children with TBI are significant. The majority of those costs relate to acute care hospitalization, but for those with significant injury, up to 47% of the hospital costs are due to inpatient rehabilitation (201).

Most children are discharged to home after TBI. Determining the appropriate services, assisting the family in obtaining them (depending on their third-party payer and network requirements), and coordination with the public school system are essential elements in this planning process. Working closely with the third-party payer case manager can be helpful in obtaining the appropriate services for optimal transition. Family or other caregiver training is imperative in medical or nursing procedures as well as the management of behavioral problems after TBI.

After discharge from the acute care setting, rehabilitation continues, with reintegration into the community. Coordination of medically and educationally based services and effective communication among providers are essential. Accommodations to facilitate effective reintegration can be physical, environmental, or instructional (208).

The use of a token economy has been reported to be of benefit in children with moderate to severe TBI, although those with severe TBI were noted to respond to this only at 1 year after injury compared to those with moderate TBI responding at 6 months and 1 year after injury (227).

COMMUNITY REINTEGRATION

SCHOOL SERVICES

Children who have experienced TBI are more likely than the general population to require special education services (228). Children with TBI have learning problems (229). Twenty-five percent to 75% of children with TBI demonstrate school failure or require special education services within the first 5 years of injury. Studies demonstrate that the severity of injury is correlated with cognitive functioning after brain injury. Areas of concern include intelligence, adaptation, adaptive problem solving, memory, academic performance, motor abilities, and psychomotor problem solving (201). Other studies have demonstrated poor overall academic performance and academic promotion despite average academic achievement scores in nearly all children who have sustained TBI (228).

Most children return to school relatively soon after TBI, and many schools have an inclusive service model so that these children are in regular classrooms receiving supportive services. The wide variety of potential impairments post-TBI makes general statements about school programming challenging. It is necessary to identify the student's needs by evaluating his or her level of function and plan strategies to address those needs. Most likely, a student with a TBI will need a program that is unique to his or her individual needs, and offers flexibility, frequent communication with family, and regular monitoring (152).

INDIVIDUAL EDUCATIONAL PLANS

The Individuals with Disabilities Education Act (IDEA) was enacted in 1990 as Public Law 101-476 and allowed for the inclusion of TBI as a condition of eligibility for special education and educational assistance within the public school system. With this law in place, emphasis was placed on the child's global functioning rather than on academic performance alone. This resulted in increased emphasis on executive function deficits, memory and attention deficits, and slowed perceptual motor functions that tend to be characteristic of children with TBI (230). A team approach to the management of the Individualized Education Program (IEP) for the child with a TBI is important. The child's team should include a rehabilitation specialist, the child's school, and the child's family, at a minimum. Preparation of the initial IEP should begin while the child is still an inpatient on the rehabilitation ward. This allows for smooth transition from the inpatient rehabilitation program back to the school system (231). The involvement of the family is essential to facilitate a sense of continuity of care, and demonstrates to the parents that return to school does not represent return to the child's previous level of functioning. Ongoing difficulties will likely persist and need to be addressed accordingly. It is imperative that the team understand the dynamic and changing needs of the child with a brain injury, such that regular review and updating of the IEP occurs. The role of family involvement and family support for these children cannot be minimized, as it has been shown that there is an increased risk for maladaptive behavior in children with

TBI who came from poorly functioning families. Therefore, individual and family counseling, parent training, and child behavior management is recommended to improve these children's outcomes (232).

In recent years, a push toward identifying the best approach to assisting children with TBI within the school system has been investigated. Some states have responded with programs that provide consulting services to the school systems and their educators with a TBI team model. The school system then presents on a case-by-case basis their concerns for a given pupil, and the consulting team assists in developing an IEP. The state brain injury team will then reassess the child and the IEP. It has been demonstrated that educators who receive training in childhood TBI have increased confidence in working with these pupils (233).

Too often, children with TBI remain underserved and, in some cases, forgotten. Sometimes educators are unaware the child had a previous TBI, or if academic performance on achievement tests was within the average or acceptable range, he or she is deemed to be unaffected by the brain injury. Their diagnosis is forgotten until the child has failed academically. This is highlighted in the research estimating that there are approximately 130,000 students in the United States with special education needs after TBI; however, the U.S. Department of Education reported only about 15,000 students receiving services under the TBI label (209).

COMMUNITY SUPPORT

When the child with TBI is discharged from the hospital, it is almost certain that the child, at a minimum, will have a need for increased supervision. Ideally, the child will be transitioned back to school full time, but the family will need to care for that child when school is not in session. Community services become paramount in caring for these family units.

IN-HOME SERVICES

There are many reasons why additional support may be needed within the family home to care for the child with TBI. If the child is dependent for all aspects of care, personal care assistants (PCAs) or skilled nursing care may be necessary for a time. Even if the child is not dependent for mobility, marked behavioral changes in the child with TBI may warrant some of these services. Furthermore, if ongoing therapy services are needed to meet active rehabilitation goals, these therapies can sometimes be provided in the home setting. Social workers and case managers may be helpful, especially when poor family functioning is present. This is especially critical to attempt to offset the development of behavioral problems in these children status post-TBI (232).

OUT-OF-HOME SERVICES

The majority of children with TBI are discharged to home in the care of their families. Some children transition to medical foster placement, group homes, or skilled nursing facilities as an alternative living situation. In these circumstances, the children still need to have school services identified and accessed locally, as well as potential outpatient therapy services for their ongoing rehabilitation goals and needs.

PLANNING FOR LONG-TERM NEEDS

Ultimately, the child with a TBI becomes an adult with a remote TBI, and often ongoing services as well as resources are still needed. The time may come for the child who is dependent for all cares to require transition out of the home and into a long-term care facility or medical foster care placement. Resources are often limited in this regard, so early planning with the help of a social work team and perhaps legal consultation is appropriate. Vocational rehabilitation services should also be identified for these patients if appropriate. Often, the school system can be helpful in accessing these resources. The school may collaborate with local vocational services, independent living centers, community-based advocacy agencies, and other support systems to establish and coordinate a transition plan from the school to the community (231).

RETURNING TO SPORTS AND RECREATIONAL ACTIVITIES

Since sports and other recreational activities are typically an integral part of the childhood lifestyle, return to the community for children often involves planning for return to these activities.

For the child who has sustained a TBI, counseling the family on the safety of returning them to playing sports is challenging. This is partly due to a lack of evidence or guidelines in the rehabilitation literature. In recent years, better guidelines have become available for the management of return to play within sports after a child sustains a concussion during sporting activities (see this chapter's section on concussion), but these recommendations do not necessarily translate to appropriate recommendations in the child who sustains a TBI unrelated to sports activities. For instance, the grading of nonsports TBIs as mild, moderate, and severe is a different rating scale than grading the sports-related concussion as mild, moderate, or severe.

For the child who was injured with a moderate to severe brain injury, the guidelines remain unclear. It is known that in certain sports, such as high school football, approximately 20% of players incur a concussion each year,

though other "collision" sports can result in concussions as well, including boxing and ice hockey (234). Furthermore, sports such as basketball and soccer may result in an inadvertent concussion if players come into contact with each other, though with less force than one would expect in the collision sports. Other high-risk sports, including downhill skiing, snowboarding, and gymnastics, can be as dangerous as contact or collision sports from potential resulting blows to the body (235). For these reasons, it is challenging as a rehabilitation clinician to allow a patient who sustained a TBI to return to these activities. It is known that cognitive impairments will follow multiple mild concussions. Mildly concussed athletes demonstrate a decline in memory compared with their baseline performance (236), and athletes with a history of multiple concussions score significantly lower on memory testing (237). In the individual with a recent TBI, risking subsequent brain injury or concussion and worsening their clinical outcome is not recommended. Furthermore, the patient may sustain other traumatic injuries in attempting to return to sports as a result of poor performance due to impaired speed, response time, and information processing (238).

OUTCOMES

MEASUREMENT TOOLS

Several measures of function have been used to assess outcomes after TBI. They are variable and can involve neuropsychological assessment as well as motor testing.

The Coma/Near-Coma Scale is useful in evaluating small changes in patients who are at a low level of consciousness. It can be applied to both children and adults, and is helpful in allowing for reproducible assessment of subtle changes over time (239).

The Functional Independence Measure (FIM) and the Functional Independence Measure for Children (WeeFIM) can be used to assess global functioning (240). The FIM is useful for children who are more than 7 years of age and the WeeFIM between 6 months and 7 years of age. This tool assesses transfers, locomotion, self-care, sphincter control, communication, comprehension, and social cognition (241). The WeeFIM is often used to demonstrate gains in children with TBI during their inpatient rehabilitation stays.

The Glasgow Outcome Scale is a scale for classifying patients with TBIs into five categories: death, persistent vegetative state, severe disability, moderate disability, and good recovery (242). This scale has been modified to differentiate outcomes as they apply to children (Table 17.3). It is divided into a cognitive component and a motor component.

The Glasgow Outcome Scale has been modified with pediatric definitions of the five levels and reported by Paget and colleagues (243). They used the King's Outcome Scale for Childhood Head Injury (KOSCHI) in a retrospective study of 97 children followed by a TBI service. Interrater reliability was reported to be good (weighted kappa 0.710) for investigators with various levels of experience. The KOSCHI score did show some change over time. Twenty-three percent of children under age 8 had a decrease in the score over time (all were under age 3.5 at the time of injury), and reasons for the decrease in the score included intellectual disability not identified during early follow-up, new behavioral problems, and the development of late onset seizures.

TABLE 17.3 MODIFIED GLASGOW OUTCOME SCALE

Cognitive Status
0—Normal
1—Verbal communication, needs help in academic setting
2—Limited language, can express needs and wants, significant adaptation of academic setting
3—No language, responds to voices
4—Persistent vegetative state

Motor Status
0—Normal
1—Near-normal ambulation, needs supervision for ADLs
2—Ambulates with assistive devices and/or needs adaptive equipment for ADLs
3—Needs assistance for ambulation or ADLs
4—Nonambulatory, assistance for transfers, dependent for ADLs
5—No purposeful movement

Abbreviation: ADLs, activities of daily living.

The Pediatric Evaluation of Disability Inventory (PEDI) is another clinical assessment tool. It describes performance in the domains of self-care, mobility, and social function. The PEDI has questions about 197 functional skills, 20 caregiver assistance questions, and 20 equipment modification questions. This scale is used in children 6 months of age to 7 years of age, and correlates well with the WeeFIM, demonstrating good validity within both of the measures (240).

SURVIVAL

In the past two decades, morbidity and mortality associated with pediatric TBI has been on the decline, with children younger than 4 years of age and adolescents greater than 15 years of age having higher mortality rates. Infants still had the highest overall mortality (244). This improved mortality rate in children and adolescents may be due to improvements in medical care and surgical treatment. Potoka and colleagues (245) reported that for children who sustain severe TBI, mortality was significantly lower if the child was treated at a pediatric trauma center or at an adult trauma center with qualifications to treat children. The mortality of patients who sustained a TBI was higher if the child was treated at a level 2 adult trauma center

instead. More than two-thirds of deaths from brain injury occur at the scene or en route to the hospital in a population in which both adults and children were studied (246), but children with acquired brain injury who survive the initial injury generally live for many years. The pediatric literature evaluating mortality after TBI suggests that death from profound brain injury is only seen in children who remained in vegetative states longer than 90 days after anoxic or traumatic injury (247). These findings stand in contrast to adults who have sustained an acquired brain injury. The adult literature notes that approximately 50% of adults in vegetative states die within 1 year of their injury, whereas in the pediatric population, one-half of the children still in vegetative states 1 year after injury were still living 7 to 8 years later (247,248).

MORBIDITY BY INJURY SEVERITY

Concussions

A concussion is the transient and immediate change in neurologic function due to a mild TBI, with or without a brief loss of consciousness (240). A concussion is often referred to as getting "dinged" or having your "bell rung." Neuroimaging is typically normal following a concussion (249), and the diagnosis is made clinically. Symptoms of concussion usually resolve within 20 minutes, but postconcussive symptoms can last for days and weeks. Common concussive symptoms include headache, memory lapses, cognitive problems, confusion, feeling dazed or "foggy," dizziness, sleep problems, behavioral changes, bizarre statements, poor attention span, photophobia, diplopia, and sadness (250).

Common causes of concussions in children are sports injuries, falls, bicycle accidents, and automobile accidents (251). Yearly in the United States, more than 300,000 TBIs, mostly concussions, occur due to youth sports (252). Female athletes have a higher rate of concussions than males, thought to be secondary to their relatively weaker neck muscles being less able to absorb head and neck trauma (253). A particularly high rate of concussion has been recently reported in middle-school-age girls who play soccer, often reported to be due to heading the ball. It was also noted that many play with symptoms (254). This is unfortunate, given that cognitive and physical rest is recommended after concussion. This is supported by a recent report that cognitive activity prolongs recovery from concussion. They also reported a mean duration of symptoms of 43 days (255).

Concussions are graded by severity (Table 17.4), and return to activities depends on the concussion severity.

TABLE 17.4 WHEN TO RETURN TO PLAY

GRADES OF CONCUSSION	GRADE 1	GRADE 2	GRADE 3
Definitions	1. Transient confusion 2. No loss of consciousness 3. Concussion symptoms last <15 minutes	1. Transient confusion 2. No loss of consciousness 3. Concussion symptoms last >15 minutes	1. Any loss of consciousness
Management recommendations	1. Remove from activity 2. Examine immediately and every 5 minutes for change in status, at rest and with exertion 3. May return to activity if symptoms clear within 15 minutes	1. Remove from activity for remainder of day 2. Examine immediately and frequently for signs of deteriorating neurologic status 3. Trained person reexamine the next day 4. Full neurologic exam by physician to okay return to activity after asymptomatic for one full week at rest and with exertion	1. Transport to nearest emergency department if still unconscious or other concerning signs 2. Thorough neurologic exam on emergent basis and appropriate neuroimaging, if indicated 3. Hospital admission if pathology detected or mental status abnormal
When to return to play (period of time being asymptomatic with normal neurologic exam at rest and with exertion)	1. One grade 1 concussion: 15 minutes 2. Multiple grade 1 concussions: 1 week	1. One grade 2 concussion: 1 week 2. Multiple grade 2: 2 weeks	1. Grade 3 with brief loss of consciousness (seconds): 1 week 2. Grade 3 with prolonged loss of consciousness (minutes): 2 weeks 3. Multiple grade 3: 1 month or longer, as per evaluating physician

Source: Adapted from Quality Standards Committee of the American Academy of Neurology. The Management of Concussion in Sports (practice parameter). *Neurology*. 1997;48:581–585.

Postconcussive symptoms (252) may resolve before cognitive functioning returns to normal (256). Neuropsychological testing can detect these persistent cognitive changes. Many youth sports programs use cognitive assessment tools such as ImPACT™ (257) prior to participation and will not allow a return to activities until cognition returns to baseline (258). In general, a person should be symptom-free for 1 week before returning to activities. A recent report indicated that Wii games might be a means to provide exertional challenge to assist in assessing exercise tolerance after concussion (259).

The latest update of the American Academy of Neurology regarding the evaluation and management of concussion in sports recommends: (a) preparticipation counseling, (b) use of checklists and screening tools to assess suspected concussion, (c) removal from activity anyone suspected of having a concussion, (d) preclude return to training or play until all symptoms have resolved, (e) and a gradual, structured return to activities (260). The Ontario Neurotrauma Foundation has recently published guidelines for the diagnosis and management of concussion in children (261). They caution that children and adolescents may not be aware of or perhaps fully able to articulate the symptoms they have related to concussion. This detailed document includes guidelines for clinicians and parents and emphasizes a planful return to both cognitive and physical activities after symptom resolution.

In the days and weeks after a concussion, the injured brain cells are vulnerable to repeat injuries, which can cause extensive neuronal loss (262). For this reason, the brain should be rested following a concussion until all symptoms have resolved. Symptoms can be exacerbated and recovery slowed by strenuous physical and cognitive activities. During this "cognitive rest," physical and academic activities should be limited. Once symptoms have resolved, the patient should gradually return to activities as tolerated (263).

Repeated concussions over months or years can lead to long-term cognitive deficits (237) and increase the risk of neurodegenerative disorders such as Alzheimer disease (264). So activities that have a higher risk of concussions, such as football, boxing, and ice hockey, should be restricted if a person has suffered several concussions. Persons who have had previous concussions may be more susceptible to recurrent concussions and slower brain healing (237). Repeat concussions over hours, days, or weeks can lead to catastrophic changes, such as second impact syndrome, previously described in the pathophysiology section.

Mild to Moderate Injury

Children who sustain minor TBI may demonstrate few, if any, consequences, or they may complain of subjective complaints such as headaches, mild memory impairment, and fatigue. This constellation of symptoms is consistent with postconcussive syndrome. Although children with a mild TBI may not require a prolonged hospital stay on the rehabilitation unit, they may still have difficulty returning to school. The challenges these children may encounter include difficulty with timed tasks, impaired attention, and impaired memory. Subtle language dysfunction and impaired prosody of speech may be notable, as well as behavioral and personality changes. For these children, neuropsychological testing to identify any deficits is imperative, lest they be allowed to fall behind in their academic progress as the effect of the injury on their cognitive function goes unnoticed (168). It is encouraging, however, to note that by 1 year after injury, children who sustained a minor TBI rarely have impairment that continues to challenge them academically (265). In 2004, Hawley and colleagues identified a group of 67 school-age children who sustained TBI (35 mild, 13 moderate, 19 severe) and gathered 14 control subjects as well. They reported that two-thirds of the children with TBI exhibited significant behavioral problems and 76% of the children with behavioral problems also had difficulties with schoolwork (266). Another study has noted that children with mild TBI also demonstrate difficulties compared to typically developing peers in some areas of metacognition—specifically in their ability to recognize semantic anomalies in spoken sentences (267). A recent review article noted that the symptoms seen after mild TBI in children are not unique to mild injury, typically resolve, and are more likely to persist if there is intracranial pathology present or preexisting cognitive impairment (268). These findings suggest that although it is encouraging that so many children do well academically after sustaining mild TBI, caution must be taken to not overlook behavioral concerns or higher executive functions that may affect academic performance.

Moderate to Severe Injury

Outcome studies regarding children who sustained significant TBI have demonstrated overall fair recovery. One such study evaluating 30 subjects noted that only 1 out of the 30 subjects failed to become ambulatory by two or more years postinjury, and 6 out of the 30 subjects ultimately attended college. The evaluators found that 13 out of 30 of those subjects returned to their previous level of functioning (269). Another study in 1980 by Brink and colleagues (66) noted 73% of pediatric survivors of severe TBI were able to demonstrate independence in ambulation and self-care within 1 year postinjury.

The literature regarding academic outcomes for children after severe TBI is less encouraging. These children demonstrate lower scores on standardized tests (201). Ewing-Cobbs (230) reported these children have lower reading recognition, spelling, and arithmetic scores compared with patients who sustained only a mild to moderate brain injury. Two years post-TBI, 39% of these patients had failed a grade and 73%

of them needed special education assistance. Ewing-Cobbs (270) also reported that moderate to severe TBI sustained prior to the age of 6 had adverse persistent consequences for intellectual and academic development. These children were assessed 5 years after injury and were found to have continuing deficits with no further recovery of function, demonstrating a persistent performance gap with no "catch-up" phenomenon. They also found that children with focal nonprogressive brain injury demonstrated relatively good intellectual and academic outcomes. They concluded that there appeared to be significant limits on neurologic and cognitive plasticity. An interesting note was that the older children did fairly well on achievement testing but demonstrated poor functional academic recovery by failing a grade and needing ongoing support services. It seems that contributing components to success at school are the comorbid behavioral problems that almost two-thirds of children display after TBI and approximately three-quarters of those children demonstrate difficulties with schoolwork (253).

Shaklai and colleagues (271) found predictors of better outcomes after severe pediatric TBI including GCS greater than 5, length of unconsciousness less than 11 days, FIM and IQ at discharge from rehabilitation, and length of acute hospitalization and rehabilitation (271).

Profound Injury

Children with a profound brain injury and unconsciousness that lasted for greater than 90 days demonstrated a less favorable prognosis for recovery. In a series evaluating profoundly injured children by Kriel, only 1 of the 36 subjects had a normal motor outcome and no children demonstrated a normal cognitive outcome. Two-thirds of the patients recovered some language function, and one-quarter recovered independent ambulation with or without assistive devices (247).

Anoxic Brain Injury

Generally speaking, the children who sustain an anoxic brain injury tend to demonstrate a worse outcome than those with TBI. In a study that evaluated children who were unconscious for greater than 90 days secondary to an acquired brain injury, 75% of the subjects who had a TBI eventually regained consciousness. Only 25% with anoxic brain injury ultimately regained consciousness. One-quarter of children with TBI became ambulatory, and most of them regained some language function. Children with an anoxic brain injury who were unconscious for more than 60 days did not regain language skills or become ambulatory. A greater percentage of the children who had anoxic injuries died during the years of follow-up (272).

MORBIDITY RELATED TO AGE AT TIME OF INJURY

Since children have a better rate of survival after TBI, it is often assumed that pediatric outcomes are more favorable than adult outcomes. This is often attributed to the plasticity theory, suggesting that the young brain has a better opportunity to recover function. As noted in the pathophysiology section, however, injury to the developing brain may affect response to injury and the ability for future development and learning to occur. Also, the pediatric brain has had less time to learn skills and overlearn skills. A recent publication confirms that young age at the time of injury does not have a neuroprotective effect and that the mechanism of improvement after injury may differ between those injured as children and those injured as adults (273).

TBI during infancy has been shown to result in difficulty developing expressive and receptive language skills. Infants sustain a higher proportion of TBI that is secondary to nonaccidental trauma and their outcomes are poor. Koskiniemi (274) reported the long-term outcome of TBI in children and identified that the worst outcomes typically occurred in those children who were younger than 4 years of age. That study demonstrated similar results to a study done by Kriel (65) in which 97 pediatric patients who were unconscious for greater than 24 hours were followed, with the worst outcomes seen in children who were younger than 6 years of age and involved both cognitive and motor impairment. However, significant functional improvement has been noted in children who have participated in rehabilitation programs after nonaccidental trauma (275).

Older children show fairly good recovery of language function and independent ambulation. This was evaluated in a study of 28 adolescents followed longitudinally after brain injury. Twenty-five of them recovered language function, and 21 of them recovered independent ambulation. However, they had a lower high school graduation rate and employment rate than an age-referenced population. Their social interactions are impaired, as two-thirds of these individuals reported that after their TBI, their social life declined, and in fact, only 1 of the 28 subjects was married at the time of the follow-up, compared with 61% of the reference population (64).

LONG-TERM OUTCOMES

Anderson and colleagues (276) published a prospective longitudinal study of a group of 40 children with TBI at ages 2 to 7 that they compared to a group of 16 healthy controls acutely and at 12- and 30-month follow-up as well as at 10 years postinjury. MRI scans were obtained for the TBI subjects at 10-year follow-up. As anticipated, those with severe TBI had the poorest outcomes and the greatest deficits in the cognitive domain, particularly in

adaptive, executive, and social abilities. Younger age was linked to lower white matter volume at 10 years, but it was unrelated to functional outcomes. As noted in previous studies, family function predicted social and behavioral outcomes and preinjury adaptive function was predictive of 10-year adaptive abilities. The authors concluded that this data supported that recovery is poorer with injury to the very young and that the immature brain does not result in an increased likelihood of recovery.

Another report about attention problems after TBI included the 40 children noted previously. Those with severe TBI had continuing concerns with attention, both those that develop early and later developing skills. Again, a comparison group of 19 noninjured controls was included. Factors that contributed to attention at 10 years after injury included age at injury and acute IQ (277).

Crowe and colleagues (278) reported on the outcomes of accidental TBI in 53 children injured prior to age 3 and assessed an average of 40 months after injury. They were compared to a group of 27 children without injury. Children with moderate and severe TBI (grouped together) had significantly lower IQ scores than the control group (although still within the average range) but no differences were seen on parent behavior ratings. No group differences were seen for processing speed as measured by coding on the Wechsler Preschool and Primary School Intelligence Scale—Third Edition coding subtest. Consistent with previous research, predictors of long-term intellectual, behavioral, and social function after early TBI were socioeconomic status, family function, and parental mental health.

Rivera and colleagues (279) reported on disability seen at 3, 12, and 24 months after TBI in children and adolescents in a large prospective cohort study that included children younger than 18 years at the time of TBI and a comparison group with an arm injury. Outcome measures included health-related quality of life, adaptive skills, and participation. The cause of injury varied with the severity of injury (falls—mild 56.6%, moderate 34.1%, and severe 23.1%; motor vehicle occupant most common cause in the moderate and severe TBI groups.) At 3-, 12-, and 24-month follow-ups, children in the moderate and severe TBI groups had PedsQL, Adaptive Behavior Assessment System—Second Edition and Child and Adolescent Scale of Participation scores that were significantly lower than baseline, and at 24 months lower than the comparison group as well.

Another study looking at quality of life noted that it was the time postinjury, not the severity of injury, that correlated with quality of life findings and that these were still significant up to 5 years after injury (280).

Ganesalingam and colleagues (281) reported executive function and social competence in young children 6 months after TBI. Children were aged 3 years 0 months to 6 years 11 months when injured. A control group included children hospitalized with orthopedic injury. They concluded that poorer performance on neuropsychological tests of complex executive functions after severe TBI might be related to impairments that indicate poor self-regulation, metacognition, and effortful control. Higher levels of behavioral aspects of executive functions were related to higher levels of social competence. Individuals with moderate TBI did not perform differently than the control group for social competence.

Sullivan and Riccio (282) published a review of language functioning and deficits following pediatric TBI in 2010. They concluded that language deficits experienced by children post-TBI are quite variable and that comparison of outcomes is complicated by the lack of standard assessments or categorization of TBI. They also noted the lack of information about the interventions used to address language deficits.

PREVENTION

Prevention campaigns against child abuse and shaken impact syndrome have largely been educational campaigns provided by perinatal hospital staff and pediatricians.

Seatbelt use has been shown to reduce fatalities by 45% in passenger cars and by 60% in light trucks. Child safety seats, like seatbelts, decrease injury and death in the pediatric population when correctly installed. Their use has been associated with a reduction in mortality by 70% for infants and by 47% to 54% for toddlers. Seatbelt use in children decreased the need for hospitalization by 69% (2). Helmet use during motorized vehicle use has been documented to decrease the number of hospital-treated head injuries and the severity of motorcycle-related TBI (283).

Aggressive injury prevention campaigns, such as the "ThinkFirst" National Injury Prevention Foundation program, aim to educate children on the effects of brain injury related to gun accidents and sporting accidents, as well as the benefits of seatbelt use and general safety (244). The use of bicycle helmets has reduced the frequency and severity of brain injuries (284–287). Greenwald (2) reported bicycle helmet use decreased the risk of serious brain injury by up to 85%. Rule changes and better equipment in football have significantly reduced severe neurologic injuries (30,258). Efforts should be made to prevent mild brain injuries by avoiding risky behavior, wearing helmets when appropriate, following sports rules, and training properly. Following these guidelines can minimize the incidence and long-term consequences of concussions. Furthermore, in sporting activities, as previously discussed, guidelines for returning to play should be followed to avoid multiple concussive events and worsening cumulative effects.

Other prevention strategies to reduce TBI include lowering the height of playground equipment to no higher than 5 feet and fabricating play surfaces on the playground out of rubber, sand, or wood chips for better

absorption of impact in the event of a fall (2). Finally, prevention of pediatric TBI begins with adults modeling safe behaviors within the home. Whenever adults are around children, safety-conscious behaviors should be demonstrated, including regular and routine safety belt use and helmet use during sporting activities.

LONG-TERM REHABILITATION FOLLOW-UP

The role of the pediatric physiatrist in caring for the child with TBI continues throughout the child's development. Cognitive deficits may not actually be evident in the very young child until higher cognitive skills are expected to develop. Follow-up should continue throughout the child's development, with the need for intervention intermittently re-evaluated by the patient's physiatrist, therapists, and school team.

PEARLS AND PERILS

1. Injury at a younger age (younger than 4–6 years) typically results in poorer outcomes. This is perhaps due to increased vulnerability of the young child's brain to injury and the injury's impact on development.
2. Following a concussion, the injured brain cells are vulnerable to repeat injuries, which can cause extensive neuronal loss. For this reason, the brain should be rested following the concussion until all symptoms have resolved.
3. The long-term outcomes in motor, cognitive, and behavioral function may be better in focal injuries versus diffuse injuries, given the isolated nature of the brain damage.
4. Context-sensitive rehabilitation, with integration across many domains of functioning, and providers using the team approach should be practiced.
5. Care needs to be taken to distinguish cerebral atrophy (hydrocephalus ex vacuo) from posttraumatic hydrocephalus.
6. In children, seizures early after injury do not correlate with late seizures.
7. Long-term anticonvulsant prophylaxis has not been shown to decrease the development of late seizures.
8. Children often perform better in an evaluation setting than in their daily life.
9. It is important to be able to distinguish among DI, SIADH, and cerebral salt wasting.

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NEUROMUSCULAR DISEASES

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INTRODUCTION

Acquired or hereditary neuromuscular diseases (NMDs) are disorders caused by an abnormality of any component of the lower motor neuron—anterior horn cell, peripheral nerve, neuromuscular junction (NMJ; presynaptic or postsynaptic region), or muscle. These diseases affect children and adults with variable onset over the life span and they are often progressive with variable severity and rates of progression. While some NMDs have pathologic abnormalities isolated to one anatomic region of the lower motor neuron, with primary or secondary changes in muscle, other NMDs have been recognized as multisystem disorders. For example, myotonic muscular dystrophy may affect skeletal muscle, smooth muscle, myocardium, brain, and ocular structures; Duchenne muscular dystrophy (DMD) gives rise to abnormalities of skeletal and cardiac muscle, the cardiac conduction system, and brain; Fukuyama congenital muscular dystrophy (CMD) affects skeletal muscle and brain; mitochondrial encephalomyopathies may affect the mitochondria of multiple tissues.

Appropriate rehabilitation management of NMDs requires an accurate diagnosis. All diagnostic information needs to be interpreted, not in isolation but within the context of relevant historical information, family history, physical examination findings, laboratory data, molecular diagnostic studies, electrophysiologic findings, and pathologic information, if obtained. The features of the diagnostic evaluation of NMDs pertinent for the physiatrist and neuromuscular medicine specialist have recently been reviewed (1). A skilled synthesis of all available information may provide the patient and family with (a) a precise diagnosis or as accurate a diagnosis as is medically possible; (b) prognostic information (if available for a specific entity); and (c) anticipatory guidance for the near future. Knowledge of the natural history of specific NMD conditions helps in the ongoing rehabilitative management of progressive impairments in body structure and body function, reduced activities, disabilities, and handicap.

This chapter summarizes the diagnostic features, natural history and impairment profiles, and rehabilitation management of the most common childhood NMDs.

SPECIFIC NEUROMUSCULAR DISEASE CONDITIONS

DYSTROPHIC MYOPATHIES

"Muscular dystrophies" are debilitating myopathic disorders that present with muscle wasting and diffuse muscle weakness. They are caused by genetic mutations which produce muscle fiber necrosis and regeneration, ultimately resulting in muscle fiber loss. They are characterized by elevated serum creatine kinase (CK), and muscle biopsies typically show variation in fiber size, muscle necrosis, and increased amounts of fat and connective tissue. Most types of muscular dystrophy are not purely muscle disorders, but multisystem disorders with disease manifestations in a variety of body systems, which may include the musculoskeletal, cardiovascular, pulmonary, and gastrointestinal systems, as well as endocrine system, skin, eyes, brain, and other organ systems. Muscular dystrophies are caused by mutations of the genes encoding for structural proteins which are located either in the subsarcolemmal region, within the sarcolemmal membrane, or in the extracellular region. These proteins are important for the stability of the sarcolemmal membrane and the maintenance of muscle fiber intracellular homeostasis. They are genetically, biochemically, and clinically diverse diseases.

In healthy muscle, muscle fiber damage leads to a moderate activation of connective tissue cells. Satellite cells are activated and replace the damaged muscle with new muscle fibers. In muscular dystrophy, the primary defect (eg, dystrophin deficiency) leads to continuous myofiber degeneration with massive activation of

connective tissue cells, resulting in fibrosis. The muscle regeneration is unable to keep pace in order to replace the damaged muscle fibers with new muscle fibers.

Dystrophinopathies

DUCHENNE MUSCULAR DYSTROPHY. Duchenne muscular dystrophy (DMD) is an X-linked recessive disorder caused by a genetic mutation in the dystrophin gene at the Xp21 gene loci which codes for the protein dystrophin. The dystrophin gene which causes DMD and Becker muscular dystrophy (BMD) has a coding sequence that contains 79 exons. The primary protein product, *dystrophin* is localized to the intracellular side of the plasma membrane of all skeletal muscle cells, and other isoforms of dystrophin are located in certain types of neurons, and in cardiac muscle cells, the Purkinje cells of the cardiac conduction system, and smooth muscle cells of the gastrointestinal tract (2). Dystrophin deficiency at the plasma membrane of muscle fibers disrupts the membrane cytoskeleton and leads to the secondary loss of other components of the muscle cytoskeleton. Dystrophin links the intracellular actin cytoskeleton to the sarcolemmal membrane and serves as a shock absorber for the muscle cells and the primary consequence of dystrophin deficiency is membrane instability, leading to membrane injury from mechanical stresses, transient breaches of the membrane, membrane leakage, and a cascade of events subsequent to loss of homeostasis. Chronic dystrophic myopathy is characterized by aggressive fibrotic replacement of the muscle and eventual failure of regeneration with muscle fiber death and fiber loss. Generally loss of the open reading frame causes complete absence of dystrophin and a Duchenne phenotype. For cases with a deletion mutation, the "reading frame" hypothesis predicts that BMD patients with in-frame deletions produce a semifunctional, internally deleted dystrophin protein, whereas DMD patients with frameshift or "out-of-frame deletions produce a severely truncated protein that would be unstable and not persist (3). Characteristics of DMD and BMD are shown in Table 18.1. Consensus care considerations for DMD using expert opinions and RAND methodology have been published (4,5).

Diagnostic Evaluation. Data from the Centers for Disease Control and Prevention (CDC) Muscular Dystrophy Surveillance, Tracking, and Research Network (MD STARnet) among 156 boys without a known family history of DMD prior to birth, first signs, or symptoms were noted at a mean age of 2.5 years. Concerns resulted in primary care provider evaluation of the child at a mean age of 3.6 years. The mean age at the time of initial CK was 4.7 years. The mean age at definitive diagnosis of DMD was 4.9 years. Thus, there is a delay of about 2.5 years between the onset of DMD symptoms and the time of definitive diagnosis, unchanged over the previous two decades (6). This delay results in lost opportunities for timely genetic counseling and initiation of corticosteroid treatment. It is recommended that clinicians check the CK level early in the evaluation of boys with unexplained developmental delay as serum CK is a useful screening test for DMD and a normal CK value rules out DMD.

Approximately 60% of mutations are large-scale deletions, 5% are duplications, and the remainder, detectable from genomic DNA, are point mutations or small deletions/insertions. The remaining ~5% of mutations are due to intronic mutations that are undetectable by standard genomic analysis but result in altered splicing only detectable by messenger ribonucleic acid (mRNA) analysis from muscle tissue. Thus, lack of a detectable DMD gene mutation using standard methodology does not exclude a DMD diagnosis. Muscle biopsy and dystrophin expression analysis remains the gold standard of diagnosis, remaining particularly useful in cases where no mutation was detected by standard clinical molecular diagnostic testing. Full gene sequencing is now the standard of care in all patients at risk of a dystrophinopathy, and in the United States funding is available for detailed molecular diagnostics if insurance coverage is insufficient. This testing is both for diagnostic purposes and to identify candidates for future molecular-based therapies such as exon skipping with oligonucleotides or morpholinos, nonsense-mediated suppression therapy for the 12% to 15% of patients with DMD and BMD with premature stop codon mutations, and other novel therapies that will require knowledge of specific gene sequence alterations.

In patients suspected of dystrophinopathy with no family history and molecular genetics which do not clearly differentiate a DMD and BMD phenotype, a muscle biopsy with immunostaining and quantitative dystrophin analysis with Western blot or novel quantitative dystrophin assays using immunostaining or mass spectrometry is critical to allow patients to be eligible for future clinical trials of novel therapeutics with rigid inclusionary criteria. Immunohistochemical, immunofluorescent, or Western blot analysis can show the relative amount of dystrophin in skeletal muscle specimen, and Western blot can reveal its size, helping to distinguish between DMD and milder muscular dystrophy phenotype such as BMD. An amount of dystrophin of less than 3% to 5% of normal has been described as consistent with DMD, and greater than 20% as consistent with BMD, but standardization of dystrophin quantification is challenging, and in current clinical diagnostic practice, dystrophin expression is frequently descriptive or semiquantitative. Intermediate DMD may be characterized by a dystrophin quantity greater than 5%, ambulation past 12 years (if steroid naïve), and 14 years (if treated with steroids).

Epidemiology and Survival. The incidence of DMD, based on a number of population studies as well as neonatal screening has been estimated to be around 1:3,500 to

TABLE 18.1 CHARACTERISTICS OF DYSTROPHINOPATHIES (DMD AND BMD)

	DMD	BMD
U.S. prevalence (est.)	15,000	2,200
Incidence rate	1/3,500 to 5,500 male births	Unknown
Inheritance	X-linked	X-linked
Gene location	Xp21 (reading frame shifted)	Xp21 (reading frame maintained)
Protein	Dystrophin	Dystrophin
Onset	2–6 years	4–12 years (severe BMD) Late teenage to adulthood (mild BMD)
Severity and course	Relentlessly progressive • Reduced motor function by 2–3 years • Steady decline in strength • Life span < 35	Slowly progressive • Severity and onset correlate with muscle dystrophin levels
Ambulation status	• Loss of ambulation: 7–13 years (no corticosteroids) • Loss of ambulation 9–16 years (corticosteroids)	• Loss of ambulation: >16 years
Weakness	Proximal > distal • Symmetric • Legs and arms (lower extremity weakness predates upper extremity weakness by approximately 2 years)	Proximal > distal • Symmetric • Legs and arms
Cardiac	Dilated cardiomyopathy first to second decade Onset of signs second decade	Cardiomyopathy (may occur before weakness); third to fourth decade frequent
Respiratory	• Profoundly reduced vital capacity in second decade • Ventilatory dependency in second decade	• Respiratory involvement in subset of patients • Ventilatory dependency in severe patients
Muscle size	• Calf hypertrophy	• Calf hypertrophy
Musculoskeletal	• Contractures—ankles, hip, and knees • Scoliosis: onset after loss of ambulation	• Contractures: ankles and others in adulthood
Central nervous system	• Reduced cognitive ability in some • Reduced verbal ability	• Minority of patients have reduced cognitive ability
Muscle pathology	• Endomysial fibrosis and fatty infiltration • Variable fiber size and • Myopathic grouping • Fiber degeneration/regeneration • Dystrophin: 3% or less • Sarcoglycans: secondary reduction	• Variable fiber size • Endomysial connective tissue and fatty infiltration • Fiber degeneration • Fiber regeneration • Dystrophin: reduced (usually 20%–60% of normal)
Blood chemistry and hematology	• Newborn CK usually > 2,000 CK: Very high (10,000–50,000) by 2 years of age CK usually < 5,000 by second decade • High AST and ALT (normal GGT) • High aldolase	• CK: 5,000 to 20,000 • Lower levels of CK with increasing age

Abbreviations: ALT, alanine aminotransferase; AST, aspartate aminotransferase; GGT, gamma-glutamyl transferase.

5,500 male births (7,8). As many as one-third of isolated cases may be due to new mutations, which is considerably higher than observed in other X-linked conditions. This high mutation rate may relate to the large size of the gene. There has been a changing natural history in DMD over the past four decades affecting survival from the 1960s when there was largely no supportive treatment to the 1970s and 1980s when survival was improved by spinal surgery and earlier provision of noninvasive ventilation (9,10). Provision of noninvasive ventilation has been recognized as the main intervention affecting survival (9,11) with ventilated mean survival increasing from 17.7 years to 27.9 years in one study (9) and 19.0 years to 27.0 years in another study (11). In addition, survival in DMD has been enhanced by glucocorticoids (12) and angiotensin-converting enzyme (ACE) inhibitors (13).

Onset and Early Signs. While the history of hypotonia and delayed motor milestones are often reported in retrospect, the parents are often unaware of any abnormality until the child starts walking. There has been variability reported in the age of onset (14,15). In 74% to 80% of instances, the onset has been noted before the age of 4 years (14–16). The vast majority of cases are identified by 5 to 6 years of age. In addition to gross motor delays, cognitive and language delays have also been documented prior to the age of 3 in DMD (17,18). The most frequent presenting symptoms have been abnormal gait, frequent falls, and difficulty climbing steps. Parents frequently note the toe walking, which is a compensatory adaptation to knee extensor weakness and a lordotic posture to the lumbar spine, which is a compensatory change due to hip extensor weakness (Figure 18.1).

Occasionally, DMD is identified presymptomatically in situations where a CK value is obtained with a markedly elevated value, malignant hyperthermia occurs during general anesthesia for an unrelated surgical indication, or a diagnosis is pursued in a male with an affected older sibling.

Difficulty negotiating steps is an early feature as is a tendency to fall due to the child tripping or stumbling on a plantar-flexed ankle or the knee buckling or giving way due to knee extensor weakness. There is progressive difficulty getting up from the floor with the presence of a Gower's sign (Figure 18.2).

Pain in the muscles, especially the calves, is a common symptom. Enlargement of muscles, particularly the calves (Figure 18.3), is commonly noted. The deltoid may also be hypertrophied. With the patient's arms abducted to 90 degrees and externally rotated, the hypertrophy of the posterior deltoid and infraspinatus frequently leaves a depression between these two muscles referred to as the "posterior axillary depression sign" in DMD (Figure 18.4). The tongue is also frequently enlarged. There is also commonly an associated wide arch to the mandible and maxilla with separation of the teeth, presumably secondary to the macroglossia.

FIGURE 18.1 "Myopathic" stance in an 8-year-old male with Duchenne muscular dystrophy. Notice the lumbar lordosis to compensate for hip extensor weakness and primarily forefoot contact to compensate for knee extensor weakness.

Pattern and Progression of Weakness. The earliest weakness is seen in the neck flexors during preschool years (Figure 18.5). Weakness is generalized but predominantly proximal early in the disease course. Pelvic girdle weakness predates shoulder girdle weakness by several years. Ankle dorsiflexors are weaker than ankle plantar flexors, ankle everters are weaker than ankle inverters, knee extensors are weaker than knee flexors, hip extensors are weaker than hip flexors, and hip abductors are weaker than hip adductors (15).

The weakness progresses steadily, but the rate may be variable during the disease course. Quantitative strength testing shows greater than 40% to 50% loss of strength by 6 years of age (15). With manual muscle testing, DMD subjects exhibit loss of strength in a fairly linear

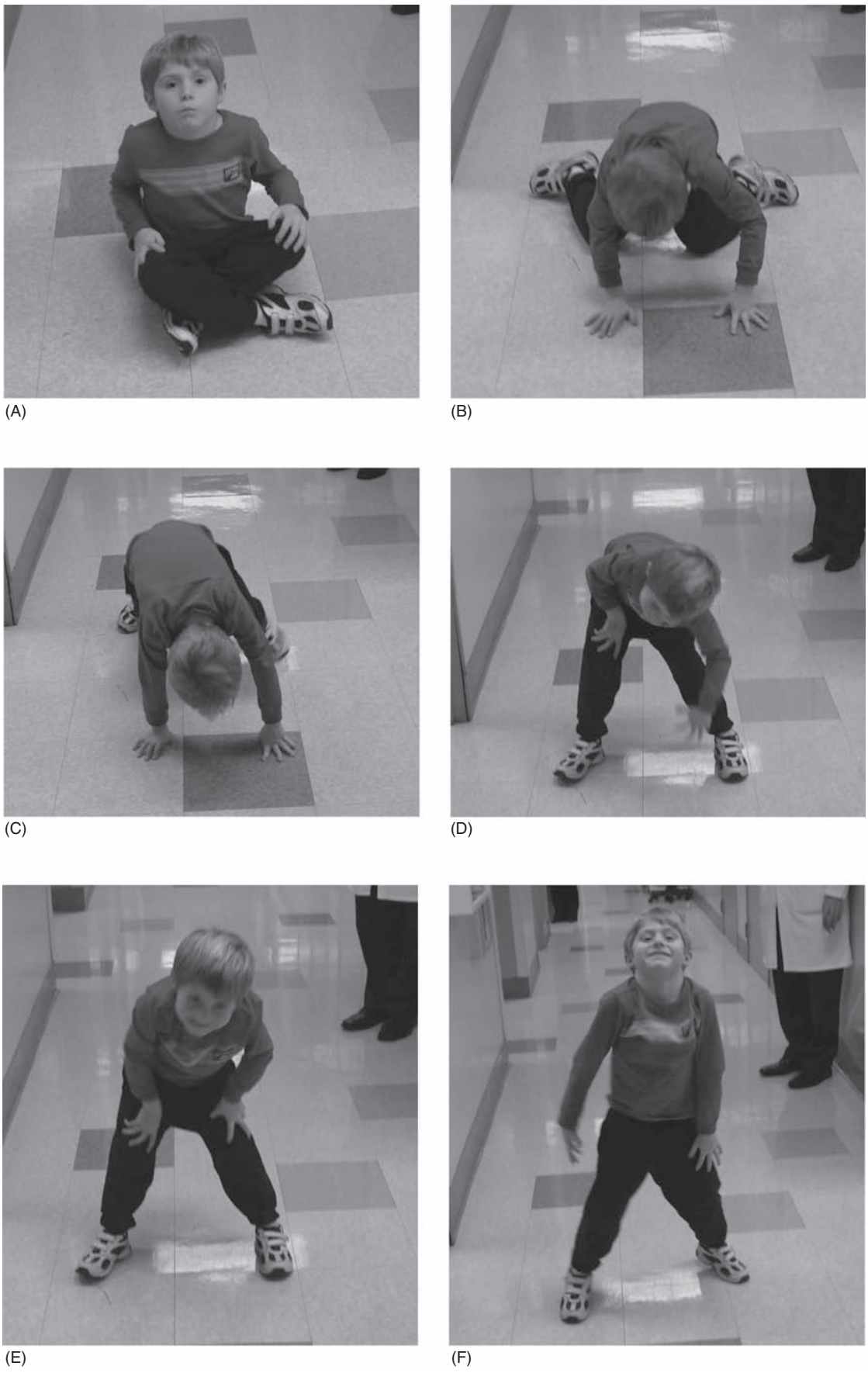


FIGURE 18.2 A–F, Gower's sign in a 7-year-old boy with Duchenne muscular dystrophy due to hip extension weakness.

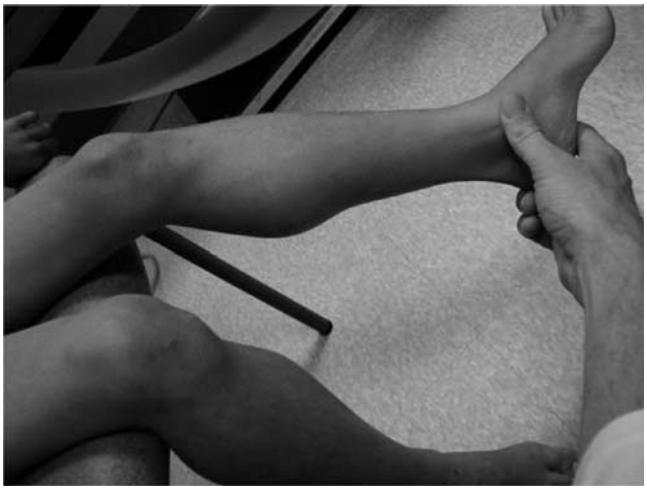


FIGURE 18.3 Calf pseudohypertrophy in a male with Duchenne muscular dystrophy.

FIGURE 18.5 Weakness of neck flexors in an 8-year-old with Duchenne muscular dystrophy makes it difficult for him to bring his chin to the chest when supine and to hold his head up when placed at the end of the examination table.

fashion from ages 5 to 13 and measurements obtained several years apart will show fairly steady disease progression. A variable course may be noted when analyzing individuals over a shorter time course (15). Quantitative strength measures have been shown to be more sensitive for demonstrating strength loss than manual muscle testing when strength is graded 4 to 5 (15).

Disease Progression and Loss of Milestones. What follows is a brief overview of the current natural history of DMD across the spectrum of disease. Furthermore, while specific functional changes are observed at different ages, it should be emphasized that the disease is due to generalized skeletal muscle involvement, and cardiomyopathy and pathologic processes involved in DMD are ongoing over the course of a patient's lifetime — while loss of ambulatory capacity and gross motor functions may be a primary focus in ambulatory boys, neuromuscular deterioration may already be measurable in the upper limb and other muscle groups. Note, the ages are approximations, and the intent is not to create artificial stages of the disease.

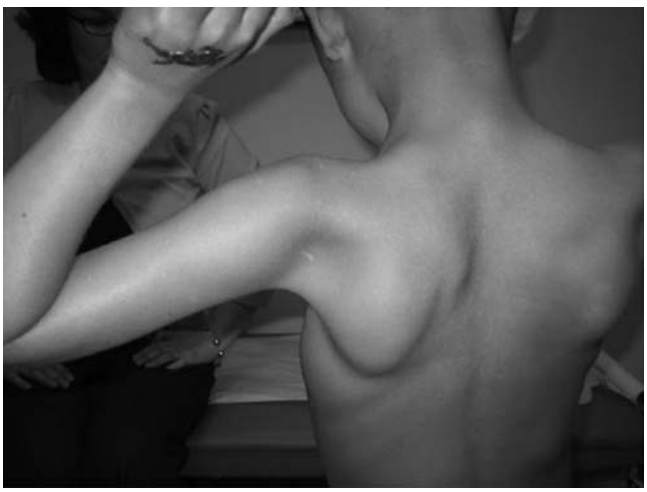


FIGURE 18.4 Posterior axillary depression sign in Duchenne muscular dystrophy. Note the prominent deltoid superolaterally and infraspinatus inferomedially.

- *Neonates/Infancy:* While DMD is rarely diagnosed in infancy, the disease is manifested at birth. Even though some of the infants detected due to family history are sometimes referred to as being asymptomatic, most will still show delayed development if evaluated with tools such as the Griffiths Mental Development Scales, an outcome measure than can be used in the very young (6–47 months) and the Bayley Scales of Infant and Toddler Development, Third Edition (Bayley-III). One study of children with DMD with mutations upstream or in exon 44 had higher Developmental Quotient (DQ) than those with mutations downstream exon 44 which are associated with involvement of dystrophin isoforms expressed at high levels in the brain. The difference was significant for total and individual subscale DQ with the exception of the locomotor subscale. Items, such as ability to run fast, or getting up from the floor consistently failed in all children, irrespective of the age or of the site of mutation.
- *Young children, early ambulant (aged 1 to 42 months):* The development of gross motor milestones is typically slower than in boys without Duchenne, and some children may show signs of delayed language and cognitive impairment. Toddlers and young children may also

be scored with developmental outcome measures such as the Bayley-III and Griffith's Developmental Scales. Gross motor scores were lower in young children with DMD at baseline compared with published controls and revealed a further declining trend at 6 months. Repeated measures analysis over 12 months revealed that gross motor scores declined further at 12 months. Cognitive and language scores were lower at baseline compared with typically developing children and did not change significantly at 6 or 12 months. Fine motor skills, also low at baseline, improved over 1 year.

- *Young ambulatory (from 4 to approximately 7 years):* A period where there may be slower gains in ambulatory function as compared to typically developing children (on 6-minute walk test (19–22) and 10-meter walk/run test) and either gains or losses in milestones as noted by end points such as the North Star Ambulatory Assessment (NSAA) (23). However, it is important to note that physiologic deterioration is ongoing and boys are increasingly falling behind normative performance levels of their normally functioning peer group.
- *Late ambulatory (from approximately seven to thirteen years of age):* Generally defined as when individuals begin to suffer a decline in their gross motor functions as well as some pulmonary function parameters, particularly maximal expiratory pressure (MEP) and maximal inspiratory pressure (MIP). During this stage of disease, there is marked progressive loss of muscle fiber in the proximal muscles, growing weakness, and the gradual loss of gross motor skills and ambulatory functions (including standing ability, stair climbing, and ultimately, the ability to walk). The six-minute walk distance (6MWD) typically declines after age 7, and a 30-meter decline in 6MWD in comparison to stability is predictive of a greater tendency to lose ambulation over the following 2 years. The average age of full-time wheelchair use in a DMD population not treated with corticosteroids has been age 10 with a range of 7 to 13 (15). Timed motor performance is useful for the prediction of time when ambulation will be lost without provision of long-leg braces. The 10-meter walk/run test has been shown to be predictive of subsequent loss of ambulation. Two large natural history studies have showed that a large proportion of DMD subjects who took 12 seconds or longer to ambulate 10 meters or approximately 30 feet lost ambulation within 1 year (15,22).

Ambulation past the age of 14 in a non-corticosteroid-treated patient should raise the suspicion of a milder form of muscular dystrophy such as BMD or limb girdle muscular dystrophy (LGMD). Ambulation beyond 16 years has been previously used as an exclusionary criterion for DMD in studies of BMD. Immobilization for any reason can lead to a marked and often precipitous decline in muscle power, rapid development of contractures, and loss of ambulatory ability. A fall with resultant fracture leading to immobilization and loss of ambulatory ability is not an uncommon occurrence. Ankle equinus contractures are the most common skeletal deformity. There is risk of osteopenia and fractures. There is also a comparative loss in height and increased weight gain in comparison to their normally functioning peer group.

- *Early nonambulatory (beginning at the age when a boy starts using a wheelchair full time)*: After boys can no longer walk, there is continued muscular deterioration throughout the upper and lower limbs, and skeletal deformities such as lower and upper limb contractures and spine deformity may become problematic. Powered mobility is required after loss of ambulation. Postural maintenance and sitting balance are initially intact and progressively lost. There is increasing loss of upper limb function (with decreasing ability to reach overhead, dress, self-feed, and perform other self-care). There is continued decline in pulmonary function with the ultimate need for mechanical cough assistance and progressive risk of nocturnal hypoventilation requiring noninvasive ventilation. Cardiomyopathy is evident by cardiac MRI in virtually all patients and in some patients by cardiac echo. After transition to a wheelchair, patients tend to put on more weight compared to their normally functioning peer group.
- *Late nonambulatory*: Postural support of the trunk and head support from a seating system are required as well as power recline. Upper extremity function is severely limited to distal fine motor function and tabletop activities. Maintaining computer access is a critical quality of life concern. Virtually all patients benefit from mechanical cough assistance and there is a high risk of nocturnal and daytime hypoventilation requiring noninvasive ventilation. Optimal nutritional management may require gastrostomy tube placement and enteral formula supplementation. There is risk for dysphagia and aspiration. Adequate phonation may become an issue late in the disease course. There may be a larger number of older DMD patients with unmet medical needs. As patients age, respiratory impairment and heart disease (heart failure and conduction abnormalities) are causes of morbidity and, eventually, mortality.

Both progressive limb weakness and decline in pulmonary function are due to skeletal myopathy; however, cardiac deterioration due to progressive cardiomyopathy may not be correlated with skeletal muscle deterioration. With increased life span due to effective ventilation interventions, cardiomyopathy has become a more common cause of death among patients with DMD. Cardiomyopathy in DMD does not usually manifest clinically until later phases of disease progression, but is likely present to some degree beginning at birth. However, the concept that cardiac disease develops only later along the spectrum of DMD progression does not appear to be the case—imaging data suggest a proportion of boys already show fibrosis in the heart at ages as young as 6.

The loss of clinical milestones is a hallmark of disease progression in DMD. Prior to and after the loss of ambulation, the difficulty performing functions and the loss of milestones, occur in a generally predictable descending order (although there may be some overlap or slight variation in some of the milestones).

Ambulatory functions and milestones (descending order):

- Unable to jump, hop, and run
- Gower's sign with standing
- Loss of standing from the floor
- Loss of transition from lying supine to sit
- Loss of stair climbing
- Loss of ability to stand from a chair
- Loss of ability to walk independently (defined by inability to perform 10-meter walk/run)
- Loss of standing in place

Nonambulatory milestones (descending order):

- Loss of ability to reach overhead
- Loss of ability to reach the scalp
- Loss of ability to self-feed without adaptations (hand to mouth)
- Loss of ability to place hands to tabletop
- Loss of ability to use a computer (distal hand function)

Pulmonary milestones are primarily measured by forced vital capacity (FVC), peak cough flow, and maximal static airway pressures (MIP and MEP)) indicating a need for interventions as outlined in the DMD Care Considerations (4,5):

- Less than 50% predicted FVC (cough assistance; monitoring required)
- Less than 40% predicted FVC (noninvasive ventilation should be a consideration)
- Less than 30% FVC (inability to sustain adequate overnight ventilation without support is likely)
- MEP less than 60 cm water (preoperative training in and postoperative use of manual and assisted cough techniques are necessary)
- Peak cough flow less than 160 L/min (manual and mechanically assisted cough techniques necessary)
- MEP less than 40 cm water (manual and mechanically assisted cough techniques necessary on a daily basis)

Cardiac milestones:

- Normal ejection fraction (afterload reduction with ACE inhibitors or angiotensin II receptor blockers (ARBs), recommended by some cardiologists)
- Less than 55% ejection fraction (most would agree that cardiac medications are indicated)

Cognition and Behavioral Phenotype in DMD. There is a dystrophin isoform present in the brain. Previous studies on intellectual function on children with DMD have generally revealed decreased IQ scores when these children are compared with both control and normative groups (15). A mean score for the DMD population of 1.0 to 1.5 standard deviation (SD) below population norms has been reported. There has generally been a considerable consistency in the degree of impairment across measures reflecting a rather mild global deficit. Some studies (24) have demonstrated relative deficits in verbal IQ. In a longitudinal assessment of cognitive function, McDonald and colleagues (15) found the IQ measure in DMD to become stable over time. On neuropsychological testing, a large proportion of DMD subjects fell within the "mildly impaired" or "impaired" range according to normative data (15). Again, there were no particular areas of strength or weakness identified. These findings likely reflect a mild global deficit rather than focal nervous system impairment (15). Hinton found that DMD is associated with poor attention to complex verbal information (more so than verbal or memory measures) and they exhibit decreased verbal span capacity, but not impaired recall (25,26). An increased incidence of autism spectrum disorder has been found in DMD (27). In one large series of DMD subjects, 11.7% were reported to have a comorbid diagnosis of attention deficit hyperactivity disorder, 3.1% had autism spectrum disorder, and 4.8% had obsessive-compulsive disorder (28). In addition, impaired facial affect recognition has been found to be a part of the phenotype associated with DMD (29).

Anthropometric Changes in DMD. Substantial anthropometric alterations have been described in DMD. Short stature and slow linear growth with onset shortly after birth have been reported (30). A set of growth curves, derived from a large cohort of male youth with DMD from the CDC MD STARnet, have been published. These curves demonstrate that DMD males are shorter and tend to the extremes of weight and body mass index compared with the general male pediatric population in the United States (31). Accurate measurement of linear height is extremely difficult in this population. Arm span measurements may be an alternative measure of linear growth; however, this measurement might also be difficult as elbow flexion contractures of greater than 30 degrees are frequently present in patients older than age 13. Ulnar length has been proposed as an alternative linear measurement in DMD patients with proximal upper extremity contractures and may be followed for those with wrist and finger contractures. Obesity is a substantial problem in DMD, subsequent to the loss of independent ambulation (15,32). Weight control during early adolescence has its primary rationale in ease of care, in particular ease of transfers during later adolescence.

Immediately following spine fusion there has been a documented tendency for DMD patients to lose significant weight. Those who lost weight were unable to self-feed. The weight loss after surgery was associated with

loss of self-feeding (33). There was no association with weight loss and loss of biceps strength. A correction of the kyphosis may actually make self-feeding problematic in DMD. A feeding evaluation and incorporation of kyphosis into the spinal instrumentation construct may help preserve self-feeding and prevent weight loss subsequent to spine fusion.

Longitudinal weight measurements in DMD confirm significant rates of weight loss in subjects aged 17 to 21 (15,34). This is likely caused by relative nutritional compromise during the later stages when boys with DMD have higher protein and energy intake requirements because of hypercatabolic protein metabolism. Protein and calorie requirements may often be 160% of that predicted for able-bodied populations during the later stages of DMD (35,36). Restrictive lung disease becomes more problematic during this time, and this may also influence caloric intake and requirements. Self-feeding often becomes impossible during this period because of significant biceps weakness. In addition, boys with DMD may develop signs and symptoms of upper gastrointestinal dysfunction (37).

Optimal Medical Management Affects the Course of DMD. Current medical management has changed the natural history in DMD affecting the timing of clinically meaningful milestones in individuals with access to high-quality care. This has largely been due to the use of glucocorticoids, management of spine deformity, pulmonary management, and cardiac management. The occurrence of contractures may impact mobility and upper limb function and efforts are made to prevent and manage contractures. Despite these interventions, the cardiomyopathy and pulmonary involvement in DMD still leads to a substantially shortened life span. In The Cooperative International Neuromuscular Research Group (CINRG) natural history cohort of 340 subjects followed prospectively from 2006 to 2011, death occurred in 5% of the cohorts (5%) and age at death ranged from 9.9 years to 29.5 years (38).

Pharmacologic treatment with corticosteroids. Treatment with prednisone or deflazacort (not approved by the Food and Drug Administration in the United States) helps maintain strength and prolongs ambulation by approximately 2 years (39–41). There does not appear to be an advantage of deflazacort over daily prednisone in terms of efficacy, but deflazacort may be associated with less weight gain and behavioral side effects. The optimal dose of prednisone is 0.75 mg/kg/day up to a maximum of 40 mg/day (39–41). The optimal dose of deflazacort appears to be 0.90 mg/kg/day (42). With both corticosteroid regimens the patients need to be monitored for cataracts, hypertension, weight gain, osteoporosis, growth retardation, diabetes, and behavioral side effects. These two daily regimens and an intermittent regimen of 10 days on and 10 days off are being studied in an international multicenter clinical trial. Recently, a high-dose weekend regimen of prednisone (5 mg/kg/day on two consecutive days each week) was shown to be equally efficacious in comparison to daily prednisone (43).

A Cochrane review has also concluded that glucocorticoid corticosteroids improve muscle strength and function over 6 months to 2 years (42). Improvements were seen in time taken to rise from the floor (Gower's maneuver time), 9-meter walking time, four-stair climbing time, the ability to lift weights, leg function grade, and FVC. In one natural history study, steroids have been shown to delay the loss of ambulatory milestones, prolonging ambulation by about 2 to 3 years over time and delayed losses in upper limb functioning — so that young men can continue to raise their hand to their mouths and feed themselves for a longer period of time (44). Steroids have also affected pulmonary function — young men treated with steroids reach an older age before requiring mechanical cough assistance or noninvasive ventilation as defined by FVC parameters outlined in the DMD care considerations.

However, as previously noted, glucocorticoid therapy also comes with substantial adverse events and are not used universally in practice. The use of corticosteroids for treatment of Duchenne and Becker muscular dystrophy in clinical practice from 1991 through 2005 was reviewed in a large population-based cohort (MD STARnet) of boys in four regional sites and six clinics of the United States (45). Corticosteroid use increased from 20% (11 of 56 individuals) in 1991 to 44% (93 of 218 individuals) in 2005. Average use varied by site and ranged from 15% to 49%. The median age of corticosteroid initiation was 6.9 years (range, 3.7–17.4 years). Dosage and growth information was available for 102 participants and showed a median dose as 0.729 mg/kg for prednisone and 0.831 mg/kg for deflazacort. The most common reasons that corticosteroids were discontinued included weight gain, behavioral side effects, and loss of ambulation, resulting in full-time wheelchair use. Substantial variations in clinical practice were identified among study sites.

Contracture management. Significant joint contractures have been found in nearly all DMD children older than 13 years (15,46,47). The most common contractures include ankle plantar flexion, knee flexion, hip flexion, iliotibial band, elbow flexion, and wrist flexion contractures (15). Significant contractures have been shown to be rare in DMD before age 9 for all joints. There is no association between muscle imbalance around a specific joint (defined as grade 1 or greater difference in flexor and extensor strength) and the frequency or severity of contractures involving the hip, knee, ankle, wrist, and elbow in DMD (15). Flexion contractures have been shown to be rare in those with extensor strength grade 3 or greater, about a joint. The presence of lower extremity contractures in DMD has been shown to be strongly related to the onset of wheelchair reliance (15).

Lower extremity contractures were rare while DMD subjects were still upright, but developed soon after they developed a sitting position in a wheelchair for most of the day. The occurrence of elbow flexion contractures also appears to be directly related to prolonged static positioning of the limb, and these contractures develop soon after wheelchair reliance. The relationship between wheelchair reliance and hip and knee flexion contractures has been noted (15). Mild contractures of the iliotibial bands, hip flexor muscles, and heel cords occur in most DMD patients by 6 years of age (47). Limitations of knee, elbow, and wrist extension occur about two years later (15,47); however, these early observed contractures were relatively mild. Given the tremendous replacement of muscle by fibrotic tissue in DMD subjects, it is not surprising that a muscle of less than antigravity extension strength, statically positioned in flexion, would develop a flexion contracture (subsequent to wheelchair reliance). The lack of lower extremity weight-bearing likely contributes to the rapid acceleration in the severity of these contractures after transition to a wheelchair. Ankle plantar flexion contractures are not likely a significant cause of wheelchair reliance, as few subjects exhibit plantar flexion contractures of 15 degrees or greater, before their transition to a wheelchair (15). Natural history data suggests that weakness is the major cause of loss of ambulation in DMD, not contracture formation.

Approaches to contracture prevention and management have been outlined in the CDC DMD care considerations (5), but the efficacy of these approaches has not been established. To the extent that contractures are directly related to antigravity strength (movement against gravity through a full range of motion [ROM]), and ambulatory capacity, any intervention that maintains strength, function, and upright mobility will likely result in decreased contractures. Although the evidence supporting the efficacy of multiple interventions to improve ROM in DMD and other NMDs in a sustained manner is lacking, there are generally accepted principles with regard to splinting, bracing, stretching, and surgery that help minimize the impact or disability from contractures (48).

Spine deformity management. The incidence of significant scoliosis requiring spinal arthrodesis has changed due to the use of glucocorticoids. In retrospective series, treatment of DMD with deflazacort and prednisone has been shown to reduce the occurrence of significant scoliosis (49–52).

In addition, timely spine surgery for curves greater than 30 to 40 degrees has impacted survival. The reported ultimate prevalence of scoliosis in DMD subjects not treated with corticosteroids has been reported to be from 33% to 100% (53). This marked variability is primarily because of retrospective selection for scoliosis, the inclusion or exclusion of functional curves, and dissimilar age groups. The prevalence of scoliosis is strongly related to age. Fifty percent of DMD patients acquire scoliosis between ages 12 and 15, corresponding to the adolescent growth spurt. Johnson and Yarnell (54) reported an association between the side of curvature, convexity, and hand dominance, an association that has been confirmed (55). Oda and colleagues (56) reported that the likelihood of severe progressive spinal deformity could be predicted by the type of curve and early pulmonary function measurements. Those with spines lacking significant kyphosis or hyperlordosis and a peak obtained absolute FVC greater than 2,000 mL tended not to show severe progressive scoliosis.

