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HANDBOOK OF PEDIATRIC REHABILITATION MEDICINE

ROBERT J. RINALDI
RAJASHREE SRINIVASAN

Handbook of Pediatric Rehabilitation Medicine

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FOREWORD

I am proud to provide the foreword to the *Handbook of Pediatric Rehabilitation Medicine,* spearheaded and edited by the capable leadership of Dr. Bob Rinaldi and Dr. Raji Srinivasan. Pediatric rehabilitation medicine (PRM) is one of the latest subspecialties to provide value-added care and to join the medical neighborhood for children with disabilities and their families. I have been part of the pediatrics and rehabilitation community for over 40 years and served on the American Academy of Physical Medicine and Rehabilitation ad hoc committee to establish the subspecialty of PRM 15 years ago. One of the overarching goals of the subspecialty was to establish a curriculum and collate a body of knowledge that pertains to the best practice, care, and management of problems related to disability in childhood. The process of defining the body of knowledge serves to improve care and to educate the future leaders in pediatric rehabilitation.

Those of us fortunate enough to choose PRM as an area of practice know that many disciplines touch the lives of and are important in maximizing the health and function of children with disabilities. PRM at its core is relationship-based with the child, parent, and the many professionals caring for the child. These relationships are highlighted in this excellent guidebook, which stresses the overlap and importance of all medical, therapeutic, and educational professionals. The *Handbook of Pediatric Rehabilitation Medicine* serves to anchor the concepts that reflect the body of knowledge in PRM and those that are essential in managing the care of children with disabilities. The content of the handbook will be helpful regardless of discipline—for the novice to established team members, including nurses, therapists, general pediatricians, and those working in the subspecialties of pediatrics.

The handbook provides essential clinical information in a concise, organized, easy-to-follow sequence for working through complex conditions frequently encountered in practice. This book positions itself between textbooks, which are characteristically in-depth and require study, and care pathways or clinical guidelines, which are formulated and designed for specific conditions and interventions. The handbook should be a first pass of necessary information that can be accessed and used in the time frame of a clinical practice.

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I predict and hope that the handbook will stay in your actual or digital pocket to be your go-to knowledge hub for teaching and clinical problem-solving for years to come.

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PREFACE

The concept behind the creation of this first edition of the *Handbook of Pediatric Rehabilitation Medicine* stemmed from our consideration of the need to provide information about key concepts in pediatric rehabilitation more effectively to students, residents, and fellows on pediatric rehabilitation rotations. As the subspecialty of pediatric rehabilitation medicine has grown in scope, so has the need to expand our knowledge base—not only within the subspecialty, but also to other subspecialties and clinicians who many times are caring for the same children that we are. Subsequently, the idea behind providing a clinical resource to trainees grew to encompass other specialties that are involved in taking care of children with complex medical needs. This handbook has been created with this focused, yet broader call to convey key clinical information to all clinicians who provide care to children with complex medical and rehabilitation needs.

The handbook has been uniquely formatted and written with the goal of providing easily accessible key information that is portable. Thus, it is provided in a compact print version as well as an online version. The latest information has been organized efficiently in each chapter through the combined use of text and bullet points to allow for rapid access in clinics and inpatient units, or to refer to while studying for boards. Information is also provided in bite-sized portions that are easier to swallow and digest. The chapters are grouped for easy reference into growth and development, central nervous system disorders, neuromuscular disorders, musculoskeletal, general, and miscellaneous sections. Finally, our collaborating authors bring years of experience across numerous subspecialties and disciplines, emphasizing the collaborative and relationship-driven nature of the care we provide in pediatric rehabilitation medicine.

The handbook is intended for medical students; trainees in rehabilitation medicine, neurology, orthopedics, and pediatrics; practicing pediatricians, pediatric subspecialists, and primary care physicians; allied health professionals, including physical, occupational, and speech therapists; and advanced practice providers and nurse practitioners. As noted, this is reflected in the varied backgrounds of the contributing authors, the nature of the information provided, and the overall scope of the included chapters.

We wish to thank our many contributing authors who have lent their considerable talent, expertise, and time to making this handbook come to life. Their input and effort are truly appreciated by all involved in this project. We also want to thank Beth Barry, Hannah Greco, Jaclyn Shultz, and the

entire editorial staff at Springer Publishing whose efforts, guidance, and patience were instrumental in making the handbook a success. Finally, pediatric rehabilitation medicine is an inspiring field where one sees catastrophic, life-altering conditions and the resilience of children who bounce back. This book is dedicated to all those wonderful children who we are lucky to care for. We hope you enjoy the book and learn from it.

Robert J. Rinaldi
Rajashree Srinivasan

PART I

Normal Growth and Development

Pediatric Development

1

VIRGINIA SIMSON NELSON

GENERAL PRINCIPLES

Growth refers to the increase in size and dimension of the body from birth to physical maturity. It occurs at different rates and is usually fastest during infancy and preadolescence. During growth, the body proportions change, with different parts and organ systems growing at different rates. Health, food intake, and abnormal conditions may affect growth, and it is important to understand growth of typically developing children before assessing growth of children with various conditions.

All children follow a set developmental pattern, from being totally dependent to becoming fully functioning. This pattern may vary in timing and completeness, but rarely in order. It is necessary that the examiner knows the patterns and timing of development of typically developing children before evaluating a child with a known or suspected condition that might affect development.

There are various scales available for assessment of growth and development of children. Some are specifically applicable to children from a specific geographic location or ethnicity (e.g., growth chart for children from a given country), while others apply to children with a given condition (e.g., Gross Motor Function Measure for children with cerebral palsy). There are many others used widely for children.

GROWTH

Growth should be measured at each clinic visit and plotted on a standardized growth chart. This should include weight and length/height and head circumference up to the age of 2. There are numerous growth scales available: length/height-for-age, weight-for-age, weight-for-length, weight-for-height and body mass index-for-age, and head circumference. Currently, the most commonly used scale is the WHO Child Growth Standards.[1,2] There are also growth charts for some conditions that affect children, as well as a training module published by the Maternal and Child Health Bureau which details how various specialized growth charts are used.[3] Examples of growth charts for specific conditions include those for children with Down syndrome,[4] cerebral palsy,[5] and other conditions that are available in the training module.[3] Other specialized growth charts can be found on the

Centers for Disease Control and Prevention website. It is important to understand the typical growth patterns of children with specific conditions in order to follow their growth and to plan for changes needed in orthoses, wheelchairs, and other adaptive equipment.

DEVELOPMENT

Children develop in a number of different ways and each can be evaluated using commonly available scales. It is helpful to know the timeline of a typically developing child before evaluating a child with a known or suspected condition that may alter development.

Reflexes

Infants progress from having primitive reflexes to integrating them as their gross motor skills develop. Evaluation of infants should include determining the presence or absence of at least the following reflexes: Moro, rooting, asymmetric tonic neck reflex (ATNR), palmar and plantar grasp, stepping, and placing. When present, these reflexes should never be obligate. Older infants should be evaluated for protective reactions when sitting. Older children with brain abnormalities, such as those seen in cerebral palsy, may demonstrate one or more of these reflexes (Table 1.1):

- *Moro:* When the head is lowered below the plane of the body, the arms abduct and extend, and then flex; the baby may cry.
- *Startle:* A loud noise leads to the same response as Moro.
- *Rooting:* The baby turns the head toward anything that touches their cheek or lips.
- *ATNR:* When the head is turned (passively or actively) to one side, the extremities on that side extend and on the opposite side flex.
- *Palmar grasp:* The fingers flex and grasp with stroking of the palm.
- *Plantar grasp:* The toes flex with stimulation of the distal sole of the foot.
- *Stepping:* When the child is supported upright, the child "takes steps."
- *Placing:* Dorsal foot stimulation leads to flexion of the hip and knee.

Protective reflexes develop in the mid to late first year of life and are necessary for an infant to assume an upright posture. These initially develop when sitting and later when standing.

Gross Motor

Motor development progresses in a stepwise manner, with speed of development varying slightly in typically developing children (Table 1.2). Children with varying conditions, such as cerebral palsy or spina bifida, may have slower development and/or may not achieve all motor milestones.

Flexor tone of the arms and legs predominates in neonates and movements are typically symmetric. The neonate can turn their head side to

Table 1.1 Reflexes

REFLEX	PRESENT	DISAPPEARS
Moro	At birth	At 6 mo
Rooting	At birth	At 3–4 mo
Startle	At birth	At 4–6 mo
Stepping	At birth	At 3–5 mo
Placing	At birth	At 12 mo
ATNR	At birth	At 4–6 mo
Palmar grasp	At birth	At 5–6 mo
Plantar grasp	At birth	At 12–14 mo
PROTECTIVE REFLEXES (all continue after appearing)		
	APPEARS	
Forward/parachute	At 5–7 mo	
Lateral	At 6–8 mo	
Posterior	At 7–8 mo	

ATNR, asymmetric tonic neck reflex.

Source: Data from Nelson MR. *Normal Development in Nelson MR, Pediatrics (Rehabilitation Medicine Quick Reference.* 1st ed. Demos Medical Publishing, 2011: 2–3; Zafeiriou DI. Primitive reflexes and postural reactions in the neurodevelopmental examination. *Pediatr Neurol.* 2004;31(1):1–8. doi: 10.1016/j.pediatrneurol.2004.01.012.

Table 1.2 Gross Motor Development

AGE	ACTIVITY
Newborn	Flexor tone, turns the head side to side
4 mo	Supports self on elbows
6 mo	Rolls
10 mo	Sits unsupported, creeps, pulls to stand, cruises
12–14 mo	Walks unsupported
2 y	Walks upstairs, runs
3 y	Pedals tricycle
4 y	Walks down the stairs alternating their feet
5 y	Skips, tandem-walks

Source: Data from Nelson MR. *Normal Development in Nelson MR, Pediatrics (Rehabilitation Medicine Quick Reference.* 1st ed. Demos Medical Publishing, 2011: 2–3; Molnar GE, Sobus KM. Growth and development. In: Molnar GE, Alexander MA, eds., *Pediatric Rehabilitation*, 3rd ed., Hanley & Belfus, 1999.

side in both supine and prone positions and gradually develops head control in supported sitting and then lifts their head up when placed prone. At about 3 to 4 months, the baby can hold their head up about 90° and keep it up when placed in prone on elbows. By 7 months, most babies can maintain a sitting position without support if the center of gravity is not disturbed. As protective reactions develop, the infant can maintain a sitting position when the center of gravity is disturbed. By about 10 months, infants can creep on their hands and knees, pull to a stand, and cruise holding on to a stable surface. Children typically walk by 12 to 14 months, although the normal is up to 18 months. Early walking is characterized by the hands held above the head (high guard position), with the hands/arms gradually lowered as the child gains more balance.

Fine Motor

Neonates have generalized movements which are typically symmetrical. The earliest purposeful fine motor movement is reaching for objects with two hands, which occurs between 2 and 4 months. Gradually, the infant gains more control of their arms and hands, with purposeful movements replacing random movements. By the end of the first year, the child has a pincer grasp and can finger-feed (Table 1.3).

Table 1.3 Fine Motor Development

AGE	ACTIVITY
2–3 mo	Reaches for objects with two hands
3 mo	Hands to midline, reaches with one hand
6 mo	Transfers, releases object
9 mo	Immature pincer grasp, raking
12 mo	Mature pincer grasp
15 mo	Builds tower of two blocks
18 mo	Builds tower of three blocks, turns pages in group, scribbles
2 y	Builds tower of six blocks, turns a single page, makes vertical and circular strokes, strings large beads
3 y	Builds tower of nine blocks, draws a circle
4 y	Draws a cross, attempts to write letters, establishes handedness

Source: Data from Nelson MR. *Normal Development in Nelson MR, Pediatrics (Rehabilitation Medicine Quick Reference.* 1st ed. Demos Medical Publishing, 2011: 2–3; Molnar GE, Sobus KM. Growth and development. In: Molnar GE, Alexander MA, eds., *Pediatric Rehabilitation*, 3rd ed., Hanley & Belfus, 1999.

Personal/Social

As the infant develops gross and fine motor skills, they also develop social interactions. The earliest social interaction is regarding faces in the first couple of months. This is followed by smiling, laughing, and interacting with the parent. Between 6 and 8 months, most children recognize strangers and develop stranger anxiety. Finger feeding occurs between 9 and 12 months (assuming the child is given a chance to do this), followed by feeding with a spoon by 12 to 15 months (Table 1.4).

Speech and Language Development

During the first year of life, the infant goes from crying to communicate their needs, to babbling, to pointing, to saying sounds and some words. By 1 year, most can speak at least a few meaningful words. By the age of 2, children can speak many words and two- to three-word phrases and can follow simple commands. By the age of 3, speech should be understandable to strangers most of the time (Table 1.5).

Table 1.4 Personal/Social and Cognitive Development

AGE	PERSONAL/SOCIAL	COGNITIVE
0–2 mo	Regards faces	Reflex stage
3–4 mo	Smiles, laughs	
6–8 mo	Recognizes familiar faces, develops stranger anxiety	
8–10 mo	Finger-feeds, plays peekaboo	Object permanence
12 mo	Feeds with spoon, removes garment	
18 mo	Imitates activities	Cause and effect

Source: Data from Nelson MR. *Normal Development in Nelson MR, Pediatrics (Rehabilitation Medicine Quick Reference*. 1st ed. Demos Medical Publishing, 2011: 2–3; Molnar GE, Sobus KM. Growth and development. In: Molnar GE, Alexander MA, eds., *Pediatric Rehabilitation*, 3rd ed., Hanley & Belfus, 1999.

Table 1.5 Speech and Language Development

AGE	SPEECH AND LANGUAGE
Neonate	Cries
3 mo	Laughs, coos, turns to sound
4–6 mo	Babbles
10 mo	Sounds for attention, waves bye-bye
12–14 mo	Speaks single words, understands names and simple commands
18 mo	Points to named body parts, says "no"
2 y	Speaks many words, speaks two- to three-word phrases, follows simple directions, parallel plays
3 y	Speaks three- to four-word sentences, understandable to a stranger, identifies as boy/girl, recognizes two to three colors
4 y	Follows two-step commands, uses pronouns, asks questions

Source: Data from Nelson MR. *Normal Development in Nelson MR, Pediatrics (Rehabilitation Medicine Quick Reference*, 1st Edition, New York, Demos Medical Publishing, 2011:, pp. 2–-3; Molnar GE, Sobus KM. Growth and development, In: Molnar GE, Alexander MA, (eds.), *Pediatric Rehabilitation.*, 3rd ed., Philadelphia, Hanley & Belfus, 1999; Berkman ND, Wallace I, Watson L, Coyne-Beasley T, Cullen K, Wood C, Lohr KN et al. *Screening for Speech and Language Delays and Disorders in Children Age 5 Years or Younger: A Systematic Review for the U.S. Preventive Services Task Force, Evidence Syntheses, No. 120.* Rockville (MD): Agency for Healthcare Research and Quality (US); 2015 Jul. Report No.: 13-05197-EF-1.

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PART II

Central Nervous System Disorders

AL Grawany

Cerebral Palsy

2

CAITLIN CHICOINE, LAUREN DESMARAIS, AMY KANALLAKAN, and JILDA VARGUS-ADAMS

GENERAL PRINCIPLES

Definition[1]

Cerebral palsy (CP) has most recently been defined as "a group of permanent disorders of the development of movement and posture, causing activity limitation, that are attributed to non-progressive disturbances that occurred in the developing fetal or infant brain. The motor disorders of cerebral palsy are often accompanied by disturbances of sensation, perception, cognition, communication, and behavior, by epilepsy, and by secondary musculoskeletal problems."

Epidemiology[2]

- The most common motor disability of childhood
- Overall prevalence: 2.11 per 1,000 live births
- Highest prevalence in children weighing less than 1,500 g at birth

Classification

CP can be classified according to topography, type of movement disorder, and functional level.

Topography

- Unilateral
  - Hemiplegia, or involvement of one side of the body, typically with upper greater than lower extremity involvement (38%)
- Bilateral
  - Diplegia, with involvement of the lower more than upper extremities (37%)
  - Quadriplegia, with involvement of the trunk and all four extremities (24%)

Movement Disorder[3]

- Spasticity (85%–91%)
- Dyskinesia, including dystonia and athetosis (4%–7%), with dystonia commonly occurring in conjunction with spasticity

- Ataxia (4%–6%)
- Hypotonia (2%)

DIAGNOSIS

Risk Factors[8]

The etiology of CP is not always clear. A number of risk factors have been identified in the prenatal, perinatal, and postnatal periods, as listed in the following. The greatest risk factor for the development of CP is prematurity. Although the risk is highest in premature infants, most children with CP were not born prematurely.

Prenatal

- Maternal disease, such as type 1 and 2 diabetes, lupus, Crohn disease, rheumatoid arthritis, and multiple sclerosis[9]
- Maternal coagulopathy
- Assisted reproductive technology
- Intrauterine growth restriction
- Multiple gestation
- Stroke
- Infections, such as chorioamnionitis
- Congenital brain anomalies

Perinatal

- Prematurity
- Intrapartum hypoxic-ischemic event
- Neonatal encephalopathy
- Low birth weight
- Kernicterus

Postnatal

- Stroke
- Accidental or nonaccidental brain injury before the age of 2 years
- Central nervous system infection

The role of genetic factors in CP has become a growing area of interest. With testing becoming more available, studies have found that CP may be associated with a genetic variant in 14% to 60% of cases.[10] Some have argued that a genetic diagnosis precludes a diagnosis of CP. However, a 2019 international multidisciplinary consensus statement recommends that a clinical diagnosis of CP be maintained even if an underlying genetic variant is identified, provided the patient "exhibits a nonprogressive permanent disorder of movement and posture."[11]

History

The clinical evaluation for CP should include a comprehensive history, including a discussion about any movement or tone abnormalities

observed by the family and an assessment of risk factors for CP, as described in the previous section. Developmental milestones should be reviewed. For children with a history of prematurity, timing of milestones should be considered in reference to their corrected age until the age of 2 years. The provider should make note of developmental delays as well as abnormal achievement of motor milestones. For example, early head control or rolling may occur as a result of hypertonia, and early handedness can occur in hemiplegia. Additionally, the provider should confirm that the child has not experienced developmental regression or loss of milestones, as CP by definition involves a nonprogressive neurologic insult.

Physical Examination

Initial physical examination of a child with suspected CP:

- Focus on assessment of tone, posture, and mobility.
- Tone abnormalities can be described by type of movement disorder and topographic distribution, as noted in the "Classification" section.

Initial and subsequent examinations:

- Evaluate tone.
- Assess range of motion (ROM) and document the presence of contractures.
- Examine the spine for kyphosis and scoliosis.
- Observe gait if the child is ambulatory. Common gait deviations in CP include scissoring due to hip adductor tone, crouch gait due to hip flexor and hamstring tightness, and toe walking with genu recurvatum due to ankle plantarflexor tone.
- Classify function using the Gross Motor Function Classification System and other scales described in Table 2.1.

Diagnostic Evaluation

CP is a clinical entity without a single diagnostic test. Basic diagnostic criteria include alterations in movement or posture, activity limitation, and a static insult that occurred in the developing brain, which may be assessed through a range of standardized assessments. A detailed discussion of best practices in early diagnosis is available at https://www.aacpdm.org/publications/care-pathways/early-detection-of-cerebral-palsy.

Standardized Neurologic and Motor Assessments

To explore a diagnosis of CP, consider[3]:

- Prechtl Qualitative Assessment of General Movements (GMA) before 5 months of age
- Hammersmith Infant Neurological Examination (HINE)
- Alberta Infant Motor Scale (AIMS)
- Developmental Assessment of Young Children (DAYC) after 5 months of age

Table 2.1 Functional Classification

CLASSIFICATION	DEFINITION	LEVEL I	LEVEL II	LEVEL III	LEVEL IV	LEVEL V
GMFCS	Usual gross mobility performance See reference for detailed breakdown by age	Walks without limitations	Walks with limitations	Walks with handheld mobility device	Self-mobility with limitations Power mobility may be used	Transported in a manual wheelchair
MACS	Typical object handling classification system Age 4–18 years	Handles objects easily and successfully	Handles most objects but with some reduced quality and/or speed of achievement	Handles objects with difficulty Needs help to prepare and/or modify activities	Handles a limited selection of easily managed objects in adapted situations	Does not handle objects and has severely limited ability to perform even simple actions

EDACS	Eating and drinking abilities Age 3 and older Efficiency: "eat a meal in the same time as peers"	Eats and drinks safely and efficiently	Eats and drinks safely but with some limitations in efficiency	Eats and drinks with some limitations to safety May have limitations in efficiency	Eats and drinks with significant limitations to safety	Unable to eat or drink safelytube feeding may be considered to provide nutrition
CFCS	Functional communication scale	Sends and receives familiar and unfamiliar partners effectively and efficiently	Sends and receives familiar and unfamiliar partners but may need extra time	Sends and receives familiar partners effectively, but not with unfamiliar partners	Inconsistently sends and/or receives even with familiar partners	Seldom effectively sends and receives, even with familiar partners

Source: Palisano R, Rosenbaum P, Bartlett D, et al. *GMFCS – E&R. CanChild*, 2021. https://canchild.ca/en/resources/42-gross-motor-function-classification-system-expanded-revised-gmfcs-e-r; Eliasson A-C, Krumlinde-Sundholm L, Rösblad B, et al. The manual ability classification system (MACS) for children with cerebral palsy: Scale development and evidence of validity and reliability. *Dev Med Child Neurol.* 2006;48(7):549. doi: 10.1017/s0012162206001162; Tschirren L, Bauer S, Hanser C, et al. The eating and drinking ability classification system: concurrent validity and reliability in children with cerebral palsy. *Dev Med Child Neurol.* 2018;60(6):611–617. doi: 10.1111/dmcn.13751; and Hidecker MJC, Paneth N, Rosenbaum PL, et al. Developing and validating the Communication Function Classification System for individuals with cerebral palsy. *Dev Med Child Neurol.* 2011;53(8):704–710. doi: 10.1111/j.1469-8749.2011.03996.x.

CFCS, Communication Function Classification System; EDACS, Eating and Drinking Ability Classification System; GMFCS, Gross Motor Function Classification System; MACS, Manual Ability Classification System.

Imaging

Although infants may be examined with ultrasound, brain MRI without contrast is the imaging study of choice for evaluation of CP. Normal imaging does not preclude a diagnosis of CP and is seen in 10% to 15% of cases.[12] Characteristic MRI findings include[3]:

- White matter injury, such as periventricular leukomalacia or periventricular hemorrhagic infarcts
- Cortical and deep gray matter lesions, such as basal ganglia or thalamic lesions, watershed injury, or stroke
- Brain malformations, such as lissencephaly, pachygyria, cortical dysplasia, polymicrogyria, or schizencephaly

The diagnosis of CP will typically be based on a combination of physical examination, standardized assessments, and imaging.

Laboratory Studies

No biomarkers for CP exist. Genetic and other laboratory testing should be considered, particularly in the case of a normal MRI, in order to rule out neurodegenerative or metabolic disorders with a similar presentation.[10,12] Genetic panels can assess for a number of genetic etiologies of CP as well as CP mimics with a single test.

TREATMENT

Guiding Principles

The goals of treatment for CP include:

- Early diagnosis and initiation of high-quality, CP-specific therapy interventions
- Maximizing function and community participation
- Minimizing complications, such as development of contractures
- Providing holistic and family-centered care

Management in Early Childhood

The importance of early identification and intervention for CP has been increasingly recognized, with the goal of maximizing neuroplasticity and preventing musculoskeletal complications. Children should receive CP-specific early intervention therapies that are task-oriented and use child-initiated movement; passive stretching is not sufficient.[3]

Ongoing management is detailed in Table 2.2.