No cause-and-effect relationship has been established between onset of wheelchair reliance and occurrence of scoliosis (15,57). Wheelchair reliance and scoliosis have both been found to be age-related phenomena. The causal relationship between loss of ambulatory status and scoliosis is doubtful, given the substantial time interval between the two variables in most subjects (scoliosis usually develops after 3–4 years in a wheelchair). Both wheelchair reliance and spinal deformity may be significantly related to other factors (eg, age, adolescent growth spurt, increase in weakness of trunk musculature, and other unidentified factors) and thus represent coincidental signs of disease progression.

Pulmonary assessment and management. Pulmonary outcome measures can be divided into measures of strength, clinical measures of restrictive lung disease (which are predictive of the need for pulmonary interventions), and measures of cough function.

- *Measures of pulmonary strength (static airway pressures):* The MIP and the MEP are standard clinical measures that assess muscle strength. MIP is largely the function of diaphragmatic strength, where MEP is more reflective of the strength of the rectus abdominis and oblique muscles (and to a lesser extent the intercostals). These measures are relatively independent of chest wall compliance and lung function.
- *Clinical spirometry measures of restrictive lung disease:* FVC is a global measure of lung function and capacity. In DMD, its clinical utility derives from thresholds of diminished function, which dictate consideration of intervention.
- *Measures of cough function:* Pulmonary management in NMDs has largely been driven by findings in more common adult diseases. Measures of cough abilities such as the peak cough flow and peak expiratory flow rate have been used in these other contexts. There are positive data predicting that measuring peak flow is a potentially useful measure that correlates with the quality of life in other NMDs—although it may be difficult to use across all populations in DMD. It has been used in DMD, but without contemporary published natural history data, and thus could be considered an exploratory pulmonary biomarker to measure in DMD.

The American Thoracic Society practice parameter (58) includes recommendations for the management of DMD with airway clearance strategies or mechanical cough assistance and noninvasive ventilation. Survival has been most impacted by ventilation—two recent studies have reported that life spans in Duchenne can be lengthened substantially due to the implementation of noninvasive ventilation. Consequently, a larger number of young men with DMD are living into their twenties and thirties but often with significant disability. In DMD, absolute FVC volumes increase during the first decade and plateau during the early part of the second decade (15). A linear decline in the percentage of predicted FVC is apparent between 10 and 20 years of age in DMD (15). Rideau and colleagues (59) reported FVC to be predictive of the risk of rapid scoliosis progression. In the most severe DMD cases, maximal FVC reached a plateau of less than 1,200 mL. This was associated with the loss of ability to walk before age 10 and severe progressive scoliosis. Moderately severe DMD cases with respiratory compromise reached maximum FVCs between 1,200 mL and 1,700 mL. Spinal deformity was present consistently in these cases but varied in severity. The least severe DMD cases reached plateaus in FVC of greater than 1,700 mL. Similarly, McDonald and colleagues (15) found that those patients with a higher peak FVC (greater than 2,500 mL) had a milder disease progression, losing 4% predicted FVC per year. Those with peak predicted FVC less than 1,700 mL lost 9.6% predicted FVC per year. Thus, the peak obtained absolute values of FVC, usually in the early part of the second decade, are an important prognostic indicator for the severity of spinal deformity, as well as the ultimate severity of restrictive pulmonary compromise due to muscular weakness. Prednisone and deflazacort both appear to reduce the loss of pulmonary function over time during the second decade in DMD (44,49–51,57).

Maximal static airway pressures (both MIP and MEP) are the earliest indicators of restrictive pulmonary compromise in DMD with impaired values noted between 5 and 10 years of age. Vital capacity typically increases concomitantly with growth between 5 and 10 years of age with the percentage of predicted FVC remaining relatively stable and close to 100% of the predicted value. DMD patients typically show a linear decline in the percentage of predicted FVC between 10 and 20 years of age. An FVC falling below 35% is associated with increased perioperative morbidity in DMD (59,60) and optimally surgery should ideally be performed with a percentage of predicted FVC greater than 40%. Recent evidence suggests that spinal arthrodesis may be safely performed in a population of DMD with a percentage of predicted vital capacity less than 30% (61).

Ultimately, respiratory failure in DMD is insidious in its onset and results from a number of factors, including: (a) respiratory muscle weakness and fatigue; (b) alteration in respiratory system mechanics; and (c) impairment of the central control of respiration. Noninvasive forms of both positive and negative pressure ventilatory support are increasingly being offered to DMD patients nocturnally and continuously with an acceptable quality of life. Airway clearance strategies such as the cough assist/in–exsufflator, TheraVest, or intrapulmonary percussion ventilation (IPV) are also important pulmonary management strategies (62).

Management of cardiomyopathy. The dystrophin protein is present in both the myocardium and the cardiac Purkinje fibers of the conduction system. Early death is still commonly observed in individuals with DMD in the early teen years to early 20s, mostly due to heart problems. Abnormalities of the heart may be detected by clinical examination, electrocardiogram (ECG), echocardiography, cardiac MRI, and Holter monitoring. Cardiac examination is notable for the point of maximal impulse palpable at the left sternal border due to the marked reduction in anteroposterior chest dimension common in DMD. A loud pulmonic component of the second heart sound suggests pulmonary hypertension in patients with restrictive pulmonary compromise. Nearly all patients over the age of 13 demonstrate abnormalities of the ECG (15). Q-waves in the lateral leads are the first abnormalities to appear, followed by elevated ST-segments and poor R-wave progression, increased R/S ratio, and finally resting tachycardia and conduction defects. ECG abnormalities have been demonstrated to be predictive for death from cardiomyopathy with the major determinants including: R-wave in lead V1 less than 0.6 mV; R-wave in lead V5 less than 1.1 mV; R-wave in lead V6 less than 1.0 mV; abnormal T-waves in leads II, III, AVF, V5, and V6; cardiac conduction disturbances; premature ventricular contraction; and sinus tachycardia (63). Sinus tachycardia may be due to low stroke volume from progressive cardiomyopathy, or in some cases may be sudden in onset and labile, suggesting autonomic disturbance or direct involvement of the sinus node by the dystrophic process (64).

Autopsy studies and thallium-201 single photon emission computed tomography (SPECT) imaging have demonstrated left ventricular (LV) lateral and posterior wall defects that may explain the lateral Q-waves and the increased R/S ratio in V1 seen on ECG. Localized posterior wall fibrosis was found to be peculiar to DMD and was not found in other types of muscular dystrophy (65). Pulmonary hypertension leading to right ventricular enlargement also is known to affect prominent R-waves in V1 and has been demonstrated in patients with DMD (66).

Ventricular ectopy and sudden death are known complications of the cardiomyopathy in DMD and this association likely explains observed cases of sudden death. Severe ventricular ectopy in DMD has been associated with LV dysfunction and sudden death. Yanagisawa and colleagues (67) reported an age-related increase in the prevalence of cardiac arrhythmias detected by

ambulatory 24-hour electrocardiographic recordings. They also noted an association between ventricular arrhythmias and sudden death in DMD. Traditional echocardiography—measures of LV function (ejection fraction [EF], dimensions, volumes) typically remain relatively stable until early teenage years, and then begins to decline. Clinically evident cardiomyopathy is usually first noted after age 10 and is apparent in nearly all patients over age 18 (15,68). Development of cardiomyopathy is a predictor of poor prognosis. Echocardiography has been used extensively to follow the development of cardiomyopathy and predict prognosis in patients with DMD. The onset of systolic dysfunction noted by echocardiography is associated with a poor short-term prognosis (68). The myocardial impairment remains clinically silent until late in the course of the disease, possibly caused by the absence of exertional dyspnea, secondary to lack of physical activity. Death has been attributed to congestive heart failure in as many as 40% to 50% of patients with DMD by some investigators (68). Regular cardiac evaluations with an ECG, echocardiography, and Holter monitoring should be employed in teenagers with preclinical cardiomyopathy.

Cardiac measures in DMD are evolving. Historically, echocardiograms have been used to assess heart health in DMD—if the echocardiogram looked normal, it was assumed that the patient with DMD was not at great risk of cardiac events. Pediatric cardiologists have increasingly evaluated cardiomyopathies with cardiac MRI. In Duchenne, cardiac MRI has been used to show that damage to the heart begins quite early. Myocardial fibrosis may be observed in 17% of 6 to 7 year old boys with DMD (69). Measures of wall strain by cardiac MRI have emerged as earlier markers of cardiac dysfunction in boys with DMD. Declines in these markers can be detected earlier while left ventricular ejection fraction (LVEF) by echo is stable. Late gadolinium enhancement to detect areas of cardiac fibrosis has detected subclinical cardiomyopathy in young boys with DMD even earlier than changes in ventricular wall motion or function are detected.

Recent studies suggest that early presymptomatic treatment to achieve afterload reduction with ACE inhibitors such as perindopril or enalapril delayed the onset and progression of prominent LV dysfunction in children with DMD (70). In another series, 43% with impaired left ventricular systolic dysfunction (LVSD) responded to enalapril with the normalization of function (71). Alternatively, angiotensin II type 1 receptor blockers (ARBs) such as losartan may be considered for afterload reduction in DMD. Animal studies show that the ARB losartan attenuates transforming growth factor beta (TGF beta)-induced failure of muscle regeneration in dystrophinopathy presenting an additional potential for therapeutic benefit vis-à-vis the skeletal muscle in DMD (72). In addition, spironolactone, an aldosterone antagonist, when used in combination with an ACE has been shown to have beneficial effects on both cardiac and skeletal muscles in DMD animal models (73). There appears to be dual intracellular overload of sodium ions and water in DMD which precedes the dystrophic process and persists until fatty muscle degeneration is complete. Eplerenone, a modern spironolactone derivative, may decrease both cytoplasmic sodium and water overload and increase muscle strength and mobility in DMD (74). Finally, beta-blockers have been advocated by some but one study showed no incremental benefit of beta-blockers in addition to ACE inhibitors for delaying progression of cardiomyopathy (75).

Exercise in DMD. Despite concerns that exercise in DMD will produce contraction-induced injury and overwork weakness, there is no evidence that either aerobic exercise or moderate resistive exercise is harmful to patients with DMD. A recent randomized trial was conducted of assisted bicycle training in boys with DMD: the randomized controlled trial—the "no use is disuse" trial (76). The investigators examined whether assisted bicycle training is feasible, safe, and beneficial in DMD. Ambulatory (n = 18) and recently wheelchair-dependent (n = 12) boys with DMD were randomized to an intervention or a control group. The intervention group (n = 17) received assisted bicycle training of the legs and arms during 24 weeks. The control group (n = 13) received the same training after a waiting period of 24 weeks. The primary study outcomes were the Motor Function Measure (MFM) and the Assisted 6-Minute Cycling Test. After 24 weeks, the total MFM score remained stable in the intervention group, whereas it had significantly decreased in the control group. No serious adverse events were observed. Results suggest that assisted bicycle training of the legs and arms is feasible and safe for both ambulant and wheelchair-dependent children with DMD and may decline the deterioration due to disuse.

Novel Therapeutic Strategies in DMD. There are many promising novel therapeutic strategies in DMD, which has led to many recent and ongoing therapeutic trials. The strategies likely to be utilized in clinical trials in the near future are summarized below.

Dystrophin restoration. This therapeutic strategy represents a clear advance in treating the underlying cause of DMD with the goal being the restoration of low levels of dystrophin to transition patients from a DMD phenotype to a more slowly progressive phenotype similar to BMD. Thirteen percent of patients have a nonsense or premature stop codon mutation. Ataluren (marketed as Translarna in the European Union) binds to the ribosome and induces read-through of a remature stop codon mutation and low levels of dystrophin. In ambulatory DMD patients aged 5 and older, ataluren has been shown to improve disease progression as measured with the 6MWD and it also produced slowing of deterioration across timed function tests. Younger patients aged 5 to 6 actually showed trends toward improved strength (77).

There is a clear bell-shaped dose–response curve with ataluren. In 2014, the European Medicines Agency provided a conditional approval for Translarna representing the historic first approved treatment of the underlying cause of dystrophinopathy for a subpopulation of DMD patients. A multicenter phase 3 confirmatory trial of ataluren was nearing completion in late 2014. Exon skipping strategies utilize either antisense oligonucleotide (ASO) or phosphorodiamidate morpholino oligomer (PPMO) chemistries to bind to mRNA and induce exon skipping with restoration of the open reading frame of the dystrophin gene and a production of a shortened but functional dystrophin protein. Exon skipping strategies could theoretically treat up to 70% of all DMD patients and many drugs are in the therapeutic pipeline which target specific genetic constructs among subpopulations of DMD patients. Drisapersen, an antisense oligonucleotide which skips exon 51, has been shown to produce low levels of dystrophin in more functional patients with a higher baseline 6MWD (78). The AO chemistry gets into cells more readily producing some dose limiting toxicity of the subcutaneous formulation. Eight of ten ambulatory patients treated 3.4 years have demonstrated stable 6MWD. A phase 3 multicenter trial did not show efficacy of drisapersen at 48 weeks but longer term 2-year follow-up data is promising. Eteplirsen exhibits PPMO chemistry that also skips exon 51 and appears to have a more favorable safety profile, but higher more costly dosing of Eteplirsen is required to achieve dystrophin production. Results of a phase 2 study of weekly IV infusions of eteplirsen in 10 patients with a higher baseline 6MWD showed dystrophin production by 24 weeks, stabilization of 6MWD at 48 weeks (79), and subsequent stabilization of 6MWD for over 2 years. Two patients treated with eteplirsen with a lower baseline 6MWD of 260 and 330 meters went off their feet. A phase 3 confirmatory trial of eteplirsen was launched in late 2014.

Utrophin upregulation. In normal muscle cells, utrophin is located at the neuromuscular synapse and myotendinous junctions. The 900 kb gene for utrophin is found on the long arm of human chromosome 6. Utrophin was discovered due to its homology with dystrophin. Utrophin expression is dramatically increased in patients with DMD (and female carriers), both in those muscle fibers lacking dystrophin and in rare, revertant fibers that express dystrophin. Animal models show amelioration of dystrophinopathy if utrophin can be upregulated and there is much active effort to produce small molecules which upregulate utrophin for the treatment of DMD.

Phosphodiesterase type 5 (PDE5) inhibitors. Dystrophin deficiency causes loss of sarcolemmal nNOSμ and reduces paracrine signaling of muscle-derived nitric oxide (NO) to the microvasculature, which renders the diseased muscle fibers susceptible to functional muscle ischemia during exercise. DMD and BMD patients have demonstrated restoration of impaired sympatholysis with PDE5 inhibitors (80,81), and a phase 3 trial of tadalafil, a PDE5 inhibitor approved for the treatment of primary pulmonary hypertension in children is underway in DMD.

Myostatin inhibition. Myostatin is a secreted growth differentiation factor that is a member of the TGF beta protein family that inhibits muscle differentiation and growth in the process known as myogenesis. Myostatin is produced primarily in skeletal muscle cells, circulates in the blood and acts on muscle tissue, by binding a cell-bound receptor called the activin type II receptor. Blocking the activity of myostatin may have therapeutic application in treating muscle wasting diseases such as muscular dystrophy. Small molecules have been developed which antagonize myostatin including antibodies, soluble receptors, and blocking aptamers. The goal of this strategy is to increase functional lean muscle mass.

Follistatin stimulation. Follistatin induces muscle regeneration, increases with exercise, is antifibrotic blocking activin receptors in the TGF beta family, is anti-inflammatory, and inhibits myostatin. Current therapeutic approaches include recombinant follistatin administration and follistatin gene therapy.

Corticosteroid replacements. There is a great deal of effort under way to produce steroid analogs which maintain the therapeutic efficacy of prednisone and deflazacort but have improved side effect profiles. These include TGF-beta-blockers which decrease NFkB activation and ameliorate DMD pathology.

BECKER MUSCULAR DYSTROPHY. The existence of a form of muscular dystrophy with a similar pattern of muscle weakness seen in DMD, X-linked inheritance, but with later onset and a much slower rate of progression, was first described by Becker and Kiener in 1955 (52). The disorder has the same gene location as the DMD gene (Xp21) and is thus allelic. On immunostaining of muscle biopsy specimens, the presence of patchy abundance of dystrophin suggests a BMD phenotype. On Western blot for quantitative dystrophin analysis, either 20% to 80% dystrophin levels or normal quantity and reduced or increased molecular weight dystrophin is consistent with BMD. Studies show that 5% to 20% dystrophin quantity is consistent with an outlier or intermediate phenotype (2).

Epidemiology. BMD has a lower incidence than DMD, with prevalence rates for BMD ranging from 12 to 27 per million and a recent estimated overall prevalence of 24 per million (7). Median survival in BMD is 67 years compared to 27 years in DMD (based on data from TreatNMD Network).

Molecular Genetics and Diagnostic Evaluation. Full gene sequencing of the dystrophin gene which demonstrates large deletions, duplications, and point mutations, identifies 99% of patients with dystrophinopathy and is now the standard of care. This is essential for the identification

of patients with stop codons and specific gene alterations that will be targeted for molecular-based therapies. Not all DMD and BMD patients have deletion mutations: Many have point mutations that cannot be detected by screening deletion testing. Thus, full sequence analysis is necessary. About 55% of DMD patients and 70% of BMD patients show large deletion mutations of the gene. A positive DNA test result (presence of a point mutation, duplication, or deletion) is diagnostic of a dystrophinopathy (Duchenne or Becker dystrophy)—there are no false positives if the test is done appropriately. While genetic testing is improving with regard to the differentiation of DMD and BMD, there remains some overlap and variability. Differential diagnosis between DMD and BMD is best done by a consideration of clinical findings, family history of clinical phenotype, and muscle biopsy with quantitative dystrophin analysis. If the patient is still ambulating at 16 to 20 years of age and has a deletion mutation, then the correct diagnosis is BMD. Mutations at the Xp21 locus, which maintain the translational reading frame (in-frame mutations), result in an abnormal but partially functional dystrophin protein, whereas in DMD the mutations shift the reading frame (out-of-frame mutations) so that virtually no dystrophin is produced. The reading frame interpretation is most accurate for deletions at the center of the gene (exons 40–60) and is least accurate for deletions at the beginning of the gene (exons 1–20).

Absent dystrophin or levels less than 5% of normal generally are considered diagnostic of DMD; however, 5% of such patients have BMD phenotypes. In BMD, dystrophin typically has an abnormally small molecular weight (less than 427 kDa). A minority of patients have dystrophin of larger than normal molecular weight (greater than 427 kDa), or normal molecular weight. Most BMD patients with larger or smaller molecular weight dystrophin also have decreased quantities of the protein. All BMD patients with normal molecular weight dystrophin have decreased quantities, usually less than 30% normal. Smaller size dystrophin typically is caused by deletion mutations, and larger size dystrophin by duplication mutations. A further refinement is the use of antibodies specific to the carboxy-terminal (C-terminal) region of dystrophin. Using such antibodies, immunohistochemistry reveals that the C-terminal region is almost always absent in DMD but invariably present in BMD. Thus, when this region of the molecule is missing, a more severe phenotype is likely.

Age of Onset and Presenting Signs. Studies have shown significant overlap in the observed age of onset between DMD and BMD (16). Although determination of the quantity and molecular weight of dystrophin has substantially improved the early differentiation among BMD, "outlier" DMD, and the more common and rapidly progressive DMD phenotype, Bushby and colleagues (82) found no clear correlation between abundance of dystrophin and clinical course within the BMD group.

A series of Bushby and colleagues (83) which included 67 BMD subjects, supported the presence of two major patterns of progression in BMD—a "typical" slowly progressive course and a more "severe" and rapidly progressive course. All of the "severe" BMD cases showed difficulty climbing stairs by age 20, whereas none of the "typical" BMD cases had difficulty climbing stairs before age 20. Abnormal ECGs were seen in 27% of typical BMD subjects and 88% of severe subjects. Bushby and colleagues (83) found BMD subjects to have a mean age of onset of 12 years in the typical group and 7.7 years in the severe group. Some patients with BMD present with major muscle cramps as an isolated symptom (83). As in DMD, preclinical cases are often identified by the finding of a grossly elevated CK value. There is also considerable overlap in CK values between DMD and BMD cases at the time of presentation. Thus, CK values cannot be used to differentiate DMD from BMD.

Calf enlargement is a nonspecific finding in BMD, as is the presence of a Gower's sign. The gait over time is similar to other NMD conditions with proximal weakness. Patients often ambulate with a lumbar lordosis, forefoot floor contact, decreased stance phase knee flexion, and a Trendelenburg or gluteus medius lurch, often described as a waddle.

Other atypical clinical presentations include a sole complaint of cramps on exercise in individuals with no muscle weakness (83). In addition, patients with focal wasting of the quadriceps, previously diagnosed with quadriceps myopathy have been diagnosed with BMD, based on molecular genetic testing and/or dystrophin analysis on muscle biopsy (16).

Age of Transition to Wheelchair. The most useful clinical criterion to distinguish BMD from DMD is the continued ability of the patient to walk into late teenage years. Those with BMD will typically remain ambulatory beyond 16 years. Some patients may become wheelchair users in their late teens or 20s, whereas others may continue walking into their 40s, 50s, or later. DMD cases usually stop ambulating by 13 years unless treated with corticosteroids. Outlier DMD or intermediate dystrophinopathy cases generally stop ambulating between 13 and 16 years of age. The ages when siblings reach clinical milestones of disease vary widely between siblings. However, the time to ceased ambulation for older brothers predicts the time to ceased ambulation for their younger brothers (84).

Pattern and Progression of Weakness. BMD patients have distribution of weakness, similar to those with DMD (16). Proximal lower limb muscles are involved earlier in the disease course. Gradual involvement of the pectoral girdle and upper limb musculature occurs 10 to 20 years from the onset of disease. Extensors have been noted to be weaker than flexors (16). The muscle groups, which are most severely involved earlier in the course of disease include the hip extensors, knee extensors, and neck flexors (16).

Contractures. Early development of contractures does not appear to be a feature of BMD (16,83). As with BMD, nonambulatory BMD subjects may develop equinus contractures, knee flexion contractures, and hip flexion contractures. Because of the tremendous replacement of muscle in BMD subjects by fibrotic tissue, it is likely that, as in DMD, a muscle with less than antigravity extension strength, which is statically positioned in flexion, is more likely to develop a flexion contracture subsequent to wheelchair reliance.

Spine Deformity. Spinal deformity is not nearly as common or severe in BMD, as compared with DMD. Spinal instrumentation is rarely required by BMD patients (16,83).

Pulmonary Function. Compromised pulmonary function is much less problematic in BMD as opposed to DMD (16,56,83). The percentage of predicted FVC does not appear substantially reduced until the third to the fourth decade. The percentage of predicted MEP appears relatively more reduced at younger ages than the percentage of predicted MIP, a finding seen in DMD and other NMDs (15,85–87). This may be caused by more relative involvement of the intercostals and abdominal musculature with relative sparing of contractile function in the diaphragm of BMD. As in DMD and other NMDs, it appears that predicted MEP may be a useful quantitative measure of impairment and perhaps disease progression early in the course of BMD.

Cardiomyopathy. The pattern of occasional life-threatening cardiac involvement in otherwise mild and slowly progressive BMD has been reported by many (83,88). A significant percentage of BMD cases develop cardiac abnormalities and the rate of progression of cardiac failure may on occasion be more rapid than the progression of skeletal myopathy (83). In fact, cardiac transplantation has been successfully performed in BMD subjects with cardiac failure. Approximately 75% of BMD patients have been found to exhibit ECG abnormalities (16,83). The abnormal findings most typically reported include abnormal Q-waves, right ventricular hypertrophy, left ventricular hypertrophy, right bundle branch block, and nonspecific T-wave abnormalities. Unlike DMD, resting sinus tachycardia has not been a frequent finding. Echocardiography has shown LV dilation in 37%, whereas 63% have subnormal systolic function because of global hypokinesia (83). Thus, the cardiac compromise may be disproportionately severe, relative to the degree of restrictive lung disease in some BMD subjects. The evidence for significant myocardial involvement in BMD is sufficient to warrant screening of all of these patients at regular intervals using ECG and echocardiography. The slowly progressive nature of this dystrophic myopathy, which is compatible with many years of functional mobility and longevity, makes these patients suitable candidates for cardiac transplantation if end-stage cardiac failure occurs.

Some cases with BMD may present with an isolated cardiomyopathy with no clinical manifestation of skeletal muscle involvement. The diagnosis can be established by the demonstration of a deletion in the Xp21 gene or by muscle biopsy. Isolated cases of cardiomyopathy in children, particularly those with family histories indicative of X-linked inheritance, should be screened for BMD with an initial serum CK estimation and molecular genetic studies of the Xp21 gene.

Cognition. Cognitive testing in BMD subjects have shown large variability in IQ scores and neuropsychological test measures. Mildly reduced intellectual performance has been noted in a subset of BMD patients; however, the degree of impairment is not as severe as noted in DMD (16).

Limb Girdle Muscular Dystrophy (LGMD)

Before the advent of genetic testing, a group of disorders in patients commonly sharing a progressive pattern of proximal greater than distal muscular weakness with either autosomal-recessive (LGMD2) or -dominant (LGMD1) inheritance were termed *limb girdle muscular dystrophies.* Recent advances in molecular and genetic analyses have now identified a number of distinct genetic mutations in these patients. LGMD1 subtypes usually have later onset in adulthood. LGMD2 usually present during childhood or adolescence, although some may present in early adulthood. Many of the LGMD2 subtypes have been linked to gene defects causing abnormalities of the sarcolemmal associated proteins including sarcoglycans (alpha-SG, gamma-SG, beta-SG, and delta-SG), dystroglycans, Calpain-3, dysferlin, fukutin-related protein (FKRP), telethonin, and titin. The most common LGMD2 subtypes include sarcoglycanopathies, dysferlinopathies, calpainopathies, and FKRP deficiencies. The distribution and pattern of weakness at onset most often affects the pelvic or shoulder girdle musculature or both. The rate of progression is slower than DMD (87,89,90). Clinical features of the most common forms of LGMD2 are shown in Table 18.2.

SARCOGLYCANOPATHIES (LGMD 2C–2F). Disruption of the sarcolemmal membrane cytoskeleton is a common feature of sarcoglycanopathies. Most of the primary sarcoglycan abnormalities lead to secondary deficiencies of alpha-sarcoglycan. The CK levels are markedly elevated. A Duchenne-like phenotype in a female should raise a suspicion of a sarcoglycanopathy. Diagnosis of sarcoglycanopathies may be made with molecular genetic studies and immunohistochemical analysis of muscle biopsies. The age of onset of sarcoglycanopathies ranges from 2 to 15 years. Progression is variable with both more severe and milder phenotypes. Loss of ambulation may vary from 10 years to young adulthood. Weakness involves proximal greater than distal musculature. Calf pseudohypertrophy

TABLE 18.2 CHARACTERISTICS OF AUTOSOMAL-RECESSIVE LIMB GIRDLE MUSCULAR DYSTROPHIES (AR-LGMDS)

	LGMD 2A	LGMD 2B	LGMD 2C	LGMD 2D	LGMD 2E	LGMD 2F	LGMD 2I
U.S. prevalence	4,200	2,850	675	1,260	675	105	450
Inheritance	AR	AR	XR	XR	AR	AR	XR
Gene location	4p21	2p12-14	13q12.12	17q21	4q12	5q33	19q13.3
Protein	Calpain-3	Dysferlin	γ-Sarcoglycan	α-Sarcoglycan (Adhalin)	β-Sarcoglycan	δ-Sarcoglycan	Fukutin-related protein
Onset	Early < 12 years • Leyden–Mobius type: 13–29 years • Late: >30 years.	12–39 years Mean 19±3 years	Mean 5–6 years • C283Y mutation: <2 years.	2–15 years	3 years–teens • Intrafamilial variability	2–10 years	0.5–27 years • 61% less than 5 years
Severity and course	Variable • Mild phenotype in majority • Early onset has more severe progression	Slow progression • Mild weakness	Variable progression (some like DMD; others like BMD) • Death common in second decade	Variable • Absent adhalin: rapid progression • Reduced adhalin: Later onset, and milder weakness	Moderate progression and severity	Rapid progression • Death in second decade	Variable • Early onset: nonambulant by teens • Later onset: slowly progressive, and milder weakness
Ambulation status	• Loss of ambulation 10–30 years after onset	• Loss of ambulation: 10–30 years after onset • Most walk until their fourth decade	• Loss of ambulation: 10–37 years (mean 16 years)	• Early onset: loss of adhalin • Later onset: Reduced adhalin	• Often in wheelchair by 10–15 years; usually by 25 years	• Loss of ambulation: 9–16 years.	• 30% nonambulant by fourth to sixth decade
Weakness	• Scapula pelvic girdle and trunk weakness • Proximal legs > arms	• Weakness in gastrocnemius, quadriceps, and psoas • Weakness in biceps after legs	• Proximal > distal • Patchy distribution with some mutations • Quadriceps: spared	• Proximal > distal • Symmetric • Quadriceps weakness	• Proximal	• Proximal • Symmetric	• Proximal > distal • Legs: proximal • Arms: proximal • Face: mild weakness in older patients
Cardiac	• No involvement	• No involvement	Occasional; especially late in disease course	• Dilated cardiomyopathy	• Occasional cardiomyopathy	• Dilated cardiomyopathy described; may occur without myopathy	• Dilated cardiomyopathy in 30% to 50% of patients

Respiratory	• Rarely involved • PFTs rarely < 80% of normal	• Rarely involved	• Functional vital capacity ranges from normal to severe	• Functional vital capacity ranges from normal to severe	• Variable respiratory involvement	• Variable respiratory involvement	• Variable respiratory involvement; some severe
Muscle size	• Limbs, pelvic, and shoulder • Atrophy of posterior compartments	• Hypertrophy: uncommon	• Hypertrophy of calf and tongue in some patients	• Calf hypertrophy in some patients	• Prominent muscle hypertrophy	• Calf hypertrophy • Cramps	• Calf, tongue and thigh hypertrophy • Wasting in regions of weakness
Musculoskeletal	• Contractures: calf (toe walking may be presenting sign)	• Contractures: calf (toe walking may be presenting sign)	• Lumbar hyperlordosis • Scapular winging	• Scapular winging	• Shoulders: scapular winging and muscle wasting	• Scapular winging	• Contractures in ankles (especially in nonambulant) • Scoliosis
Central nervous system	• Intelligence: normal to mild mental retardation	• No intellectual defect reported	• No intellectual defect reported • Hearing loss in some	• No intellectual defect reported	• No intellectual defect reported	• No intellectual defect reported	• No intellectual defect reported
Muscle pathology	• Myopathic Necrosis and regeneration with fiber size variability Endomysial fibrosis • Type I predominance with increasing weakness • Normal dystrophin and sarcoglycan	• Myopathic • Necrosis and degeneration with variable fiber size ↑Endomysial connective tissue • Absent or ↓ dysferlin staining • Normal dystrophin and sarcoglycan	• Myopathic • Inflammation: occasional • Severe disease: absent γ-sarcoglycan • Slowly progressive: • Reduced γ-sarcoglycan • Dystrophin: normal or reduced	• Myopathic • Degeneration and regeneration • Variable fiber size ↑Endomysial connective tissue • Myopathic grouping of fibers • Absent or reduced adhalin α-sarcoglycan • Dystrophin: Normal or reduced	• Myopathic • Sarcoglycans: usually absent Dystrophin: often reduced, but not absent	• Myopathic • Fiber degeneration • Fiber regeneration • δ-Sarcoglycan absent • Other sarcoglycans absent or reduced	• Myopathic • Necrosis and degeneration • Variable fiber size • ↑ connective tissue • Type 1 fiber predominance • ↓ staining for sarcoglycans
Blood chemistry and hematology	CK: 7–80 times normal	CK: 10–72 times normal	CK: Very high	CK: Very high (often >5,000)	CK: Very high (often >5,000)	CK: 10 to 50 times normal	CK: Very high (1,000–8,000)

Abbreviation: AR-LGMD, autosomal recessive limb girdle muscular dystrophy.

scapular winging, progressive contractures, and scoliosis often occur (89). A dilated cardiomyopathy may occur particularly in alpha-SG and delta-SG. Intelligence is often normal.

DYSFERLINOPATHIES (LGMD 2B). Dysferlin is a skeletal muscle protein localized in the muscle cell membrane (91). It is involved in muscle contraction and contains C2 domains that play a role in calcium-mediated membrane fusion events, suggesting that it may be involved in membrane regeneration and repair. Specific mutations in this gene have been shown to cause autosomal-recessive limb girdle muscular dystrophy type 2B (LGMD2B) with proximal muscle involvement as well as Miyoshi myopathy which presents with distal weakness involving the distal legs including the gastrocnemius and soleus muscles (89). In LGMD 2B no specific genotype–phenotype correlations have been established. Patients show markedly elevated CK from 10 times normal to approximately 30,000. Onset is from age 10 to 39 years. Mild weakness occurs in a distal lower extremity and/or pelvic-femoral distribution. Weakness is usually mild early in the course of disease. Proximal leg weakness associated with exercise is common. Asymmetrical weakness is common. Distally the gastrocnemius weakness may cause the inability to toe walk and run. Proximally the glutei, quadriceps, and psoas are affected. The lower limbs are involved 9 to 10 years before upper limbs. The shoulder girdle and biceps are often affected with sparring of the deltoids. In the trunk the erector spinae are affected. The face is generally normal. There is atrophy of the pelvic and shoulder girdle muscles. Equinus contractures are common. There is no scapular winging. Loss of ambulation is typically after 30 years. Respiratory muscles are affected with longer disease durations. Cardiac function is spared. Intelligence is normal.

CALPAINOPATHIES (LGMD 2A). Heterogeneous dystrophies due to mutation of the Calpain-3 gene are termed calpainopathy (90). Calpain-3 is a nonlysosomal calcium-dependent proteinase specifically expressed in muscle. Muscle biopsies reveal that calpainopathy patients have normal dystrophin and sarcoglycan labeling, but lack Calpain-3. Some may present with asymptomatic hyperCKemia. Labs show normal to markedly elevated CK up to 11,000. Toe walking is often a presenting sign. The age of presentation is 6 to 18 in 70% of patients. An early-onset form occurs before 12 years of age and has the most severe progression. The "Leyden–Mobius" subtype has an onset between 13 and 30 years and weakness is in the pelvic-femoral girdle distribution. In a rare subset with Erb dystrophy type there is scapular-humeral weakness, the onset age is 16 to 37 years, and weakness presents early in the shoulder girdle. The Miyoshi muscular dystrophy phenotype may be caused by Calpain-3 deficiency and those individuals present with gastrocnemius weakness and wasting. Others with later onset have been reported. The general pattern of symmetrical weakness in Calpainopathy involves scapula, pelvic girdle and trunk weakness with normal facial strength. Milder distal weakness is common and lower extremity weakness is greater than upper extremity at the outset. Weakness of the rectus abdominis is common as is symmetric scapular winging. Lower extremity weakness affects the gluteus maximus, hip abductors, knee flexors, and ankle dorsiflexors while upper extremity weakness is seen in shoulder adduction; elbow flexion, and wrist extension. Hip adductors may be spared. The quadriceps, face, ocular and bulbar muscles are selectively spared. FVC declines over time but is rarely less than 80%. Progression is slow and patients demonstrate an inability to walk on heels, lumbar lordosis, and loss of walking typically occurs in the late second or third decade. Scapular winging is usually present from the early stages. The rate of deterioration varies between families. Wheelchair dependency typically occurs at 10 to 30 years after the onset of symptoms. The disease is predominantly symmetrical and atrophic, with prominent calves seen in only a minority of cases. Achilles tendon contractures may be an early sign, and spine deformity may also develop. Respiratory, but not cardiac, complications have been reported.

FUKUTIN RELATED PROTEIN—FKRP (LGMD 2I). This dystrophy is caused by pathogenic mutations in the gene for FKRP, which is involved in the glycosylation of cell surface molecules in muscle fibers (91). The majority of the LGMD2I patients carry a common C826A missense mutation in the FKRP gene. In the LGMD 2I patients, different mutations in the FKRP gene are associated with several secondary muscle protein reductions and the deficiencies of α2-laminin and α-DG on sections are prevalent, independent of the mutation type or the clinical severity. Onset ranges from 0.5 to 27 years with 61% less than 5 years. Onset may be at birth in patients with congenital muscular dystrophy syndrome (MDC1C), an allelic disorder. Weakness presents with waddling gait, difficulty with stairs, hypotonia, greater proximal than distal weakness of the legs and arms, and mild facial weakness in some older patients. Respiratory failure has been reported in 30%, occasionally occurring in the ambulant. The onset may be variable with intrafamilial variability described. Patients with early onset become nonambulant by teenage years. In those with later onset, they become nonambulant by the fourth to the sixth decade. Serum CK is elevated. Contractures are often at the ankles and more common in nonambulant patients. Scoliosis occurs in some. Cardiomyopathy is common in up to 80% and echocardiography may show a dilated left ventricle with mild LV failure. The cardiomyopathy shows no relation to skeletal muscle severity. Intelligence is preserved, although structural brain changes have been reported.

LAMIN A/C (LGMD 1B). Lamin A/C deficiency may present in those younger than 20 years with symmetric, proximal weakness of the lower limb, and slow progressive

weakness. The upper limbs are involved by the third or the fourth decade. There are typically no contractures. Cardiomyopathy has been reported in approximately 60% manifested by A-V conduction block and occasional dilated cardiomyopathy. Serum CK is normal to mildly elevated.

CAVEOLIN-3 (LGMD 1C). Onset may occur from 5 years to adulthood. Weakness is usually proximal and moderate in severity, There is difficulty walking, progression is slow, and some adults may present with a Gower's maneuver. Calf hypertrophy is often observed and a history of cramps after exercise is reported. The CK values are elevated 3 to 40 times. Variable clinical manifestations may occur in a single family. Caveolin-3 mutations have been linked to variant syndromes including hyperCKemia, rippling muscle disease, distal myopathies, dilated hypertrophic cardiomyopathy, and long QT syndrome.

Congenital Muscular Dystrophy

The term congenital muscular dystrophy (CMD) has been widely used for a group of infants presenting with hypotonia, muscle weakness at birth or within the first few months of life, congenital contractures, and immunohistochemical finding of dystrophic changes on muscle biopsy: muscle fiber necrosis and regeneration, increased endomysial connective tissue, and replacement of muscle with fat tissue. The early contractures may include equinovarus deformities, knee flexion contractures, hip flexion contractures, and tightness of the wrist flexors and long finger flexors. The contractures can become more severe over time with prolonged static positioning and lack of adequate passive ROM and splinting/positioning. Classical CMDs are clinically confined to the musculoskeletal system but other CMDs are characterized by significant cerebral neuronal migration defects and eye abnormalities. Classical CMDs are further subdivided according to the presence or absence of merosin (laminin-2) (92). An additional subgroup with collagen VI abnormalities has been identified and referred to as *Ullrich's congenital muscular dystrophy.*

MEROSIN-DEFICIENT CMD. This condition (CMD 1A) has been linked to chromosome 6q22 and accounts for around a half of classical CMD (92). These children show a consistently severe phenotype with multiple contractures and joint deformities (arthrogryposis) at birth. Weakness correlates with the level of residual merosin (laminin α2) protein. If there is absent laminin α2 protein weakness is severe, symmetric, proximal greater than distal and involves the facial muscles. Contractures are present at multiple joints. CK is mildly to moderately elevated. Infants may present with respiratory failure, but if adequately supported, they can be weaned off ventilatory support. A proportion will achieve independent sitting but independent standing or walking is almost never achieved if laminin α2 is severely reduced. Progressive spine deformity is common. The condition tends to remain relatively static, but some subjects may show slow progression. Mental development is usually normal although minor learning disabilities and seizures do occur. Brain MRI commonly shows diffuse white matter signal changes. Nerve conduction velocities are frequently slowed reflecting the ubiquitous expression of merosin in basement membranes. Merosin (laminin α2) is an extracellular glycoprotein that interacts with surface receptors on the sarcolemmal membrane of the muscle cell. The diagnosis of merosin-deficient CMD is dependent on the demonstration of absent merosin staining on muscle immunohistochemistry.

MEROSIN-POSITIVE CMD. This is generally a milder disorder than merosin-deficient CMD and the clinical phenotype is more heterogeneous. Intellectual function is normal and the brain MRI is normal. Most of these children present with weakness and hypotonia, and they achieve the ability to stand and walk independently by age 4. The course is static with little or no progression; however, contractures and scoliosis may develop. Respiratory failure is uncommon, as is cardiomyopathy.

FUKUYAMA CMD. These patients present in infancy with severe hypotonia, weakness and wasting of the face and limbs, occasional spasticity, large cheeks, contractures, kyphoscoliosis, microcephaly, seizures (50%), severe mental retardation (IQ 30–50), and occasionally progressive hydrocephalus. Muscle biopsy shows dystrophic changes. While rare in North America, the condition is common in Japan with an incidence approaching 40% of DMD (93). Brain malformations are frequently seen on MRI including polymicrogyria, pachygyria, and agyria. Frontal white matter lucencies are also evident on MR or computed tomography (CT) imaging. The gene loci have been identified to be at 9q3133.

MUSCLE–EYE–BRAIN DISEASE (MEB). This is a syndrome comprising CMD, marked mental retardation due to neuronal migration defects, and ocular abnormality. Infants present with congenital hypotonia, muscle weakness, elevated CK, myopathic electromyography (EMG), and dystrophic changes on muscle biopsy. Children with MEB are usually able to stand and ambulate. Severe visual impairment is present caused by severe myopia, retinal dysplasia, cataracts, and optic atrophy. Patients often deteriorate around 5 years of age with progressive occurrence of spasticity. CT scans have shown ventricular dilation and low density of the white matter. Death is usually in the first or the second decade but some individuals survive well into adulthood.

WALKER–WARBURG SYNDROME (WWS). This is a severe condition leading to blindness at birth and early death.

Infants present with CMD, mental retardation, and consistent central nervous system (CNS) abnormalities on imaging (type II lissencephaly, abnormally thick cortex, decreased interdigitations between white matter and cortex, and cerebellar malformation). Ocular abnormalities and cleft lip or palate may also be present. Muscle involvement is less prominent in WWS than other CMDs. Several gene abnormalities with autosomal-recessive inheritance have been linked to WWS including O-mannosyltransferase 1 (POMT1) linked to chromosome 9q34.1 and O-mannosyltransferase 2 (POMT2) linked to chromosome 14q24.3.

ULLRICH CONGENITAL MUSCULAR DYSTROPHY (SCLEROATONIC MUSCULAR DYSTROPHY). An emerging common group of CMD patients have a unique combination dystrophic changes on muscle biopsy in association with weakness, low tone, and selected early joint contractures, and other joints and skin demonstrating clinical laxity caused by a primary collagen VI abnormality (92). The term "collagen myopathy" is increasingly being utilized to describe these conditions. Three subunits of collagen VI have been found to be abnormal in these patients: collagen, type VI, subunit α1 (COL6A1) linked to chromosome 21q22.3; collagen, type VI, subunit α2 (COL6A2) also linked to chromosome 21q22.3; and collagen, type VI, subunit α3 (COL6A3) linked to chromosome 2q37. Inheritance for all three groups may be recessive or dominant. Clinical features are variable as some patients show severe weakness and some families with COL6A3 mutations have milder disease. Onset is often at birth with congenital proximal contractures and arthrogryposis caused by reduced fetal movements, hypotonia, and early hyperlaxity of distal joints (Figure 18.6). Knee contractures may limit walking in some. Spine rigidity and kyphoscoliosis have been noted. Torticollis may improve with increasing age. Weakness is diffuse and affects distal muscles greater than proximal and neck flexors. A minority of patients walk by age 1 to 2 years but the majority never walk. Respiratory insufficiency and hypoventilation may begin in the first decade and respiratory failure is not correlated with the degree of weakness. The course is slowly progressive. Death has been reported in the first or the second decade due to respiratory failure but many patients live to adulthood. The skin is soft, lax and a classic rash on the extensor surfaces of the upper arm and back can often be found described as "keratosis pilaris." Patients may also show keloids, atrophic scars, striae, and petechiae. There is no associated cardiomyopathy and intelligence is usually normal. Serum CK is normal to 10 times elevated and EMG is usually myopathic. Muscle biopsy and skin biopsy should be obtained to make the diagnosis. Muscle biopsy shows varied muscle fiber size, some very small muscle fibers, and an increase in endomysial connective tissue. Rare or occasional necrotic muscle fibers may be found. Collagen VI expression may be absent in skeletal muscle and capillaries or absent on the surface of muscle fibers but present in connective tissue. There has been no correlation between pattern of pathology and clinical phenotype.

FIGURE 18.6 Joint laxity in Ullrich congenital muscular dystrophy with collagen VI abnormality.

CONGENITAL MUSCULAR DYSTROPHY WITH EARLY SPINE RIGIDITY. This is a recessive condition caused by a defect in selenoprotein N, 1 (SEPN1) and linked to chromosome 1p35-p36. Clinical severity is variable with early-onset cases in infancy and later-onset cases in the later first decade. Patients present with hypotonia and poor head control. The weakness is symmetric and involves the neck, face, proximal and distal musculature.

Respiratory function is compromised with vital capacity below 55% by the end of the first decade. Patients often shows signs of nocturnal hypoventilation and central apnea. Respiratory failure may develop. Some patients never develop walking. Muscle size is small, especially in the inner thighs and calves. Many children show early improvement with development followed by nonprogressive or slow decline. The rigid spine develops by 3 to 7 years and is manifested by limited flexion of the neck and spine. Progressive scoliosis occurs with onset 4 to 12 years. Contractures of the elbow flexors; hip extensors, ankles, and knees are common. The rate of insulin resistance is increased and intelligence is normal. Serum CK is usually normal. The optimal muscle to biopsy can be best identified by MR imaging with involved muscles often being the vastus lateralis and biceps femoris. Clinically there is overlap with minicore congenital myopathy syndromes, and mutations in this SEPN1 gene also cause minicore congenital myopathy, congenital myopathy with desmin inclusions, and congenital fiber type size disproportion (small type I fibers).

Facioscapulohumeral Muscular Dystrophy (FSHD)

Facioscapulohumeral muscular dystrophy (FSHD) is a slowly progressive dystrophic myopathy with predominant involvement of facial and shoulder girdle musculature (94). There are two subtypes of FSHD: FSHD1 observed in 95% of FSHD patients and linked to the chromosome 4q35 locus, and FSHD2 genetically distinct with a more complex digenic (2 gene) inheritance pattern (chromosomes 10 and 18). Approximately 10% to 30% of FSHD cases are caused by sporadic mutations. To date, based on a relatively small study, FSHD1 and 2 appear to be clinically indistinguishable; however, larger studies are needed to confirm this observation.

FSHD is the third most common of the dystrophies, behind Duchenne and myotonic dystrophies with an incidence of between 10 and 20 per million live births (7). The age of presentation is generally before 20 years. Changes on muscle biopsy are relatively slight, with the most consistent finding being the presence of isolated small atrophic fibers. Other fibers may be hypertrophied. Serum CK levels are normal or slightly elevated in the majority of patients. Diagnosis is confirmed in over 90% of cases by molecular genetic testing.

Facial weakness is an important clinical feature of FSHD muscular dystrophy. The initial weakness affects the facial muscles, especially the orbicularis oculi, zygomaticus, and orbicularis oris. These patients often have difficulty with eye closure but not ptosis. An individual may assume an expressionless appearance and exhibit difficulty whistling, pursing the lips, drinking through a straw, or smiling (Figure 18.7). Even in the very early stages, forced closure of the eyelids can be easily overcome by the examiner. Masseter, temporalis, extraocular, and pharyngeal muscles characteristically are spared in FSHD.

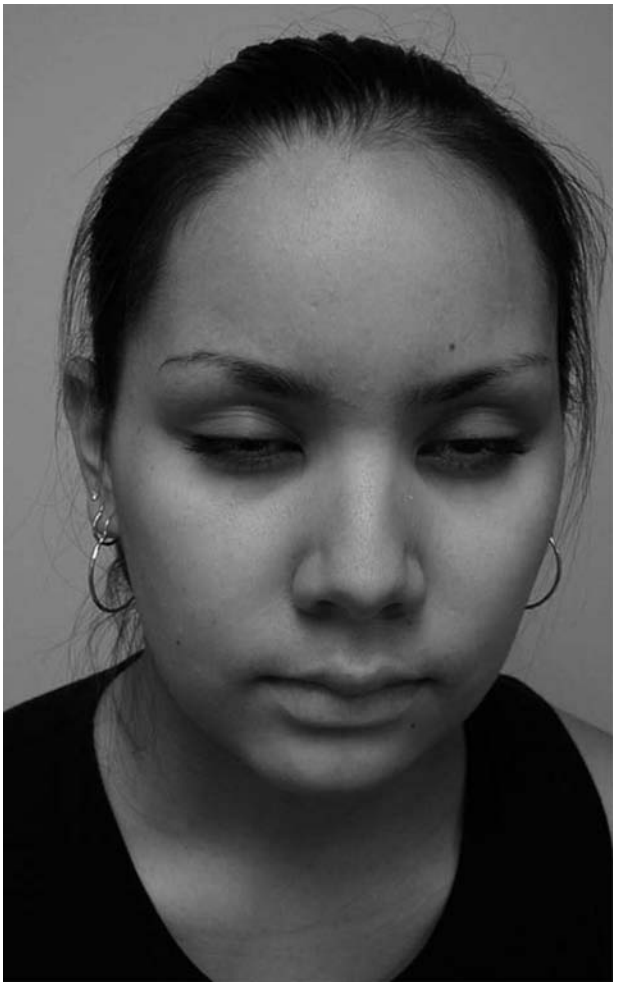


FIGURE 18.7 Facial weakness and expressionless facies in FSHD muscular dystrophy. Both father and daughter demonstrate difficulty whistling and pursing their lips.

Scapular stabilizers, shoulder abductors, and shoulder external rotators may be significantly affected, but at times the deltoids are surprisingly spared if tested with the scapulae stabilized. Both the biceps and triceps may be more affected than the deltoids. Patients with FSHD show characteristic patterns of muscle atrophy and scapular displacement. Involvement of the latissimus dorsi, lower trapezius, rhomboids, and serratus anterior results in a characteristic appearance of the shoulders, with the scapula positioned more laterally and superiorly, giving the shoulders a forward-sloped appearance (Figure 18.8). The upper border of the scapula rises into the trapezius, falsely giving it a hypertrophied appearance. From the posterior view, the medial border of the scapula may exhibit profound posterior and lateral winging. The involvement of shoulder girdle musculature may be quite asymmetric. Some authors have found asymmetric weakness in the dominant upper extremity (95).

A sensory neural hearing deficit was originally observed in Coates syndrome (early-onset FSHD). These individuals have a myopathy that presents in infancy. The disease progression is fairly rapid with most individuals becoming wheelchair reliant by the late second or third decade. These individuals also have a progressive exudative telangiectasia of the retina. Early recognition and photocoagulation of the abnormal retinal vessels may prevent loss of vision. Several audiometry studies have demonstrated hearing deficits in many later-onset FSHD patients in addition to those with Coates syndrome, suggesting that impaired hearing function is more common than expected in FSHD muscular dystrophy (96). Thus, all patients with FSHD should have screening audiometry and ophthalmologic evaluation. Contractures are relatively uncommon in FSHD muscular dystrophy. FSHD patients with scoliosis have mild and nonprogressive curves. Rarely, severe and progressive hyperlordosis is associated with FSHD. The patients with severe hyperlordosis may utilize their lordotic posturing to compensate for hip extensor weakness.

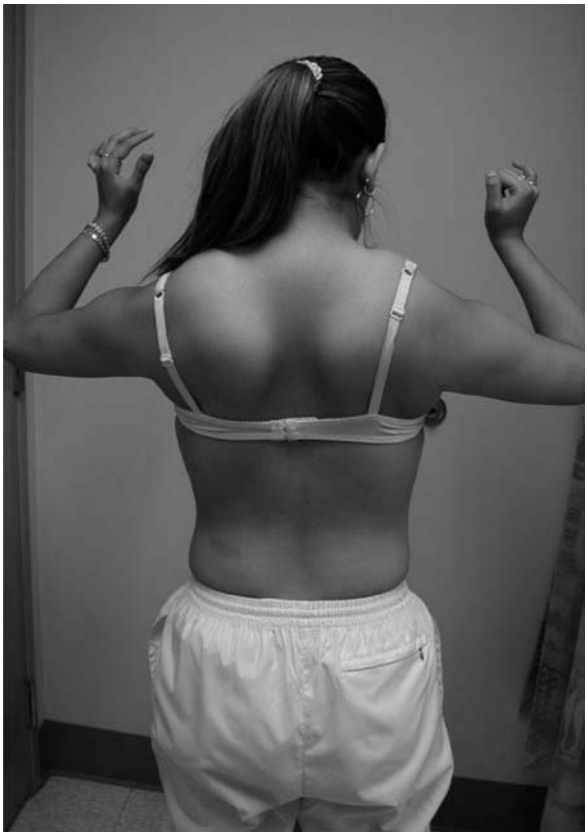


FIGURE 18.8 Posterior and lateral scapular winging and high riding scapula.

Mild restrictive lung disease has been reported in nearly one-half of FSHD patients (94). The expiratory muscles involved in respiration appear to be more affected than inspiratory muscles in FSHD (95). Patients rarely require nocturnal ventilatory support.

The presence of cardiac abnormalities in FSHD muscular dystrophy is debated. While diverse ECG abnormalities have been noted, one study showed no abnormalities on ECG, chest radiography, Holter monitoring, and echocardiography (97). Nuclear scanning with thallium-201 has demonstrated diffuse defects consistent with diffuse fibrosis (65). Abnormalities in systolic time intervals on echocardiography and elevations in atrial natriuretic peptide are consistent with subclinical cardiomyopathy. Cardiac complications in FSHD muscular dystrophy are rare and patients in general have normal longevity. There is usually no associated intellectual involvement in this dystrophic myopathy.

MOLECULAR GENETICS AND THERAPEUTIC TARGETS IN FSHD. The complex molecular genetics of FSHD is becoming increasingly understood (98). D4Z4 repeat contractions on chromosome 4 are consistently identified in FSHD1 patients, but not in persons with FSHD2. A DUX4 mRNA polyadenylation sequence (PAS) is necessary for FSHD, providing genetic proof that the DUX4 mRNA is necessary for FSHD. Normally DUX4 is repressed in unaffected individuals. Repression of DUX4 is lost in FSHD and resulted in a burst of DUX4 expression with either subsequent silencing or the death of the cell. Expression of DUX4 retrogene is harmful and causes FSHD pathogenesis. A *retrogene* is defined as a DNA gene copied back from RNA by reverse transcription. Normally there is repression of the DUX4 retrogene embedded in the D4Z4 repeat units. In FSHD1 there is a PAS region of D4Z4 distal to the final DUX4 retrogene which initiates transcripts in the antisense (opposite) direction. For FSHD1 to occur, (a) there needs to be a smaller D4Z4 repeat (1–10), (b) there needs to be inefficient epigenetic repression of the DUX4 retrogene (normally a silenced gene) in the D4Z4 repeat region, and the last repeat must be adjacent to a PAS in the subtelomeric region of chromosome 4. This results in abnormal expression of DUX4 protein (a transcription factor) in skeletal muscle nuclei in FSHD.

Within the control population, the size of the D4Z4 repeat array on chromosome 4 varies between 11 and 100 units, while the array on chromosome 10 can vary from 1 to 100 units. Most patients with FSHD1 have one array of 1 to 10 units on chromosome 4 (some with 11 reported). Symptomatic hearing loss and retinal vascular disease occur almost exclusively in FSHD individuals with only one to three residual D4Z4 repeats.

FSHD2 individuals have a classical FSHD phenotype and do not have a D4Z4 repeat array contraction, but do show a strong reduction of D4Z4 methylation suggesting that the common feature of FSHD1 and FSHD2 was decreased epigenetic repression of D4Z4 (increased DUX4 expression) (98). Subsequent whole exome sequencing in selected FSHD2 families identified mutations in the Structural Maintenance of Chromosomes Hinge Domain Containing 1 (SMCHD1) gene. Analysis of a larger cohort confirmed that SMCHD1 mutations account for approximately 85% of FSHD2 families. Thus, in FSHD2 there is digenic inheritance of a normal-sized D4Z4 repeat array on a DUX4 PAS containing chromosome 4 and a heterozygous SMCHD1 mutation on chromosome 18.

The pathogenesis of FSHD is becoming increasingly understood (98). Skeletal muscle cell apoptosis is perhaps the most dramatic consequence of DUX4 expression. The expression of DUX4 might also inhibit normal muscle regeneration. DUX4-induced misexpression of genes in FSHD muscle would be expected to induce an immune response in some tissues. RNAs and novel protein encoding transcripts could have biologic activity related to FSHD pathophysiology. Finally there may be modifier gene loci that protect the muscle cell. This has led to the following therapeutic targets for FSHD: (a) drugs that enhance the epigenetic repression of the D4Z4 region (silence the region), (b) oligonucleotide-based therapies that target the DUX4 mRNA and prevent it from making the DUX4 protein, or small molecules that might alter

RNA splicing or polyadenylation of a specific transcript, (c) drugs that block the activity of the DUX4 protein and DUX4 transcriptional activity, and (d) drugs that build lean muscle mass such as myostatin inhibitors.

Emery–Dreifuss Muscular Dystrophy (EMD)

Emery–Dreifuss muscular dystrophy (EMD) refers to a group of muscular dystrophies with weakness, contractures, and cardiac conduction abnormalities. Inheritance pattern is variable among subtypes.

EMERY–DREIFUSS 1 (EMD1). EMD1 is an X-linked recessive progressive dystrophic myopathy due to an abnormality of the protein "emerin" with a gene locus identified at Xq28 (99,100). The protein is associated with the subcellular nucleus and cytoplasm membranes and is found in muscle, nerve, mucosal epithelium, skin, and cardiac tissue. Patients usually present in teenage years but the age of presentation may vary from the neonatal period with hypotonia to the third decade. Early elbow flexion contractures are a hallmark of the disease. Severe contractures including elbow flexion, ankle equinus, rigid spine, and neck extension contractures are often more limiting than weakness which begins in a scapulohumeral peroneal distribution. The biceps and triceps show wasting and weakness and the deltoids and forearms are often more spared. The calf frequently shows wasting. Ankle dorsiflexors often are weaker than ankle plantar flexors leading to the equinus contractures. Scapular winging is frequent. Tightness of the cervical and lumbar spinal extensor muscles, resulting in limitation of neck and trunk flexion, with inability to flex the chin to the sternum and to touch the toes, also has been reported in EMD. The face is either spared or affected late. Functional difficulties are experienced walking or climbing stairs. Progression is slow and loss of ambulation is rare. Some cases with EMD1 may show evidence of nocturnal hypoventilation, as a result of restrictive expansion of the chest in association with the rigid spine, and partly due to involvement of the diaphragm.

Progressive cardiac disease is almost invariably present with onset in the early second decade to the forties. Arrhythmia may lead to emboli or sudden death in early adult life. The cardiomyopathy may progress to LV myocardial dysfunction or four-chamber dilated cardiomyopathy due to fibrosis with complete heart block and ventricular arrhythmias (101). Initially, atrial arrhythmia usually appears prior to complete heart block. Reported features include first-degree heart block, followed by the Wenckebach phenomenon and then complete atrial ventricular dissociation and atrial fibrillation or flutter with progressive slowing of the heart rate (101). Frank syncope may develop in the late second and early third decade and patients often require a cardiac pacemaker by age 30 with an indication being bradycardia with a heart rate below 50. EKG changes include slow heart rate, absent or small P-waves, arteriovenous (AV) block, and atrial fibrillation/flutter. Evidence of cardiac arrhythmia, sometimes only present at night, may be detected on 24-hour Holter monitoring. A significant percentage of female carriers have conduction defects and arrhythmias, so they warrant monitoring with annual EKGs.

Laboratory evaluation is usually with molecular genetic studies and/or muscle biopsy. Serum CK is mildly elevated to less than 10 times the normal value and levels decrease with age. MRI of posterior calf shows the soleus to be involved and the gastrocnemius to be relatively spared. Muscle biopsy shows variable muscle fiber size, endomysial fibrosis, inflammation, Type I fiber atrophy, Type I or II predominance, and nuclear membrane pathology on electron microscopy. Other muscle changes in some patients include rimmed vacuoles as seen in inclusion body myositis ("IBM-like"). Immunohistochemistry reveals Emerin loss in muscle in greater than 95% of patients.

EMERY–DREIFUSS MUSCULAR DYSTROPHY 2. EMD2 is due to a lamin A/C protein abnormality and it has been linked to chromosome 1q21.2. Inheritance may be dominant or recessive and lamin A/C mutations may be either frameshift or missense (99). Those with missense mutations have childhood onset with a mean age of onset of 2.4 years. Weakness is in a scapuloperoneal and facial distribution. Patients demonstrate paravertebral weakness or rigidity, and tendon contractures are common. Those with frameshift mutations producing a truncated protein have adult onset with a mean age of 30.5 years, and cardiomyopathy is more frequent than weakness (99). Contractures are rare and weakness is in a limb girdle distribution. The disorder is allelic with autosomal-dominant LGMD 1B.

CONGENITAL MYOPATHIES

The term *congenital myopathy* is used to describe a group of heterogeneous disorders usually presenting with infantile hypotonia due to genetic defects causing primary myopathies with the absence of any structural abnormality of the CNS or peripheral nerves and a specific diagnosis of each entity is made on the basis of specific histologic and electron microscopic changes found on muscle biopsy. While patients may be hypotonic during early infancy, they later develop muscle weakness that is generally nonprogressive and static. The weakness is predominantly proximal, symmetric, and in a limb girdle distribution.

The serum CK values are frequently normal and the EMG may be normal or may show mild, nonspecific changes, usually of a myopathic character (small amplitude polyphasic potentials). The only congenital myopathy consistently associated with spontaneous activity is

myotubular (centronuclear) myopathy. In this disorder, the EMG reveals myopathic motor unit action potentials with frequent complex repetitive discharges and diffuse fibrillation potentials. These myopathies may be considered primarily structural in nature and thus patients do not actively lose muscle fibers, as is the case in dystrophic myopathies.

Central Core Myopathy

This is an autosomal-dominant disorder with gene locus at 19q13.1, the same gene locus as the malignant hyperthermia gene (ryanodine receptor gene, RYR1). Indeed, these patients have a high incidence of malignant hyperthermia with inhalational anesthetic agents. Histologically, the muscle fibers have amorphous-looking central areas within the muscle that may be devoid of enzyme activity. There are densely packed disorganized myofibrils ("cores") at the center of the majority of type 1 fibers. Electron microscopy shows the virtual absence of mitochondria and sarcoplasmic reticulum in the core region, reduced muscle enzymes (cytochrome oxidase, NADH), a marked reduction in the interfibrillary space and an irregular zigzag pattern (streaming) of the Z-lines. This gives rise to the characteristic central pallor. There is a predominance of high oxidative, low glycolytic type I fibers and a relative paucity of type II fibers resulting in a relative deficiency of glycolytic enzymes.

Clinically, patients generally demonstrate mild and relatively nonprogressive muscle weakness, either proximal or generalized, and areflexia which presents in either early infancy or later. There may be mild facial weakness, but normal extraocular movements. Patients often achieve gross motor milestones, such as walking, rather late and they continue having difficulty going upstairs. Proximal limb weakness is typical and patients may show a Gower's sign. The disorder remains fairly static over the years. There may be a frequent occurrence of congenital dislocation of the hip, kyphoscoliosis, and pes cavus. The condition is largely nonprogressive with affected children remaining ambulatory into adult life. One-third show anesthesia-related malignant hyperthermia. Central core myopathy and familial malignant hyperthermia appear to be allelic as the ryanodine receptor chain implicated in malignant hyperthermia has the same locus. Individuals within the same family can exhibit one or both phenotypes.

Nemaline Myopathy

Nemaline myopathy, also referred to as rod body myopathy, represents a varied group of disorders with different modes of inheritance, but the most typical form is autosomal recessive. While the rods may be easily overlooked on routine H&E staining, they can be rarely demonstrated with the Gomori trichrome stain. The rods are readily demonstrated on electron microscopy. They are thought to be an abnormal depositions of Z-band material of a protein nature and possibly alpha-actinin. The disease has been linked to at least seven distinct genes. The severe congenital form has been linked to α-Actin, Nebulin, and Troponin T1 mutations. A milder childhood form has been linked to α-Actin, Nebulin, α-tropomyosin 3 (TPM3), and β-tropomyosin (TPM2) mutations.

A severe form of the disease may present in the neonatal period with very severe weakness, respiratory insufficiency, and often a fatal outcome. Most cases present with a mild, nonprogressive myopathy with hypotonia and proximal weakness. In more severe cases, swallowing difficulty may be present in the neonatal period. Skeletal abnormalities, such as kyphoscoliosis, pigeon chest, pes cavus feet, high-arched palate, tent-shaped mouth, and an unusually long face has been noted. Cardiomyopathy has been described in both severe neonatal and milder forms of the disease. Autosomal-dominant inheritance has been described in a few instances.

Centronuclear (Myotubular) Myopathy (Non X-Linked)

Patients with non-X-linked myotubular myopathy have muscle biopsies which show a striking resemblance to the myotubes of fetal muscle. Patients typically present with early hypotonia, delay in motor milestones, generalized weakness of both proximal and distal musculature, and ptosis with weakness of the external ocular muscles as well as weakness of the axial musculature. The author has seen severe cardiomyopathy in an adult female with documented autosomal-dominant inheritance. Nocturnal hypoventilation has been described.

Several gene loci with autosomal-dominant inheritance have been identified in centronuclear congenital myopathy including Dynamin 2 (DNM2) linked to chromosome 19p13.2, and MYF6 linked to chromosome 12q21.

Severe X-Linked Centronuclear (Myotubular) Myopathy

Cases with neonatal onset and severe respiratory insufficiency have been identified with an X-linked recessive mode of inheritance. The gene for this disorder codes for Myotubularin (MTM1), and has been linked to chromosome Xq27.3-q28. Muscle biopsy shows characteristic fetal-appearing myotubes with rows of centrally placed internal nuclei.

Patients present with severe generalized hypotonia, associated muscle weakness, swallowing difficulty, and respiratory insufficiency. They often become ventilator-dependent at birth. If they are able to be weaned from the ventilator, subsequent death due to pulmonary complications is not uncommon. The mean age of death is 5 months but some children survive for many years with mechanical ventilation. Aspiration pneumonias are common. Additional clinical features include congenital contractures,

facial weakness with an elongated expressionless face, tent-shaped mouth, high-arched palate, weakness of the external ocular muscles, and long digits. Progressive kyphoscoliosis is common. Systemic features in some survivors above 1 year of age include pyloric stenosis, spherocytosis, gallstones, renal stones or calcinosis, a vitamin K responsive bleeding diathesis, rapid linear skeletal growth, advanced bone age, and hepatic dysfunction. EMG shows many fibrillations and positive sharp waves.

Minicore Disease (Multicore Disease)

This is a relatively rare congenital myopathy with muscle biopsies showing multiple small randomly distributed areas in the muscle with focal decrease in mitochondrial oxidative enzyme activity and focal myofibrillar degenerative change. Characteristic changes are present on electron microscopy. There is a predominance of type I fiber involvement.

Clinically, patients present with hypotonia, delays in gross motor development and nonprogressive symmetric weakness of the trunk and proximal limb musculature. There may be mild facial weakness, ptosis, and ophthalmoplegia. There is also associated diaphragmatic weakness, placing patients at risk for nocturnal hypoventilation. Subtle ultrastructural changes allow this condition to be distinguished from central core disease. The cores are smaller in size (minicores) and not confined to the center of the fiber. Inheritance is usually autosomal recessive, and two genes, the Ryanodine receptor gene (RYR1) linked to chromosome 19q13.1 and the SEPN1 gene linked to chromosome 1p35-p36, account for 50% of cases.

Congenital Fiber-Type Size Disproportion

Congenital fiber-type size disproportion represents a heterogeneous group of conditions most likely with varied genetic defects. The condition was initially delineated by Brooke (102) on the basis of the muscle biopsy picture demonstrating type I fibers which are smaller than type II fibers by a margin of more than 12% of the diameter of the type II fibers. The mean reduction in fiber diameter is 41% and ranges up to 78%. A number of disorders such as congenital myopathies (nemaline rod, centronuclear, and multiminicore), EMD and myotonic dystrophy 1, rigid spine syndromes, congenital muscular dystrophy (SEPN1), LGMD 2A, and severe spinal muscular atrophy (SMA) all may show small type I fibers and should be excluded. The diagnosis of congenital fiber-type disproportion should be made only in the presence of normal-sized or enlarged type II fibers and not in cases where both type I and type II fibers were small. Serum CK has been normal to less than three times the upper limit of the normal value.

Patients typically present with infantile hypotonia and delay in gross motor milestones. The severity has been noted to be quite variable, but it is generally nonprogressive or improves with time. Limb weakness of variable severity may be diffuse or affect proximal muscles. Deep tendon reflexes (DTRs) are reduced. Ophthalmoplegia, facial weakness, and bulbar weakness are rare findings but associated with more severe cases. Intelligence is normal. There is generally short stature and low weight. Patients may exhibit a long narrow face, high-arched palate, and deformities of the feet, including either flat feet or occasionally high-arched feet. Kyphoscoliosis has been reported. Lenard and Goebel (103) documented a case with fairly severe weakness and associated respiratory deficit, necessitating tracheostomy. The author has managed two cases (a mother and son with presumed autosomal-dominant inheritance), who both developed nocturnal hypoventilation requiring bilevel positive airway pressure (BIPAP).

Patients with muscle biopsies indicative of congenital fiber-type disproportion and ptosis should be evaluated for a congenital myasthenic syndrome as the author has seen a number of cases in recent years of congenital structural NMJ disorders which have associated nonspecific changes on muscle biopsy, interpreted to be congenital fiber-type disproportion. This is an important distinction, as some of these patients with congenital myasthenia respond to pharmacologic intervention.

The mode of inheritance for congenital fiber-type disproportion is varied with both autosomal-recessive and autosomal-dominant patterns of inheritance reported.

MYOTONIC DISORDERS

Myotonic Muscular Dystrophy 1 (DM1)

Myotonic muscular dystrophy 1 (DM1) is an autosomal-dominant multisystem muscular dystrophy with an incidence of 1 per 7,500 to 8,000 (7,104). It represents the most common inherited NMD of adults and may be the most common disorder of skeletal muscle. The disorder affects multiple systems including skeletal muscle, smooth muscle, myocardium, brain, and ocular structures. Associated findings include baldness and gonadal atrophy (in males), cataracts, and cardiac dysrhythmias. Insulin insensitivity may be present. DM1 is caused by expansion of a cytosine-thymine-guanine (CTG) triplet repeat in the 3′ noncoding region of myotonin protein kinase, the gene encoding the DM protein kinase (DMPK). The gene has been localized to chromosome 19q13.3. Patients demonstrate expansion of an unstable CTG trinucleotide repeat within the region. Molecular genetic testing is available for diagnosis. Normal individuals generally have less than 37 repeats which are transmitted from generation to generation. DM1 patients may have 50 to several thousand CTG repeats with remarkable instability. The age of onset is inversely correlated by the repeat links (105). Mild, late onset DM1 usually is associated with 50 to 150 repeats, classic adolescent or young adult onset

(A) (B)

FIGURE 18.9 (A) Typical facial characteristics in myotonic muscular dystrophy 1 (DM1) and congenital DM1. The symptomatic mother has 660 trinucleotide CTG repeats at the DM protein kinase (DMPK) gene loci in chromosome 19q13.3, while the child has 1,560 repeats. (B) Temporal wasting and myopathic facies with tent-shaped mouth in children with DM1.

DM1 shows 100 to 1,000 repeats and congenital DM1 patients show greater than 1,000 repeats. The expanded CTG repeat further expands as it is transmitted to successive generations, providing a molecular basis for genetic anticipation. Both maternal to child and paternal to child transmission occurs. Repeat size in offspring exceeding 1,000 CTG repeats is generally seen in maternal rather than paternal transmission. Affected fathers seldom transmit alleles larger than 1,000 copies to offspring owing to a lack of sperm containing such alleles.

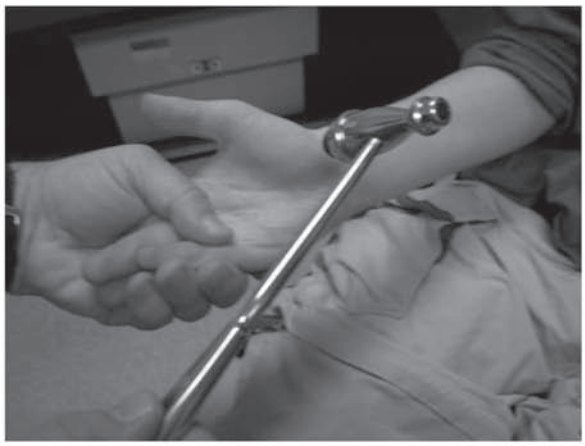
Several characteristic facial features of DM1 may be noted on inspection (Figure 18.9). The adult with long-standing DM1 often has characteristic facial features. The long thin face with temporal and masseter wasting is drawn and has been described by some as "lugubrious." Adult males often exhibit frontal balding. Children with congenital myotonic muscular dystrophy often exhibit a triangular or "tent-shaped mouth" (Figure 18.9).

Myotonia. Myotonia is a state of delayed relaxation or sustained contraction of skeletal muscle is easily identified in school-age children, adolescents, and adults with DM1. Grip myotonia may be demonstrated by delayed opening of the hand with difficult extension of the fingers following tight grip. Percussion myotonia may be elicited by percussion of thenar eminence with a reflex hammer, giving an adduction and flexion of the thumb with slow return (Figure 18.10). Symptomatic myotonia may be treated with agents such as mexiletine, or membrane stabilizers such as carbamazepine or Dilantin, which have been shown to impact the symptoms; however, patients treated have shown little functional gain (106). A randomized placebo-controlled crossover study of mexiletine showed reduction of grip myotonia by up to 50% in DM1 patients after 7 weeks of treatment (107).

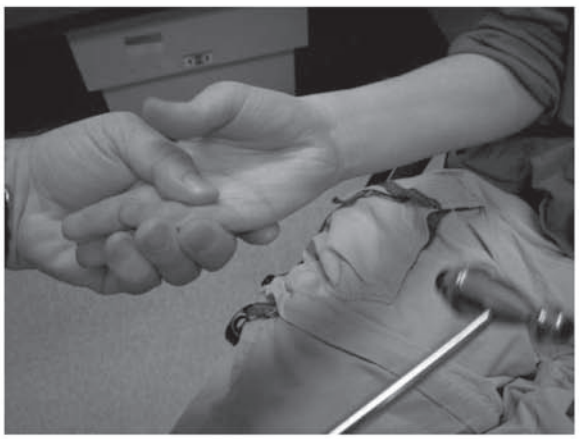
DM1 is one of the few dystrophic myopathies with greater distal weakness than proximal weakness, although neck flexors, shoulder girdle musculature, and pelvic girdle musculature may become significantly involved over decades, weakness initially is often most predominant in the ankle dorsiflexors, ankle everters and inverters, and hand muscles (108). As with other dystrophic myopathies, significant muscle wasting may occur over time. In DM1 patients with infantile onset, a congenital club foot or talipes equinovarus is a fairly common deformity (Figure 18.11). In patients with noncongenital DM1, contractures at the wrist, ankle, and elbows are relatively uncommon and mild (108). Patients with congenital onset DM1 may develop spinal deformity requiring surgical spinal arthrodesis (108).

Hypersomnolence. Excessive daytime sleepiness is commonly seen in DM1 (109) and it is thought to be related to the loss of serotonergic neurons in the dorsal raphe and superior central nucleus of the brainstem. Treatment of the hypersomnolence with modafinil has been helpful.

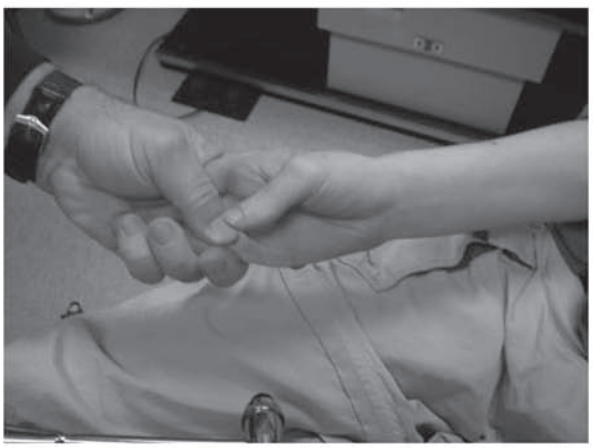
Cardiac involvement. The cardiac impact of DM1 falls mainly on the conduction system. Abnormalities on ECG and echocardiography are demonstrated in approximately 70% to 75% of patients (110). Prolongation of the PR interval, abnormal axis, and infranodal conduction abnormalities are all suggestive of conduction system disease which may explain the occurrence of sudden death which is a risk in adult DM1 patients (111). Ventricular tachycardia may also contribute to the syncope and sudden death associated with DM1. Cardiac dysrhythmia,



(A)



(B)



(C)

FIGURE 18.10 A–C, Percussion myotonia of the thenar eminence.

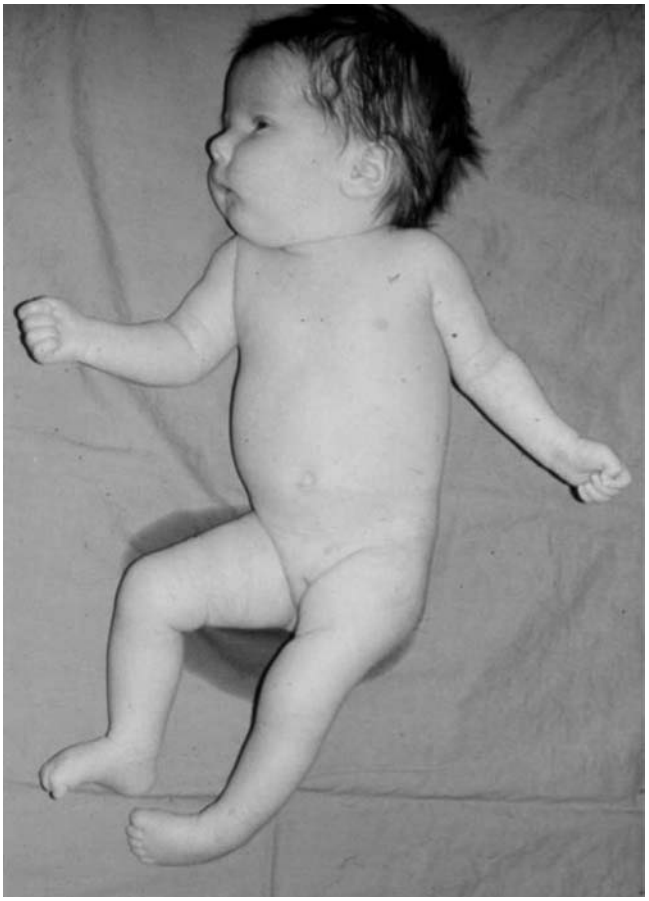


FIGURE 18.11 Talipes equinus in congenital myotonic muscular dystrophy 1 (DM1).

particularly heart block, is the second leading cause of death after respiratory failure (112). In a prospective study, the risk of sudden death in a clinic population was 1.1% per year (113). Sixty-five percent of patients show prolongation of the PR interval or QRS duration. The conduction defects are progressive and may lead to severe bradycardia or asystole due to atrioventricular block. Atrial tachycardias (flutter, fibrillation, or sinus tachycardia) are relatively common, and risk of ventricular tachycardia is also elevated. Although the cardiac contractility is relatively preserved, heart failure may occur at later ages. Ten percent of patients in a large study had clinical or echocardiographic evidence of LVSD (113). LVSD was rare before 40 years of age, but after this age the frequency steadily increased, reaching a high of 30% by the age of 70 years. Some patients have required implantation of cardiac pacemakers. Q-waves have been reported on screening ECGs in DM1 patients and this abnormality may reflect myocardial fibrosis (111,114). Occasionally teenagers may present with atrial arrhythmias. Any DM1 patient with dyspnea, chest pain, syncope, or other cardiac symptoms should receive thorough cardiac evaluation. Care should be taken during general anesthesia in DM1 due to risk of cardiac arrhythmias and malignant hyperthermia.

Ocular involvement. Cataracts before the age of 55 years, or family history of premature cataracts, suggest the diagnosis of DM1 or DM2 in patients with muscle symptoms. By direct ophthalmoscopy, the cataracts of DM are nonspecific and appear as punctate opacities. By

slit lamp examination, they have a multicolored iridescent appearance and are located in the posterior lens capsule, findings that are highly suggestive of DM1 or DM2. Premature cataracts may also occur in mitochondrial, centronuclear, or myofibrillar (aB-crystallin) myopathies.

CNS involvement. Congenital DM1 show severe cognitive impairment and mental retardation is common. In noncongenital DM1, there is evidence for a generally lower intelligence of a mild degree (full-scale IQs have been reported in the 86–92 range) (110). There is a wide range of IQ values found in this population with many subjects scoring in the above-average range. Cognitive functioning also appears to be related to the size of the CTG expansion at the DM1 gene locus. The most common CNS symptom, affecting around 80% of patients, is daytime hypersomnolence. In some individuals this is coupled with a global disorganization of sleep habits and diurnal rhythm. Studies have shown sleep-onset rapid eye movement in 26% to 54% of patients (115,116). DM1 is also associated with a variable constellation of behavioral and cognitive changes, which may include anxiety, avoidant behavior, apathy, memory impairment, executive dysfunction, and problems with visuospatial processing.

Endocrinopathies. Endocrine disorders are frequently found in DM1. Hypothyroidism is common as is insulin resistance and type 2 diabetes. DM1 patients should be screened regularly with thyroid stimulating hormone (TSH) levels and a fasting insulin, fasting glucose, and HgbA1C. There is often reduced insulin-like growth factor-1 (IGF-1) levels, and pituitary deficiency with reduced growth hormone release and increased FSH levels. In males there is hypogonadism with testicular atrophy, reduced testosterone levels, oilgospermia, reduced fertility, and erectile dysfunction.

Pulmonary disorders. Individuals with congenital and noncongenital DM1 have a very high incidence of restrictive lung disease (110). Involvement of respiratory muscles is a major cause of respiratory distress and mortality in affected infants with DM1. Early ventilator support may be required but children frequently wean from the ventilator over the first few years of life. Swallowing difficulties that produce aspiration of material into the trachea and bronchial tree, along with weakened respiratory muscles and a weak cough have been reported as factors that may result in pulmonary complications in DM1 patients.

Gastrointestinal disorders. Gastrointestinal symptoms are highly prevalent in DM1 (117). The frequency of cholelithiasis is increased, which may reflect involvement of smooth muscle in the gallbladder (118). Intestinal dysmotility is common, producing symptoms of bowel urgency and diarrhea, often alternating with constipation. Whether these symptoms result from involvement of smooth muscle, enteric neurons, or both has not been determined.

Hair loss. Male pattern balding can occur in men and women with DM1. It rarely occurs in children with DM1.

Metabolic disorders. DM1 patients often show increased cholesterol and hypertriglyceridemia (119,120). Abnormal liver function tests are common in DM1 and DM2 (120,121). Modest elevations of alanine and aspartate aminotransferase levels, gamma-glutamyltransferase, and alkaline phosphatase may occur. Generally these abnormalities are nonprogressive and do not require liver biopsy unless there is corollary evidence of another disease process. It is unknown whether these changes represent a primary effect of DM on hepatocytes or a secondary consequence of metabolic derangements, biliary stasis, or fatty liver.

Congenital DM1. Twenty-five percent of infants born to myotonic mothers have congenital DM1 and 10% to 15% of all DM1 patients have congenital presentations. CTG repeats in these cases may range from 1,000 to over 4,000 repeats. Obstetric problems are inversely related to the age of presentation of the mother with DM1 and they include polyhydramnios, decreased fetal movements, breech presentation, and preterm labor. Infants show hypotonia, failure to thrive due to an inability to suck, bilateral facial and jaw muscle weakness, craniofacial changes including a tented upper lip and high-arched palate, neonatal respiratory distress (50%), delayed motor milestones, and delayed speech. Equinovarus deformities are common. Most children are weaned from the ventilator and walk independently. Clinically children with congenital DM1 usually show no myotonia over the first five years of life. Those with congenital DM1 usually show significantly reduced IQ, often in the mentally retarded range (108,122). The cognitive impairment is nonprogressive. Behavioral abnormalities include attention deficit hyperactivity and autistic behavior. Hydrocephalus may be seen in nearly half of patients with congenital DM1. MRI may show hypoplasia of corpus callosum and cerebral white matter changes, and diffuse cerebral atrophy. Diagnosis of congenital DM1 is made by molecular genetic studies as EMG shows no myotonia and CK is usually normal. Muscle biopsy is normal or nonspecific.

Proximal Myotonic Myopathy (PROMM; DM2)

Proximal myotonic myopathy (PROMM), also referred to as myotonic muscular dystrophy 2 (DM2), is a disorder with clinical similarities to DM1 (123). The abnormal protein in this autosomal-dominant disorder is the zinc finger protein 9 (ZNF9) with genetic loci at chromosome 3q21. Clinical severity is unrelated to variable size CCTG repeats. The prognosis is more benign than DM1 and there is not a severe congenital onset form. Onset is 8 to 60 years and there is intrafamilial variability. Patients present with muscle stiffness and pain. Weakness involves the proximal legs (hip flexors and extensors) greater than the distal legs as well as thumb and finger flexors. Facial weakness is seen in a minority of patients. Distal legs and respiratory muscles are not clinically affected. A hallmark is the enlargement of calf muscles. Muscle pain may be exercise-related, or at rest and increases with cold. The

myotonia is severe, asymmetric, and intermittent from day to day. The myotonia actually increases with warmth and decreases with cold. There is both grip and percussion myotonia. Cataracts are noted in all patients over 20 years with slit lamp examinations. Few studies have examined the cardiac effects of DM2, but it is clear that conduction disease and heart failure may both occur. One study found that the frequency of conduction disease was lower in DM2 than in DM1, but LVSD was more common (124). DM2 is also associated with increased risk of sudden death (125). Diabetes mellitus and/or hearing loss are observed in DM2. MRI shows white matter hyperintensity on T2 weighted images. CK is normal to less than 10 times elevated. EMG shows profound myotonia and compound muscle action potential (CMAP) amplitudes increment by 60% with exercise and reduce by 40% with rest. There is no decrement on short exercise, or slow or rapid repetitive stimulation. Myopathic motor units are seen proximally. MRI shows selective muscle involvement of the erector spinae and gluteus maximus. Diagnosis is confirmed by molecular genetic studies.

COMMON PATHOGENESIS OF MYOTONIC MUSCULAR DYSTROPHY (DM1 AND DM2). RNA toxicity has been implicated in both DM1 and DM2. The DM1 and DM2 gene discoveries were perplexing because DMPK and ZNF9 have no functional connections, yet the clinical features are similar. Also, the repeat expansions in both disorders are located in genomic regions that do not encode proteins. The evidence now supports a unifying theory of RNA-mediated pathogenesis in which both disorders result from toxicity of repetitive RNA (126,127). DM1 has been examined in more detail but it appears that the disease process is broadly similar in DM2.

Sequestration of muscle blind proteins (MBNL proteins) results from repetitive RNA. The expanded repeat in DM1 is located in the terminal part of the DMPK gene, close to the signal for polyadenylation. Even when highly expanded, the repeat sequence does not block the synthesis or processing of DMPK RNA. This results in production of a mutant mRNA that contains several thousand CUG repeats. These unusual transcripts are not exported to the cytoplasm, but instead are retained in the nucleus in discrete clumps or "foci." (128,129). These collections of mutant RNA were not previously observed by conventional histochemical stains. They were first revealed by staining tissue with probes that hybridize to the repetitive RNA. As expected, the foci are most conspicuous in cells with large expansions and high levels of DMPK expression: muscle fibers, smooth muscle cells, cardiomyocytes, and neurons (130–132). Proteins in the MBNL family bind to CUG repeat RNA with high affinity (133). These proteins normally act to regulate splicing of several hundred transcripts (134). They also have a role in regulating RNA transport and decay (135). However, these functions are lost when MBNL proteins are trapped in nuclear foci of CUG repeats (133,136). This results in expression of many incorrect splice products and protein isoforms. For example, missplicing of the ClC-1 chloride channel leads to reduced chloride conductance in muscle fibers (137,138), a physiologic alteration that is known to produce myotonia. Splicing defects of other transcripts, including insulin receptor, BIN1, dystrophin, and L-type calcium channels, are suspected to cause insulin resistance and myopathy (139–142). CUG repeat RNA can interfere with myogenic differentiation (143). These observations are consistent with the concept that RNA toxicity, and possibly MBNL sequestration, may contribute to developmental phenotypes of DM1. However, if that is the case, it is unclear why congenital disease does not also occur in DM2, considering that CCUG repeats are similarly effective for sequestering MBNL proteins.

EXPERIMENTAL TREATMENTS FOR DM1 AND DM2. Elucidation of disease mechanisms in DM1 and DM2 has led to the identification of novel targets for therapeutic intervention. In preclinical studies, evidence for engagement of these targets and therapeutic benefit has been obtained using several different approaches. Antisense oligonucleotides (ASOs), gene therapy vectors, and small molecules have been used to reduce the levels of toxic RNA (102–105). Small molecules and ASOs have also been used to inhibit MBNL binding to CUG/CCUG repeats or block the signaling pathways that lead to overexpression of CUGBP1 (144–147). Gene therapy vectors have been used to increase the expression of MBNL1 protein in animal models (148). Taken together, these studies have suggested that DM-associated biochemical and physiologic defects are reversible in transgenic mouse models. Further, chemical optimization and preclinical testing are necessary, but it seems possible that several of these therapeutic strategies may advance to clinical trials.

Myotonia Congenita

Myotonia congenita (Thomsens disease) presents in infancy and is inherited as an autosomal-dominant condition. There is an abnormality of the muscle chloride channel and the disease is linked to the 7q35 loci. There is variable penetrance. Symptoms may be present from birth, but usually develop later. The myotonia is relatively mild and may be manifest as difficulty in releasing objects or by difficulty walking or climbing stairs. Most patients do not show overt weakness. Functional difficulties in climbing stairs may be present. The myotonia is exacerbated by prolonged rest or inactivity. There is a "warm-up" phenomenon with reduced myotonia after repeated activity. Myotonia may be aggravated by cold, hunger, fatigue, and emotional upset. Patients may demonstrate grip myotonia or lid lag following upward gaze or squint and diplopia following sustained conjugate movement of the eyes in one direction. Nearly all have electrical myotonia by EMG but there is a warm-up

phenomenon with the myotonia reduced after a period of maximal contraction. Half of individuals have percussion myotonia. Patients may be symptom-free for weeks to months. The other common feature of myotonia congenita is muscle hypertrophy. Patients may exhibit a "Herculean" appearance. Patients have shown some benefit from treatment with quinine, mexiletine, Dilantin, procainamide, carbamazepine, and acetazolamide.

A recessive form of myotonia congenita (Becker form) also exists with later onset (ages 4 to 12), more marked myotonia, more striking hypertrophy of muscles and associated weakness of muscles, particularly with short exercise. EMG shows myotonia in distal muscles and less myotonia after maximal contraction. On repetitive stimulation there is a decremental CMAP response at high stimulation frequency (30 Hz) and following exercise. The dominant form seems more prone to aggravation of the myotonia by cold. Diagnosis is suspected based on clinical information and the presence of classical myotonic discharges on EMG. Diagnosis is confirmed with molecular genetic testing. Muscle biopsy is essentially normal apart from the presence of hypertrophy of fibers and an absence of type II-B fibers.

Paramyotonia Congenita

Paramyotonia congenita is an autosomal-dominant myotonic condition with at least two distinct genetic etiologies involving the sodium channel—α subunit (SCN4A) located at chromosome 17q35 and a muscle chloride channel (CLCN1) located at chromosome 7q35. The worsening of the myotonia with exercise is referred to as paradoxical myotonia. Weakness or stiffness may occur together or separately, there is cold and exercise aggravation, hypertrophy of musculature, and more severe involvement of hands and muscles of the face and neck. Myotonic episodes usually subside within a matter of hours but may last days. Some patients become worse with a potassium load. On electrodiagnostic studies there is a drop in CMAP amplitude with cooling. Dense fibrillations disappear below 28°C, myotonic bursts disappear below 20°C, and electrical silence may occur below 20°C. Treatment has involved mexiletine or tocainide.

Schwartz–Jampel Syndrome (Chondrodystrophic Myotonia)

Schwartz–Jampel syndrome is an autosomal-recessive disorder with myotonia, dwarfism, diffuse bone disease, narrow palpebral fissures, blepharospasm, micrognathia, and flattened facies. Onset is usually before age 3. Patients have respiratory and feeding difficulties with impaired swallowing. Limitation of joint movement may be present along with skeletal abnormalities, including short neck and kyphoscoliosis. Muscles are typically hypertrophic and clinically stiff. There is a characteristic facies with pursed lips, micrognathia, and small mouth. Patients may be difficult to intubate. Ocular changes include myopia and cataracts. There may be hirsutism and small testes. The symptoms are not progressive. The protein perlecan with gene loci at chromosome 1p34-p36 has been implicated.

Electrodiagnostic studies show continuous electrical activity with electrical silence being difficult to obtain. There is relatively little waxing and waning in either amplitude or frequency of complex repetitive discharges. Abnormal sodium channel kinetics in the sarcolemma of muscle has been demonstrated. Some therapeutic benefit has been reported with procainamide and carbamazepine.

Metabolic Myopathies

Inborn errors of glycogen metabolism and fatty acid metabolism may result in neuromuscular disorders. The major clinical presentations include *(a) fixed and progressive weakness or (b) exercise intolerance, cramps, and myalgias; and myoglobinuria*.

Fixed and progressive weakness may be caused by (a) glycogenoses (acid maltase deficiency or "Pompe disease," debrancher deficiency, brancher deficiency, and aldolase A deficiency) or (b) disorders of lipid metabolism (primary systemic carnitine deficiency, primary myopathic carnitine deficiency, secondary carnitine deficiency, short-chain acyl-coenzyme A synthetase deficiency [SCAD], medium-chain acylocoenzyme A synthetase dehydrogenase deficiency [MCAD], etc.).

Exercise intolerance, cramps/myalgias, and myoglobinuria may be caused by (a) glycogenoses (myophosphorylase deficiency or "McArdle's disease," phosphorylase kinase deficiency, phosphofructokinase [PFK] deficiency, phosphoglycerate mutase deficiency [PGAM], etc.), (b) disorders of lipid metabolism (carnitine palmitoyltranferase II deficiency [CPT II], VLCAD deficiency, TP deficiency, etc.), and (c) respiratory chain defects (coenzyme Q10 deficiency, complex I deficiency, complex III deficiency, and complex IV deficiency). While an exhaustive review of metabolic myopathies is not presented here, two prototypical metabolic myopathies—McArdle's disease and Pompe disease deserve mention.

MYOPHOSPHORYLASE DEFICIENCY (MCARDLE'S DISEASE). The most common glycogen storage disease is myophosphorylase deficiency, also known as McArdle's disease or glycogenosis type 5. The autosomal-recessive disorder has been linked to chromosome 11q13. and over 65 different disease causing mutations have been identified. Initial onset of symptoms often occurs during childhood and consists of poor endurance, fatigue, and exercise-induced cramps and myalgia that mainly affect active muscle groups. Myoglobinuria may also be absent during childhood with prevalence of fixed muscle weakness increasing as the patient ages. Symptoms can be precipitated by activities such as lifting heavy weights or climbing long flights of stairs. The "second wind phenomenon is

characteristic of this disorder. With the onset of myalgia, patients who rest briefly are then able to continue their physical activity with few or no symptoms. The normal function of muscle myophosphorylase is to catalyze the removal of 1,4-glycosyl residues from glycogen to produce glucose-1-phosphate. Its absence leads to decreased metabolic substrate for glycolysis to produce adenosine triphosphate (ATP). CK is persistently elevated between episodes of myoglobinuria. EMG is normal when patients are asymptomatic, but can show myotonic discharges and fibrillation potentials during an acute attack. Non-ischemic forearm exercise testing shows only an increase in ammonia and stable levels of lactic acid and pyruvate. Diagnosis is made by demonstrating the absence of myophosphorylase on muscle biopsy or by genetic mutation analysis. Possible treatments include high protein diet, pyridoxine, and creatine monohydrate.

ACID MALTASE DEFICIENCY (GLYCOGENOSIS TYPE 2; POMPE DISEASE). Acid maltase deficiency, also referred to as glycogenosis type 2 or "Pompe disease," is caused by a deficiency of acid α-1,4-glucosidase (GAA). Inheritance is autosomal recessive with linkage to chromosome 17q23. Disease incidence is 1 in 40,000 to 50,000 live births. The level of residual enzyme activity correlates with the severity of disease. Those with infantile onset (birth to 1 year) show less than 1% GAA activity, childhood and juvenile onset (1 year to teens) show 2% to 6% GAA activity, and those with adult onset (third decade or later) show 1% to 29% GAA activity. There is no clear correlation with residual activity within the adult population. There is glycogen accumulation in tissues. Clinically in those with infantile onset, symptoms and signs usually present within the first six months with hypotonia, weakness, cardiomegaly, congestive heart failure, and arrhythmia. There is liver involvement and pulmonary involvement. Anesthesia risks with succinylcholine include arrhythmia, hyperkalemia, and rhabdomyolysis. Propofol also produces risks. Safer anesthetics include ketamine and etomidate. Death occurs within the first year of life in 80% to 95% of untreated patients. In childhood onset there is mildly enlarged tongue, symmetric proximal weakness, and calf hypertrophy. Death occurs between 3 and 24 years due to respiratory failure. There is glycogen accumulation mainly in muscle. In adult-onset Pompe patients present with lower extremity weakness, restrictive lung disease from diaphragm involvement, headache, somnolence, and increased dyspnea when supine. Sleep disordered breathing is common. Expiration is more involved than inspiration due to chest wall muscle involvement. Nocturnal noninvasive ventilation is occasionally necessary. There is atrophy of paraspinous muscles and scapular winging. The disease course is one of slow progression over years. Pain, fatigue, and cramps are common complaints. There may be mild calf hypertrophy and diffuse muscle atrophy more proximally. Progressive disability is related to disease duration rather than the age of onset. Eventual respiratory involvement is common and many need wheelchairs or walking devices. Death is most often due to respiratory failure.

Diagnosis of Pompe disease is confirmed with either molecular genetic studies or biochemical analysis of acid maltase activity with muscle biopsy. However, new methods using blood samples to measure GAA activity are rapidly becoming adopted because of their speed and convenience (149,150). Typically serum CK is elevated (less than 10 times) in infants and less elevated in adults. EMG shows an irritative myopathy with fibrillations, complex repetitive discharges, and myotonic discharges. Treatment now involves enzyme replacement with intravenous administration of recombinant α-glucosidase (Myozyme® for infant onset and Lumizyme® for later onset). Better outcomes are seen with earlier initiation of therapy. Myozyme has been shown to benefit infantile disease with improved strength of distal and proximal muscles, improved pulmonary function, improved cardiomyopathy, and improved survival (151,152). Lumizyme is the formulation of recombinant α-glucosidase that benefits later-onset disease.

MITOCHONDRIAL DISORDERS

Mitochondrial encephalomyopathies, also referred to as "mitochondrial cytopathy," represent a complex group of disorders that affect multiple organ systems. Mitochondria are essential cellular organelles that convert carbohydrates, lipids, and proteins into usable energy in the form of ATP via aerobic metabolism. Although the human mitochondrial genome is only 16.5 Kb and encodes 13 proteins, many different clinical syndromes can result from mutations of these genes. Mutant mitochondrial DNA can be present in different proportions in various cell populations in a phenomenon known as heteroplasmy. The pathogenic effect of the mutation will only be manifested when a critical level of mutation is reached. Mutant and normal mitochondrial DNA segregate randomly during cell division, thus changing the proportion of mutant DNA in different cells over time. All mitochondria and mitochondrial DNA are derived from the mother's oocyte. Thus a family history compatible with maternal inheritance is strong evidence for a primary mitochondrial DNA mutation. Different family members in the maternal lineage may be asymptomatic or oligospermatic.

Of the many clinical features of mitochondrial disorders that involve multiple organ systems, some are frequently present together and should alert the clinician to a mitochondrial etiology. Ptosis, progressive external ophthalmoplegia (PEO), or both are hallmarks of Kearns–Sayre syndrome (KSS) which produces diplopia and blurred vision. Myopathy is common among patients with mitochondrial disorders. Neck flexors may be affected earlier and more severely than neck extensors. Progressive fixed proximal weakness is more common and patients may develop decreased muscle bulk. Premature fatigue, exercise intolerance, myalgia, and recurrent

myoglobinuria can be symptoms of mitochondrial disorders. Serum lactate and pyruvate often are elevated at rest and these levels may increase significantly after moderate exercise. Sensorineural hearing loss is frequently associated with mitochondrial encephalomyopathies. The hearing loss may be asymmetric and fluctuating in severity. Maternally inherited deafness and diabetes (DAD) is another phenotypic combination in patients with mitochondrial DNA mutations. Dementia can be a prominent feature in mitochondrial cytopathy.

The diagnostic workup of a mitochondrial disorder often includes a complete blood count, serum electrolytes (including calcium and phosphate), liver function tests, blood urea nitrogen, creatinine, blood lactate and pyruvate, ECG, lumbar puncture for cerebrospinal fluid (CSF) protein, glucose, lactate, and pyruvate, EMG and nerve conduction study, brain imaging with MRI, and muscle biopsy for histology and electron microscopy. Histochemical stains for mitochondrial enzymes (SDH, NADH-TR, and COX) may be obtained and the activities of mitochondrial respiratory chain enzymes can be measured in muscle tissue. Diagnosis of these disorders has been increasingly confirmed by complete mitochondrial DNA sequencing. The identification of numerous mitochondrial DNA mutations including duplications, deletions, multiple deletions, and pathogenic point mutations, provides specific genetic diagnoses.

Treatment is symptomatic for seizures (with avoidance of valproic acid which is contraindicated because of depletion of carnitine and direct inhibitory effects on the mitochondrial respiratory chain). Electrolyte disturbances related to hypoparathyroidism and diabetes mellitus are corrected. Thyroid replacement alleviates hypothyroidism and cardiac pacemaker placement prolongs life in KSS with conduction defects. Impairments in the oxidative phosphorylation pathway may generate increased amounts of free radical; therefore, antioxidants are prescribed which include beta carotene, vitamin C, vitamin E, and CoQ_{10}. CoQ_{10} shuttles electrons from complexes I and II to complex III and may stabilize the oxidative phosphorylation enzyme complexes within the inner mitochondrial membrane. The dose for CoQ_{10} in adults is 50 to 100 mg, three times per day. L-carnitine is also recommended. Dichloroacetate increases the pyruvate dehydrogenase complex and reduces lactate. Aerobic training is recommended for some conditions.

More common mitochondrial disorders presenting in childhood include the following:

Kearns–Sayre Syndrome (KSS)

These patients show PEO, retinitis pigmentosa on fundoscopic examination, and complete heart block. Onset is usually before 20 years of age. Cerebellar findings may be present on physical examination and patients may show limb weakness, hearing loss, diabetes mellitus, hypoparathyroidism, irregular menses, and growth hormone deficiency. Dementia may be progressive. CSF protein is frequently greater than 100 mg/dl.

Myoclonus Epilepsy With Ragged-Red Fibers (MERRF)

This clinical syndrome is defined by the presence of myoclonus, generalized seizures, ataxia, and ragged-red fibers (RRFs) on muscle biopsy. Symptoms usually begin in childhood. Other common clinical manifestations include hearing loss, dementia, exercise intolerance, and lactic acidosis. Multiple lipomatosis is common. Multiple members of a pedigree usually show the full syndrome.

Mitochondrial Encephalopathy, Lactic Acidosis, and Stroke-Like Episodes (MELAS)

This clinical syndrome is characterized by (a) stroke-like episodes at a young age and typically before 40 years; (b) encephalopathy evident as seizures, dementia, or both; and (c) lactic acidosis, RRFs on biopsy, or both as manifestations of the respiratory chain defects. Other frequent clinical features include normal early development, myogenic limb weakness, ataxia, myoclonus, migraine-like headaches, recurrent nausea and vomiting, and hearing loss. The abrupt onset strokes often affect the occipital cortex but may involve other regions of the brain. These patients often describe an antecedent history of migraine headaches that often occur prior to the stroke-like event. Patients may experience improvement over weeks to months but events virtually always recur. The lesions do not conform to territories of large vessels, a finding that favors the term *stroke-like episodes*. Dementia may occur and be progressive. There is infrequent occurrence of the full syndrome in more than one member of a pedigree. Based on the hypothesis that MELAS is caused by impaired vasodilation in an intracerebral artery, investigators have evaluated the effects of administering L-arginine, a NO precursor to patients acutely with the first signs of stroke-like episodes. Oral L-arginine administration within 30 minutes of a stroke was shown to significantly decrease frequency and severity of stroke-like episodes (153).

Neuropathy Ataxia and Retinitis Pigmentosa (NARP)

This disorder consists of the variable combinations of proximal neurogenic limb weakness, sensory neuropathy, ataxia, pigmentary retinopathy, developmental delay, dementia, and seizures. The onset occurs in teens and young adults and the course is gradually progressive.

Mitochondrial Neurogastrointestinal Encephalomyopathy (MNGIE)

This syndrome is clinically recognized by the unusual combination of six features: PEO, severe gastrointestinal

dysmotility, cachexia, peripheral neuropathy, diffuse leukoencephalopathy on MRI, and evidence of mitochondrial dysfunction (histologic, biochemical, or genetic). The peripheral neuropathy and the prominent gastrointestinal dysmotility are defining features. Lactic acidosis at rest is present in two-thirds of patients. Both axonal and demyelination polyneuropathy is frequent. Muscle biopsy reveals RRFs and neurogenic changes.

NEUROMUSCULAR JUNCTION DISORDERS

Transient Neonatal Myasthenia

Transient neonatal myasthenia occurs in about 10% to 15% of infants born to myasthenic mothers and is due to transplacental transfer of circulating acetylcholine receptor (AChR) antibodies from the myasthenic mother to the fetus. Symptoms appear within the first few hours of birth; however, occasionally onset may be delayed for 3 to 4 days. Typical clinical characteristics include feeding difficulty, generalized weakness and hypotonia, respiratory difficulties, fetal cry, facial weakness, and less frequently, ptosis.

The author prefers diagnostic confirmation by evaluating the response to edrophonium or neostigmine with repetitive nerve stimulation studies performed at baseline and subsequent to infusion of the anticholinesterase agent. A response decrement with slow rates of stimulation (2–5 Hz) over a train of four to five stimuli may be repaired by the edrophonium (Tensilon®) or neostigmine.

Treatment is largely supportive and the condition itself limiting with resolution generally occurring within 2 to 3 weeks, although occasional cases may persist longer.

Congenital Myasthenic Syndromes

Congenital myasthenia syndrome (CMS) is a term used for a heterogeneous group of disorders which are genetically determined rather than autoimmune mediated. Patients may present in the neonatal period, later in childhood, or even in adult life. Patients often exhibit ptosis, external ophthalmoparesis, facial weakness, general hypotonia, proximal greater than distal muscle weakness, and variable degrees of functional impairment. Patients show absence of anti-AChR antibodies. Over 20 subtypes have been described and congenital myasthenia may be classified according to the following: ***(a) presynaptic defects*** (eg, CHAT—choline acetyltransferase deficiency causing CMS with epidodic apnea; paucity of synaptic vesicles and reduced quantal release; or congenital Lambert–Eaton-like syndrome), ***(b) synaptic basal lamina defects*** (eg, endplate acetylcholinesterase (AChE) deficiency at NMJs), and ***(c) postsynaptic defects:*** (eg, AChR disorders involving α, β, δ, e subunits; kinetic abnormalities in AChR function caused by AChR deficiency; slow AChR channel syndromes; fast-channel syndromes; endplate rapsyn deficiency, etc.).

Several congenital myasthenic syndromes have been associated with arthrogryposis syndromes. For example "multiple pterygium syndrome" (Escobar syndrome) has been associated with AChR gamma, alpha 1, and delta subunit mutations.

For diagnostic workup, standard EMG with repetitive nerve stimulation is utilized initially and subsequently stimulated single fiber EMG may be useful. Ultrastructural evaluation of the NMJ with electron microscopy usually is performed on a biopsy of the deltoid or biceps, including the muscle region containing the NMJ or the "motor point." For in vitro electrophysiologic and immunocytochemical studies of the NMJ, a short muscle usually is removed from the origin to insertion along with its motor branch and NMJ (a "motor point biopsy"). Muscles obtained have included the anconeus muscle near the elbow, the external intercostal muscle in the fifth or sixth intercostal space near the anterior axillary line or the peroneus tertius muscle in the lower extremity. Such in vitro electrophysiologic studies allow specific delineation of the congenital myasthenic syndrome into one of the numerous specific subtypes. More recently, the diagnostic evaluation of CMS has increasingly relied upon molecular genetic studies as the gene loci have been identified for the majority of subtypes.

For treatment of a CMS subtype a definitive diagnosis is important because some CMS syndromes deteriorate with empiric treatment with AChE inhibitors such as pyridostigmine (Mestinon®). For example, slow-channel syndromes may deteriorate on pyridostigmine and endplate AChE deficiency may deteriorate or show no response. Some presynaptic syndromes may show response to 3,4 diaminopyridine, which increases release of acetylcholine at the presynaptic terminal. This drug has been used in the Lambert–Eaton syndrome and in presynaptic CMS on a compassionate use basis.

Autoimmune Myasthenia Gravis

This disorder is similar to the autoimmune myasthenia gravis observed in adults. The onset is often insidious, but at times patients may present with acute respiratory difficulties. Patients usually present with variable degrees of ophthalmoparesis and ptosis. In addition, patients may exhibit facial weakness, swallowing difficulties, speech problems, and weakness of the neck, trunk, and limbs. Proximal muscles are more affected than distal, and the upper limits are more affected than the lower.

Fluctuation in the disease course with relapse and remission is common. Patients often complain of fatigue and diplopia, as well as progressive difficulty with chewing or swallowing. Patients are often worse with fatigue toward the end of the day. Thymoma, which occurs in about 10% of adult cases, is not a feature of the childhood onset disease.

Serum AChR antibodies are an important diagnostic screening tool. Anti-AChR antibodies can be detected

in the serum in about 85% to 90% of patients with generalized myasthenia gravis and greater than 50% of those with ocular myasthenia. The most common antibodies detected are AChR binding, followed by AChR modulating, and then striational AChR antibodies. MUSK antibodies are an additional marker present in some seronegative patients and many patients with ocular myasthenia.

Diagnosis may also be confirmed by clinical response to an anticholinesterase drug such as edrophonium (Tensilon). Alternatively, neostigmine, a longer acting agent, can be used. Repetitive nerve stimulation studies show a characteristic decrement in the compound muscle action potential with slow stimulation rates (2–5 Hz) over a train of four to five stimuli. A decrement greater than 12% to 15% is often noted. Electrophysiologic studies may be more sensitive with proximal muscle groups such as the accessory nerve to the trapezius or study of the facial nerve. Abnormal repetitive nerve stimulation studies may also be seen in the Lambert–Eaton syndrome, botulism, and congenital myasthenic syndromes. Single fiber EMG is usually impractical in children; however, stimulated single fiber EMG may be performed under anesthesia. Management may include treatment with anticholinesterase drugs, such as pyridostigmine, corticosteroids (prednisone), IV immunoglobulin, immunosuppressants (azathioprine, cyclosporine, mycophenolate mofetil, or cyclophosphamide), plasma exchange, or thymectomy.

Infantile Botulism

Infants with botulism usually present between 10 days to 6 months with an acute onset of hypotonia, dysphagia, constipation, weak cry, and respiratory insufficiency. The neurologic examination shows diffuse hypotonia and weakness, ptosis, ophthalmoplegia with pupillary dilation, reduced gag reflex, and relative preservation of DTRs. The diagnosis may be made by electrodiagnostic studies (see Chapter 6) or by measurement of Clostridium Botulinum toxin in a rectal aspirate containing stool.

Noninfantile Acquired Botulism

Older children and adults acquire botulism through poorly cooked, contaminated food with the toxin or through a cutaneous wound that becomes contaminated with soil-containing Clostridium Botulinum. The toxin can often be identified in the serum and the food source. Clinical findings include acute onset of constipation, ptosis, diplopia, bulbar weakness, respiratory difficulties, ophthalmoparesis, pupillary dilation, and diminished DTRs. Recovery may take months. The diagnosis is generally made from electrodiagnostic studies.

PERIPHERAL NERVE DISORDERS

Acute Inflammatory Demyelinating Polyneuropathy (AIDP; Guillain–Barré Syndrome)

Acute inflammatory demyelinating polyneuropathy (AIDP) is a primarily demyelinating neuropathy with autoimmune etiology. Motor axons are affected more than sensory axons. Incidence in children is similar to that seen in adults. Children often have a prodromal respiratory or gastrointestinal infection occurring within 1 month of onset. Common precipitating infections include mycoplasma, cytomegalovirus, Epstein–Barr virus, campylobacter jejuni, and various vaccinations. Weakness generally begins distally in the lower extremity with a progressive ascending paralysis ultimately involving the upper limbs. Pain and sensory symptoms are not uncommon. The most common cranial nerve abnormality is an ipsilateral or bilateral lower motor neuron facial paralysis. Objective sensory loss has been documented in the minority of children (154). In one series, only 15% required mechanical ventilation (155). The maximal degree of weakness generally reaches a peak within 2 weeks of onset and time to maximum recovery was 7 +/− 5 months in one series (156). Complete recovery occurs in most children. Classic criteria for poor recovery in adults (low median CMAPs and fibrillation potentials) may not apply to children (157).

Disturbances of the autonomic nervous system are common in children, including transient disturbances of bowel and bladder, excessive sweating or vasoconstriction, mild hypertension or hypotension, and occasionally cardiac arrhythmias.

The acute motor axonal neuropathy (AMAN) involves predominantly motor nerve fibers with a physiologic pattern suggesting axonal damage, whereas the AIDP involves both motor and sensory nerve fibers with a physiologic pattern suggesting demyelination. Another clinical variant is the Miller Fisher syndrome characterized by acute onset ataxia, ophthalmoparesis, and areflexia.

Diagnosis is generally confirmed by electrodiagnostic studies (see Chapter 6), and the CSF protein is characteristically elevated in a majority of children. Serum autoantibodies which may be elevated include IgM and IgG versus beta-Tubulin and heparin sulfate. AMAN patients may show increased IgG antibodies to GM1 ganglioside. The Miller Fisher syndrome is associated with a high frequency of the IgG GQ1b antibodies. The major considerations in differential diagnosis of AIDP or AMAN include transverse myelitis, toxic neuropathies, tick paralysis, infantile botulism, myasthenia gravis, and dermatomyositis.

Treatment has typically included corticosteroids, plasma exchange, or more recently, intravenous immunoglobulin (IVIG; 158–161). AIDP patients respond to both plasma exchange and IVIG. Patients with AMAN respond

preferentially to IVIG over plasma exchange. Recovery is often quite good in children without treatment. After standard IVIG therapy, children with axonal forms of Guillain–Barré syndrome recover more slowly than those with the demyelinating form, but outcome at 12 months appears to be equally favorable in two groups (162).

Chronic Inflammatory Demyelinating Polyradiculoneuropathy (CIDP)

Children with chronic inflammatory demyelinating polyradiculoneuropathy (CIDP) often have a presentation similar to AIDP; however, the disorder continues with a chronic or relapsing course. The disorder may begin as early as infancy, but is seen in children and adults. Electrophysiologic studies show focal conduction block, temporal dispersion of CMAPs, prolongation of distal motor latencies, markedly slow conduction velocities, and absent or prolonged H-wave and F-wave latencies. CIDP cases often demonstrate axonal loss on EMG. The CSF protein is elevated in most cases.

The differential diagnosis usually includes Charcot–Marie–Tooth (CMT) types I and III. The presence of acute relapsing episodes points toward CIDP. Due to the more severe involvement of proximal nerves and nerve roots, a distal sural nerve biopsy may not always show inflammatory changes and demyelination.

Treatment may include corticosteroids (prednisone) and IVIG as first-line approaches and subsequently plasma exchange.

Charcot–Marie–Tooth—CMT (Hereditary Motor Sensory Neuropathy)

Charcot–Marie–Tooth (CMT) neuropathy (also called hereditary motor sensory neuropathy [HMSN]) is a heterogeneous group of inherited diseases of peripheral nerve that affects both children and adults and causes significant progressive neuromuscular impairment (162,163). It has been estimated that 1 per 2,500 to 3,000 persons has a form of CMT. CMT 1 denotes individuals with a hypertrophic demyelinating neuropathy ("onion bulbs") and reduced nerve conduction velocities, whereas CMT 2 refers to individuals with an axonal neuropathy and normal or slightly reduced nerve conduction velocities. Individuals with CMT 3 (Dejerine-Sottas disease) have a primarily demyelinating peripheral neuropathy with a more severe phenotype presenting in infancy. Historically types 1, 2, and 3 were felt to be autosomal-dominant conditions with type 3 CMT patients exhibiting point mutations with frameshift and either dominant or recessive inheritance. CMT 4 refers to autosomal-recessive CMT. However, recently axonal forms of CMT have been identified with autosomal-recessive inheritance (deemed AR-CMT 2A, 2B, etc.)

In general, in most CMT subtypes onset is usually during the first or second decade of life. Both motor and sensory nerve functions are affected. The clinical features include distal muscle weakness, impaired sensation, and absent or diminished DTRs. Weakness usually is greatest initially present in the foot and hand intrinsics and distal lower extremities and subsequently in the distal upper extremities. Slow progressive weakness, more proximally in the knees, elbows, and pelvic and shoulder girdles may occur over decades (85). There is variable penetrance in most subtypes. The various gene locations and known protein abnormalities associated with various forms of CMT (HMSN) are given in Table 18.3.

The majority of ***CMT 1*** pedigrees (70%), demonstrate linkage to chromosome 17p11.2-12 and are designated CMT 1A (164). CMT 1A duplication results in increased expression of peripheral myelin protein-22 (PMP-22). Conduction velocities are uniformly slow in all nerves with a mean of 17 to 20 M/s and a range 5 to 34 M/s. Onset is typically in the first decade with leg areflexia, gait disorder (toe walking or steppage gait), foot muscle atrophy or pes cavus, occasionally short Achilles tendons, and enlarged nerves owing to onion bulb formation in half of patients. Distal weakness develops initially in intrinsic muscles of the feet and hands with development of wasting of musculature occurring slowly over time (Figure 18.12). Ankle dorsiflexion, ankle eversion, and extensor hallucis longus weakness develop with more normal strength proximally. Progress cavus foot deformities with clawing of the toes often develop (Figure 18.13). Orthopedic procedures are limited to soft tissue procedures and correcting wedge osteotomies and joint fusion should be avoided if possible to avoid late pain. Late in the disease, diaphragm or bulbar weakness may develop in rare cases. Progression is slow over many decades. Defects in the human myelin zero gene (P_0) on chromosome 1q22-q23 leads to CMT 1B. P_0 is the major protein structural component of peripheral nervous system myelin. The clinical presentation is similar to CMT 1A; however, onset may lag into the second to third decade in a minority of patients and there is more variability in severity. Nerve conduction velocities are usually less than 20 m/s. P_0 mutations may lead to other clinical variants referred to as CMT 1E (demyelinating CMT with deafness), and predominantly axonal neuropathy with late adult onset (eg, CMT 2I and CMT 2J with hearing loss and pupillary abnormalities).

CMT 2 is a less common disorder than CMT 1. Generally, CMT 2 patients demonstrate later age of onset, less involvement of the small muscles of the hands, and no palpably enlarged nerves. Wasting in the calf and anterior compartment of the leg may give rise to an "inverted champagne bottle" or "stork-leg" appearance. Conduction velocities are mildly reduced and CMAP amplitudes and sensory nerve action potential (SNAP) amplitudes are usually reduced. CMT 2A2 with mitofusin abnormality

TABLE 18.3 HMSN TYPES: COMPARISON OF CLINICAL FEATURES

DISORDER	GENE	LOCATION	USUAL ONSET	EARLY OR DISTINCT SYMPTOMS	TENDON REFLEXES	AVERAGE NCVS
CMT1: Dominant; Demyelinating						
CMT 1A	PMP-22	17p11	First decade	Distal weakness	Absent	15–20 m/s
CMT 1B	P_0	1q22	First decade	Distal weakness	Absent	<20 m/s
CMT 1C	LITAF	16p13	Second decade	Distal weakness	Reduced	16–25 m/s
CMT 1D	EGR2	10q21	Second decade	Distal weakness	Absent	26–42 m/s
CMT X (S-D*)	Connexin-32	Xq13	Second decade	Distal weakness	Absent distal	25–40 m/s
HNPP	PMP-22	17p11	Third decade	Focal episodic weakness	Normal	Entrapments
Dejerine-Sottas (HMSN 3)	PMP-22 8q23 EGR2	17p11 8q23 10q21	2 years	Severe weakness	Absent	<10 m/s
CMT Intermediate NCV	DNM2 10q24 1p34 P0 CMT-X	19p12 10q24 1p34 1q22 Xq13	First or second decade	Distal weakness		25–50 m/s
CMT2: Dominant; Axonal						
CMT 2A	KIF1Bβ Mitofusin 2	1p36 1p36.22	10 years	Distal weakness	Absent distal	>38 m/s
CMT 2B	RAB7	3q13	Second decade	Distal weakness Sensory loss acromutilation	Absent distal	Axon loss
CMT 2C	*TRPV4*	12q23-q24	First decade	Vocal cord and distal weakness	Absent	>50 m/s
CMT 2D	GARS	7p15	16–30 years	Distal weakness Arms > legs	Reduced	Axon loss
CMT 2E	NF-68	8p21	1–40 years	Distal weakness	Reduced	Axon loss
CMT 2F/Distal HMSN	HSPB1 (HSP 27)	7q11	6–54 years	Difficulty walking	Reduced ankle	Axon loss
CMT 2G	12q12-q13.3	12q12	15–25 years	Distal weakness	Reduced	42–58 m/s
CMT 2L	HSPB8	12q24	15–33 years	Distal weakness	Reduced	Axon loss
HMSN-Proximal	Trk-fused Gene (TFG)	3q13	17–50 years	Proximal weakness cramps	Absent	Axon loss
HSMN + Ataxia	IFRD1	7q22	13–27 years	Gait ataxia	Absent	Axon loss
CMT 2 P_0	P_0	1q22	37–61 years	Leg weakness Pupil or hearing	Reduced	<38 m/s–normal
AR-CMT2: Recessive; Axonal						
AR-CMT2A	Lamin A/C	1q21	Second decade	Distal weakness	Reduced	Axon loss
AR-CMT2B		19q13	Third and fourth decades	Distal weakness	Absent distal	Axon loss
AR-CMT2 Ouvrier		Autosomal	First decade	Distal weakness	Reduced	Axon loss
HMSN 3: Infantile						
Dejerine-Sottas (HMSN 3)	P_0 PMP-22 Periaxin	Autosomal Dominant/ recessive	2 years	Severe weakness	Absent	<10 m/s

(*continued*)

TABLE 18.3 HMSN TYPES: COMPARISON OF CLINICAL FEATURES (*CONTINUED*)

DISORDER	GENE	LOCATION	USUAL ONSET	EARLY OR DISTINCT SYMPTOMS	TENDON REFLEXES	AVERAGE NCVS
Congenital Hypomyelinating Neuropathy	P_0 EGR2 PMP-22	Autosomal Recessive	Birth	Severe weakness	Absent	<10 m/s
CMT4: Recessive; Demyelinating						
CMT 4A	GDAP1	8q13	Childhood	Distal weakness	Reduced	Slow
CMT 4B	MTMR2	11q22	2–4 years	Distal and proximal weakness	Absent	Slow
CMT 4B2	SBF2	11p15	First two decades	Distal weakness Sensory loss	Absent	15–30 m/s
CMT 4C	KIAA1985	5q23	5–15 years	Delayed walking	Reduced	14–32 m/s
CMT 4D (Lom)	NDRG1	8q24	1–10 years	Gait disorder	Absent	10–20 m/s
CMT 4E	EGR2	10q21	Birth	Infant hypotonia	Absent	9–20 m/s
CMT 4F	Periaxin	19q13	1–3 years	Motor delay	Absent	Absent
CMT 4H	FGD4	12q12	10–24 months	Walking delay	Absent	<15 m/s
CCFDN	CTDP1	18q23	First or second decade	Distal leg weakness	Reduced	20–34 m/s

Abbreviation: HMSN, hereditary motor sensory neuropathy.

accounts for approximately 20% of CMT 2 probands. CMT 2C linked to chromosome 12q23-q24 has interesting features of early onset in the first decade, and diaphragm and intercostal weakness producing shortness of breath. Vocal cord paralysis may alter the voice of these patients. The disease may progress to proximal and face muscles. Arthrogryposis is present in some patients. Phrenic nerve CMAPs are often reduced. CMT 2E with abnormality in neurofilament light chain (NFL) linked to chromosome 8p21 may have associated hearing loss in 30% of cases. While most axonal CMT is autosomal dominant, emerging pedigrees are being identified with recessive inheritance.

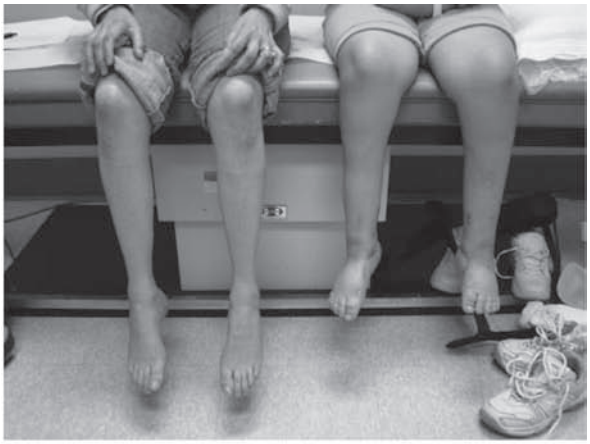
Dejerine-Sottas disease (CMT 3) is a severe hypertrophic demyelinating polyneuropathy with onset in infancy or early childhood. Most achieve ambulation but some may subsequently progress to wheelchair reliance. Nerve conduction velocities are greatly slowed (often below 10 m/s), and elevations in CSF protein may be present. Dejerine-Sottas disease may be associated with point mutations in either the PMP-22, P_0, or EGR2 genes (164). While this disorder was previously felt to be autosomal recessive, many cases are due to de novo point mutations and actually have dominant inheritance.

Congenital hypomyelinating neuropathy is a severe and often fatal newborn disorder that often presents with respiratory distress in the delivery room. These infants often have severe generalized hypotonia and associated arthrogryposis. Diagnostically, these infants have absent SNAPs or low amplitude SNAPs with prolonged distal latencies. Compound muscle action potentials are either absent or low amplitude with motor conduction velocities ranging from 3 to 10 meters per second. The disorder has been linked to PMP-22, P0, and EGR2 genes. Sural nerve biopsy may be useful. Inheritance is usually autosomal recessive with some dominant inheritance linked to EGR2.

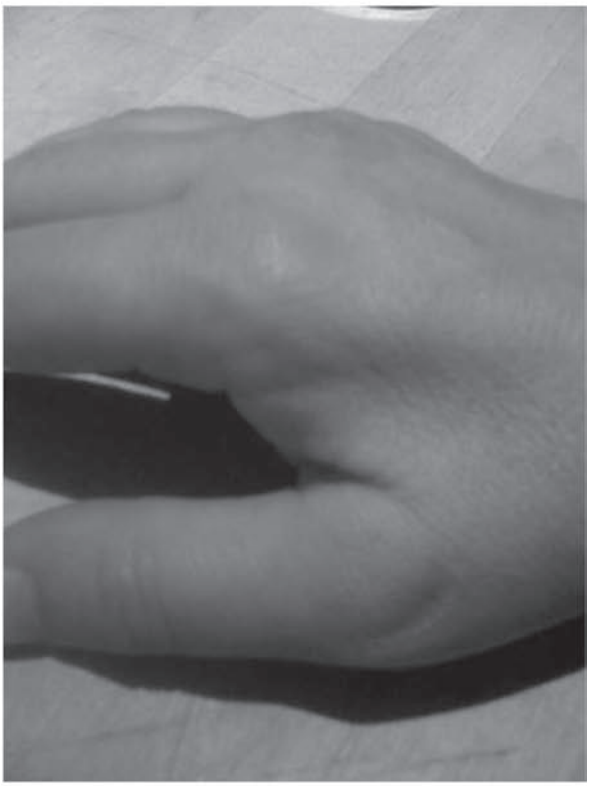
Autosomal-recessive CMT 4 is relatively rare. Most are demyelinating with more severe phenotypes and onset is often in childhood. CMT 4C linked to 5q23 is a relatively more common form of CMT 4.

Hereditary neuropathy with liability to pressure palsies (HNPP) is an autosomal-dominant disorder that produces episodic recurrent nerve entrapments with focal demyelination. Patients may present with peroneal palsies, carpal tunnel syndrome, and other entrapment neuropathies. A positive family history of entrapments often exists. Peripheral nerve biopsies may demonstrate segmental demyelination and tomaculous or "sausage-like" formations. A deletion at the PMP-22 gene locus (chromosome 17p11.2-12) causes this autosomal-dominant condition, in contrast to a duplication of this gene which causes CMT 1A.

Patients with an ***X-linked dominant form of CMT (CMT-X)*** have been described. Male-to-male transmission



(A)



(B)

FIGURE 18.12 Distal weakness of intrinsic muscles of the feet (A) and hands (B) with wasting in CMT.

is not observed and the disorder generally shows earlier onset and faster rate of progression. The gene locus coding for connexon 32 protein is Xq13, which encodes a major component of gap junctions which provide a pathway for the transfer of ions and nutrients around and across the myelin sheath.

DNA testing for many of the CMT subtypes (particularly CMT 1) is available but the ordering of extensive CMT genetic batteries is very expensive unless guided by nerve conduction study findings. Nerve conduction studies may be more expeditiously carried out on an affected

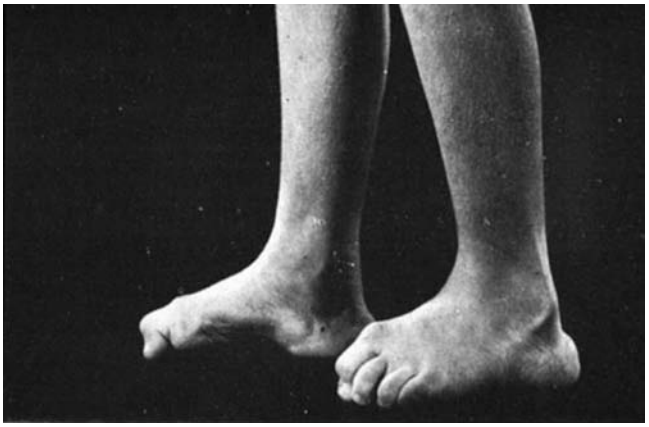


FIGURE 18.13 Progressive cavus foot deformities with clawing of the toes on CMT.

parent to guide the molecular genetic workup of an affected child. Given the overlap of some gene abnormalities with several CMT clinical subtypes it is often difficult to make a definitive diagnosis on genetic study results without the additional information provided by nerve conduction testing. HMSN remains one clinical entity that continues to warrant electrodiagnostic evaluation.

Toxic Neuropathies

Toxic polyneuropathies are rare occurrences in children in North America. Toxic exposure to heavy metals and environmental toxins may be more common in other regions of the world. Expeditious diagnosis is critical to identify and remove the source of the toxicity and to establish treatment with agents such as penicillamine. ***Arsenic polyneuropathy*** is a sensorimotor neuropathy that may be axonal or, at times, predominantly demyelinating, simulating Guillain–Barré syndrome or CIDP. Gastrointestinal symptoms are common as well as tachycardia and hypotension. Mee's lines may be seen in nails along with other skin changes and alopecia. The diagnosis is established by obtaining levels of arsenic in blood, urine, hair, and nail samples.

Lead polyneuropathy is most commonly observed in children who have ingested old lead-based paint. Acute exposures cause lead encephalopathy more commonly. Clinical findings may include anorexia, nausea and vomiting, gastrointestinal disturbance, fatigue, clumsiness and ataxia, and occasionally cognitive impairment, seizures, mental status changes, papilledema, and coma. The weakness is predominantly in the lower limbs, but the upper limbs may be involved. Electrophysiologic studies show a primarily axonal degeneration affecting motor greater than sensory axons. A microcytic hypochromic anemia with basophilic stippling of red blood cells establishes the diagnosis. Lead lines may be evident in long bone films. Lead levels may or may not be elevated

in urine and blood but levels of delta aminolevulinic acid are usually elevated in the urine.

Mercury poisoning may occur from the ingestion of mercuric salts, exposure to mercury vapor or use of topical ammonia mercury ointments. Patients present with a generalized encephalopathy, fatigue, and occasionally a skin rash. A predominantly distal motor axonal neuropathy occurs. DTRs may be absent and the gait is often ataxic. Sensory examination is often normal, although patients may complain of distal paresthesias. Electrophysiologic studies show motor axonal degeneration with normal sensory conduction studies.

Organophosphate poisoning may be due to exposure to insecticides or high-temperature lubricants or softeners used in the plastic industry. Patients present with an encephalopathy manifested by confusion and coma. In acute exposure cholinergic crisis, manifested by sweating, abdominal cramps, diarrhea, and constricted pupils, may be present. A predominantly motor polyneuropathy is a late effect. However, the disorder may present as a rapidly progressive polyneuropathy mimicking Guillain–Barré syndrome. Severe paralysis with respiratory failure requiring ventilatory support may occur and in this situation there may be a superimposed postsynaptic defect in neuromuscular transmission.

Glue-sniffing (N-Hexane) neuropathy may be seen in teenage recreational glue sniffers. Repeated use may cause symptoms and signs of a predominantly distal motor and sensory polyneuropathy which is predominantly demyelinating. Motor and sensory nerve conduction studies demonstrate moderate slowing.

Chemotherapeutic agents, in particular, vincristine, often produce a relatively pure motor axonal polyneuropathy. Severity is dose-dependent. Clinical findings include distal weakness, absent DTRs, and at times foot drop. The disorder is often readily apparent by clinical examination and electrophysiologic studies or nerve biopsy is usually not necessary. The neuropathy usually improves with discontinuation of the medication, although significant electrophysiologic abnormalities (reduced CMAP amplitudes and neuropathic recruitment) may persist. Vincristine may be particularly troublesome for children with HMSN.