Therapy

- After early childhood, episodic, goal-based physical and occupational therapy is preferred over weekly models of therapy that focus on stretching and ROM.
- Intermittent consultative visits can be beneficial in assessing interval changes in ROM, evaluating for fit of orthoses and other adaptive equipment, and refining home exercise programs.

Table 2.2 Ongoing Care

COMORBIDITY	COMPLICATIONS	MANAGEMENT
Visual abnormalities: retinopathy of prematurity, optic neuropathy, cortical visual impairment	Impaired visual perception, difficulty with motor coordination, mobility, and social interaction	Pediatric ophthalmologist for monitoring, specialized therapy for visual impairment, environmental adaptations
Hearing impairment	Speech and learning delays	Audiology evaluation and regular screening assessments, augmentative alternative communication assessment
Oropharyngeal dysphagia, oromotor feeding impairment[8,13]	Aspiration pneumonia, malnutrition, GERD	Monitor weight, speech therapy for motor feeding therapy, modified barium swallow study, thickened oral feeds, gastrostomy tube
Dental problems[14]	Dental caries, malocclusion, gingival disease	Pediatric dental evaluations every 6 months, GERD management, oral care at home daily
Drooling[15]	Skin breakdown, dental caries, odor, social stigmatization, dehydration	Drooling Impact Scale for monitoring, OT oromotor therapy, postural management with equipment, medications, botulinum toxin injections to salivary glands Severe: ENT referral for salivary gland excision (risk of dysphagia)
Recurrent aspiration, impaired airway clearance, restrictive lung disease[16,17]	Bronchopulmonary dysplasia Recurrent respiratory infection as a major cause of morbidity and mortality	Pulmonary assessments, swallow evaluation/nutrition support, airway clearance/chest physiotherapy, scoliosis monitoring and treatment, ventilation support

(*continued*)

Table 2.2 Ongoing Care (*continued*)

COMORBIDITY	COMPLICATIONS	MANAGEMENT
GERD[18]	Dental erosions, pain, respiratory issues (cough, pneumonitis)	Positioning, thickened feeds, medications (proton pump inhibitors, histamine 2 receptor antagonists) Severe: surgical fundoplication
Constipation, fecal incontinence[19]	Pain, spasticity, feeding problems, poor appetite	Bowel history monitoring, abdominal x-ray, bowel medications, dietary fiber and fluids
Dysfunctional voiding[18,19,20]	Urinary incontinence, urgency, frequency, retention, and infection Toilet training takes longer due to motor impairments	Monitor for UTI, adaptive equipment
Pressure ulcers[21]	Mobility impairments limit weight shifts in seating or bed Ulcers can lead to infection	Custom seating systems with pressure relief cushion, pressure relief bed mattress, daily skin checks, keep skin dry
Osteoporosis[21,22]	Fragility fractures, bone pain	Calcium and vitamin D supplementation, monitor vitamin D 25-OH levels, bone labs, x-rays (long bones, spine), DEXA scan
Behavioral: ADHD, anxiety, depression[21]	Impact on school and quality of life	Cognitive behavioral therapy, psychiatry consultation for medical management

Pain[8]	Etiologies: hip subluxation, muscle spasms, dystonia, constipation Decreased participation in school and recreation Impaired quality of life	Treatment of underlying medical cause: tone and constipation x-rays to screen for hip subluxation or long bone fracture
Epilepsy[23]	Partial epilepsy, generalized tonic-clonic seizures	Antiepileptic medication determined by seizure type, refer to child neurologist, EEG or video EEG Seizure rescue medication at home and school, school seizure plan
Sleep problems: insomnia, central or obstructive sleep apnea, nocturnal seizures, dysregulated sleep (pain, muscle spasms, positioning)[24]	Academic or behavioral problems, headaches, caregiver sleep disruption	Sleep hygiene, spasticity management, seizure management Sleep/bed positioning systems, sleep-safe bed, sleep study, medications[3]
Cognitive	Cognitive impairment	Neuropsychology testing by age 5–6 years old, then repeat testing every 2–3 years Management of medical factors such as seizures Advocacy for inclusion in school classroom

(*continued*)

Table 2.2 Ongoing Care (*continued*)

COMORBIDITY	COMPLICATIONS	MANAGEMENT
Hip dysplasia	Structural changes result in increased risk of hip subluxation and dislocation[25]	Refer to orthopedic surgery: migration percentage >30%, <30° of hip abduction, or if hip pain during examination (AACPDM guideline) See Table 17.1 for hip surveillance timing guidelines; see also www.aacpdm.org/publications/care-pathways/hip-surveillance-in-cerebral-palsy
Neuromuscular scoliosis	Scoliosis can progress beyond skeletal maturity, can cause challenges in seating and positioning	Obtain baseline AP/lateral spine image and repeat every 1–2 years until the Cobb angle plateau is reached after skeletal maturity Nonsurgical management: improve seating positioning; bracing is not effective Refer to surgery for Cobb angle >50° or if progression of curve is beyond skeletal maturity[27]
Contractures	Can occur at any joint, limit function, and cause pain	Mild: trial stretching and splinting in addition to serial casting +/– neurotoxin injection for plantarflexion contractures Moderatesevere: refer to orthopedic surgery[28]
Spasticity and dystonia: see Chapter 9, "Spasticity Management"		

25-OH, 25-hydroxy vitamin D; OT, occupational therapy

- Specific therapy interventions that are considered particularly beneficial include:
  - Constraint-induced movement therapy (CIMT)
  - Bimanual training
  - Partial body weight support ambulation training
  - Strengthening programs
  - Speech therapy for feeding skills, language development, and evaluation for augmentative communication devices
  - Vision therapy for children with CP and cortical visual impairment or other etiologies of low vision

Surgery

Orthopedic Surgery

Children with CP are at risk of muscle imbalance and musculoskeletal deformities, including scoliosis, hip dysplasia, and joint contractures. Orthopedic surgery is usually delayed until the age of 4 to 7 due to risk of recurrence in younger children, and attempts are made to combine procedures into a single-event multilevel surgery.[25] The goal of surgery is to reduce pain and improve care and function, and includes procedures such as tendon lengthening, tendon transfers, as well as bone surgery for contractures and guided growth.

Neurosurgery

Please see Chapter 9, "Spasticity Management" for common surgical procedures to address spasticity and dystonia.

ADDITIONAL READING

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Brain Injury

3

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TRAUMATIC BRAIN INJURY GENERAL PRINCIPLES

Definition

Moderate to severe traumatic brain injury (TBI) results from damage to the brain tissue caused by an external mechanical force,[1] and includes non-penetrating (closed head injury) and penetrating (open head injury) injuries (Table 3.1).

Etiology

The most common causes by age are falls in infants, falls or blunt trauma in school-age children, and motor vehicle collisions (MVC) in adolescents. In children under 5 years of age, nonaccidental trauma (also known as abusive head trauma) is considered.[2,3]

Epidemiology

Prevalence[4]*:* TBI-related *hospitalizations* (rate: 25/100,000) most commonly caused by falls (ages 0–9) and motor vehicle collisions (ages 10–17); TBI-related *deaths* (rate: 3/100,000) most commonly caused by motor vehicle crashes (all ages 0–17)

Risk factors: alcohol and/or substance abuse, high-risk behaviors, male gender, TBI history, history of psychiatric illness, diagnosis of attention deficit hyperactivity disorder, and low socioeconomic status

Classification of Traumatic Brain Injury

Rancho Los Amigos Level of Cognitive Functioning Scale

The Rancho Los Amigos Level of Cognitive Functioning Scale (RLA) describes the cognitive and behavioral patterns observed in patients after TBI. This 10-point scale can be accessed at www.neuroskills.com/education-and-resources/rancho-los-amigos-revised.

Pathophysiology and Mechanism of Injury[5]

Primary injury: due to mechanical damage that occurs at the time of injury; in closed head injury, result of contact between the brain and the interior skull, at the area of impact, or at contrecoup areas, and includes

Table 3.1 Classification of TBI

	GLASGOW COMA SCALE	LOSS OF CONSCIOUSNESS	POST-TRAUMATIC AMNESIA
Mild TBI	13–15	<1 hr	<24 hr
Moderate TBI	9–12	1–24 hr	1–7 d
Severe TBI	≤8	>24 hr	>7 d

TBI, traumatic brain injury.

Source: Data from Yeates KO, Brooks BL. Traumatic brain injury in children and adolescents. In: Morgan JE, Ricker JH, eds. *Textbook of Clinical Neuropsychology*. 2nd ed. Routledge; 2018:141–157.

lacerations (tears in brain tissue), contusions (bruising or microscopic hemorrhages), axonal injury (internal rotation and velocity lead to tearing and stretching of the axons within the white matter), and skull fracture
Secondary injury/metabolic cascade: direct or indirect injury occurring days to weeks following injury and can include hypoxia, ischemia, swelling/edema, hypotension, mass lesions, increased intracranial pressure, and poor cerebral perfusion pressure

Second impact syndrome: additional brain injury while still symptomatic leading to disruption of brain vasculature autoregulation; can result in coma and death[6]

DIAGNOSIS

Clinical Features[7]

- Dependent on the location and size of the lesion/injury; greatest rate of recovery seen within the first 6 months; continued recovery for 6 to 24 months but at a slower pace
- Motor impairments in balance, coordination, gait, tone abnormality, and response speed
- Sensory impairments in olfactory dysfunction, hearing, and vision
- Cognitive impairments in attention, arousal, memory, behavior, communication, and executive function
- Common medical impairments including neuroendocrine dysfunction, respiratory dysfunction, inadequate nutrition, neurogenic bowel and bladder, paroxysmal sympathetic hyperactivity (PSH), hydrocephalus, and seizures

Diagnostic Evaluation

Monitor duration of coma/unconscious state as well as the time in days it takes one to follow a command (*time to follow commands* [TFC]) and form new memories. These variables (duration of coma + time to follow commands and form new memories) combine to determine the length of time in days from the date of injury that one is in *posttraumatic amnesia* (PTA). The length of PTA and TFC are two of the best early predictors of outcome

after TBI.[8–10] TFC >26 days has been associated with poorer outcomes at 1-year follow up.[8,9,11]

Glasgow Coma Scale (GCS)[12]: scores range from 3 to 15; lower scores indicate greater impairment; less helpful after the acute period[13]

Rappaport Coma/Near Coma Scale14: used to assess responsiveness in those with very low function (RLA 1–3)

Coma Recovery Scale-Revised (CRS-R)[15]: used to assess responsiveness, following commands, and emergence from PTA; used serially to monitor clinical changes; moderate research support indicating JFK scores can predict recovery and outcomes[16,17]

Children's Orientation and Amnesia Test (COAT)[18]: screening for general orientation (person and place), temporal orientation (time and date), and memory; appropriate for ages 3 to 15

Other cognitive testing: neuropsychological screening completed in the postacute period as needed to assist with discharge recommendations or return to school

Laboratory Studies[19]

Endocrine dysfunction is common and most resolve by 1 year after injury. Screening for moderate to severe TBI-induced hypopituitarism is recommended (at the time of injury, at 3 months and 6 months, and yearly after) and includes serum cortisol, free thyroxine (FT4), thyrotropin (TSH), and insulin-like growth factor (IGF-1).

- *Pubertal age:* includes prolactin, follicle-stimulating hormone (FSH), luteinizing hormone (LH), testosterone, and estradiol
- *For polyuria:* urine-specific gravity, sodium, and plasma osmolality after 12 hours of fasting

Radiographic Assessment

CT: acutely, assesses the need for emergent neurosurgical intervention; detects fractures, large parenchymal hemorrhages, acute hydrocephalus, and extra-axial hemorrhages,[20–23] with contusion being the most common intracranial finding reported[24]; can see hemispheric hypodensity with abusive head trauma[25]

MRI: more sensitive to intraparenchymal lesions[23]; susceptibility-weighted and diffusion-weighted sequences (DWI) help detect diffuse axonal injury[26]; lesions found on fluid-attenuated inversion recovery (FLAIR) MRI correlate with poorer functional outcome[27]

EEG: seizures more common with known risk factors[28]

ANOXIC BRAIN INJURY

GENERAL PRINCIPLES

Definition

Anoxic brain injury occurs when there is an interruption of oxygenated blood supply to the brain.[29] Hypoxic-ischemic encephalopathy (HIE) and anoxic brain injury are often used interchangeably.[29,30]

Etiology

Perinatal anoxia (see Chapter 2, "Cerebral Palsy") and ischemic stroke (see Chapter 5, "Stroke")

Cardiac arrest, most commonly due to respiratory arrest,[29–31] although can also be due to congenital cardiac disease, hypovolemic or septic shock, arrhythmias, and trauma,[31] with other causes including nonfatal drowning,[29,32,33] hanging or near-hanging,[34] smoke inhalation or carbon monoxide poisoning or other acute lung injury,[29,30] and drug overdose[29,30]

Epidemiology

- Across all pediatric ages, the incidence is 8.3/100,000. In infants, the incidence is 75.3/100,000.[35]
- Children aged 0 to 4 years are at the highest risk of nonfatal drowning (16/100,000).[32] Near-drowning is associated with more severe impairments than other causes of HIE.[36]
- Accidental hanging or near-hanging injuries are more common in younger children. Intentional injuries are more common in young adolescent males.[34]

Pathophysiology and Mechanism of Injury

- With interruption of blood supply to the brain, oxygen and glucose delivery is also impaired. The brain is unable to store energy, and with ongoing ischemia or hypoxia the production of adenosine triphosphate (ATP) is greatly diminished, resulting in cell death from excitotoxicity.[29,30]
- Infarcts in watershed areas from ischemia lead to damage in areas that are high in metabolic demand, such as the pyramidal neurons of the hippocampus and cerebral cortex, basal ganglia, and cerebellum.[29,37]

DIAGNOSIS

Differential

TBI, intracranial hemorrhage, ischemic stroke, seizure or postictal state, metabolic abnormalities (hypoglycemia, hyponatremia, and hypernatremia), sepsis, drug overdose or alcohol intoxication, locked-in syndrome, and akinetic mutism[29,30]

Clinical Features

- Clinical symptoms include coma, altered level of consciousness, seizures, and strokes.[37]
- Thalamocortical injury leads to long-term memory deficits and persistent vegetative state.[37]
- Basal ganglia injury leads to movement disorders such as myoclonus, rigidity, chorea, tics, dystonia, ataxia, dysarthria, and dysphagia.[37,38]
- PSH presents as episodic agitation, hypertension, tachycardia, and tachypnea. It results from injury to autonomic centers such as the hypothalamus or its connections to the brainstem, cortex, or subcortical regions.[37]

Diagnostic Evaluation

Cranial nerve examination along with pupillary reactivity, corneal reflex, and oculocephalic reflex to determine brainstem function if able[37]

Laboratory Studies

Initially include electrolyte panel, complete blood count, blood glucose, blood urea nitrogen, creatinine, liver function tests, arterial blood gas, urine drug screen, and/or blood alcohol level depending on the underlying etiology[29]

Radiographic Assessment

MRI: T2 and/or DWI identify abnormalities due to anoxic injury and help with outcome prognostication.

CT: Noncontrast evaluation for structural lesions that require intervention should be done on all with altered levels of consciousness, and may be unremarkable in hypoxic injury but may see loss of graywhite differentiation. If acute stroke or vascular injury is suspected, CT angiography and/or perfusion may be warranted.[29,30,37]

EEG: Status epilepticus, burst suppression, generalized suppression, and alpha coma patterns are correlated with poorer outcomes.[29,33,37,39]

Somatosensory evoked potentials (SSEPs): The median nerve stimulation SSEPs with subsequent absent bilateral cortical N20 responses are specific for poor prognosis.[29,30,33,37,39]

ENCEPHALOPATHY

GENERAL PRINCIPLES

Definition

Encephalopathy refers to the damage or disease of the brain parenchyma due to infection or inflammation.

Etiology[40]

- *Infectious:* bacteria, viruses, fungi, and parasites such as *Streptococcus*, pneumococcus, *Haemophilus influenzae*, *Enterovirus*, arboviruses, and *Naegleria fowleri*
- *Noninfectious:* autoimmune and rheumatologic conditions such as autoimmune disseminated encephalomyelitis (ADEM) and systemic lupus erythematosus (SLE)

Epidemiology

- The incidence of viral encephalitis is 3.5 to 7.4 cases per 100,000.[41] The incidence of pediatric encephalitis (from all causes) is 10.5 to 13.8 cases per 100,000. Arboviruses, transmitted by insects, are seen in summer and fall. *Enterovirus* spreads via the feco-oral and respiratory routes. Adenovirus spreads via respiratory route, lymphocytic choriomeningitis through direct contact with rodents, and herpes by epithelial or sexual contact.[42]

- Of 2,600 sporadic meningitis cases per year, the incidence of pneumococcal meningitis is 1.1/100,000 and *H. influenzae* meningitis .2/100,000. Worldwide, 350,000 deaths from meningoencephalitis with an incidence of 700,000 cases have been reported.[43]
- *Risk factors:* Viral meningitis in <5-year-olds, bacterial meningitis in >20-year-olds, community settings, and swimming in freshwater increase the risk.[42] Encephalitis is seen in older adults and children <1 year old, living in areas with ticks and mosquitoes, with recent vaccinations, and in immunocompromised individuals.
- *Protective factors:* These include handwashing, avoiding sharing of food and objects, vaccinations such as measles, mumps, and rubella (MMR) and pneumococcal vaccines, and prophylactic antibiotics if exposed, as well as protective measures against mosquitoes/insects and avoiding standing bodies of water.[44]

Pathophysiology and Mechanism of Injury

Bacterial and viral infections can lead to hematogenous spread to the central nervous system (CNS), with the choroid plexus as the potential entry location.[40] This increases inflammation, which causes leukocyte invasion and disruption of the bloodbrain barrier.[40]

DIAGNOSIS

Differential[45]

Acute toxic or metabolic encephalopathy, hepatic encephalopathy, SLE cerebritis, multiple sclerosis, granulomatous angiitis, CNS vasculitis, CNS tumor, stroke, seizures, Colorado tick fever, mycoplasma meningoencephalitis, *Legionella*, subacute bacterial endocarditis, Rocky Mountain spotted fever (RMSF), and malaria

Clinical Features[40]

- Presents fever, malaise, headache, neck stiffness (meningismus), photophobia, phonophobia, and vomiting as signs of meningeal irritation
- Can also present with focal neurologic signs such as seizures, limb paresis, impaired consciousness, or coma
- Meningococcal petechiae, Osler nodes, fulminant Gram-negative sepsis with prominent cardiovascular insufficiency, and disseminated intravascular coagulation

Laboratory Studies[40,46]

- *CSF studies:* Gram staining, culture and sensitivity, polymerase chain reaction, and latex agglutination tests
- *In bacterial meningitis:* CSF with elevated white blood cell count (WBC) >500 cells/μL, predominant neutrophils, elevated protein >1 g/L, increased lactate >.3g/L, and decreased glucose CSF to blood ratio <.4; increased peripheral WBC, erythrocyte sedimentation rate, C-reactive protein, and procalcitonin
- *In viral meningitis:* slightly elevated CSF protein and normal glucose

Radiographic Assessment

MRI: shows edematous changes in the orbital surfaces of the frontal lobes and the medial temporal lobe in herpes simplex virus (HSV) encephalitis and is helpful in evaluating other causes of encephalopathy[47]

CT: shows cerebral edema, hydrocephalus, and infarcts; bone window imaging can identify sinusitis, mastoiditis, or odontogenic abscess

EEG: most sensitive early in infection and may show slow waves over the brain[40]; HSV encephalitis showing characteristic nonspecific diffuse high-amplitude slow waves[47]

TREATMENT OF TRAUMATIC AND ANOXIC BRAIN INJURY AND ENCEPHALOPATHY

Concurrent Injuries in Traumatic Brain Injury

Spinal cord injury (SCI): Dual diagnosis in pediatrics is unknown, although recent study estimates about 50% in pediatric inpatient rehabilitation (IPR).[48] This may be due to the anatomical characteristics of the cervical spine and the head to torso ratio and MVC as a common etiology for both diagnoses.[48,49]

Fractures (polytrauma): Due to cognitive impairment after TBI, children may not be able to indicate pain from undetected fractures.[50]

General Principles in Anoxic Brain Injury

- Resuscitation, treatment of underlying cause (metabolic derangements, confounding drug exposure, hypovolemia, hypotension, and hypoxia[29]), prevention of ongoing brain injury, and supportive care are recommended.[29]
- Therapeutic hypothermia (32°C–34°C) used in comatose adults after ventricular fibrillation and resuscitation may also provide some neuroprotection in infants after HIE.[30,37,51]
- Consider cervical cord injury if anoxia resulted from hanging or near-drowning/diving.[29]

Initial Management

Traumatic and Anoxic Brain Injury

Disorders of Consciousness

Children who do not emerge from RLA 1 to 3 within the acute or postacute time frame are considered to have a disorder of consciousness (DoC). Long-term functional outcomes in DoC cannot be accurately predicted in the early stages after the injury.[52] Progress (from RLA 1 to 2 or from 2 to 3) can still occur at 5+ years postinjury.[53,54]

Arousal/Awareness

The primary goal is to enhance arousal and responsiveness by[55]:

- Minimizing sedating medications for pain, seizures, tone, PSH, and agitation by finding the least sedating alternatives[55,56]

- Introducing neurostimulants such as dopamine agonists (amantadine or bromocriptine), carbidopa/levodopa, pramipexole, methylphenidate, and zolpidem[55,56]
- Optimizing environmental factors, such as lighting to assist with orientation and sleepwake cycle, clustering overnight care, limiting screens/alarms, and weaning sedating medications during the day

Agitation

Always evaluate for underlying medical causes or factors (e.g., pain) that exacerbate agitation.[56] Trial less sedating medications such as propranolol, valproate, or carbamazepine,[55] or a short duration of benzodiazepines. Selective serotonin reuptake inhibitors may treat both post-TBI depression and agitation, although not acutely.[56] Antipsychotics remain controversial due to their side effect profiles.[56]

Paroxysmal Sympathetic Hyperactivity

The goals of treatment include blunting excessive sympathetic outflow, avoiding triggers (movement, pain, and urinary retention), and managing the effects on organ systems.[57] Depending on clinical features being targeted, options include[55,57,58] beta-adrenergic blockers (propranolol), neuromodulators (bromocriptine, gabapentin, and baclofen), or dantrolene, and sedating medications such as benzodiazepines (diazepam) and alpha-adrenergic agonists (clonidine and dexmedetomidine), and can be used for a limited duration.