Metabolic Neuropathies

Uremic neuropathy often occurs in children with end-stage renal disease. If clinical manifestations are present, they consist of a predominantly distal motor and sensory polyneuropathy with glove and stocking loss of sensation, loss of vibratory sense, and distal weakness, particularly involving peroneal innervated musculature. With successful renal transplantation, clinical findings and electrophysiologic abnormalities normalize. ***Diabetic polyneuropathy*** usually is a mixed motor and sensory polyneuropathy with both axonal changes and mild demyelination. The polyneuropathy is less common in children with diabetes mellitus, as compared with adults. The severity of the neuropathy may be related to the degree of glucose control (165).

MOTOR NEURON DISORDERS

Predominantly Proximal Spinal Muscular Atrophy

Spinal muscular atrophy (SMA) is a term used to describe a varied group of inherited disorders characterized by weakness and muscle wasting, secondary to degeneration of both anterior horn cells of the spinal cord and brainstem motor nuclei without pyramidal tract involvement. Three subtypes of autosomal-recessive predominantly proximal SMA have been described, all linked to chromosome 5q. A common nomenclature subdivides SMA into types I, II, and III, based on the age of onset and the age of death, whereas the other approach classifies cases as severe, intermediate, and mild, based on the ability to achieve independent sitting, independent standing, and walking. The International Consortium on SMA attempted to standardize the classification of childhood SMA to provide a rational basis for linkage studies and therapeutic trials (Table 18.4) (166).

SMA type I (Werdnig–Hoffmann, severe form) was defined by the International Consortium on SMA as follows: Onset from birth to 6 months, no achievement of sitting without support, and death usually prior to age 2 years. In SMA type II (intermediate form), onset is before 18 months, sitting is usually obtained, but standing and ambulation are never obtained and death occurs above the age of 2 years, usually much later in adulthood. In SMA type III (Kugelberg–Welander, mild form), the onset is after the age of 18 months, patients develop the ability to stand and walk, and death is in adulthood. There is considerable variability and severity within each of the three groups and occasionally some overlap exists. For example, patients with onset prior to 6 months may exhibit prolonged survival well past 4 years of age. Patients with onset between 6 and 18 months may ultimately achieve standing and independent ambulation. An adult-onset type of SMA with mild disease phenotype presenting usually in the second or third decade, has been recognized. These patients usually are able to ambulate with minor motor impairments. Although the adult-onset SMA is not classified formally by criteria set forth by the consortium, among clinicians, SMA type IV has been used widely to classify later-onset patients with mild disease features. A modified classification has been proposed by Zerres and Rudnik-Schoneborn which defines adult SMA as type IV (167).

The carrier frequency for SMA in the general population is estimated at about 1 in 40 to 50 individuals. Autosomal-recessive inheritance has long been documented in proximal SMA with childhood onset. In 1990, all three forms of SMA were mapped to chromosomal

TABLE 18.4 CHILDHOOD ONSET PROXIMAL SPINAL MUSCULAR ATROPHY

	SMA I (WERDNIG–HOFFMANN)	SMA II (INTERMEDIATE SMA)	SMA III (KUGELBERG–WELANDER)
Onset	<6 months	6–18 months	>18 months IIIa < 3 years IIIb > 3 years
Genetics	SMN1: AR homozygous deletion SMN2: <3 copies	SMN1: AR homozygous deletion SMN2: three copies	SMN1: AR homozygous deletion SMN2: 4–8 copies
Phenotype	Severe hypotonia, weak suck, weak cry, proximal weakness, absent reflexes, respiratory failure common	Hypotonia, proximal weakness, muscle wasting, contractures, scoliosis, absent reflexes, tongue fasciculations	Proximal symmetric weakness, lordotic gait, Gower's sign, decreased reflexes, tremor, tongue fasciculations
Milestones	Poor head control; Never sit independently	Sit with head control; never stand unassisted; may require ventilatory support	Stand and walk unassisted; may lose standing or continue to walk IIIa: onset 18 months to < 3 years (80% not walking at age 40) IIIb: onset > 3 years (40% not walking at age 40)
Life Expectancy	1–2 years 10% living at age 20	Most live to third decade; many live to fourth to fifth decade	Normal life expectancy

region 5q13, indicating that allelic variance of the same disease locus accounts for the clinical heterogeneity (99,100). During the past two decades, tremendous advances have been made in our understanding of the genetic basis for SMA (168–176). A detailed analysis of the 5q13 region revealed that this chromosomal region in humans contained a large inverted duplication, with at least two genes present in telomeric and centromeric copies.

Further studies have identified the SMA causative gene as the survival motor neuron (SMN) 1 gene (SMN1, telomeric copy), along with a disease modifying gene (SMN2, centromeric copy) (168–170). Briefly, the two SMN genes are nearly identical except for a difference of only five nucleotides in their 3′ regions, without any alteration of the amino acid sequence of the protein. However, the critical difference between the SMN1 and SMN2 genes is a C–T transition located within the exon-splicing region of the SMN2 that affects the splicing of exon 7. This change results in frequent exon 7 skipping during the splicing of SMN2 transcripts (174,175). It is thought that the resulting truncated SMN protein, without its exon 7 contribution, is a less stable form of SMN protein, and therefore, rapidly degraded. In about 95% of SMA patients, both copies of SMN1 exon 7 are absent because of mutations. In the remaining SMA-affected patients, other small or subtle mutations have been identified (169).

Genetic studies have now established that SMA is caused by mutations in the telomeric SMN1 gene, with all patients having at least one copy of the centromeric SMN2 gene. At least one copy of the SMN2 must be present in the setting of homozygous SMN1 mutations; otherwise, embryonic lethality occurs. The disease severity is primarily determined by the copy number of the SMN2 gene which produces a protein with similar function to the SMN1 gene but in much smaller quantities (176). The copy number of SMN2 varies in the population, and this variation appears to have some important modifying effects on SMA disease severity (177–179). All SMA patients have more than two SMN2 genes. It appears that a higher number of SMN2 copies in the setting of SMN1 mutations is associated with a less severe clinical SMA phenotype: SMA I (severe): two or three gene copies of SMN2; SMA II: three copies of SMN2; SMA III: four to eight copies of SMN2. However, substantial variations in SMA phenotype and disease severity can exist with a given SMN2 copy number, so it is not recommended that disease severity be predicted based solely on SMN2 copy numbers. Although we now know that SMN protein is expressed widely in many tissues throughout the body, its function is still not completely understood at this time.

Spinal Muscular Atrophy I (Werdnig–Hoffmann Disease)

The majority of cases of SMA I present within the first two months with generalized hypotonia and symmetrical weakness. The age of onset of symptoms is less than 4 months in the vast majority of cases. Weak sucking, dysphagia, labored breathing during feeding, frequent aspiration of food or secretions, and weak cry are frequently noted by history.

Examination shows generalized hypotonia and symmetric weakness involving the lower extremities earlier and to a greater extent in the upper extremities. Proximal muscles are weaker than distal extremities. In the supine position, the lower extremities may be abducted and externally rotated in a "frog-leg" position. The upper extremities tend to be adducted and externally rotated at the shoulders with a semiflexed elbow. Volitional movements of fingers and hands persist well past the time when the shoulders and elbows cannot be flexed against gravity. The thorax is flattened anteroposteriorly and bell-shaped as a result of intercostal weakness. Pectus excavatum may be variably present. The diaphragm is usually preserved, relative to the intercostal and abdominal musculature. This results in a diaphragmatic breathing pattern during respiration with abdominal protrusion, paradoxical thoracic depression, and intercostal retraction. Neck flexor weakness may result in persistent posterior head lag when the trunk is lifted forward from the supine position. Neck extensor weakness may result in forward head lag when the infant is positioned in the horizontal prone position. With advanced disease, the mouth may remain open as a result of masticatory muscle weakness. Facial weakness may be noted in up to half of patients. The diagnostic criteria for SMA outlined by the International SMA Consortium (166) list marked facial weakness as an exclusionary criterion for SMA, but this is not an absolute criterion. Tongue fasciculations have been reported in 56% to 61% of patients (180), so the absence of this finding does not necessarily exclude the disease. In one series (180), DTRs were absent in all four extremities in 74% of cases. Thus, the preservation of DTRs does not exclude the diagnosis of SMA. Appendicular muscle fasciculations and distal tremor are also associated examination findings. Extraocular muscles are spared, as are the myocardium. Mild to moderate hip flexion, knee flexion, and elbow flexion contractures may be observed in some patients along with wrist contractures and ulnar drift of the fingers. Severe arthrogryposis is not typically observed.

Diagnosis is confirmed by a consideration of clinical findings, molecular genetic studies and, occasionally, electrodiagnostic studies. Muscle biopsy is generally not required to confirm the diagnosis.

In a large series from Germany (167), 197 patients classified as type I (never sits alone) had the following survival probabilities: 32% at age 2; 18% at age 4; 8% at age 10; and 0% at age 20.

Spinal Muscular Atrophy II

Spinal muscular atrophy II disease onset is usually more insidious than that of SMA I. The findings of generalized hypotonia, symmetrical weakness, and delayed motor milestones are hallmarks of SMA II. Weakness also involves proximal muscles more than distal muscles, and lower extremity more than upper extremity. A fine tremor of the fingers and hands occurs in a minority of patients. This "polyminimyoclonus" may be attributed to spontaneous, repetitive rhythmical discharges by the motor neurons that innervate a large territory of muscle. Wasting tends to be more conspicuous in SMA II versus SMA I. DTRs are depressed and usually absent in the lower extremities. Appendicular or thoracic muscle wall fasciculations may be observed. Tongue fasciculations have been observed in 30% to 70% of SMA II patients (166,180,181). Progressive kyphoscoliosis and neuromuscular restrictive lung disease are almost invariably seen in the late first decade. Contractures of the hip flexors, tensor fasciae latae, hamstrings, triceps surae, and elbow and finger flexors are quite common. Hypotonic hip dislocations have been noted commonly in SMA II patients. Sensory examination is completely normal and extraocular muscles and the myocardium are spared. In a large series from Germany (167), of 104 cases classified as SMA II (sits alone, never walks), 98% survived to the age of 10 and 77% to the age of 20. Thus, a longer life span is possible with adequate supportive care.

SMA II is a slowly progressive condition affecting proximal musculature more than distal. The calculated grade of progression for SMA may be less than one-half manual muscle testing units decline per decade (86). Longitudinal series of 12 to 39 months' duration have shown essentially stable strength measurements but slow loss of function (182,183).

Pathologic changes on muscle biopsy have been consistent with hypotrophic change in fetal muscle development. Other changes are consistent with a more active denervating process. Thus, SMA includes a component of myofiber atrophy, comparable to that seen in other denervating diseases and is not a pure hypotrophic process occurring during early fetal development.

Spinal Muscular Atrophy III (Kugelberg–Welander Disease)

In more chronic SMA III, also referred to as Kugelberg–Welander Syndrome, weakness usually initially occurs between the ages of 18 months and late teens. Motor milestones may be delayed in infancy. Proximal weakness is observed with the pelvic girdle being more affected than the shoulder girdle (86). There is an exaggerated lumbar lordosis and anterior pelvic tilt owing to hip extensor weakness. There is also a waddling gait pattern with pelvic drop and lateral trunk lean over the stance phase side, secondary to hip abductor weakness. If ankle plantar flexion strength is sufficient, the patients may show primarily forefoot or toe contact and no heel strike similar to patients with Duchenne dystrophy. This is a compensatory measure for knee extensor weakness to maintain a stabilizing knee extension moment at the knee. The patient may exhibit a Gower's sign when arising from the

floor; stair climbing is difficult due to hip flexor weakness. Facial weakness is sometimes noted. Fasciculations are noted in about half of the patients (166) and are more common later in the disease course. Fasciculations in the limb muscles and thoracic wall muscles are common. Calf pseudohypertrophy has been occasionally noted, but wasting of affected musculature is more prominent. DTRs are diminished and often become absent over time. Contractures are generally mild as long as patients remain ambulatory. Scoliosis may be observed in SMA III, but it occurs less frequently and is less severe than scoliosis and SMA II. While no survival data exists for patients with SMA III, cases have been followed into the eighth decade without mechanical ventilation (86,167). Ventilatory failure due to neuromuscular restrictive lung disease is a rare event in SMA III, occurring only in adulthood (86,184).

Zerres and Rudnik-Schoneborn (167) have proposed further subtypes, including SMA IIIa (walks without support; age of onset less than 3 years) and SMA IIIb (walks without support; age of onset 3–30 years). In their series, only 44% of SMA "IIIa" patients remained ambulatory 20 years after onset of weakness, whereas 89% of "IIIb" patients remained ambulatory after a similar 20-year duration.

Therapeutic Strategies in Predominantly Proximal SMA. Single nucleotide polymorphisms (SNPs) in exon 7 of SMN2 cause a high proportion of exon 7 skipping during pre-mRNA splicing leading to an unstable truncated SMN protein. As a result, small levels of functional SMN protein are produced by the SMN2 gene. Restoring the splicing to achieve exon 7 inclusion has the potential to produce sufficient quantities of functional protein to compensate for the defect of SMN protein (185,186). Splice intervention therapies to promote exon 7 retention and increase amounts of full-length SMN2 transcript offer great potential as a treatment for SMA patients. Several splice silencing motifs in SMN2 have been identified as potential targets for antisense oligonucleotide mediated splice modification. A strong splice silencer is located downstream of exon 7 in SMN2 intron 7. Intrathecal antisense oligonucleotides targeting this motif have promoted SMN2 exon 7 retention in the mature SMN2 transcripts, with increased SMN expression detected in SMA fibroblasts. Systematic optimization of phosphorodiamidate morpholino oligonucleotides (PMOs) promotes exon 7 retention to levels that rescue the phenotype in a severe mouse model of SMA after intracerebroventricular (ICV) delivery. Furthermore, the PMO gives the longest survival reported to date after a single dosing by ICV (187,188). Clinical trials in humans with SMA are under way. Results of an open-label, escalating dose study to assess the safety, tolerability, and dose range finding of a single intrathecal dose of ISIS-SMN_{Rx} in patients with SMA, showed that ISIS-SMN_{Rx} was well tolerated when administered intrathecally as a single dose directly into the spinal fluid and that no safety concerns related to the drug were identified. Concentrations of ISIS-SMN_{Rx} measured in cerebral spinal fluid were consistent with levels predicted from preclinical studies, indicating that the drug half-life in nervous system tissues is very long and that dosing once every six to nine months is feasible. Although the study was not designed to provide evidence of functional activity, clinically significant, dose-dependent improvements in the Hammersmith Functional Motor Scale-Expanded (HFMSE), a measure of muscle function, were observed in these children.

Through chemical screening and optimization, PTC Therapeutics, Inc. and Roche Pharma Research and Early Development have identified orally available small molecules that shift the balance of SMN2 splicing toward the production of full-length SMN2 messenger RNA with high selectivity. Administration of these compounds to Δ7 mice, a model of severe SMA, led to an increase in SMN protein levels, improvement of motor function, and protection of the neuromuscular circuit. These compounds also extended the life span of the mice. Selective SMN2 splicing modifiers may have therapeutic potential for patients with SMA (189). A clinical trial of the lead compound is being planned.

Adeno-associated virus type 9 (AAV9) is a powerful tool for delivering genes throughout the CNS. To specifically target the CNS, Kaspar and colleagues have explored AAV9 delivery of the SMN gene to CSF. CSF injection efficiently targeted motor neurons, and restricted gene expression to the CNS, providing an alternate delivery route and potentially lower manufacturing requirements for older, larger patients (190). Their findings support the use of AAV9 for gene transfer to the CNS for disorders such as SMA in pediatric populations.

Distal Spinal Muscular Atrophy

Distal SMA is an increasingly recognized group of rare diseases with varied genetic etiologies. Over 20 distinct genetic subtypes have been identified. The patients may be clinically misdiagnosed as having CMT due to the distal weakness of the foot and hand intrinsics. Some subtypes of distal SMA have predominant upper extremity involvement. Other variants of distal SMA may present initially with distal lower extremity weakness. Sensory function is always normal clinically and electrodiagnostically. The course is usually slowly progressive although some patients may experience a prolonged period of stability. Other associated features in some subtypes include vocal cord paralysis and diaphragm weakness. Some subtypes have associated pyramidal signs.

Juvenile Segmental SMA (Benign Focal Amyotrophy; Hirayama Disease)

This disease was originally described by Hirayama as a slowly progressive focal motor neuron disease affecting the upper extremities. Most cases occur on a sporadic

basis. The onset of this syndrome is typically between 15 and 25 years with a range of 2 to 30. Wasting and weakness develop segmentally in C8-T1 hand and forearm muscles, unilaterally and often but not always the dominant extremity. Sensation is completely normal. The disease progresses to more proximal upper extremity muscles. The lower extremities are never affected and typically the disease progression plateaus after 2 to 6 years. Symptoms worsen in the cold ("cold paresis"). Tremor may occur due to distal weakness. Hyperhidrosis of the involved limb is a common complaint. Reflexes are typically spared but not brisk. EMG studies are consistent with an anterior horn cell disorder. MRI abnormalities of the cervical spinal cord (segmental atrophy, stenosis, or foraminal narrowing) have been described in a proportion of patients. The disease is more common in Asian populations.

(Fazio–Londe Disease) Progressive Bulbar Paralysis of Childhood

Fazio–Londe disease or progressive bulbar paralysis of childhood is a progressive bulbar paralysis which is probably genetically transmitted. This is a disorder of bulbar motor neurons. Patients present with cranial nerve findings including ptosis, facial weakness, dysphagia, normal hearing, and respiratory stridor. They may show hyperreflexia. Dominant transmission is rare. One group with recessive inheritance has early onset in infancy and rapid progression with death from respiratory failure less than 2 years from the age of onset. Another group with recessive inheritance shows later onset (3–12 years), less respiratory involvement, but slowly progressive dysarthria; dysphagia and facial weakness. These patients may have progressive motor neuron disease with primary involvement of the anterior horn cells in the cervical and upper thoracic core segments. In addition, there may be widespread degenerative changes in the brainstem. Of cranial nerves, cranial nerve VII is almost always affected. These patients develop dysphagia secondary to cranial nerve XII involvement. The nuclei of cranial nerves III, IV, VI, and X may also be involved; however, clinical impairment of extraocular movement is rare.

SPINOCEREBELLAR DEGENERATION DISEASES

Friedreich's Ataxia

Friedreich's ataxia is a spinocerebellar degeneration syndrome with the onset of symptoms before age 20 years. This autosomal-recessive condition has been linked in one subtype to chromosome 9q13-21.1 (FRDA) with the protein implicated being termed "frataxin." A second subtype referred to as FRDA2 is linked to chromosome 9p23-p11.

The incidence of Friedreich's ataxia is 1 in 25,000 to 50,000. Carrier frequency is 1 in 60 to 110. The age of onset is usually less than 20 years, typically around puberty with a range from 2 to 25 years. Obligate signs and symptoms include progressive ataxic gait, cerebellar dysfunction with tremor and dysmetria, dysarthria, decreased proprioception or vibratory sense (or both), muscle weakness, and absent DTRs. Other common signs include cavus foot deformity, cardiomyopathy, scoliosis, and upper motor neuron signs, such as a Babinski sign and spasticity. Weakness is progressive, and it affects lower extremities and small muscles in hands and feet. Sensory loss is typical and especially affects vibration and joint position sensation. Tendon reflexes are often absent. An occasional patient may have chorea without ataxia. With electrodiagnostic studies, sensory nerve potentials may be absent or reduced. Progression is slow, and the mean time to wheel chair is 15 years of age and death from cardiomyopathy ranges from the third to the seventh decade.

The prevalence of scoliosis approaches 100% but some cases have more severe progressive spinal deformity than others. Those Friedreich's ataxia cases with onset of disease before the age of 10 years generally have more severe progressive scoliosis. Those with the onset of disease during or after puberty have later-onset spinal deformity which may not require surgical intervention.

Frataxin is a mitochondrial protein located on the inner mitochondrial membrane. It is likely required for maintenance of mitochondrial genome and it is involved in iron homeostasis and iron transport into mitochondria. Idebenone is a power antioxidant and a synthetic analogue of coenzyme Q. It may improve iron homeostasis and mitochondrial function in Friedreich's ataxia. In randomized clinical trials, longer term idebenone treatment has been shown to prevent progression of cardiomyopathy and cardiac hypertrophy in both pediatric and adult patients with Friedreich's ataxia. Its stabilizing effect on neurologic dysfunction has been shown to be present only in the pediatric population, mainly before puberty. This suggests that the age at which idebenone treatment is initiated may be an important factor in the effectiveness of the therapy (191). The findings of the open-label IONIA-E study combined with the double-blind IONIA study indicate that idebenone at a dose of 1,350/2,250 mg/day may offer a therapeutic benefit to pediatric FRDA patients by stabilizing the overall neurologic function and improving fine motor skills and speech (192,193).

Other Hereditary Ataxias

The hereditary ataxias are a group of genetic disorders characterized by slowly progressive incoordination of gait and often associated with poor coordination of hands, speech, and eye movements. Frequently, atrophy of the cerebellum occurs. The hereditary ataxias are categorized

by mode of inheritance and causative gene or chromosomal locus. The genetic forms of ataxia are diagnosed by family history, physical examination, and neuroimaging. Molecular genetic tests are available for the diagnosis of many but not all **spinocerebellar ataxias (SCA)**. At least 28 genetically distinct autosomal-dominant SCA subtypes and four other autosomal-dominant hereditary ataxias have been identified. Childhood onset has been found commonly in SCA7, SCA13, SCA17, and SCA21 and more rarely in SCA1, SCA2, SCA3, SCA5, and SCA22. Other pedigrees of SCA with childhood onset have been identified in only single families. Autosomal-dominant episodic ataxia 1 (EA1) with episodic attacks of myokymia and ataxia and linkage to chromosome 12p13.3 has onset in the first decade. At least seven autosomal-recessive ataxias in addition to Friedreich's ataxia have been identified, most of which have childhood onset. The most common of these is **ataxia telangiectasia**, with linkage to chromosome 11q22.3, which presents in the first decade with ataxia, dysarthria, ocular telangiectasias, immune deficiency, and risk of cancers.

MANAGEMENT OF CHILDHOOD NEUROMUSCULAR DISEASES

Diseases affecting the lower motor neuron, including those primarily affecting anterior horn cell, peripheral nerve, NMJ (presynaptic or postsynaptic), or muscle ultimately lead to progressive loss of functional muscle fiber over time. This loss of functional muscle fiber may lead to progressive weakness, decreased endurance, limb contractures, spine deformity, body composition changes, decrease in mobility, decreased pulmonary function, and occasionally cardiac impairment if the myocardium is affected. Genetic defects causing CNS structural protein alterations may lead to intellectual impairment. Rehabilitation approaches directed at improving impairment and/or resultant disability may substantially improve the quality of life and community integration of children with NMDs. The following discussion emphasizes general principles in the rehabilitation management of childhood NMD with several specific conditions used to illustrate key concepts.

EXERCISE IN NEUROMUSCULAR DISEASE

Exercise prescriptions and recommendations in childhood NMD need to consider the specific disease condition as well as the developmental and maturational status of the child.

Strengthening Exercise in Rapidly Progressive Disorders

The more rapidly progressive neuromuscular disorders of childhood generally include the dystrophic myopathies. The inherent instability of the sarcolemmal membrane predisposes to membrane injury due to mechanical loads. Theoretically, eccentric or lengthening contractions produce more mechanical stress on muscle fiber than concentric or shortening contractions. Indeed, many of the muscle groups, which show the greatest weakness early in the course of DMD, are muscle groups which perform a great deal of eccentric activity such as the hip extensors, knee extensors, and ankle dorsiflexors. In addition, lower extremity muscles in this population experience more mechanical loads than upper extremity muscle groups, and weakness in the lower extremities generally predates weakness in the upper extremities. Edwards and colleagues (194) proposed that routine eccentric contractions occurring during gait are a likely source of the pattern of weakness typically seen in myopathies.

There may be increased weakness following strengthening exercise in DMD (195). There are other instances which have raised concerns regarding overwork weakness in dystrophic myopathies. The dominant upper limb has been found to be weaker in persons with FSHD muscular dystrophy than the nondominant, providing circumstantial evidence for overwork weakness (72,196). A single subject with scapuloperoneal muscular dystrophy had a reversal of rapid strength decline after reducing daily physical activity. Other studies evaluating strengthening intervention in DMD subjects have shown maintenance of strength or even mild improvement in strength over the period of the investigation. However, these studies are limited by use of primarily nonquantitative measures (197), lack of a control group (198), and use of the opposite limb as a control without considering the effects of cross training (199). Animal work utilizing dystrophic dogs has shown significant increases in CK values immediately following exercise.

No systemic studies using the DMD population have shown any deleterious effects of mild or moderate resistance exercise. The assisted cycling protocol described above did not result in any overwork weakness in DMD (76). Based on the theoretic susceptibility of the dystrophin-deficient sarcolemmal membrane to mechanical injury, and the relative paucity of investigations, it is prudent to recommend a submaximal strengthening program in DMD and other rapidly progressive dystrophic disorders. A great concern is how to incorporate these activities effectively into the daily routine of the child, avoiding the use of mundane and tedious regimens that employ progressive resistive exercises. Incorporation of the activity into recreational pursuits and aquatic-based therapy are probably the most reasonable approaches for the preadolescent child.

Strengthening Exercise in Slowly Progressive Neuromuscular Diseases

Only supervised strengthening programs in this population have been advocated. Recently, a moderate resistance

home exercise program (using a less supervised approach) was devised that demonstrated similar strength gains in both NMD patients and normal control subjects without evidence of overwork weakness (200). Based on this encouraging result, the home program was advanced to high resistance training in similar subjects without apparent additive beneficial effects; in fact, eccentrically measured elbow flexor strength actually decreased significantly (201).

Based on the above investigations, the author believes that there is adequate evidence to generally advocate a submaximal strengthening program for persons with slowly progressive NMD. There seems to be no additional benefit to high-resistance, low-repetition training sets, and the risk of actually increasing weakness becomes greater. Improvement in strength will hopefully translate to more functional issues such as improved endurance and mobility.

Aerobic Exercise in Neuromuscular Disease

Aerobic exercise refers to rhythmic, prolonged activity of the level sufficient to provide a beneficial training stimulus to the cardiopulmonary and muscular systems but below the threshold where anaerobic metabolism of fuels is the primary source of energy. The response of normal skeletal muscle to this type of training includes: increased capillary density in the muscle to improve substrate transfer, increased skeletal muscle mitochondrial size and density, higher concentrations of skeletal muscle oxidative enzymes, and improvement in utilization of fat as an energy source for muscular activity. Patients with NMD have a diminished capacity for exercise. Children with DMD have been demonstrated to have low cardiovascular capacity and peripheral oxygen utilization with higher resting heart rate compared with controls (202). Physical ability and exercise capacity are more likely to be limited by muscle strength than by deterioration of cardiorespiratory function. In a recent study using a home-based aerobic walking program, slowly progressive NMD subjects showed modest improvement in aerobic capacity without evidence of overwork weakness or excessive fatigue (203). The benefits of assisted cycling in DMD were previously discussed. It is likely that alternative exercise approaches, such as aquatic-based therapy, will need to be utilized in children with more severe NMDs who are nonambulatory and have less than antigravity muscle strength.

One group recently studied the effect of endurance training on conditioning and strength in adult BMD. Eleven patients with BMD and seven matched, healthy subjects cycled 50- and 30-min sessions at 65% of their maximal oxygen uptake (VO_2max) over 12 weeks, and six patients continued cycling for 1 year. Endurance training for 12 weeks significantly improved VO_2max by 47 +/- 11% and maximal workload by 80 +/- 19% in patients. This was significantly higher than in healthy subjects (16 +/- 2% and 17 +/- 2%). CK levels did not increase with training. Strength in muscles involved in the cycle exercise (knee extension, and dorsi- and plantar flexion) increased significantly by 13% to 40%. Cardiac pump function, measured by echocardiography, did not change with training. All improvements and safety markers were maintained after 1 year of training. Endurance training was demonstrated to be a safe method to increase exercise performance and daily function in patients with BMD and the findings support an active approach to rehabilitation of patients with BMD (204).

MANAGEMENT OF LIMB CONTRACTURES AND DEFORMITY

The management of limb contractures in progressive NMD and the role of stretching, orthotics, and surgery have recently been comprehensively reviewed (48). Contracture is defined as the lack of full passive ROM due to joint, muscle, or soft tissue limitation. Contractures may be arthrogenic, soft tissue or myogenic in nature, and a combination of intrinsic structural changes of muscle and extrinsic factors leads to myogenic contractures in selected NMD conditions. These factors include the following: (a) degree of fibrosis and fatty tissue infiltration; (b) static positioning and lack of full active and passive ROM; (c) imbalance of agonist and antagonist muscle strength across the joint; (d) lack of upright weight-bearing and static positioning in sitting; (e) compensatory postural changes used to biomechanically stabilize joints for upright standing; and (f) functional anatomy of muscles and joints (multijoint muscle groups in which the origin and insertion cross multiple joints). In general, dystrophic myopathies have a high degree of fibrosis and fatty infiltration, placing these patients at higher risk for contractures. Significant contractures have been most commonly identified in DMD, BMD, EMD, CMD, autosomal-recessive LGMD, FSHD muscular dystrophy, myotonic muscular dystrophy, HMSN, and SMA.

Contractures and progressive NMD conditions should be managed with the following concepts in mind:

1. Prevention of contractures requires early diagnosis and initiation of physical medicine approaches such as passive ROM and splinting while contractures are still mild.
2. Contractures are inevitable in some NMD conditions, such as DMD.
3. Advanced contractures become fixed and show little response to stretching programs.
4. A major rationale for controlling contractures of the lower extremity is to minimize the adverse effect of contractures on independent ambulation. However, the major cause of wheelchair reliance in NMD is generally weakness, not contracture formation.
5. Static positioning of both upper and lower extremity joints in patients with weak musculature is the most important cause of contracture formation.

6. Passive stretching for control of lower limb contractures is most successful in ambulatory patients with early mild joint contractures.
7. Upper extremity contractures may not negatively impact the function if they are mild.
8. Joint ROM should be monitored regularly by physical therapists and occupational therapists using objective goniometric measurement.

Principle therapy modalities must be regularly carried out to prevent or delay the development of lower extremity contractures for those at risk for musculoskeletal deformity. These include: (a) regularly prescribed periods of daily standing and walking if the patient is functionally capable of being upright; (b) passive stretching of muscles and joints with a daily home program; (c) positioning of the leg to promote extension and oppose joint flexion when the patient is non-weight-bearing through the lower extremities; and (d) splinting, which is a useful measure for the prevention or delay of ankle contracture.

In the upper extremity, elbow flexion contractures in dystrophic myopathies may occur soon after transition to the wheelchair, secondary to static positioning of the arms and elbow flexion on the armrests of the wheelchair (15). Other associated deformities in DMD and other dystrophic myopathies include forearm pronator tightness and wrist flexion–ulnar deviation in the later stages of the disease. The regular palm-down position of the hand increases the occurrence of forearm pronator contracture. Mild elbow flexion contractures of 15 degrees or less are of no functional consequence to the patient using crutches or a wheelchair. Contractures of the elbows over 30 degrees can interfere with the use of crutches in ambulatory patients with NMD. Severe elbow flexion contractures of greater than 60 degrees are associated with decreased distal upper extremity function and produce difficulty when dressing.

Passive stretching of the elbow flexors may be combined with passive stretching into forearm supination to help prevent contractures. Prophylactic occupational therapy management of the wrist and hand is recommended in NMD to slow down the development of contractures and to maintain fine motor skills. Daily passive stretching of the wrist flexors and intrinsic and extrinsic muscles of the hand and wrist is recommended, as are active ROM exercises for the wrist and long finger flexors. Nighttime resting splints, which promote wrist extension, metacarpophalangeal extension, and proximal interphalangeal flexion are recommended. Daytime positioning should emphasize wrist and finger extension, but any splinting should not compromise sensation or function.

Shoulder contractures are less problematic in patients with profound proximal muscle weakness. Combined shoulder internal rotation, adduction contracture, and elbow flexion deformity may interfere with self-feeding. Severe shoulder internal rotation deformities may complicate dressing, produce pain on passive ROM, and cause pain during sleep.

BRACING/ORTHOTIC MANAGEMENT AND ORTHOPEDIC SURGICAL MANAGEMENT OF LIMB DEFORMITY

Management in Neuromuscular Diseases With Proximal Weakness.

The prototypical disorder in which bracing and surgical management of contractures for prolonged ambulation has been applied is DMD. In this population, wheelchair reliance is imminent when knee extension strength becomes less than antigravity and time to ambulate 10 meters or 30 feet is greater than 12 seconds (15). A number of principles should be emphasized for these populations. First, with an appropriate and aggressive home-based therapy program, equinovarus contractures generally are absent or very mild in DMD at the time walking ability ceases (15). In addition, hip and knee flexion contractures are also absent or extremely mild in ambulatory DMD patients at the time of transition to a wheelchair. The wide-based Trendelenburg gait, exhibited by these patients with gluteus medius weakness places the hip in an abducted position, leading to iliotibial band contractures. The late phase of ambulation often is associated with more marked joint contractures involving the iliotibial bands and heel cords, because DMD patients spend more time sitting and less time standing. The release of contractures at both the heel cord and iliotibial band generally is necessary to obtain successful knee ankle foot orthosis (KAFO) bracing (205–207). Other authors have reported bracing of DMD patients without surgical release of the iliotibial bands (208,209). Hip and knee flexion contractures generally are not severe enough to interfere with bracing at the time of transition to a wheelchair (15). The iliotibial band contractures may be released with a low Young fasciotomy and a high Ober fasciotomy.

The ankle deformity may be corrected by either a tendo-Achilles lengthening (TAL) or a TAL combined with a surgical transfer of the posterior tibialis muscle tendon to the dorsum of the foot. The posterior tibialis tendon transfer corrects the equinovarus deformity but prolongs the time in a cast and recovery time, and it increases the risks of prolonged sitting.

Orthopedic surgical release of these contractures allows the DMD patient to be braced in lightweight polypropylene KAFOs with the sole and ankle set at 90 degrees, drop-lock knee joints, and ischial weight-bearing polypropylene upper thigh component. DMD patients who are braced may or may not require a walker for additional support. At times, DMD patients who have had excellent home stretching programs can be placed immediately into KAFO bracing without surgical tenotomies.

While DMD subjects are still ambulating independently without orthotics, they often use their ankle equinus posturing from the gastrocnemius-soleus group to create a knee extension moment at foot contact, thus stabilizing the knee when the quadriceps muscle is weak. Several authors have cautioned against isolated heel cord tenotomies while DMD patients are still ambulating independently. Overcorrection of the heel cord contracture in a DMD patient may result in immediate loss of the ability to walk without bracing unless the quadriceps are grade 4 or better (210).

The duration of ambulation in DMD has been successfully prolonged by prompt surgery and bracing, immediately implemented following loss of independent ambulation. Generally, the gains in additional walking time have been variable, but generally reported between 2 and 3 years.

Long-term benefits of prolonged walking include decreased severity of heel cord and knee flexion contractures at age 16 (211). This may ultimately improve shoe wear tolerance and foot positioning on the wheelchair leg rests. Prolonged ambulation by lower extremity bracing in DMD has never been documented to be an independent factor in the prevention of scoliosis. Disadvantages of braced ambulation center around the excessive energy cost of braced ambulation and safety concerns in the event of falls. DMD subjects with KAFO bracing usually need gait training by physical therapy, and they need to be taught fall techniques.

Weakness is the major cause of loss of ambulation in DMD, not contracture formation. Thus, the primary indication of orthopedic surgical tenotomies and posterior tibialis tendon transfers likely is the provision of optimal alignment for KAFO bracing. Little evidence supports the efficacy of early prophylactic lower extremity surgery in DMD for independently producing prolonged ambulation (15,210,212).

In general, with the increased utilization of corticosteroids in DMD, there has been a trend over the past two decades toward reduced use of lower extremity surgery and long leg bracing to prolong ambulation.

Management of NMD Patients With Distal Lower Extremity Weakness

Ankle dorsiflexors are often clinically weaker than ankle plantar flexors in NMD because of selective involvement of the peroneal nerve in many neopathies and isolated anterior and lateral compartment weakness in several myopathic conditions such as FSHD, scapuloperoneal distribution LGMD, DMD, and EMD. Ankle foot orthoses (AFOs) often are used for patients with distal weakness. AFOs are generally contraindicated in situations where NMD patients utilize equinus posturing with forefoot initial contact to maintain a knee extension moment in the setting of quadriceps weakness. Heel cord contractures may need to be surgically lengthened to allow for AFO or KAFO bracing. Cavus feet are common in peripheral neuropathies. Intrinsic muscle weakness of the foot results in hyperextension at the metatarsophalangeal joints and flexion at the interphalangeal joints with resultant claw toe deformities. This constellation of deformities may cause difficulty in walking, lack of balance and painful callosities. Treatment of the cavus foot depends on the patient's age, flexibility of the foot, bony deformity, and muscle imbalance. A supple foot can be managed nonoperatively by serial casting in a walking cast, followed by an AFO with a solid ankle in neutral position and a lateral heel wedge if significant hindfoot varus exists. Fixed soft tissue or bony deformity may require orthopedic surgery to produce a plantigrade foot. In skeletally immature children, triple arthrodesis is contraindicated. Triple arthrodesis should only be considered as a salvage procedure for severe heel varus and severe mid-foot deformity with the goal being achievement of hindfoot stability in a skeletally mature patient.

MANAGEMENT OF SPINAL DEFORMITY

The management of spinal deformity in progressive NMDs has recently been reviewed (213). Severe spinal deformity and progressive NMD lead to multiple problems, including poor sitting balance, difficulty with upright seating and positioning, pain, difficulty in parental or attendant care, and potential exacerbation of underlying restrictive respiratory compromise (Figure 18.14). Severe scoliosis and pelvic obliquity can, in some instances completely preclude upright sitting in a wheelchair. Populations at risk for scoliosis include DMD, autosomal-recessive LGMD, CMD, FSHD muscular dystrophy, congenital myotonic muscular dystrophy, SMA II and III, and Friedreich's ataxia. While previous estimates of incidence of severe scoliosis in DMD approached 80% to 90%, recent evidence suggests that corticosteroids (specifically deflazacort) may significantly decrease the incidence of severe progressive scoliosis in DMD (51).

Close clinical monitoring is essential for children with NMD at risk for scoliosis. Curves may progress rapidly during the adolescent growth spurt, and children need to be monitored every 3 to 4 months during this time with clinical assessment and spine radiographs if indicated. In addition, patients who are likely to require surgical arthrodesis at some point should be monitored with pulmonary function tests every six months. An FVC falling below 30% to 40% of predicted does not contraindicate surgery (61), but is associated with increased perioperative morbidity and the likely need for prolonged noninvasive ventilatory support during the postoperative recovery period (214). Thus, there is often a critical window of time where the spinal deformity is evident and likely to continue to progress, and the restrictive

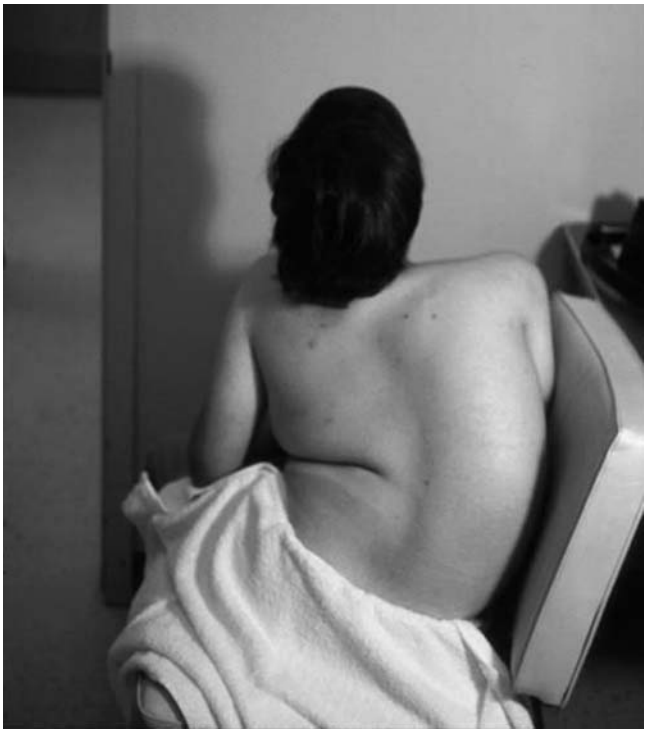


FIGURE 18.14 Scoliosis in Duchenne muscular dystrophy compromising long-term comfortable supported sitting in a power wheelchair.

lung disease is not of a severity which would contraindicate surgery or be associated with perioperative complications.

The management of spinal deformity with orthotics is ineffective in DMD and does not change the natural history of the curve. Spinal orthoses are often reported to be uncomfortable and poorly tolerated by DMD patients. Further, vital capacity potentially can be lowered with constrictive orthoses. On the other hand, in NMDs, with spinal deformity beginning in the first decade of life, such as SMA, CMD, congenital myotonic muscular dystrophy, some congenital myopathies, and congenital myasthenic syndromes, spinal bracing is generally used to improve sitting balance in patients who are unable to walk. In addition, spinal orthotics are employed in these younger patients in an attempt to halt curve progression until children are 10 to 11 years of age when a single posterior spinal arthrodesis procedure is sufficient. Children younger than the age of 10 generally require both anterior and posterior spinal arthrodesis because of continued spinal growth which decreases in rate after age 11 to 12. If a younger child has a severe progressive curve and severely compromised pulmonary function, a posterior fusion may be considered with acceptance of the fact that some rotational "crank shaft deformity" will ensue.

Spinal arthrodesis is the only effective treatment for scoliosis in DMD, autosomal-recessive LGMD, CMD, congenital myotonic muscular dystrophy, SMA, and Friedreich's ataxia. The decision to pursue posterior spinal instrumentation involves a consideration of the severity of the restrictive lung disease, severity of the cardiomyopathy, severity and flexibility of the spinal deformity, and the likelihood that the spinal deformity will continue to progress. Surgical spinal arthrodesis should be deferred to a later date in marginally ambulatory patients with LGMD, CMD, FSHD and SMA type III as these individuals may use significant lumbar lordosis during gait to compensate for hip extensor weakness.

PROVISION OF FUNCTIONAL MOBILITY

Mobility assistive technology has recently been reviewed in progressive NMD (215). Generally, antigravity quadriceps are required for community ambulation in childhood NMD. Short distance ambulation may be achieved by some patients with more severe weakness using KAFO bracing with or without a walker. Such orthotic intervention is often provided to children with SMA type III, Severe Childhood Autosomal Recessive Muscular Dystrophy (SCARMD), CMD, DMD, and BMD during adulthood. Children with DMD SMA type II, CMD, congenital myopathies, some myasthenic syndromes, and more severe HMSNs utilize power mobility devices for functional mobility. Generally, children can be taught to safely operate a power wheelchair when they are at the developmental age of approximately 2 years (216,217). The initial power wheelchair prescription needs to consider the natural history of the NMD condition over the following five years as some children will subsequently develop the need for a power recline system and the chair needs to be able to accommodate such a recline or be retrofit. In more severe disability, the power wheelchair electronics should be sufficiently sophisticated to incorporate alternative drive control systems, environmental control adaptations, and possibly communication systems in patients who are unable to vocalize.

ROBOTICS AND ASSISTIVE TECHNOLOGY TO IMPROVE UPPER LIMB FUNCTION

The state of the art in therapeutic and assistive robots and orthoses for the upper and lower extremity in neuromuscular disorders has been recently reviewed by Rahman and colleagues (218). As an example, a novel, articulated upper extremity orthosis, the Wilmington Robotic EXoskeleton (WREX), helps people with NMD overcome upper limb movement deficits secondary to weakness. The WREX uses elastic bands to negate the effects of gravity; it allows a person with neuromuscular weakness to move their arm in three dimensions. The WREX can be fixed on a brace for ambulatory patients and on the wheelchair for nonambulatory patients. The WREX

provided an increase in functionality and improved the quality of life of the patients. The JACO™ robotic arm (Kinova; Innovations Health, Roseville, California) uses the joystick as a controller for patients with distal hand function allowing operation of a power chair. The arm is mounted on the chair and provides reach in multiple dimensions and functional grasp.

PULMONARY MANAGEMENT

Pulmonary complications are recognized as the leading cause of mortality in childhood NMD. Respiratory insufficiency in NMD results from a number of factors, including: (a) respiratory muscle weakness and fatigue; (b) alteration of respiratory system mechanics; and (c) impairment of a central control of respiration. Progressive muscle weakness and fatigue lead to restrictive lung disease and ultimately to hypoventilation, hypercarbia, and respiratory failure. Increase in elastic load on respiratory muscles occurs because of chest wall stiffness, airway secretions, and ineffective cough mechanism. This may result in atelectasis and increased airway resistance, and kyphoscoliosis can further alter respiratory mechanics. Defects in central control of respiration may be secondary to hypoxemia and hypercarbia, associated with severe restrictive lung disease. Significant nocturnal decreases in partial pressure of oxygen, as well as elevations in arterial partial pressure of carbon dioxide occur in more severe restrictive lung disease. Hypercapnia or hypoxemia occurring at night may have a role in reducing daytime central respiratory drive. A chronic increase in the bicarbonate pool may blunt the stimulus to breathe, generated by respiratory acidosis and perpetuates the hypercapnic state. Expiratory muscle weakness may produce ineffective cough, problems with clearance of secretions, and predispose patients to pulmonary infections.

Respiratory failure may present acutely or insidiously. Respiratory difficulties in the delivery room or early infancy may be seen in acute infantile type I SMA, myotubular myopathy, congenital hypomyelinating neuropathy, congenital infantile myasthenia, congenital myotonic muscular dystrophy, transitory neonatal myasthenia, and severe neurogenic arthrogryposis. In most other childhood NMDs, the respiratory insufficiency develops more insidiously unless an acute decompensation occurs from an event such as an aspiration episode or acute onset of weakness, as seen in Guillain–Barré syndrome, botulism, and myasthenic syndromes. Signs and symptoms of significant respiratory difficulties may include subcostal retractions, accessory respiratory muscle recruitment, nasal flaring, exertional dyspnea or dyspnea at rest, orthopnea, generalized fatigue, and paradoxic breathing patterns. A history of nightmares, morning headaches, and daytime drowsiness may indicate nocturnal hypoventilation with sleep disordered breathing. Pulmonary function tests have been used to help in the decision-making process regarding the institution of mechanical ventilation. In a study of 53 patients with proximal myopathy, hypercapnia occurred when the MIP was less than 30% of predicted and when vital capacity was less than 55% of predicted (219). Other authors (220,221) have noted lower values for vital capacity measurements in their patients with DMD at the time they require institution of mechanical ventilatory support. Hahn and colleagues (222) have reported the predicted value of maximal static airway pressures in predicting impending respiratory failure. Splaingard (223) reviewed a series of 40 patients with a diverse group of NMD conditions. They noted that all their patients who required mechanical ventilation had a vital capacity of 25% or less with at least one of the following associated findings: (a) $PaCO_2$ greater than 55 mmHg; (b) recurrent atelectasis or pneumonia; (c) moderate dyspnea at rest; or (d) congestive heart failure.

Noninvasive forms of both positive and negative pressure ventilation are being increasingly applied to children with NMDs (Figure 18.15). Initially, patients may require ventilatory support for only part of the day. Noninvasive nocturnal ventilation has become a widely accepted clinical practice, providing ventilatory assistance for patients while sleeping and allowing them to breathe on their own during the day. Intermittent ventilation may ameliorate symptoms of respiratory failure, reduce hypercarbia, increase oxygenation (even during periods off the ventilator), and prolong survival in patients with NMD. The long-term use of noninvasive ventilation (see Figure 18.12) may be associated with fewer complications than ventilation via a tracheostomy; however, bulbar muscle function should be adequate for safe swallowing (184). Ventilatory support has allowed prolonged survival and acceptable quality of life in SMA I, SMA II, and DMD (221,224–226).

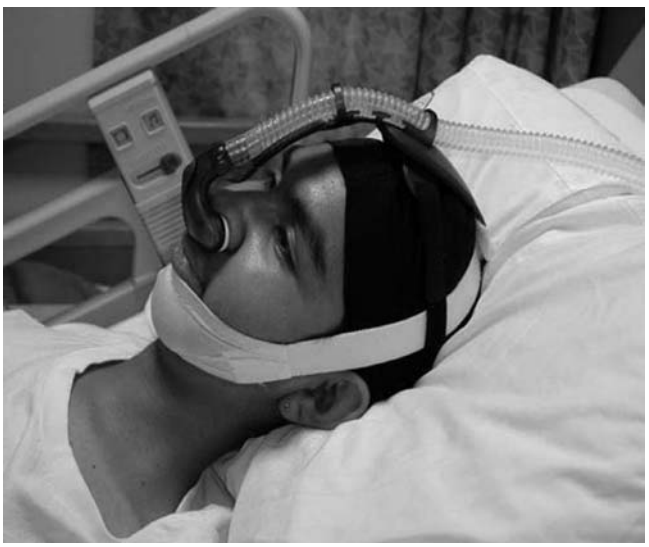


FIGURE 18.15 Noninvasive ventilatory support using BIPAP and nasal pillows mask interface in young adult with DMD.

Improved pulmonary toilet and clearance of secretions can be achieved with assisted cough, deep breathing and set-up spirometry, percussion and postural drainage, and in more severe cases, the additional use of interpulmonary percussive ventilation (IPV), given two to three times daily.

NUTRITIONAL MANAGEMENT

Management of Swallowing Problems

Involvement of palatal and pharyngeal muscles may produce dysphagia. Patients at particular risk include those of SMA, myasthenia gravis, congenital myasthenic syndromes, congenital myopathies, such as myotubular myopathy, oculopharyngeal muscular dystrophy, late-stage DMD, and late-stage SCARMD. The presence of dysphagia in NMD patients has been documented by others (37,227). The function of the swallowing mechanism is best evaluated with a fluoroscopic video dynamic swallowing evaluation. DMD patients have a high prevalence of dysphagia during the late stages of the disease (37). DMD patients may also rarely develop acute gastric dilation secondary to gastric paresis (228). Bulbar dysfunction and/or respiratory distress may affect feeding in SMA patients. In SMA I, therapeutic modifications may include the use of a premature baby nipple with a large opening, use of proper head and jaw position, along with a semireclined trunk position and the use of frequent small feedings to minimize fatigue. These larger bolus feeds may distend the stomach and encroach on the diaphragm, thus affecting respiratory status. Improved nourishment in SMA leads to a feeling of well-being and therefore a better quality of life. Poor nutritional status, labored feeding, and/or symptoms of dysphagia are indications for initiation of supplemental enteral feedings via a nasogastric tube or gastrostomy. Gastroesophageal reflux with risk of aspiration may be an indication for placement of a gastrojejunostomy tube.

Energy and Protein Supplementation

Severe deficits in energy and protein intake have been documented in DMD (35,36) during the second decade. Substantial weight loss has been documented in DMD to occur between the ages of 17 and 21 (see Figure 18.16). Protein and calorie needs in DMD may be approximately 160% of that required for able-bodied adolescents. Beneficial effects in weight gain, anthropometric measurements, and nitrogen balance were documented for DMD patients aged 10 to 20 years, subsequent to a 3-month nutritional supplementation which consisted of an additional 1,000 kcals and 37.2 grams of protein (229). The positive effects on metabolism observed in this study warrant further investigation.

Branched-Chain Ketoacid Supplementation

Based on the observations that muscle protein degradation is accelerated in DMD, and administration of branched-chain ketoacids reduces protein breakdown in fasting obese subjects, Stewart and colleagues (230) conducted a trial of branched-chain ketoacid supplementation. The ketoacids of the branched-chain amino acids leucine, valine, and isoleucine were administered orally as ornithine salts at a dosage of 0.45 gm/kg body weight/day for 4 days in nine boys with DMD, aged 5 to 9 years. An equivalent amount of protein was removed from the diet during this time. A small but significant reduction in muscle protein degradation was observed as a result of the treatment, and no negative effects were noted. The results warrant further investigation regarding the effects of longer term branched-chain ketoacid supplementation on muscle protein degradation.

Weight Reduction

DMD patients typically gain excessive weight between 9 and 13 years of age, subsequent to the onset of wheelchair

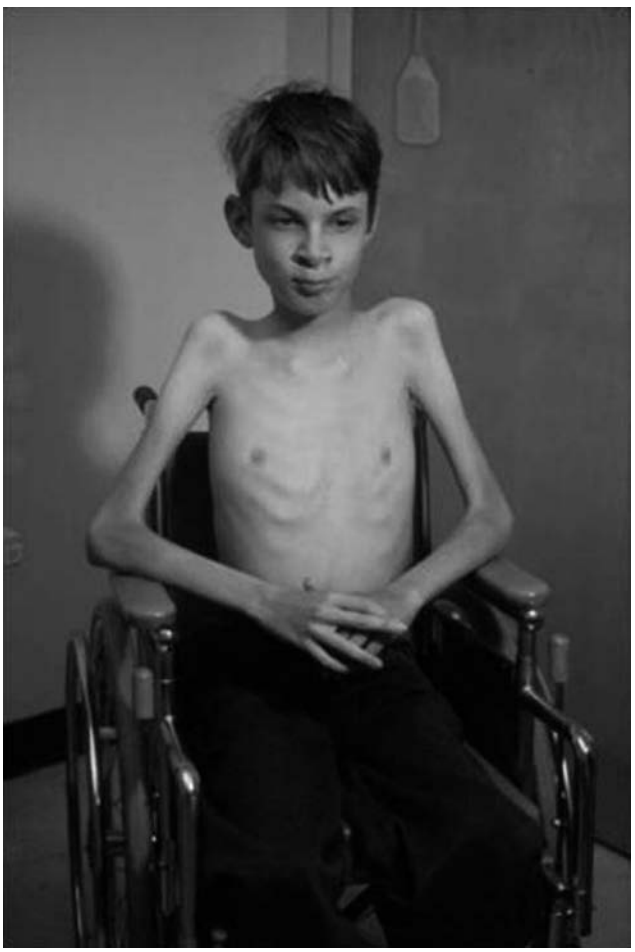


FIGURE 18.16 Severe weight loss in young adult with DMD.

reliance. This is likely due to a reduction in total daily energy expenditure with increased sedentary existence. Edwards and colleagues (231) demonstrated weight reduction through a medically supervised decrease in energy intake could be achieved successfully in DMD without compromising skeletal muscle mass. Obesity has also been observed in SMA III patients and has been attributed to a relatively sedentary lifestyle. Increased adiposity has been documented in adults with slowly progressive NMDs (232). Approaches to weight reduction in slowly progressive NMD patients have been previously reviewed (233).

BONE HEALTH IN NMDS

Joyce and colleagues (234) have reviewed the recent literature regarding bone health as it relates to the patient living with NMD. Poor bone health is common in patients with NMD and is the cause of significant morbidity, including increased fracture rates and severe scoliosis. Bone health depends on a complex interplay of both local and distant mechanisms, including genetic, endocrine, neurologic, and lifestyle factors. Osteoporosis in NMD may be due to disease-specific pathophysiology, but appears to frequently be complicated by hypovitaminosis D with osteomalacia, as evidenced by incomplete improvements in bone density when serum vitamin D is replete. The use of glucocorticoids in DMD extends independent ambulation; however, its effects on bone health have not been completely studied, and may have adverse effects on bone density and increase fracture risk. Further studies are warranted. Further research is needed to assess the extent of poor bone health across all NMDs and to evaluate the efficacy of known osteoporosis treatments in this unique patient population with NMD.

MANAGEMENT OF CARDIAC COMPLICATIONS

Early treatment with ACE inhibitors is probably warranted in DMD when the measured ejection fraction falls below 55% (70,71). The benefits of earlier protective treatment with either ACE inhibitors or ARBs are under investigation. Digitalis has been demonstrated to be effective in decreasing morbidity from heart failure, but not mortality, and probably is also indicated for the treatment of heart failure observed in DMD patients with cardiomyopathy. Beta-blockers may also have a role in DMD. Treatment with coenzyme Q10 remains controversial. Cor pulmonale, confirmed on echocardiography, may benefit from continuous supplemental oxygen. Patients with known arrhythmias who are at risk for fatal tachyarrhythmias may benefit from anti-arrhythmic medication. DMD patients with mitral valve prolapse and mitral regurgitation should be given antibiotic prophylaxis for dental and surgical procedures in accordance with current guidelines.

The management of cardiomyopathy, seen in BMD, is similar to that seen in DMD; however, in cases of severe end-stage cardiomyopathy, cardiac transplantation should be considered.

Cardiac conduction abnormalities observed in myotonic muscular dystrophy may ultimately require implantation of cardiac pacemakers. In rare instances with cardiomyopathy, treatment may consist of ACE inhibitors, digitalis, and diuretics, based on proven efficacy in cardiomyopathies of other etiologies.

EMD patients with symptomatic bradycardia or heart block should undergo implantation of a permanent cardiac pacemaker. Atrial stand-still, atrial fibrillation, and atrial flutter are all disorders in which blood can pool in the atria, leading to thrombus formation and possible embolic events, including stroke. Anticoagulation with warfarin to an international normalized ratio (INR) of 2 to 3 has demonstrated a reduction in the incidence of stroke in patients with atrial fibrillation. Prompt referral to a cardiologist should be made for children with cardiac signs on symptoms or screening ECG, echocardiography, or for those with Holter recording abnormalities suggestive of cardiac disease. Late-stage DMD, BMD, and EMD patients should be followed by a cardiologist on a regular basis. There are selected subtypes of LGMD with cardiomyopathy warranting close follow-up and aggressive management. Appropriate management of cardiac complications in childhood NMD will likely lead to increased life expectancy and quality of life.

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19

NEURODEGENERATIVE AND DEMYELINATING DISEASES AND OTHER CNS DISORDERS

Amy Houtrow

This chapter covers numerous conditions that pediatric rehabilitation medicine physicians may encounter during their training and career. Because data contained in other chapters of this book are particularly relevant to the conditions presented here, the reader will be guided back to other chapters from time to time. This chapter will address pediatric demyelinating diseases of the central nervous system (CNS) and acquired brain injury from encephalitis (including anti-N-methyl-D-aspartate receptor [NMDAR] encephalitis), pediatric brain tumors, and seizure focus resection.

PEDIATRIC DEMYELINATING DISEASES

In this section of this chapter, the following pediatric demyelinating diseases of the CNS will be described: multiple sclerosis (MS), transverse myelitis, neuromyelitis optica (NMO), and acute disseminated encephalomyelitis (ADEM). Together, these conditions have incidence in children of 1.66 per 100,000 person-years (1). They are more common among Whites and Blacks and less common among Hispanics and Asians (2). Acute inflammatory demyelinating polyradiculoneuropathy (Guillain–Barré syndrome), a peripheral process, is described in Chapter 18. Generally, demyelinating diseases are described as monophasic or polyphasic and monofocal or polyfocal. Monophasic disease is also often referred to as a clinically isolated syndrome (3). Some children with monophasic disease go on to develop chronic demyelinating conditions such as MS or NMO. And it is important to note that there is considerable clinical overlap between these conditions (4).

PEDIATRIC MULTIPLE SCLEROSIS

Multiple sclerosis (MS) is an autoimmune chronic inflammatory disease characterized by demyelination and axonal degeneration. Approximately 5% of the total population of individuals with MS are children (5). In 2007, the International Pediatric Multiple Sclerosis Study Group proposed provisional definitions for CNS demyelinating disorders for children that were updated in 2012 and published in 2013 (6). The diagnosis requires the presence of CNS inflammatory disease that is disseminated in time and space. Pediatric MS can be diagnosed when any one of the following exists:

- Two or more nonencephalopathic clinically identified CNS events with presumed inflammatory cause, which are separated by more than 30 days and involve more than one area of the CNS
- One nonencephalopathic episode with associated MRI findings consistent with the revised 2010 McDonald criteria (7) (see Table 19.1), and follow-up MRI demonstrates one or more lesions consistent with the disseminated in time criteria
- One encephalopathic (ADEM) attack followed by a nonencephalopathic attack three or more months after the onset of symptoms with MRI lesions that meet the disseminated in space criteria
- For children 12 years old or older, a single nonencephalopathic event with MRI findings consistent with the criteria for dissemination in time and space

EPIDEMIOLOGY OF PEDIATRIC MS

While 1.7% to 5.6% of the total MS population is under the age of 18, diagnosis under the age of 10 is very rare. The incidence of pediatric MS worldwide is not known, but in California, the reported incidence is 0.51/100,000 person-years (1). There is greater racial/ethnic diversity among children with MS compared to adults, among whom most are non-Hispanic Whites (5). The remote

TABLE 19.1 2010 MCDONALD MRI CRITERIA FOR THE DIAGNOSIS OF MULTIPLE SCLEROSIS

DEMONSTRATION OF DISSEMINATION IN SPACE	DEMONSTRATION OF DISSEMINATION IN TIME
At least two of four areas of the CNS with one or more T2 lesions • Periventricular • Juxtacortical • Infratentorial • Spinal cord (excluded if patient has a brainstem or spinal cord syndrome)	1. One or more new T2 and/or gadolinium-enhanced lesion(s) on follow-up MRI in comparison to baseline scan 2. Simultaneously present gadolinium-enhancing asymptomatic and nonenhancing lesions at any time as long as the asymptomatic gadolinium-enhancing lesion is not due to non-MS pathology

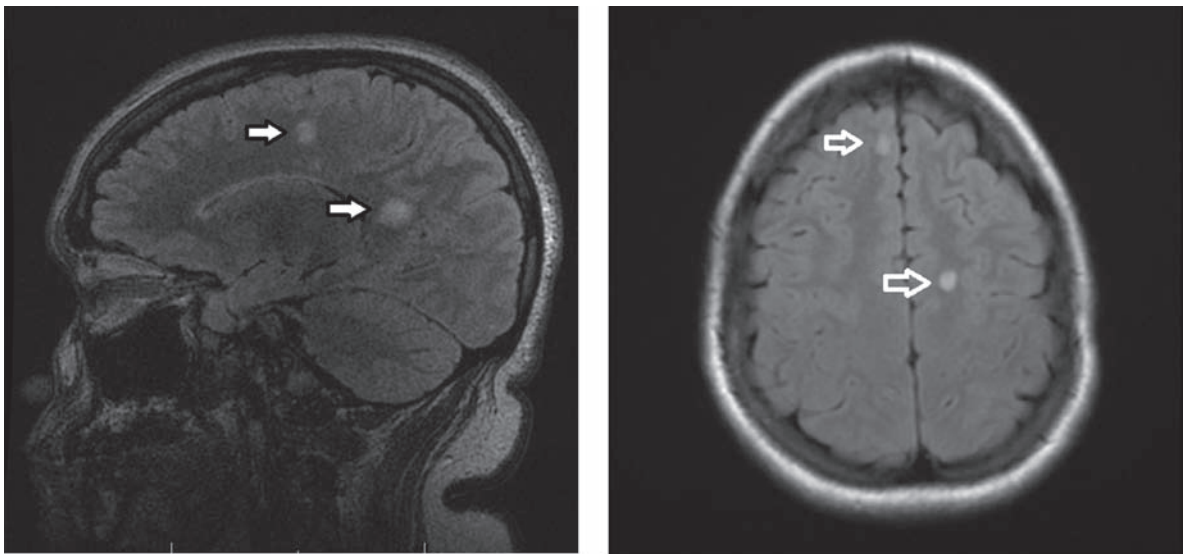
Source: Adapted from Ref. (7). Polman CH, Reingold SC, Banwell B, et al. Diagnostic criteria for multiple sclerosis: 2010 revisions to the McDonald criteria. *Ann Neurol*. 2011;69(2):292–302.

history of Epstein–Barr virus is associated with pediatric MS and many studies have supported the gene-viral environment hypothesis (7). The exact mechanisms that lead to MS are not understood and ongoing research is being conducted to identify triggers and the pathophysiology.

CLINICAL COURSE OF PEDIATRIC MS AND DIAGNOSIS

Almost uniformly, children with MS have relapsing–remitting type (3). The rate of relapses in children are higher than adults with MS, but recovery seems to occur more quickly (3). Frequently, children are polysymptomatic at presentation. Common presenting symptoms include sensory deficits, optic neuritis, gait disturbances, and brainstem symptoms (3). There is clinical overlap with ADEM raising concern that children who present with ADEM may eventually meet criteria for MS. In addition, children with MS have cognitive deficits more commonly than adults. Thirty-five percent of consecutively enrolled subjects from the Pediatric Multiple Sclerosis Centers for Excellence met diagnostic criteria for cognitive impairment (8).

When MS is suspected, a complete neurologic exam is necessary as is MRI with gadolinium of the brain and spinal cord. MRI findings consistent with MS include ovoid T2 lesions, and fluid-attenuated inversion recovery (FLAIR) hyperintensities in the periventricular white matter are typical. Cortical lesions are less common in children with MS compared to adults (9). See Figure 19.1 for an MRI of a teenaged girl with MS.



(A) (B)

FIGURE 19.1 The sagittal (A) and axial (B) views of the MRI of a teenaged girl with MS demonstrate T2-FLAIR hyperintensities separated in space. The arrows point to the lesions.

Source: Images courtesy of Guilio Zuccoli, MD, Associate Professor of Radiology, University of Pittsburgh.

Recommended blood work includes a complete blood count (CBC) with differential, C-reactive protein, erythrocyte sedimentation rate, antinuclear antibody, angiotensin-converting enzyme, folate, B-12, and thyroid-stimulating hormone (3). Depending on the clinical presentation, a fundoscopic exam and MRI of the orbits may be indicated. A lumbar puncture with opening pressure may be performed especially if the presentation is atypical. Cerebral spinal fluid examination often reveals a lymphocytic predominance, normal glucose, and elevated protein levels (5). The differential diagnosis of pediatric MS includes other autoimmune conditions, vascular disorders, space-occupying lesions (tumors), metabolic disorders including nutritional deficiencies, mitochondrial disorders, and leukodystrophies (3). It is important to evaluate for the stigmata of neurofibromatosis (NF) and tuberous sclerosis (TS). Complicated migraines and CNS vasculitis may mimic MS and should also be on the differential when headaches are the predominant presenting symptom (6).

Children with MS experience two to three times more frequent relapses than adults with an annualized relapse rate of 1.12 to 2.76 (10). Their MRI lesion burden is higher than adults, as well (11). Additionally, they have higher rates of cognitive impairments and longitudinally demonstrate worsening of cognitive functioning (12). Conversely, children with MS accrue locomotor disability more slowly than adults (13). At the time of disease onset, a progressive initial course is predictive of a statistically shorter time to irreversible disability (13). Two years after onset, the number of relapses was predictive of increased rate of disability as was having a progressive initial course (13).

TREATMENT FOR PEDIATRIC MS

The International Pediatric Multiple Sclerosis Study Group recommends beta-interferon or glatiramer acetate as first-line therapy for pediatric MS (14). In the presence of inadequate treatment response, defined as a 6-month minimum of full dose fully adherent therapy with either persistent or increased relapse rate, or two or more confirmed relapses in 12 months, the study group recommends switching between first-line agents or switching to or adding a second-line agent (14). High-dose steroids are often used for short periods and natalizumab and cyclophosphamide are also second-line options (15,16). Some providers use mitoxantrone, fingolimod, or rituximab as second-line options although the evidence is more limited regarding their use (14,17).

REHABILITATION FOR PEDIATRIC MS

Since the 1980s, the Expanded Disability Status Scale (EDSS) and the Functional Systems Score (FSS) have been used to track MS-related disability (18). The systems in the FSS are: pyramidal functions, cerebellar functions, brainstem functions, sensory function, bowel and bladder function, visual function, and cerebral (mental) functions (18). See Table 19.2 for the FSS.

The scores on the EDSS range from 0 (normal neurologic exam) to 10 (death due to MS) and focus on mobility skills and FSS dysfunction. See Table 19.3 for the complete EDSS.

The EDSS does not map well to functional assessment tools typically used in pediatric rehabilitation such as the Functional Independence Measure for Children (WeeFIM), Pediatric Evaluation of Disability Inventory (PEDI), and Functional Rehabilitation Evaluation of Sensori-Neurologic Outcomes (FRESNO) (19–21). Nonetheless, the EDSS is used in most research related to the clinical management of MS and is important for the rehabilitation physician to understand. Developmentally speaking, EDSS is not appropriate for children who would not be expected to be ambulatory or independent with their self-care. This is rarely a problem because MS is extremely uncommon among very young children.

The rehabilitation interventions depend on the functional impairments, activity limitations, and participation restrictions experienced by the child with MS. Common impairments include: weakness, spasticity, neurogenic bladder, neurogenic bowel, pain, fatigue, and depression. Although physical and occupational therapy are the primary way weakness is addressed, potassium channel blockade has been shown to improve ambulation due to the improved conduction in demyelinated fibers (22). Because of the narrow therapeutic window, it is not recommended for use in children. When considering an antispasticity agent such as baclofen, it is important to evaluate if the spasticity is benefiting function such as for ambulation when spasticity in the lower extremities is compensating for weakness (23). It is also important to consider the side effect of fatigue because fatigue is a common symptom of MS and may be made worse with the initiation of an antispasticity agent. For intractable spasticity, intrathecal baclofen pump placement may be a consideration. For more localized spasticity, botulinum toxin injection should be considered (24). Botulinum is also used for neurogenic detrusor overactivity (25). But the first-line management remains anticholinergic agents such as oxybutynin (23).

Symptomatic neurogenic bladder impacts about 80% of all patients with MS with detrusor hyperactivity and detrusor-sphincter dyssynergia as the most common diagnoses (23). There is little consensus about when to start intermittent straight catheterizations (26), but issues regarding continence should be discussed both from the medical and social perspectives. For the adolescent with MS, discussions about sexual functioning should occur. Management of neurogenic bowel and constipation follows the recommendations for management of upper motor neuron bowel (see Chapter 16 for details). Neuropathic pain and musculoskeletal pain are common. Antiepileptic medications play an important role in the

TABLE 19.2 KURTZKE FUNCTIONAL SYSTEMS SCORES

KURTZKE	0	1	2	3	4	5	6
Pyramidal Functions*	Normal	Abnormal signs without disability	Minimal disability	Mild to moderate paraparesis or hemiparesis; or severe monoparesis	Marked paraparesis or hemiparesis; or moderate quadriparesis or monoplegia	Paraplegia, hemiplegia, or marked quadriparesis	Quadriplegia
Cerebellar Functions*	Normal	Abnormal signs without disability	Mild ataxia	Moderate truncal or limb ataxia	Severe ataxia in all limbs	Unable to perform movements due to ataxia	
Brainstem Functions*	Normal	Signs only	Moderate nystagmus or other mild disability	Severe nystagmus, marked extraocular weakness, or moderate disability of other cranial nerves	Marked dysarthria or other marked disability	Inability to speak or swallow	
Sensory Function*	Normal	Vibration or figure writing decrease only in one or two limbs	Mild decrease in touch or pain or position sense, and/or moderate decrease in vibration in one or two limbs; or vibratory decrease alone in three or four limbs	Moderate decrease in touch or pain or position sense, and/or essential lost vibration in one or two limbs; or mild decrease in touch or pain and/or moderate decrease in all proprioceptive tests in three or four limbs	Marked decrease in touch or pain or loss of proprioception alone or combined, in one or two limbs; or moderate decrease in touch or pain and/or severe proprioceptive decrease in more than two limbs	Loss (essentially) of sensation in one or two limbs; or moderate decrease in touch or pain and/or loss of proprioception for most of the body below the head	Sensation essentially lost below the head
Bowel and Bladder Function*	Normal	Mild urinary hesitance, urgency, or retention	Moderate hesitancy, urgency, retention of bowel or bladder, rare urinary incontinence	Frequent urinary incontinence	In need of almost constant catheterization (and constant use of measures to evacuate stool)	Loss of bladder function	Loss of bowel and bladder function
Visual Function*	Normal	Scotoma with visual acuity corrected better than 20/30	Worse eye with scotoma with visual acuity corrected 20/30–20/59	Worse eye with large scotoma, or moderate decrease in fields, but with maximal acuity corrected of 20/60–20/99	Worse eye with marked decrease of fields and maximal acuity of 20/100–20/200; grade 3 plus maximal acuity of better eye of 20/60 or less	Worse eye with maximal acuity corrected less than 20/200; grade 4 plus maximal acuity of better eye of 20/60 or less	Grade 5 plus maximal visual acuity of better eye of 20/60 or less
Cerebral (or Mental) Functions*	Normal	Mood alteration only	Mild decrease in mentation	Moderate decrease in mentation	Marked decrease in mentation	Dementia or chronic brain syndrome	

* Score of 9 given if unknown.

Source: Adapted from Ref. (18). Kurtzke JF. Rating neurologic impairment in multiple sclerosis: an expanded disability status scale (EDSS). *Neurology*. 1983;33(11):1444–1452.

TABLE 19.3 THE EXPANDED DISABILITY STATUS SCALE

	KURTZKE EXPANDED DISABILITY STATUS SCALE
0	Normal neurological exam: FSS = 0 in all systems (score of 1 on cerebral function is allowable)
1	No disability: minimal signs in functional system
1.5	No disability: minimal signs in more than one functional system
2	Minimal disability: FSS = 2 in one functional system, other 0 or 1
2.5	Minimal disability: FSS = 2 in two functional systems, others 0 or 1
3	Moderate disability: FSS = 3 in one functional system, others 0 or 1, or mild disability in three to four functional systems
3.5	Fully ambulatory but FSS = 3 in one functional system and FSS = 2 in two other functional areas; FSS = 3 in two functional areas, other 0 or 1; or FSS = 2 in five functional systems
4	Fully ambulatory without assistive device, able to walk 500 m without rest, self-sufficient despite relatively severe disability: FSS = 4 in one functional system, others 0 or 1; or combination of lesser grades exceeding previous levels
4.5	Fully ambulatory without assistive device, able to walk 300 m without rest, able to work, may otherwise have some limitations of full activity or need minimal assistance: usually FSS = 4 in one functional system or combination of lesser grades exceeding previous levels
5	Ambulatory without assistive device or rest for 200 m, full day activities impaired: FSS = 5, others 0 or 1, or combination of lesser grades exceeding level 4
5.5	Ambulatory without assistive device or rest for 100 m, full day activities impaired: FSS = 5 for one functional domain or combination of lesser grades exceeding level 4
6	Intermittent or constant assistive device required to walk 100 m with or without rest: usually FSS = 3+ in more than two functional systems
6.5	Constant bilateral assistive device(s) required to walk 20 m: usually FSS = 3+ in more than two functional systems
7	Unable to walk 5 m with assistive device(s), transfers independently, can propel manual wheelchair for full day: usually combinations of functional system impairments with more than one FSS = 4+
7.5	Unable to take more than a few steps, assistance for transfers, cannot self-propel manual wheelchair: usually combinations of functional system impairments with more than one FSS = 4+
8	Essentially restricted to bed or wheelchair but retains many self-care functions, generally has functional use of arms: usually combinations of functional system impairments with FSS = 4+ in several systems
8.5	Essentially restricted to bed, some effective use of arms, retains some self-care skills: usually combinations of functional system impairments with FSS = 4+ in several systems
9	Dependent but can communicate and eat: usually combinations of functional system impairments with FSS = 4+ in most systems
9.5	Totally dependent: almost all FSS = 4+
10	Death due to MS

Abbreviation: FSS, Functional systems score.

Source: Adapted from Ref. (18). Kurtzke JF. Rating neurologic impairment in multiple sclerosis: an expanded disability status scale (EDSS). *Neurology*. 1983;33(11):1444–1452.

modulation of neuropathic pain and traditional analgesics are helpful in musculoskeletal pain (7). In adults, the use of cannabis extract has been used with success in the management of painful spasticity refractory to other interventions (27). When a child presents with debilitating fatigue, a close examination of the medications should be conducted to determine whether they are contributing to the fatigue. A sleep hygiene regime may be initiated and medications modafinil and amantadine may be considered, but their efficacy is limited (23). Mood disorders are frequent among individuals with MS and likely under-recognized in children. Psychological interventions, including cognitive behavioral therapy have been shown to be helpful (23). Medications to address mood may be considered.

To address activity limitations and participation restrictions, the rehabilitation program should be family-centered and goal-directed. The child and the family should be actively involved in goal setting. These goals should include skill attainment/reacquisition/refinement, enhancement of endurance, and adaptations to the environment as necessary (23). Occasionally, the child with MS will present for comprehensive inpatient rehabilitation, but more commonly, the child will be engaged in outpatient services. It is important to coordinate care between the treating therapists, the neurologist, and the rehabilitation physician. Regular re-evaluation is essential because MS is not a static disease. Close attention to the cognitive sequelae of MS is necessary because school programming frequently needs to be adapted. Some children may do well with a 504 plan and others may require an Individualized Education Program (IEP). Neuropsychological testing can help guide cognitive therapy and classroom adaptations. For older adolescents, vocational rehabilitation may be an adjunct to their therapeutic program. Additionally, structured drivers' education may be necessary.

Pediatric MS is a rare and complicated condition that can impact functioning in multiple different domains. The pediatric rehabilitation physician has many resources available to help address the disability associated with MS. These include medications to address the common symptoms of MS and the coordinated goal-directed rehabilitation team.

TABLE 19.4 THE DIAGNOSTIC CRITERIA FOR TRANSVERSE MYELITIS

Development of dysfunction attributable to the spinal cord
Bilateral signs and symptoms (not necessarily symmetric)
Inflammation of the spinal cord evidenced by: • CSF pleocytosis, or • Elevated IgG index, or • Gadolinium enhancement
If none are present at symptom onset, repeat MRI and lumbar puncture are warranted.
Progression to nadir between 4 hours and 21 days

Abbreviation: CSF, cerebrospinal fluid.

Source: Adapted from Ref. (29). Transverse Myelitis Consortium Working Group. Proposed diagnostic criteria and nosology of acute transverse myelitis. *Neurology*. 2002;59(4):499–505.

TRANSVERSE MYELITIS

Transverse myelitis, also called acute transverse myelitis, is an acute monophasic or clinically isolated syndrome of the spinal cord (4). Transverse myelitis may also be associated with polyphasic condition such as neuromyelitis optica, MS, or systemic lupus erythematosus (SLE) (28). The diagnosis relies on the exclusion of other causes of myelitis and spinal cord compression. Once other etiologies are excluded, the diagnosis depends on meeting the inclusion criteria listed in Table 19.4 (29).

When working up myelopathy, there are three priorities (30):

1. Ruling out compressive etiology
2. Determination of evidence of cord inflammation, and
3. Determination of the extent of the demyelination (beyond the cord or not).

Epidemiology of Transverse Myelitis

Transverse myelitis is a rare condition but one that often requires comprehensive inpatient rehabilitation. Approximately 20% of the 1,400 new cases in the United States each year are children (31). Children with transverse myelitis tend to be older at the age of diagnosis compared to children with ADEM (1). There appears to be a bimodal distribution with peaks in early childhood and a board peak between school-age and older children/adolescents (32). A considerable percentage of children with transverse myelitis go on to be diagnosed with a polyphasic demyelinating disorder such as MS (33).

Clinical Course and Diagnosis of Transverse Myelitis

If there is clinical concern for transverse myelitis, a thorough history should be gathered to document time course and extent of the symptoms, as well as any recent illness or prodromal symptoms (29). A recent mild illness is reported in a vast majority of cases; less frequently the patient may report a vaccine or allergy shot (28). The location of the lesion is most commonly the thoracic spine, but any and all segments may be involved (3,32). Therefore, a majority of patients present with complaints of lower extremity weakness. Rarely conus medullaris syndrome presentation may occur (32). In cases of posterior column involvement, fine motor discoordination and ataxic gait may mimic cerebellar disease (28). Children with posterior cord syndrome should be worked up for B_{12} and copper deficiency (33). In addition to weakness, most children present with constipation and bladder dysfunction consistent with neurogenic bowel and bladder, respectively. Children often also present with sensory disturbances such as allodynia and paresthesias (28,32). Horner syndrome, in lesions above T2, has also been reported (28).

A full neurologic exam is warranted with delineation of motor and sensory levels. The child's neurologic level of the injury should be determined to the best of the examiner's ability recognizing the challenges of conducting a comprehensive spinal cord examination on a very young child. The

neurologic level is the most caudal segment with normal sensation and antigravity motor function bilaterally (34). The American Spinal Injury Association impairment scale classifies the completeness of the injury based on sacral sparing, which is the presence of motor or sensory function in the most caudal sacral segments (34). The grading of the degree of impairment is important not only to anticipate function, but also because partial transverse myelitis is associated with a higher likelihood of future diagnosis of a polyphasic demyelinating disorder (31,33). In the very early phases of the disease, the child may be in spinal shock and have absent reflexes. Guillain–Barré Syndrome can present similarly with diminished reflexes, dysautonomia, and bowel and bladder dysfunction, but usually is quickly distinguished from transverse myelitis (28). See Table 19.5 for the differential diagnosis of myelitis (28,30,33,34).

TABLE 19.5 THE DIFFERENTIAL DIAGNOSIS OF TRANSVERSE MYELITIS

DIAGNOSTIC GROUP	SPECIFIC DIAGNOSES (NONEXHAUSTIVE LISTS)
Compressive myelopathy from extramedullary mass effect (excluding neoplasms)	• Vertebral body compression • Disk herniation • Abscess • Epidural hematoma

Neoplasm	• Extramedullary:
	  • Ewing sarcoma
	  • Lymphoma
	  • Neuroblastoma
	  • Granulocytic sarcoma
	  • Metastatic disease
	• Intramedullary:
	  • Astrocytoma
	  • Ependymoma
	  • Glioma
	  • Hemangioblastoma
	  • Metastatic disease

Paraneoplastic (exceptionally rare in children)	• Lung cancer • Breast cancer • Ovarian cancer
Toxin	• Radiation associated myelopathy (usually presents within 15 years after radiation)

Infectious	• Viral:
	  • DNA viruses (herpes)
	  • RNA viruses:
	  • Flaviviruses (West Nile)
	  • Orthomyxoviruses (influenza A)
	  • Paramyxoviruses (measles)
	  • Picornaviruses (coxsackie)
	• Bacterial
	• Fungal
	• Parasitic

Ischemic/hemorrhagic	• Anterior spinal artery syndrome—presents with motor and spinothalamic-related sensory deficits with sparing of proprioception and vibratory senses (34) • Surfer's myelopathy • Fibrocartilaginous embolism • Arteriovenous malformation
Metabolic	• Copper deficiency • B_{12} deficiency
Autoimmune	• Systemic lupus erythematosus • Sjogren syndrome • Mixed connective tissue disorder • Scleroderma • Neurosarcoidosis • Behcet's disease

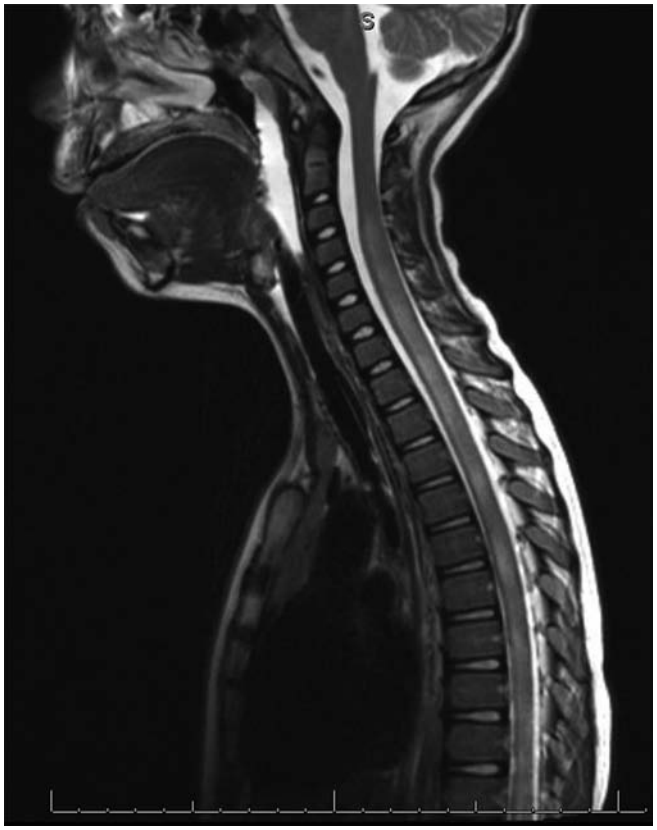


FIGURE 19.2 This sagittal view MRI demonstrates diffuse alteration of the signal intensity of the cord from C2 downward. Multilevel cord expansion is also noted.

Source: Image courtesy of Guilio Zuccoli, MD, Associate Professor of Radiology, University of Pittsburgh.

A gadolinium-enhanced MRI of the spine should be conducted as soon as possible. Depending on the timing, a brain MRI can be conducted simultaneously or at a later time. If a compressive myelopathy is identified, then urgent surgical referral is warranted and steroids may be considered (29). The MRI of transverse myelitis usually reveals T1-isointense and T2-hyperintense signals over multiple contiguous segments (3,33). The entire cord may be involved and there can be substantial swelling with effacement (3). See Figure 19.2 for the MRI of a child with tetraplegia from transverse myelitis.

Children with transverse myelitis tend to have more severe presentations than adults (30). Complete recovery can occur, but occurs in a minority of children (30). A number of factors have been identified as poor prognostic indicators, including: rapidity to nadir (in some studies), need for ventilator support, higher lesion level, larger number of segments involved, and spinal shock (28,30,32). Symptom nadir tends to occur within a week of presentation. Around that time, children with transverse myelitis may experience autonomic instability with fluctuations in heart rate, temperature, and respirations (28). Recovery is most rapid during the first 6 months after onset, but improvements have been documented years after the disease (30). As mentioned previously, some children, especially those with partial lesions will go on to develop a polyphasic demyelinating condition despite good recovery from the first lesion.

Treatment for Transverse Myelitis

High-dose steroids given for 5 to 7 days are the mainstay of treatment for transverse myelitis (3), although there is insufficient evidence to determine the efficacy of steroids (31). In fulminant disease, plasmapheresis and intravenous immunoglobulin (IVIg) may be considered (3,31). Many children present with pain and sensory changes, which may persist long-term. Medications to manage spasticity and pain are commonly needed. Children with cervical lesions may need ventilatory support.

Rehabilitation for Transverse Myelitis

The rehabilitation management of the child with transverse myelitis is similar to the management of children with spinal cord injuries of other etiologies (see Chapter 16). Consultations with therapies and pediatric rehabilitation medicine should occur early in the acute period to help maximize recovery (28). As with other children with spinal cord injury, children should receive interdisciplinary, family-centered, goal-directed rehabilitation that focuses on recovery of lost function and making adaptations when function does not return. Depending on the timing of admission to rehabilitation and the capabilities of the rehabilitation unit, some children with transverse myelitis may still be receiving steroids, IVIg, or plasmapheresis for disease management. If a child is transitioned to rehabilitation early in the disease course, vigilance is required to monitor for worsening symptomatology because the time to reach nadir is variable (28). The child with cervical transverse myelitis is at particular risk for decompensation and should be monitored closely for changes in respiratory status due to disease progression. If the lesion is cervical or thoracic, the aggressive pulmonary toilet is recommended. Children with tetraplegia may need cough assist; therefore families should be trained to assist with "quad coughing." Later in their rehabilitation course, children with transverse myelitis lesions above T6 are at risk for autonomic dysreflexia (see Chapter 16) (28).

Chronic sensory changes have been noted in nearly half of patients (32). If bothersome, long-term antiepileptic mediation should be considered. Amitriptyline may also be used in the management of pain (28). A desensitization program may be considered for the child experiencing allodynia or dysesthesia that is interfering with activities. During the acute rehabilitation phase and long term, children with transverse myelitis are at risk for skin breakdown due to limited sensory input and limited mobility. Prevention of skin breakdown is the most effective management and requires keen attention to at risk

areas. The patient and family need to be educated extensively and equipment that minimizes risk should be provided.

Neurogenic bladder is noted in most cases in the acute period and may persist for a majority of patients (32). Intermittent straight catheterization remains the mainstay of management. Chronically, children may have detrusor muscle spasticity, dyssynergia, or an areflexive bladder. Urodynamics and routine renal and bladder ultrasound should be a part of the long-term management to ensure urinary system function and health (35). In the very young child with neurogenic bladder associated with transverse myelitis, social continence is not an issue, but catheterizations and medications may still be indicated for urinary system health. For older children, urinary system health and achieving continence should be the goals of treatment. Similarly, neurogenic bowel and the associated constipation often require long-term management (35). Children with spastic bowels usually benefit from an oral agent to improve transit and an agent administered rectally to initiate evacuation.

While not immediately evident by the clinical presentation of transverse myelitis, cognitive functioning of the patient with transverse myelitis requires special attention because some patients will go on to develop polyphasic disease. Additionally, in a cohort of children with transverse myelitis, most had neuropsychological function in the normal range but attention and memory problems were identified in a large minority of patients, with a third of parents endorsing subclinical attention problems (36). Neuropsychological screening can identify the need for administration of a full neuropsychological battery. Furthermore, children with transverse myelitis are at elevated risk of depression and anxiety symptoms. Therefore screening and counseling if necessary should be part of the multidisciplinary management team (28). Children deemed at increased risk of progression to polyphasic disease may benefit from baseline neuropsychological testing even in the absence of neurocognitive or neurobehavioral concerns.

Because transverse myelitis affects multiple areas of function and can be associated with future polyphasic disease, routine multidisciplinary management is warranted (36). Unlike for children with pediatric MS, where the neurologist is likely the subspecialist with the most active management responsibilities, children with transverse myelitis have needs similar to other children with spinal cord injury, which are often best met by the rehabilitation physician and team.

NEUROMYELITIS OPTICA

NMO is a polyfocal and often polymodal demyelinating condition characterized by optic neuritis and transverse myelitis (3,33). A relapsing course of NMO, previously known as Devic disease, is more common than a monophasic course (37). NMO used to be considered a rare variant of MS but is now understood to be a separate disease (38). The key to diagnosing NMO includes ensuring that the child's presentation is not more consistent with MS. This is important because management differs, NMO is less responsive to treatment, and it has a poorer prognosis than MS (38,39). Seropositivity for NMO-IgG is not required for the diagnosis but is highly indicative. Children are seropositive less frequently than adults (3). The binding of NMO-IgG (aquaporin-4-IgG) to aquaporin-4 results in a biological cascade that results in inflammation and demyelination characteristic of the disease (37). The understanding of NMO has expanded exponentially in the past decade and consensus has been reached regarding the diagnosis (40). Table 19.6 details the diagnostic criteria for NMO (41).

TABLE 19.6 THE DIAGNOSTIC CRITERIA FOR PEDIATRIC NEUROMYELITIS OPTICA

Optic neuritis
Acute myelitis
Two of the following:
• Spinal MRI demonstrating contiguous lesion extending at least three vertebral segments • NMO-IgG seropositivity • Brain MRI does not meet the criteria for MS

NMO spectrum disorders are those disorders that do not meet diagnostic criteria for NMO and include recurrent optic neuritis and recurrent transverse myelitis, with seropositivity (6,40). The child with an NMO spectrum disorder may go on to meet the criteria for NMO. Because of this, children with NMO spectrum disorders need to be monitored very closely for disease progression. Children who have relapsing optic neuritis without evidence of other neurologic deficits or systemic disease have chronic relapsing optic neuropathy (CRION), which is often bilateral and steroid dependent (42). These children also have to be closely monitored for progression.

Epidemiology of NMO

NMO is exceptionally rare; for every individual with NMO, there are 50 to 100 individuals with MS (38). NMO can affect children and adults of any age, but children tend to present in late childhood or adolescence (3). There is a strong female predominance, even stronger than the female predominance for MS (37,39). NMO is more common among those of Asian and African descents (33). NMO tends to occur sporadically but familial cases have been identified (40). A large majority of patients with NMO have serologic evidence of autoimmunity (37). Children with NMO are often seropositive for antinuclear, SS-A,

antiacetylcholine receptor, or double stranded DNA antibodies (38). Autoimmune conditions such as SLE, Sjogren syndrome, juvenile idiopathic arthritis, and Graves disease may coexist or precede NMO (37).

Clinical Course of NMO and Diagnosis

The hallmarks of NMO are transverse myelitis (see preceding section) and optic neuritis. Children with optic neuritis can present with unilateral or bilateral blurry vision, a black spot in the central visual field, or visual acuity loss (43). Compared to adults, vision loss is more severe in children. Children may also report pain with ocular movement (3). If the diagnosis of optic neuritis is suspected, the child should be seen by an ophthalmologist or neuro-ophthalmologist, if one is available. A normal fundoscopic exam does not rule out the diagnosis as some cases are due to retrobulbar inflammation. Visual acuity, visual field, low contrast sensitivity, and color vision should all be assessed. The neuro-ophthalmologist may also perform visual evoked potentials and may recommend optical coherence tomography and an orbital MRI (3). Figure 19.3 demonstrates active optic neuritis.

The differential diagnosis list for optic neuritis includes vitamin B_{12} deficiency, adrenoleukodystrophy, Leber hereditary optic neuropathy (usually pain is absent but a family history is often present), and optic glioma. Additionally, sarcoidosis may present with cranial nerve defects and optic neuritis (3).

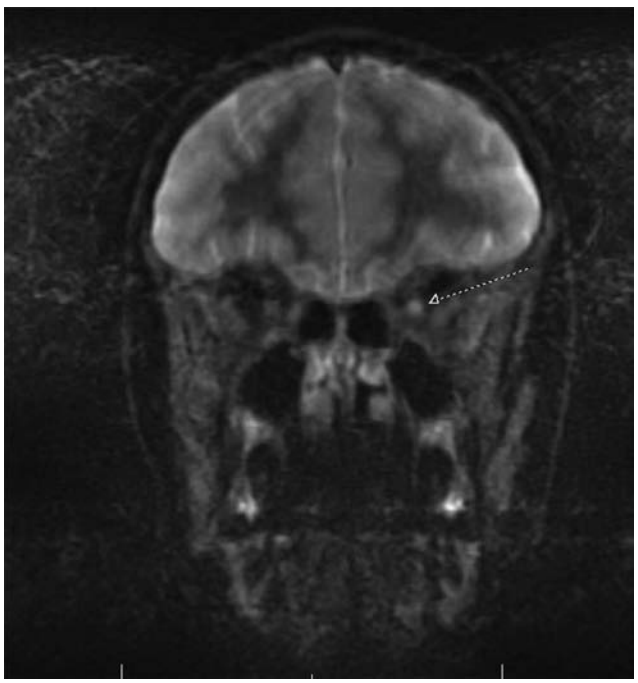


FIGURE 19.3 This coronal view MRI demonstrates left-sided optic neuritis in a child with NMO.

Source: Image courtesy of Guilio Zuccoli, MD, Associate Professor of Radiology, University of Pittsburgh.

The treatment of isolated optic neuritis is high-dose steroids (3,44). Refractory cases may be treated with IVIg or plasmapheresis (3). Children tend to recover well from optic neuritis with a majority achieving at least 20/40 vision at follow-up (43,45). Children with bilateral optic neuritis have an increased likelihood of developing MS, as are children who are older (45,46). Children with relapsing optic neuritis may go on to develop either MS or NMO. The case of seropositivity and relapsing optic neuritis without other signs of NMO is NMO spectrum disease. Because children with optic neuritis can later develop MS or NMO, the workup should include a gadolinium-enhanced MRI of the brain and spinal cord and a lumbar puncture with opening pressure (3). Because NMO can occur concurrently with other autoimmune conditions, additional serologies should also be ordered.

Similar to other pediatric demyelinating conditions, children often present after a prodromal flu-like illness (37). The child with NMO tends to present with complete transverse myelitis. This is in contrast to MS in which children tend to present with partial lesions (37). Occasionally, though, central cord syndrome may be the presentation of transverse myelitis in NMO (33). A careful and thorough neurologic examination is necessary to delineate the findings of NMO. While transverse myelitis and optic neuritis are the hallmarks of the condition, children with NMO may also have signs and symptoms of intracranial pathology. The MRI of the brain typically demonstrates lesions in the areas of the brain that are aquaporin-4 rich such as the hypothalamus, subcortical white matter, and the periventricular gray matter (3). Brain lesions are more common in pediatric NMO than adult NMO (37). In addition to the typical MRI findings, "cloud-like" enhancements, linear lesions extending from the brainstem to the spinal cord, and posterior reversible encephalopathy syndrome (PRES)-like lesions may be seen (40). Necrosis can occur in the areas of the brain with severe antibody-mediated inflammation and demyelination (37). Children with NMO may also present with encephalopathy and can therefore resemble ADEM. The MRI lesions may be quite large and be radiographically difficult to distinguish from ADEM (3,37,40). In patients with MRI findings in the area postrema, the clinical presentation often includes weeks to months of intractable emesis (37).

Treatment of NMO

The treatment of NMO includes management of acute episodes, prevention of relapses, symptom management, and rehabilitation (40). The medical management for pediatric NMO is based on the treatment recommendations for adults. First-line treatment is IV methylprednisolone 30 mg/kg/day for 5 days. Plasmapheresis and IVIg are also treatment options in the acute period (37,40). Plasmapheresis is considered a first- or second-line agent and may be given concurrently with steroids or instead

of steroids if the child does not tolerate steroid treatment. Five exchanges scheduled every other day is the typical administration and can lower NMO titers (3). IVIg at a total dose of 2 g/kg given over 2 to 5 days is a third-line agent (3). Long-term immunosuppression is the standard of care but is associated with significant side effects about which the patient and parents should be educated (40). Children should be treated with calcium and vitamin D to promote bone health, and they need surveillance because of the risk of bone demineralization (37).

For children who are seronegative and fully recovered, prophylactic medications can be considered versus periodic monitoring for new symptoms. Children who are seropositive or who have had a relapse should be placed on prophylactic medication after treatment of the acute episode. Usually oral steroids are given for several months while transitioning to a steroid-sparing agent such as azathioprine, mycophenolate mofetil, or rituximab (3,37). Rituximab dosing tends to follow the schedule developed for rheumatoid arthritis and mitoxantrone, a potent immunosuppressant, has been used in refractory cases (40). Other medication options include methotrexate, IVIg, cyclophosphamide, cyclosporine, and tacrolimus. Plasmapheresis on a routine basis may also be considered (40). It is important to note that many conventional MS medications (interferon-beta, natalizumab, and fingolimod) are contraindicated in NMO because they have been associated with worsening of symptoms (40).

Rehabilitation of NMO

Unfortunately disability in NMO tends to be more severe than in MS. Disability associated with NMO maps to the severity and location of the lesions with vision loss and paraplegia being the most common (37). Disability tends to accrue with each subsequent attack, so it is important to minimize the impact of each attack and prevent future attacks (40). Pain, stiffness, weakness, fatigue, and bowel and bladder dysfunction are common with NMO. Patients with NMO can experience painful tonic spasms of the limbs that look dystonic in presentation. Carbamazepine has been shown to be effective in reducing these spasms (40).

The rehabilitation management of NMO includes the management of transverse myelitis as described in the previous section and in Chapter 16. Additionally, children with brain lesions should receive appropriate brain injury rehabilitation. The rehabilitation program should be interdisciplinary, family-centered, and goal-directed. Neuropsychological testing may be conducted after the acute recovery period to guide classroom adaptations. Like the other polymodal pediatric demyelinating conditions, periodic neuropsychological screening is warranted and repeat testing is often indicated. Unlike the other pediatric demyelinating conditions, except occasionally pediatric MS, low vision rehabilitation may need to be incorporated into the rehabilitation program.

Optimally, low vision rehabilitation is a multidisciplinary endeavor that provides assessments, adaptive techniques and strategies, and training (47). Functional vision assessment is the measurement of how well patients use their existing vision to conduct day-to-day activities (47,48). The ophthalmologic assessment should include measurement of visual acuity, contrast sensitivity, perimetry, light characteristics, refractory errors, oculomotor functions, and cortical visual integration (47). Based on the findings, optical assistive devices may be prescribed. Occupational therapists with specialized low vision rehabilitation training teach patients how to use the assistive devices in everyday activities and help patients strategize compensatory strategies (49).

Community reentry, including return to school, may be particularly complicated for the child with NMO because of the combination of motor, cognitive, and vision deficits. Environmental assessments and adaptations of the home and school can help improve participation and quality of life by assuring a safe environment for optimal functioning (48,49).

While the prognosis remains relatively poor for NMO, advances in the management will hopefully lead to improvements in prognosis in the next several years (40). Actively employing rehabilitation strategies to maximize function in conjunction with immunosuppression and careful multidisciplinary surveillance should improve clinical outcomes and the quality of life of children with NMO.

ACUTE DISSEMINATED ENCEPHALOMYELITIS

ADEM is a polyfocal demyelinating disease more commonly diagnosed in children than adults (3). To be diagnosed with ADEM, the encephalopathy (behavioral change or altered consciousness) cannot be explained by fever, an infectious process, or a postictal state (6). Additionally, the presentation must be the first demyelinating illness for that child, the MRI must be abnormal in the acute phase (first 3 months), and new findings cannot emerge after the acute phase (6). The typical brain MRI findings in ADEM are diffuse, large white matter lesions with poor demarcation. There may also be focal punctate lesions, bithalamic or basal ganglia lesions, or more rarely, hemorrhagic lesions consistent with hemorrhagic leukoencephalopathy (50).

Epidemiology of ADEM

The estimated incidence of ADEM is 0.4/100,000/year (51), making it a rare disease. Children commonly present between the ages of 5 and 8 years old and are more often affected than adults. Some cases of ADEM can be linked to preceding viral illness or vaccine administration (Semple rabies vaccine, small pox vaccine, and older measles vaccines), but most are not linked to a specific pathogen (3,52,53).

Clinical Course of ADEM and Diagnosis

A rapid onset of encephalopathy with multifocal neurologic deficits is the typical presentation with maximal deficits noted between 2 and 5 days after onset (51). A viral infection or vaccine may be in the recent history (2–4 weeks prior) (3,52). Prodromal symptoms can include nausea, vomiting, fever, headache, and general malaise. The level of encephalopathy can range from behavioral changes such as irritability to coma. Children may also present with seizures. Common neurologic signs and symptoms involving the brain and spinal cord include hemiplegia, pyramidal signs, vision changes (due to optic nerve involvement), and speech impairments (3).

When ADEM is suspected, a full neurologic examination is warranted. An MRI of the brain and spinal cord should be ordered—lesions may or may not enhance with gadolinium. The MRI images in Figure 19.4 demonstrate multifocal lesions associated with ADEM in a young boy.

Rapid determination of a potential viral etiology such as herpes simplex virus (HSV) is essential. Empiric treatment with acyclovir is warranted if a viral etiology is suspected. Cerebral spinal fluid and serologic studies should be collected to help rule out an infectious etiology. Bacterial, viral, and fungal cultures, as well as assays for HSV, Epstein–Barr virus, enterovirus, West Nile virus, and varicella zoster virus (VZV) should be sent (54,55). If anti-NMDAR encephalitis is suspected, then titers should be drawn. Collecting extra cerebrospinal fluid (CSF) for additional studies is wise as it may eliminate the need for a repeat lumbar puncture. The cerebral spinal fluid of ADEM is characterized by normal opening pressure, normal glucose, and moderately elevated cell count and protein (3). Rarely children will experience severe decompensation with increased intracranial pressure warranting surgical intervention (3). Approximately 2% of children with ADEM experience fulminant hemorrhagic demyelination with cerebral edema (50). This condition, called acute hemorrhagic leukoencephalitis is often fatal (55).

Treatment for ADEM

The first-line treatment for ADEM is corticosteroids intravenously, usually given as 1 g/day for 5 days in children over 40 kg or 20 to 30 mg/kg/day for smaller children. In addition to acyclovir, antibiotic coverage may also be started (3). IVIg is a second-line treatment which can be used in refractory cases or in situations where steroid treatment is not tolerated (3,55). The typical dose of IVIg is 2 g/kg over 2 to 5 days. Plasmapheresis with three to five exchanges may also be used in refractory cases (56). Cyclophosphamide has been used in some cases (55). In the acute period, supportive interventions in the intensive care setting may be necessary to maintain adequate ventilation in cases of respiratory failure, to control blood sugars, and to manage nutrition. After the

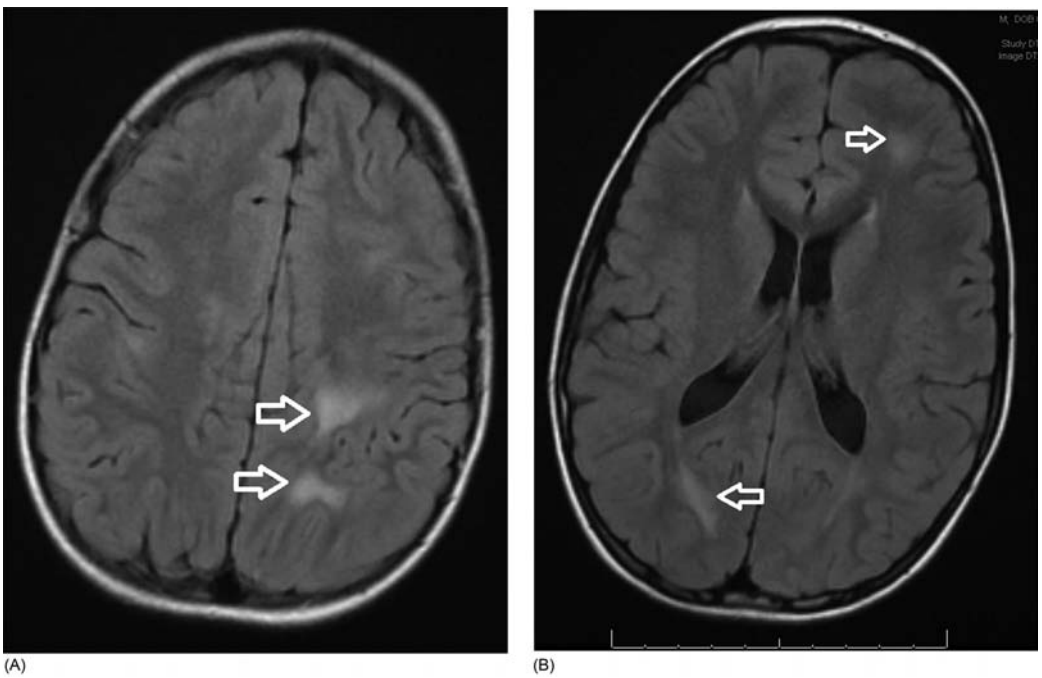


FIGURE 19.4 These two axial MRI FLAIR views of the brain demonstrate multiple lesions with increased diffusivity consistent with ADEM. The two largest lesions on each view are indicated with arrows.

Source: Images courtesy of Guilio Zuccoli, MD, Associate Professor of Radiology, University of Pittsburgh.

nadir is reached and the child is stable from a respiratory standpoint, they may be transferred to an acute floor or directly to comprehensive inpatient rehabilitation if it is warranted.

Recovery from ADEM is variable. Children with a history of viral illness may have worse outcomes (57). Poorer outcomes are seen with higher lesion load (58). Usually after improvement with treatment in the acute period, children make gradual improvements over time (weeks to months) (3,58). Although some researchers consider ADEM to be a relatively benign condition (50), children may be left with neurocognitive and behavioral consequences (59). Neurocognitive findings may persist after lesions are fully resolved on imaging (59,60). Children with a history of ADEM are at risk for recurrent disease. Approximately 10% of children with an initial diagnosis of ADEM have another demyelinating episode (50). Children with two ADEM episodes separated by at least 3 months and not associated with additional demyelinating episodes are considered to have multiphasic ADEM (6). Pediatric ADEM may be the first manifestation of MS or NMO. Periventricular lesions can occur in ADEM, but if present, raise suspicion for the possibility of an MS diagnosis in the future. Similarly, hypodense white matter lesions that persist are predictive of MS (6). The presence of a positive anti-aquaporin-4 IgG titer with ADEM is strongly suspicious for a subsequent NMO diagnosis.

Rehabilitation for ADEM

The literature on the rehabilitation of ADEM is sparse (61). Children hospitalized with ADEM who have persistent functional deficits should be considered for comprehensive inpatient rehabilitation. Debility may be superimposed upon the neurologic findings of ADEM and children treated with steroids may also have evidence of myopathy. Cognitive and language recovery lags behind motoric recovery, although some children will have lasting physical disabilities (56,58,61). Comprehensive inpatient rehabilitation should focus on recovery of lost function, be it family-centered and goal-directed. Speech therapists, neuropsychologists, and school reentry specialists are particularly important members of the rehabilitation team as many children may have lasting difficulties with attention and executive functioning (62,63). Children with ADEM may have subtle or more overt cognitive challenges and may also present with ongoing behavioral problems. Younger children tend to present and persist with behavioral problems more frequently than older children (3). Children with ADEM may benefit from adaptations in the classroom short term or long term depending on the persistence of their cognitive deficits. Neuropsychological testing may be performed shortly after the acute phase of the disease and should be repeated if abnormalities are identified.

PEDIATRIC ENCEPHALITIDES

PEDIATRIC ENCEPHALITIS

In this section of this chapter, encephalitis in children will be discussed. The first portion will address encephalitis generally. Then specific types of encephalitis will be described in more detail with particular emphasis on autoimmune encephalitis (excluding ADEM, which was described in the "Pediatric Demyelinating Diseases" section). The newly identified condition, anti-NMDAR encephalitis, will be highlighted.

Encephalitis is inflammation of the brain tissue with evidence of neurologic dysfunction (54,64). Encephalitis is different than encephalopathy and causes of encephalopathy such as toxic exposure should be ruled out (55,65). Generally speaking, encephalitis may be associated with infections or other inflammatory processes (54). There are well over 100 known pathogens that cause infectious or postinfectious encephalitis (64). There are also numerous noninfectious processes that cause encephalitis. A definitive etiology is identified in only about half of the cases (64). To be diagnosed with encephalitis, a child should exhibit neurologic dysfunction evidenced by altered mental status (behavior or level of consciousness) and have neuroimaging abnormalities consistent with inflammation and/or inflammatory cells in the cerebral spinal fluid (54,64).

Epidemiology of Pediatric Encephalitis

The estimated incidence of encephalitis has been reported to be 10.5 per 100,000, but is more common in infants (64). Surveillance systems vary internationally and likely underestimate the incidence of encephalitis (54). Additionally, the risks vary between populations. For example, neonates are at much higher risk of HSV encephalitis than other populations. Boys and girls tend to be equally at risk except for autoimmune encephalitis for which girls are at increased risk (54,55). Measles, mumps, and VZV can cause encephalitis (54) and may be on the rise, given the increasing loss of herd immunity associated with the decreased rates of immunization in the United States.

Clinical Course of Pediatric Encephalitis and Diagnosis

The typical presentation for encephalitis is prodromal "flu-like" illness followed by changes in behavior or consciousness, headaches, and nausea and vomiting. Children may also present with focal neurologic signs and/or seizures (54). Some presentations are more likely to be associated with particular types of encephalitis. For example, children with ADEM often have multifocal neurologic signs, HSV is often characterized by temporal lobe seizures, and arboviruses typically present with fever

and early obtundation (64). Important aspects of the history taking at the time of presentation include detailing potential exposures, recent illnesses and vaccines, history of autoimmune disorders, and immunosuppression status (64). In addition to a detailed physical with a complete neurologic examination, neuroimaging and CSF and serum studies are necessary. Blood work should include a culture, CBC with differential, renal, liver, and coagulation studies (54,64). The CSF usually shows lymphocytic pleocytosis, normal glucose, and elevated protein. Oligoclonal IgG bands are rarely seen. Serum and CSF IgM antibodies and/or rising IgG may help the identification of specific viral etiologies (54). CSF viral detection by polymerase chain reaction (PCR) amplification should also be sought to identify specific viral etiologies (54). Samples of both blood and CSF should be stored for future study and a repeat lumbar puncture may be needed when an etiology is not readily identified. Because of the challenges and expense associated with determining a specific etiology, investigations should be directed toward determining if the encephalitis is caused by an autoimmune process versus a viral process to help guide treatment (65). MRI findings vary with etiology. Figure 19.5 demonstrates several small lesions in a child with encephalitis.

For the child with suspected seizures, an EEG should be ordered. Nonspecific findings of diffuse high-amplitude slow waves are often seen (54). Although not pathognomonic for HSV encephalitis, temporal lobe periodic lateralizing epileptiform discharges are highly suggestive (54,64). The clinical course is variable depending on the etiology. Some children may require prolonged care in the intensive care setting and may have long-lasting consequences of their disease.

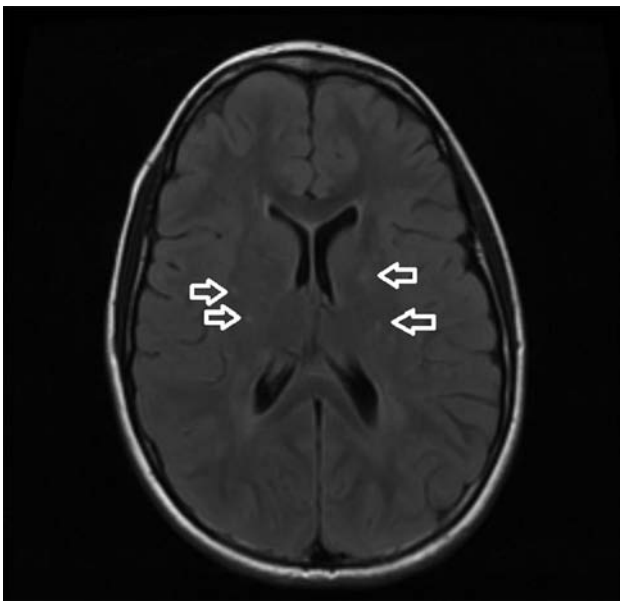


FIGURE 19.5 This axial MRI FLAIR image demonstrates multiple small enhancing lesions; the four most notable are indicated with arrows.

Source: Image courtesy of Guilio Zuccoli, MD, Associate Professor of Radiology, University of Pittsburgh.

Treatment of Pediatric Encephalitis

Empiric antibiotics should be administered urgently and all children with suspected encephalitis should be placed on acyclovir (64). The typical dose of acyclovir is 10 mg/kg every 8 hours for 3 weeks (55). If it is determined that the etiology is not viral, acyclovir may be stopped. Survivability and outcomes are markedly improved with aggressive early treatment with acyclovir for HSV encephalitis (54,55,65). Supportive care should be directed based on the child's needs. Children may need short- or long-term seizure management. Children with seizures at presentation, abnormal imaging, and focal neurologic signs are at higher risk for epilepsy (66). Children are often hospitalized for several weeks before disposition home or to comprehensive inpatient rehabilitation.

Rehabilitation of Pediatric Encephalitis

As described in the section on ADEM, comprehensive rehabilitation should be family-centered and goal-directed. Lasting motoric and cognitive sequelae are common (66). Having HSV encephalitis or another identified viral etiology, a longer hospital stay, and abnormal imaging are predictive of long-term deficits (66). The team approach employed in brain injury rehabilitation (see Chapter 17) should attend to the cognitive sequelae and school reentry. Neuropsychological testing can help identify the needs for adaptations in the classroom (66). Follow-up after disposition from comprehensive rehabilitation is important to ensure successful community integration and to maximize functional outcomes.

It is important to note that many children with encephalitis recover in the acute period and do not meet criteria for comprehensive inpatient rehabilitation. Despite having a good recovery in the acute period, many children have long-term cognitive and behavioral issues that can impact their learning and performance in school (54,66). Children with a history of encephalitis have higher rates of headaches, behavioral problems, sleep difficulties, attention deficit hyperactivity disorder, low IQ, and learning difficulties compared to other children (66). The pediatric rehabilitation medicine physician may be asked to evaluate a child with a remote history of encephalitis, and therefore should be knowledgeable about the long-term consequences.

HERPES SIMPLEX VIRUS ENCEPHALITIS

HSV encephalitis is the most common cause of viral encephalitis not associated with epidemics (54). Children

tend to present with fever, focal neurologic signs, and altered mental status. The common neurologic signs at presentation include headaches, seizures, vision changes, cranial nerve palsies, motor changes, and aphasia (64). Infants often have seizures and may present with apnea (64). The temporal lobe is most commonly affected, which can be associated with the symptom of déjà vu phenomenon (55). The MRI typically demonstrates gadolinium enhancement in the temporal lobe. MRI findings elsewhere in the brain do not exclude HSV encephalitis (55). Treatment with acyclovir has markedly improved survivability (54). Relapses of HSV encephalitis can occur. Despite the advances in medical management of HSV encephalitis, children often have lasting deficits. These deficits can include seizures, impaired cognitive function, motor deficits (especially hemiplegia in the child with unilateral involvement), and aphasia (55). Depending on the severity of the presentation, many children with HSV encephalitis will benefit from comprehensive inpatient rehabilitation. Infants tend to be discharged home and may need extensive services as an outpatient such as Early Intervention. In whatever setting the infant/child with encephalitis is seen, it is important to take a family-centered interdisciplinary approach to optimize functional outcomes.

VARICELLA ZOSTER VIRUS (VZV) ENCEPHALITIS

VZV, the cause of chicken pox, may invade the CNS during primary infection (55). VZV is now much more rare since widespread vaccination but may become more common as vaccination rates decline and herd immunity diminishes. Children who are immunosuppressed are at increased risk for VZV encephalitis and therefore immune status should be assessed as part of the history taking process (64). For VZV encephalitis, vasculitis is the major pathologic process (54). Invasion of the vascular endothelial cells can result in ischemic injury or vessel wall necrosis leading to dissection, hemorrhage, or aneurysm formation (55). Encephalitis can be a part of the reactivation presentation. Zoster ophthalmicus is often the initial complaint and then is followed by vasculitis, which can present as hemiplegia associated with stroke (55). The vasculitis can involve any vascular territory in the CNS, as well as the cranial nerves. Involvement of the middle cerebral artery and carotid is common (55). Similar to HSV encephalitis, the treatment of VZV encephalitis is acyclovir (54). The dose is 10 mg/kg every 8 hours for 2 weeks. Steroids may be considered to decrease the inflammation associated with the vasculitis (54,55). Children with VZV encephalitis frequently need comprehensive inpatient rehabilitation to address the functional deficits associated with their presentation. The principles of stroke rehabilitation are particularly relevant for these children. Long-term motor impairment is common after VZV encephalitis (54). Child status post VZV encephalitis should be monitored routinely to ensure optimal functional outcomes. Reactivation should be treated aggressively if it occurs to minimize cumulative disability.

ARBOVIRUS ENCEPHALITIS

The arthropod-borne viruses (arboviruses) tend to present in clusters (55). Most arboviral infections are subclinical, meaning that infected individuals do not know they have an infection. Arboviral encephalitis usually presents with a prodromal "flu-like" illness with fever, headache, and myalgia (64). Neuroinvasive West Nile virus infection can present as meningitis, encephalitis, and/or poliomyelitis-like anterior horn cell disease (55). In addition to the typical workup for encephalitis, EMG is indicated if the presentation is flaccid paralysis. Patients may have respiratory compromise when severely affected and death is not uncommon (67). Individuals with dysarthria and dysphagia early in their presentation may be at increased risk of respiratory compromise (67). Treatment is supportive and long-term functional sequelae are common. Rehabilitation is exceptionally important for the child with neuroinvasive West Nile. The prognosis for functional recovery after encephalitis is good. On the other hand, recovery after poliomyelitis-like presentation is poor (55).

There are over 100 other etiologies of infectious and postinfectious encephalitis. Table 19.7 details a subset of them with cues about their presentation and the treatment basics (54,55,64).

AUTOIMMUNE ENCEPHALITIS

This section will address immune-mediated encephalitis with emphasis on anti-NMDAR encephalitis. In the past several years, the understanding of autoimmune encephalitis has expanded considerably, although there are some forms of encephalitis that are presumed to be immune-mediated for which a mechanism has yet to be elucidated (68,69).

Anti-NMDA Receptor Encephalitis

Anti-NMDAR encephalitis is an immune-mediated encephalitis in which antibodies target the NR1 receptor subunit (68). The condition was first described in 2005 with an elucidation of the etiology in 2008 (70,71). The first cases were in young women with ovarian teratomas, but relatively quickly the diagnosis was made in children with no evidence of paraneoplastic processes (70–76).

Epidemiology

Although anti-NMDAR encephalitis was first identified among young women, it is now understood to be a leading

TABLE 19.7 THE PRESENTATION AND TREATMENT FOR COMMON TYPES OF ENCEPHALITIS

ETIOLOGY	PRESENTATION	TREATMENT
Viral		
• St Louis encephalitis	• Encephalitis or meningoencephalitis	• Supportive
• Eastern Equine encephalitis	• Early stupor or coma, very young	• Supportive
• Mumps	• Unimmunized, meningoencephalitis	• Supportive
• Measles	• Unimmunized, hemorrhagic component	• Supportive
• Parvovirus	• B19, Fifth disease	• Supportive
• Enterovirus	• Children on rituximab, immunosuppressed, hand–foot mouth disease can involve brainstem	• Pleconaril (not available in the United States)
• Cytomegalovirus	• Infants and immunosuppressed children	• Ganciclovir
• Influenza A&B	• During outbreaks	• Oseltamivir
Bacterial		
• *M. pneumoniae*	• Postinfectious encephalitis	• Doxycycline
• Listeria	• Brainstem (rhombencephalitis)	• Ampicillin
• Rickettsia	• Cerebral vasculopathy	• Doxycycline

cause of encephalitis among adults and children. It is the second leading cause (after ADEM) of immune-mediated encephalitis (77). The California Encephalitis Project found that among individuals younger than 30 years old the rates of anti-NMDAR encephalitis surpassed all viral etiologies (78). Among young children there does not appear to be a strong gender predominance, but girls are more commonly affected than boys in adolescence (79,80). Except for among young women, identification of a neoplasm is rare (68).

Clinical Course of Pediatric Anti-NMDAR Encephalitis and Diagnosis

The clinical presentation of anti-NMDAR encephalitis frequently includes mood and behavior changes, seizures, altered levels of consciousness, dyskinesia, and functional deterioration (70–76). Younger children tend to present similarly but may have more subtle behavioral symptoms as their initial presenting complaint (68). The differential diagnosis for anti-NMDAR is broad. Table 19.8, adapted from Armangue (68), details the differential.

Prodromal symptoms are often vague and nonspecific. Usually within 3 weeks, psychiatric symptoms begin for a vast majority of patients. These symptoms often include hallucinations, delusions, confusion, agitation, amnesia, anxiety, paranoia, sleep disturbances, and bizarre behavior (68,80). Because individuals with anti-NMDAR encephalitis often present with psychotic symptoms, the diagnosis, and therefore, the appropriate management is frequently delayed. It is not uncommon for a patient to present from a psychiatric facility

TABLE 19.8 THE DIFFERENTIAL DIAGNOSIS OF ANTI-NMDAR ENCEPHALITIS

DISEASE	CHARACTERISTICS
Viral encephalitis	Tends not to present with movement disorders and psychosis. Usually the presumed diagnosis. Empirically treated with acyclovir.
Limbic encephalitis	Paraneoplastic, autoimmune (described later in the chapter).
Encephalitis lethargica	Poorly understood (described later in the chapter).
New onset psychosis	Psychiatric features in anti-NMDAR encephalitis tend to present before more overt neurologic signs. Early misdiagnosis is common.
Drug use	Ketamine and phencyclidine can present with behavioral changes and involvement of dopaminergic pathways.
Neuroleptic malignant syndrome (NMS)	Rigidity, altered consciousness, hyperthermia, and autonomic instability can be among the symptoms of both NMS and anti-NMDAR encephalitis.
Inborn errors of metabolism	Many may present with acute or subacute encephalopathy.
Childhood disintegrative disorder (CDD)	CDD presents with rapid loss of language skills, cognitive decline, seizures, and autistic features. The prognosis is poor.

Source: Adapted from Ref. (68). Armangue T, Petit-Pedrol M, Dalmau J. Autoimmune encephalitis in children. *J Child Neurol.* 2012;27(11):1460–1469.

TABLE 19.9 THE DIAGNOSTIC CRITERIA FOR CATATONIA USING THE *DSM-5* AND *DSM-IV*

***DSM-5* (NEW CRITERIA)**	***DSM-IV***
At least three of the following	At least two of the following
• Catalepsy (passive induction of a posture held against gravity) • Waxy flexibility (slight and even resistance to positioning) • Stupor (no psychomotor activity; no engagement with environment) • Mutism (cannot co-occur with aphasia) • Negativism (opposing/not responding to external stimulus) • Posturing (active spontaneous maintenance of posture against gravity) • Stereotypies (repetitive, abnormally frequent, nongoal-directed movements) • Grimacing • Echolalia (mimicking others' speech) • Echopraxia (mimicking others' movements)	• Motor immobility (catalepsy or stupor) • Excessive motor activity • Extreme mutism or negativism • Peculiarities of voluntary movements (posturing, sterotypies, prominent mannerisms, prominent grimacing) • Echolalia or echopraxia

Source: Adapted from Ref. (81). Tandon R, Heckers S, Bustillo J, et al. Catatonia in *DSM-5*. *Schizophr Res*. 2013;150(1):26–30.

after symptom progression (80). Most patients develop symptoms within the first month in at least four of the following areas: memory, speech, behavior, cognition, seizures, movement, and consciousness (79). Catatonia can occur in anti-NMDAR; therefore the clinician should be knowledgeable in making the diagnosis of catatonia (80). The diagnostic criteria for catatonia have been updated in the *Diagnostic and Statistical Manual for Mental Disorders Five* (*DSM-5*). Table 19.9 details the criteria from the *DSM-IV* and the *DSM-5* (81).

The psychiatric symptoms of anti-NMDAR encephalitis typically progress toward frank neurologic symptomatology. Patients develop seizures, dyskinesias, decreased level of consciousness, and autonomic instability (68,82). Younger children are more likely than older children or adults to present with neurologic features at the onset of presentation (82), but many younger children present with temper tantrums, aggression, and other behavioral changes (80). The movement disorders of anti-NMDAR encephalitis, which occur in a majority of pediatric cases, include choreoathetoid movements, rigidity, parkinsonism, myorhythmia (a resting tremor with a lower frequency than a parkinsonian tremor), opisthotonus, orofacial dyskinesias, and myoclonus (80,83,84). When movement disorders dominate the presentation, the clinician should be aware of other autoimmune conditions that also present with new onset movement disorders such as Sydenham's chorea, pediatric autoimmune neuropsychiatric disorder associated with streptococcal infections (PANDAS), SLE, Rasmussen's encephalitis (discussed in the following), and Sjogren syndrome (83). Children, most commonly girls, with Sydenham's chorea usually present in childhood after streptococcal infection with hemichorea, ballism, or generalized chorea. Children with PANDAS, on the other hand, are usually boys presenting with tics, obsessive compulsions, and abrupt onset after streptococcal infection (83). Chorea can be the presenting symptom of SLE and may be unilateral or bilateral (83). Children with SLE often have associated cognitive dysfunction and psychiatric issues (85). Up to 25% of individuals with Sjogren syndrome experience movement disorders including parkinsonism, akinesia, chorea, and dystonia (83), which may look similar to the movement disorders of anti-NMDAR encephalitis.

The autonomic instability of anti-NMDAR encephalitis, which is more common in adults, can occur in children and is often characterized by tachycardia, hypertension, and hyperthermia (68). Hypoventilation and decreased consciousness can necessitate mechanical ventilation that may be needed for a prolonged period of time (80). Time in the intensive care setting can lead to additional complications including disuse muscle wasting and loss of mobility. Time needed in the pediatric intensive care unit (PICU) varies and can last days to weeks to months.

Some authors believe that three phenotypes of anti-NMDAR encephalitis exist and that by categorizing patients into these phenotypic groups, outcomes can be better predicted (86). DeSena and colleagues describe Type 1 (classic type) as characterized by seizures, movement disorders, catatonia, agitation/aggression, mood and behavioral disturbances, and a predominantly catatonic/stuporous state lasting less than 2 months (86). Type 2 is characterized by primarily psychiatric symptoms with very limited, if any, time in a catatonic state, while Type 3 is primarily catatonic. Patients with Type 3 spend more than 2 months in a catatonic or stuporous state and often have severe movement disorders (86). In their review of cases, DeSena and colleagues found the most favorable recovery trajectory for Type 2 and the least favorable for Type 3 (86).

The diagnosis of anti-NMDAR encephalitis is confirmed by the identification of autoantibodies against the NMDA-type glutamate receptors in the CSF and/or serum (82,87). The brain MRI is normal in approximately half of all cases and approximately a third of pediatric

cases (68,88). When abnormal, the MRI frequently shows cortical and subcortical T2-FLAIR signal abnormalities typically involving cortical and subcortical regions of the brain (68,88). EEG is abnormal in a vast majority of patients (82,87). Almost all patients have diffuse background slowing in the delta–theta range (68). Some patients exhibit extreme delta brush, which consists of essentially continuous delta activity with superimposed fast activity (usually in the beta range) in the frontal regions (68,82). The identification of a tumor is rare in children but a screen should be carried out. The role of repeat screening for boys and for girls under the age of 12 is less clear. In teenaged girls, screening MRI of the abdomen and pelvis every 6 months for 4 years is recommended (79).

There are cases of anti-NMDAR encephalitis that overlap with demyelinating syndromes (88). Therefore, the pediatric rehabilitation physician should be knowledgeable about overlapping symptoms as they may present during outpatient follow-up. In a cohort of 691 cases of confirmed anti-NMDAR encephalitis, 23 (3.3%) had clinical or MRI findings consistent with demyelination (88). Twelve individuals, five of whom were children, had a demyelinating syndrome episode separated in time from their episode of anti-NMDAR encephalitis. The remaining 11, 6 of whom were children, had co-occurring anti-NMDAR encephalitis with multifocal or extensive T2-FLAIR abnormalities (88). These cases indicate the presence of two active immune mechanisms. If concurrent demyelination is suspected, testing for aquaporin-4 and myelin oligodendrocyte glycoprotein antibodies is recommended as the treatment of demyelinating disease is often more intense (88).

Treatment of Anti-NMDAR Encephalitis

Early identification with aggressive immunotherapy limits morbidity (87). The recovery course is often prolonged and multiphasic with autonomic instability resolving first (87). First-line treatment is intravenous (IV) steroids with an oral taper in combination with or followed by IVIg and/or plasmapheresis (80). In patients with inadequate responses or deterioration, rituximab and/or cyclophosphamide should be considered (80). In pediatrics, cyclophosphamide is often considered a last resort because of the side effects, which include infertility, premature gonadal failure, and malignancies (68). In a multi-institutional study of 577 pediatric and adult patients, aggressive immunotherapy with tumor removal (if present) resulted in substantial recovery in over 80% of patients (79). Patients who failed first-line treatment had worse outcomes, but among those who failed first-line treatment, those who received second-line treatment fared better than those who only received first-line therapy (79). Relapses occur in approximately 20% of cases and are more common among individuals without identifiable tumors (68,80). Second-line therapy reduces the risk of relapses (79). Mycophenolate mofetil has been used in refractory cases (82,84,87).

Symptom management for movement disorders, sleep irregularity, and psychiatric issues is often necessary, as is seizure management. There is no standard approach to address these symptoms. Multidisciplinary experts should collaborate to manage the predominant symptoms that change over time. The psychiatrist may be heavily involved at presentation, followed by management by neurology with an eventual shift to the rehabilitation team. The movement disorders of anti-NMDAR encephalitis tend to improve with immune therapy and tetrabenazine (84). Catatonia should be managed with benzodiazepines and electroconvulsive therapy can be considered as a last resort for malignant catatonia (80). Atypical antipsychotics and mood stabilizers, such as valproic acid, have some utility (80). Trazodone, clonidine, benzodiazepines, and diphenhydramine can be helpful in the management of sleep disturbances (80). Full symptom control is unlikely, but the employing symptom management techniques are important adjuncts to immune therapy.

Rehabilitation of Anti-NMDAR Encephalitis

Patients with anti-NMDAR encephalitis benefit from a multidisciplinary team of professionals including rehabilitation specialists (80). The timing of comprehensive rehabilitation depends on the capability of the rehabilitation center to manage the medical issues especially the autonomic instability and medication infusions, along with the rehabilitation needs of the patient (89). In a case series of 20 pediatric patients, the median length of the acute care hospitalization was 56 days (82). Of those 20 patients, it is unknown how many required comprehensive inpatient rehabilitation. Autonomic instability tends to resolve first along with improved consciousness; once this has happened, the psychiatric manifestations may reemerge, which can pose challenges in the rehabilitation setting (68). The rehabilitation program should be interdisciplinary, family-centered, and goal-directed. Having an interdisciplinary team with behavioral health specialists is important because children frequently have impulsivity, inattentiveness, behavioral disinhibition, and challenges with social interaction (68). Cognitive recovery tends to occur gradually (80), highlighting the value of speech therapy and neuropsychological services. Because behavioral issues, sleep disturbances, and movement disorders are frequently present during the recovery phase, the rehabilitation physician should be well versed in managing these issues.

Although a majority of patients make a good recovery, a large proportion of children with anti-NMDAR encephalitis do not make a full recovery (80), and children frequently leave the inpatient rehabilitation setting with persistent functional deficits (89). Ongoing post-discharge therapy and behavioral services are often warranted especially since a protracted but full recovery is possible. Recovery can occur a year after initial presentation (82). Those who have incomplete recovery can still continue to make functional progress 2 years after initial presentation (79).

Therefore, ongoing follow-up with rehabilitation medicine is of value to monitor therapeutic needs over time. Neuropsychological testing should be performed at intervals that allow for the tracking of cognitive recovery and planning for school adaptations if needed. In cases of relapse, comanagement with neurology should be considered because management options include starting first-line treatment again or moving directly to second-line management with rituximab, cyclophosphamide, or mycophenolate mofetil (82).

LIMBIC ENCEPHALITIS

Prior to the elucidation of the etiology of anti-NMDAR encephalitis, cases of anti-NMDAR encephalitis fell under the umbrella of limbic encephalitis (69). Limbic encephalitis is an inflammatory process of the limbic system (medial temporal lobes, amygdala, and cingulate gyrus) (68), with MRI abnormalities classically in the medial temporal lobes (80). Individuals with limbic encephalitis tend to present with rapid memory loss, temporal lobe seizures, and psychiatric symptoms (90). Like anti-NMDAR encephalitis, the paraneoplastic presentation is more common in adults. True limbic encephalitis is rare in children (68). Ophelia syndrome, limbic encephalitis associated with Hodgkin's lymphoma, can affect children. Proper identification of the syndrome is important because it can facilitate the diagnosis of lymphoma and is very responsive to treatment (68).