Tone

Increased tone is a common occurrence after moderate to severe TBI and anoxic brain injury[59] and can present as spasticity or dystonia.[55,59–61]

- *Spasticity:* This refers to velocity-dependent resistance to stretch that varies depending on the direction of joint movement.[60,61] See Chapter 9, "Spasticity Management" for more information.
- *Dystonia:* Abnormal involuntary co-contractions result in abnormal posturing.[59,61] Systemic management includes carbidopa-levodopa, trihexyphenidyl, dantrolene, clonidine, benzodiazepines, and intrathecal baclofen.[55,59] Deep brain stimulation (DBS) has limited results in secondary dystonia.[61]
- Physical management includes stretching with therapies, casting, orthoses, modalities, and assistive technology.[61]

Sleep–Wake Cycle

Difficulties in maintenance of sleep and delayed sleep onset are often seen.[62,63] Melatonin is often used for sleep initiation[55,62] and trazodone for sleep maintenance.[55] Alternative therapies include cognitive behavioral therapy for insomnia, acupuncture, blue light therapy, and moderate-intensity aerobic exercise.[63]

Neurogenic Bowel/Bladder

This is due to overactive (most common) or atonic detrusor muscle. Early recognition and evaluation through urodynamic studies and management can improve quality of life.[64,65] Renal function tests are recommended due to predisposition to intrinsic renal problems.[64] Functional constipation and fecal incontinence have been reported after acquired brain injury (ABI).[66] Various types of laxatives such as osmotics, contacts, emollients, and those promoting defecation reflex may promote normal defecation patterns.[66]

Chronic pain is prevalent after brain injury, most commonly of the head.[67] Musculoskeletal or neuropathic pain is typically due to fractures, dislocations, soft tissue trauma, or nerve damage.[67] Central pain can result after a CNS lesion and present as pain in body regions without pathology.[67] Children may not be able to reliably communicate pain complaints, but careful evaluation and management with least sedating medications is imperative.[55]

Seizures

A higher risk of seizures is associated with severity of brain injury.[59] Posttraumatic epilepsy (PTE) is the occurrence of two or more late seizures (>7 days after injury).[68] Risk factors include age ≥10 years, family history of epilepsy, early seizures, severe TBI, depressed skull fractures, penetrating injuries, and intracranial hemorrhage.[68,69] Seizure prophylaxis with levetiracetam or phenytoin/fosphenytoin is recommended for 7 days post-TBI,[70] but this does not prevent the development of PTE.[68]

Heterotopic Ossification

The prevalence increases in diffuse lesions or postanoxic children[71] and most frequently forms in the hip or knee.[72] Risk factors include concurrent fractures, prolonged coma, spasticity, younger age, male gender, autonomic dysregulation, and surgery.[60,71] Increased levels of alkaline phosphatase, bone scan, and MRI can help with diagnosis.[60,64] Nonsteroidal anti-inflammatory drugs and bisphosphonates can reduce symptoms.[64] Surgical excision is considered in those with significant pain, functional limitations, and hygiene/wound issues.[60]

Nutrition

The current recommendation is to start enteral feeding within 72 hours.[70] This often requires gastrostomy or gastrojejunostomy feeds in acute and postacute stages.

Encephalopathy

Initial treatment includes immediate antibiotic therapies (2–4 weeks depending on the cause). Corticosteroids reduce cerebral edema, intracranial hypertension, and meningeal inflammation. Supportive treatment includes maintenance of hydration, nutrition, thromboembolism prevention, and treatment of associated infections.

Ongoing Care

Severe brain injury is a chronic disease process and long-term needs vary based on residual deficits.[59,55,73] This requires an ongoing multidisciplinary approach.[55]

Medical

Medical management of complications mentioned under Initial Management.

Cognitive

Full neuropsychological testing in language and verbal reasoning, visual-spatial processing, learning and memory, attention, processing speed, executive functions, social functioning, behavioral/emotional functioning, and academic achievement is recommended postinjury to capture gains and guide intervention. Cognitive rehabilitation is appropriate for children with adequate insight into their deficits. For children without insight into their deficits, intervention focuses on environmental modifications (caregiver-driven).[74,75]

Therapies

Speech/language, occupational, and physical therapies are recommended within the postacute period to support regain of prior function and teach accommodation/modification strategies. Periodic episodes of care throughout key developmental sensitive periods are indicated to preserve/maintain function.

Behavioral/Emotional

Children may have new social, emotional, and behavioral difficulties related to increased executive deficits (increased impulsivity, decreased inhibition, decreased attention, apathy, etc.). Young children are vulnerable to late-onset executive deficits and can demonstrate new/evolving behavioral problems as they age.[76] Executive deficits are linked to broader difficulties with social adjustment and behavior regulation.[77]

School[78,79]

Schools can provide physical (transportation, mobility), cognitive (attention, memory, learning), and medical (nursing, support for activities of daily living, vision, hearing) accommodations as well as curriculum modifications. School-based therapies are also available but should not take the place of medically based therapies.

ADDITIONAL CONSIDERATIONS

Additional considerations include guardianship for young adults and palliative and advance care planning.

Prevention

Home: secure furniture/electronics and baby gates, remove blinds with looped cords, cover outlets, working carbon monoxide/smoke detectors; drowning: swimming lessons, use of life jackets, supervised access to standing water[80,81]; transportation: helmets and seat belts, car seats; and weapon-related injury: storage of ammunition separately from unloaded weapons in a locked box

Prognosis/Outcomes

Improved outcomes: associated with less severe injury, traumatic versus anoxic injury,[82,83] presence of intracranial pressure (ICP) monitoring with TBI,[84] and enteral feeding started within 72 hours of admission[85]

Poorer outcomes: associated with younger age at injury,[82,83] extension (or no motor response) to painful stimuli on GCS 48 to 72 hours after discontinuation of sedatives and paralytics,[33,37] GCS <5 at 24 hours after injury,[33] absence of corneal and/or pupillary reflexes at 72 hours,[29] hemispheric hypodensity on head CT,[25] lesions in multiple brain lobes (DWI) or basal ganglia and cortex (T2-weighted MRI) in the first 2 weeks after injury,[30,51,86] presence of PSH,[58,87] public insurance payor (Medicaid),[88] receiving care at nonpediatric facility,[88] and lower socioeconomic status[89]

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Concussion

4

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GENERAL PRINCIPLES

Definition

There is no universally accepted definition for mild traumatic brain injury (mTBI) or concussion, and the terms are often used interchangeably. Sports-related concussion (SRC) is generally felt to be a subset of mTBI. mTBI committees from the Centers for Disease Control and Prevention (CDC), the World Health Organization, and the American Congress of Rehabilitation Medicine defined mTBI as "an acute brain injury resulting from mechanical injury to the head from external forces including:

- One or more of the following: confusion or disorientation, loss of consciousness (LOC) for 30 minutes or less, post-traumatic amnesia for less than 24 hours, and/or other transient neurological abnormalities, such as focal signs, symptoms, or seizure;
- Glasgow Coma Scale (GCS) score of 13 to 15 after 30 minutes post-injury or later upon presentation for health care."[1]

Epidemiology

- *General:* Traumatic brain injury (TBI) is a leading cause of childhood morbidity, with at least 75% being classified as mTBI.[2]
- *Incidence/Prevalence:* The true prevalence of mTBI in children is unknown due to multiple healthcare entry points, no universally accepted definition for mTBI, limited injury surveillance systems, and underreporting of injuries.[3] It appears the incidence of mTBI has been increasing,[1,4] and the rising incidence is likely due to increased awareness among medical personnel, coaches, parents, and athletes, as well as increased media exposure. This has led to improved diagnosis and increased reporting.[3] A review of three nationally representative surveys evaluating lifetime pediatric concussions/head injuries estimated the prevalence to range from 3.6% to 7.0% in children ages 3 to 17 years and from 6.5% to 18.3% in adolescents 13 to 17 years.[5] The review also found that the increased prevalence was associated with increasing age, male sex, and non-Hispanic white race/ethnicity.[5] In gender-comparable sports such as soccer, females have been found to have a higher rate of SRC.[6]

Etiology

Sports and recreational-related injuries are the leading cause of TBI-related ED visits among children and teens.[7] The most common mechanism of injury varies by age, with 0- to 9-year-olds most commonly presenting with injuries related to playground activities and bicycling. Football and bicycling were the most common activities in 10- to 19-year-old males, and soccer and basketball in 10- to 19-year-old females.[7] SRCs are often reported using athletic exposures, which are equivalent to an athlete participating in some or all of one practice or game. High school sports with the highest reported rates of SRC per 1,000 athletic exposures are football, lacrosse, and ice hockey for boys, and soccer, lacrosse, and basketball for girls.[3]

Mechanism of Injury

Concussion results from rapid acceleration/deceleration forces of the brain. It can be caused by a hit to the head, neck, or elsewhere on the body with forces transmitted to the head.[8]

Pathophysiology

The above forces result in an acute neurometabolic cascade. Recent reviews published on the pathophysiology of mTBI describe a number of contributing factors including ionic shifts, release of excitatory neurotransmitters, ongoing axonal and cytoskeletal problems, impaired synaptic plasticity, neuroinflammation, blood–brain barrier dysfunction, and potential for cell death.[9,10] It is thought that individuals experience symptoms of concussion due to changes in pathophysiology. Symptoms improve as the neuronal homeostasis is restored.

DIAGNOSIS

Risk Factors for Prolonged Symptoms

The most consistent predictor of prolonged recovery is the severity of the acute and subacute symptoms.[11] The CDC mTBI guideline states that clinicians should screen for known risk factors for persistent symptoms, including older children/adolescents, children of Hispanic ethnicity, children of lower socioeconomic status, children with more severe presentation, and children reporting more acute postconcussive symptoms (PCS).[1] Additional risk factors that have been reported in the literature include female gender,[12,13] number of previous concussions,[13] premorbid history of mental health problems,[12,14] history of developmental problems,[15] dizziness,[16] and migraines.[14]

Initial Clinical Evaluation

If mTBI is suspected, the child should be removed from play or activity and an evaluation should be arranged by a healthcare provider with training in concussion. The child should not be left alone in the first few hours after mTBI. Concerns for more serious head injury ("red flags"), including LOC, declining mental status, focal neurologic deficit, severe or worsening

headache, repeated emesis, seizures, or suspicion of significant cervical spinal cord injury, should trigger immediate emergency evaluation.

Symptoms

Signs and symptoms of altered mental status (AMS) may be subtle and transient and typically occur at the time of impact but may evolve over minutes. Examples of AMS include LOC, posttraumatic amnesia, confusion, slowed thinking, inattention, slurred speech, or dazed feeling.[4,8] LOC is not necessary in the diagnosis of mTBI. Some symptoms may go unrecognized by the child, or an athlete may underreport symptoms to avoid losing play time. PCS are typically broken down into four main categoriescognitive, physical, neurobehavioral, and sleepand may present immediately or may develop over several hours or days (see Table 4.1). A number of conditions have symptoms similar to concussion, such as cervical strain, posttraumatic stress, depression, anxiety, sleep disorders, attention deficit disorder, and migraines, and it is important to differentiate the symptoms of these conditions from those that are due to mTBI.[4]

Physical Examination

A comprehensive neurologic examination should be performed and is commonly normal. A cervical spine examination, including passive range

Table 4.1 Concussion Signs and Symptoms

COGNITIVE IMPAIRMENT	PHYSICAL SIGNS	NEUROBEHAVIORAL IMPAIRMENT	SLEEPWAKE DISTURBANCE
Difficulty thinking clearly or feeling mentally foggy Feeling slowed down Difficulty concentrating Difficulty remembering new information Loss of consciousness	Headache Fuzzy or blurry vision Nausea or vomiting Dizziness Sensitivity to light and/or noise Balance problems Neck pain Hearing problems and/or tinnitus	Irritability Sadness Nervousness or anxiety Lability	Sleeping more than usual Sleeping less than usual Trouble falling asleep Feeling tired

of motion and assessment of any muscle or spinous process tenderness, should be performed. The scalp should be examined for ecchymosis, hematoma, or evidence of skull fracture. Additional tests for balance or oculomotor dysfunction should be considered.

Standardized Diagnostic Assessments

Symptoms are the most important and sensitive indicator of mTBI.[17] There are a number of standardized assessments available, but psychometric evidence is limited in children.

Sideline assessment: The Sport Concussion Assessment Tool, Fifth Edition (SCAT5) and the Child SCAT5 are standard sideline evaluation tools that are recommended by the Concussion in Sport Group in the Berlin 2016 guideline because they were deemed to be the most well-established and rigorously developed.[14] They did note, however, that the utility of the SCAT appears to decrease significantly 3 to 5 days after injury.[14] The SCAT5 is a multimodal screening assessment composed of the GCS, Maddocks Questions, Postconcussive Symptom Scale, Standardized Assessment of Concussion, a balance assessment (the modified Balance Error Scoring System), neck examination, and coordination testing.[18] The Child SCAT5 was developed for children ages 5 to 12 years old and incorporates simpler symptom questions, a parent coreport, and a more age-appropriate cognitive screening.[19]

Computerized neuropsychological testing (CNT): Examples of these tests are the Immediate Post-Concussion Assessment and Cognitive Test (ImPACT) and the CogSport. Testing after injury is compared either with an individual's preinjury results or population normative data. Comparisons of CNT with preinjury testing are limited by ongoing cognitive development and other factors that may be dissimilar, such as motivation, test setting, fatigue, mood, and practice effect.[16,20] The Berlin 2016 Conference systematic review made the following specific recommendations regarding CNT: The widespread use of baseline CNT is not recommended in children and adolescents given problems with reliability and insufficient evidence of diagnostic and prognostic value. CNT may be used under appropriate qualified supervision as an adjunct to clinical assessment in adolescents with SRC, and if it is used it should be combined with a multimodal clinical assessment.[16]

Neuropsychological testing: Postinjury formal neuropsychological testing is not recommended to all athletes following SRC.[1,14] It may be useful in a number of settings, including cognitive and attentional comorbidities, individuals with persistent PCS, and those with a history of repeated mTBI or impact exposure.[21] It can also be helpful in providing more detailed guidance regarding school or work accommodations.

Serum Biomarkers

The CDC mTBI guideline and the 2017 Concussion in Sport Group consensus statement do not recommend the use of biomarkers outside the research setting for the diagnosis of mTBI in children due to insufficient evidence to support their use.[1,14]

Radiographic Assessment

The results of conventional imaging are typically normal following mTBI.[1,3] The CDC guidelines do not recommend skull radiographs be used in the diagnosis or screening of intracranial injury (ICI) in mTBI.[1] A head CT can better detect ICI and characterize skull fractures, making it a better test when indicated.[1,22] Because CT studies are associated with ionizing radiation, it is recommended that validated clinical decision rules be used to identify children who may be at either high or low risk of clinically important ICI and thus guide the need for a head CT.[1] A commonly used example is the Pediatric Emergency Care Applied Research Network (PECARN) decision rules.[22] MRI is often noted to be the test of choice if neuroimaging is needed outside of the acute setting and can be considered in patients who are not recovering as predicted to assess for other pathology that can cause similar symptoms, such as a Chiari malformation.[3]

TREATMENT

Guiding Principles

The majority of children and adolescents experiencing concussion or mTBI will recover within the first 2 to 4 weeks postinjury.[4] In addition to the aforementioned risk factors for prolonged symptoms, recovery can be stalled by either prolonged rest or by return to full physical or cognitive activity while at the peak of symptoms. Rest is recommended for 24 to 48 hours, with gradual reintroduction to physical and cognitive activity.[4,23] The activities should not exacerbate symptoms or cause new symptoms to arise. For children returning to any sport or physical activity, they should follow the graded return to play (RTP) guidelines. An example of a graduated RTP strategy provided by the Berlin Conference on Concussion is presented in Table 4.2.[14] To facilitate a successful return to school, the following could be implemented:

- Modified assignments
- Providing written notes of lessons
- Increased time for tests/assignments
- Rest breaks throughout the day[4]

Patient/Family Education

The CDC mTBI guideline recommends that the healthcare professional include the following information when educating and providing reassurance following mTBI:

- Warning signs of a more serious injury
- Description of injury and expected course of symptoms and recovery
- Instructions on how to monitor PCS
- Prevention of further injury
- Management of cognitive and physical activity/rest
- Instructions on RTP, recreation, and school
- Clear clinician follow-up instructions[1]

Table 4.2 Example of Graduated Return to Sport Strategy

STEPS	RECOMMENDED ACTIVITIES
Symptom-limited activity	Start with typical daily nonsport activities as long as they do not cause worsening of symptoms.
Light aerobic exercise	Introduce activities that mildly increase heart rate, such as walking or using an exercise bike.
Sport-specific exercise	Participate in running or simple training drills that avoid risk of head impact.
Noncontact training drills	Participate in agility or more complex training drills with increasing demands for coordination and cognition. Resistance training can be introduced and advanced.
Full contact practice	Participate in normal sport training activities. Medical clearance should be obtained for this step.
Return to competition	Participate in typical competitive sports activities.

Source: Data from consensus statement on concussion in sport – the 5th International Conference on Concussion in Sport held in Berlin, October 2016.

Initial Management/Symptom Management

Headaches: Lifestyle modifications are the first line of treatment. This can include proper nutrition, hydration, getting adequate sleep, and reducing environmental triggers such as loud noises and activities that are overstimulating.[4,23] Typically, patients will self-medicate with over-the-counter medications such as acetaminophen or nonsteroidal anti-inflammatory drugs (NSAIDs). However, patients should be educated on the potential for medication overuse that can cause headaches to persist—the so-called "rebound headache"—which occurs if these medications are used more than three times per week.[23–25] In addition to graded return to physical and cognitive activity, treatment of headaches should target the specific etiology:

- *Migraine:* triptans
- *Cervicogenic/whiplash:* physical therapy, muscle relaxants, and NSAIDs
- *Tension:* nerve blocks and trigger point injections[23,25,26]

When headaches are refractory to abortive medications and environmental modifications, prophylactic medications and supplements such as amitriptyline, magnesium, and riboflavin can be useful. Other medications such as gabapentin, topiramate, and coenzyme Q10 (CoQ10) have been used, but the evidence to support their use in pediatric posttraumatic

headache is limited.[23,25,27] If headaches persist despite these interventions, referral to a headache specialist is warranted.

Cognitive impairments: Cognitive symptoms can be attributed to a number of factors. If a child is not sleeping well, has persistent headaches, or is feeling anxious or depressed, this can manifest as inability to problem-solve, plan, memorize, or sustain attention.[23] Often, treating the causative factors can improve the cognitive symptoms. However, if cognitive impairment persists, neuropsychological testing can be helpful in identifying the impairments and their severity so that proper school support can be put into place.

Sleep: Sleep impairment contributes to other symptoms of concussion: headache, fatigue, cognitive impairment, and mood changes. Environmental and lifestyle modifications are the first line in restoring an individual's typical sleepwake cycle. Examples of modifications include:

- Setting routine sleep and wake times
- Minimizing screen time an hour before bed
- Limiting naps[4,24]

When sleep problems persist, melatonin can be used for an extended period to reset the natural sleepwake cycle.[4,23,24] One can also take advantage of medication side effects (e.g., drowsiness from amitriptyline prescribed for headache) to help with sleep onset. If sleep problems persist, referral to a specialty clinic is recommended.

Oculomotor/vestibular dysfunction: Research on the effectiveness of vestibular rehabilitation has been limited and at times conflicting.[28,29] However, the general consensus is that vestibular rehabilitation is useful and may allow patients to return to activities faster. Strategies involved in the treatment of oculomotor or vestibular dysfunction involve dynamic movements of the head and eyes to provoke symptoms and retraining the vestibular system to tolerate these movements.

- For vestibularocular reflex impairment, gaze stability training involves gaze fixation while the head is moving.
- For motion sensitivity, patients are exposed to visually stimulating environments.
- For impaired postural control, balance training is used.
- For dizziness due to benign paroxysmal positional vertigo, the Dix-Hallpike maneuver is used.
- For oculomotor impairments, exercises include speeded saccadic eye movements, visual pursuit tasks, alternating monocular and binocular tasks, visual field scanning, near-far focal shifting, attention tasks, and reading.

Physical activity: There is a growing body of evidence to support reintroduction to physical activities early in the recovery process, and these are typically started immediately after the rest period, following the steps outlined in Table 4.2. A prescriptive exercise program that has demonstrated improvement in recovery time is the subsymptom threshold

aerobic exercise (SSTAE), where patients exercise at a target heart rate that is 80% of the heart rate that causes symptom exacerbation based on treadmill testing with a trained physical therapist.[30] Exercise is progressed based on symptom tolerance until patients are exercising at full capacity. For patients with protracted symptoms and delayed recovery, there is evidence that the SSTAE in conjunction with other rehabilitation therapies reduces symptom burden.[31]

Ongoing Care

There are a subset of patients who may experience persistent symptoms despite the above interventions. For these patients and those with risk factors for protracted recovery, a multidisciplinary approach to care can be useful. One study demonstrated a reduction in overall symptoms and depression scores and improvement in quality of life scores using a collaborative care model with cognitive behavioral therapy, care management, and psychopharmacologic intervention.[32]

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Stroke

5

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INTRODUCTION

The incidence of pediatric stroke is relatively rare; however in those affected, it has a high morbidity with long-standing impact on health, wellness, independence, and community reintegration. Evidence behind the recommended diagnostics and interventions in pediatric stroke is limited, although it continues to expand with growing interest in the topic. Variations in initial presentation and its relative rarity pose challenges to prompt identification and early treatment of stroke. Treatment requires an interdisciplinary approach to diagnosis and treatment for the best long-term outcomes. The team includes ED providers, neurologists, physiatrists, physical therapists, occupational therapists, speech therapists, neuropsychologists, and geneticists.

GENERAL PRINCIPLES

Definition and Epidemiology

Cerebrovascular accident (CVA), or stroke, is any cerebrovascular injury caused by occlusion or rupture of cerebral blood vessels.[1] Pediatric strokes are often defined according to the developmental stage in which they occur and by the mechanism of damage. Strokes that occur between gestation (28 weeks) and shortly after birth (≤28 days) are classified as perinatal strokes, while strokes that occur between >28 days and 18 years of age are classified as childhood strokes.[2] Stroke is one of the top 10 causes of death in children, with the highest mortality rate in the first year of life.[3] Of children with stroke, 10% to 25% die, up to 25% have recurrence, and up to 66% have persistent neurologic deficits, seizure disorders, or learning/developmental problems that they carry into adulthood.[4] Pediatric stroke is more common in boys and there is predominance in black children. The incidence in the United States is as follows: total: 1.2 to 13.5 per 100,000; neonatal/perinatal: 1 per 4,000 live births per year; childhood: 2 to 4 per 100,000 children; ischemic: 7.8 per 100,000; hemorrhagic: 2.9 per 100,000; cerebral sinus venous thrombosis: 0.7 per 100,000.[1,3,5,6]

Classification

Pediatric CVA can be classified into three broad pathophysiologic subtypes, namely arterial ischemic stroke, hemorrhagic stroke, and cerebral sinovenous thrombosis (CSVT).[2] *Arterial ischemic stroke* results from disruption in normal blood flow due to arterial blockage or narrowing (i.e.,

embolism, thrombus, or vasospasm), resulting in damage to the brain due to lack of oxygen and nutrients. *Hemorrhagic strokes* are bleeds in the brain following rupture or leakage of blood vessels. These can be further classified according to the location of the bleed: intracerebral (ICH), intraventricular (IVH), or subarachnoid (SAH). Spontaneous intraparenchymal hemorrhage is more common than nontraumatic SAH (i.e., aneurysm).[7] *CSVT* is a rare venous thromboembolism resulting in blockage of blood leaving the brain through the venous system. CSVT leads to a mixture of brain damage due to primary infarction (often due to swelling) and secondary bleeding (hemorrhagic transformation).[6]

Etiology

The vast majority of perinatal and childhood ischemic and hemorrhagic strokes have no identified cause. Ischemic stroke is commonly associated with cardiac disease and coagulation disorders, while hemorrhagic stroke is commonly associated with vascular malformation and trauma.[3,7] Some commonly associated conditions are discussed in the next section.

DIAGNOSIS

In contrast to adult stroke, children often encounter delay in medical attention, imaging, and diagnosis.[4] Identifying stroke is challenging, especially in early childhood, as children often present with nonlocalizing, atypical, or nonspecific symptoms, although they may present with focal neurologic deficits similar to adults beyond early childhood.