OPSOCLONUS MYOCLONUS

Opsoclonus myoclonus is also called dancing eye syndrome because of the characteristic chaotic, rapid multidirectional eye movements. In children, presentation is usually in the first 2 years of life. Irritability, myoclonus, tremor, ataxia, drooling, and other oral motor problems are frequent presenting symptoms (68). Approximately half of the children with opsoclonus-myoclonus will have an underlying malignancy, most commonly neuroblastoma (91). Though direct evidence is lacking, the condition is assumed to be autoimmune in nature and is somewhat responsive to immune therapy (92). Despite treatment and tumor resection, many children remain quite symptomatic with ongoing problems with behavior and cognition (68). In addition, some children experience insomnia and abnormal pain responses (93). Relapses are frequent, occurring in about 50% of patients (68).

RASMUSSEN'S ENCEPHALITIS

Rasmussen's encephalitis is a very rare condition of unilateral inflammation of the cerebral cortex. Children present with partial seizures and focal myoclonus (83). It is a progressive disease in which children have partial seizures that are refractory to antiepileptic medications (94). Additionally, children have progressive hemiplegia, cognitive decline, and unilateral brain atrophy (68,94). Children present usually around the age of six (94). There are three stages of the disease—a nonspecific prodromal phase with low seizure frequency and mild hemiplegia; an acute phase with frequent seizures (may be in epilepsia partialis continua), progressive hemiplegia and cognitive deterioration; and a residual phase in which seizures persist and the neurologic deficits are stable (95). The diagnosis is made by the presence of (a) focal seizures and unilateral cortical deficits, (b) EEG findings of unihemispheric slowing and unilateral seizure onset, and (c) MRI findings of focal cortical atrophy and T2-FLAIR hyperintense signal abnormality and/or hyperintense signal or atrophy of the ipsilateral caudate head (94,95). Treatment with high-dose steroids, plasmapheresis, IVIg, and rituximab can help ameliorate the progression of symptoms (68). T-cell inactivating medications such as tacrolimus and azathioprine have been shown to be helpful as well (94). Definitive treatment is hemispherectomy (94). The rehabilitation of hemispherectomy will be discussed later in this chapter.

There are other encephalopathies that are associated with refectory seizures that are suspected to be induced by an inflammatory process. They include acute encephalitis with refractory repetitive partial seizures (AERRPS), devastating epilepsy of school-aged children (DESC), and fever-induced refractory epileptic encephalopathy syndrome (FIRES) (68).

PEDIATRIC BRAIN TUMOR REHABILITATION

The focus of this section of the chapter is on pediatric brain tumor rehabilitation. There is considerable overlap with Chapter 17; therefore the focus of this section will be the unique aspects of brain tumor rehabilitation. In particular, medulloblastomas and posterior fossa syndrome will be highlighted.

Brain tumors can be benign or malignant and some benign tumors can recur as malignant tumors. In 2007, the World Health Organization (WHO) updated their classification schema for brain tumors (96). Table 19.10 details the WHO grading system (96). Whether benign or malignant, the space-occupying nature of brain tumors can have profound impacts on children.

EPIDEMIOLOGY

Collectively, brain tumors are the second most common type of childhood cancer, the most common type of solid pediatric tumors, and the most common cause of

TABLE 19.10 WHO GRADING SYSTEM OF BRAIN TUMORS

Grade I	Nonmalignant, slow growing with good long-term survival
Grade II	Relatively slow growing, may recur as higher grade tumors, may be malignant or nonmalignant
Grade III	Malignant and often recur as Grade IV tumors
Grade IV	Aggressive rapidly reproducing malignant tumors

Abbreviation: WHO, World Health Organization.

childhood death from cancer (97). The incidence rate of pediatric brain tumors is between 3.3 and 4.9 per 100,000 person-years (97,98). The most common pediatric brain tumor is astrocytoma (97). Medulloblastoma is the most common malignant brain tumor in children (99), and the second most common brain tumor overall (97). Before the age of 2, supratentorial tumors predominate; thereafter infratentorial tumors do until late adolescence (100).

Some children are predisposed to the development of brain tumors (101). Between 10% and 20% of children with TS develop slow growing subependymal giant-cell astrocytomas (102). Children with neurofibromatosis types 1 and 2 (NF-1 and NF-2) are also at increased risk of brain tumors (103,104). Approximately, 15% of children with NF-1 develop optic gliomas and pilocytic astrocytomas (105). Tumors are much more common in NF-2. Bilateral vestibular schwannomas are characteristic of NF-2; multiple meningiomas occur in over 50% of individuals with NF-2 and tend to be more complicated than in children without NF; and spinal cord tumors, especially ependymomas, are common (105,106). Von Hippel–Lindau is characterized by hemangioblastomas, which rarely occur supratentorially (105,107). Other genetic syndromes that predispose children to CNS tumors include Li–Fraumeni syndrome, Gorlin syndrome, and Turcot syndrome (101,105). Table 19.11 details the common brain tumors of childhood, their WHO grade, and their characteristics (96,101,108–113)

CLINICAL COURSE AND DIAGNOSIS OF PEDIATRIC BRAIN TUMORS

The presenting symptoms for brain tumors can be highly focal or more generalized. Many children present with

TABLE 19.11 WHO GRADING OF COMMON PEDIATRIC CENTRAL NERVOUS SYSTEM

TUMOR TYPE	GRADE	CHARACTERISTICS
Juvenile pilocystic astrocytoma	I	Often involves the cerebellum, usually complete resection is possible, well-defined borders, excellent prognosis
Diffuse fibrillary astrocytoma	II	Usually in the midbrain, hard to fully resect, not well-defined margins
Glioblastoma	IV	Rare in children, histologically different than Grade IV gliomas in adults, poor prognosis with 2-year survival in 10% of cases
Medulloblastoma	IV	Neuroectodermal tumor of the posterior fossa with at least four variants, heterogeneous, frequently relapses
CNS primitive neuroectodermal tumor (PNET)	IV	Very poor prognosis (worse than medulloblastoma), usually supratentorial, when resection is attempted care given to preserving neurological function versus total resection
Atypical teratoid/rhabdoid tumor (ATRT)	IV	Rare tumor type (1%–2%) of pediatric brain tumors but 20% of brain tumors in young children, very poor prognosis
Ependymoma	II	Can occur in the posterior fossa, supratentorial brain, or spinal cord; cure rates improving, surgical resection is key, resection and radiation lead to cure in about 65% of cases, third most common brain tumor in children
Meningioma	I	Rare in children, but commonly multifocal in children with NF-2
Hemangioblastoma	I	Very rare but seen in children with von Hippel–Lindau
Craniopharyngioma	I	Arise from Rathke's pouch, variants may be WHO grade II, side effects common (such as diabetes insipidus)
Schwannoma	I	Seen in NF-2, especially bilateral vestibular schwannomas
Neurofibroma	I	Benign peripheral nerve tumors that may be seen in individuals without NF-1, but commonly present in NF-1, spinal neurofibromas are often not symptomatic

Abbreviation: WHO, World Health Organization.

symptoms associated with increased intracranial pressure such as headache (especially in the morning), vomiting, vision changes, and cranial nerve abnormalities (108). Hydrocephalus occurs in approximately half of pediatric brain tumor cases and is almost always obstructive in nature (114). Depending on the location of the tumor, children may have ataxia, motor impairments, seizures, and growth abnormalities. Children with posterior fossa tumors frequently present with discoordination and ataxic gait, but they may also present with cognitive impairments and behavioral issues (115). The very young child may present with failure to meet milestones, loss of milestones, irritability, lethargy, and failure to thrive (108). Seizures are more common among certain tumor types such as astrocytomas, which are slow growing (116). Definitive diagnosis is made through histologic evaluation of the tumor tissue but clinical findings and neuroimaging can provide a preliminary diagnosis in most situations.

The neurologic sequelae of pediatric brain tumors depend on the tumor location and size, the presence of metastases, and the treatment course, which often includes resection, chemotherapy, and neuraxis radiation. Additionally, the developmental stage of the child predicts outcomes (100). The brain of the young child is especially vulnerable because of the rapid cell proliferation, dendritic and axonal growth, and rapid myelination that occurs during infancy and early childhood (117). Table 19.12 details some of the late symptoms of pediatric CNS tumors (118).

It is important to note that the treatment for pediatric brain tumors can lead to substantial neurologic consequences that can negatively impact function and quality of life. CNS radiation can have progressive and potentially devastating impacts. The amount of injury is related to the dose and extent of the CNS irradiated, as well as the age of the child. Children can experience endocrinopathies, vasculopathies, hearing loss, cognitive deficits, radiation-induced necrosis, radiation-induced neoplasms, and if the spinal column is irradiated, children may experience radiation myelitis (100,108,119). Whole brain radiation in the very young child limits the child's ability to achieve functional independence later in life (118).

The long-term impacts of chemotherapy on the nervous system are less well understood and the research evidence limited (100,120). Leukoencephalopathy is a rare complication. Other toxic complications include peripheral neuropathy and hearing loss. See Table 19.13 for neurologic side effects of common chemotherapy agents (100,120).

TABLE 19.12 COMMON LATE SYMPTOMS OF PEDIATRIC CENTRAL NERVOUS SYSTEM TUMORS

Central tumors	• Visual impairments/optic atrophy/abnormal eye movements • Diabetes insipidus • Hypothalamic/pituitary axis deficiencies • Hydrocephalus • Short stature • Weight gain • Vertigo
Hemispheric tumors	• Weakness • Sensory changes • Seizures • Cognitive deficits
Posterior fossa tumors	• Dysarthria or mutism • Discoordination • Visual impairments • Behavioral problems
Brainstem tumors	• Cranial nerve dysfunction • Visual impairments • Pyramidal signs • Discoordination
Spinal cord tumors	• Paralysis • Spinal deformities • Neurogenic bowel and bladder

Source: Adapted from Ref. (118). Walker DA, Finlay J. Central nervous system tumors of childhood and adolescence: the rehabilitation challenge of survival and "true cure". *J Pediatr Rehabil Med*. 2011;4(1):23–29.

MEDULLOBLASTOMAS AND POSTERIOR FOSSA SYNDROME

Because medulloblastomas are the most common malignant pediatric brain tumor and the tumor type most commonly associated with posterior fossa syndrome, they will be discussed in greater detail here. The peak incidence of medulloblastoma is around 6 years of age (112). Some conditions such as ataxia telangiectasia, Rubinstein–Taybi syndrome, Turcot syndrome, Gorlin syndrome, and Li–Fraumeni syndrome are associated with increased risk of medulloblastoma (99). Children typically present with ataxia and discoordination (115).

TABLE 19.13 CHEMOTHERAPY NEUROLOGIC SIDE EFFECTS

Methotrexate	Stroke-like encephalopathy, chronic leukoencephalopathy, ascending radiculopathy (if given intrathecally), neurocognitive compromise
Vincristine/Vinblastine	Axonal sensorimotor polyneuropathy
Cisplatin and Carboplatin	High-frequency sensorineural hearing loss
Cyclophosphamide/Ifosfamide	Reversible neurotoxicity characterized by somnolence, disorientation, and hallucinations
Steroids	Myopathy

Medulloblastoma is a primitive neuroectodermal tumor (PNET) (101). There are different subtypes of medulloblastoma identified by gene expression patterns and also different histologic subtypes (99). These subtypes help stage the tumor clinically, which helps determine which treatment protocol to use. Additionally, the amount of residual tumor and the presence of metastatic disease help determine whether the tumor is considered standard or high risk. Metastatic disease is identified in approximately 30% of cases and disseminates along the CSF pathway (99). Metastases outside of the CNS are very rare (99). Current therapy includes maximal safe tumor resection and craniospinal radiation with adjuvant chemotherapy (112). The surgical goal is maximal resection with reduction of mass effect and restoration of CSF flow, while minimizing the damage to the neuroanatomy (99). Current protocols for standard-risk disease have over an 80% cure rate (112). Unfortunately, relapsed medulloblastoma carries a dismal prognosis (112). The treatment for medulloblastoma (as is true of other tumor types) can lead to extensive neurologic sequelae that can worsen over time (99,120). Table 19.14 details some of the reported consequences (99,108,119–122).

Medulloblastomas need to be distinguished from other PNETs and atypical teratoid rhabdoid tumors (ATRT) (123). In the past, these three tumors were classified together. Newer research demonstrates that they are distinct entities. PNETs often occur supratentorially and can be differentiated from medulloblastomas based on their genetic profile (123). They also carry a worse prognosis than medulloblastomas. ATRTs are very aggressive tumors that tend to affect the very young (124). Most patients with ATRT die within a year of diagnosis (123).

TABLE 19.14 NEUROLOGIC SEQUELAE OF MEDULLOBLASTOMA TREATMENT

Cognitive	• Mental processing speed • Fine motor skills • Memory • Concentration
Psychological	• Internalizing behavior • Global distress • Depression
Neurologic	• Cerebellar dysfunction • Posterior fossa syndrome • Sensorineural hearing loss (due to radiation and/or ototoxic chemotherapy)
Endocrine	• Hypothyroidism • Growth hormone deficiency • Precocious or delayed puberty • Hypocortisolism
Secondary neoplasms	• Malignant gliomas • Meningiomas

Posterior fossa syndrome, a consequence of approximately 25% of posterior fossa tumor resections, is very common in medulloblastoma (99,125). It was first described in 1979 in children with medulloblastoma (126). The literature often used the term cerebellar mutism to describe the temporary loss of speech postoperatively (127). For a time, the term "mutism and subsequent dysarthria" was employed to describe the transient mutism followed by severe dysarthria that eventually resolved (128). With the recognition that children also experienced neurobehavioral and emotional problems, as well as dysphagia, decreased initiation of voluntary movements, and other neurologic problems, the term posterior fossa syndrome was adopted (129). Emotional liability (including apathy and irritability), ataxia, axial hypotonia, and brainstem dysfunction all commonly occur in posterior fossa syndrome (125,128). Children may also have cranial nerve findings and substantial dysphagia (99,125). Usually children have a period of normal speech followed by mutism, which manifests between 1 and 2 days after surgery (115,130). Children tend to experience variable recovery, with some children having persistent symptoms beyond 1 year (99,125,130). The recovery of speech typically includes dysarthric monotonous and ataxic speech with subsequent language disturbances such as agrammatism, word finding difficulties, lack of verbal initiative (adynamic language), and reading and writing difficulties (128).

The pathogenesis of posterior fossa syndrome has not been fully elucidated (99,115,128). That being said, splitting the cerebellar vermis and damage to the dentate–thalamic–cortical tracks are associated with posterior fossa syndrome (99,125,130). Hypoperfusion is also a potential culprit (128). The cerebellum is involved in many of the complicated neural functions of the brain including equilibrium, movement, attention, memory and other cognitive functions, and emotional control (115). Although more commonly reported in adults, children with cerebellar damage may be considered to have cerebellar cognitive-affective syndrome characterized by executive dysfunctions, mood disturbances, visual-spatial difficulties, and language impairments (128,131). There is considerable overlap between posterior fossa syndrome and cerebellar cognitive-affective syndrome, which can lead to confusion of terms. In general, posterior fossa syndrome is the acute syndrome that occurs very shortly after posterior fossa surgery (128). Refinement of the definitions of mutism and subsequent dysarthria, posterior fossa syndrome, and cerebellar cognitive-affective syndrome is warranted as is continued research into the etiologies of these conditions.

TREATMENT OF PEDIATRIC BRAIN TUMORS

The oncologic management depends on the histology of the tumor, its resectability, and the age of the child. The

current management strategy for brain tumors is to maximize survival while minimizing treatment effects (110). The mainstay of immediate management of an identified brain tumor is surgical (108). Surgical treatment may be required to control hydrocephalus. Usually an external ventricular drain (EVD) is placed temporarily (114), but there is no consensus for the optimal management of hydrocephalus (99). Depending on the location of the tumor, surgery may be performed to achieve a gross-total resection, a near-total resection, a subtotal resection or a biopsy. For example, a gross-total resection of a craniopharyngioma may not be desirable if the resection would cause substantial hypothalamic damage (108). Newer technologies including intraoperative MRI, navigational guidance, and functional mapping are advancing neurosurgical care for brain tumors (108). When ongoing management of hydrocephalus is required, a ventriculoperitoneal (VP) shunt is usually placed, although often, a third ventriculostomy may be performed (114). Some patients will require VP shunting in the face of third ventriculostomy failure (114).

After the tumor type is histologically identified, oncologic management may include monitoring or initiation of radiation and/or chemotherapy. During active treatment with chemotherapy, children can experience substantial hematopoietic suppression that can put them at risk for infections, fatigue, and hemorrhage. Therefore therapies should be tailored to minimize the risk of hemorrhage while the platelet count is low and should accommodate the patient's need for isolation when their absolute neutrophil count is low. For children with symptomatic anemia, a transfusion may be warranted. Children undergoing active treatment may have debilitating fatigue from a variety of causes. Good sleep hygiene should be promoted as should adequate pain management, exercise, and treatment of anxiety. In addition, certain chemotherapy agents, such as vincristine, can cause peripheral neuropathy, which can complicate the recovery trajectory. Treatment for neuropathic pain may be warranted. Some children may be placed on deep vein thrombosis prophylaxis due to the increased risk of clotting associated with neoplasms and immobility (132). The rehabilitation team should be monitoring for the signs and symptoms of thrombosis in all patients with brain tumors.

Radiation treatment deposits energy into the tissue and disrupts cells (119). Although radiation indiscriminately damages the DNA of both normal cells and cancer cells, the cancer cells are less capable of repairing double stranded breaks (119). The use of radiation has markedly improved survival (122). The acute side effects of radiation include headaches, fatigue, hair loss, skin redness and throat irritation (119). The long-term effects of radiation are of a greater concern. Radiation to the very young brain leads to substantial neurocognitive impairments (99). Because of this, some protocols exclude very young children from receiving neuraxis radiation. Traditionally, craniospinal radiation is followed by a boost to the tumor bed for malignant tumors (108). For lower grade tumors, radiation to the tumor bed is frequently warranted (108). The dosing of radiation is dependent on the tumor type, stage, and the age of the child (108). Proton radiation therapy, compared to standard photon radiation, provides a more targeted and tissue-sparing approach (133). Standard photon radiation has infinite range, compared to proton radiation, which has a finite range meaning that the radiation dose is deposited at the end of the penetration range (119). Success with using proton radiation has been documented (134). But access to proton therapy is limited (108). Given that proton radiation is the most promising form of radiation to decrease treatment sequelae (119), the pediatric rehabilitation physician should expect to see more patients receiving this treatment as availability increases.

The neurocognitive consequences of radiation emerge in the years after treatment. Developmental stagnation rather than regression is the likely reason for lower IQ scores as children age (117,120). In addition, children often experience emotional and behavioral problems including inattentiveness and internalizing emotional symptoms (120). Because the frontal lobes are more susceptible to radiation-induced injury, acquired frontal lobe symptoms may become apparent (122).

Seizures associated with brain tumors are notoriously difficult to control. In addition, there are special considerations when recommending antiepileptic medications. Enzyme-inducing medications (carbamazepine, phenytoin, and phenobarbital) can increase drug–drug interactions and are therefore often prohibited in research trials (116). Recent studies have demonstrated the antineoplastic effect of valproic acid as well as the antiepileptic effects of temozolomide, an antineoplastic agent (116). For intractable epilepsy, especially in the temporal lobe, epilepsy surgery may be indicated with intraoperative electrocorticography to identify the epileptogenic tissue (116).

REHABILITATION OF PEDIATRIC BRAIN TUMORS

The goal of cancer rehabilitation is to achieve optimal functioning taking into consideration the limits imposed by the disease itself and its treatment (100). Creation of a rehabilitation plan for children should be initiated as soon as possible. Ideally, the rehabilitation team would be familiar with the patient and family prior to tumor resection and would be involved in functional goal setting during each of the stages of treatment (100). The establishment of specific goals by an interdisciplinary team helps enhance the functional autonomy of pediatric patients with brain tumors (100). Similar to the impairments seen in other acquired brain injuries, children with brain tumors can experience a wide range of deficits. Debility

TABLE 19.15 DIETZ CATEGORIES OF CANCER REHABILITATION

Preventative Rehabilitation	Early intervention to prevent or delay the symptoms related to the disease or its treatment
Restorative Rehabilitation	Goal to achieve premorbid level of functioning
Supportive Rehabilitation	Goal to optimize function in light of substantial impairments related to the disease and/or its treatment
Palliative Rehabilitation	Primarily focuses on comfort, caregiver education, and training and provision of appropriate equipment

is a common problem especially if rehabilitation is occurring simultaneously with radiation or chemotherapy.

The interdisciplinary goals should be adapted and refined during the recovery trajectory. In 1969, Dietz developed four categories of cancer rehabilitation as shown in Table 19.15 (135).

During preventative rehabilitation, even prior to oncological management, the rehabilitation team can help establish a plan of care, provide education, train the family and patient in therapeutic exercises, and help prognosticate regarding function after treatment. If a child is suspected to lose speech, voice banking can be performed presurgery to assist with augmentative communication postoperatively (100).

During the treatment phase, a child may be appropriate for a comprehensive inpatient rehabilitation stay depending on the level of disability and ability to participate in intense therapies (100). Children receiving concurrent rehabilitation and radiation may need their radiation scheduled late in the day to accommodate for the level of fatigue often experienced after radiation. For younger children who may require sedation for radiation, the timing of therapy sessions before radiation becomes especially important. Some children require temporary cessation of the intense rehabilitation as acute medical issues are addressed (136). As is true for other children undergoing rehabilitation for acquired brain injury, children with brain tumors should have services directed at successful community reintegration including school accommodations, adaptive equipment planning, and training to incorporate compensatory strategies as needed (99,100,120). A formal plan that encompasses schooling during treatment, school reentry after treatment and continued monitoring to update school accommodations should be set (120).

The role of the pediatric rehabilitation physician continues after the child completes treatment. During surveillance, the rehabilitation team may be able to provide more intense therapies as tolerance and endurance improve (100). Neuropsychological batteries should include general IQ testing as well as tests of attention, working memory and memory, processing speed, and visual-perceptual skills (120). Executive functions (volition, planning, purposeful action, and effective performance) are the cognitive processes principally controlled by the prefrontal cortex and are often impacted by radiation (122). Ideally, the first neuropsychological battery should be given before radiation or immediately following its completion (120). It is well established that children with pediatric brain tumors have lower performance and verbal IQs than their peers (117). Performance IQ, which relies more heavily on motor function, tends to be more vulnerable than verbal IQ (117). Therefore, adaptations to the academic curricula of children with brain tumors are often warranted. Children may also need psychological support for optimal functioning due to the neurobehavioral consequences of their disease and treatment. Table 19.16 details some of the cognitive consequences of pediatric brain tumor treatment (122).

In the face of recurrent disease, the rehabilitation team may be challenged by new functional deficits and the impacts of very aggressive treatment. Oncologic treatment may be directed toward cure, temporization, or palliation depending on the aggressiveness of the tumor, its responsiveness to treatment, and the child's and family's wishes. While active rehabilitation is often overlooked during palliation, children continue to benefit from optimization of functioning and their families from caregiver training. For children who are no longer mobile, families may need specialized equipment to assist with transfers. The rehabilitation team can collaborate actively with the palliative care team to help maximize comfort and quality of life.

The pediatric rehabilitation physician should always be attending to the quality of life of their patients. This is particularly true for the child with a brain tumor because the treatment of pediatric brain tumors can have profound impacts on the child's well-being. More aggressive tumors that require more aggressive treatment are associated with lower quality of life scores than less aggressive tumors (137). Parents tend to report lower quality of life scores than their children (133,137). This perhaps relates to their understanding of the long-term consequences of pediatric brain tumors, or the association of perceived quality of life and the impact on families with children undergoing treatment for brain tumors (133). Because of the longitudinal relationships that pediatric rehabilitation physicians tend to have with their patients and the general rehabilitation goals to improve functioning and quality of life, the pediatric rehabilitation physician is in a unique position to help children and their families to adapt to the consequences of having a brain tumor and its treatment. The rehabilitation treatment goal at this stage is to minimize the impact of the sequelae on daily life (99). The pediatric rehabilitation physician may need to provide guidance about independent living and employment because the likelihood of maintaining successful employment is lower for pediatric brain tumor survivors (120).

TABLE 19.16 NEUROCOGNITIVE PROCESSES IMPACTED BY PEDIATRIC BRAIN TUMOR TREATMENT

COGNITIVE PROCESS	DESCRIPTION	IMPACTS/FINDINGS	INTERVENTIONS
Attention	Selectively focusing on one aspect of the environment while ignoring others	• Academics—math and reading • Friendships • Self-esteem • Achievement	• School accommodations • Methylphenidate
Working Memory	Temporary mental workspace where information is manipulated or maintained	• Greatest decline noted shortly after treatment • Limits academic learning • Poor social outcomes	• School accommodations • Cognitive training
Processing Speed	How quickly one can perform relatively automatic mental tasks	• Continues to decline over time • Impacts ability to interact with peers and others	• School accommodations • Cognitive training
General Executive Functions	• Metacognition • Organization • Reasoning • Problem solving	• Worsening deficits over time	• School and work accommodations • Cognitive training

SURGICAL TREATMENT OPTIONS FOR INTRACTABLE EPILEPSY

This section of the chapter will be devoted to nonmedical options for the management of intractable epilepsy. Intractable epilepsy goes by many terms including medically refractory, treatment resistant, and drug resistant epilepsy. Although there tends to be a general consensus regarding what constitutes intractable epilepsy, there is not a universally accepted definition clinically or in research. Epilepsy that fails to respond to the first two to three antiepileptics with at least one seizure every month for 2 years is considered intractable (138). One caveat to the definition is that trials of antiepileptics need to be appropriately tailored based on the seizure syndrome and type (138). Therefore, drug resistant epilepsy is defined by failure to control despite adequate trials of two appropriate and well-tolerated antiepileptic medications (139). Surgical intervention for intractable epilepsy may be curative or palliative (140).

EPIDEMIOLOGY

Seizure disorders occur in 1% of children and adolescents (141). Disability associated with seizures has declined in the past decade (142), but for some children seizures are debilitating and difficult to control. Seizures are intractable in less than one-third of children with epilepsy (140,143,144). Some genetic conditions such as TS are associated with a strong predisposition toward developing intractable epilepsy (145). A vast majority of children with TS will develop seizures that are frequently medically refractory (145,146). Focal seizures in TS may precede or evolve into infantile spasms, or may coexist. First-line treatment for infantile spasms in babies with TS is vigabatrin (147). Nearly all young children with TS who present with focal seizures or infantile spasms develop other seizure types as they age that are challenging to treat medically.

For children with hemimegalencephaly and hemispheric cortical dysplasia (rare congenital brain malformations characterized by early intractable epilepsy, intellectual impairment, and contralateral hemianopsia and hemiplegia), anatomic or functional hemispherectomy is usually indicated (148). Children with acquired conditions such as brain tumors and traumatic or otherwise acquired brain injuries are also at risk for intractable epilepsy. For example, for children with Rasmussen's encephalitis surgery is curative by hemidisconnection in which the affected hemisphere is disconnected by either hemispherectomy or functional hemispherectomy (94).

DIAGNOSIS OF INTRACTABLE EPILEPSY AND PRESURGICAL WORKUP

Before proceeding with surgical evaluation, medical management should be optimized. Antiepileptic medication management should be determined first by the seizure syndrome, if one can be identified, and then the seizure type. Monotherapy is ideal and when a second agent is added because of inadequate response, removal of the first agent should be considered (substitution monotherapy) (138). Some antiepileptics are synergistic and are therefore given together as rational polytherapy (138). Table 19.17 is adapted from McTague and details commonly occurring issues to consider when making a diagnosis of intractable epilepsy (138).

TABLE 19.17 COMMON ISSUES FACED WHEN MAKING THE DIAGNOSIS OF INTRACTABLE EPILEPSY

DIAGNOSTIC FACTORS	TREATMENT FACTORS	PATIENT FACTORS
Misclassification of seizure type	Inappropriate type of antiepileptic drug (AED)	Medication adherence
Misclassification of seizure syndrome	Inadequate dosing of AED	Unrealistic expectations about seizure frequency
Failure to identify an underlying cause	Inappropriate combination of AED	Patient lifestyle
	Failure to address provoking factors	
	Drug–drug interactions	

Source: Adapted from Ref. (138). McTague A, Appleton R. Treatment of difficult epilepsy. *Arch Dis Child*. 2011;96(2):200–204.

Children with medically refractory epilepsy should be referred early for surgical evaluation. Outcomes are improved at high volume centers (149), so if possible a referral should be made to a highly experienced center. Occasionally, children without refractory epilepsy should be referred for operative evaluation. Potential reasons for referral include significant toxicity of antiepileptics, desire for pathologic diagnosis, notable growth and mass effect, and risk of rupture from arteriovenous malformation (150).

The presurgical workup should include EEG, neuroimaging, neuropsychological testing, a complete neurologic and functional examination, and additional corroboration if needed (138). Neuroimaging maps to the interictal and ictal EEG findings (147,151). When the seizure focus is not lesional, peri-ictal single-photon emission computed tomography and/or a subtraction image coregistered to the MRI may help localize the onset location (151). Magnetoencephalography is also a helpful tool in localizing seizure foci and may be more accurate than EEG (145,147,151). Intracranial EEG is often used to localize or refine localization, which can help minimize the size of the resection (145). See Figure 19.6 for a plain film x-ray of a child status post grid placement for EEG monitoring and mapping.

Neuropsychological testing should evaluate intelligence, memory, executive functioning, attention, and behavior (151). Focused speech and language assessments should also be performed as well as assessments of motor and sensory functions. Language and memory lateralization may be determined by functional MRI or Wada testing (amobarbital sodium is injected into the carotid to inactivate the ipsilateral cerebral hemisphere to allow for testing of the contralateral hemisphere temporarily) (151). Many centers exclusively use functional MRI due to the invasive nature of the Wada test.

Cortical resection can include functional (eloquent) cortex that is associated with predictable neurological deficit such as hemiplegia and homonymous hemianopsia (94). Understanding the consequences of sacrificing eloquent cortex is important as patients and families decide to pursue surgical management (140). The risks and benefits need to be thoroughly discussed (138). When deciding to pursue seizure surgery, it is important to look beyond the likelihood of seizure freedom and also consider the potential impacts on function, quality of life, and other outcome domains (152).

TREATMENT OF INTRACTABLE EPILEPSY

The Ketogenic Diet

The mainstay of nonpharmacologic nonsurgical treatment is the ketogenic diet. The ketogenic diet has been used for seizure management since the 1920s, first a primary management and then as secondary treatment after the development of antiepileptic medications. The ketogenic diet is an attractive adjunct when surgery is contraindicated (147). It has been shown to decrease seizures in numerous observational studies (138). In a randomized controlled trial, over 40% of treated children had greater than 50%

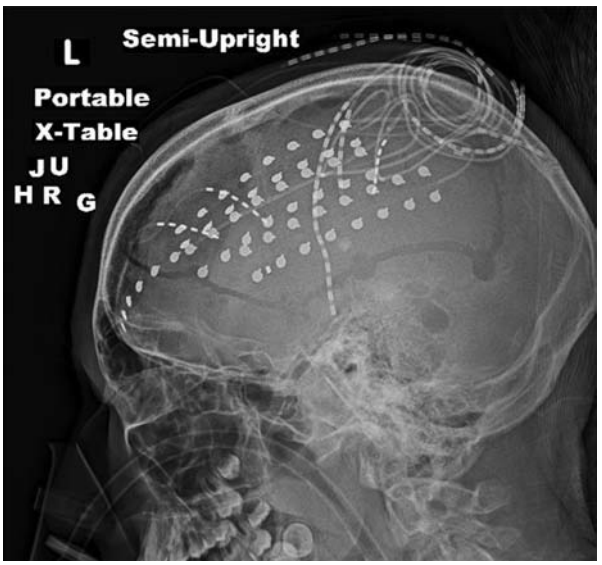


FIGURE 19.6 X-ray of a child's skull with EEG grid placed for mapping the seizure focus.

Source: Image courtesy of Guilio Zuccoli, MD, Associate Professor of Radiology, University of Pittsburgh.

TABLE 19.18 VARIATIONS OF THE KETOGENIC DIET

DIETS	CHARACTERISTICS
Classic Ketogenic Diet	3:1 or 4:1 fat to protein and carbohydrate ratio
Medium chain triglycerides (MCTs)	Allows more flexibility with protein and carbohydrates because MCTs provide more ketones
Modified Atkins	1:1 fat to protein and carbohydrates, which is easier to manage
Low glycemic index therapy	10% of calories may be carbohydrates but only ones with low glycemic index (<50)

reduction in seizure frequency (153). Unfortunately, the classic ketogenic diet is challenging to maintain, but variations have been shown to be effective (151). Table 19.18 details the variations of the ketogenic diet (151).

Vagus Nerve Stimulation

Vagus nerve stimulation (VNS) was approved by the Food and Drug Administration in 1997 for patients 12 years and older as adjunctive therapy for partial onset seizures (143,154). The VNS device is implanted in a subcutaneous pocket near the left clavicle with the lead implanted along the vagus nerve in the carotid sheath (154). Ascending fibers from the vagus nerve track to the tractus solitarius and then from there to the limbic, reticular, and autonomic regions of the brain (154). The mechanism of action is poorly understood but may be through desynchronization activity through repetitive stimulation to these fibers (143). In an analysis of 3,321 patients' status post-VNS, approximately 50% of patients obtained a clinically significant reduction in seizure frequency (143). A clinical response to VNS can take up to a month to be notable (154). The best outcomes were achieved among patients with post-traumatic epilepsy and TS (143). Seizure reduction with VNS seems to be somewhat better among children than adults (143). In addition to reducing seizure frequency, seizure severity and recovery time may be decreased (151). Common side effects include hoarseness and cough, with pain, dyspnea, and headache being less commonly reported (143,154). Individuals with a history of drooling or dysphagia may have worsening of those symptoms (154). VNS malfunction is rare and batteries last 7 to 10 years (154).

Surgical Resection

The classical management was anatomic hemispherectomy in which the entire cerebral hemisphere, including the deep structures, was removed (148). Typically, though, when a patient is reported to have a hemispherectomy, he or she has had a functional hemispherectomy, which involves removal of the affected temporal and parietal lobes and a disconnection of the frontal and occipital lobes from the unaffected hemisphere (148,155). Variations include modified lateral hemispherectomy, vertical parasagittal hemispherectomy, or peri-insular hemispherectomy (156). Overall though, the most common procedures performed are focal or lobar resections in the frontal and temporal lobes (138).

Other operative options include corpus callosotomy and multiple subpial resections. Corpus callosotomy prevents interhemispheric propagation of epileptic discharges and therefore prevents generalization of seizures (156). It is used as a palliative intervention for some children with intractable generalized seizures. It is especially beneficial among children with Lennox–Gastaut syndrome (151). Multiple subpial transections may be performed when typical epilepsy surgery is contraindicated due to the consequences to the eloquent motor, sensory, visual, or speech areas. This procedure interrupts the horizontal synchronizing neuronal networks while preserving vertical ones (151). Multiple subpial resections have been shown to reduce seizure frequency in adults and children, but the durability of the efficacy has been called into question (156).

TREATMENT OUTCOMES AND REHABILITATION

Surgical success is graded on the Engel Classification (146). The Engel Classification is provided in Table 19.19 (157).

In a meta-analysis of outcomes, 56% of patients achieve Engel I and 13% get Engel II (146). The outcomes tend to be better among children with TS, among whom two-thirds achieve seizure freedom (Engel 1) and among children undergoing temporal lobectomy (145,150). In general, predictors of good outcomes (Engel I or II) are absence of generalized seizure semiology, mild or no developmental delay, abnormal preoperative MRI, and unifocal ictal scalp EEG abnormality (146,150). Patients with lesional epilepsy were more likely than those with nonlesional epilepsy to achieve seizure freedom (150). Rates of seizure freedom are higher with temporal lobe focus resections than frontal ones (138). When a tumor was the epileptogenic focus in the temporal lobe, gross-total lesion resection was associated with better outcomes

TABLE 19.19 ENGEL EPILEPSY SURGERY OUTCOME SCALE

Class I	Free of disabling seizures
Class II	Rare disabling seizures (almost seizure-free)
Class III	Worthwhile improvement of seizures
Class IV	No worthwhile improvement of seizures

than subtotal resection. Approximately 94% of children who had gross-total resections were seizure-free versus 38% of children who had subtotal resections (150). Importantly, better outcomes are achieved with nondominant hemisphere surgery (94). Seizure surgery is rarely fatal. Mortality after seizure surgery is 0% to 2% in children, higher than reported in adults (156).

The functional outcomes after seizure surgery have been evaluated in observational studies. In children with pre-existing hemiparesis, the hemiparesis was not worsened in about half of the cases, with over one-third reporting worsening hemiparesis (155,158). In the cases where hemiparesis worsened, the upper extremity tended to be more affected than the lower extremity (155). Some children have preservation of motor function. Two possible mechanisms have been proposed to explain this: (a) in abnormal brains, ipsilateral pathways are strengthened and refined to address functional demand, and (b) axonal sprouting and cortical reorganization of the ipsilateral pathways develop new functional pathways (155). Cognitive changes postoperatively were noted in about 50% of cases nearly equally divided between improvement and worsened cognitive status (155). Everyday memory skills do not appear to be impacted by seizure surgery (159). Postoperative outcomes are better among children with higher levels of development presurgery and children with shorter seizure disorder durations. But outcomes tend to be better among young children compared to young adults likely due to incomplete brain maturation in young children (155). Contralateral brain abnormalities predict poor motor and language outcomes (158).

Predictably, there is scant literature on pediatric rehabilitation after seizure surgery. In the treatment of Rasmussen's encephalitis, with rehabilitation after hemispherectomy, ambulation is usually achievable (94). A comparative analysis of adult patients' status post temporal lobe epilepsy surgery who did or did not participate in comprehensive inpatient rehabilitation demonstrated better employment outcomes for the rehabilitation group (160). In terms of outpatient rehabilitation, a small case series of constraint-induced movement therapy led the investigators to consider it a feasible method of rehabilitation (161). Additionally, locomotor gait training was associated with subjective mobility improvement and functional MRI evidence of cortical remodeling (162).

When considering the rehabilitation treatment options for children after epilepsy surgery, it is important to evaluate their deficits and the likelihood of functional recovery to tailor the rehabilitation program. As is true in other types of acquired brain injury rehabilitation, the treatment approach should be interdisciplinary, goal-directed, and family-centered. Optimally, the pediatric rehabilitation physician should know of the child's functional status preoperatively as well as postoperatively to assist with treatment planning, especially as it relates to hemiplegia. Children with a history of intractable epilepsy experience worse cognitive and developmental outcomes than children with controlled epilepsy (140); therefore, even after the postoperative phase, the rehabilitation team has the opportunity to develop strategies to help children be as successful as possible. Interventions in the school setting may be indicated to help address cognitive and motoric deficits. Additionally, the child may have persistent seizures that may impact school performance and attendance. Long-term follow-up is warranted when the child has ongoing deficits that impact his or her function.

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REHABILITATION OF CHRONIC PAIN AND CONVERSION DISORDERS

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Chronic pain and conversion disorders continue to be a diagnostic and treatment challenge for many physicians. They both require a creative, multidisciplinary approach for optimal outcomes. This chapter will summarize the current opinions, diagnostic strategies, and treatment protocols regarding these two conditions.

A. CHRONIC PAIN

UNDERSTANDING PAIN

Historically, pain was considered a symptom of a disease and if the underlying cause were cured it would no longer exist. The International Association for the Study of Pain (IASP) defines pain as "An unpleasant sensory and emotional experience associated with actual or potential tissue damage, or described in terms of such damage" (1). As our knowledge about pain continues to expand, there is evidence that chronic pain may cause more long-term harm than the initial disease or injury (2). Chronic pain is further defined as persistent and recurrent; it is a significant problem in the pediatric population estimated to affect 20% to 35% of children under the age of 18 around the world (3). Chronic pain can cause significant disruption in many facets of the patient's life. It is not uncommon to observe in the pediatric population that suffers from chronic pain to be avoidant of school and social activities and demonstrate symptoms of clinical depression (4).

The system that controls pain is complex and involves both ascending and descending pathways. A change in the pain conducting system occurs after tissue injury. Chronic pain may be due to neurons being pushed to their limits. Nociceptors that once only responded to noxious stimuli begin to fire in response to nonpainful stimuli causing receptors to evoke activity in the nociceptive system that is then interpreted as pain or central sensitization (2,5). Pain may be upregulated in order to receive immediate attention and withdraw from a stimulus or downregulated during a "fight or flight" response.

Pain is necessary, as it allows us to process a potentially harmful situation and withdraw from or investigate the source to prevent further injury. Pain is one of the most common reasons that people seek medical attention. Inadequately controlled pain may result in unnecessary suffering resulting in compromised care of the underlying disease and lead to depression (6).

Caregivers of children who do not feel pain or have "congenital insensitivity to pain," need to be consistently vigilant to maintain a safe environment for their young (7). However, not all circumstances are preventable and these children may suffer severe injuries from painful events such as biting their tongue or burning their mouth with a hot beverage. Fortunately, this is an uncommon condition. Not all insensitivities to pain are congenital and some can be developed over time, for example, diabetic peripheral neuropathies or other peripheral nerve disorders/injuries.

BRIEF HISTORY

There was a time when many health professionals believed that babies could not perceive pain and their response was reflexive with little if any emotional ties. It was theorized that the nervous system of babies is underdeveloped at birth and their ability to experience pain was altered (8,9). During the 1980s in the United States, pediatric pain became a topic of focus after infant Jeffrey Lawson underwent surgical correction of a patent ductus arteriosus (PDA) with only pancuronium

bromide (neuromuscular blocker) and no analgesia, causing his mother to go to the press. After Jeffrey Lawson's story made national headlines, pain control for even the youngest of patients became a priority.

PREVENTION OF CHRONIC PAIN

Treating pain acutely may be the best way of preventing chronic pain. Studies of male infants undergoing circumcision displayed an altered, intensified reaction to vaccination at 4 to 6 months compared to male infants who did not undergo circumcision (10). Long-term effects on the memory with subsequent painful experiences in neonates are unknown; however, memory is thought to play an important role in later pain events. Early exposure to painful stimuli may have long-lasting effects on perception and response to pain (11,12).

PAIN SCALES

Commonly, pain is accepted as the "fifth vital sign"; scoring systems for pain have become the expectation of patients and families and now are used as a benchmark by hospital accrediting bodies to improve the quality of care that patients receive on a daily basis (2). Despite their utility, pain intensity scores are inherently an oversimplification as they disregard elements such as the location, quality, and the emotional experience of pain (13). Currently, many pain assessment tools are available for evaluation of neonates, infants, children, and adults. Unfortunately, the majority of these scales require self-report as the primary means of assessment. Common self-report scales that are used include: the numerical rating scales (NRS), Visual Analog Scales (VAS), and Wong–Baker FACES Scale (Figure 20A.1). Observational scales for the developing child include: Faces, Legs, Activity, Cry, and Consolability Scale (FLACC) (14) and the Child Facial Coding System. Other assessment tools that may be used for children include: Échelle Douleur Enfant San Salvador (DESS), the Pediatric Pain Profile, the Non-Communicating Children's Pain Checklist (NCCPC), and the Pain Indicator for Communicatively Impaired Children (15–17). Providers should be aware of the many available pain scoring resources and should make their assessment with the most appropriate scoring systems given the patient's age, intellectual ability, and experience.

As previously mentioned, pain is complex and incorporates both sensory and emotional experience. Therefore, further research and validation of these tools are necessary, as they do not fully encompass the pain experience.

CHRONIC PAIN IN THE DEVELOPMENTALLY DELAYED

Evaluating the level of pain a developmentally delayed child is experiencing can be challenging in both the acute and chronic setting. Unfortunately, the term *developmentally delayed* tends to group many different intellectual and developmental disabilities into one category despite the possibility that characteristics of each condition may differ. Commonly, developmentally delayed children have multiple comorbidities that increase their exposure to health care providers and the likelihood of undergoing painful procedures. Many patients will not be able to verbally express their perception and experience thus leading the health care provider to make inferences of the level of pain and proper treatment modalities. Also, many impaired patients may express pain differently than those without limitations. In fact, some may lack expression completely or have a unique way expressing discomfort (18,19).

			2	3	4	5	6	7	8	9	10
Verbal Description Pain	No Pain	Mild Pain			Moderate Pain			Severe Pain			Worst Pain Possible
Wong–Baker Facial Grimace Scale*	Face 0 is very happy because he or she does not hurt at all.		Face 2 hurts just a little bit.		Face 4 hurts a little more.		Face 6 hurts even more.		Face 8 hurts a whole lot.		Face 10 hurts as much as you can imagine, although you do not have to be crying to feel this bad.
Activity Tolerance Scale	No Pain	Can be ignored			Interferes with tasks and concentration			Interferes with basic needs			Bed rest required
*The Wong–Baker Facial Grimace Scale is commonly used for pediatric patients and can be found at wongbakerfaces.org											

FIGURE 20A.1 Wong–Baker FACES scale example.
Source: Adapted from Perret D, Chang E, Hata J, et al. *The Pain Center Manual*. New York, NY: Demos Medical Publishing LLC; 2014: 163.

Early recognition and treatment of painful conditions is key to prevention of chronic pain states. Utilization of specialized assessment tools may prove to be helpful but cannot be used in isolation. A thorough assessment can be difficult to obtain in this population, given the frequent challenges in effectively communicating somatic complaints. Self-report pain scales for developmentally delayed children in most circumstances cannot be recommended as a first-line assessment tool, rather observational scales may be more appropriate (20). Studies have also shown that caregiver reports and beliefs are often incomplete or inaccurate, thus necessitating the use of multiple tools for the most accurate assessment (21,22).

A multidisciplinary approach that includes the patient's primary care provider and other specialists who are working closely with the patient can prove to be beneficial to accomplish a common goal. It is important to involve the patient's caregivers to gain insight into what they believe the patient may or may not be going through, but even then, requires significant inference. The overall goal of treatment should be directed toward quality of life improvement.

NEUROPATHIC PAIN IN CHILDREN

Neuropathic pain in children is well documented with an incidence reported to be around 6% (23). While there are some disease processes and causes of neuropathic pain that adults and children share, the picture of neuropathic pain in children is different. Many common causes of neuropathic pain in adults such as diabetes and trigeminal neuralgia are rare in childhood. Disease processes that are common in both include complex regional pain syndrome (CRPS), which will be discussed later, and phantom limb pain. There are multiple known causes of neuropathic pain in children (Table 20A.1) in addition to these, such as Fabry's disease, HIV, and cancer to name a few. Children more frequently, although still rarely, may have neuropathies due to metabolic and toxic causes such as heavy metal positioning or due to neurodegenerative or mitochondrial disorders. Of these, CRPS is likely the most recognized.

TABLE 20A.1 CAUSES OF NEUROPATHIC PAIN IN CHILDREN

Cancer	1° Nervous system tumor, tumor invasion of nerve, chemotherapeutic and radiation therapy, surgery
Genetic	Erythromelalgia, Fabry's disease
Infectious	HIV, Herpes zoster
Neurologic	Multiple sclerosis, Guillain–Barré, CIDP
Toxic	Mercury, lead
Trauma	Direct nerve injury, phantom limb, CRPS

Abbreviations: CIDP, chronic inflammatory demyelinating polyneuropathy; CRPS, complex regional pain syndrome.

PHANTOM LIMB PAIN

Children may undergo traumatic or iatrogenic amputation just as their adult counterparts. Children who have undergone surgical amputation of limbs have up to a 40% incidence of phantom limb pain (24). In the pediatric population, it is rarely associated with complications of diabetes of vascular disease, as these disease processes have not been long-standing to lead to amputation. Phantom pain can also occur in children with congenitally missing limbs; however, they are much less likely to experience pain compared to children who lose their limbs later in life (25,26).

Phantom limb pain is children has been described as sharp, stabbing, piercing, squeezing and can lead to significant disability and affect functionality (25).

There are several factors that predispose children to phantom limb pain including older age at the time of amputation, concomitant use of chemotherapy, and preoperative pain. Similar to phantom limb pain is brachial plexus injury.

POSTOPERATIVE AND TRAUMA-RELATED PAIN

The incidence of neuropathic pain in children following surgery is not clear, but is well reported. Common causes of persistent postoperative pain in children are postthoracotomy pain as well as pain following various orthopedic procedures especially those associated with fracture.

CANCER-RELATED NEUROPATHIC PAIN

The incidence of neuropathic pain in children with cancer is unknown. However, it is known that neuropathic pain in children with cancer can be difficult to treat just as in their adult counterparts. A significant number of children with primary nervous system tumors report neuropathic pain. In addition, chemotherapy is a major cause of neuropathic pain in cancer patients, adult and children alike. Common chemotherapeutics that result in peripheral neuropathy are platinum agents such as cisplatin, and vinca alkaloids such as vincristine and vinblastine.

FABRY'S DISEASE

While Fabry's disease is overall an uncommon cause of neuropathic pain, it merits mention, given it is a disease in which pain is often the presenting symptom and the age of presentation is in the first decade (25,27). Pain is initially intermittent but will generally become constant over time. Fabry's disease is an X-linked recessive disease that leads to accumulation of glycolipids in multiple

organs including the central nervous system (CNS). Pain incidence in this disease is quite high with up to 70% of affected males reporting pain (27). Furthermore, treatment of Fabry's disease with enzyme replacement will lead to a decrease in pain scores although it does not decrease the incidence of pain (25).

MULTIPLE SCLEROSIS (MS)

It is common to find adults with MS-related pain. Of those with MS, approximately 5% will have their first symptoms prior to the age of 16 (25). There are multiple types of pain children with MS may experience including neuropathic pain. They, as their adult counterparts, may suffer from trigeminal neuralgia.

POSTHERPETIC NEURALGIA (PHN)

Although the herpes zoster infection or "shingles" is mostly known in adults, children too can experience PHN. While the incidence of herpes zoster in children does not reach that of adults, immunocompromised children can develop this infection and may go on to develop PHN. The incidence of herpes zoster is particularly prevalent in children undergoing treatment for acute lymphoblastic leukemia (25).

RADICULOPATHY

Although seen less frequently in children than the adult population, children can present with radicular pain secondary to herniated discs. Children with vertebral anomalies such as pars defect, transitional or sixth vertebrae can be prone to further injury just as the adult population. In addition, children can suffer traumatic fracture of vertebrae or traumatic spondylolysis, which we have seen in contact sport injuries.

TREATMENT OF NEUROPATHIC PAIN IN CHILDREN

As in adults, neuropathic pain in children can be difficult to treat. Tricyclic antidepressants, gabapentin, and pregabalin have the most frequent use in children. There are also reports of use of intravenous (IV) and intrathecal ketamine as well as topical treatments including lidocaine and capsaicin cream. In addition to common pharmacologic management, it is also important to remember that sometimes treatment of the underlying disease can lead to improved neuropathic pain. As is discussed in the following, pediatric pain is best treated within the context of the biopsychosocial model with the best responses seen with a multimodal approach to pain.

Although less commonly performed in children than in adults, procedural interventions may be warranted. Procedures such as epidural steroid injections or spinal cord stimulation may be considered in the treatment of children; however, such interventions do not have the same supporting evidence that is present for adults.

COMPLEX REGIONAL PAIN SYNDROME (CRPS)

CRPS, formerly known as reflex sympathetic dystrophy, is well described in pediatric literature. Diagnostic criteria for CRPS can be defined by methods typically used to assess adult patients utilizing either The Budapest Criteria or the IASP Criteria most frequently. There are two defined types of CRPS: CRPS-1, where there is no defined nerve injury, and CRPS-2, which can be traced to a direct nerve injury (28). The disease can be debilitating, commonly characterized by allodynia, hyperalgesia, and burning pain (29). Children with CRPS are prone to significant emotional distress, school absenteeism, and decreased social interaction. There are some notable differences seen in children compared to the disease in adults. The first is a large gender gap with a female-to-male ratio between 3:1 and 6:1 (30). Also found in children is a large preference of the disease for the lower extremity (31). In addition, the long-term prognosis for children with CRPS is more favorable in comparison to their adult counterparts. Specifically, greater than 90% of children can achieve remission of the disease and this is almost always accomplished with physical therapy (PT), desensitization, mirror visual feedback, and cognitive behavioral therapy (CBT) alone (26,32,33).

It is now better understood that CRPS is not only a peripheral problem but strongly involves the neocortex, hence the role of multidisciplinary treatment (34). Children with CRPS respond best to noninvasive interdisciplinary programs with a focus on PT. Children tend to do well without any intervention apart from those used to facilitate PT in the early rehabilitation period. Desensitization therapy with continued use and mobilization of the limb is the mainstay of PT. Psychological therapy is also a cornerstone in the treatment of CRPS in children. Multiple models exist for accomplishing aggressive PT and behavioral therapy including inpatient models, day hospital treatment programs, and outpatient programs (35). Mainstay medication therapy used in children with CRPS mirrors those used in adults, which includes antidepressants, antiepileptic drugs (AEDs), as well as nonsteroidal anti-inflammatory drugs (NSAIDs) and tramadol. Opioids are of limited benefit and are best avoided.

Although the basis of treatment is noninterventional, a small percentage of children will fail noninvasive therapy or will at some point in their treatment undergo interventional pain management. Various types of intervention such as peripheral nerve blockade, lumbar sympathetic nerve blockage, epidural infusion, and spinal

cord stimulation are reported in children (32,36). However, the majority of children will recover without such interventions and mainstream therapy remains focused on PT and CBT with positive outcomes.

MUSCULOSKELETAL PAIN IN CHILDREN

Musculoskeletal pain is a common complaint in older children and adolescents with the most common complaint being low back pain (37). It is important to assess for possible underlying causes when first evaluating children with such complaints. A thorough exam can lead to the discovery of a hypermobility syndrome, which can be assessed with the Beighton scoring system, or early symptoms of an underlying rheumatologic disorder may be revealed.

Children with musculoskeletal pain are prone to becoming adults with chronic pain. The basis of pain in children is often multifactorial in nature with psychosocial and lifestyle factors in addition to physiologic causes (37). There is a strong prevalence for musculoskeletal pain for adolescent females (38). Variable evidence exists to support the idea that having examples of chronic pain, such as a parent with chronic back pain, predisposes children to pain.

The mainstay of treatment of musculoskeletal pain is best accomplished with multidisciplinary treatment consisting of CBT, PT, and pharmacotherapy. Appropriate pharmacotherapy for musculoskeletal pain in children is with the use of NSAIDs with possible role adjuvants such as antidepressants, AEDs, or medications such as tramadol. Of key importance in the use of these therapies is a focus on function and improved school attendance. If functionality is not achieved, adolescents with chronic pain are almost ensured to become adults with chronic pain.

PEDIATRIC FIBROMYALGIA

Fibromyalgia, also referred to as pain amplification syndrome, follows a similar yet distinct pattern in children compared with adults and requires specialized knowledge on the part of practitioners. Currently the diagnosis is based on clinical parameters; however, there is a potential autoimmune mechanism that is being considered as a contributing factor of childhood fibromyalgia (39). The major difference when comparing adult fibromyalgia to juvenile fibromyalgia is that children may have a better prognosis; specifically, they may outgrow the condition. Fibromyalgia is characterized as persistent diffuse pain, fatigue, poor sleep, and the presence of tender points on physical exam (40). Standard diagnostic criteria for fibromyalgia in children have yet to be determined. Previous diagnostic criteria for adults included specific tender points; however, in 2010 The American College of Rheumatology revised the criteria, which are now generalized to widespread pain with associated symptomology (41).

There are a limited number of studies on juvenile fibromyalgia and most treatment strategies stem from adult research. Treatment for fibromyalgia should focus primarily on nonpharmacologic therapies and utilizing pharmacologic therapies only if deemed appropriate by the treating practitioner(s). A multidisciplinary approach including medical management, psychotherapy with a focus on CBT, and physical/occupational therapy has proven beneficial. There is evidence that coping skills training can improve overall functioning in pediatric patients (42). The patients and their families should be educated on self-management strategies that focus on quality sleep, stress reduction, and exercise (43).

INTERDISCIPLINARY CARE IN CHILDREN

It is our belief that the treatment of children with pain is best accomplished with an interdisciplinary approach. Children and adolescents who present for evaluation of recurrent or chronic pain often exhibit functional disability just as their adult counterparts. In children this manifests by school avoidance, and decreased school attendance in addition to decreased physical activity and social interaction. Just as in adults, children too are prone to alteration of sleep patterns and appetite changes. While children can have specific diagnoses that cause pain, in many children there is often a lack of a discernible physical cause. Parents along with their children may have seen several providers by the time they arrive at a pediatric center. Often, at these major centers the focus becomes on treating pain in the setting of family, social, and school environments (35).

Various models exist for interdisciplinary treatment of pain in children from strictly intermittent outpatient therapy, inpatient therapy, or intensive outpatient rehabilitation programs. Components of an interdisciplinary team often include PT, occupational therapy, medication management, psychology, psychiatry, and sometimes alternative therapies such as acupuncture. CBT is a cornerstone in treatment with a focus on teaching children how to self-regulate their pain by various techniques such as progressive muscle relaxation, cognitive restructuring, problem solving, and biofeedback (44). These all serve to improve function as well as decrease the fear associated with pain, giving children a sense of control.

Use of interdisciplinary programs not only leads to decreased pain scores in children but also leads to improved school attendance (45–47). Other benefits include less medication use, reports of better social and emotional functioning, decreased catastrophizing, and decreased health care utilization (44). There is also recent data to suggest that intensive inpatient therapy improves function and pain more so than intermittent outpatient therapy (47). This is not to say that this is always the ideal

treatment path, as many factors must be evaluated including severity of pain, parental support, functional disability, and school attendance. Given that these programs are limited, they should be reserved for those children with severe impairment.

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B. CONVERSION DISORDERS

The term conversion disorder was first used by Breuer and Freud to describe the transformation of unresolved psychological conflicts and unprocessed emotions into physical manifestations (1). Throughout history, other terms have also been used to describe the disorder and its presentations, including hysteria, hysterical, functional, psychogenic, pseudo-, medically unexplained, and psychosomatic. The *Diagnostic and Statistical Manual of Mental Disorders* fourth edition (*DSM-IV*) classifies conversion disorder as a somatoform disorder, whereas the *Diagnostic and Statistical Manual of Mental Disorders* fifth edition (*DSM-5*) classifies it in the somatic symptom and related disorders. It is defined as a condition in which symptoms and deficits in voluntary motor or sensory function suggest a neurologic or other physical condition which is not present (2,3). *DSM-5* has also proposed functional neurological symptom disorder as a more accurate name for the disorder (3).

It is differentiated from factitious disorder in which there is deliberate production of symptoms for psychological gain and from malingering in which the symptoms are deliberately produced for external or material gain (4).The diagnosis should not be a diagnosis of exclusion as this often leads to excessive medical testing and extends the time to appropriate intervention. For example, in one study, 22% of children diagnosed with conversion disorder underwent unnecessary surgery (5).

PATHOPHYSIOLOGY

There are a number of theories regarding the pathophysiology and some recent work in imaging which might indicate a neurobiologic cause for conversion disorder. The psychodynamic theory suggests that the symptom has a symbolic value for the underlying psychological conflict. This conflict is suppressed out of awareness with the focus instead on the presenting symptom. Alternatively, the learning theory states that the symptom is a maladaptive learned response to stress (6). From a neuroanatomic perspective, Paus et al. screened normal adults with positron emission tomography (PET) scanning and noted that the anterior cingulate gyrus plays an important role in the suppression of inappropriate motor responses (7). In a study of adults with conversion disorder and in a separate study of adults with hypnotic-induced paralysis using PET scanning, abnormal blood flow was noted in two distinct areas of the prefrontal cortex; the right orbitofrontal and the anterior cingulate gyrus (8,9). Vuilleumeir et al. (10) found decreased cerebral blood flow in the contralateral thalamus and basal ganglia of seven individuals with sensorimotor conversion disorder. However, it is unknown whether these abnormalities in blood flow reflected the cause of the paralysis.

EPIDEMIOLOGY

Kozlowska et al. estimated a prevalence of 2 to 4/100,000 children (11). However, there are wide variations in the reported prevalence of conversion disorder since children are evaluated in a variety of venues. Up to 20% of school children will complain of abdominal or limb pain and/or headache without an obvious organic cause. Most of these children are evaluated and treated by their pediatrician and do not have persistent symptomatology. They are often not included in prevalence studies for conversion disorder. The age at presentation is most commonly between 10 and 15 years of age and is rare under the age of eight. In general, females are diagnosed more frequently at a ratio of 2 to 3:1. However, a study in Taiwan noted a significant change in gender distribution as the incidence of child abuse rose in that country. The ratio of female to male children and adolescents with conversion disorder changed from 5:1 to 1:1 over the 20-year study period. This was associated with a significant increase in child abuse and reports of school bullying of males (12).

CLINICAL PRESENTATION

Children may present to their pediatrician or the hospital with symptoms such as weakness, difficulty walking, pseudoseizures, sensory abnormalities or pain. The presentation may have a cultural basis. For example, individuals in Pakistan often presented with "unresponsiveness," while those in Southern Africa demonstrated an agitated dementia (13). Table 20B.1 lists the more common symptoms.

In most cases, the symptoms are of acute onset and appear overnight. In younger children, this is often preceded by a seemingly minor injury or illness. Children who may not demonstrate concern for their impairment are called "la belle indifference." However, this is not specific for the diagnosis of conversion disorder. "La belle indifference," which is often reported in the adult population, is not as prevalent in the pediatric population and children are often very concerned about their symptoms. There are also a significant number of children who have associated symptoms of an anxiety disorder or major depression.

Risk factors for conversion disorder have been well reported in the literature and include a rigid obsessional personality trait (14), anxiety state or depression (15), and previous sexual abuse (16). Environmental risk factors include domestic stress, perceived parental rejection, poor communication within the family, unresolved grief (17), difficulty with peer relations and unhappiness at

TABLE 20B.1 COMMON PRESENTING SYMPTOMS OF CONVERSION DISORDER

MOTOR	SENSORY	OTHER
Paralysis	Numbness or anesthesia	Seizure-like activity
Weakness	Blindness	Syncope
Gait disturbance	Tunnel vision	Headache
Incoordination	Double vision	Fatigue
Tremor	Paresthesias	Coma
Loss of speech or dysarthria	Hearing loss	
Dystonic movements	Dysphagia	

school and/or academic failure (18). These children are often very intelligent, high achievers, and set very high personal expectations.

The diagnosis should be made after a detailed history and physical examination with appropriate testing as needed. Exam findings which are incongruous with neuroanatomic function support the diagnosis of conversion disorder. On physical examination, for example, normal reflexes may be noted in a flaccid limb. The gait disturbances do not correlate with a known organic disorder. Despite symptoms of balance impairments, weakness, or blindness, the individual does not collide with objects or sustain an injury. The sensory loss is not in a dermatomal or stocking-glove pattern. Diplopia does not resolve with covering one eye or is present in all directions. There may be periods of normal function or inconsistencies in function. It should also be remembered that a conversion disorder can be present with a confirmed neurologic condition; they are not mutually exclusive. Practitioners should try to avoid an extensive medical workup. However, Moene et al. noted that 11% of adult patients were later diagnosed with an organic disorder and the incidence of an incorrect diagnosis of conversion disorder has been reported in 4% to 6% of patients (19).

TREATMENT

In many cases, the physician may be able to simply give reassurance that the symptoms will resolve quickly and parents should downplay or ignore the symptoms. It is very beneficial to explain the diagnosis to parents so that they do not inadvertently reinforce the symptoms. However, giving the diagnosis to the child is often counterproductive as children feel that others think they are making up the symptoms or crazy. Instead, it is helpful to tell the child that testing reveals that the nerves and muscles are normal, but the communication to and from the brain is not working well; mind–body concept. This communication can be restored and symptoms can resolve. A referral for counseling is helpful in teaching children coping techniques for stress and if there are associated symptoms of anxiety or depression, a referral to psychiatry for further treatment is indicated (20).

When reassurance and education are not enough, there are two different treatment approaches; psychiatric intervention and physical rehabilitation. Symptoms such as pseudoseizures, syncope, and blindness, for example, are difficult to treat with physical rehabilitation and a psychiatric program is preferred. Psychiatric programs typically involve psychotherapy, hypnosis, counseling, and medication. The length of treatment is significantly longer than the physical rehabilitation approach and it may take years before symptoms improve and/or resolve. A more in-depth discussion of the psychiatric approach to treating conversion disorder is beyond the scope of this chapter and the reader is referred to the psychiatric literature.

If the conversion disorder has a physical manifestation such as a gait abnormality, for example, a structured rehabilitation program may be beneficial. However, parents must accept the diagnosis and agree to the program prior to initiation for the best results. The treatment team may include PT, occupational therapy, recreational therapy, school, psychology, psychiatry, and physiatry. Treatment is aimed at a systematic reacquisition of skills to meet the expected functional outcome; walking normally, for example. Speed demonstrated the effectiveness of a behavioral management approach (21) in a comprehensive program when used in conjunction with PT in the adult population (22). The behavioral approach uses positive reinforcement of healthy behaviors or functions and ignores those behaviors which are maladaptive. When the individual is not in therapy, he or she is instructed to rest so that the symptoms (eg, abnormal gait pattern) are not reinforced. In children, the program is kept very structured and children receive positive reinforcement only when they have achieved the individual goals set for them on their goal mountain (Figure 20B.1). Every goal mountain is individualized to sets steps to correct the physical abnormality, reinforce the mind–body concept of control, and reflect something of interest to the child.

Symptoms are largely ignored and children may not progress up the goal mountain until the goal is performed correctly on a set number of consecutive therapies. The goals should be achieved in the specified sequential order so that increased individual independence is achieved as skills are attained. Children are referred back to their goal mountain daily with praise for each achievement. The use of physical modalities such as biofeedback may be helpful in establishing the mind–body connection. In addition to PT and occupational therapy, children receive daily counseling with a psychologist to evaluate stressors and learn coping techniques. When children are not in therapy, they

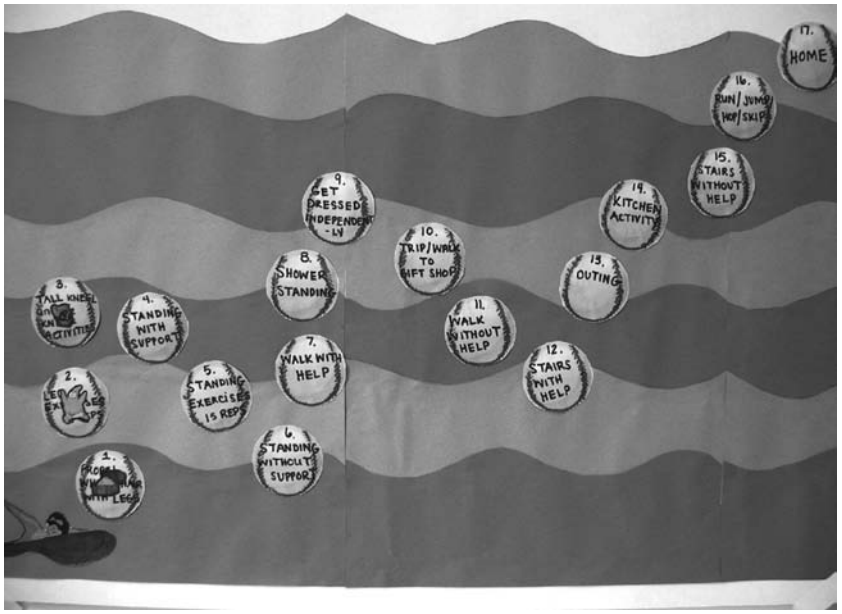

FIGURE 20B.1 This goal mountain outlines the expected functional progress and steps to attaining functional independence for a boy with interests in baseball and swimming.

are instructed to rest or complete schoolwork. Parents also participate in counseling for education regarding the disorder and how to address any regression once children return home. Oftentimes, children require ongoing counseling after discharge from the rehabilitation program.