Risk Factors

In the pediatric population, risk factors are more diverse and abundant as compared with the adult population.[1] In neonates, there is an increased risk of stroke associated with maternal and labor factors. In young adults, modifiable risk factors are similar to those seen in older adults.[5]

Predisposing risk factors differ significantly between childhood and adulthood. Strokes that occur in adulthood are highly associated with lifestyle-related risk factors (e.g., hypertension, atherosclerosis, diabetes, smoking, and obesity). In contrast, pediatric strokes are more often related to nonlifestyle-related risk factors, including trauma, congenital cardiopulmonary defects or diseases, blood cell disorders (e.g., sickle cell disease and anemia), arteriopathies (e.g., arterial dissection and moyamoya disease), thrombophilias (e.g., factor V Leiden), connective tissue disorders (e.g., EhlersDanlos type IV), inflammatory or systemic diseases (e.g., lupus), and other genetic and metabolic disorders.[5]

Clinical Presentation

Clinical presentation can be quite varied, with more nonspecific symptoms associated with younger age. At all ages, stroke most commonly presents as a headache. It may also present as seizure or altered level of consciousness.[1] Infants may present with lethargy within the first few

days after birth and many remain neurologically asymptomatic until they develop voluntary hand use at 4 to 5 months of age.[1,3] Toddlers may present with general deterioration, feeding difficulties, vomiting, sepsis-like symptoms, or irritability.[1] Older children may present with more focal symptoms, including unilateral weakness, sensory deficits, difficulty walking, or sudden-onset vision changes or aphasia.

Symptoms

Acute presenting symptoms are varied and are outlined in the "Clinical Presentation" section. Poststroke symptoms affect multiple body systems and are outlined along with specific treatment recommendations in the "Treatment: Medical" section.

Physical Examination

Physical examination can be useful in identifying focal neurologic deficits such as cranial nerve involvement, hemiplegia, or hemisensory impairment, as well as vertigo, ataxia, nystagmus, bulbar dysfunction, and dysarthria. In the neonatal period, special attention should also be given to dilated scalp veins, large anterior fontanelle, or bulging fontanelles. Throughout childhood, presentation may also include neck pain, meningismus, or photophobia. At all ages, irritability may also be noted.[1]

Diagnostic Evaluation

Because the etiology is so varied, diagnostic evaluation is focused on identification of life-threatening comorbid conditions related to the acute injury (e.g., increased intracranial pressure leading to herniation) and factors that may lead to short-term recurrence.[5] Diagnostic tools commonly utilized include the following:

- *Pediatric National Institutes of Health Stroke Scale (NIHSS) score:* a useful aid in determining severity, tracking progress, and predicting future disability[4]
- Laboratory/imaging studies (see the "Laboratory Studies" and "Radiographic Assessment" sections)
- EKG/echocardiogram
- *EEG:* useful in identifying seizure and predicting the development of hemiplegia (if abnormal during the first week of life)[3]
- Neuropsychological assessment (see the "Treatment: Neuropsychology" section for more details)

Laboratory Studies

At present, there are no clearly established guidelines for laboratory assessment.[1] Considerations in addition to routine metabolic and hematologic studies include varicella titer, serum for autoimmune disorders (erythrocyte sedimentation rate [ESR], antinuclear antibodies (ANA), complement profile),[5] coagulation studies,[7] and glucose.[3]

Radiographic Assessment

Common imaging studies include the following:

- *Noncontrast head CT:* sensitive to acute bleeding; used to identify stroke size and estimate the likelihood of symptomatic intracranial hemorrhage following tissue plasminogen activator (tPA)[3,7]
- *Minimum sequence MRI:* includes axial diffusion-weighted imaging (DWI), fluid-attenuated inversion recovery (FLAIR), susceptibility-weighted imaging (SWI), and time-of-flight magnetic resonance angiography (MRA) of head and neck; helpful in identifying large venous or sinus thromboses[3,7]
- *Lumbar puncture:* useful in ruling out SAH if CT is negative, but clinical suspicion remains high, especially if the patient is febrile; presence of red blood cells is most helpful; opening pressure may also be elevated[1,5,7]
- *Cranial ultrasound:* useful in intraventricular and germinal matrix hemorrhage[3]
- *Catheter angiography:* gold standard; provides precise detail about vascular anatomy, including small vessel disease; superior to MRA and CT angiography for tertiary branches and small cerebral arteries, although it involves an invasive procedure[1,5]

TREATMENT: MEDICAL

Guiding Principles

Specific guidelines for pediatric stroke based on clinical studies are lacking, with the exception of stroke in children with sickle cell disease.

Initial Management

After obtaining a quick history and performing a focused examination, it is important to intervene early as early detection and intervention reduces morbidity and mortality. Initial resuscitation followed by supportive care is important in the early treatment of pediatric stroke. Acute management includes stabilization of cardiorespiratory status, management of comorbidities such as hydration, infections, seizures, and fever, maintenance of blood pressure, circulation, and glucose levels, and monitoring for intracranial hypertension (see Table 5.1).[2]

Ongoing Care

Once stabilized, depending on the etiology, various measures are instituted in the management, with treatment options largely extrapolated from adult data. Areas of medical management to focus on are based on the etiology of the stroke and the associated symptoms (see Tables 5.2 and 5.3). Appropriate therapeutic interventions are essential to a comprehensive management plan and include an interdisciplinary approach, including physical therapy (see Table 5.4), occupational therapy (see Table 5.4), speech therapy (see Table 5.5), and neurocognitive rehabilitation. The role

Table 5.1 Medical Considerations in Acute Management of Pediatric Stroke

CONDITION	SIGNIFICANCE OF MONITORING
Hyperglycemia	This is associated with adverse outcome.
Pyrexia	There is no influence on pediatric stroke unlike in adults.
Blood pressure	Hypertension in the acute period after stroke is associated with worse outcomes. It is important to not allow patients to become hypotensive as this is equally dangerous for patients with pressure-dependent stenosis.
Seizures	Subclinical seizures should be identified and treated.

Table 5.2 Interventions in Pediatric Stroke by Condition[2]

Acute ischemic stroke or hemorrhage: hemicraniectomy	Rare in pediatric AIS, lifesaving in children, with large supratentorial stroke, decided based on each case
Acute ischemic stroke: antithrombotic therapies and endovascular thrombectomy	Aspirin: 3–5 mg/kg/d, most commonly used; LMWH and vitamin K antagonists; oral anticoagulants: dabigatran; anti-factor Xa agents: rivaroxaban, apixaban, and edoxaban being studied in children with venous thrombosis tPA: there is no consensus on use; use may vary based on availability of resources Has a longer poststroke window for intervention; can be used for patients <18 years using adult parameters
Moyamoya: surgical revascularization	Two types: direct: transecting a donor vessel and anastomosing directly to a single recipient cortical vessel; and indirect: vascularized tissues like a vessel, muscle, and pericranium used to stimulate growth of a new vascular network upon contact with the brain
Sickle cell disease: avoid hyperviscosity (maintain hemoglobin S <30%)	Blood transfusion and phlebotomy (if hemoglobin >12 g/dL); hydroxyurea for children with SCD and stroke who cannot or will not be on long-term transfusion, or in low-resource settings where long-term transfusion is not possible; screen for cerebral infarcts with MRI of the brain

AIS, acute ischemic stroke; LMWH, low molecular weight heparin; SCD, sickle cell disease; tPA, tissue plasminogen activator.

Table 5.3 Interventions in Pediatric Stroke by Symptom[2]

SYMPTOM	INTERVENTION
Arousal	Amantadine: dopamine agonist Bromocriptine: dopamine D2 receptor agonist, can also be used in patients with associated paroxysmal sympathetic hyperactivity
Attention	Methylphenidate: noncompetitively blocks reuptake of dopamine and noradrenaline into the terminal by blocking the noradrenaline transporter Dextroamphetamine: dopamine agonist and norepinephrine agonist
Agitation	Quetiapine: antipsychotic used to calm patients and as a sleep aid Behavioral therapy and environmental modifications
Spasticity	Baclofen: GABA agonist used to relax muscles
Behavioral problems, impulsivity	Sertraline Seizure medications such as carbamazepine and valproic acid Behavioral therapy and environmental modifications
Sleep disturbance	Melatonin, quetiapine, trazodone, and zolpidem Behavioral therapy and environmental modifications

GABA, gamma-aminobutyric acid.

and key interventions within each domain in the context of stroke are discussed later in this chapter.

Treatment Controversies

Due to lack of adequate data in the pediatric population, there are considerable controversies in treatment type, timing, and approach.

No Clear Consensus on Timing and Type of Intervention

- There are no clear indications for use of transesophageal echocardiography in children with stroke.
- There is no precise nomenclature in describing intracranial arteriopathies.
- The threshold to screen patients for rare metabolic/genetic causes of stroke in the pediatric population is not clear.

Table 5.4 Physical and Occupational Therapy Intervention and Treatment for Pediatric Stroke

Guiding principles	Prior level of functioning Patient/family goals Clinical presentation Location of injury Neural plasticity
Initial management	General strengthening, ROM, stretching, and weight-bearing[8] Modalities (NMES, FES, and Kinesio tape)[9–11] Orthotic management: splinting[12] and bracing (AFO, SMO, and SaeboStep)[13]
Ongoing care	General upper/lower extremity strengthening, ROM, stretching, and weight-bearing CIMT: a rehabilitation strategy to increase functional use of paretic upper extremity through repetitive and adaptive task practice while the nonparetic upper extremity is restrained, usually by a cast or stockinette; part of a "Therapeutic Package" used to improve overall use and quality of movement in a patient's arm/hand when impairment or weakness is present using the following methods: shaping, task practice, adherence-enhancing, and behavioral strategies, and constraining the less affected upper extremity; with varying protocols that determine the amount of time in cast in and outside of therapy, home practice activities completed while in cast, etc.[14] Modalities (NMES, FES, and Kinesio tape)[9–11] Gait training[15] Orthotic management: splinting[12] and bracing (AFO, SMO, and SaeboStep)[13]
Treatment controversies	"Yes/No" list (compilation of activities that a patient should or should not participate in following a stroke, e.g., "yes" to card/board games and going for a walk, "no" to skateboarding and contact sports)
Additional considerations	Family support and caregiver buy-in Subluxation of the shoulder Contracture prevention

Note: A more detailed explanation of specific physical and occupational therapy techniques can be found in Chapters 20 and 28.

AFO, ankle foot orthotic; CIMT, constraint-induced movement therapy; FES, functional electric stimulation; NMES, neuromuscular electrical stimulation; ROM, range of motion; SMO, supramalleolar orthotic.

Table 5.5 Speech Therapy Intervention and Treatment for Pediatric Stroke

Guiding principles	Prior level of functioning Patient/family goals Clinical presentation Location of injury Neural plasticity
Initial management	Modalities (NMES, Kinesio taping, myofascial release, etc.) Diet tolerance Communication system for basic wants and needs (verbal, low-tech AAC, gestures/sign, etc.) Safety precautions/plans based on cognitive-communication needs Support functional executive functioning, problem-solving, and recall skills
Ongoing care	Modalities (NMES, Kinesio taping, myofascial release, etc.) Sustainable communication system (verbal, high-tech AAC, sign, etc.) Diet tolerance and upgrade to least restrictive diet Repeat imaging for swallow function Compensatory strategies for cognitive-communication needs Recommendations for transition to home/work/school settings Support functional executive functioning, problem-solving, and recall skills
Treatment controversies	NMES for dysphagia Compensation versus rehabilitation
Additional considerations	Family support and caregiver buy-in School accommodations Respiratory status (tracheostomy/ventilator use, PMV appropriateness)

Note: A more detailed explanation of specific speech therapy techniques can be found in Chapters 21 and 22.

AAC, augmentative and alternative communication; NMES, neuromuscular electrical stimulation; PMV, Passy Muir valve.

Secondary Stroke Prevention Strategies Remain Controversial

- Use of steroids in focal cerebral arteriopathy–inflammatory type (FCA-I), anticoagulation for pediatric arterial dissection, and timing/type of surgery for moyamoya are controversial.
- Significance of management of a patent foramen ovale is controversial.
- Consider genetic screening in children with pial arteriovenous fistula, arteriovenous malformations, and cavernous malformations, especially if there is associated family history, multiple or unusually complex malformations, or vascular birthmarks on physical examination.
- In arteriovenous fistulas not related to trauma, screening for the presence of a thrombus should be performed.

ADDITIONAL CONSIDERATIONS

Development of educational programs to improve knowledge and skills in the diagnosis and management of pediatric stroke is key for ED physicians, pediatricians, and emergency medical technicians. Multidisciplinary approach with hematologists, neurologists, cardiologists, pediatric intensivists, and nursing staff caring for this patient population is important for patient care. Systems and pathways, including criteria for consideration of endovascular thrombectomy in children with acute ischemic stroke (AIS) and large vessel occlusion, should be developed. Appropriate referral networks should be established with frontline workers and communities. Ongoing research is important to establish safe guidelines and collect data regarding treatment options.

TREATMENT: PHYSICAL AND OCCUPATIONAL THERAPY, SPEECH THERAPY, AND NEUROPSYCHOLOGY

The Role of the Neuropsychologist, Assessment, and Outcomes

As a member of the pediatric stroke rehabilitation team, the neuropsychologist plays a key role in the evaluation and management of a child's injury from acute care to poststroke, long-term follow-up.

The role of the neuropsychologist varies across the following treatment phases:

- *Acute phase:* The neuropsychologist monitors the recovery of the patient and provides initial feedback and education to the patient, their family, and the treatment team.
- *Postacute phase:* The neuropsychologist provides a brief, targeted evaluation prior to discharge from acute care or inpatient rehabilitation aimed at (a) monitoring the level of recovery, (b) assisting with short-term treatment planning, and (c) supporting reintegration into daily life (e.g., home, school, and the larger community).
- *Long-term follow-up:* After the rate of recovery begins to slow (often between 6 and 12 months post stroke),[16] the neuropsychologist may

provide an in-depth evaluation of the neurocognitive and functional impact of the injury and continue to educate patients and their families and provide support for longer term treatment planning. They also collaboratively work with neurology, physiatry, rehabilitation therapists, school personnel, and caregivers to optimize the patient's treatment course and address evolving socioemotional, behavioral, and cognitive deficits that impact the patient's functioning.

As part of the patient's broader rehabilitation services, the neuropsychologist or another rehabilitation specialist (e.g., speech language therapist or occupational therapist) may provide direct treatments aimed at the patient's neurobehavioral and cognitive impairments.[17–19] In general, cognitive rehabilitation uses four broad treatment approaches,[20] namely (a) compensatory, (b) restorative, (c) ecologically based,[21] and (d) pharmacologic interventions (under the direction of the treating physician).[22] The evidence for these broad interventions is growing but remains limited in the pediatric population.[20,23,24]

Ongoing Care

Factors Predicting Neurologic Outcomes After Pediatric Stroke

A large number of children who have suffered from stroke will continue to have neurologic deficits at long-term follow-up.[25–27] The long-term impact of stroke on a child's cognitive, behavioral, physical, and socioemotional functioning is dependent on multiple factors, including the mechanism of injury; size, location, and severity of the injury; type and timing of treatment following the injury; other diseases or comorbidities; age of the child at the time of stroke; the child's prestroke development and functioning; and time since the injury.[28–31]

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Spinal Cord Dysfunction

6

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GENERAL PRINCIPLES

Definition

Spinal cord injury (SCI) occurs when there is damage to the spinal cord resulting in disruption of sensory and motor signals between the brain and the body.[1] Damage can subsequently lead to temporary or permanent changes in strength, sensation, and autonomic function below the level of injury.

Epidemiology

In the United States, pediatric SCIs comprise <10% of all injuries,[2] and are mostly seen in adolescents (6× greater) and only 3% to 5% in those younger than 15 years.[3,4] Pediatric SCIs can be further classified into traumatic SCI (tSCI) and nontraumatic SCI/spinal cord dysfunction (ntSCI/SCD).[5]

- *Traumatic:* highest global incidence in the United States and Canada (13.2 cases/million)[5]; more likely in males>5 years (~3×)[6]; increased risk in White and Black American populations[7]
- *Nontraumatic:* damage, lesion, or myelopathy, among other terms[8]; paucity of data due to heterogeneous etiologies[9]; estimated incidence: 2.1 cases/million (in North America)[5]

Etiology

- *Traumatic:* The most common causes are motor vehicle collisions (MVCs), followed by falls (higher rates in ages 6–12), violence (including assault and firearm injuries; ages 13–17), sports, and medical/surgical factors.[5,6,10]
- *Nontraumatic:* This is further classified by the International Spinal Cord Injury Data Set[11] into the following:
  - *Congenital and genetic:* Congenital includes spinal dysraphism (see Chapter 7, "Spina Bifida"), skeletal malformations (e.g., foramen magnum stenosis, atlantoaxial instability), and congenital syringomyelia.[11] Genetic includes hereditary spastic paraparesis, spinocerebellar, adrenomyeloneuropathy, other leukodystrophies, and spinal muscular atrophy (see Chapter 11, "Neuromuscular Disorders").
  - *Acquired:* See Table 6.1.

Table 6.1 Acquired SCI/D[11]

Neoplastic (most common cause; 30%–63%; see Chapter 24, "Cancer Rehabilitation")	Primary sites: spinal cord, meninges, and cauda equina
Neuroimmunological (second most common cause; 28%–35%; see Chapter 12, "Neuroimmunological Disorders")	ATM, AFM, and ADEM
Vascular[11–14]	Epidural hematoma Dural AV fistulas AV malformations Ischemic events (fibrocartilaginous embolism, surfer's myelopathy)
Metabolic/toxic[15]	Leukodystrophies (adrenoleukodystrophy, Alexander disease, Krabbe disease, metachromatic leukodystrophy) Mitochondrial diseases (Leigh syndrome, MELAS, MERRF, Kearns–Sayre syndrome, Leber hereditary optic neuropathy) Vitamin deficiencies (biotinidase, vitamin E ataxia, cerebral folate deficiency) Miscellaneous disorders (Friedrich ataxia, Wilson disease, HHH syndrome, arginase deficiency, nonketotic hyperglycemia)
Infectious[16,17]	Bacterial myelitis, spondylodiscitis, spinal tuberculosis, and spinal epidural abscess

ADEM, acute demyelinating encephalomyelitis; AFM, acute flaccid myelitis; ATM, acute transverse myelitis; AV, arteriovenous; HHH, hyperornithinemia-hyperammonemia-homocitrullinuria; MELAS, mitochondrial encephalopathy, lactic acidosis, and stroke-like episodes; MERRF, myoclonic epilepsy with ragged red fibers; SCI/D, spinal cord injury/dysfunction.

Pathophysiology and Mechanism of Injury

The anatomical and biomechanical characteristics of the spine of children are factors in the unique etiologies of pediatric SCI. The pathophysiology of ntSCI is variable and dependent on the etiology.

- *Cervical (≤8 years):* ligament hyperlaxity, shallow and more horizontal facet joints, incomplete ossification, underdeveloped uncinate processes, anterior wedging of the vertebral bodies, larger head to torso

ratio, and weaker neck muscles[6,18,19]; in neonates due to torsional/traction forces[18]
- *Thoracic (older):* use of lap and shoulder belts[6]
- *Lumbar (L2 and L4):* children <60 lbs; lap belt placement above the pelvic brim leads to flexion/distraction injuries[18]

DIAGNOSIS

Risk Factors

Pediatric Anatomy

Cervical spine does not reach adult proportions until 8 to 10 years old. By 10 to 12 years old, an adult and a pediatric spine appear similarly after a traumatic event. In children, the fulcrum of motion is higher (C2–C3).[20]

Rheumatologic Conditions

Juvenile rheumatoid arthritis most commonly affects the cervical axial spine (~25%–80%), and may present with atlantoaxial subluxation/impaction, vertebral body growth changes, and ankylosis.[21] Systemic lupus erythematosus or antiphospholipid antibody syndrome may present with acute transverse myelitis.[22,23]

Other Predisposing Factors

- *Trisomy 21:* Atlantoaxial instability (10%–20%) and myelopathy may develop (1%–2%).[18]
- *Skeletal dysplasia:* This can present as scoliosis, kyphosis, hyperlordosis, vertebral instability, and central stenosis, with increased risk with minimal trauma.[24]

Clinical Presentation

- A majority of tSCIs are incomplete injuries (90.3%); cervical injuries are most common (40.5%; 19.8% high cervical, 20.7% low cervical); high cervical (C1–C4) injuries in ≤5 years[6]
- *Spinal cord injury without radiographic abnormality (SCIWORA):* symptoms without visible fracture or ligamentous instability on plain radiographs or CT scans[25]; MRI findings can be present (90%)[26]; thought to be caused by hyperextension, flexion, distraction, and spinal cord ischemia[27]; 50% of tSCIs present as SCIWORA[10]
- *Concomitant traumatic brain injury (TBI):* 50% to 60% of tSCIs may present with TBI, especially those with cervical and thoracic injuries[28]; more likely to have motor complete injuries and less likely to have lumbar and sacral injuries compared with patients with SCI alone[28]

Symptoms

Symptoms include neck pain, muscle tightness, torticollis, transient or permanent symptoms of radicular pain, or myelopathic symptoms (strength or sensation impairments with bowel/bladder involvement).[29]

Physical Examination

Initial assessment should include evaluation of airway, circulation, and breathing, and a thorough neurologic evaluation. The International Standards for Neurological Classification of Spinal Cord Injury (ISNSCI), known as the American Spinal Cord Injury Association (ASIA) Impairment Scale, is the gold standard for evaluating, classifying, and determining completeness of injury.[30,31]

- This is not shown to be reliable in children <6 years old (the level should be documented as suspected). Children up to 8 to 10 years old may have difficulty with understanding the examination due to poor reliability of the anorectal examination.[31,32] Modified 16-dermatome sensory examination with pinprick and light touch may provide good correlation with the traditional sensory examination.[33]

Diagnostic Evaluation

Laboratory Studies

- See Table 6.2.

Table 6.2 Laboratory Studies

LABORATORY STUDIES	PERTINENT NOTES IN PEDIATRIC SCI/D
Calcium	Related to bone health; hypercalcemia (acute injury), hypocalcemia (chronic)[34,35]
25-hydroxy vitamin D	Related to bone health; hypovitaminosis D (high incidence); should be checked annually[36,37] (goal level: 50–70)
Prealbumin	Measure for nutrition; check and optimize if there are concerns for malnutrition, especially if pressure ulcers present (goal: >21)
Alkaline phosphatase	Often increased in heterotopic ossification; not a reliable screening method[38,39]
Others	Workup for infections; rheumatologic and neuroimmunological causes should be considered for injury in the absence of trauma

SCI/D, spinal cord injury/dysfunction

Radiographic Assessment

Imaging is indicated if the Glasgow Coma Scale is ≤13, with neurologic deficits, posterior midline cervical tenderness, hypotension without explanation, signs of intoxication, history of MVC, fall from greater than 10 feet, or nonaccidental trauma.[29]

- *X-rays:* Initial evaluation may include anteroposterior (AP) and lateral cervical radiographs ± flexion-extension films to evaluate spine stability.[29]

- *CT:* CT has increased sensitivity in detecting SCI acutely prior to surgical intervention if indicated.[29]
- *MRI:* MRI is the gold standard for assessment of ligamentous damage, inflammatory process, vascular, tumor, or other pathologic lesions.[29]

TREATMENT

Guiding Principles

Traumatic Injury

Immediate stabilization aims to reduce injury and preserve neurologic function. The heads of children are large in comparison with their bodies, and the trunk may need to be elevated or the head recessed to prevent unintentional cervical flexion during immobilization.[19,29] Surgery, if indicated, should be performed within 24 hours to minimize secondary injury. Methylprednisolone is no longer routinely recommended.