Potential reinforcers of the sick role must also be carefully evaluated and removed at the beginning of the program. For example, gifts for being in the hospital or increased visits from friends and family are discouraged. If gifts are brought in, they should be associated with the achievement of specific goals. Cell phone privileges and access to the Internet should be curtailed as these communications may either positively reinforce the sick role or may be a stressor. Families may also be a source of stress and limitation of visitation initially may be helpful with eventual reintegration as the child progresses and learns better coping strategies.

If the child is not progressing with the behavioral approach, a double bind should be considered. Teasell and Shapiro describe this as a strategic behavioral intervention in which the child is told that lack of progress "proves" a psychological cause for their symptoms. They reported improvements in several patients with long-standing symptoms using this approach (23).

In a review of the literature, Fitzgerald et al. found limited and poor quality evidence to establish efficacy for PT in the treatment of conversion disorder in children because most studies were observational, included PT as a component of a larger treatment regimen, and did not have control groups or well-defined outcome measures (24). The commonly used modalities in these studies included contracture management, electrotherapy, biofeedback, hydrotherapy, strength training, and behavioral programs. The study indicated the need for further research to provide an evidence base for treatment, but did not demonstrate that PT was ineffective or inappropriate for the treatment of this disorder.

With early recognition, recovery is usually within a few days or weeks and 85% to 97% of children will have a full recovery (13). Table 20B.2 lists the positive and negative prognostic factors for children with conversion disorder.

TABLE 20B.2 PROGNOSTIC FACTORS FOR CHILDREN WITH CONVERSION DISORDER

FAVORABLE	UNFAVORABLE
Recent onset of symptoms	Longer duration of symptoms
Monosymptomatic manifestation	Multiple symptoms or changing symptoms
Early diagnosis	History of conduct disorder
Good premorbid personality	History of sexual abuse
Parental acceptance of the diagnosis	Anxiety or depression
	Family persists in the belief that symptoms are due to an organic cause
	Recurrence of symptoms after successful treatment

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21

AGING WITH PEDIATRIC ONSET DISABILITY AND DISEASES

Margaret A. Turk, Jan Willem Gorter, and Lynne Romeiser Logan

Aging is a fact of life, and although many may not be well prepared for typical aging changes, current wisdom suggests this is not an unexpected event. However, aging with a disability can be an overwhelming and alarming situation, especially for those experiencing changes in function or health at an earlier-than-expected time. These changes can also mean the difference between living alone with minimal to no support and requiring a more restrictive living environment, including a move to an institutional setting, at a young age. It is essential to understand the developmental course of disability over the life span, and to provide comprehensive health care that focuses on primary and secondary prevention, as well as medical treatment and psychosocial care for aging people with pediatric-onset disability and conditions.

For years, children with disability and their families have been told that health and functional status, mobility, and musculoskeletal problems essentially stabilize by early adulthood. Recent longitudinal studies have noted that trajectories of functional status differ by gross motor function and intellectual ability in children with cerebral palsy (CP) (1–4). And as more people with lifelong mobility and other impairments live through their adult years, it is apparent that mobility, functional status, and musculoskeletal changes commonly continue into adulthood (5–7). In fact, questions and concerns about mobility, function change, and pain are common among the majority of adults with mobility impairments caused by any etiology (8).

Despite the personal accounts and experiences of those with disability, their families, and many clinicians, there are no longitudinal studies on disability crossing the life span and few surveys or statistics that can document these aging changes and risk factors for them. Present statistics estimate that the number of Americans of all ages with disability (broadly defined by impairment, functional limitation, or participation restriction) exceeds 40 million, and may be closer to 50 million (8). However, these are estimates using multiple national surveys, cross-analyzed in an attempt to cover all ages and living situations. Many of these surveys exclude those living in institutions or assisted living programs (where a number of adults with congenital or childhood-onset conditions may live), and many exclude young children or adults younger than retirement age. There are no U.S. national surveillance programs that monitor the trajectory of aging with a disability by specific disability condition, by severity, or by age of onset over a lifetime, although there are international registries that are beginning to provide information about adolescents and young adults. Data do identify that more infants, children, and young adults are surviving with conditions that were at one time fatal such as CP (9–11), spina bifida (SB) (12,13), muscular dystrophy (14), and childhood-onset epilepsy (15). Moreover children and young adults are completing and surviving long-term risk treatments (eg, chemotherapy, radiation, surgery). In the United States, approximately 500,000 children and youth with special health care needs turn 18 years annually (16). Thus, there is an increasing population of adults with disability, with accompanying risks for long-term complications and additional conditions. There have been changes in a few health conditions in childhood that contribute to adults with disability statistics. The incidence of SB dropped from 24.9 to 18.9 per 1,000 live births with the use of folic acid supplements in women of childbearing age (17). Lead exposure, a risk factor for neurodevelopmental problems, has dropped significantly, with reported lead levels now below 2% (18). Autism spectrum disorder (ASD) and attention deficit hyperactivity disorder (ADHD) have both increased in prevalence, likely related to service availability and classification, better definition of diagnosis and early recognition, and increased prevalence of prenatal risk factors (19). Advances in medical care and public health practices will continue to change the face of the types of disability seen in adults with early-onset conditions in the future.

Table 21.1 identifies the leading chronic health conditions as causes of activity limitations, using the 2008–09 National Health Interview Survey (20). As is noted, the listed chronic conditions do not necessarily list disability

TABLE 21.1 DECREASING PREVALENCE OF CHRONIC PHYSICAL HEALTH CONDITIONS ASSOCIATED WITH LIMITATION IN ACTIVITIES: U.S. CHILDREN UNDER AGE 18, 2008–09 (NATIONAL HEALTH INTERVIEW SURVEY)

CHRONIC CONDITION*	NUMBER OF CASES PER 100,000 CHILDREN (SE)
Speech problems	1,815 (87.5)
Learning disability	1,775 (86.8)
Attention deficit hyperactivity disorder	1,715 (74.7)
Other mental, emotional, or behavioral problems	1,452 (75.9)
Other developmental problems	779 (57.1)
Asthma/breathing problem	632 (48.4)
Other impairment/problems	431 (36.5)
Birth defect	423 (35.7)
Bone/joint/muscle problem	260 (31.0)
Hearing problem	256 (29.9)
Vision problem	244 (27.1)
Intellectual disability	207 (25.9)
Epilepsy/seizures	173 (24.6)
Injuries	76 (16.4)

* Categories are not mutually exclusive—more than one condition could be reported as contributing to the child's activity limitation.
Abbreviation: SE, standard error.
Source: Modified from Ref. (20). Halfon N, Houtrow A, Larson K, Newacheck PW. The changing landscape of disability in childhood. *Future Child.* 2012;22(1):13–42.

types typically identified in medical rehabilitation systems as identified by diagnosis, International Classification of Diseases (ICD) codes, or diagnostic-related groups, but rather by more generalized conditions. The pediatric chronic conditions listed are largely cognitive and mental-health-based. The only estimate of adults with early-onset disabilities is by Verbrugge and Yang (21) using data from the 1994 National Health Interview Survey Disability Supplement, Phase 1, suggesting that 7% to 9% of adults reporting a disability had onset before the age of 20 years. The surveillance data available imply that the prevalence of disability diagnoses typical of rehabilitation program settings is small compared to other conditions, and usually not the primary focus of public health, surveillance, and policy programs.

Most of the health and function information regarding aging in congenital and childhood-onset disability that is known is derived from existing databases developed for service or financial reasons, case studies and series, limited survey information, cross-sectional studies, opinion pieces, and the like. Much of the conventional wisdom in this area has been communicated through the network of persons with disabilities and, more recently, through books and texts. There is minimal systematic information regarding the impact of commonly practiced interventions over a lifetime, including environmental approaches to barriers. Health care providers receive minimal education regarding disability and/or aging with a disability during undergraduate and graduate education (22,23). Therefore, health care providers and consumers have limited knowledge from which to base decisions regarding adult health issues and anticipated changes in function. However, individuals with disability do require ongoing medical, dental, and specialized services to maintain health, decrease morbidity, and improve quality of life (QOL) (24).

This chapter will provide a conceptual framework regarding aging as it relates to congenital and childhood-onset disability, review general issues of health and function across early-onset disability, discuss lifelong functional status and health issues of adults with specific early-onset disability, and consider the issues surrounding health care access and transitioning from pediatric to adult care services. Since the last edition, there has been a significant increase in research about adults with CP, SB, and intellectual disability (ID). Additionally, there has been an increasing population of adult survivors of childhood-onset cancers, especially brain tumors, who are seeking care from physiatrists. These advances have been included along with research about participation and QOL as it relates to health and adults with pediatric-onset disability.

LIFE-SPAN PERSPECTIVE

Improved medical care, increased life expectancy, and better services for lifelong care and support in society have provoked an interest and need for long-term future planning. This includes transitions in care from typical nurturing pediatric care systems to more traditional adult self-directed services. Although the notion of transition of youth with chronic conditions and disabilities is not new, the complexity of transitions, the possible negative outcomes in health, and the need for understanding aging with disability are now being recognized (25). Retrospective reviews and anecdotal experiences also question some long-held beliefs of "use it or lose it" to one of "conserve it to preserve it (26)." Choice of health care providers for adults with early-onset disability and special health care needs is, unfortunately, often limited by insurance and expertise.

Clinicians with an understanding of the natural history of disabling conditions can be helpful in monitoring, keeping vigilance for, and prevention of some general health conditions and aging or secondary conditions seen in disability. This public health model of prevention also includes tertiary prevention with the use of environmental modifications and technologies and removal of barriers to participation. There are general aging, associated conditions, secondary conditions, and health concepts that are helpful in understanding a life-span perspective.

Aging is a developmental process. It begins at birth and continues to death. Typically, however, children and adolescents are said to develop, whereas adults, especially adults over 50 or 60, are said to age. During the early stages of aging (infancy, childhood, adolescence), attainment of skills and capabilities is on the rise; in the middle stages (adulthood), maintaining and retaining function is the focus. Over a normal life span, natural physiologic declines are not truly preventable, although they may be accelerated or slowed by a variety of individual genetic factors, personal behaviors (eg, diet and exercise), health care practices, and environmental conditions. Aging changes in motor performance seem to be accelerated in some adults with early-onset disability, with earlier-than-typical manifestation of slowed or decreased motor performance and pain complaints. Persons with disability follow a course of aging, although likely with a slower and lower attainment of skills and a smaller capacity to adjust to acute or intercurrent health or medical and surgical intercedents (Figure 21.1). This is a conceptualization, so it is important to recognize that not all disability types respond in the same way to aging. Nevertheless, the emphasis here is on *aging with disability*, not aging into disability (27).

There is also a need to appreciate the different time dimensions at play (8). These include the age of onset of disability in relation to developmental maturity (congenital onset versus acquired onset), the number of years spent with a disability (hemiparesis onset noted at birth versus onset at age 17 years), cumulative effects of medications or treatments (long-term steroid use), and era of disability onset (CP onset in 1950s versus 1990s versus 21st century), including different treatments, opportunities, and attitudes. Anticipated aging changes and treatment strategies will be modified by these temporal concepts.

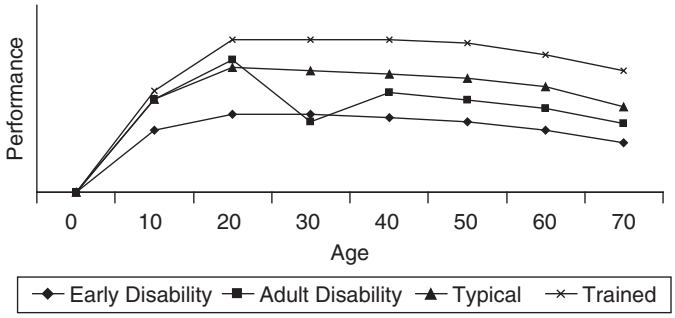

FIGURE 21.1 Conceptual model of aging and performance. Performance is a conceptual quotient of multiple skills. The trained person will achieve a higher level of performance than typical and, assuming ongoing exercise, will have a slower decline with age. With the onset of disability in adult years, there is more loss of skill than improvement, but often not achieving the previous typical level. Those with early-onset disabilities do not achieve full "performance" and are slower to achieve the maximum level.

Secondary conditions are defined as "any additional physical or mental health condition that occurs *as a result* of having a primary disabling condition (8,28)." The initial concept and intended use (29) distinguishes secondary health conditions from the social and economic consequences that may follow a primary disabling condition (societal limitations and barriers—for example, poverty with disability, social isolation, limited transportation). There are key common features of secondary conditions (28):

- Causal relationship to the primary disability—the primary disability is a risk factor for the secondary condition
- Preventable or modifiable conditions
- Variability in expression and timing of manifestation
- Capability to increase the severity of the primary condition
- Potential to become the primary health concern

Many secondary conditions are linked across several primary disabling conditions through common physiologic processes or functional characteristics. As an example, disabilities with sensation changes and immobility are risk factors for pressure ulcers, such as spinal cord injury (SCI), SB, multiple sclerosis, and severe brain injury. Three common secondary conditions noted through cross-disability studies are fatigue, chronic pain, and depression (30,31).

Secondary conditions are distinct from associated conditions or residual deficits and comorbidities. Associated conditions describe elements that result from the defect, injury, disease, or pathology, although the expression may be variable. These conditions are the residual from the original pathology, and are often present at the time of diagnosis of the primary disability, although by development or evolution may not be expressed or expressed fully at initial diagnosis. For CP or other brain injuries, the list of associated conditions includes seizures, spasticity, learning disabilities, ID, sensory problems, and oral motor and communication problems. These conditions may not be present for all people with the specific disability, are fairly well known to require monitoring by clinicians, and their presence is confirmed through typical timely evaluation. Persons with a primary disabling condition may have any combination of associated conditions, all of which will affect their ultimate functional capabilities. Comorbidities are other medical conditions unrelated to the primary disabling condition, and not a feature of the primary disability. As an example, persons with CP may also develop congestive heart failure or colon cancer should they have the risk factors or genetic predisposition for these conditions. As research continues, especially through longitudinal studies, links may be identified between primary disabilities and specific health conditions.

The health of aging people with childhood-onset conditions can be seen as a dynamic balance between opportunities and limitations, shifting through life (32). Health perception is an individual determination, and is affected by personal expectations, experiences, sense of vulnerability, support, and locale. How people with disability self-rate their health has been questioned (33). This self-concept may also direct consideration of engagement in typical health and wellness activities. Often, the health of persons with disability is perceived as poor by clinicians and providers when individuals report a positive perception of their own health. This health provider concept may limit the offer of screening or health promotion opportunities. Perception of health in adults with disability may be related to time of onset; it is reported that adults with early-onset disability may identify better health than those with adult-onset disability (34,35). Research further suggests that adults with disability likely have a different construct of and self-rating process for health (36). Therefore, there has been growing interest in concepts associated with QOL.

QOL is a multidimensional construct reflecting subjective perceptions of the individual's position in life in the context of the culture and value systems in which he or she lives and in relation to the individual's goals, expectations, and concerns (37). QOL refers to the notion of holistic well-being, whereas health-related quality of life (HRQOL) specifically focuses on health-related aspects of well-being (37). HRQOL includes elements about physical and mental functioning, as well as the person's appraisal of their effect on daily life and social functioning. For individuals with disability, HRQOL may be restricted. In general, persons with nonprogressive disabilities should be considered healthy, with a shift of the health care model from an illness and disability paradigm to one of wellness and prevention or early identification of secondary conditions, aging issues, and/or comorbidities.

GENERAL KNOWLEDGE REGARDING HEALTH AND PERFORMANCE

A body of literature has accumulated regarding health, aging, and secondary conditions for adults with disabilities and for some specific disabilities of early onset. Most research has focused on disabilities and impairments that have relative higher prevalence rates (eg, CP, ASD, speech–language disorders); are easy to associate with a disability group (eg, SB, Down syndrome [DS]); benefit from organized, dedicated service programs (eg, muscle disorders); and therefore have attracted research funding to generate a significant body of knowledge about the condition. The literature includes a combination of scientifically observed and anecdotal information as the database, often involving a "convenience" sample and a cross-sectional approach. Conclusions are drawn from patient reports, clinical observations, and ICD codes. More recently, standardized surveys or severity measures are used, and these provide a glimpse into individual characteristics or outcomes. Most studies identify health issues or concerns, with few challenging prevention or intervention strategies. Each factor in the interaction of disability and aging or secondary conditions has the capability to become a "negative feedback loop" (38) that may lead to further disability or a new health condition. There are studies using cross-disability groups that may have a higher representation of certain disability groups or may be small sample sizes, and consequently generalization to other disability groups should be considered with caution. In a similar manner, prevalence rates for some aging, secondary, and health conditions in disability-specific studies cannot be applied to all disability groups.

Pain is a common health condition for adults with disability, as noted earlier, and may be seen earlier in early-onset disability groups, especially those with mobility impairments. Pain is also a common complaint in adults without disability, and there is an expected response from health care providers, including intensity of evaluation and treatment. This should also be the expectation for those with disability, especially at younger ages. Any significant decrease or loss of motor skill, change in continence, change in typical activities, direct pain complaint, or "sluggishness" (39) requires further evaluation. Common musculoskeletal etiologies include poor ergonomics and biomechanics in tasks (secondary to

deformity or limited motor control), underlying weakness and therefore overuse, hypertonia depending on the primary disability, and degenerative joint disease. Neurologic etiologies may also need to be considered, including general neuropathies, focal neuropathies (eg, carpal tunnel syndrome, ulnar entrapments), radiculopathies, and myelopathy or stenosis. Appropriate evaluation should be completed to determine the treatment strategy. Typical treatment strategies should be implemented and modified as needed, given the disability and improvement noted. Management may include traditional noninvasive interventions (eg, analgesics, nonsteroidal anti-inflammatory drugs [NSAIDs], therapy modalities, massage), more aggressive pain management strategies (eg, manual medicine, trigger point injection, spinal injections), and re-evaluation of functional activities or positioning that may predispose to the pain complaints. For spasticity-related problems, use of tone management techniques can be helpful, including oral antispasticity medications, use of botulinum toxin injections for focal problems, or intrathecal baclofen. Surgical interventions should also be considered, and will require preplanning for rehabilitation, living arrangements, supports postprocedure, and above all good outcome evaluation.

There are anticipated health and performance changes with aging. The risk for additional health problems should be monitored and addressed as with the general population. However, people with disability are often not afforded typical screening as in the general population. Iezzoni et al reported those with mobility impairments did receive pneumonia and flu vaccines, but were less likely to receive other preventive services. Women with severe mobility impairments in particular were less likely to receive Pap smears and mammography screenings (40). Cryptorchidism in males may be missed if examination is dependent on standing for examination. Women with disabilities had less knowledge about cardiovascular risks and no screening for risk factors, despite their higher risk with low activity levels (41). However, Cooper described a minor modification in office-screening techniques for adults with intellectual disability (ID) that improved the identification of risk factors and health needs, with improved health determinants (42). Lennox has shown that a Comprehensive Health Assessment Program (CHAP) resulted in increased health promotion, disease prevention, and case-finding in both community living and supported living adults with ID (43). Additional health risks and conditions can affect general performance.

Performance changes with aging include decrease in strength, balance, flexibility, coordination, and cardiopulmonary function, to name a few. The impact of these known aging changes on a person with mobility impairment is not well understood. Use of equipment, modifications to environment or activities, and joint protection all contribute to maintaining function over time. It is, however, known that persons with mobility impairments use more energy to perform mobility activities than their nondisabled peers. Therefore, exercise and activity to improve performance and maintain those improvements would seem intuitively obvious. In fact, there is scientific evidence that exercise and activities are effective for people with mobility impairments and that these activities can be managed through home programs and health clubs or aquatic activities, not just traditional physical therapy programs (44,45). Simply continuing typical activities, even though considered "strenuous," will not increase strength, conditioning, or performance. Exercise and activities should be a part of a health maintenance program for adults with mobility impairments.

Participation (eg, social engagement, life role, employment) is one of the constituent elements of health and is increasingly considered a fundamental rehabilitation and health promotion outcome for child-onset disability. This is an important aspect of health and performance that should be queried when evaluating adults with pediatric-onset disability. We now have increasing generations of adults with childhood-onset disability who have the same aspirations and rights to participate fully in society. The literature on the unique needs and experiences of people with childhood-onset disability has taught us that we need to focus on physical, social, cognitive, and emotional aspects of health, with participation as one of the ultimate measures of outcome (ie, involvement in a life situation) (46). There is strong evidence that participation contributes to children's physical, mental, and social well-being and to their QOL (47), and there is no reason to assume that this will be different when these children develop and live an adult life. Youth with physical disability, speech and language delay, or developmental disorders are at risk for lower community participation and social isolation, since they do not experience the widening social world of other teens (48). Tan et al (3) found that younger age, severe physical disability, and ID are important determinants of social participation in individuals with CP. Positive social experiences from early in childhood and continuing through adulthood prepare and support individuals with pediatric-onset disability to achieve positive sexual health and adult sexual roles (49). It is through these opportunities that people with pediatric-onset disability will develop friendships and romantic relationships. The results of the National Longitudinal Transition Survey Wave 2 showed that engagement in work or time with friends was most common for youth with learning disabilities or speech, visual, or other health impairments compared to other disabilities; more than three-fourths of youth in these categories were engaged in some type of employment including part-time jobs and unpaid volunteer work (50). In contrast, youth with ID (52%), multiple disabilities (54%), ASD (56%), and orthopedic impairment (59%) had the lowest rates of engagement.

DISABILITY-SPECIFIC HEALTH

There is increasing information about specific early-onset disability conditions and adults' health and expectations for functional performance with aging. This chapter will highlight those conditions commonly managed by pediatric physiatrists, or those for which we have useful information. There is actually considerable information available for clinicians; however, it is important to recognize the quality of the study or report (eg, case series, a convenience cohort with single-point assessments, or registry data with standardized intake and follow-up). Nonetheless, it does begin to provide a picture of the health of adults with childhood-onset conditions and the need for modifications to our health monitoring and interventions for some conditions. Table 21.2 identifies the more common health conditions and management strategies for the disabilities described in this chapter.

CEREBRAL PALSY

CP is the most common condition that pediatric physiatrists manage, although it is not the most common reason for childhood disability, as noted earlier. Recent estimates from the United Cerebral Palsy Association indicate that there are about 750,000 or more people in the United States with CP. Over the past 10 years, there has been increasing information available about the life course in CP, and adult issues and health are better defined.

The health of adults with CP is generally good. Although CP may affect multiple organ systems, in general, long-term health problems are related to physical impairments such as pain, fatigue, and the musculoskeletal system as well as mental health issues such as depression and anxiety (see Table 21.2).

Mortality

Mortality for people with CP appears to be related to severity of impairments. This is very clear in the pediatric population, but less so for adults who have survived into their late twenties and thirties. In a recent study in Sweden, Westbom et al showed that virtually all children with good motor abilities, and 96% of the whole population of children with CP, survived into adulthood. Although the risk of death is the highest in fragile children with CP, their estimated survival is 60% at 19 years of age (11). There is also an obvious cohort bias when comparing mortality data from those born prior to the 1980s to mortality data of a younger adult population, and it is not clear that the information about the adults of today may be used specifically for predicting life expectancies.

Through a large database in California defined by financial and service support needed and especially representative of the more severely impaired individuals with CP, survival of higher-functioning adults was close to that of the general population (51). Strauss et al also reported with this same database that older subjects who had lost the ability to walk by age 60 years had poorer survival and that those who had the most severe disabilities rarely survived to age 60 years (52). This group has found a trend toward improved survival continuing over the past 3 decades, highlighting improved care for the most fragile individuals with high levels of impairment, including use of gastrostomy tubes (53,54). The mortality ratio for most adolescents and adults with CP, relative to the general population, has increased at 1.7% (95% CIs [1.3, 2.0]) per year during the recent study period (1983–2010), while a decline in childhood mortality was noted. Life expectancies for adolescents and adults are lower for those with more severe limitations in motor function and feeding skills, and decreases with advancing age. Life expectancies for tube-fed adolescents and adults have increased by 1 to 3 years, depending on age and pattern of disability, over the past 20 years (55).

Reports from countries other than the United States also identify life expectancies for adults with CP to be close to the general population for those with mild to moderate impairments. The Western Australia Cerebral Palsy Registry noted the strongest single predictor of mortality was ID (56). This study noted motor impairment severity increased the risk of early mortality, with mortality declining after age 5 to 15 years, and remaining steady at 0.35% for the next 20 years. Providing insights on era of disability onset, Hemming et al reported on adults with CP in the 1940 to 1950 birth cohort in the United Kingdom. Assuming survival to age 20 years, almost 85% survived to age 50 years compared to 96% of the general population (57). Again comparing to the general population, many of the deaths noted in the age range 20s to 30s were respiratory, and deaths in 40s to 50s were as a result of circulatory conditions and neoplasms. Few deaths in adulthood were directly attributed to CP, although the nervous system was implicated more than in the general population. The notion of increased neoplasm as the cause for death rates is echoed by the large California database noting a three-times-higher rate for breast cancer in CP than in the general population, and this may be related to severity as well as poor screening (58). Survival rates for children of today may not necessarily be extrapolated from any of these studies.

Health and Quality of Life

The general health of adults with CP is self-reported as good or satisfactory to excellent (59,60), and this can be comparable to that of the community at large (34). In a population-based study of adults with CP in a mid-sized

TABLE 21.2 AGING HEALTH AND PERFORMANCE CHANGES

COMMON RELATED HEALTH CONDITIONS	PREVENTION STRATEGIES	TREATMENT STRATEGIES
Pain Fatigue	Routine exercise Monitor and query routinely Work simplification Ergonomic evaluations Energy conservation	Exercise prescription Query/evaluate sleep; manage as needed Evaluate for pain etiology and treat Modify equipment or workplace Evaluate mental health and manage Progress to pain management program
Musculoskeletal Contractures Hip pathology Knee pathology Foot or ankle pain Back pain	Monitor and query routinely Joint protection strategies Routine exercise Biomechanic and ergonomic assessments	Focal musculoskeletal evaluation Tone management Modify equipment, workplace, biomechanics of function Therapy prescription Adjust orthoses and wheelchair
Bone health Osteoporosis Fractures	Routine exercise, especially weight-bearing Calcium/vitamin D supplement Fracture and fall prevention; education	DXA evaluation Consider treatment when multiple fractures Exercise when appropriate
Neurologic Spasticity Seizures Spinal stenosis Nerve entrapments	Routine monitoring Adjust medications with reported change Query for changes—high index of suspicion for pathology	Tone management—medications, botulinum toxin injections, intrathecal baclofen Seizure management Radiological evaluation Electrodiagnosis Surgical referral when appropriate
Genito/urinary conditions Incontinence UTIs	Monitor and query routinely Routine gynecologic follow-up for women and follow-up for men	Urodynamic evaluation Scans/radiographs Medications and CIC when needed Urology referral as appropriate
Cardiovascular health	Monitor blood pressure and typical serum panels Query for risk factors	Treat cardiovascular symptoms and events
Obesity/overweight	Healthy nutrition and weight management Measurement of body fat—consider waist circumference, DXA, or BIA Monitor for metabolic syndrome symptoms/signs	Manage weight; promote exercise
Respiratory conditions Infection Sleep apnea	Routine monitoring Immunization Query sleep hygiene	Scoliosis evaluation Sleep study and management Specialty referral as needed
Gastrointestinal Constipation GERD Obstruction	Monitor and query routinely—recognition of severity Nutritional management	Adjustment to bowel program regimen Specialty referral when appropriate
Oral motor problems	Dental monitoring, preventive care	Dental treatment Drooling management, including botulinum toxin injections and possible ENT referral
Deconditioning Falls	Query about changes in function Routine exercise Education and prevention	Therapy prescription—focus on strength and aerobics Reconsideration of equipment
Mental health	Routine monitoring, especially for depressive or anxiety symptoms Query support, living arrangements	Specialty referral as appropriate Referral for psychological and social support Use of community resources

(*continued*)

TABLE 21.2 AGING HEALTH AND PERFORMANCE CHANGES (*CONTINUED*)

COMMON RELATED HEALTH CONDITIONS	PREVENTION STRATEGIES	TREATMENT STRATEGIES
Sexual functioning Fertility/reproduction Interference spasticity or pain Emotional/body image	Engage in discussion re: sexuality Provide education about sexuality and function—appropriate modality for cognition and function Assist with environmental modification for routine assessments as able Ensure pregnancy high-risk needs are met	Following pregnancy, support may be needed in the home
Health maintenance	Monitoring—see Table 21.9	

Abbreviations: BIA, bioelectric impedance analysis; CIC, clean intermittent catheterization; DXA, dual energy x-ray absorptiometry; ENT, Otolaryngologist; GERD, gastroesophageal reflux disease; UTI, urinary tract infection.

U.S. metropolitan area, persons with CP were generally healthy (based on clinical information and self-report), but noted worries and concerns about their health status and future (61). Self-perceived health ratings and life satisfaction may be related to the presence of pain or functional changes over time, but not to the severity of impairment (62–64). A cross-sectional study of youth and young adults with CP in Canada using standardized measures noted youth were somewhat more positive about their health than young adults, although QOL scores were similar. Severity of CP was a strong predictor of health and QOL. Similarities between the groups were notable suggesting self-reported health and QOL outcomes may remain relatively stable across the transition to adulthood (65). HRQOL also remains fairly stable over time for people with CP. As individuals with CP grow and mature, many changes take place in their psychosocial development, which accordingly changes their expectations and those of their caregivers, peers, and professionals. The functional effect of CP seems particularly predictive of physical HRQOL, whereas the associated ID may affect their HRQOL in social functioning (2).

Health outcomes are also evaluated by the use of medical services. Despite reports of good health, a Canadian publication notes adults with CP visited outpatient physicians 1.9 times more than age-matched peers. Annual hospitalization admission rates were 10.6 times higher for adults with CP compared to their peers (66). The presence of other medical conditions is associated with increased odds of hospital or emergency department (ED) use (67). Analysis of the Canadian Institute for Health Information notes epilepsy and pneumonia are the top two reasons for hospital admissions for youth and young adults, and for young adults only, mental illness is the third most common admission diagnosis (68). Additional adult diagnoses included lower gastrointestinal (GI) problems or constipation, malnutrition or dehydration, upper GI problems, and two unique problems seen in the adult group: fractures and urinary tract infections (UTIs). These represent conditions for surveillance in an adult population of people with CP.

Functional Status and Performance

The functional status of adults with CP is not static over time, and with aging there can be modest decreasing function, as there is for the general population. A number of studies, both in the United States and abroad, with small to large convenient samples, have noted that about a third of subjects report modest to significant decreases in walking or self-care tasks (34,52,69–71). Changes in dressing and walking with relative sparing of other self-care or social activities were reported in two of these studies (34,52). More recent studies provide data by Gross Motor Function Classification System (GMFCS), which allows better definition. Day et al used the large California database to determine the probabilities of loss or gain of walking skills into adulthood for those with CP (72). They noted that by age 25, there would unlikely be any improvement in walking skill and most would not change over the next 15 years, although there could be some decline. Therefore, the reason for even modest decreasing skill is not clear and may be related to progressive neurologic problems (eg, cervical spine stenosis, radiculopathy), lack of environmental modifications, pain, no access to or participation in exercise or activity programs, aging, or other medical conditions.

Decreased independence (increased need for assistance) in mobility and self-care is a common complaint of adults with mobility impairments. The reasons for change are varied, and may include those related to age changes (eg, decreased endurance, flexibility, strength, or balance), progressive pathology or secondary conditions (eg, pain, contractures, spasticity, osteoporosis and fractures, stenosis), or personal choices (eg, use of powered

mobility to conserve energy). The change in mobility is often a response to a secondary condition or age-related change. Falls may also be such a response. Significant change in mobility or falls should not automatically be accepted as a part of a congenital or childhood-onset disabling condition in adult years; treatable etiologies should be sought.

It has been suggested through cross-sectional and convenience samples that adults with congenital or childhood-onset disability may show musculoskeletal, mental health, or performance changes typical of advanced aging earlier than their nondisabled peers (51,59,73) (7,74,75). These observations require confirmation through longitudinal controlled studies. While risk factors may predispose a person to these changes, they are, as yet, unproven. If these earlier-than-expected aging changes are confirmed, they should be considered secondary conditions.

Pain and Fatigue

Pain is the most consistent health condition reported by adults with CP (35,59,74,76,77). It has been reported in a number of samples of adults with CP at a variety of ages to be 30% to 80%, with activity limitation from this at greater than 50%. For this reason, it will be covered as a separate topic. Communication challenges and multiple pain etiologies complicate diagnosis and treatment. Pain in CP may be present for a variety of reasons; it may be acute, recurrent, or chronic (78). Increased spasticity, weakness, falls, or progression of contractures or deformities can result from pain, particularly when pain is not reported because of communication difficulties or severe ID. Because of the high prevalence, the health care provider should try to elicit complaints or indications of pain, and evaluation, diagnosis, and intervention should ensue (78). Pain is often the reason for a change in function, living arrangement, or social interaction.

Pain is usually identified by proximity to a joint, and less often a limb. Most people report "arthritis" as the etiology of these pain complaints; however, these pains may originate from either joints or muscles. A good history and clinical exam will help sort out the issues and direct appropriate treatment. Back, leg, and hip pain complaints are common in persons with CP (79,80). There are usually more pain complaints in those with spasticity (79). It has been reported that fatigue often incites pain, and exercise most commonly relieves pain (79,81).

Fatigue is a common complaint of adults with CP, and is associated with pain (74,82). It is also associated with deterioration of skills, depression, and low life satisfaction, with no association with any specific type or severity of CP. As noted, it may incite pain. The fatigue may also be associated with the reported coping strategies sometimes used for chronic pain by adults with CP (83). Sleep disruption should also be questioned since it is commonly seen with pain and fatigue. Anecdotally, the pain/fatigue complex appears to respond positively to directed pain management, good sleep hygiene, medications, and exercise.

Appropriate management includes early identification of the problem and its source. Common musculoskeletal etiologies include poor ergonomics and biomechanics in tasks (secondary to deformity or limited motor control (71)), underlying weakness and therefore overuse (84), hypertonia (85), and degenerative joint disease (86). Typical management strategies should be offered, and referral for additional interventional, orthopedic, or neurosurgical consultation should be considered. However, adults with CP tend to self-manage their pain complaints (87), and for those who seek medical care, the report is minimal improvement and few options offered (88).

Musculoskeletal and Neurologic Conditions

CONTRACTURES. Contractures are a common secondary condition and may develop in childhood/adolescence and progress over the life span. Many individuals may have had interventions, including multiple surgeries in their life. Their impact on functional status or general health care needs is variable. Increasing contractures, particularly when associated with pain or increased spasticity, may be an indication of progressing pathology. Aging changes include decreased flexibility, and the clinician must distinguish pathologic causes of increasing contracture through appropriate diagnosis.

OSTEOARTHRITIS. Because of the significant pain complaints that adults with CP report, it is often stated that there is an early onset of osteoarthritis. Conceptually, this has been explained by unusual and possibly increased forces on joints that may be malaligned and/or have deformity; these forces are associated with underlying weakness and poor selective motor control (59). In fact, health care providers often will make a presumed diagnosis of "arthritis" for pain complaints in adults with disability. Clinically, it is not surprising to find significant arthritic changes with radiographs of painful joints, and sometimes at young adult ages. However, the presence of early-onset arthritic changes has been documented by case reports, and studies that report arthritis among subjects base this information on self-report of arthritis or presence of pain. Often, the pain complaint is not evaluated fully, and may have an etiology in soft tissue injuries or problems and not degenerative changes within the joint. There may, in fact, be premature osteoarthritis, but it has not been documented definitively. Of importance is the recognition of pain, appropriate evaluation, and treatment.

HIP PATHOLOGY. Degenerative changes have been noted radiographically in dislocated and subluxed hips, not always related to weight-bearing activities, in persons with CP (76,89,90). Use of tone reduction strategies may be helpful. Femoral head resection as a treatment strategy for control of pain in hip disease for persons with CP has been suggested; however, pain often persists or recurs postoperatively (91–93,94). Total hip and knee replacements as a treatment option for pain from severe arthritis in adults with CP are becoming more common; however, as their lifelong efficacy remains unknown (95,96), revision may be anticipated with placement at younger ages.

KNEE PATHOLOGY. Knee contractures are common in those who do not walk and in those who walk with obvious knee flexion and crouch. Not all knee contractures are painful. Tone management may improve range, function, and pain. Patella alta may develop from adolescence onwards over time, and pain or chondromalacia may result. Joint laxity may also be present. Modalities, exercise, kinesiotaping, and other interventions may be helpful. There are advocates for patellar tendon advancement surgeries, with or without distal femoral extension osteotomies, in adolescents and young adults to improve pain and restore knee function in gait, confirmed on gait analysis (97).

FOOT OR ANKLE PAIN. Again from biomechanical factors, contractures and pain may develop. Typical interventions may assist including orthoses, but not all bracing or shoe inserts are helpful, and biomechanics must be taken into account. Plantar fasciitis with appropriate treatment should be considered.

SPINE PATHOLOGY. In people with CP, severe motor impairment is associated with scoliosis and other deformities (98,99). Scoliosis may progress during adulthood, and those at 50 degrees or greater at skeletal maturity may deteriorate more rapidly (100). Scoliosis can cause seating and pressure problems, impaired respiratory function, and pain (85,100), and may be associated with windswept hips and pressure sores (85). It has been reported that spinal fusion improves the QOL for those with CP (101).

Spinal stenosis must be ruled out whenever significant functional change is noted, particularly for change in or loss of walking skills, increased leg spasticity, change in bladder habits, neck pain, vague sensory changes, and (late) change in arm and hand function (102,103). A tethering effect on the spinal cord also may occur, resulting in cranial nerve changes. Some early reports noted a higher risk in those with an athetoid or dyskinetic component (104,105); however, more recent reports show these problems are present in spastic forms of CP as well. While it is generally held that stenosis is due to early spondylosis and compression, there may also be a predisposition to it in those with a congenitally narrow canal, especially at C4 to C5 (102,104). Diagnosis is made through imaging studies, while comparatively evoked potentials may also be helpful in determining neurologic function. Surgical decompression may prevent further, often catastrophic, loss of function, but does not ensure return of lost function, particularly in cases of long-standing compression with spinal cord atrophy. Recurrence at levels above or below surgical correction may be noted (106,107). Postoperative management planning should accommodate changes in functional capabilities and care needs. The presence of an athetoid movement component will affect postoperative spine stabilization and possibly head positioning and neck mobility. When no surgical intervention is undertaken, a frank discussion of possible respiratory compromise and the future need for ventilator assistance should be provided.

PERIPHERAL NEUROLOGIC COMPRESSION. Radiculopathies may be a cause for painful complaints, and appropriate evaluation and treatment should ensue. It is most important that treatment strategies are based on the person's history of function, that there is effective input from that person or their care provider, and that practical outcome goals are identified. Although not as common as a musculoskeletal etiology, nerve entrapment is also a cause of pain. The most common nerves and areas of entrapment as reported by adults with CP are the same as those susceptible to compression in the nondisabled population: the median nerve at the carpal tunnel and the ulnar nerve in the hand distally and at the elbow. Compression points are often related to use of crutches, transfer techniques, propelling wheelchairs, or existing deformity. Work-related or positional activities may also cause entrapments, just as in the nondisabled population. There is no reported increased incidence in CP. All hand pains or sensation changes do not represent nerve entrapment. Often, these complaints are actually problems of repetitive motion or are position-related. While they may be ascribed to carpal tunnel syndrome, they often respond poorly to surgery (108). Appropriate testing (including electrodiagnostic testing) is necessary to determine their etiology. Where treatment options are similar for disabled and nondisabled adults, some modification of management will be required if functional independence is changed by or during treatment.

OSTEOPOROSIS. Osteoporosis has been documented in at least 50% of children and adults with CP (109). The aging process may exacerbate this issue, as does anticonvulsant use and mobility impairment. Fractures were noted to be common reasons for hospital admission (68). Pathologic fractures occur typically in the long bones, but frequency data vary and no large studies of people with CP have been reported. Low serum 25-OH vitamin D concentrations are not identified as a cause in most cases described in the literature (110). Typical screening devices or schema do not accurately identify osteoporosis risk in women with disability (111); therefore, bone mineral density testing and counseling on fall risk are important for

both women and men with disability. Dual energy x-ray absorption (DXA) scans must be read with caution, since contractures often skew results. The recommendation is to use the scan results of the distal femur, as is used in children with CP and contractures (112). Use of bisphosphonates is described, but the functional improvement derived from these drugs over the long term is unknown.

Additional Health Conditions

There are no comorbidities known to be associated with CP. As noted, general health is good. Little is reported about cardiovascular health outcomes or risk factors such as reduced habitual physical activity levels and/or increased sedentary time in adults with CP. A study of adults living in group homes from upstate New York notes increasing health conditions with age for adults with CP as would be expected: cardiovascular, respiratory, and hearing/vision (113); this has been replicated in Taiwan and Israel (114,115). Of interest is that in comparison to U.S. national norms, there are fewer cardiovascular risk factors than seen in the general population. Peterson et al postulate that it is possible that adults with CP age more quickly than their healthy peers, resulting in an earlier onset of cardiovascular disease (CVD) (75). Either this is a healthier population or, more likely, there has not been effective screening and monitoring. Longitudinal research is necessary to determine risk factors and causation of cardiovascular events in aging adults with CP. In looking more critically at this population, the severity of the CP was related to increasing health problems with aging more than the diagnosis of CP alone (116). Vision and hearing problems may have been present early, and as anticipated, there is an increase in vision and hearing problems with age (113).

Dental issues are reported for adults with CP (79). Medications, nutrition problems, poor dental hygiene, and difficulty with access to dental care all contribute to the ongoing problems into adulthood.

Previously known associated conditions persist into adulthood. Dysphagia continues, and monitoring is required. Constipation also persists, and adjustments to bowel programs may be needed, in particular in individuals with more severe CP (117). From the latter study in children we can learn laxative use is high but dosing is frequently inadequate to prevent symptoms. Gastroesophageal reflux is often reported, but has not been present at increased rates. Intestinal obstruction is reportedly common in CP, and in an upstate New York cohort living in group homes, adults with CP had an increased rate compared to other adults with developmental disability (DD) (116).

Urinary incontinence may also continue, and it must be ensured that there is no dyssynergia or overflow with retention (118). Rosasco et al reported adults with CP had a higher incidence of UTIs that was related more to severity than the presence of CP (116), compared to other adults with DD living in group homes in upstate New York. UTI has been found to be a common reason for hospital admission for adults with CP (68). Neurogenic bladders in adults with CP are only infrequently associated with upper tract pathology (119). Some women report that incontinence consistently occurs at a particular point of their menstrual cycle, often associated with increased spasticity (120). Urinary incontinence can be effectively addressed through well-established diagnostic and intervention approaches. There are no available data that assess the adverse impact of urinary incontinence on sexual functioning and social integration in CP, but anecdotal support for this association is abundant. In both men and women, urinary incontinence should be identified and addressed, regardless of age or other conditions.

Respiratory problems have been implicated as the cause of death early in life and in early adulthood. Use of vaccinations may be helpful, along with vigilance and monitoring. Respiratory problems may increase with progressive scoliosis, and aspiration from gastroesophageal reflux disease (GERD) or dysphagia must be recognized. Sleep disorders related to pulmonary problems should be considered with progressive scoliosis, especially with complaints of poor sleep, morning headache, or daytime sleepiness.

There has been suggestion that obesity is a problem in CP, and yet studies are inconclusive (75,121). In fact, a small study of adults with CP identified mean body fat percentages and body mass indexes (BMIs) were within normal range, although 40% had heights below the fifth percentile for age and gender. Fifty-five percent reported dysphagia (122). Measurement of the percentage of body fat, which is the mark of healthy weight, is not routinely done, and state-of-the-art techniques (eg, DXA, bioelectric impedance analysis) have not been standardized for this population.

Sexual Functioning

Empirical data on sexual development and functioning of persons with CP are scarce. The most recent information comes from research on romantic relationships and sexual development of a well-documented cohort of mainly ambulatory persons with CP with average intelligence. About 30% of participants were found to be below their age level for dating and intimate relationships (49). Women's sexual health and functioning is better described than men's. Women with CP typically have limited participation in health maintenance activities such as routine pelvic examinations, Pap smears, and breast examinations (60,123). Office visit planning is required for those with significant motor impairments to ensure a complete examination. Attitudinal barriers of health care providers often limit services and education. However, women with CP are typically able

to conceive and carry pregnancies to term without the expectation of major complications related to their CP. Use of contraceptives has not been well studied, and consideration of thrombotic effects must be considered in the choice of options. A commonly offered contraception is injectable nonestrogenic formulations such as depot medroxyprogesterone acetate (DMPA), although long-term effects are not well defined (120,123). DMPA can reduce bone mineral density, so may not be indicated in those with existing low bone densities or other risk factors for osteoporosis. An intrauterine device (IUD) may reduce menstrual flow and provide effective contraception, but its insertion may necessitate sedation (124). Women with CP report fewer sexual encounters as compared to other women with disability (35,125). Women with early-onset disability also experience high levels of sexual desire compared to other women with disability, postulated as being related to reduced social opportunities, frustrated satisfaction of sexual urges, discouragement of childhood sexual expression, or perceived social stereotypes (125).

Men with CP also should receive information on sexual functioning, including information on contraception and protection. There have been no reported problems with fertility. Most young men with CP reported a sexual interest (fantasizing or seeking erotic material), experienced feelings of sexual arousal, although some young adults with CP, however, experienced anorgasmia (49). Men with CP have a higher incidence of cryptorchidism (53%) at the age of 21 years compared with the general population (126), but this problem can easily be missed when the testis cannot be visualized or palpated with the patient in the standing position.

Adults with CP have many questions, including the use of contraception, heredity, pregnancy, assistive aids, spasticity, and pain. They also reported to have worries about interaction with a partner, experiencing a deviant body image, shame about their body, reactions of their body, and how to stimulate their partner sexually (127).

The topic of sexuality does not come up easily at appointments with physicians. Young adults with CP request an active role for the physician or other health care professionals to openly communicate on sexuality and provide the necessary information (127). Topics specific to CP that should be discussed are the following (127):

- Information about fertility, pregnancy, parenting
- Influence of spasticity (stimulation, masturbation, orgasm, etc.)
- Advice on alternative ways of making love, adapted postures
- Use of specific assistive devices
- Possibility of medication and operations to facilitate postures
- How to ask for assistance in activities of daily living in preparation for sex
- Information about sexual caregiving

SPINAL CORD DYSFUNCTION

SB and SCIs are the most common etiologies for spinal cord dysfunction (SCD) in childhood, although infectious, rheumatologic, demyelinating, and tumor etiologies are also seen. The incidence and prevalence of SCD in general is low in a pediatric population. Earlier chapters have identified the decreasing incidence of both SB and SCI.

The prevalence for either, and for SCD in general, is only an estimate, and is well below estimates for ID and CP. It is also estimated that life expectancy is increasing, and therefore, it is important to understand the lifelong health and functional issues of adults with childhood-onset SCD. SCD usually involves multiple organ systems, often to a significant degree. These medical conditions are fairly well described but not well defined over a lifetime. Consequently, there will be more medical monitoring than in other conditions. There is significant overlap in the long-term management of those with SB and SCI, although there are disability-specific health issues and risks. There is much more research and information available about adults with SB than adults with childhood-onset SCI. Information about adults who have sustained SCI may be useful in supporting and managing the health issues of adults with congenital and childhood- onset SCD; in a similar manner, identified issues for either SB or childhood-onset SCI may have applicability for both. This section will highlight what is known about the health of adults with SB and childhood-onset SCI independently. For both subsets, adults are presenting with health challenges, such as renal dysfunction, musculoskeletal problems, neurologic complications, pulmonary conditions, pressure ulcers, and sexuality and reproduction issues (see Tables 21.3 and 21.4).

Spina Bifida

MORTALITY. As noted, in general, both early and late survival has improved over the past 20 years. The majority of U.S. information available is through databases that involve specific sites of care for programs serving people with SCD, some state malformation registries, and international registries. Of note is that a new registry, the National Spina Bifida Patient Registry, has been initiated among a select number of programs in the United States. Mortality remains high early in life, and continues at a higher-than-typical rate throughout adulthood (128,129). About 75% of children born with an open SB today reach early adulthood; survival has a positive association with a lower level of spinal involvement and less neurologic deficit; presence of a shunt decreases survival probability in adulthood (128,130,131). Common causes of death in adulthood are renal failure, cardiac dysfunction, respiratory complications, infections, injuries, neoplasm, and causes related to the central nervous system (CNS); many deaths can be unexpected, including those from seizures,

TABLE 21.3 SPINA BIFIDA: COMMON AGING HEALTH AND PERFORMANCE CHANGES

COMMON RELATED HEALTH CONDITIONS	PREVENTION STRATEGIES	TREATMENT STRATEGIES
Urologic/renal disease UTIs Incontinence Vesicoureteral reflux Urolithiasis End-stage renal disease Bladder cancer	Routine monitoring—UTI frequency, renal scans, urodynamics Maintain routine urology appointments Monitor for bladder cancer following augmentation surgery	Appropriate management, consideration of alternatives for treatment with increased frequency With change consider neurologic evaluation for cause New hematuria, pain—screen for bladder cancer
Musculoskeletal Shoulder pain/overuse Scoliosis Joint pain	Routine exercise, especially strengthening posterior shoulder Monitor and query routinely Joint protection strategies Routine exercise Biomechanic and ergonomic assessments	Focal musculoskeletal evaluation Evaluate for neurologic change with new symptoms Modify equipment (possible power wheelchair) at the workplace, biomechanics of function Therapy prescription Adjust orthoses, shoe wear, wheelchair
Osteoporosis/fracture	Calcium/vitamin D supplement Education and fall prevention	DXA evaluation Consider treatment with multiple fractures Exercise when appropriate
Neurologic Hydrocephalus Chiari malformation Tethered cord syndrome Epilepsy Vision changes	Routine monitoring Query for changes, recognition of symptoms related to central structures Maintain neurosurgery appointments Routine neurology appointments with active epilepsy	Neurosurgical evaluation Postsurgery, may require rehabilitation admission Cognitive and functional assessments post intercurrent events to ensure safe community living
Obesity Metabolic syndrome	Monitor body fat—consider waist circumference, DXA, BIA Routine exercise Nutrition management Monitor cardiovascular health early, metabolic syndrome symptoms	Nutrition referral Exercise prescription
Cardiovascular health	Early monitoring of blood pressure, serum panels for risk factors	Treatment of symptoms and events
Pressure ulcers	Frequent position change Monitor skin, nutrition, equipment, change in function	Modify positioning or pressure relief equipment Ensure good nutrition Appropriate care for ulcer staging May need change to tone management Surgical referral
Pulmonary restriction Pulmonary infection	Monitor for infection Immunizations	Evaluate for neurologic change with new symptoms Consider sleep study or O_2 supplement
Bowel incontinence	Monitor and adjust program for change	Evaluate for neurologic change with new symptoms
Lymphedema	Monitor; use of compression and elevation at first sign	Referral for lymphedema program and prescribe compression garments Modify equipment if needed

(*continued*)

TABLE 21.3 SPINA BIFIDA: COMMON AGING HEALTH AND PERFORMANCE CHANGES (*CONTINUED*)

COMMON RELATED HEALTH CONDITIONS	PREVENTION STRATEGIES	TREATMENT STRATEGIES
Latex allergy	Limit exposure to latex	Acute event treatment Appropriate recognition in medical record, personal acknowledgment
Mental health	Monitor routinely, especially for depressive or anxiety symptoms	Specialty referral as appropriate Referral for psychological and social support Use of community resources
Sexual functioning Fertility/reproduction Interference spasticity or pain Emotional/body image	Engage in discussion regarding sexuality Provide education about sexuality and function—appropriate modality for cognition and function Assist with environmental modification for routine assessments as able Ensure pregnancy high-risk needs are met	Urology referral for male fertility or performance Following pregnancy, support may be needed at home
Health maintenance	Monitoring—see Table 21.9	

Abbreviations: BIA, bioelectric impedance analysis; DXA, dual energy x-ray absorptiometry; UTI, urinary tract infection.

TABLE 21.4 CHILDHOOD-ONSET SPINAL CORD INJURY: COMMON AGING HEALTH AND PERFORMANCE CHANGES

COMMON RELATED HEALTH CONDITIONS	PREVENTION STRATEGIES	TREATMENT STRATEGIES
Urologic/Renal disease UTIs Urolithiasis Incontinence Reflux	Routine monitoring—UTI frequency, renal scans, urodynamics Maintain routine urology appointments	Appropriate management, consideration of alternatives for treatment With change consider neurologic evaluation as cause
Musculoskeletal Shoulder pain/overuse Scoliosis Other pain complaints	Routine exercise, especially strengthening posterior shoulder Monitor and query routinely Joint protection strategies Routine exercise Biomechanic and ergonomic assessments	Focal musculoskeletal evaluation Evaluate for neurologic change with new symptoms Modify equipment (possible power wheelchair) at the workplace, biomechanics of function Therapy prescription Adjust orthoses, shoe wear, wheelchair
Osteoporosis/fracture	Calcium/vitamin D supplement Education and fall prevention	DXA evaluation Consider treatment with multiple fractures Exercise when appropriate
Neurologic Spasticity Autonomic dysreflexia	Routine monitoring Query for changes Adjust medications with reported change	Tone management—progress to more aggressive strategies Evaluate for neurologic change, painful symptoms, bowel/bladder etiologies, fractures, pressure ulcers with more frequent AD symptoms
Obesity Metabolic syndrome	Monitor body fat—consider waist circumference, DXA, BIA Routine exercise Nutrition management Monitor cardiovascular health early, metabolic syndrome symptoms	Nutrition referral Exercise prescription

(*continued*)

TABLE 21.4 CHILDHOOD-ONSET SPINAL CORD INJURY: COMMON AGING HEALTH AND PERFORMANCE CHANGES (*CONTINUED*)

COMMON RELATED HEALTH CONDITIONS	PREVENTION STRATEGIES	TREATMENT STRATEGIES
Pulmonary conditions Ventilator dependency	Monitor for infection Immunizations Consider diaphragm or phrenic nerve pacing	Evaluate for neurologic change with new symptoms Consider sleep study or O_2 supplement
Pressure ulcers	Frequent position change Monitor skin, equipment, change in function	Modify positioning or pressure relief equipment Appropriate care for ulcer staging May need change to tone management Ensure good nutrition Surgical referral as appropriate
Bowel incontinence	Monitor and adjust program for change	Evaluate for neurologic change with new symptoms
Latex allergy	Limit exposure to latex	Acute event treatment Appropriate recognition in medical record, personal acknowledgment
Mental health	Monitor routinely	Specialty referral as appropriate Referral for psychological and social support Use of community resources
Sexual functioning	Engage in discussion regarding sexuality Provide education—appropriate modality for level of function Assist with environmental modification for routine assessments as able Ensure pregnancy high-risk needs are met	Urology referral for male fertility or performance Following pregnancy, support may be needed at home
Health maintenance	Monitoring—see Table 21.9	

Abbreviations: AD, autonomic dysreflexia; BIA, bioelectric impedance analysis; DXA, dual energy x-ray absorptiometry; UTI, urinary tract infection.

unrecognized shunt malfunction, and acute renal sepsis (129,131). With decreasing birth prevalence of SB and improved survival over the past 40 years, the majority of people living with SB in the United States are adults, aged 18 years and older (132,133).

HEALTH AND QUALITY OF LIFE. Individuals with SB have neurosurgical, urologic, musculoskeletal, and cardiopulmonary issues that demand a primary care provider well versed in care for those with SB, routine specialty care, and periodic team evaluations. Youth and young adults with SB access health care services more than their age-matched peers, with hospitalizations significantly higher as well (134). Health care expenditures are high for children with SB, but adult expenditures are increasing and are about three to six times greater than those for adults without SB (132). Young adults tend to seek health care in the ED, and few have a medical home that can coordinate care needs (134). UTI is the single most common acute diagnosis among patients with SB seen in the ED, followed by neurologic issues in the United States (135). A U.S. claims data report noted that people with SB averaged 0.5 hospitalizations/year with more than 50 times the number of admissions for UTIs compared to those without SB (136). An Australian study noted common diagnoses for hospital admissions were chronic renal failure, pressure ulcers, respiratory conditions, and acute shunt dysfunction (137). Dicianno et al report UTI as the most common primary diagnosis for hospitalization, followed by complications from devices/grafts/implants and skin wounds through a U.S. Agency for Healthcare Research and Quality national inpatient sample (138). A higher number of hospitalizations are associated with a higher number of pressure ulcers and lower self-management abilities, as noted in a U.S. cross-sectional multicenter study (139). There may be potentially preventable causes for these ED visits and hospitalizations, and the need for self-management skills has been promoted (139).

However, the complexity of care needs, lack of knowledge about risk factors for these young adults with SB, and nonstandardized transition programs make tackling this problem difficult.

Health and wellness behaviors, such as self-management skills and fitness programs, are being promoted for adolescents and adults. In general, adults with SB have less healthy diets, less exercise, and more sedentary behaviors, with a peak in substance abuse in the late 20s (140). Aerobic fitness in adults with SB has been associated with decreased cardiovascular risks (141). Adults with SB report sex education in school, but not routinely through family or physicians (142). Cardenas et al further report that in their small U.S. sample, adults with SB who reported smoking were more likely to be sexually active. Self-management and goal setting for independence are being promoted through a variety of techniques, including camps, counseling, and education modules (139,143,144).

Using a prospective cohort study of a nationally representative sample of adolescents with SB in the United States, Liptak et al report that self-reported health of individuals with SB declined during adolescence and early adulthood (145). This corresponds to a smaller self-report and use of standardized instruments study comparing youth and young adults in Canada, noting a more common report of fair to poor health by the young adults (146). Younger age and lower level of surgical lesion were associated with higher self-reported health.

Reports on HRQOL are difficult to synthesize. A variety of measures are used; most are standardized for a general population biased by societal values or are condition-specific instruments with notable weaknesses (147). Young et al report that single global health ratings for individuals with SB are more favorable than HRQOL scores because of the inherent bias in those instruments (146).

In general, adolescents and adults score lower on domains that relate to physical functioning. The theme of low scores on physical functioning scales compared to a general population is not surprising. The relationship of the level of lesion in SB to report of health is important to note. However, there are discrepancies within domains related to emotional, social, or school issues. Two studies from the Netherlands used the Medical Outcomes Study 36-item Short-form Health Survey and noted the mental component or emotional health scores were equal to or exceeded scores of a general Dutch population (148,149). Thus, as has been found with other congenital and childhood-onset disabilities, living with SB assumes a different sense of health or QOL compared to those without a disability over a lifetime.

The larger sense of life satisfaction and participation for adults with SB shows areas of concern. In general, adults with SB tend to be less likely to move away from home, attend college, maintain employment, or have romantic relationships (150). Overall there are low rates of employment and independent living for adults with SB, regardless of sex (151). This low employment is in the face of relatively high service use, which may counterintuitively restrict independence and employment options (152). It has been proposed that use of assistive technology and universal design might improve life satisfaction (153). Using the Life Satisfaction Questionnaire, the highest proportion of dissatisfaction for Dutch adults with SB was with financial situation, partnership relations, and sex life, and those with hydrocephalus were less satisfied with self-care ability and partnership relationships than those without hydrocephalus (154). Dissatisfaction with life often leads to participation in health-risk behaviors (142). Over time, family satisfaction plays a large role in a favorable QOL determination (155), and pain, especially increasing pain, is associated with a lower sense of QOL. Zukerman et al note predictors for achievement of adult milestones include higher executive functioning level, higher intrinsic motivation (which has a relationship to cognitive functioning), less intrusive parents, and higher socioeconomic status (150).

FUNCTIONAL STATUS AND PERFORMANCE. There are no large studies to identify change in function over time; however studies report less walking in middle-aged adults compared to adolescents and early adults (146). It is expected that mobility will change over time, as will the need for assistance with daily care. However any significant change in function or insidious onset of changes should initiate a workup for the presence of tethered cord syndrome (TCS), shunt malfunction, Chiari II malformation progression, syringomyelia, or an acute process. Studies identify mobility based on a variety of schemas: defect level; muscle strength testing; sensory level; distance able to walk (ie, community, household, exercise, nonwalker); or a standard nomenclature: lower (S1 and below), middle level (L3-5), and higher level (L2 and above). American Spinal Injury Association (ASIA) levels or declaration of complete or incomplete function are not typically used. Lower lesions are associated with higher walking abilities, with or without aids (131,156). A small cohort of adults with sacral-level myelomeningocele (MMC) was noted to have maintained their walking abilities for low-sacral lesions, and almost 90% maintained walking in the high-sacral group (157). Complications reported included scoliosis, osteomyelitis, amputations, and spinal cord tethering. Small studies in Sweden and the United States have identified that across the spectrum of SB, mobility decreased from early childhood to early teen years (131) and through adulthood (158). Common reasons for change in walking included deterioration of neurologic function, spasticity, contractures, low back pain, acute medical events, and deconditioning (158). At least three-fourths who walked during their teen years continued walking as adults.

The associated cognitive effects of SB influence the level of functional independence in adults. A small cohort

report showed that most young adults with hydrocephalus and lesions at L2 or above were dependent for sphincter control, locomotion, and self-care, with an additional number requiring assist with transfers and social interaction and communication (159). Those without hydrocephalus or with hydrocephalus and lesions below L2 required assist with sphincter control only. An additional small study reported more difficulties in independence and QOL, with increasing numbers of shunt revisions (160). Upper limb performance is decreased for coordination in general, and worse in higher levels and with visual or cognitive challenges (161). In general, prospective memory (ie, recall required to initiate an activity) is impaired in older and younger adults with SB, and this memory declines sharply with age (162). Typical reminders (eg, written prompts) are not used readily. Since prospective memory relates to self-management capabilities, acknowledgment of needed supports at any age is required.

Rehabilitation and use of assistive technology should be focused on improving or maintaining physical functioning and independence. There are high physical demands for walking, even with gait aides, and also for performance of daily care (163). With age, there is increasing use of a wheelchair, and increasing wheelchair dependence is associated with higher-level lesions and hydrocephalus (164). Adults with SB using wheelchairs have lower energy cost compared to adults with and without SB, making use of a wheelchair more efficient (163). It is not clear that wheelchair use results in less activity, since increased efficiency may promote activity. In a small study of adolescents and young adults with SB and SCI, there was reduced aerobic capacity and reduced upper limb strength (165). Therefore, it is important to promote exercise (eg, stretching, resistance training, and aerobic exercise) both for those who use wheelchairs as well as those who walk. Therapeutic interventions reduce pain, increase biomechanical efficiency for wheelchair use, and improve balance and physical activity (166). Sports participation does not require a certain level of physical ability, but relates more to personal preference. Having good social support from family, a perception of athletic competence and appearance, and high global self-worth is associated with more sport participation. Those who state they participate report higher physical activity (although not substantiated with the use of accelerometer), and demonstrate better performance on some physiologic measures (141).

UROLOGY/NEPHROLOGY. Urinary and renal issues are the most common health problems for those with SCD, and demand routine monitoring. UTIs are the most common health condition noted both for outpatient management and hospitalization in adults with SB (136,138). Renal damage and renal failure are among the most severe complications in SB (167), and contribute to early and late mortality. Adolescents and early adults with SB often experience bladder and renal function deterioration following puberty (168), and this is the time health care transitioning takes place.

In general, typical strategies for management of neurogenic bladders are used with goals of achieving continence, preventing UTIs, preventing renal calculi, managing detrusor pressures to prevent upper tract problems, and monitoring renal function to prevent renal failure. Clean intermittent catheterization (CIC) is an effective long-term management strategy for properly selected persons with neurogenic bladders from SB (167,169), usually concomitant with medications. Medication choices are similar for neurogenic bladder management, and side effects may limit compliance in early adults with SB. There is no consensus for the evaluation, follow-along studies, and general management or management of bacteriuria among SB programs responding to a national U.S. survey (170), and there is no registry data about long-term outcomes. Typical surveillance schedules during adolescence and adulthood include annual urology consultation with ultrasound and serum creatinine (Cr) studies, and urodynamics annually during growth and when indicated due to symptomatology, change in function, and ultrasound changes (168). Renal function, as measured by Cr clearance, intravenous pyelogram (IVP), ultrasound, or scan, has been found to be normal in 47.7% of patients with SB and abnormal in 46.1%, without regard for symptoms (156). In patients with lumbar-level SB who undergo CIC and are dry between catheterizations, only 38% have normal renal ultrasound and an abnormal Cr clearance greater than 1.5 mg/dL (171). This correlates well with the fact that renal failure is the leading cause of death among patients with SB despite proper management and follow-up (172).

For adults with SB, almost 60% of hospital admissions are for urologic reasons, with neurologic problems accounting for almost 21%, and dermatologic problems (likely wounds and ulcers) almost 20%. Of the urologic admissions, almost half of these were for conditions such as UTI and renal calculi (173). In one study, urinary tract stones were responsible for about 30% of all renal complications (172). Repeated UTI, along with pyelonephritis and an already compromised kidney, can lead to acute renal failure with loss of nephrons. Unfortunately, by the time the serum Cr begins to rise, the patient will have already lost up to two-thirds of their nephrons (174). It is important to note serum Cr is dependent on muscle mass, so in adults with SCD who often have low muscle mass, the serum Cr may not be indicative of the true renal function (175).

A study comparing long-term urologic outcomes among children and adults with neural tube defects noted the type of neural tube defect influenced the urologic outcome (176). Neurogenic bladder was seen in practically all those with MMC, with caudal regression syndrome (CRS) above 50% and spinal lipoma (SL) below 50%. Vesicoureteral reflux was most common in MMC, with CRS surprisingly close behind. The incidence of renal agenesis

was highest in CRS. Subjects with SL were best controlled with CIC and medications.

There have been a variety of procedures to assist with acute and long-term management of neurogenic bladders developed and/or promoted over the past 20 years in those with SCD. However, aside from a few retrospective cohort studies regarding specific interventions (177–179), there are no large or randomized controlled studies to identify best treatment strategies or factors that may indicate the procedure of choice (180,181). Furthermore, there are no reports of the effectiveness of surgical interventions over a lifetime. Newer interventions include urethral collagen injections and surgical slings to decrease incontinence (164), enterocystoplasty and detrusorectomy to increase capacity or decrease pressures (182,183,184); continent urinary diversions to promote continence (185), and botulinum toxin injection to the bladder or urethra to decrease intravesicular pressures (164); procedures may be used concomitantly or serially. Urinary diversions continue to have a place in neurogenic bladder management, although there is more investment of health care resources compared to augmentation surgery (183). Augmentation surgeries by enterocystoplasty carry a high complication rate, including bowel dysfunction, B12 malabsorption, metabolic changes, difficulty distinguishing bacteriuria from infection, persistent mucus, increased bladder and renal calculi, delayed spontaneous bladder rupture, and possible risk for bladder malignancy (182). A small study from a center in British Columbia noted no significant increase in HRQOL in SB patients who underwent reconstruction for incontinence, compared to those who did not (186).