- *SCIWORA:* Majority of patients are managed conservatively with rigid bracing (×12 weeks) and weaned if MRI is normal and symptoms have resolved.[40]

Nontraumatic Injury

If confirmed to have an autoimmune or infectious etiology, appropriate treatment should be initiated.[41]

Initial Management

Acute Recovery

The goals are to maximize function and independence in mobility and activities of daily living (ADLs) and to educate patients and their families. This should include bracing and equipment needs with assistive and mobility devices.[42]

Autonomic Dysreflexia[43,44]

This classically manifests as pallor, piloerection, vasoconstriction, elevated blood pressure (≥15 mmHg in children; ≥15–20 mmHg in adolescents), headache, blurred vision/visual spots, nasal congestion, bradycardia, cardiac arrhythmias, diaphoresis, and flushing above the level of injury. Young children may present with atypical symptoms (head grabbing, behavioral changes). Normative data are limited in children with SCIs. Management is similar to adults. If systolic blood pressure continues to be elevated (>120 mmHg in <5 years old; >130 mmHg in 6–12 years old; >140 mmHg in ≥13 years old), pharmacologic management (short-acting antihypertensive) should be initiated:

- *Nitropaste:* ½-inch increments for patients <13 years old; 1-inch for ≥13 years (preferred choice)
- *Nifedipine:* 0.25 to 0.5 mg/kg per dose for <13 years; 10 mg per dose for ≥13 years; may repeat every 20 to 30 minutes, as needed
- *Other agents:* captopril and hydralazine; IV medications require ICU

Orthostatic Hypotension

This is more common in tetraplegia.[45] There is a decrease in systolic blood pressure or diastolic blood pressure with transitional movements such as sitting and standing. Interventions include abdominal binder and compression stockings/ACE bandages and pharmacologic management with fludrocortisone or midodrine.

Spasticity/Tone

Initial treatment consists of stretching, positioning, and bracing. Systemic management includes baclofen, tizanidine, or benzodiazepines.[3,46]

Neurogenic Bladder

- Dysfunction may present as upper motor neuron type (UMN; reflexic) or lower motor neuron type (LMN; areflexic). UMN results in detrusor overactivity or detrusor-sphincter dyssynergia and can result in high bladder pressures and reflex voiding. LMN results in atonic urinary sphincter and predisposes patients to urinary leakage.[46] The long-term goal is to maintain kidney health and provide social continence.[46] Maximal bladder capacity is estimated using the Koff equation[47] (until the age of 12): bladder capacity [in mL] = (age + 2) × 30.
- Postvoid residuals (PVRs) should be obtained in patients who can void volitionally. Catheterization is required with incomplete emptying (⅓–½ of maximum bladder capacity). Intermittent straight catheterization (ISC), every 4 to 6 hours, is the preferred method of bladder elimination.[48] Indwelling catheters (suprapubic) can be utilized immediately following injury or in individuals who are unable to perform ISC.[46,48]
- *Medication*: Anticholinergics increase the capacity of bladder storage and filling (oxybutynin, tolterodine, and solifenacin).[48] Alpha-blockers reduce bladder outlet resistance (prazosin, terazosin, doxazosin, tamsulosin, and alfuzosin).

Neurogenic Bowel

Prolonged transit time, increased gastric emptying, and orocecal transit time can occur. UMN results in increased external anal sphincter and rectal tone, and rectal contractility; LMN presents with relaxed external anal sphincter and rectal tone, and decreased rectal contractility.[46,49]

- *Bowel program:* This is performed daily to every other day at a scheduled time to avoid incontinence. Rectal medications (bisacodyl, glycerin, and docusate) are typically integrated to aid in social continence around the age of 3 to 4 years, unless significant constipation is not managed with oral options.
  - *UMN:* Digital rectal stimulation is needed due to increased external anal sphincter and rectal tone. Commonly used medications include surfactant laxatives (docusate sodium), osmotic laxatives (polyethylene glycol, lactulose), and stimulant laxatives (senna,

bisacodyl). Bulk laxatives should be used carefully in younger patients due to difficulties with hydration which may render them inefficient.
  - *LMN:* The goal is firmer stool to reduce incontinence between bowel programs. This may need to be performed one to two times a day. Manual evacuation is typically needed.

Venous Thromboembolism

Children are less likely to develop venous thromboembolism (VTE). Mechanical prophylaxis is recommended for all. Incidence appears to increase during adolescence (13–15 years old or Tanner pubertal stage >2). Postpubertal youth should receive chemoprophylaxis (LMWH), per the Paralyzed Veterans of America guidelines, especially in the setting of a lower extremity or pelvic fracture, unless contraindicated.[50,51]

Immobilization Hypercalcemia

This commonly occurs in adolescent males 4 to 6 weeks (can be seen from 2 weeks to 6 months) following SCI. Symptoms include constipation, nausea, vomiting, polydipsia, polyuria, decreased appetite, fatigue, polydipsia, and changes in behavior; treated with IV fluids, furosemide, glucocorticoids, and bisphosphonates.[34]

Maintenance Care (Chronic Management)

Neurodevelopmental Optimization

Activity-based interventions promote neuroplasticity below, above, and across the level of injury. Locomotor training has shown improved trunk control and neuromuscular capacity in acute and chronic SCIs.[52]

- *Spasticity/tone:* This may lead to joint contractures and musculoskeletal deformities. Long-term management is most effectively provided with mechanical and pharmacologic interventions.
  - *Mechanical:* This includes stretching and bracing. Daily activity through standing programs and electrical stimulation (functional electrical stimulation [FES], therapeutic electrical stimulation [TES]) may synergistically assist in the management of spasticity and tone.[53]
  - *Medications:* These include oral baclofen, tizanidine, or benzodiazepines.[54] Monitoring liver function tests (LFTs) is important with tizanidine (risk of transaminitis). Botulinum toxin chemodenervation may be used for localized spasticity management. Intrathecal baclofen may be an option.
- *Syrinx/tethered cord:* This may be present in the area of initial injury (mostly after traumatic or tumor-related).[55] It is an accumulation of fluid in a cyst within the spinal cord (caused from degeneration). Expansion may lead to neurologic decline. Significant scarring tissue around the cord may lead to tethering with growth. Presenting signs typically include new hyperreflexia (Hoffmann sign) or weakness in

the upper limbs, early or quickly progressive scoliosis, and changes in bowel/bladder function. Imaging is typically indicated. Surgery may be needed.

- *Concomitant TBI:* This is also pertinent in patients with ntSCI/SCD from disease processes such as acute demyelinating encephalomyelitis (ADEM), in which the presenting widespread inflammation throughout the central nervous system leads to brain and spinal cord dysfunction. Cognitive screening is essential.
- *Sexual function:* Typical in males, majority will recover some erectile function, and approximately half of patients may be able to have orgasms, despite completeness. Chronic ejaculatory dysfunction may be typical. Mechanical and pharmacologic interventions may assist with erectile dysfunction (sildenafil).[56] It is essential to include sexual health education for all youth.
- *Neurogenic bowel:* Long-term persistent chronic constipation or fecal incontinence can be present despite efforts. Advanced procedures include the following:
  - *Irrigation systems:* Use of cone enemas is reported to be safe in spinal cord injury/dysfunction. Rectal irrigation system trial may require an inpatient stay and documentation.
  - *Surgical interventions:* There are limited reports on patients with spinal cord injury/dysfunction, although they may be safe and efficient. Options include antegrade colonic enema (ACE) stoma or colostomy.
- *Neurogenic bladder:* ISC is typically initiated at the age of developmentally appropriate social continence efforts (potty-training). ISC is not indicated in young children (<3 years) without history of recurrent urinary tract infections and high-pressure bladder/reflux. Patients >6 years old may be independent with ISC.
  - Pediatric urologist care should be established. Guidelines recommend surveillance with renal ultrasound at least annually (red flags: hydronephrosis, nephrolithiasis, other high-pressure bladder clinical stigmata), voiding cystourethrogram (VCUG) (red flag: reflux), or urodynamic testing. UMN bladders develop decreased compliance. Storage capacity should be followed as the child ages. Procedures may further assist with social continence.
    - *Intravesical botulinum toxin:* for high-pressure bladders or inability to improve bladder storage capacity, poor tolerance to anticholinergics
    - *Urinary diversion surgery (with and without bladder augmentation):* leads to improved quality of life during adulthood[57]; bladder augmentation linked to increased risk of developing bladder cancer at an earlier age; surveillance cystoscopy recommended starting 10 years after surgery; risk needs to be discussed during transition to adult-based services; transition to adult urologist experienced in complex reconstruction given potential future complications

  - *Suprapubic tubes (SPT):* patients develop chronic cystitis and bladder stones; have generally low bladder infection rates[58,59]; will often lead to a small and spastic bladder and decreased compliance and affect storage capacity

- *Pressure ulcers:* These are less frequent in children. Common locations include the sacrum (22.1%), heels (14.8%), ears (12.9%), elbows (10.6%), and buttocks (6.8%). Extrinsic risk factors include friction and shear forces and prolonged pressure over bony prominences. Nutrition is integral to prevention and management.[60] Turning every 2 to 3 hours while in bed and weight shifts every 15 to 30 minutes for 15 to 30 seconds, depending on the level of injury, are recommended.[61] Twice-daily skin checks should be performed.[10] Children may require cues, such as timers or charts to remind them. Adults who provide supervision should be trained in pressure relief, early recognition and treatment, moisture avoidance, and potential risks.[10,60,62]
- *Orthopedic complications:* Injury prior to puberty increases the risk.
  - *Scoliosis:* This is the most common orthopedic complication (100% if prepubertal onset, ~4× will require spinal fusion).[63,64] Thoracolumbosacral orthosis can delay the need for surgery. Surgery is indicated at age >10 years old, Cobb angle >40°, rapid curve progression, and pain or functional problems.[3,64]
  - *Hip dysplasia (subluxation/dislocation):* This is the second most common complication (>90% if the SCI onset is ≤10 years old).[3,64,65] Surgical treatment is controversial; spastic hip instability should be treated similar to cerebral palsy; flaccid paralysis should be treated similar to spina bifida.[65]
  - *Heterotopic ossification (HO):* This is not typically found in children with SCI (3%), although they may have delayed presentation.[65,66] This is most common in the hip. Risk factors include motor complete lesions,[3,66,67] cervical or thoracic injury, spasticity, pelvic trauma, pressure ulcers, tracheostomy, and infections (pneumonia and urinary tract infections).[38,67,68] It is visible on x-rays in 3 to 6 weeks or as early as 2 weeks on a three-phase bone scan. Alkaline phosphatase increases 6 to 12 weeks after injury.[38]
  - *Pathologic fractures:* Dual-energy x-ray absorptiometry (DEXA) is evaluated via z-scores rather than t-scores, as peak bone mass is not reached until just before the age of 30.[37] Childhood-onset SCI patients have 40% less bone density than age-matched controls. Bone mineral density (BMD) fracture threshold in children is unknown.[3,69] The prevalence is 10% to 20%, especially in the hips,[65] and is more common in complete injuries and those with paraplegia.[69] There is increased risk of fractures if SCI onset is <16 years old, greater time since injury, body mass index <19, and previous pathologic fracture.[69] Preventive measures include education, monitoring, weight-bearing activities, proper equipment for transfers, avoiding increased immobilization, and screening for adequate calcium and vitamin D intake.[3,65] Surgical intervention for hip fractures

is controversial; however, if this is pursued, sideplate with lag screw and derotational screw is suggested.[65]
  - *Joint contractures:* These are frequent (30%–50%) and lead to difficulty with sitting and standing tolerance, and skin hygiene.[65] Surgical intervention can be considered after discussion on functional goals and strong commitment to postoperative therapy.
  - *Tendon transfers:* These are often used to restore some upper limb function (active elbow extension, wrist extension with tenodesis grip, or re-establish pinch) with the goal of being able to perform ADLs without requiring adaptive equipment.[3,70]
- *Respiratory insufficiency:* This is a significant source of morbidity and mortality in cervical SCIs.[3,71] Initial ventilator support can often be weaned, but those who cannot be weaned often require long-term support with tracheostomy (high tidal volumes) for chronic respiratory insufficiency.[3,71] Sleep studies may be indicated due to high risk of sleep-disordered breathing.[72] Aggressive secretion management can be done with insufflation-exsufflation machines (such as CoughAssist), percussion vests, and/or manual percussion and postural drainage.[73,74] Education can be provided on how to perform pulmonary hygiene, including manually assisted cough.[71] Annual influenza and pneumococcal vaccines are recommended.[74] Pacing (diaphragmatic or phrenic) can be considered.[75] Children may take longer to wean from ventilation despite pacing.

Treatment Controversies

Steroids and Traumatic Spinal Cord Injury

Steroids are not recommended for acute tSCI. Studies showed increased risk of pneumonia, ICU length of stay, and mechanical ventilation duration, in addition to other adverse reactions.[76] Steroids may still be indicated for ntSCI/SCD.

Timing for Surgical Stabilization and Traumatic Spinal Cord Injury

Management of unstable vertebral fractures may vary, with specific recommendations provided for growing children (pediatric instrumentation, growth potential, size of implants). Vertebral fractures from gunshot wound-related (GSW) cervical tSCIs are typically nonoperative (even if the midcolumn is involved) due to concerns of cerebrospinal fluid leak, infection, and prolonged stay.[77,78] The potential benefit of surgical exploration and stabilization to avoid further neurologic injury when presenting neurologic function is considered.[79]

Epidural Stimulation

Studies have reported compelling outcomes with regard to muscle activation or bladder control.[80,81] These are available only through clinical trials (adults). Long-term complications include encapsulation, migration, infection, and fibrosis, which need to be further weighted for use in youth.[82]

Stem Cells

There is a multitude of reports around the world with some intriguing data. Transplantation trials for SCI are in their infancy. There is no consensus regarding cell type, dose, route, timing for transplantation,[83] and the potential need for additional interventions.[84] Safety concerns abound due to lack of standardization, including potential for worsening SCI with intraspinal transplantation.

Electrical Stimulation and Lower Motor Neuron

There have been small studies that have reported potential for motor output improvement with long-term use of TES/FES in patients with LMN lesions.[85,86]

ADDITIONAL CONSIDERATIONS

Long-Term Prognosis

- *Ambulation potential:* Greater likelihood of ambulation in children after SCI is associated with younger age at time of injury, total ISNSCI motor score, and ASIA Impairment Scale (AIS).[87]
- *Functional recovery:* There are very minimal data in children.

Psychosocial

- *School/educational:* Elementary school students are more likely to participate in school-based therapy due to age-expected independence. Older students may require modifications to assignments or increased time to complete tasks per Rehabilitation Act 504.[88] Teachers have rated student performance to be on par or greater than peers within primary and secondary school students.[88]
- *Sexual health:* Hormonal birth control should not be used in smokers and in those with circulatory or cardiovascular disease or injury onset <1 year. Depot-medroxyprogesterone acetate can negatively affect BMD. Intrauterine devices carry an increased risk of pelvic inflammatory disease.[89]
  - *Fertility considerations:*
    - *Females:* Return of menses is usually 6 months post injury; fertility is not affected.[90] Risks of thrombosis increase with high-dose hormonal stimulation during in vitro fertilization, but not in low dose. Pregnant females can deliver vaginally.[91] Specialized care is recommended to avoid complications from autonomic dysreflexia.
    - *Males:* Biological children are possible if semen can be obtained; motility is often impaired and analysis can be helpful.[89]
- *Quality of life:* Individuals with pediatric-onset SCI are less likely to live, be married, or drive independently. They have similar rates in obtaining college degrees.[92] As compared with adult-onset SCI, individuals with pediatric-onset SCI have higher employment rates. Employment is associated with higher levels of education, functional independence, and community mobility, and fewer medical complications.[93]

- *Transition to adulthood:* Compared with adult-onset, adults with pediatric-onset SCI are noted to have less pain, greater functional independence, and less overall physician visits in the year prior, with greater values for participation in social, occupational, and physical activities, although below normative values for nondisabled individuals.[94]

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Spina Bifida

7

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GENERAL PRINCIPLES

Definition

The term *spina bifida (SB)* encompasses both open and closed spinal neural tube defects (NTDs) but most commonly refers to myelomeningocele (MMC), an open spinal NTD with a lesion draining cerebrospinal fluid (CSF), with exposed neural tissue and malformations of the brain and spinal cord.[1–4] A closed spinal NTD typically has a lesion with no CSF leakage and sometimes contains malformations of the spinal cord alone.[4]

Epidemiology

- 222,000 neonates born annually with a NTD, the second most common birth defect[5,6]
- 33.86 cases per 100,000 live births in countries with mandatory folic acid (FA) fortification and 48.35 cases per live births in countries without FA fortification mandates[7]
- *United States:* overall prevalence 3.17 cases per 10,000 live births, 2.73 cases per 10,000 live births in non-Hispanic blacks or African Americans, 3.09 per 10,000 live births in non-Hispanic whites, and 3.80 per 10,000 live births in Hispanics[8]

Classification

The classification of spinal NTDs as either open or closed is of practical value in understanding its pathology and clinical implications.[4]

- *Open NTD:* fluid-filled sac leaking CSF, with protruding neural elements
  - Results in lower extremity weakness and neurogenic bowel and bladder[4]
- *Closed NTD:* closed sac without involvement of neural elements
  - Minimal to no neurologic involvement or weakness[4]

Table 7.1 outlines the clinical classification of the various forms of open and closed spinal NTDs.[4]

From McComb[4]:

Table 7.1 A Practical Clinical Classification of Spinal Neural Tube Defects

Open neural tube defect
Myelomeningocele
Myeloschisis
Hemimyelomeningocele
Closed neural tube defect
Meningocele
Posterior: lumbar, sacral, and thoracic
Posterior: cervical and limited dorsal myeloschisis
Anterior: sacral
Myelocystocele
Lipomatous malformation
Abnormal filum terminale
Congenital dermal sinus
Split cord malformation
Neurenteric cyst
Neural tube defect associated with caudal regression

Etiology

Spinal NTD may develop due to failure in the gastrulation, preneurulation, neurulation, or postneurulation stages of embryological development.[4] Primary neurulation begins at approximately 18 days postfertilization and is complete by 26 to 28 days postfertilization, with failure during this stage resulting in an open NTD.[9] Failure during secondary neurulation, where the distal portion of the neural tube forms, results in closed or skin-covered lesions and often presents with tethering of the spinal cord and ectopic lipomatous material.[9]

DIAGNOSIS

Risk Factors

Folate status is the best known risk factor influencing NTD development, and FA supplementation has been shown to have a 72% preventive effect.[2,10] Approximately 30% of NTDs remain resistant to FA supplementation.[10] Supplementation with 400 mcg of FA daily at least 1 month prior to conception is recommended for women with no prior risk factors, while women with a previously affected pregnancy or personal history of NTD should take 4,000 mcg beginning at 3 months prior to

conception.[11] *Vitamin B_{12} deficiency* is also a potential risk factor for the development of a NTD.[10] Certain medications that alter folate metabolism increase the risk of NTD, including antiepileptic drugs (AEDs), methotrexate, sulfamethoxazole-trimethoprim, antacids, sulfasalazine, and rifampin.[10] As well, vitamin A has been implicated as a risk factor.[10] Other medical conditions known to increase risk include diabetes mellitus, substance abuse, and chronic illness.[12] Genetic factors including female sex, Hispanic descent, Irish descent, prior pregnancy affected with a NTD, consanguinity of parents, and single-gene disorders have also been identified as risk factors.[10,13]

Clinical Presentation

Ultrasound (US) is used for noninvasive diagnosis of MMC prenatally, detecting 90%–98% of cases during the second trimester, although it rarely detects closed lesions in utero.[2] Features on US, such as ventriculomegaly and small biparietal diameter for gestational age, can also aid in prenatal diagnosis.[2,14]

Laboratory Studies

Elevated alpha-fetoprotein and acetylcholinesterase have been found in the amniotic fluid samples from pregnancies with MMC.[2] However, due to a 1% chance of miscarriage and with US having greater sensitivity and specificity, biochemical screening for MMC is only utilized in cases where maternal obesity limits US examination.[2]

Physical Examination

Initial functional motor level assessment allows for anticipatory guidance regarding an infant's long-term motor development.[15] Motor level can be assessed based on observed movements of the infant or manual muscle testing as it becomes more reliable after the age of 5.[13] In MMC, function is typically related to the level of the lesion in the spine (Table 7.2).[13] Infants may also present with impaired swallowing or airway protection due to brainstem compromise requiring urgent evaluation.[16]

Radiographic Assessment

Imaging modalities including head US and brain MRI to assess for hydrocephalus; brain and spine MRI to assess for Arnold Chiari II malformation, tethered cord, and syringomyelia; renal US to assess for neurogenic bladder; and x-rays to assess for scoliosis and other orthopedic abnormalities are common throughout childhood.[3]

TREATMENT

Guiding Principles

Management of patients with MMC can best be addressed by a multidisciplinary team. Coordinating care can be challenging due to the medical

Table 7.2 Rehabilitation Needs By Functional Motor Level

REHABILITATION NEEDS BY FUNCTIONAL MOTOR LEVEL				
FUNCTIONAL MOTOR LEVEL	**EXPECTED MUSCLE FUNCTION**	**FUNCTIONAL MOBILITY**	**EQUIPMENT USE**	**ORTHOTIC USE**
Thoracic	Abdominal, paraspinal, quadratus lumborum	Nonfunctional ambulation/ standing during therapy, school, or at home, wheelchair for mobility	Standing frame, wheelchair, parapodium	Trunk-hip-knee-ankle-foot orthosis
High lumbar (L1L3)	Hip flexion, hip adduction	Limited household ambulation, wheelchair for mobility	Wheelchair, walker, forearm crutches	Reciprocating gait orthosis Hip-knee-ankle-foot orthosis
Midlumbar (L3L4)	Knee extension	Household, limited community ambulation	Wheelchair, walker, forearm crutches	Knee-ankle foot orthosis
Low lumbar (L4L5)	Hip abduction, knee flexion, ankle dorsiflexion, ankle inversion, toe extension	Household and community ambulation, wheelchair for long distances	Wheelchair, forearm crutches	Ankle-foot orthosis
Sacral (S1S2)	Hip extension, ankle plantar flexion, ankle eversion, toe flexion	Community ambulation	–	Supramalleolar foot orthosis, foot orthotic

Source: Apkon SD, et al. Advances in the care of children with spina bifida. *Adv Pediatr*, 2014;61(1):33–74.

complexities of the disease, as well as the economic and sociocultural barriers.[3,14] The goal of treatment is to preserve, maintain, or improve function while managing the complications of the condition.

Initial Management

- Following prenatal diagnosis, parents should have the opportunity to determine whether they may qualify for intrauterine repair (see the "Prenatal Versus Postnatal Closure" section).[3,17]
- Following delivery, back closure should occur within 48 hours, with subsequent ongoing evaluation for an increase in ventricle size and head circumference or signs of hydrocephalus.[18]
- Assess for symptomatic Arnold Chiari II malformation, where elongated cerebellar tonsils displaced inferiorly can lead to obstructive hydrocephalus and pseudobulbar palsy, causing an inability to regulate breathing or difficulty swallowing.[4]
- Evaluate for neurogenic bladder including obtaining renal/bladder US, urodynamic testing, and serum creatinine within the first 3 months of life, with initiation of clean intermittent catheterization (CIC) and antimuscarinic therapy for treatment of mixed incontinence when test results indicate.[3]
- Perform hip US to evaluate for subluxation or dislocation. The spine and feet are also examined for deformities, although interventions such as Ponseti casting or kyphectomy are usually deferred until after initial hospital discharge.[19]

Ongoing Care

Habilitation and Rehabilitation

Throughout childhood, individuals with SB, and MMC in particular, should be reassessed to determine what medical interventions, therapy services, orthotics, durable medical equipment (DME), and community and educational resources will best support their development or address new-onset impairments.[6] Therapy services typically begin during the initial hospital stay and include physical therapy, occupational therapy, and speech language pathology as appropriate.[6] Infants with MMC are discharged from the hospital with a home exercise program to maintain range of motion in all affected parts of the body and are typically referred for early intervention services. These services become school-based at the age of 3 and are supplemented by outpatient therapy as needed throughout infancy and childhood.[3] Functional motor level, determined at the time of initial evaluation, should be reassessed at each visit using standardized assessment tools in order to provide appropriate clinical management and guide orthotic and DME prescription (Table 7.2).[13]

Orthopedics

Commonly encountered orthopedic and musculoskeletal problems in children with MMC include scoliosis, kyphosis, hip subluxation, contractures, tibial torsion, and foot deformities.