Moderate hypertension and proteinuria are associated with increased risk of progression of renal dysfunction, eventually leading to chronic renal failure in patients with SB. For this reason, as well as cardiovascular protective effects, even moderately elevated blood pressure (BP) should be treated. An angiotensin-converting enzyme (ACE) inhibitor should be considered, except in cases of advanced renal disease, due to the risk of hyperkalemia and further advancement of the renal disease (187). There are increasing numbers of children with SB who have undergone renal transplantation, with or without lower urinary tract reconstruction (188), and mortality and morbidities into adulthood are not well characterized.

Bladder cancer has been reported in adults with long-term SCI, and it has also been reported in adults with SB. The characteristics appear to differ from adult-onset SCI patients with younger age onset, variable tumor histology and advanced stage, and poor survival. The possible association with enterocystoplasty is a source of concern, but carcinogenesis in this setting is likely multifactorial. Thus, for individuals who have undergone enterocystoplasty, recommendation is for lifelong surveillance including annual cytological evaluation and biannual cystoscopy (182). A small case series of adults with SB and bladder cancer, unrelated to augmentation procedures, cautions health care providers to consider cystoscopy and biopsy in the face of pain, increased urinary leakage, new recurrent infections, or gross hematuria (189).

Urolithiasis is also reported in adults with SB. A retrospective center study noted recurrent UTIs and incorporation of bowel tissue into the continent reservoir were independent risk factors for bladder stones. The independent risk factor for upper tract stones was an indwelling catheter (190).

Urinary incontinence in adults with childhood-onset SCD can persist into adulthood, and can be a socially limiting condition. Up to 80% of adults with SB can achieve social urinary continence (76), although a survey of persons identified in a state registry as having spina bifida reported only a slight majority of the adults had achieved independence in urinary management (191). For adults with SB, incontinence has been shown to be associated with partial employment or unemployment (192). There has been an attempt to begin to explore the issues of incontinence and QOL. For adults with SB, urinary or fecal incontinence does not appear to play a major determinant role in HRQOL (193).

Many patients with SCD receive regular urologic follow-up as children, but not necessarily as adults. Adults with SCD who do not have urinary calculi or urinary incontinence are often assumed to be urologically stable. However, many adults with SB (and likely SCI) have been found to have urologic abnormalities, such as abnormal renal ultrasound or elevated serum Cr, which put them at increased risk of further renal problems, especially in the upper urinary tract (171). Many of these patients were also found to have increased pressure (>40 cm H_2O) in their bladder with the storage of urine at normal volumes. It is imperative that individuals with SB maintain at least annual urologic evaluations through their lifetime.

MUSCULOSKELETAL COMPLICATIONS AND CONDITIONS. Level of motor function and musculoskeletal abnormalities are typically the areas of concern during growth and development, and often changes are not anticipated during adult years. Pain is a common complaint, and may be related to musculoskeletal issues, although in SB, tethered cord must be considered.

Overuse syndromes are common for wheelchair users, and have been identified in adults with SCI at shoulders, wrists, and hands. In a comparison with adult wheelchair users, those with childhood-onset disability had fewer shoulder pain complaints than those with adult-onset disability, even though lifestyles were no different (194). Shoulder pain in adults and adolescents with SB is not as common as in adult-onset SCI wheelchair users, although older SB subjects had more pain than younger ones (195). It is important to identify the risk for shoulder pain, recognize the onset, evaluate, and treat appropriately.

Scoliosis is common in SB, and is a common contracture noted in adults. It rarely progresses in adulthood.

Spinal fusion has usually been performed prior to adulthood, but does not appear to improve the QOL for those with SB (101). A combined anterior and posterior approach is reported to be more effective in older adolescents and adults with pelvic obliquity (196). Seating difficulties, back pain, and pressure ulcers arise from the scoliosis and pelvic deformities. Adults with SB report back pain less frequently than those with SCI, although, in general, pain complaints increased with age. In adults with SB, back pain may presage tethered cord.

Hip dislocation is related to thoracic or high lumbar neurologic-level abnormalities, and hip contractures notable in high neurologic level, but also in thoracic and high lumbar levels (197). There are no published reports of hip or knee pain in adults with SB, although this should not be unexpected in those who walk, given muscle imbalances and poor skeletal alignment. Charcot joints can be seen given the lack of sensation and muscle imbalances, especially in adults with SB, and particularly those with lower-level defects (95). Other contractures and deformities include equinus foot in thoracic-level lesions, clubfoot with mid-lumbar lesions, calcaneal foot in sacral lesions, knee flexion contractures with any lesion but especially thoracolumbar, and hip flexion with lesions above the sacral level (164). All relate to muscle strength imbalances, and long-standing changes in biomechanics may cause abnormal stresses, instabilities, and degenerative changes. New onset contractures or spasticity should suggest evaluation for TCS, hindbrain changes, hydrocephalus, or other CNS acute processes.

Osteoporosis with associated fractures has become an area of interest and evaluation. For adults with SB, the high incidence of renal dysfunction is an added component for osteoporosis, given that renal dysfunction can lead to impaired bone mineralization (174). Renal dysfunction can also lead to metabolic acidosis as well as hyperparathyroidism; there may also be hyperphosphatemia, which can enhance the secondary hyperparathyroidism. This often necessitates the requirement for a low-phosphate diet and may also include taking phosphate-binding agents. For those with SB, bone mineral density is one to two standard deviations below the normal population, without a difference between ambulatory and nonambulatory patients (198). Treatment is not definitive. There are proponents for managing with calcium and vitamin D and/or using bisphosphonates, although no long-term information is available. Continued walking with muscle activity and weight-bearing has a positive effect on bone mineral density in those with SB (198). Again, long-term follow-up is not available to identify dosing to achieve and maintain improvements.

Fractures may be more concerning than the risk factor of osteoporosis. Few studies detail incidence and prevalence. In an SB program cohort in upstate New York, where the vast majority are adults and late adolescents, the overall fracture prevalence was 200/1,000, most common during adolescence and least likely during adulthood. In comparing adult and childhood fractures, there was no significance to sex, BMI, defect level, functional independence, shunted hydrocephalus, epilepsy, or other congenital anomalies (199). It has also been noted that patients with a higher level of defect have more of a risk for fractures (198,199). Most of the fractures reported in SB involved the tibia or femur, with 75% occurring in children after casting for an orthopedic procedure (200). Postorthopedic procedure and fracture management must be tailored to the situation. Environmental modifications to prevent fractures may be more effective than pharmacologic interventions (199).

NEUROLOGIC COMPLICATIONS AND CONDITIONS. Adults with SB are at an increased risk for neurologic complications because of the pathophysiology of their disability. Among the neurologic abnormalities seen in SB, the most common for which to monitor are hydrocephalus and TCS, although Arnold–Chiari malformation/hydrosyringomyelia complex, seizures, or headaches also may be seen. The vast majority with hydrocephalus have some form of shunting, possibly contributing to the increased survival rates seen today. However, shunt malfunctions are not uncommon, are often unsuspected, and can lead to significant morbidity and mortality (201). Recommendation is that adults with SB have routine neurologic evaluations and periodic computed tomography (CT) scans to monitor the shunt (76), and it is reported that only 40% of adults with SB with a shunt have regular follow-up (202). Symptoms often seen with a shunt malfunction include headache, vomiting, lethargy, or change in mental status, with other neurologic sequelae also possible (76). Chronic headaches may be seen in adults with SB, and recurrent hydrocephalus or shunt malfunction must be excluded through intracranial pressure (ICP) monitoring if necessary (131). In the absence of increased pressure, further treatment options should be considered for pain management. Presence of hydrocephalus is associated with more dependence for self-care (including bladder and bowel care), for mobility, and for communication and cognitive assistance into adulthood (159).

Tethering of the spinal cord is expected following repair, with adhesions between the spinal cord and dura mater. Tethered cord syndrome (TCS) can be seen at any age for those with SB who report changes in bladder or bowel habits, increase in leg weakness, change in sensory level, onset or increase of spasticity, report of pain (usually backache), or progression of deformities. TCS is associated with lower-level lesions and previous surgeries (164). In adults, an antecedent event such as trauma to the back or buttocks often initiates symptoms. Prominent changes for adults are diffuse leg pain with referral to the anorectal area, and changes in bladder or bowel habits, often difficult to detect given reconstructive surgeries; progressive deformity usually is not noted, as is reported in children (203). Physical examination changes

noting new upper motor neuron signs may not be present until later in the progression. Studies report that tethering, cord thinning, lipomas, cavities within the cord, and diastematomyelia are common in this population, with or without symptoms, so identification on scan may not be definitive (204). Treatment consists initially of conservative management of symptoms, such as medications for dysesthesias or pain, with monitoring or neurosurgical intervention. Neurosurgical intervention is undertaken only when symptoms and clinical presentation support the diagnosis since radiographic studies can be deceiving. Surgical release is usually associated with improvement in pain, urinary symptoms, and weakness, and poorer outcomes are associated with repeat procedures (205); however, not all postsurgical outcomes are good. TCS should be considered with changes seen in urinary function postpuberty.

Adults with SB can have worsening neurologic symptoms from progression of an Arnold–Chiari malformation, with or without the hydrosyringomyelia complex. Presenting symptoms in adults with SB may not be those seen typically with brainstem compression, but may include upper limb weakness, sensory symptoms or reflex changes, ataxia, and lower cranial nerve palsies (156). Postsurgical outcomes vary, including some level of recovery, stabilization of symptoms, further deterioration, and even death.

Epilepsy may remain an active problem in adulthood for those with SB. Seizures are associated with shunts. Most series identify program cohorts with less than 15% requiring active seizure management with anticonvulsants (131,155).

OBESITY AND OVERWEIGHT. Obesity is a reported medical condition in motor disabilities in general. It is commonly seen clinically in an SB population across the life span at similar rates as the general population, although higher in adults especially women (206). Determination of healthy weight is somewhat problematic since anthropometry measurement can be difficult and measures are not standardized for people with SB; BMI is not likely the best proxy measurement, although there has not been standardization of other measurement systems. Obesity is associated with a lower HRQOL (207). There are higher levels of body fat in adults with SB who do not walk, and there is an association of increased body fat with previous hydrocephalus (208).

Metabolic syndrome is a cluster of several cardiometabolic risk factors that can lead to coronary artery disease and diabetes. Diagnosis is made using a combination of increased percentage of body fat; increased serum high-density lipoproteins, triglycerides, and/or cholesterol; increased BP; excess waist body fat; and diabetes mellitus usually with insulin resistance. In a small study of 34 adolescents with spina bifida that used dual x-ray absorptiometry (DXA) to define obesity, investigators identified metabolic syndrome in 32.4% of youth with SB; with presence of obesity, almost 50% were diagnosed with metabolic syndrome (209). Increased BP has been reported in a cross-sectional retrospective analysis of an outpatient program for adults with SB, with less than half being normotensive (210). However, many outpatient programs rarely document anthropometry (209), and better measures for percentage of body fat (eg, waist circumference, DXA) are not considered. Evaluation for metabolic syndrome should be considered, especially in an adult population.

Obesity can often be an associated factor with onset and management of pressure ulcers. It is associated with obstructive sleep apnea, hypoxemia, and consideration for tracheostomy (210). A single case study reports successful gastric bypass surgery in a 15-year old with SB, with improvement in CVD risk factors, reversal of sleep apnea, reduction in body fat mass, and subjective improvement in breathing and reported QOL.

Improved monitoring for healthy percentage of body fat and other parameters may be effective for prevention or at least management (211,212). Appropriate nutrition and adequate exercise and activity should be a lifelong goal in persons with disability.

PRESSURE ULCERS. Pressure ulcers are a commonly occurring secondary condition in adults with SCD related to their impaired protective sensation. For adults with SB, pressure ulcers may be associated with higher level spinal cord involvement and with hydrocephalus (213,214). A recent cohort study reported one in three SB program patients had at least one wound episode over 13 years, with at least a 50% chance of future ulcers (214). Wounds were more common in adolescents, but persisted into adulthood, and foot and ankle ulcers were most common at any age. Key risk factors identified include adolescence, wheelchair use, and bare feet, and possibly obesity and reduced executive functioning. Osteomyelitis is a complication of recurrent or chronic pressure sores, and may ultimately require amputation for management (191).

Despite significant attention from health care systems and increased options for management, pressure ulcers continue to present in this population and incur significant cost. Increased wound education and use of and monitoring of assistive devices remain the mainstay of prevention.

ADDITIONAL MEDICAL CONDITIONS. Pulmonary conditions may be seen in adults with childhood-onset SCD, although not typically reported in SB cohorts. Restrictive lung disease occurs as a consequence of scoliosis, and decreasing pulmonary function with age in the general population is well documented. For adults with SB, changing pulmonary function may indicate further neurologic progression of a Chiari malformation. Obesity is associated with sleep apnea and hypoxemic syndromes. Pulmonary embolism has been identified as a cause of mortality.

Cardiovascular risk factors must be monitored, whether or not metabolic syndrome is present. As noted, hypertension is common and in the cross-sectional study, was associated with diabetes, renal dysfunction, bladder procedures, and renal dysfunction (240). Deep venous thrombosis (DVT) is not common, but there have been reports especially in mid-teens with high-level lesions (164).

GI conditions can be seen in adults with childhood-onset SCD. Usually, they are chronic rather than new or late-onset problems, unless related to progressive neurologic conditions. Adults with SB also report problems with fecal incontinence in about 50% of reported cohorts (131,213). Constipation and diarrhea may continue through adulthood; megacolon can develop if management is inadequate. It has been noted that assistance is commonly required for bowel management in adulthood (159,191,215). Bowel continence is often difficult to achieve, and lack of continence can influence the ability to participate in community activities. There is no management specific to adults with SB, and medications, manual evacuation, enemas, suppositories, large-volume retrograde colonic enemas, and biofeedback have all been suggested (164). Surgeries have also been described, which result in high satisfaction, although there must be monitoring for complications such as stomal stenosis, tube dislodgement, granulation tissue formation, and leakage (164). Of concern is ensuring appropriate evaluation and management of acute abdominal symptoms; a case series of children and young adults notes etiologies included underlying neurogenic bladder or bowel, shunt, and complications from previous surgeries and a substantial mortality rate (216).

Latex sensitization/allergy is an important issue for adults with SB, and the rate may be higher for adults than children (217). The risk of sensitization increases with more surgical procedures being performed (218); the percentage of patients sensitized to latex ranges from 2.97% to 64.5%. Radioallergosorbent testing has been found to be more sensitive with a higher negative predictive value and more accurate than skin prick testing. The prevalence of latex allergy in the SB population is almost 19%, while the prevalence of latex sensitization is 32.4% (218); therefore, every effort should be made to limit exposure to latex.

Lymphedema was recently reported in an outpatient program for adults with SB cohort noting a higher prevalence than in the general population at 9.2% (219). Significant associations were trauma, cellulitis, cancer, obesity, wounds, and hypertension. The presence of power wheelchair seat functions such as tilt, recline, or elevating leg rests showed no protective response. Simple over-the-counter compression garments are not useful, and most adults with SB are unable to apply daily Ace wraps. The edema is often responsive to lymphedema wrapping followed by tailored compression garments. There are two reports of severe and unresponsive lymphedema in adult women with SB—one responsive to suction-assisted lipectomy (220), and the other progressed to a diagnosis of lipedema, which has no successful treatment regimen (221).

Vision may be altered in association with hydrocephalus, and response following shunting is not clear in adults (164). Chiari malformations are associated with brainstem ocular motor disorders such as strabismus, nystagmus, and esotropia. Changes in vision or ocular control may indicate progressive central pathology.

Poor psychological functioning has been acknowledged in an adolescent population, but only recently has there been study of psychological symptoms and risk for depression in adults with SB. The majority of adults have restricted living experiences, usually living with family and not employed. High rates of depressive and anxiety symptoms were found in a small sample of young adults with SB through surveys and self-report measures (222). Greater family satisfaction and more positive attitude toward SB were associated with less depressive symptoms, and pain was associated with more depressive and anxiety symptoms. The presence of pain is associated with reduced physical health and psychological QOL (155,222). Major depressive symptoms are associated with alcohol drinking (140). Improved family functioning and increased self-management of SB issues showed reduced depressive symptoms over time. Health care providers need to have increased awareness of high-risk profiles and the need for mental health and health risk screening and counseling.

SEXUAL FUNCTIONING. The number of adults with SB and childhood-onset SCI is increasing; therefore, the health care community can no longer ignore dealing with the medical and social issues of sexuality (223). A recent report noted that sexual education was received at school, and far less at home or from health professionals (224). Urinary incontinence may limit sexual participation (142,224), although this is not a consistent report (225). Higher neurologic level and presence of hydrocephalus was associated with less participation for both genders, but more problems with sexual functioning for men (213,224). Women appear to be more likely to be sexually active than men (142). There is no published data regarding contraception, but for women, contraception or suppression can be offered considering risks (eg, thrombotic risk, lack of sensation for IUDs), side effects, and need for follow-up (226). Sexual education should be offered, with consideration for cognitive impairments when appropriate.

Many men with SB are able to achieve erections, but only about 53% are able to ejaculate (223). As anticipated, a lower defect gives men a greater chance of being able to sustain an erection, and there are normal testosterone levels. Erectile dysfunction is treatable with medications, although men with SB in a study did have some adverse effects after taking sildenafil, including dyspepsia, nausea, headache, flushing and nasal

congestion, hematologic changes, and UTI. The dyspepsia was treated with antacids, and the UTI was treated with antibiotics. The remainder of the adverse events did not require treatment (227).

Women with SB had fewer problems with sexual functioning and were able to maintain pregnancies. Arata et al reported that there was no increase in back pain, no changes in neurologic or motor function, and no changes in bowel or bladder function during or following pregnancy (228). There were two commonly seen secondary conditions during pregnancy: UTI—but only in women who did not have normal voiding patterns—and pressure ulcers sometimes requiring hospitalization. Women with SB also had more emergent and elective C-sections than in the normal population. Women with SB were also found to have more antenatal admissions than women without SB, and it was noted that women with SB using wheelchairs exclusively had an average of 2.8 admissions antenatally per pregnancy, with an average stay of 25.8 days, while women with SB who walked had an average of 1.9 admissions antenatally per pregnancy, with an average stay of 17.3 days. More women with SB are admitted with preeclampsia than in the normal population, but given the incidence of renal dysfunction in this population, the prevalence is not overly high (228). Further study is needed to fully address the possible complications of pregnancy and childbirth in patients with SB. Pregnant women with SB may be evaluated through a high-risk pregnancy service.

There is no information specifically regarding typical gynecologic screening and prevention practices for women with childhood-onset SCD; however, national data regarding women with mobility impairments, especially those requiring use of a wheelchair, clearly demonstrate minimal participation, likely due to environmental and attitudinal barriers. Pregnant women with childhood-onset SCD should be at least evaluated through a high-risk pregnancy service.

Childhood-Onset Spinal Cord Injury

MORTALITY. Using data from the National Spinal Cord Injury Statistical Center over a 30-year period, it has been determined that life expectancy for adults injured as children appears to be slightly lower than that of those with comparable functional levels incurred through SCI as adults (229). More specifically, for those injured at a young age with incomplete injuries and minimal deficits, there is about an 83% chance of normal life expectancy, and for those with high cervical injuries without ventilator dependence, the estimate is about 50% of normal.

HEALTH. In general, adults with childhood-onset SCI note increasing occurrences of SCI-related complications and overuse syndromes. There are also the accompanying aging issues related to CVD. Recent longitudinal studies have reported prevalence and developed odds ratios (ORs) for a variety of medical conditions, related to ASIA impairment scale (eg, A, B, C, D) groupings. UTIs are commonly reported in all groups, but less so in those with ASIA impairment scale D. There are increased ORs over time for severe UTIs for those with C1-4 A, B, C. Pressure ulcers commonly present in all groups, but especially in those with ASIA impairment scales A, B, and C (not D). Spasticity was more common in those with C1-8 A, B, C, and increased ORs for those with C1-4 A, B, C. Shoulder pain was common at all levels and impairment scales, and ORs increased over time for all groups. Those with C1-4 A, B, C and C5-8 A, B, C had increased risk of occurrence over time for pneumonia/respiratory failure and autonomic dysreflexia (AD). Hypertension and cardiac disease were more likely in those with C1-4 A, B, C and T1-S5 A, B, C impairments. AD and spasticity decreased in the T1-S5 groups over time, and those with ASIA impairment scale D noted decreasing ORs of UTI, pressure ulcers, urolithiasis, spasticity, and bowel accident occurrences over time (230,231). Adults with pediatric-onset SCI may experience more medical complications over a lifetime than those with adult-onset SCI; the cumulative effect of this is not well understood.

LIFE SATISFACTION. Adults with childhood-onset SCI show relatively high satisfaction with life and relate this to independent living, education, income, satisfaction with employment, and social/recreation opportunities (232,233). Medical complications adversely affect satisfaction, especially the presence of pressure ulcers, severe UTIs, and spasticity (233,234). Those with paraplegia are more satisfied than those with tetraplegia, and there appears to be no gender difference (233). Depression symptoms have been reported in adults with childhood-onset SCI, and are associated with medical complications, social participation, and incomplete injury (235). Life satisfaction is not associated with level of injury, age at injury, or years with disability (233), and QOL is not altered by obesity (207). QOL scores, unlike life satisfaction scores, are lower when compared to the general population (207).

Of interest is that adults with childhood-onset SCI self-perceptions may not be as significantly altered as clinicians anticipate (236,237) and likely due to enrichment by services and providers that emphasize education, employment, and long-term health management (233).

UROLOGY/NEPHROLOGY. The most common reported health complication for adults with childhood-onset SCI was UTI (238). Typical strategies for management of neurogenic bladders are used, as previously noted, and CIC continues to be the typical management. Adults with childhood-onset SCI also frequently receive reconstructive lower tract surgeries; however, the decision factors determining best treatment options have not been determined. There are studies reviewing specific interventions

(177), but there is no information regarding long-term effectiveness of surgical options.

Adults with childhood-onset SCI have some association of urologic complications that relate to age or years with disability, and consequently, regular urologic follow-up is recommended. In a large study of adults followed at Shriners Hospital for Children in Chicago, Vogel and colleagues report older age at interview and longer years with disability were associated with orchitis or epididymitis (238). Also, greater impairment was related to UTI, severe UTI, and renal stones. Severe UTIs were also related to poor life satisfaction (234). Although not reported in this cohort, bladder cancer and pseudotumors of the bladder may also be present. Multiple and serial interventions may be undertaken (239).

MUSCULOSKELETAL COMPLICATIONS AND CONDITIONS. For adults with childhood-onset SCI, pain at any site was the most common complaint, and shoulder pain was noted in almost half of the respondents in interviews, as reported by Vogel et al. (240). As was noted earlier, overuse syndromes must be considered, especially at the shoulder. In general, for adults with childhood-onset SCI, longer years with disability and increasing age are associated with shoulder pain (240). Etiology must be identified, and evaluation and treatment are essential. An outpatient physical therapy program and/or a home exercise program for shoulder pain (with or without impingement) (241) have been shown to be effective in pain management for adults with SCI.

For adults with childhood-onset SCI, younger age at injury and longer years with disability have a correlation with scoliosis (240,242). More severe and frequent scoliosis has been reported in paraplegia and complete lesions, and lordosis has been noted to be greater in paraplegia and incomplete lesions (242). There is no evidence that bony injury at the time of childhood-onset SCI influences the development of scoliosis or lordosis (243).

For adults with childhood-onset SCI, younger age at injury and longer years with disability were associated with hip subluxation, and older age at injury was associated with elbow and ankle pain (240). Back pain may be seen in about 20% of patients unrelated to scoliosis, and ankle pain and elbow contractures are associated with tetraplegia, and hip contractures with paraplegia (240). For those who walk, the presence of hip or knee pain should be questioned, and for any pain complaint, appropriate workup and management should ensue. Shoulder pain is the most common musculoskeletal pain complaint across all ASIA levels and impairments, and elbow and wrist pain is more commonly seen in those with paraplegia (230). There is increasing odds that shoulder pain will occur over time in all groups, and wrist and elbow pain will increase in those with paraplegia over time (231). Preservation of upper limb function, management of pain, and recognition of biomechanical risks from wheelchair propulsion and transfer techniques are of utmost importance.

There are few reports detailing osteoporosis in adults with childhood-onset SCI and none that compare to SB or adult-onset SCI; however, there are studies that identify osteoporosis as a common secondary condition in SCI and reports of high prevalence in children and adolescents with childhood-onset SCI. Treatments studied have included bisphosphonates and functional electrical stimulation (FES) exercise, and the most effective treatment has not been established. Case series have advocated for the use of cycling with FES to improve bone mineral density, with mixed results (244,245). Fractures are usually the complication of osteoporosis. Risk factors for fractures include longer years with disability, age less than 16 years at the time of injury, increasing age, prior history of fractures, BMI less than 19, increased alcohol consumption, and family history (241,245).

NEUROLOGIC COMPLICATIONS AND CONDITIONS. Neurologic sequelae for adults with childhood-onset SCI appear to be limited, as seen from reports in the literature. The presence of AD is not related to increasing age, age at injury, or years with a disability.

AD is associated with greater neurologic impairment and is a common health condition for adults with childhood-onset SCI (238). Spasticity is a problem for more than 50% of those with childhood-onset SCI; older age at injury, years since injury and additional neurologic changes all are associated with increased spasticity (241). Monitoring for changes in function and adjustment to spasticity or other forms of management must be part of routine medical care, with consideration for all possible options, including injections, pain management, medications, and surgical considerations.

ADDITIONAL MEDICAL CONDITIONS. Pulmonary conditions may be seen in adults with childhood-onset SCI. Restrictive lung disease occurs as a consequence of scoliosis, and the addition of weakness or paralysis of secondary respiratory muscles may further increase risk for recurrent respiratory infections (229). Survival for childhood-onset SCI requiring ventilator support has improved in recent years, with reported survival up to 23 years (246). Deaths in this cohort were related to respiratory complications, followed by unknown causes and suicide. There have been rare unscheduled hospitalizations, and life satisfaction is associated with better mental health.

Obesity is a reported medical condition in motor disabilities in general, and is mentioned in a recent series of adults with childhood-onset SCI. Metabolic syndrome is the most significant complication, both for individuals with pediatric and adult-onset SCI. Nelson and colleagues report that adolescents with SCD had a high prevalence of metabolic syndrome, and in particular those with childhood-onset SCI had higher frequency; 100% of the subjects who were obese were diagnosed with metabolic syndrome and 10% of those not obese also received the

diagnosis (209). The increased risks of CVD and diabetes mellitus cannot be overstated, and this syndrome must be at a high index of suspicion for health care providers. Appropriate nutrition and adequate exercise and activity should be a lifelong goal for persons with disability.

Pressure ulcers were reported in just less than 50% of adults with childhood-onset SCI, were more common in men, and more common in greater neurologic impairment (238). Well-fitting wheelchairs, seating, appliances, and orthotics; routine monitoring; regular education; and attaining continence all contribute to successful prevention.

GI conditions are not common, other than neurogenic bowel-related issues. Bowel incontinence is reported in more than 50% of adults with childhood-onset SCI, and is seen with older age and greater impairment, although not with increasing years with disability.

Latex sensitization/allergy is seen in SCI, but seemingly not as frequently as SB. It is unclear what the incidence of latex allergy is in the childhood-onset SCI population, although it is known that women more commonly report a latex allergy (238).

SEXUAL FUNCTIONING. There are less data about men and women with childhood-onset SCI. Although the general information available about adults with SCI can be helpful, it is not clear if it can be generalized. It is known that semen quality decreases at about 2 weeks postinjury, which could imply decreased fertility for adult men with childhood-onset SCI (247). Fertility is also affected by bladder care (248).

There are no menstrual cycle difficulties known for women with childhood-onset SCI (249). A multicenter study of women's self-reported reproductive health after SCI, likely adult-onset injuries, reported complications from pregnancy, labor, and delivery to be more frequent than what was noted preinjury, and babies of low birth weight (250). Women reported increased bladder spasms, muscle spasms, and autonomic symptoms at some time during their menstrual cycle. Experience of orgasms and methods of contraception varied. The effects of menopause are unknown.

There is no specific information about typical gynecologic screening and prevention practices for women with childhood-onset SCI; however, national data concerning women with mobility impairments, especially those requiring use of a wheelchair, clearly demonstrate minimal participation likely due to environmental and attitudinal barriers. Risks for use of contraception options are not known; however, combined hormone oral therapy carries a risk for thrombophlebitis; progestin-only medications have early irregular bleeding and long-term suppression effects; and IUDs with lack of sensation require vigilance for correct placement and risk of rare complications such as perforation, infection, or ectopic pregnancy (226). Given the information self-reported by women with SCI, pregnant women should be at least evaluated through a high-risk pregnancy service.

LIMB DEFICIENCY

Pediatric-onset limb deficiency is not uncommon, with 4/10,000 in upper extremity congenital limb deficiency alone. In addition, lower extremity hemimelia, traumatic amputations, and childhood cancers are associated with pediatric limb deficiency. Very little is known about aging with this disability. However, certain comorbidities and secondary conditions are typical for this group (see Table 21.5). Weight control is important to prevent osteoarthritis (251,252). One author describes increased velocity and lower effort in elderly amputees if a locked knee is used (253). Changes in gait or use of upper limb prostheses with aging in this population may be due to a variety of typical disorders of aging, including arthritis, sensory deficits, muscle weakness, or heart disease. Typical surveillance for these disorders is important to maintain ambulation status. Querying for pain may reveal treatable musculoskeletal issues.

INTELLECTUAL DISABILITIES

ID is a common reason for disability in childhood, although less prominent in adult surveillance. People with ID experience age-related health impairments at a higher rate and earlier age than people without disability (254). Depending on the etiology of their disability, they may be at much higher risk for both secondary conditions and comorbidities. These conditions can be life-threatening or life-altering. Some may be prevented or treated if identified early. DS will be discussed as a separate entity, as more is known about aging with this condition. Strategies for minimizing functional limitations will be highlighted. Rehabilitation surveillance and treatments will be discussed (see Tables 21.6 and 21.7).

Intellectual Disability

Individuals with ID are living longer and experiencing most of the same illnesses as the general population (255). Their life expectancy remains somewhat less than the general population, but has steadily increased with the move away from institutionalized care (256). Community-based health care for people with ID is not well organized, and people with ID experience poorer health than the general population (257). Difficulties with communication may make routine visits less than comprehensive. Tools to guide practitioners in best practice are now available (43).

CARDIOVASCULAR COMPLICATIONS AND CONDITIONS. Janicki and colleagues noted that CVD and respiratory diseases were more common causes of death in older adults with ID than in the general population, with cancers in

TABLE 21.5 LIMB DEFICIENCY: COMMON AGING HEALTH AND PERFORMANCE CHANGES

COMMON RELATED HEALTH CONDITIONS	PREVENTION STRATEGIES	TREATMENT STRATEGIES
Overweight or obesity	Monitor weight and nutrition Routine exercise	Exercise or therapy prescription Referral to nutritionist if indicated Modify prosthesis as needed
Pain	Routine exercise Monitor and query routinely Work simplification Ergonomic evaluations Energy conservation	Focal examination and evaluate/treat Exercise prescription Modify equipment or workplace Progress to pain management program Adjust prosthesis as needed
Deconditioning Falls	Education and falls prevention Routine exercise	Therapy prescription—focus on strength and aerobics Adjust prosthesis as needed Consider other equipment
CVD/PVD	Reduce risks Routine exercise Monitor as indicated	Referral and management as indicated
Health maintenance	Monitoring—see Table 21.9	

Abbreviations: CVD, cardiovascular disease; PVD, peripheral vascular disease.

a less prominent role (255). Although there have been discussions of significant rates of chronic health conditions and general poor health for adults with DD, more recent studies of adults receiving state or national support in New York state, Taiwan, and Israel (113,114,258) note gradual increases in health conditions, but not with higher incidence than in the general population, and in some cases lower.

OBESITY/UNDERWEIGHT. In a cross-disability study of a South Carolina primary care practice that included almost 50% adults with DD, there was a lower OR for coronary artery disease, cancer, and obesity for adults with DD in comparison to those without disability and compared to other disability groups (259). Although obesity was reported as low in the South Carolina study, other studies report obesity as being more common in adults with DD. Bhaumik et al studied 1,119 adults with ID and found that women were 1.5 times as likely to obese and twice as likely to be underweight; men with ID were eight times as likely to be underweight (260). In this study obesity was associated with self-feeding, living independently, and hypertension; underweight was associated with younger age, absence of DS, and no medication. Obesity in people with ID is higher, compared to those age-matched without ID (35.4% vs. 20.6% in one survey) (261). Other researchers have found twice as many people with ID to be obese as those without ID within the same community (262–264). Those with mild ID have more obesity than those with severe ID, and there can be a move out of the obesity state (181). The combination of increased obesity and mortality due to CVD leads to a recommendation of increased surveillance and prevention strategies for obesity-related disease.

RESPIRATORY SYSTEM. Several authors describe respiratory ailments as important factors in morbidity and mortality of aging adults with ID (254,255,265). Janicki and colleagues identified pneumonia as the most prevalent cause of death due to respiratory illness and second only to CVD (255). Maintaining immunizations and routine health checks may prevent some of these deaths (266). Sleep apnea due to obesity is mentioned as a comorbidity and may require separate screening or sleep studies.

HEALTH MAINTENANCE. People with ID require the same screening for cancers, diabetes, hyperlipidemia, hypertension, bone density, and ophthalmologic and hearing disorders as the general population. Communication about the results of these screenings and plans for treatment of any abnormalities may need to be through a proxy. Prevention strategies for diseases related to obesity may need to start earlier than in the general population. Pre-existing conditions of epilepsy and poor oral health should be monitored closely (266). GERD and *Helicobacter pylori* infection is increased in prevalence and undertreated in people with ID (267,268). Symptoms of GERD should be queried in people with ID and treatment undertaken, as with the general population. Osteoporosis also is more prevalent in people with ID, with precipitating factors of small size, hypogonadism, and anticonvulsant therapy (269–271). Fractures are associated with frequency of falling. Screening for

TABLE 21.6 INTELLECTUAL DISABILITY: COMMON AGING HEALTH AND PERFORMANCE CHANGES

COMMON RELATED HEALTH CONDITIONS	PREVENTION STRATEGIES	TREATMENT STRATEGIES
CVD	Routine monitoring	
Obesity/underweight	Routine exercise Monitor closely Nutrition management	Exercise prescription Referral for nutritional consultation
Respiratory disorders	Routine monitoring Immunizations	Consideration of sleep apnea, need for O_2 supplement
Epilepsy	Routine neurology appointments	Assist with change in community living arrangement as needed
Osteoporosis/fractures	Query and monitor Routine exercise Calcium/vitamin D supplement Fracture and fall prevention; education	DXA evaluation Consider treatment when multiple fractures occur Exercise when appropriate
Poor oral health	Monitoring Assist with environmental accessibility if able	
Mental health	Routine monitoring Reduce life events Query of support, living arrangements	Specialty referral as appropriate Referral for psychological and social support Use of community resources
Sexual functioning	Provide with education—appropriate modality for level of function Assist with environmental modification for routine assessments as able Ensure pregnancy high-risk needs are met	Following pregnancy, support may be needed at home
Health maintenance	Monitoring—see Table 21.9	

Abbreviations: CVD, cardiovascular disease; DXA, dual energy x-ray absorptiometry.

osteoporosis and falling should commence during early adulthood, with follow-up depending on the results.

MENTAL HEALTH. Mental health impairments are prevalent in elderly people with ID. Estimates vary from 20% to 70%, depending on which assessments were used and the exact population studied (272–275). Dementia, depression, and general psychiatric symptoms are all more prevalent in the elderly population with ID. Each of these groups also had high numbers of health comorbidities, such as CVD, sensory impairment, and mobility problems. Researchers note that life events, such as relocation, were more frequent in adults with ID than in comparison groups (274). Medication review is a priority for clinicians treating people with ID. Polypharmacy is a significant problem for people who may not have adequate understanding of the need to report side effects or efficacy of medications. Medications should not be prescribed unless a system is in place to ensure compliance, safety, and monitoring of efficacy (266). Surveillance for mental health problems in aging people with ID should be a priority. Dementia-like symptoms occur at an earlier age than with the general population, and depression-like symptoms may be difficult to define. Comorbidities may contribute to or appear as mental health concerns, so early recognition is imperative.

SEXUAL FUNCTIONING. People with ID are often not afforded typical education, contraception options, or sexual health screening. They face a high risk of sexual abuse, are unaware of protection from sexually transmitted diseases, and are generally unsupported in attaining healthy sexual relationships (226,276,277). Women are often prescribed suppression therapy (277,278). Sterilization for women with ID is more common abroad, and related to severity and living arrangement (279). Women and men with ID can be provided with education and support for sexual functioning, and regular health screenings can be accomplished with modifications and support (226).

Down Syndrome

More than half of the people with DS will survive to age 50, and half of those will be alive at age 65 (280). Most people with DS are living in the community with family

TABLE 21.7 DOWN SYNDROME: COMMON AGING HEALTH AND PERFORMANCE CHANGES

COMMON RELATED HEALTH CONDITIONS	PREVENTION STRATEGIES	TREATMENT STRATEGIES
Mental health Alzheimer disease/Dementia Depression	Monitor Reduce life events such as moves	Behavior management Medications as needed Specialty referral as appropriate Referral for needed supports Use of community resources
Endocrine Hypo- or hyperthyroid Diabetes	Routine monitoring Annual TSH monitoring	Medication management Diet management
CVD Mitral valve prolapse	Reduce vascular risk Routine exercise	Evaluation, treatment per study
Celiac disease	Monitor	Management by gastroenterologist
Hearing loss	Monitor	Consider amplification if appropriate
Sleep apnea	Monitor	Sleep study and management
Musculoskeletal Arthritis Osteoporosis Atlantoaxial instability	Query and monitor Calcium and Vitamin D Routine exercise Monitor for urologic change, dysphagia, spasticity, weakness, bowel changes, pain	Evaluate pain complaints appropriately Medications as appropriate Therapy prescription Consider treatment if multiple fractures occur Full evaluation of performance changes; radiographs and referral as appropriate Referral to neurosurgery with acute loss
Obesity	Routine exercise Nutrition management	Referral for exercise program Referral for diet management
Respiratory infections	Monitor Immunizations	Medications, possible O_2 supplement
Sexual functioning	Provide education—appropriate modality for level of function Assist with environmental modification for routine assessments as able	
Health maintenance	Monitoring—see Table 21.9	

Abbreviations: CVD, cardiovascular disease; TSH, Thyroid stimulating Hormone.

or in supported living. They require increased health care surveillance as they age due to higher prevalence of numerous clinical conditions. Access to appropriate health care may prove difficult for people with DS, as they may have difficulty with communication or behavior and typical primary care practices may not meet their needs. Specific health screening programs have shown a dramatic increase in recognition of unmet health care needs (42). Rehabilitation clinicians can assist families and primary care physicians to provide optimal maintenance of function throughout life.

MENTAL HEALTH. Mental health problems in people with DS have been well described in the literature. An elderly (>65) group was well described by Cooper and colleagues as having increased dementia, anxiety, and depression when compared to a younger group (273,281). Symptoms of Alzheimer disease may be seen as early as age 35 and will be noted in 75% of people with DS by age 60 (191). A variety of causes have been postulated for the high incidence of Alzheimer disease/dementia in people with DS, including antioxidant stress (282) lower bioavailable estradiol in women (283), and decreased alpha and beta secretase activity (284).

Treatable comorbidities, which may look like Alzheimer disease, must be ruled out. These include hypothyroidism, visual and hearing impairments, depression, and epilepsy, all of which are significantly more common in DS than in other populations with Alzheimer disease (285). Likewise, systemic illness, infection, drug effects, and alcoholism must also be eliminated as possible treatable causes of Alzheimer disease symptoms (286).

Depression may cause decreased function in people with DS (259,281,286,287). Experiences of loss may trigger depression, as may changes in work or living situations. Depression may be treated with counseling; however, training or experience with this population will be needed for counseling to be effective. Treatment may also include medications. The use of selective serotonin reuptake inhibitors (SSRIs) in DS has been anecdotally described, but no randomized controlled trials have been reported to date (284,288–290).

ENDOCRINE SYSTEM. Thyroid disease is well described as a comorbidity of DS (42,254,255,272–274,280,285–287,291). Hypothyroidism is found in 15% to 50% of adults with DS (280,285,286,292–294). Thyroid-stimulating hormone levels should be assessed annually in patients with DS (286).

Diabetes mellitus may have a higher prevalence in adults with DS, but is rarely discussed in the literature (285,286,295,296). McDermott and colleagues found fewer developmentally disabled adults with diabetes than control adults in a large primary care practice (259). Typical yearly testing and treatment as needed should suffice for surveillance.

OTOLARYNGOLOGY. Hearing loss is extremely common in people with DS and may not develop until adulthood (272,280,286,297,298). Poor hearing may exacerbate pre-existing communication difficulties and present as behavior problems. Auditory testing is recommended at least every 2 years in adults with DS.

Sleep apnea is also a common problem for adults with DS (299–303). The cause is likely multifactorial, with obesity (302), central (brainstem respiratory control) mechanisms (301), and obstructive (300,302,304) sources all implicated. Sleep apnea is associated with worsened cognitive skills (305), and may be successfully treated in a variety of ways (304,306,307). A sleep study is indicated to identify the cause and therefore predict the successful treatment for sleep apnea.

MUSCULOSKELETAL COMPLICATIONS AND CONDITIONS. Premature arthritis has been reported in adolescents and adults with DS and may be associated with joint subluxations and dislocations (280,308). Hip instability may occur or worsen in adults with DS and is associated with decreased ambulation status (309). Foot pain and arthritis may be associated with severe pronation and atypical gait; however, very little research has been done in this area (310). X-rays are indicated if ambulation status deteriorates. Treatment may begin with NSAIDs, but further evaluation and possible referral are indicated if typical arthritis pain relief strategies are not sufficient to maintain function.

Osteoporosis is also more common in adults with DS and is found in both men and women at a significantly younger age than in the general population (311). Long bone and vertebral compression fractures are common (312). Decreased physical activity, short stature, early menopause, low muscle tone, and increased incidence of thyroid disease may all be factors in osteoporosis in DS (283).

ATLANTOAXIAL INSTABILITY. One to two percent of individuals with DS will have cervical subluxation or symptomatic atlantoaxial instability (AI) (313). Routine monitoring via x-ray is no longer recommended, but vigilance for progression is recommended. Related symptoms include new torticollis, weakness, neck pain, change in gait, change in bowel or bladder function, increased reflexes, or other symptoms of spinal cord compression (314). Presentation of these symptoms requires immediate stabilization and referral for surgery consideration (280,313,314). Outcomes from surgery are not always acceptable (315,316).

CARDIAC COMPLICATIONS AND CONDITIONS. Nearly half of the infants born with DS will have a structural heart anomaly. Most of the typical congenital heart abnormalities will have been corrected in infancy. Increased incidence of mitral valve prolapse in adults with DS has been reported (280,286,291). Careful auscultation should reveal any change in heart murmurs, and electrocardiogram and chest x-ray can follow.

CVD is not well studied in people with DS. As people with DS live longer, become more obese, and less active, it is reasonable to expect to see increasing rates of CVD (255,317,318). Several authors have noted decreased cardiovascular capacity in people with DS (319–321). A 2010 Cochrane review of exercise training programs for people with DS revealed three small trials of good quality. Of these, only maximal treadmill grade was improved after the training program (322).

OBESITY. Obesity is a lifelong issue for many people with DS. As many as 70% of adults with DS are reported to be obese (304,323). Health promotion and group exercise classes have been successful at significantly reducing body fat percentages in short-term programs (319,320,324–326).

SEXUAL FUNCTIONING. There is little published information regarding sexual functioning in adults with DS. It has long been held that males are infertile and females are fertile or subfertile based on histology of gonads and serum levels (327,328). There are case reports of men and reports of small series of women who have been fertile (329,330). The male offspring are reported to have no abnormalities, congenital or genetic. In contrast, the female offspring are reported to have DS, be chromosomally normal, or have other congenital defects or ID. The need for education and counseling, monitoring for sexual abuse, and social support is obvious.

HEALTH MAINTENANCE. People with DS require the usual screenings for testicular and cervical or breast cancer

and hypertension. Celiac disease is now recognized as a common condition associated with DS, and monitoring should be a part of health maintenance (331,332). Dental health is important, as gingivitis and periodontal disease are more common in people with DS (333–336). Cataracts and keratoconus both occur with increased frequency in people with DS. Regular ophthalmologic examinations are indicated to evaluate for these conditions. Health care screening and promotion programs have demonstrated improved detection of symptoms and compliance with health recommendations (254,285,304,324,325,337). Sexual health should not be ignored, and often contraception or suppression is prescribed for women for hygiene problems with menstrual cycles (226,278).

Williams Syndrome

Williams syndrome (WS) is caused by a gene deletion on chromosome 7. It is rare, occurring in 1 of 20,000 live births (338). Devenny and colleagues have been following a group of 15 adults with WS, some of whom have participated in a 15-year longitudinal study on aging in adults with ID. The participants with WS demonstrated early and rapid decline in long-term episodic memory not found in other adults with ID. Verbal short-term memory was better than their peers with ID and did not decline with age (338,339). No association was found with physical or mental comorbidities. Chernisky and colleagues evaluated 20 adults (ages 30–52) with WS on a variety of physical and psychological parameters. They found significant health concerns including: abnormal body habitus ranging from underweight to obese; mild–moderate high frequency sensorineural hearing loss; CVD (13 had supravalvar aortic stenosis); hypertension; GI symptoms including diverticular disease; diabetes and abnormal glucose tolerance on standard oral glucose tolerance testing; subclinical hypothyroidism; 12/20 met the criteria for osteopenia on DXA scanning; and a high frequency of both acute and chronic psychiatric symptoms. The most frequent diagnosis was anxiety but several subjects met criteria for several diagnoses (340). Because WS has only been clearly described within the current generation of adults, few people have been extensively studied, and we do not yet know the causes of the apparent precocious aging noted in this population. Clinicians managing people with WS may find extensive guidance on management in the appendix attached to a recent summary by Pober on this disorder (341).

CHILDHOOD CANCER AND ADULT SURVIVORS

The majority of children diagnosed with cancer now survive well into adulthood. Late effects of treatment appear to mimic aging as they include all body systems. Therefore childhood cancer survivors require surveillance to identify expected health concerns at an earlier age than would otherwise be expected (342), and some of their health issues may require the assistance of a physiatrist and rehabilitation services. Rural cancer survivors are at particular risk for poor outcomes (343). Many countries have established registries to allow this long-term follow-up. The Childhood Cancer Survivor Study (CCSS) (www.cancer.gov/cancertopics/coping/ccss) involves more than 20 U.S. and Canadian centers, and monitors survivors (and siblings often used as comparisons) originally diagnosed more than 30 years ago by cohorts.

Less than 20% of adult survivors of childhood cancer are followed at a specialty follow-up clinic, so it is critical that all medical practitioners be familiar with risks to their health (344). Key additional health issues include: subsequent primary cancers (greater than 50% risk), infertility with increased miscarriage rate but no known increase in risk to infants, cardiac risk, osteopenia, metabolic syndrome without obesity, cognitive and psychosocial issues, poor growth in adolescence, and thyroid dysfunction (344,345). In addition, risk of serious infection is increased for all childhood cancer survivors and particular in those who received total body radiation (346).

The Children's Oncology Group has created an exhaustive, research-based guideline for adult follow-up of these patients (347). This group recommends that each survivor have a summary of dates and treatments used to treat the original cancer. This is then used to predict the most likely concerns and follow-up needed on an individual basis. This reference also includes specific guidance for following practitioners relative to potential late effects of each type of cancer treatment. Written materials for patients on typical concerns are easily accessed electronically (www.survivorshipguidelines.org).

A large review of survivors and siblings reported a very high rate of severe chronic health conditions in survivors. The treatment most associated with risk of severe or life-threatening conditions is core radiation. In general, women survivors had worse health status. The need for major joint replacements was the most common severe health condition reported (348).

Table 21.8 describes some common late effects, associated treatments, and screening/management methods. This should be considered an introduction to this topic; to develop a comprehensive algorithm for surveillance for an individual patient, the method described previously from the Children's Oncology Group should be used.

Childhood-Onset Brain Cancer

Adult survivors of childhood brain cancer are likely the most common cancer patients seen through a Physical Medicine & Rehabilitation Residency (PM&R) program. Survivors of childhood CNS cancers have a high risk for late mortality > 5 years, and are noted to have late effects or secondary conditions related to neurologic, sensory

TABLE 21.8 CHILDHOOD CANCER: COMMON LATE EFFECTS

TYPE OF CANCER	ADULT CONCERN	SURVEILLANCE NEEDED
Bone tumor	Joint deterioration	Pain inquiry/x-rays
	Congestive heart failure	Routine EKG/echocardiogram
Acute lymphatic Leukemia	Coronary artery disease	Routine EKG/echocardiogram
	Cerebrovascular accident	Activity program
	Obesity	Routine labs early, routine measurement of body fat
	Metabolic syndrome	
Hodgkin's disease	Coronary artery disease	Routine EKG/echocardiogram
	Cerebrovascular accident	
Brain tumor	Cognitive impairments	Monitor for fatigue, independence, depressive symptoms
	Poor fitness/strength	Exercise and activity program
	Obesity	Activity and nutrition program
All types	Second neoplasm	No consensus
	Infection	Immunization/suspicion
	General premature aging and fragility	Healthy habits, suspicion of chronic conditions
	Joint deterioration	Pain inquiry/x-rays
	Subclinical hypothyroidism	Screening

(especially auditory), endocrine, and musculoskeletal systems (349,350). They have a high risk for late mortality more than 5 years postdiagnosis and for developing other neoplasms, especially second primary brain tumors (349,351). There are physical performance limitations for adult survivors of childhood brain tumors as noted in a cross-sectional study (352). Survivors of medulloblastoma and osteosarcoma have the highest rates of inactivity. Inactivity is associated with cranial radiation, amputation, female gender, Black race, older age, lower educational attainment, extremes of weight, smoking, and depression (353). Ness et al noted that muscle strength and fitness values for the survivors were similar to standard and comparative values for individuals above 60 years, with limited physical performance and poorer management of home and school activities (352). Survivors of CNS tumors are at risk for obesity, inactivity, and an increasing number of chronic conditions (354). Natural target areas for rehabilitation activities would be within the realms of activity level, weight management, and performance.

Using data from the CCSS network, associations have been found among radiation dosing, site of radiation, neurocognitive issues, and social participation; radiation exposure to temporal brain regions is related to increased risk for memory and social functioning impairments and general health problems, while frontal brain exposure is associated with physical performance and general health problems (355). Cognitive function is also negatively affected by shunt placement, gender, and diagnosis at less than 2 years of age (356). Survivors of pediatric leukemia and CNS tumors are at higher risk for depression and anxiety, attention deficits, oppositional behavior, and social withdrawal, compared to other pediatric cancer diagnoses (354).

Thus, adult survivors of childhood-onset brain cancer may be at risk for limited social participation. Using data from a U.K. registry, survivors of childhood cancers are at a higher risk for lower educational attainment with cranial radiation, CNS tumor diagnosis, older at completion of the questionnaire, younger at diagnosis, having epilepsy, and being female (357). Cranial radiation has been found to be associated with higher unemployment (358**)**. Pronounced reductions in marriage/cohabitation are seen in survivors of CNS tumors (359). Adult survivors of pediatric brain tumor tended to limit their participation and interaction with the environment, which is associated with reduced HRQOL (360). In general, low physical and cognitive performance and less social participation are common for adult survivors of pediatric-onset CNS cancer.

While there has been documentation of late effects of cancer and treatments, there is limited report of successful interventions. Medications are advocated for symptoms of poor attention and emotional or social issues, but their use is often unrelated to behavior reports (354). Rehabilitation and physiatric strategies may be effective. There is an opportunity for research to determine the most effective strategies or comparisons of responsiveness to other disability groups.

TRANSITIONS AND ACCESS TO HEALTH CARE

TRANSITION OF CARE TO ADULT SERVICES

Improved medical care and increasing numbers of adults with childhood-onset disability have led to much interest and concern about the transitioning of care of young adults from a family-centered pediatric approach to a self-directed adult care model (2). Evidence suggests that the related health care delivery has not been able to keep pace with their multidimensional needs (25). For over two decades, various professional groups and initiatives have called for improvements in the process in their disciplines and internationally (361). Adults with childhood-onset disability continue to face adverse health outcomes that not only compromise their own life course transitions, but also increase the burden on the health care systems. A consensus policy statement, adopted by the American Academy of Pediatrics, American Academy of Family Physicians, and the American College of Physicians—American Society of Internal Medicine, states that the transition of care should "maximize lifelong functioning and potential through the provision of high-quality, developmentally appropriate health care services that continues uninterrupted as the individual moves from adolescence to adulthood." (362) Barriers to transitions include lack of adult provider training about health issues and disability, focus on medical issues without regard for the broader developmental aspects of transition into adulthood, poor communication between pediatric and adult providers, and the patient's need for self-direction while navigating the adult system (363,364). There have also been suggestions for specific elements to support a transition, such as preparation, flexible timing, care coordination, transition clinic visits, a network of interested adult care providers, and institutional and health care systems support and resources; however, this remains theoretic and programs vary across regions and countries (365,366,367). Transition programs strive for supportive environments built around the individual with a childhood-onset disability and his or her family, and recognize the need for a life-span approach (32). Barriers to programs include lack of insurance, lack of funding or dedicated time for professionals, patients' uncertainty about need for follow-up, and patients' choice (368). Resource centers with tools and tips are being developed for the transition process, focusing on young people with disability, their families, their health care team, and other service providers; these are easily accessed electronically (www.healthytransitionsny.org and www.gottransition.org) (32). At present, the science is at an early stage of development, and no evaluation plans to determine successes are available (363,366,369). Children and adolescents with early-onset and chronic health care needs usually receive an organized level of care, and maintaining coordination of often complex care is an important part of quality health care over a lifetime.

There have been reports of successful transition of service models. Successes related to planned and evaluated transitions (370), personal health records management (370,371), and provision of education on health and needs (372). There remain questions regarding shared responsibilities for the transition (373–375), need for protocols (376), and timing for planning and implementation. Pediatric physiatrists can often provide the stability for this transition. Table 21.3 identifies challenges for transitioning health care from pediatric to adult systems of care (377).

ACCESS TO HEALTH CARE

Access to health care for young adults has been problematic for funding reasons as well as transition-of-care difficulties (see Figure 21.2). Lack of insurance has been highlighted, and is as common among young adults without disability as those with disability, as noted through the National Health Information Survey (378). However, adults with disability had eight times greater odds of reporting unmet health care needs and six times greater odds of having no usual source of care, compared to those without disability. The majority of young adults with disability reported a gap in their insurance coverage, and many were uninsured over a 3-year period (379).

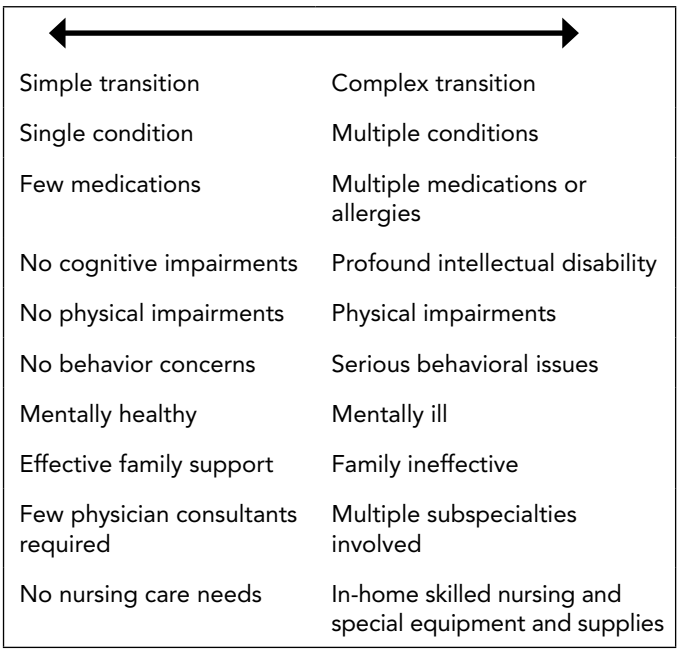

FIGURE 21.2 Characteristics that affect successful transition of care.

Source: Adapted from Ref. (377). Kelly AM, Kratz B, Bielski M, Rinehart PM. Implementing transitions for youth with complex chronic conditions using the medical home model. Pediatrics. 2002;110:1322–1327.

Access also involves environment, attitudes, and systems. Architectural barriers have been addressed through the Americans with Disabilities Act, although accessible health care providers' offices and accessible examination and procedure tables continue to be available on only a limited basis. Attitudinal barriers are more difficult to remedy, and involve both consumers and providers. To this day, there are no requirements in undergraduate and graduate medical school education to acquire disability-specific knowledge or skills, or to experience routine interactions with people with disability (22). Rehabilitation clinicians may need to ask more direct questions of their patients regarding secondary conditions and additional health concerns to better identify conditions, begin management, or discuss management with a primary care physician. Physiatrists can act as a resource for primary care providers, who likely have limited knowledge regarding persons with lifelong disability. Consumers with communication or cognitive impairments (eg, hearing impairment, speech production impairment, brain injury, ID) may need more time to communicate, require an interpreter, or require personal preparation time for the appointment in order to have their needs conveyed; modification of appointment times, with preplanning and written lists of concerns, can often be helpful. Consumers may seek help only late in the course of an acute medical condition or change because of previous difficulties managing the system. Specifically, consumers report that their routine health care providers know little about their disability and its impact on health and function (43).

HEALTH AND WELLNESS AGENDA

As a result of the steady improvement in medical care and social support systems during the past 50 years, persons with disability are healthy, conducting active and productive lives, and generally living longer. The medical paradigm must now shift from that of illness and disease to one of health and wellness. The health care delivery system must view persons with disability through a typical health maintenance and preventive medicine approach. This requires a change in attitudes and care models. Both prevention and promotion strategies should be employed: prevention of activities that lead to illness and disease (eg, smoking cessation, dietary discretion, routine laboratory and examinations, protected sexual activity) and promotion of activities that improve general well-being (eg, stress management, exercise) adapted to meet individual requirements and performance (Table 21.9) (380,381,382). However, positive health behaviors require social, health, and community resources. The more resources a person has, the more likely that individual will engage in health promotion and protective behaviors (383). Again, access is an important issue. While the Affordable Care Act has placed prevention as a high priority, it is not clear how these services will be available and modified for people with disability (384). Availability of information in appropriate modalities and the education of consumers are important. To participate in positive health behaviors, one must be interested, be ready to make changes, have the needed resources, and have a supportive environment. Early involvement of adolescents with mobility impairments in health promotion activities may pave the way for maintaining these behaviors into adulthood.

Since musculoskeletal conditions are the most common age-related changes and secondary conditions that affect performance, it would seem most reasonable to view typical physiatric strategies and interventions as preventive management techniques. Use of adaptive equipment, energy-conservation techniques, joint protection, and ergonomic positioning may enhance function, decrease musculoskeletal complaints, and possibly prevent or delay some functional changes. Personal attitudes (of the person with a mobility impairment or his or her personal support system) may have to change before a person with impaired mobility will consider such assistance or be supported in considering the value of employing supportive (less independent) techniques.

Exercise is a well-known health-promoting behavior, and its effects are positively demonstrated in persons with disability (44,385–389). Benefits of a regular exercise program include improved fitness, weight reduction, improved mood, and improved sleep. It is also known that persons must be judicious in participating in exercise programs, given the issues of fatigue and pain. Of course, care must be taken in prescribing exercise for persons with impaired mobility; they should participate in an appropriate program of exercise or activity, especially keeping in mind their risk factors for musculoskeletal injury. Jogging or running started by young adults without disability more often resulted in discontinuation of exercise because of joint pain than for persons who started a similar exercise program in their middle years, leading one to believe that long-term, high-impact exercise may result in pain. Aquatics programs can eliminate the wear and tear to joints. Adults with CP tend to report perceived changes in balance and then fear of falling, which usually improves with a general fitness program. Exercises, including strengthening exercises, are not contraindicated for persons with spasticity. Generally, adults and young adults with DD do not participate in routine fitness or exercise programs. This may be as much from limited knowledge in this area as from attitudes of care providers and persons with disability relative to exercise as a self-directed, nonmedical, or leisure activity. Exercise programs at home, in a health club, or as part of an individual recreation program (with or without modifications) must be initiated earlier than adulthood to achieve long-term participation. And, just as in the nondisabled population, priorities for persons with mobility impairment should include exercise and fitness.

TABLE 21.9 HEALTH PREVENTIVE SCREENING SERVICES

HEALTH CONDITIONS	RECOMMENDATION FOR GENERAL POPULATION	MODIFICATION NEEDED
Hypertension	>18 years and annually; screening for asymptomatic sustained BP >140 mmHg systolic or 90 mmHg diastolic	Consider earlier and more frequent check with risk for metabolic syndrome; use appropriate sphygmomanometer for limb length and circumference—automatic digital models may give spurious result, watch for spasticity trigger
Immunizations	Age and chronic condition dependent; recommendations are for those never vaccinated in childhood and those previously vaccinated, with adult schedules for: influenza, tetanus/diphtheria/pertussis booster, zoster, HPV, pneumococcal, meningococcal, hepatitis, and Hemophilus influenza	Consider immunizations; no recommendations based on disability; recommendations based on pregnancy, weakened immune system, HIV, renal disease, heart disease, diabetes, asplenia, chronic alcoholism, and chronic liver/lung disease
Cardiovascular Lipid	Men: >35 years; possibly 20 years with CAD risks Women: >45 years with CAD risks; possibly 20 years with risks	None known
Abdominal aortic aneurysm	Men: age 65–75 years if they have ever smoked	Accessible procedure environment
Cancer Men and women: colorectal	Screening ages 50–75 years; fecal occult blood testing, sigmoidoscopy, or colonoscopy used with varying risks/benefits	Level-appropriate education for all tests and procedures Accessible examination and procedure environment May need sedation to complete procedure—must consider risks/benefits before proceeding with screen and sedation May need personal assistant for procedures
Men and Women: lung	Screening adults aged 55–80 years with 30 pack-year history, who currently smoke, or have smoked within the past 15 years with CT lung; not recommended if smoking was discontinued 15 years previously	
Women: breast	Screening for positive family history of breast, ovarian, tubal, peritoneal cancer; may include genetic counseling and testing in high-risk individuals Biennial mammogram for women aged 50–74 years; earlier mammogram depends on context Insufficient data to support clinical breast exam and self-exam—not recommended Insufficient data to support use of MRI	
Women: cervical	Screening with Pap smear: ages 21–65 years every 3 years—or—for ages 30–65 years every 5 years with cytology and HPV testing; HPV testing, if positive, may prolong screening to women older than 65 years No screening for women > 65 years and if they have no risk No screening if they have had hysterectomy and no history of high-grade precancer or cervical cancer Use of HPV vaccine does not alter screening	
Men: prostate	PSA not recommended to screen for prostate cancer	
Metabolic/endocrine Obesity	Screening for all using BMI, counseling and behavior interventions offered if BMI > 30 kg/m^2	BMI does not reflect body fat, which is the measure of interest; height/weight measurement may not be accurate in disability May need modifications for activity and behavior interventions Monitor for metabolic syndrome

(*continued*)

TABLE 21.9 HEALTH PREVENTIVE SCREENING SERVICES (*CONTINUED*)

HEALTH CONDITIONS	RECOMMENDATION FOR GENERAL POPULATION	MODIFICATION NEEDED
Diabetes mellitus type 2	Screening for asymptomatic sustained BP >135/80 mmHg, with or without treatment Progress to fasting plasma glucose, 2-hour postload plasma glucose, or hemoglobin A1C	Use appropriate sphygmomanometer for limb length and circumference—automatic digital models may give spurious result, watch for spasticity trigger
Mental health Depression	Screening if able to diagnose, treat, follow-up	May require modification to queries; may need modifications in support to diagnose and treat
Dementia	Growing data to recommend screening in general population	Important to question in ID of all types at younger-than-expected ages
Violence	Screening not recommended for general population	High incidence of violence and abuse in disability; offer opportunity to discuss
Tobacco use	Recommend regular screening and offer cessation interventions	None known
Exercise	No screening recommendation for the general population; exercise guidelines available through www.cdc.gov/physicalactivity/everyone/guidelines/adults.html	Exercise is an important activity for those with motor impairments; has been shown to be effective for improved performance, pain control, weight management; important to query about exercise/activity, refer to accessible program
Aging Vision	Presbyopia, cataract, macular degeneration, and glaucoma increase with increasing age—unclear that screening is effective	Accessible examination; concern in CP, SB (related to progressive CNS pathology), ID, DS
Hearing	>50 years, hearing decreases—unclear that screening is effective	Accessible examination; concern in CP, ID, DS, brain cancer survivors

Abbreviations: BMI, body mass index; BP, blood pressure; CAD, coronary artery disease; CNS, central nervous system; HPV, human papillomavirus; PSA, prostate-specific antigen.

Source: Adapted from Refs. (380–382).

SUMMARY

Adults with early-onset disability are generally healthy. Not all adults have serious health problems, and many now recognize the aging process as a natural course of events. The most common age-related changes and secondary conditions involve physical performance and the musculoskeletal system. Prevention strategies require knowledge of expected changes, recognition of changes that alter function and require intervention, and an understanding of interventions that positively impact on function. This requires that a person with a disability have access to knowledgeable health care providers. Physiatrists may offer that knowledge through direct clinical service or indirectly functioning as a resource in the community. Environmental, communication, attitudinal, and systems barriers must be overcome in order for health care providers and people with disability to work together for the best possible outcomes.

It is time to reconsider the model of illness and disease for persons with lifelong disability. Particularly in the realm of mobility, a health and wellness model should be developed. Use of prevention strategies must be considered in childhood and adolescence to address the more frequent secondary conditions. Programs of fitness and exercise have been proven beneficial in nondisabled groups and disability groups alike. Health promotion strategies should be employed for persons with congenital and childhood-onset mobility impairments.

PEARLS

- Most adults with early-onset disability are healthy with aging. Significant or acute loss of function should not be expected, and evaluation must ensue.

- Adults with early-onset disability view themselves as healthy, although this is dependent on the number of health conditions. Life satisfaction is usually not associated with disability. This is within the context of measurement instruments that have not been standardized for those with disability.
- Urinary/renal issues for adults with childhood-onset SCD are of primary concern. However, management of pressure ulcers, obesity (with concern for metabolic syndrome), and lymphedema can be most problematic.
- Consider newer tone management options (such as botulinum toxin injections or intrathecal baclofen) to manage pain or improve function, with concomitant therapy. With decreased tone, additional focused therapy can improve function.
- Pain is common in adults with childhood-onset disability. All pain is not arthritis, and there can be many etiologies. Never miss the opportunity to question, evaluate, diagnose, and treat. Although most pain is musculoskeletal in origin, if there is no improvement, consider neurologically based etiologies, such as stenosis, tethering, or entrapments.
- Exercise can improve performance, and any person with a disability can participate, with modifications. Do not just consider therapy—home- and community-based programs can be effective.
- Adult survivors of many types of childhood cancer report premature aging issues and have significant likelihood of general infection. Adults who have survived pediatric cancer often require early joint replacement. Those who have survived brain cancers may need additional support to maintain health.
- Transition to adult health care is more successful with planned approaches and involvement of families and caregivers (when appropriate). Adults with pediatric-onset disability should receive typical preventive care, with modifications where needed.

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