Kyphosis and Scoliosis

Kyphectomy may be needed if kyphosis is present at birth to facilitate skin closure. Scoliosis is generally proportional to the level of neurologic impairment and is more common in those who are nonambulatory.[19–21] Scoliosis screening is by clinical examination and monitoring with supine x-rays until the patient is able to sit for x-rays, with repeat x-rays every 1 to 2 years or when indicated clinically.[19] Rapid development or progression of a spinal curve may indicate a tethered cord or syrinx.[22,23] Body casting and bracing have not been shown to be effective in the management of neuromuscular scoliosis and may increase the risk of skin breakdown in children with insensate skin.[24] There is no current consensus on any specific surgical intervention or on timing of intervention for scoliosis.[19]

Hip Subluxation

Hip subluxation and dislocation are common in patients with MMC.[25] Gait analysis studies have shown that hip contractures have a more negative impact on ambulation than hip subluxation or dislocation.[25]

- Neither routine hip surveillance nor surgery is currently recommended for hip dislocation.[19] Potential exceptions are a unilaterally dislocated hip and infants with hip instability in low lumbar or sacral lesions.
- Management is on a case-by-case basis and consideration may be given to using a rigid abduction orthosis.[19]

Contractures

Lower extremity contractures can limit ambulation and function and may be amenable to contracture release. These can be at the hip, knee, or ankle.

- Consider gait analysis before proceeding with surgical intervention for low lumbar and sacral lesions with atypical gait patterns or deterioration of gait once syrinx and tethered cord have been ruled out.[19]

Tibial Rotation

Tibial rotation may contribute to gait abnormalities in children with MMC. Twister cables are often trialed to support ambulation, although they can be cumbersome to children and families.[26,27] For external tibial rotation interfering with motor development or bracing, derotation osteotomies can be very effective in improving gait.[28]

Foot Deformities

Foot deformities are common in MMC[29] and are managed both non surgically (serial casting, including Ponseti casting for clubfoot) and

surgically. The *Guidelines For the Care of People With Spina Bifida* provides recommendations for orthopedic management within discrete time frames.

Neurogenic Bladder

Neurogenic bladder affects the majority of infants and children with MMC. The goals of urologic management include maintaining normal renal function, guidance through the stages of continence, and ultimately development of independence in bladder management.[3]

- CIC and antimuscarinic therapy for treatment of mixed incontinence are initiated when test results indicate. Surveillance evaluations are repeated throughout childhood and in adulthood.[3] The most prescribed antimuscarinic drug as first-line therapy for detrusor overactivity in children is oxybutynin.[30]
- Intravesical injection of botulinum A toxin is an option for patients who fail antimuscarinic therapy.[31]
- The goal of surgical management for neurogenic bladder is to protect renal function, with the secondary goal of helping patients achieve continence.[32]

Neurogenic Bowel

Children with MMC experience neurogenic bowel, with constipation and incontinence the two most common issues.[33] Independence with toileting for patients with SB is of utmost importance and has been shown to improve quality of life and family functioning and to reduce caregiver depression and anxiety.[34]

- Increasing fiber and fluid intake is effective in improving stool consistency and frequency in typically developing children.[34]
- Oral medications including osmotic (polyethylene glycol and lactulose) or stimulant (sennoside and bisacodyl) laxatives are frequently used and should be given at the same time each day to establish a predictable pattern. Dosing is based on stool consistency, frequency, and degree of incontinence.[34]
- When oral medications are not able to produce social continence, interventions including digital disimpaction, suppositories (glycerin and bisacodyl), and enemas (sodium phosphate, glycerin, bisacodyl, mineral oil, docusate-based, and cone enema) are often recommended.[34]
- If warranted, a surgically constructed catheterizable channel allows for administration of an antegrade continence enema and has success rates of between 69% and 93%.[34]

Skin Integrity Management

Skin breakdown is present in up to 82% of patients with MMC. Commonly impacted areas include the feet, sacrum, ischium, and greater trochanter.[35]

Risk factors include higher level of lesion, presence of a shunt, wheelchair use, urinary incontinence, recent surgery, above-knee orthopedic procedures, and male sex.[36] Parents should be taught to inspect the skin, especially over weight-bearing surfaces and areas of decreased sensation, with children becoming increasingly involved in skin surveillance as they age.[36]

Cognitive Assessment

Children with MMC should be monitored closely for physical, cognitive, communication, and social development. Individuals who have more severe hydrocephalus, repeated shunt malfunctions, and higher lesion levels have more severe neuroanatomical brain malformations and higher rates of intellectual disability and poorer outcomes.[37,38] A full neuropsychological assessment is recommended to document cognitive functioning. Typical areas of difficulty include construction or integration of information (math, problem-solving, and reading comprehension), use of language in context, visual-spatial reasoning, complex procedures with multiple steps, algorithms, problem-solving, and attention.[39–46] Medications for inattention may be effective at lower doses, although children with MMC do not appear to respond as robustly as children with attention deficit hyperactivity disorder.[47]

Obesity Management

Individuals with MMC have higher rates of overweight and obesity in childhood and adulthood compared with the general population, placing them at risk of metabolic syndrome, cardiovascular disease, type 2 diabetes,[48] and sleep apnea,[49] and reduced mobility, independence, and quality of life.[48] The higher risk of obesity is likely related to multiple factors, including decreased lean body mass,[50] limited intake of fresh fruits and vegetables due to potential cross-reactivity with latex,[48] decreased physical activity,[51] slower linear growth,[13] and lower resting energy expenditure.[52,53] Parents should be cautioned against using food as a reward or positive reinforcement.[54] Noncaloric fluids should be used for hydration. Recreation and physical activity at the community level should be encouraged.[55] Children with MMC over the age of 6 should engage in 60 minutes or more of physical activity each day, with at least 3 days per week of vigorous aerobic activity and with muscle and bone strengthening activities on at least 3 of those days.[56]

Endocrine Management

Short Stature

Compared with their peers, children with MMC have decreased growth of the lower body segments, leading to short stature.[13,57] Around 30% of children with MMC have growth hormone deficiency.[13] If growth hormone deficiency is noted, families and providers should take part in a risk–benefit discussion regarding treatment with human growth hormone.[57]

Precocious Puberty

Children with SB and hydrocephalus are at increased risk of precocious puberty.[58] Concerns about precocious puberty should be referred to a pediatric endocrinologist[59] for consideration of treatment of central precocious puberty.[60]

Self-Care

Progression toward independence in self-care should be a common goal for individuals with SB, their parents, and the multidisciplinary team. This process begins in childhood and should continue through adolescence.

- Incremental involvement of the child in self-management activities and transition from the parent performing to the child performing with parental oversight and to the adolescent performing without parental oversight is recommended.[61]

Sleep Assessment

Sleep-related breathing disorders (SRBDs), including central apnea, periodic breathing, obstructive apnea, and central hypoventilation, are common in people with MMC. SRBDs are associated with significant neurocognitive, psychologic, metabolic, immunologic, and cardiovascular consequences, and even death.[62] Patients with MMC have been noted to have absent arousal responses to hypoxia and hypercapnia and absent ventilatory responses to hypoxia and hypercapnia.[63]

- Overnight polysomnography in all children with MMC is recommended regardless of whether they are symptomatic.[3]

Treatment Controversies

Prenatal Versus Postnatal Closure

Recent follow-up results of the Management of Myelomeningocele Study (MOMS) of prenatal neurosurgical intervention for MMC reported that children in the prenatal surgery group walked without orthotics or assistive devices more often, had improved gross and fine motor function, had lower rates of hindbrain herniation, Chiari II malformations, and hydrocephalus, had fewer shunts placed for hydrocephalus, and among those with shunts fewer shunt revisions, better quality of life, and reduced negative impact on families than those in the postnatal group.[64] However, in the initial MOMS study, there were higher rates of maternal morbidity and premature delivery, and an increased risk of invasive care and obstetric complications in future pregnancies.[65] Subsequent studies and refinement of surgical techniques appear to have reduced the rates of prematurity and maternal morbidity.[66,67] There are also challenges with widespread implementation of prenatal surgical repair due to the relatively small number of centers performing the procedure, as well as continued evolution of techniques, making ongoing follow-up study more difficult.[17,68]

Treatment of Hydrocephalus

Although ventriculoperitoneal shunt (VP) remains the gold standard of management in hydrocephalus, with approximately 80% of patients with MMC typically requiring VP shunt to treat hydrocephalus,[69] endoscopic third ventriculostomy/choroid plexus coagulation (ETV/CPC) is being explored as a promising technique.[23] There is also controversy and variability with regard to the threshold for surgical intervention for hydrocephalus. Tolerance of larger ventricles has reduced VP shunt rates at some centers. Although the long-term impact on cognitive development is not yet known, initial outcomes indicate limited negative impact in the short term, with the benefit of avoiding the known long-term morbidity of ongoing VP shunt management.[70]

Delivery Method for Children with Myelomeningocele

There is no current evidence clearly indicating the superiority of vaginal delivery or cesarean section for children with MMC.[68,71] However, infants that undergo intrauterine repair should be delivered by cesarean section.[68]

ADDITIONAL CONSIDERATIONS

Transition to Adult Care

The primary goal of transitioning patients from pediatric to adult care is to optimize lifelong functioning through delivery of quality, developmentally appropriate healthcare.[3] Adolescents and young adults with SB have increased hospitalizations for conditions such as urinary tract infections and skin breakdown and have more difficulty accessing healthcare services.[3] They are also less likely to leave home, attend college, become employed, or have romantic relationships.[3] Positive predictors of a successful transition to adulthood include higher executive functioning and socioeconomic status, intrinsic motivation, and parental fostering of independence.[3]

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Movement Disorders

8

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GENERAL PRINCIPLES

Pediatric movement disorder is an emerging specialty within child neurology. Abnormal movements are often encountered following injury, requiring rehabilitation professionals to characterize and treat these disorders. In children, primary movement disorders are often due to underlying genetic causes, while secondary disorders are typically a result of structural brain injuries, metabolic abnormalities, or medication side effects. Movement disorders are classified into two major categories: hyperkinetic and hypokinetic disorders. Hyperkinetic disorders are more common in children than in adults and include chorea, dystonia, myoclonus, tremor, and tics. Parkinsonism is the most common hypokinetic disorder; however, it is less frequently seen in children. This chapter provides a brief overview of common pediatric movement disorders, their causes, and management.

CHOREA

- Chorea is characterized by involuntary, nonrhythmic series of movements that flow continuously from one body part to another and can vary over seconds, in type and in location.
- Patients often incorporate chorea into voluntary movements; thus, it is less functionally limiting than other movement disorders.
- Athetosis is a variant that is described as slow, writhing movements.
- Ballismus is another variant, with large amplitude, forceful, flinging-type movements.
- Chorea typically follows injury to the caudate or subthalamic nuclei[1]; however, the differential for chorea is broad and includes structural lesions, metabolic abnormalities, medication side effects, and autoimmune conditions.

Etiology

- Structural lesions
  - Injury to the basal ganglia (often the caudate) secondary to ischemia, hemorrhage, tumor, or demyelinating lesion
  - *Ballismus:* typically, but not uniformly, follows lesions of the contralateral subthalamic nucleus[1]
- Metabolic abnormalities

- Hypoglycemia/hyperglycemia, hyponatremia/hypernatremia, hyperthyroidism, uremia, and (very rarely) hypoparathyroidism
- Check comprehensive metabolic panel, thyroid panel, and parathyroid hormone

- Drug-induced
  - Dopamine agonists (DA), levodopa, anticonvulsants (phenytoin, carbamazepine, valproate, gabapentin), anticholinergics, or lithium
  - Should resolve with discontinuation of the offending agent; if multiple medications involved, remove one at a time, starting with the most recently added medication
- Autoimmune
  - Sydenham chorea (most common cause of acute-onset chorea), lupus, antiphospholipid antibody syndrome, *N*-methyl-D-aspartate encephalitis (especially following herpes simplex virus encephalitis), and paraneoplastic syndromes
  - Can check antistreptolysin, throat culture, and anti-DNase B; these have low sensitivity/specificity as symptoms may present weeks to months after streptococcal infection, but should still be checked; have a low threshold for sending cerebrospinal fluid/serum autoimmune panels

Treatment

- Treatment options include:
  - Dopamine antagonists such as tetrabenazine (side effects: parkinsonism, depression, akathisia) or benzodiazepines (clonazepam)
  - Neuroleptics, but are used less frequently due to risk profile, including tardive dyskinesia
  - Valproic acid, carbamazepine, and steroids found to be effective in Sydenham chorea and may also help in other forms[2]
- Treatment may not be needed if movements are not bothersome to the patient or interfering with activities of daily living.

DYSTONIA

- Dystonia is characterized by co-contraction of agonist:antagonist muscles causing twisting and contorting movements, abnormal postures, or both. It can be sustained or intermittent, but is typically triggered by specific movements.
- Distinguishing features include patterned movements, lack of premonitory sensation, lack of voluntary suppressibility, triggered by specific voluntary movements, and identification of a *geste antagoniste* ("sensory trick" sometimes identified by patients that reduces severity of dystonia for a few minutes).[2]
- Distribution can be focal (single body region), segmental (two or more contiguous regions), multifocal (two or more noncontiguous regions), hemidystonia (involvement of half of the body), or generalized (trunk plus two other body regions).
- It is primarily caused by damage to the basal ganglia, but also to the thalamus, brainstem, parietal lobes, cerebellum, and white matter.[1]

Etiology

- Genetic
  - 50+ genes that can lead to dystonia have been identified, from a wide range of biological pathways. Some of these inherited disorders have specific treatments; thus, early identification is important.
  - Genetic testing should be considered if (a) dystonia presents in isolation, with no other neurologic symptoms; (b) MRI suggests a focal brain degeneration or deposition (e.g., iron or manganese in the globus pallidus); or (c) history and/or MRI are not convincing for structural brain injury (e.g., a "soft call" of cerebral palsy).
- Structural abnormality
  - MRI demonstrating injury to the basal ganglia (commonly putamen) or thalamus[1] due to hemorrhage, infarct, or hypoxic injury
- Drugs/toxins
  - Dopamine receptor antagonists (antipsychotics, antiemetics), antidepressants (selective serotonin reuptake inhibitors), and exposures such as carbon monoxide, cyanide, or methanol
- Autoimmune
  - *N*-methyl-D-aspartate receptor encephalitis; less commonly acute disseminated encephalomyelitis (ADEM) or multiple sclerosis; accompanies signs/symptoms of encephalitis: confusion, obtundation, seizures, or behavioral changes

Treatment

- *Levodopa:* should be trialed in all children with dystonia; low doses (1 mg/kg/d) may be sufficient, but for an adequate trial dose should be titrated over weeks to 10 mg/kg/d; if no response at 10 mg/kg/d after 1 month, can wean off and conclude that dystonia is nondopamine-responsive
- *Anticholinergics (trihexyphenidyl):* start at .05 mg/kg/d, increase weekly to .75 mg/kg/d, divide BID or TID
- *Baclofen:* most helpful in patients with coexisting spasticity and can be used intrathecally or enterally
- *Benzodiazepines:* typically less efficacious than other medications listed

MYOCLONUS

- Quick jerk, involuntary, irregular, usually variable in location and nonsuppressible
- Can be physiologic (benign), epileptic, or a movement disorder, and can be primary or secondary (symptomatic of an underlying injury/disorder)
- Origin can be cortical, subcortical, brainstem-spinal cord, or peripheral[3]
  - *Cortical:* originates from the sensorimotor cortex, typically action-induced or stimulus-sensitive, and is often multifocal (distribution parallels the motor homunculus)

- *Subcortical:* originates from the basal ganglia or thalamus; often stimulus- or action-sensitive, often generalized, and more pronounced in the axial muscles
- *Brainstem-spinal cord:* originates from the brainstem or spinal cord, often associated with a focal lesion, usually with slower frequency, and can persist in sleep
- *Peripheral:* originates from a damaged peripheral nerve and thus has a dermatomal distribution, usually not stimulus-sensitive

Etiology

- Physiologic
  - Occurs as normal phenomena in otherwise healthy individuals and can occur during sleep; examples: hiccups, hypnic jerks, startle response, and benign myoclonus of infancy
- Essential
  - Chronic but nonprogressive myoclonus with no encephalopathy, which frequently has autosomal dominant inheritance
- Epileptic
  - Occurs in epileptic syndromes such as juvenile myoclonic epilepsy (JME) and absence epilepsy, with eyelid myoclonus (Jeavons syndrome) the most common
  - *JME:* adolescent-onset, seizures usually occur early in the morning, often generalized, and can have other seizure types (generalized tonicclonic, absence)
  - *Atypical absence:* brief absence spell with eyelid myoclonus and upward eye deviation and can be precipitated by eye closure
  - Can confirm with EEG and history consistent with epileptic syndrome
- Secondary
  - Myoclonus secondary to underlying medical or neurologic illnesses, such as posthypoxic injuries, structural brain injury (rare), neurodegenerative diseases, opsoclonus-myoclonus-ataxia syndrome, metabolic abnormalities, or drug-induced

Treatment

- Treatment is typically based on the physiology of the myoclonus.
  - *Cortical:* levetiracetam, clonazepam, and perampanel[4]
  - *Cortical-subcortical:* valproic acid, lamotrigine, and levetiracetam
  - *Segmental:* responds poorly to treatment, but clonazepam and other antiseizure medications can be tried
  - *Peripheral:* botulinum toxin injections and carbamazepine

TREMOR

- Tremors are rhythmic, oscillatory, nonsuppressible movements that are predominant in one body state (e.g., rest, action, posture).
- Tremors can have many causes; thus, history and examination are key to diagnosis. Ask about the time course, presence of distractibility,

ability to be induced or amplified by other movements, family history, and response to medications and/or ethanol.
- Examination should include various positions at rest, against gravity, and performing voluntary movements (e.g., drinking from a cup, drawing a spiral, or writing a sentence).
- Tremors can arise from injuries to the cortex, thalamus, cerebellum, or brainstem, as well as any of the circuits between these structures.

Etiology

- Essential tremor
  - Kinetic tremor that typically increases with the need for precision; can also have a postural tremor that occurs when a body part is held motionless against gravity
- Dystonic tremor
  - Tremor that results when a person resists the pull of dystonia, with tremor resolving if patients "relax into" the direction of the dystonic pull
- Secondary tremor
  - Due to central nervous system injury, degenerative disorders (e.g., Wilson disease, juvenile Huntington disease), metabolic abnormalities (hyperthyroidism, hypoglycemia), or drugs/toxins
  - Action tremors produced or exacerbated by medications such as immunosuppressants (cyclosporine, tacrolimus), stimulants, corticosteroids, valproic acid, thyroid hormone, and antidepressants/mood stabilizers
  - MRI needed if there is concern for structural lesion or injury, or with concomitant tone disorders
- Resting tremor
  - Rare in the pediatric population; can be seen with juvenile-onset parkinsonism (almost always >20 years old) or drug-induced parkinsonism

Treatment

- *First line:* propranolol and primidone
- *Second line:* topiramate, benzodiazepines, and botulinum toxin injections of affected muscles
- Deep brain stimulation reserved for severe, pharmacologically refractory tremors

TICS

- Sudden, rapid, stereotyped but nonrhythmic movements or vocalizations that often occur in irregular bursts
- *Typical features:* premonitory urge, voluntary suppression for brief periods, waxingwaning course, and exacerbation by fatigue and anxiety
- A clinical diagnosis with normal imaging/testing, but may emerge or worsen following brain injury

Treatment

- *First line:* clonidine, guanfacine, and topiramate
- *Second line:* atypical neuroleptics (such as pimozide and aripiprazole), which are better at controlling tics, but have significantly greater side effect profile
- *Third line:* tetrabenazine and first-generation (typical) neuroleptics such as haloperidol; effective for refractory tics but have the highest probability of side effects
- Habit reversal therapy effective for many, but difficult to find practitioners

ATAXIA

- Incoordination of movement that can manifest as gait abnormalities, truncal instability, difficulty with fine motor skills, dysarthria (scanning speech), or abnormal eye movements (e.g., nystagmus, changes in smooth pursuit or saccades)
- Caused by dysfunction of the cerebellum or abnormal cerebellar afferents/efferents; can result from diminished proprioceptive sensory activity, sensory-motor integration between the cortex, basal ganglia, cerebellum, and thalamus, or structural abnormalities of the cerebellum itself

Etiology

- Drug-induced
  - Acute onset of symptoms occurs within 24 hours.
  - It can occur with prescribed medications, illicit drugs, and toxin exposure (alcohol, lead, mercury, lithium, organophosphates).
  - Obtain urine toxicology screen and evaluate for other signs/symptoms of ingestion.
- Structural lesion
  - MRI shows evidence of infarct, hemorrhage, or mass effect.
  - Embolic and hemorrhagic events have an acute onset of symptoms and will often have other symptoms beyond ataxia, depending on vascular territory.
  - Posterior fossa lesions present with a more slowly progressive ataxia and may also have signs of increased intracranial pressure (headaches, nausea, vision changes).
- Infection/Inflammatory
  - This includes acute cerebellar ataxia (ACA), cerebellitis, and ADEM).
  - ACA is the most common and is a self-limited disease, presenting 1 to 2 weeks after a viral infection.
  - Cerebellitis and ADEM require prompt diagnosis (MRI) and treatment. They also present with other neurologic symptoms (encephalopathy, headache, symptoms of increased intracranial pressure).

- Hereditary
  - Two of the most common causes are spinocerebellar ataxia (SCA) and Friedreich ataxia (FA), which have chronic, progressive presentations.
  - SCA can have dominant or recessive inheritance, and many have substantial overlap between forms.
  - *FA is an autosomal recessive disorder with multisystem involvement, including symptoms such as pes cavus, scoliosis, hyporeflexia, cardiomyopathy, and diabetes.*
  - Inborn errors of metabolism may also cause ataxia. However, these are more episodic and are typically triggered by stress or illness.

Treatment

- Treatment should be focused on the underlying cause. If the cause is unclear or genetic, focus should be on symptom management.
- Symptom management includes physical therapy, occupational therapy, or use of assistive devices.
- Medications typically have little effect on the progression of ataxia, although there has been some evidence that Coenzyme Q10 may be beneficial in patients with hereditary ataxias such as SCA or FA.[5]

PAROXYSMAL DYSKINESIAS

- Paroxysmal dyskinesias are characterized by mixed abnormal movements (dystonia, chorea, myoclonus, tremor) with abrupt onset from a normal background. Episodes last from seconds to hours, with no impairment of consciousness.
- Examination, MRI, and EEG are normal.
- It has an autosomal dominant inheritance pattern and thus family history and videos of episodes are essential to the diagnosis.
- The pathophysiology is unclear, but it is thought that it may be related to neuronal network instability or hyperexcitability in the basal ganglia. It is typically inherited but rarely can occur as a result of infection, inflammation, metabolic derangements, or structural damage (e.g., trauma, infarct, mass).

Etiology

Inherited forms are classified into four types based on frequency, duration, and triggers.

- Paroxysmal kinesigenic (PKD)
  - Episodes last seconds up to 5 minutes, triggered by initiating voluntary movements after a brief period of rest (e.g., sitting to standing, running after standing)
  - May occur >100 times a day, rarely missing a day

- Paroxysmal nonkinesigenic (PNKD)
  - Episodes last tens of minutes to days, triggered by stress, fatigue, excitement, caffeine, or alcohol
  - May go weeks to months between episodes
- Paroxysmal exertion-induced (PED)
  - Episodes last 10 to 20 minutes, triggered by prolonged exercise (>15 minutes), fasting, or emotional stress
  - With caution regarding exertion, may go months with no episodes
- Paroxysmal hypnogenic
  - Typically occurs from the same phase of sleep and is thought to be a form of nocturnal frontal lobe epilepsy; some familial cases, but most cases are sporadic

Treatment

- *PKD:* anticonvulsants (carbamazepine, phenytoin, oxcarbazepine)
- *PNKD:* avoidance of triggers (although benzodiazepines have been found to be helpful)
- *PED:* avoidance of prolonged exercise as medications are often ineffective, although there are reports of response with gabapentin, acetazolamide, carbamazepine, and ketogenic diet[2]

STEREOTYPIES

- Repetitive, rhythmic, movements with a fixed pattern and regular frequency that can be stopped by distraction or redirection (e.g., calling the patient's name) and often occur during periods of excitement or stress; examples: finger tapping, hand flapping, and body rocking
- More common in children with developmental delays and autism (~80%), but are also common in neurotypical children (~45% of children)
- Unlike tics in that the child does not have a premonitory urge and thus cannot voluntarily suppress the movement
- Treatment not necessary since they are not distressing, but behavioral therapy has been effective in some cases

PARKINSONISM

- Pediatric parkinsonism consists of any two of the following four features: bradykinesia, rest tremor, rigidity, and postural instability.
- It occurs secondary to loss of dopamine in the nigrostriatal pathway (presynaptic or postsynaptic) as a result of brain injury, medication side effects, or genetic causes.

Etiology

- Juvenile-onset Parkinson disease
  - Mutations in *parkin* produce childhood-onset parkinsonism, although other genetic forms of Parkinson disease can produce early symptom onset.

- It presents with bradykinesia, focal dystonia (typically in the legs), rigidity, and tremor (although rest tremor is rare in childhood parkinsonism).
- MRI can be helpful but is not essential. Genetic testing may reveal specific mutations.

- Juvenile Huntington disease
  - It is caused by expansion of cytosine/adenine/guanine repeats (typically >50) in the *huntingtin* gene.
  - As repeats increase and age at onset decreases, patients increasingly present with the Westphal variant: nonmotor symptoms (cognitive decline, behavioral disturbances) and distinct motor symptoms (parkinsonism, myoclonus, dystonia; chorea is less common).
  - MRI shows atrophy of the caudate, putamen, and globus pallidus in symptomatic patients, with global atrophy seen in late-stage disease.[6]
- Wilson disease
  - Autosomal recessive inheritance; disorder of copper excretion leading to cellular excess
  - First neurologic signs are often dysarthria, dystonia, ataxia, tremor, or parkinsonism
  - MRI usually showing T2 hyperintensities in the caudate, putamen, thalamus, cerebellum, midbrain, and pons[7]; should test serum copper, ceruloplasmin, and 24-hour urine copper
- Drug-induced
  - Associated with dopamine receptor blocking agents such as antipsychotics, metoclopramide, and tetrabenazine
  - Often seen in a dose-dependent fashion, with symptoms resolving a few months after the offending agent is discontinued

Treatment

- Treat with carbidopa/levodopa (25/100 formulation) and monitor for side effects (nausea, dyskinesias, orthostasis, behavioral changes).
- If levodopa produces gastrointestinal side effects, increasing the ratio of carbidopa to levodopa, dividing the total over more daily doses, and using extended-release preparations can be helpful.
- It can also be treated with anticholinergics such as benztropine or trihexyphenidyl.

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Spasticity Management

9

REZA FARID

SPASTICITY MANAGEMENT

History

In the mid-1800s, Dr. William John Little began treatment for those suffering from clubfoot and other contorted limbs.[1] Subcutaneous tenotomy as developed by Stromeyer has been an effective treatment to date.[2] Progress in the treatment of spasticity slowly followed. In the late 1800s, August Bier studied intrathecal cocaine as an anesthesia method.[3] Neurosurgeons experimented with a variety of techniques hoping to quell the discomfort of those with severe spasticity. By the late 1960s, oral medications became available, and in the late 1990s use of intrathecal baclofen (ITB) and botulinum toxin was approved for treatment of spastic conditions. Interest in spasticity management has grown significantly over the past few decades. Therapeutic interventions have replaced surgery as the treatment of choice, with an increased focus on using medications, injections, and devices that are beneficial when problematic spasticity is present.

Common pediatric conditions presenting with spasticity:

- Cerebral palsy
- Spinal cord injury, complete and incomplete
- Brain injury, traumatic and nontraumatic
- Cerebrovascular accident/hemorrhage

Prior to initiating therapy, clear delineation of goals should be jointly agreed upon by the patient, their family, and their doctor. Patients should be monitored for treatment efficacy at routine intervals. Common goals of spasticity management include the following:

- Reduce complications and enhance acquisition of new skills[4]
- Reduce skeletal deformity and improve mobility
- Ease burden of care
- Improve quality of life
- Reduce feeling of stiffness or muscle pain
- Improve sleep and wellness
- Enhance self-image, inclusion, and participation

Definitions

- Lance, in 1980, put forth the following concept: "Spasticity is a motor disorder characterized by a velocity-dependent increase in tonic stretch reflexes (muscle tone) with exaggerated tendon jerks, resulting from hyperexcitability of the stretch reflex, as one component of the upper motoneuron syndrome."[5]
- In 2003, Sanger et al.[6] provided separate definitions for spasticity, dystonia, and rigidity, specifically listing spasticity as the following:
  - "'Spasticity' is defined as hypertonia in which 1 or both of the following signs are present: (1) resistance to externally imposed movement increases with increasing speed of stretch and varies with the direction of joint movement, and/or (2) resistance to externally imposed movement rises rapidly above a threshold speed or joint angle."
  - Dystonia is defined as "a movement disorder in which involuntary sustained or intermittent muscle contractions cause twisting and repetitive movements, abnormal postures, or both."
  - Rigidity requires all of the following: (a) resistance to externally imposed joint movement is present at very low speeds of movement, does not depend on imposed speed, and does not exhibit a speed or angle threshold; (b) simultaneous co-contraction of agonists and antagonists may occur and this is reflected in an immediate resistance to a reversal of the direction of movement about a joint; (c) the limb does not tend to return toward a particular fixed posture or extreme joint angle; and (d) voluntary activity in distant muscle groups does not lead to involuntary movements about the rigid joints, although rigidity may worsen.
- The European Thematic Network to Develop Standardized Measures of Spasticity (SPASM) consortium, in 2005, offered a revised definition: "disordered sensori-motor control, resulting from an upper motor neuron lesion, presenting as intermittent or sustained involuntary activation of muscles."[7]
- In 2017, a group of 37 European experts from 12 countries proposed using the term "hyper-resistance" to describe the phenomenon of impaired neuromuscular response during passive stretch.[8] It has been further suggested that "spasticity" is outdated in the practice of caring for patients.[9]

Treatment Considerations

The first step in proper spasticity management is to remove any inciting or noxious stimuli, the most common of which is constipation. Many other conditions can exacerbate spasticity, including infection, pressure sores, ingrown toenails, nephrolithiasis or cholelithiasis, venous thrombosis, fracture, bladder distension, fatigue, stress, malposition, seizure, poor orthotic fitment, and poor climate regulation. Some patients have no concerns relating to their spasticity, while others are seemingly debilitated. Treatment should be individualized. The adverse impact of undertreatment is a concern in the pediatric population because tight muscles grow slower than the corresponding bone. As the bone grows, the tightening

muscles lead to progressive loss of range of motion. With prolonged shortness of the muscle, the sarcomeres shorten and eventually the muscles will develop contracture and the joint will become fibrosed.

Therapy Interventions

Strength training should not be overlooked when desiring to improve function as it has been demonstrated to improve mobility[10,11]; however, we do not know the specific characteristics of exercise training that produce an optimal outcome. A Cochrane database review examining the potential benefit of exercise in cerebral palsy (CP) found little evidence of effectiveness.[12] An unfortunate reality of therapeutic interventions for spasticity is that we do not know the optimal amount, intensity, frequency, or duration of various interventions to produce the desired outcome.

- Physical therapy
  - Stretching, serial casting, bracing, taping, seating systems, sleeping systems, electrical stimulation, mechanical gait training, electromyography-guided robotic systems, computer-aided devices for movement, whole body vibration, and muscle cooling
- Occupational therapy
  - Stretching, splinting, casting, neurodevelopmental techniques, sensory integration, proprioceptive neuromuscular facilitation, modalities, and computer-aided devices
- Complementary and alternative therapies
  - Limited data, at this time, to support use of hyperbaric oxygen, stem cell therapies, acupuncture, apitherapy, hippotherapy, or aquatic therapy in the treatment of spasticity

Assessment Tools (Appendix 9.1)

Measures to assess spasticity[13]

- Modified Ashworth Scale
- Tardieu Scale
- Penn Spasm Scale
- Pediatric Evaluation of Disability Inventory
- Patient-Reported Impact of Spasticity Measure
- Canadian Occupational Performance Measure

Measures to assess dystonia

- BurkeFahnMarsden Dystonia Rating Scale
- BarryAlbright Dystonia Scale
- Movement Disorder Childhood Rating Scale
- Dyskinesia Impairment Scale

Measures to assess the impact of hypertonia, regardless of type

- Reflex and manual muscle testing
- Gait analysis

- Global Impression of Change
- Numeric Pain Scale

Oral Medications

Several medications are available for treatment of spasticity (Table 9.1). In general, they are inexpensive, but may have undesirable systemic side effects. Medications should be considered, in particular, for patients who have generalized, as opposed to focal, spasticity.

- Baclofen is a gamma-aminobutyric acid (GABA)-B agonist that binds to the receptors at the spinal level. Its action reduces the release of excitatory neurotransmitters and inhibits spinal reflexes.
- A Cochrane review on the use of baclofen in CP was published in the *Archives of Physical Medicine and Rehabilitation*[14] and concluded that "[t]here are insufficient data to support or refute the use of oral baclofen for reducing spasticity or improving motor function in children and adolescents with spastic cerebral palsy." Still, baclofen remains a mainstay of oral therapy for spasticity.
- Dantrolene is the only peripherally acting muscle relaxer, but weakens all skeletal muscles. The Quality Standards Subcommittee of the American Academy of Neurology (AAN) concluded that "there was conflicting evidence regarding the effectiveness of dantrolene in reducing spasticity in children with CP." Dantrolene frequently causes side effects in children with spastic CP, such as weakness, drowsiness, and irritability. Their recommendation was that "[t]here is insufficient evidence to support or refute the use of dantrolene for the treatment of spasticity in children with CP."[15]
- The Quality Standards Subcommittee of the AAN published a medication review and concluded that "[d]iazepam is probably effective for the short-term treatment of spasticity in children with CP (one Class I study and one Class II study)."[15]
- With regard to tizanidine, the same subcommittee publication noted "One small Class II placebo-controlled parallel study treated 10 children with a mean age of 4.1 years (range 2–15) with tizanidine 0.05 mg/kg/day and 30 children with placebo for 6 months" and concluded that "[t]izanidine is possibly effective to treat spasticity in children with CP. No toxicity was found in this small study."[15]
    - Medicinal marijuana has gained popularity for treatment of refractory spasticity in the adult population, but few studies have assessed responses in children. Nabiximols are available as oromucosal sprays containing tetrahydrocannabinol (THC) and cannabidiol (CBD). One study reported no significant reduction in spasticity in a pediatric population with CP or traumatic brain injury.[16] Another study found insufficient evidence to inform clinical practice in the pediatric population due to the low quality of published material.[17]

Table 9.1 Common Anti-Spasticity Oral Medications

MEDICATION	SITE OF ACTION	HALF-LIFE	SIDE EFFECTS	CLINICAL PEARLS
Baclofen	GABA-B agonist	3–4 hr	Sedation, fatigue	May inhibit brain plasticity, withdrawal may be severe
Tizanidine	Alpha-2 agonist	2.5 hr	Sedation, hypotension	Hallucinations, may prolong QT interval
Diazepam	GABA-A agonist	Biphasic	Somnolence	Tremor, nystagmus, tolerance
Dantrolene	Sarcoplasmic reticulum	8–9 hr	Generalized weakness	Liver failure at high doses
Cyproheptadine	Serotonin antagonist	8 hr	Antihistaminic	May cause weight gain
Gabapentin	Unknown	4.7 hr	Nystagmus, drowsiness	Clearance directly proportional to renal function

GABA, gamma-aminobutyric acid.

Injection Therapies

- Botulinum toxins have become the first-line treatment for patients with focal spasticity.
- A 2019 Cochrane database review found that "there is very low quality evidence that botulinum toxin A (BoNT-A) is more effective than a non-placebo control at improving gait, function, joint range of motion, spasticity, and caregiver satisfaction in the treatment of lower limb spasticity in children with cerebral palsy."[18]
- When injected into the muscle, botulinum toxin produces weakness via blockade of the SNAP-25 protein within the neuromuscular junction. Three botulinum toxin subtype A preparationsonabotulinum toxin (Botox, Allergan), abobotulinum toxin (Dysport, Ipsen), and incobotulinum toxin (Xeomin, Merz)have Food and Drug Administration (FDA) approval for use in the pediatric population in the United States. All carry a required FDA black box warning relating to the distant spread of toxin and resulting risk of aspiration pneumonia or death.
- Prabotulinum toxin (Jeuveau, Evolus) has recently been approved for adult cosmetic use but has no pediatric indication. Rimabotulinum toxin B (Myobloc, Supernus) is approved for adult use only. Botulinum toxin type B has a mechanism of action on the vesicle-associated membrane protein (VAMP or synaptobrevin).
- Botulinum toxin subtype F is not yet commercially available but may be under investigation as a toxin with a shorter duration of action.

Phenol and Alcohol Injections

- Phenol and alcohol are chemicals, not medications. As such, there is no FDA-approved use. They can be custom-ordered for off-label use in motor point blocks and chemical neurolysis of peripheral nerves.
- The mechanism of action is destruction of efferent peripheral neurotransmission, essentially producing a lower motor neuron lesion in the presence of upper motor neuron pathology.
- Commonly used concentrations for phenol injection are 3% to 6%, while ethyl alcohol concentrations may vary from 10% to 100%.
- These are used as an adjunct to spasticity management or as a primary technique for, among other conditions, hip adductor or humeral flexion hypertonicity. Appropriation of botulinum toxin to other sites may be allowed.
- These are inexpensive but require greater technical expertise for proper administration.
    - Administration is done either via ultrasound guidance, using the least amount of medication required to produce a hydrodissection of the perineurium, or via a nerve stimulator at a setting of 1 mA or less.
    - It can usually be accomplished with <1.5 mL.
    - The onset of action is within minutes. The duration of benefit is 6 to 9 months or longer.

- An adverse effect of injecting nerves carrying sensory fibers is the risk of developing dysesthesia. Patients should be counseled about loss of sensation to the lateral forearm (terminal branch of the musculocutaneous nerve).
- Less common neurolysis targets include the pectoral nerves and motor branches to the medial and lateral gastrocnemii, the hamstrings, and the posterior tibialis and soleus.
- Intrathecal injection of phenol can be beneficial in case of refractory spasticity, but there is limited literature reporting its use in the pediatric population.

Hyaluronidase

- The injection of human recombinant hyaluronidase may facilitate improvement in joint range of motion of patients experiencing spasticity with muscle stiffness.[19]
- Hylenex is commercially available and diluted 1:1 with saline and then injected into the muscle, where it may reduce muscle tone.
- It operates on the premise that muscle stiffness is a result of hyaluronan accumulation in the extracellular matrix.
- Further research will determine whether specific localization using ultrasound guidance provides an additional benefit.

Orthopedic Interventions

- Surgical procedures to lengthen muscle can be used to address secondary musculotendinous deformities (muscle contractures, joint deformities) that result from unsuccessful spasticity management.
- Clinicians must consider the following:
  - A lengthened muscle is a weakened muscle.
  - Muscle lengthening in muscles spanning two joints will impact both joints:
    - A hamstring lengthening for knee range of motion may ultimately lead to decreased posterior pelvic stability and anterior pelvic tilt.
- Tendon lengthening surgeries may decrease spasticity.[20]
- Common orthopedic surgical interventions include:
  - Tenotomy
  - Tendon lengthening
  - Tendon transfer
  - Osteotomy
  - Joint fusion
- Patients should have good selective motor control to receive the optimal outcome in terms of increased function.[21]
- Timing of orthopedic intervention is dependent on the procedure being completed. The ideal time for pelvic reconstruction, for example, is prior to skeletal maturity. Femoral reconstructive surgeries done prior to the age of 6 in lower functioning children, however, have a higher likelihood of needing repeat surgery.[22]

Neurosurgical Interventions

Selective dorsal rhizotomy (SDR) is the hallmark neurosurgical procedure for spasticity. Sir Charles Sherrington won the 1932 Nobel prize for his work on neuronal function as it related to muscle tone and the pathophysiology of spasticity. He discovered the presence of a negative feedback system that allowed for relaxation of antagonist muscles.[23] These fibers traveled through the dorsal spine and laid the groundwork for future research into decreasing afferent input into the spine through surgery. Other neurosurgical techniques attempted to treat spasticity include stereotactic encephalotomy[24] and cerebellar stimulation.[25]

- SDR involves the severing of a portion of afferent neurons within the dorsal root. There is practitioner variability in determining which neurons are cut, the levels of the surgical field, and the percentage of overall fibers that are destroyed.
- The primary adverse effects of SDR are weakness and skeletal deformity.

Peripheral neurectomy was first described in 1913[26] and remains an option for treatment of spastic conditions such as ankle clonus and adductor spasticity.

Intrathecal Baclofen

- Oral baclofen poorly penetrates the bloodbrain barrier.
- Primary advantages relate to:
  - Greater effectiveness of spasticity control as compared with oral medication or other therapies
  - Delivery of ITB which allows for direct delivery of medication to the target receptors within the spinal cord
  - Medication compliance
  - Efficacy at a much lower dose and without the side effects of oral medications
- Disadvantages include:
  - Dependence on a technological device which requires specialized expertise
  - Implantation of a relatively large pump in a confined space
    - Pumps made by Medtronic and Flowonix are FDA-approved for treatment of spasticity. Both are large devices and could be a site of discomfort unless optimally implanted.
  - Risk of infection, rejection, and catheter-related concerns such as fracture, kinking, and tip migration
  - Cerebrospinal fluid flow is an evolving field, so optimal catheter tip level and location are not yet clear. Catheters are not steerable. Placement can be done in the intrathecal space but cannot be directed posteriorly if desired.
- ITB therapy should be considered in patients with severe, generalized spasticity who are not adequately managed with a combination of therapy and oral medications and in whom injection therapy is not realistic or does not provide a desirable benefit.

Investigational Therapies

- Spinal cord stimulation[27]
  - Ongoing area of research in adults that may ultimately help children
- Mollii suit[28]
  - A reciprocal inhibition garment
- Electrical stimulation advancements[29]
  - Noninvasive stimulation as a means of spasticity reduction
- Extracorporeal shockwave therapy[30]
- MPH-220[31]
  - Selective fast skeletal myosin-2 inhibitor that affects skeletal muscle but not cardiac or smooth muscle functions
- Stereotactic dorsolateral irradiation of the spinal nerve roots[32]
- Pulsed radiofrequency ablation of the dorsal root ganglion[33]
- Cryoneurotomy[34]

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APPENDIX 9.1 MEASURES OF SPASTICITY AND ITS IMPACT

1. Modified Ashworth Scale
- **0** No increase in muscle tone
- **1** Slight increase in muscle tone, manifested by a catch and release or by minimal resistance at the end of the range of motion (ROM) when the affected part(s) is moved in flexion or extension
- **1+** Slight increase in muscle tone, manifested by a catch, followed by minimal resistance throughout the remainder (less than half) of the ROM
- **2** More marked increase in muscle tone through most of the ROM, but affected part(s) easily moved
- **3** Considerable increase in muscle tone, passive movement difficult
- **4** Affected part(s) rigid in flexion or extension

2. Tardieu Scale
- **a.** Measures the quality and angle of the muscle reaction to stretch
  - **i.** Quality
    - **0.** No resistance throughout passive movement
    - **1.** Slight resistance throughout with no clear catch at a precise angle

2. Clear catch at a precise angle followed by release
3. Fatiguable clonus (<10 seconds) occurring at a precise angle
4. Unfatiguable clonus (>10 seconds) occurring at a precise angle
5. Joint immobile

ii. Velocity
V1 Stretch of the muscle at a slow speed
V2 Stretch of the muscle at the speed of gravity
V3 Stretch of the muscle at a speed > gravity

iii. Angle
R1 Point of first resistance upon rapid stretch (V2 or V3)
R2 Point of terminal resistance with slow stretch (V1)

3. Penn Spasm Scale
   a. Frequency of spasticity
      0. No spasm
      1. Spasm induced only by stimulation
      2. Infrequent, occurring less than once per hour
      3. Spontaneous spasms occurring more than once per hour
      4. Spontaneous spasms occurring more than 10 times per hour
   b. Severity of spasticity
      0. Mild
      1. Moderate
      2. Severe
4. Pediatric Evaluation of Disability Inventory
   a. www.pearsonclinical.com.au/products/view/165
5. Patient-Reported Impact of Spasticity Measure
   a. https://journals.lww.com/jnpt/fulltext/2005/12000/the_patient_reported_impact_of_spasticity_measure.56.aspx
6. Canadian Occupational Performance Measure
   a. www.thecopm.ca
7. BurkeFahnMarsden Dystonia Rating Scale
   a. https://pubmed.ncbi.nlm.nih.gov/3966004
8. BarryAlbright Dystonia Scale
   a. https://pubmed.ncbi.nlm.nih.gov/10400175
9. Movement Disorder Childhood Rating Scale
   a. https://eprovide.mapi-trust.org/instruments/movement-disorder-childhood-rating-scale-revised-4-18-yrs
   b. https://eprovide.mapi-trust.org/instruments/movement-disorder-childhood-rating-scale-0-3-yrs
10. Dyskinesia Impairment Scale
    a. www.aacpdm.org/UserFiles/file/Dyskinesia-Impairment-Scale-Scoring.pdf
11. Global Impression of Change
    a. https://en.wikipedia.org/wiki/Clinical_Global_Impression
12. Numeric Pain Scale
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PART III

Neuromuscular and Autoimmune Disorders

AL Grawany

Peripheral Nerve Injuries and Acquired Disorders

10

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GENERAL PRINCIPLES

Peripheral nerve injuries (PNIs) in the pediatric population are a condition frequently encountered by pediatric physiatrists and pediatric providers. Seen in both inpatient and outpatient settings, evaluation and management of these injuries can vary based on the age of the patient, the mechanism of injury (MOI), and the severity of injury. Collaborative management between the medical team and the surgical team is paramount to facilitating optimum physical and functional outcomes in all PNIs. This chapter aims to present the key components in the general evaluation of PNIs in pediatrics, as well as the core elements of rehabilitation and surgical management in more common pediatric PNIs.

Epidemiology

Direct trauma is the primary cause of PNI in the pediatric population, with other causes including inflammatory and immunologic insults. Research into the prevalence of PNI in children is scant, although a recent retrospective study using the National Trauma Data Base suggested a prevalence of .6% in children who had experienced falls or had been involved in road accident trauma. This is less than a trauma-related prevalence of 5% in the adult population.[1] Older children appear to be at higher risk of trauma-related PNI.[1] Across all pediatric age groups, motor vehicle accident-related trauma and neonatal brachial plexus injuries (BPIs) account for the majority of causes.

Pathophysiology

Injury to the peripheral nerves occurs most commonly due to direct trauma. The severity of injury, and thus the prognosis, is predicated on which elements of the peripheral nerve have been affected and to what degree. It is beneficial clinically to think of the injuries in two ways: the

MOI and the reaction of the nerve to the injury (whether or not the injury is reversible or non/partially reversible). Understanding both aspects will help guide clinical decision-making and recovery expectations. The Seddon and Sunderland classification systems are commonly used to describe the category of PNI based on the damaged/affected structures in the nerve (see Table 10.1).

Mechanism of Injury

The MOI to the peripheral nerves most commonly includes *traumatic* etiologies (lacerations, crush, or traction) and/or *ischemic* injury from strictures or compressions (compartment syndrome, hematomas, entrapments, etc.).

Reaction to Injury

- *Reversible injury:* no lasting damage to the epineurium, endoneurium, or perineurium, with the axon demonstrating complete to near-complete recovery
  - *Ischemia-induced conduction block (neuropraxia):* full recovery of nerve function[2]
  - *Prolonged pressure-induced conduction block with focal demyelination:* can last for days to months[2]
- Nonreversible/degenerative injury
  - *Compression, severe traction, heavy crush injuries (axonotmetic and neurotmetic):* regeneration variable, but incomplete[2]

Table 10.1 Classification of Nerve Injury

SEDDON CLASSIFICATION	SUNDERLAND CLASSIFICATION	AFFECTED STRUCTURES
Neuropraxia	I	Myelin: conduction block
Axonotmesis	II	Axon damaged with intact endoneurium
Axonotmesis	III	Axon and endoneurium damaged with intact perineurium
Axonotmesis	IV	Axon, endoneurium, and perineurium damaged with intact epineurium
Neurotmesis	V	Complete nerve transection

EVALUATION

Evaluation of the pediatric patient is distinctly different from the adult population. A child requires an age-specific approach while being mindful of the developmental milestones they have achieved. In contrast to the adult population, a physicians assessment may be based mainly on the historical information given by the caregiver and by observation of the child.

HISTORY KEY POINTS

A detailed history is important as it will aid the examiner in localizing the site and extent of nerve injury. In addition, a PNI secondary to an overuse injury is exceedingly rare in pediatrics unlike in the adult population. Key elements to address with the patient during history taking include:

- *Identification of the MOI:* trauma (lacerating and fractures most common) versus ischemic
- Identification of the distribution of deficits and symptoms: numbness, tingling, pain, weakness, and associated functional impairments
- Hand dominance (should be age-appropriate; ~2–4 years old)
- Effect on age-appropriate functional activities: how it is affecting the patient

Physical Examination

In children less than 6 years old, the physical examination focuses mostly on motor deficits as pain and sensory status are more difficult to accurately assess.[3] The examination should be tailored to be appropriate for the child's age and developmental status.

- Observation
  - Assess for spontaneous use and integration of the extremity, posture, and muscle atrophy.
  - Evaluate for skin changes, including temperature, hair/nail growth, and anhidrosis.
- Motor evaluation
  - *0 to 6 years old:* Perform *visual observation* to detect muscle weakness during functional activities and play (abnormal limb positioning and compensatory movement patterns).
  - *≥6 years old:* Perform *formal manual muscle testing* with focus on key muscles.
  - "Pearl" movements to elicit and observe in older children for specific nerve function:
    - While lying supine, activate the serratus anterior by having the patient lift their shoulders away from the examination table ("forward shrug").[2]
    - Form an "OK" sign between the index finger and the thumb (anterior interosseous branch of the median nerve).
    - Form an "O" between the thumb and the fifth digit (the median nerve distal to the carpal tunnel).

  - Form a "Thumbs Up" sign (posterior interosseous branch of the radial nerve).
  - Perform abduction and adduction of fingers like a "fan" (ulnar nerve).
- *Sensory evaluation:* The ability to accurately test sensory function is age-dependent.
  - *0 to 6 years old:* Assessment of *sympathetic function* can be used as cutaneous sensory testing in this cohort can be unreliable.[3]
    - Observe for sweat on the palmar or plantar surfaces.
    - Wrinkle test: Immerse the affected hand or foot in 40°C water for 20 to 30 minutes and observe for presence of wrinkling (present if the sympathetic function is intact).[3]
  - *≥6 years old:* Assess for *cutaneous sensory modalities* in the distribution of the affected and surrounding peripheral nerves.
- Deep tendon reflexes: These will be asymmetrically absent or decreased in the presence of a PNI.
- Special tests
  - *Tinel sign:* Percussion (repeated moving distal to proximal) of the soft tissue over a nerve which has sustained a degenerative lesion will elicit neuropathic sensations.[2]

Ancillary Testing

While serial physical assessments performed over months allow the examiner to track the patient's clinical progression, ancillary diagnostic testing can help categorize the severity of the injury based on the peripheral nerve's structural integrity, physiology, and function. The addition of these tests is not mandatory but should be based on the nature of the diagnosis and progression (or lack thereof) of the patient's recovery.

- Electromyography (EMG) and nerve conduction studies (NCS)
  - Performed at 2 to 3 weeks postinjury to characterize injury severity and distribution, then repeated at 3 months postinjury to gauge recovery[4]
- MRI
  - Can be used to assess for mass effect (hematoma, edema) and soft tissue abnormalities around the nerve at the site of injury[5]
- Magnetic resonance neurography (MRN)
  - Can be used to characterize an injury's location and severity
  - Recommended to be performed emergently in children with penetrating trauma[4]

BRACHIAL PLEXUS INJURIES

Direct trauma is the primary cause of BPI in the pediatric population. This trauma can result in open or closed injuries and may require surgical intervention depending on the extent of the injury. As a result, management by a multidisciplinary team allows optimization of functional recovery while avoiding unnecessary interventions.[6]

BRACHIAL PLEXUS BIRTH INJURY

Brachial plexus birth injury (BPBI) is the result of traction on the brachial plexus during the birthing process, either by exogenous or endogenous applied forces.[7] The most common risk factors are shoulder dystocia and macrosomia.[7] BPBI encompasses a wide range of neurologic impairments and clinical presentations in both acute and chronic settings. Most BPBIs are neurapraxic and involve the upper trunk (vs. the lower trunk or the entire plexus).

Initial/Newborn Assessment

Physical examination is the primary diagnostic and prognostic tool for BPBI in the newborn period. The diagnosis can be suspected when there is lack of spontaneous upper extremity movement on examination.

- Inspect the newborn for asymmetric upper extremity active movements, abnormal spontaneous positioning, and signs of pain/discomfort.
  - *Active range of motion:* Assess range of motion using primitive reflexes (e.g., Moro), stretch reflex, and stimulation of the skin opposite the concerning muscle.[8]
  - Evaluate for Horner syndrome (miosis, ptosis, enophthalmos, anhidrosis), which is linked to injury of the sympathetic trunk (e.g., T1 nerve root) and the lower roots of the plexus.
  - A chest x-ray can be ordered to assess for a raised ipsilateral hemidiaphragm.[9]
- Distinguish BPBI from pseudoparalysis in the setting of a birth fracture (humeral or clavicular) or neonatal radial nerve paralysis.[7]

Early Management

Monthly clinical examinations with an objective assessment of movement allow the physician to track and quantify the infant's neurologic recovery and upper limb function. The main goals during this time (first 6 months postinjury) are to identify cases in which early nerve repair could contribute to a better functional result than conservative treatment and to prevent secondary complications.[8] The Active Movement Scale is commonly used to track recovery progression (Exhibit 10.1).

- Conservative management should start at 3 weeks after birth.[10]
  - Maintain *passive range of motion* (PROM) at the shoulder, elbow, wrist, and digits through daily stretching (critical to help limit/prevent glenohumeral dysplasia, subluxation, or dislocation secondary to shoulder muscle imbalance) and orthotic intervention/static positioning (indicated to help maintain joint integrity and prevent contracture formation).[10]
  - Daily upper extremity *strengthening* is facilitated through guided bimanual play, developmental positioning,[10] and electrical stimulation.
  - Optimize *sensory awareness* of the affected limb through tactile stimulation with different textures, vibration, and brushing.
- Further evaluation

Exhibit 10.1 Active Movement Scale

SCORE		SCALE	
SHOULDER ABDUCTION	_____	**GRAVITY ELIMINATED**	**SCORE**
Shoulder adduction	_____	No contraction	0
Shoulder flexion	_____	Contraction, no motion	1
Shoulder external rotation	_____	<50% motion	2
Shoulder internal rotation	_____	>50% motion	3
Elbow flexion	_____	Full motion	4
Elbow extension	_____		
Forearm supination	_____	AGAINST GRAVITY	
Forearm pronation	_____	<50% motion	5
Wrist flexion	_____	>50% motion	6
Wrist extension	_____	Full motion	7
Finger flexion	_____		
Finger extension	_____		
Thumb flexion	_____		
Thumb extension	_____		

If function has not fully recovered by the third to fourth month, ancillary testing can be requested to augment the workup and this can include *EMG/NCS* and/or advanced imaging such as *CT myelography* (to evaluate for root avulsions), *MRI* (to evaluate for root avulsions and neuromas), and *ultrasound of the glenohumeral joint*.[8] Ultrasound is recommended in all infants with incomplete recovery who demonstrate a progressive loss of external shoulder rotation, to evaluate for glenohumeral dysplasia, subluxation, or dislocation.[10]

Longitudinal Management

Children with incomplete recovery can experience lifelong functional impairment. Therefore, the focus of clinical care shifts from impairment to age-appropriate functional ability and assistance with participation in desired activities. Possible long-term sequelae include weakness, glenohumeral joint deformity, limb length discrepancy, pain, and psychological stress.[7,9] Due to altered sensation, children are at risk of temporary or long-lasting postural and developmental motor learning disorders and should be monitored accordingly.[9]

- *Conservative management:* Patients may benefit from episodic therapy throughout childhood and adolescence to address motion, strength, activity adaptation, and motor training to minimize habitual compensatory movements.[7] Therapeutic interventions to consider include:
  - Constraint-induced movement therapy (CIMT)
  - Botulinum toxin injections to treat co-contractions
  - K-taping to facilitate normalized movement patterns
  - Orthotics to prevent or manage growth-related contractures

Surgical Management

Surgery is indicated for persistent defects that are a result of neurotmesis, with demonstrated absence of hand function by 3 to 6 months of age or absent shoulder function by 3 to 9 months. The priorities for reconstruction include recovery of hand grasp, elbow flexion, shoulder abduction/external rotation, and elbow/wrist/finger extension. Surgical treatment options for the primary injury include:

- *C5 and C6/upper trunk neuroma-in-continuity:* neurolysis versus excision and sural nerve (SN) graft
- *C5 injured and C6 avulsed:* SN graft from C5 to the upper trunk, nerve transfer from the spinal accessory nerve (SAN) to the suprascapular nerve (SSN)
- C5 and C6 both avulsed: nerve transfers from multiple donor nerves

Sequelae of Brachial Plexus Injuries

The long-term effects and sequelae associated with incomplete recovery from BPI are numerous and include skeletal changes as well as functional weakness and impairment. Secondary surgeries may be considered to treat

functional impairments of the upper extremity and include muscle/tendon transfers and arthrodesis (>3–4 years old). The patient must have redundant expendable muscles of grade ≥M4 on the Medical Research Council Muscle Scale (MRC), with adequate PROM. Skeletal abnormalities include:

- *Glenohumeral dislocation:* This presents as sudden, painful loss of passive shoulder abduction and external rotation.
  - Assess with physical examination and dynamic ultrasound.
  - Treat initially with closed reduction, casting, and chemodenervation.
  - Treat recalcitrant cases with open release ± tendon transfer to augment external rotation.
- Glenohumeral deformity: Consider derotational osteotomy of the humerus to reorient the arc of shoulder rotation and place the hand into more functional position.
- Elbow flexion weakness
  - If wrist and finger flexors are strong, perform Steindler flexorplasty (proximal transfer of flexor-pronator origin) for elbow flexion.
  - If hand function is weak, perform bipolar pedicled transfer of latissimus dorsi or pectoralis major to replace the biceps.
  - If there are no local donors, replace biceps using free functional muscle transfer (FFMT) with gracilis neurotized by intercostal, SAN, or contralateral C7 motor nerve transfer.

OTHER ACQUIRED INJURIES OF THE BRACHIAL PLEXUS AND PERIPHERAL NERVES

Much of the conservative management strategy considerations for BPBI are applicable to all PNIs. Surgical interventions for both acquired BPIs and general PNIs include the following:

Surgical Treatment for General Peripheral Nerve Injury

- Open injuries (sharp or tissue loss)
  - Early exploration/reconstruction should be considered.
    - Sharp injuries (e.g., laceration, stab, iatrogenic) should be reconstructed ideally within 48 to 72 hours due to retained electrical stimulation of the distal stump for surgical identification.
    - Blunt/avulsive injuries (e.g., crushing or tearing) may require 3 weeks to determine the zone of injury.
  - Gunshot wounds are evaluated for nerve discontinuity versus blast/stretch injury.
- Closed injuries (avulsive or stretch)
  - Evaluate with serial physical examination. Acquire imaging (ultrasound or MRI) after 3 weeks to allow local wound trauma to resolve.
  - If there are signs of improvement (e.g., progressing Tinel sign, early motor return) at 3 months, continue hand therapy and serial physical examination every 3 to 6 months.

- If there is no improvement after 3 months, obtain an EMG. Begin surgical planning if there are no motor unit potentials on EMG.

Surgical Treatment for Supraclavicular Acquired Brachial Plexus Injuries

- Explore and determine nerve root avulsion versus rupture if blunt injury.
- Scar/neuromas/gliomas are excised from the stumps of the injured nerves.
- Nerve root ruptures and nerve gaps (unable to approximate proximal and distal stumps of the injured nerve) are reconstructed with nerve grafts.
- Nerve root avulsion requires nerve transfer from preserved brachial plexus elements and nonplexus motor nerves (e.g., ipsilateral or contralateral C7 root, SAN, intercostal nerves, phrenic nerve).

Surgical Treatment for Infraclavicular Acquired Brachial Plexus Injuries or Peripheral Nerve Injuries

- *Axillary nerve injury:* The goal is active shoulder abduction.
  - Triceps to axillary nerve transfer
  - Medial pectoral or thoracodorsal to axillary nerve transfer
- *Musculocutaneous nerve injury:* The goal is active elbow flexion.
  - *Oberlin transfer:* ulnar nerve fascicle to biceps motor nerve
  - Mackinnon or double fascicular transfer: ulnar and median nerve fascicles to biceps and brachialis motor nerves; may provide improved elbow flexion
- Median nerve injury
  - *Motor reconstruction:* The goal is pinch and grasp function.
    - *Radial nerve donor:* extensor carpi radialis brevis (ECRB) and supinator motor nerves to anterior interosseous nerve (AIN) transfer
    - *Musculocutaneous nerve donor:* brachialis motor nerve to AIN transfer
  - *Sensory reconstruction:* The goal is thumb and index sensation.
    - *Ulnar nerve donor: fourth* webspace sensory nerve to the first webspace sensory nerve transfer
    - *Median nerve donor, upper trunk injury:* third webspace sensory nerve to the first webspace sensory nerve transfer
- Ulnar nerve injury
  - *Intrinsic muscles:* distal AIN (pronator quadratus motor nerve) to ulnar motor fascicle nerve transfer for the intrinsic muscles of the hand
  - *Digit flexion:* index/long flexor digitorum profundus (FDP) to ring/small FDP tendon transfer
  - Sensation to the ulnar border of the hand: third webspace sensory nerve to the fourth webspace sensory nerve and small finger ulnar digital nerve transfer

- *Radial nerve injury:* The goal is digit/wrist extension.
  - *Median nerve donor:* flexor carpi radialis (FCR), palmaris longus (PL), and flexor digitorum superficialis (FDS) motor nerve transfer to posterior interosseous nerve (PIN) and ECRB motor nerves
  - *Radial nerve donor:* ECRB and supinator motor nerves to PIN transfer
  - Ulnar nerve donor: flexor carpi ulnaris (FCU) motor nerve to PIN transfer

Surgical Treatment for Delayed Presentation

Motor reinnervation of acutely denervated muscles must occur by 18 months postinjury to preserve neuromuscular junction viability. Nerve reconstructive surgery must be initiated before 9 to 12 months have elapsed. In cases with delayed presentation or incomplete recovery, tendon or muscle transfers may provide additional options for functional restoration.

- FFMT
  - Neurotized gracilis muscle microsurgical free flap can be used to restore elbow flexion and finger flexion or extension (less common muscle donors are latissimus dorsi and rectus femoris).
  - FFMT reconstruction requires coaptation of the motor nerve of the transferred muscle to a "recipient" motor nerve in the reconstructed limb.
- *Nonmicrosurgical muscle/tendon transfers:* For a donor to be eligible, a muscle/tendon should have an MRC >M4 muscle strength.
  - Musculocutaneous nerve functions
    - Pedicled latissimus dorsi or pectoralis major muscle transfer
    - Steindler flexorplasty-proximal relocation of flexor-pronator muscle origin to increase moment arm for elbow flexion
  - Median nerve functions
    - *Opponensplasty for thumb opposition:* tendon transfer to re-create function of abductor pollicis brevis (APB) tendon
    - *Finger flexion:* ring/small FDP to index/long FDP tendon transfer
  - Ulnar nerve functions
    - *Finger flexion:* index/long FDP to ring/small FDP tendon transfer
    - Intrinsic function/correct "clawing"
      - *Zancolli 2 tendon transfer:* ring FDS tendon transferred to ring/small finger A1 or A2 pulley
      - *Zancolli 1 procedure:* ring/small finger metacarpophalangeal capsulodesis
  - Radial nerve functions
    - *Wrist extension:* pronator teres (PT) to ECRB tendon transfer
    - *Thumb extension:* PL or ring finger FDS to extensor pollicis longus (EPL) tendon transfer
    - *Finger extension:* FCR (Brand), FCU, or FDS (Boyes) to extensor digitorum communis (EDC) tendon transfer

ACUTE FLACCID MYELITIS

Acute flaccid myelitis (AFM) is an anterior horn cell/lower motor neuronopathy characterized by acute-onset limb weakness or paralysis accompanied by predominately gray matter lesions involving one or more spinal cord segments.[11,12] It can rapidly progress over the course of hours or days, potentially resulting in permanent paralysis and life-threatening respiratory failure.[13] The enterovirus D68 (EV-D68) is suspected to be the main driver of the seasonal (late summer to early fall) biennial outbreaks (since 2014), although other enteroviruses (particularly enterovirus A71) and some coxsackie virus strains have also been associated.[14]

Initial Assessment

In most cases, a prodromal illness, manifesting with fever and respiratory symptoms, precedes the onset of limb weakness. The clinical presentation is distinct and can include the following:

- Flaccid weakness, typically asymmetric, and can affect one or more limbs, with a tendency to affect the proximal muscle groups and upper limbs
- Bowel and bladder dysfunction common in the acute phase[14]
- Respiratory symptoms (cough, rhinorrhea, pharyngitis, or asthma-like illness)[14]
- Autonomic dysfunction seen in some cases
- Sensory deficits uncommon
- Neuropathic pain or paresthesias may be present in the limb(s), neck, and low back[14]

The diagnosis may require a multimodal assessment and typically includes *MRI of the spinal cord* with hallmark demonstration of T2 hyperintensity of the spinal cord gray matter and eventual localization to the anterior horn cells,[14] *lumbar puncture* with identification of cerebrospinal fluid pleocytosis in most patients in the acute phase, *respiratory and stool or rectal swabs* testing positive for EV-D68 and EV-A71, and *EMG/NCS* changes consistent with an acute motor axonal neuropathy.[14]

Early Management

Patients with suspected AFM should be immediately hospitalized and monitored for respiratory deterioration.[13] There are currently no studies with enough evidence to endorse or discourage the use of corticosteroids, intravenous immunoglobulin, or therapeutic plasma exchange.[12] Supportive treatment with careful monitoring focused on potential emerging vital complications is the mainstay of early management.[14] This can include:

- Optimizing and maintaining stable cardiorespiratory status
- Treating bladder, bowel, or other autonomic dysfunctions
- Assessing for the presence of dysphagia and managing with supplemental fluids and nutrition if indicated

- Managing pain , with gabapentin demonstrating control of neuropathic pain[15]
- Preventing complications of immobility (e.g., contractures, pressure wounds, and venous thromboembolism)
- Early initiation of rehabilitation programs (physical therapy/occupational therapy/speech therapy), including electrical stimulation modalities, early mobilization, and activity-based therapy

After the acute phase, medically stable patients with significant residual neurologic deficits should be admitted to inpatient rehabilitation. While evidence is minimal, it is recommended that the therapeutic approach draw on methods used to treat other monophasic neurologic injuries (e.g., spinal cord injury) and other motor mononeuropathies (e.g., poliomyelitis).[14] Studies have suggested that children who participated in activity-based restorative therapy (ABRT) demonstrated significant neurologic and functional gains.[16,17]

Longitudinal Management

Most patients show some improvement in motor strength. Recovery is most rapid within the first few months after onset[14] and tends to plateau around 6 to 9 months. However, recovery can be significantly asymmetrical, with few patients (<10%) having complete recovery[14] and with persistent deficits expected in >75% of patients. Potential complications include:

- *Neurologic:* neuropathic pain, chronic constipation, chronic ventilator dependence, dependence on artificial nutrition, and hydration[14]
- Musculoskeletal: joint subluxation or dislocation (proximal > distal), profound muscle weakness and atrophy, limited joint range of motion, scoliosis, limb length discrepancy, and chest wall abnormalities[14]
- Psychological: anxiety and/or depression

Surgical Treatment

- Nerve transfer
  - Must be performed within 9 to 12 months of onset to prevent permanent loss of neuromuscular junction functionality
  - Maintains native muscle joint biomechanics, including excursion and line of pull of reinnervated muscles
  - *Nerve transfer donors:* include SAN, branches/motor fascicles of radial, ulnar and/or median nerves, contralateral C7 nerve root, and intercostal nerves
  - *Nerve transfer targets:* include SSN, axillary nerve, musculocutaneous nerve, median nerve/AIN, PIN, and triceps motor nerves
- Tendon transfer
  - *Secondary reconstructive option:* performed if there is delay in the patient presenting for reconstruction, at >12 months after onset, and usually performed after spontaneous nerve recovery has plateaued at 18 to 24 months after onset
  - Options often more limited when compared with nerve transfer

- *Joint fusion*
  - Used to stabilize joints to allow for modified function

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Neuromuscular Disorders

11

DIANA CASTRO

GENERAL PRINCIPLES

Neuromuscular disorders (NMDs) include both inherited and acquired diseases of the peripheral nervous system. These encompass a wide variety of conditions affecting the anterior horn cell, such as spinal muscular atrophy (SMA); the peripheral nerve, including GuillainBarrè syndrome (GBS) and CharcotMarieTooth (CMT); the neuromuscular junction (NMJ), such as myasthenia gravis (MG) or congenital myasthenic syndrome (CMS); or the muscle, as seen in myopathies or muscular dystrophies such as Duchenne muscular dystrophy (DMD). In many of these disorders, there can be involvement of other organ systems, either related to the condition itself or as a secondary complication due to disease progression. Several of these conditions have targeted treatments with recent scientific advances, thereby increasing the longevity of children with NMDs. As such, accurate and prompt diagnoses of these disorders, knowledge of specific therapies, and continued standard of care are becoming more crucial. This chapter aims to describe the essential parts of the neuromuscular examination, the initial diagnostic workup, and directed therapeutic options for specific conditions when available, and provides recommendations for medical management, therapy, rehabilitation strategies, and multidisciplinary care.

DIAGNOSTIC EVALUATION

Diagnostic evaluation begins with a careful review of the patient's history. Depending on the patient's age, signs and symptoms of a neuromuscular condition may be reported differently. Infants will often be described as "floppy" due to hypotonia and may have failure to thrive, difficulty eating, or respiratory distress secondary to neuromuscular weakness. In toddlers, parents may report that motor milestones are delayed or that the child is clumsy, has frequent falls, or has difficulty standing up from the floor.[1,2] Older children may have fatigue, difficulty keeping up with their peers, pain or muscle cramps, and develop orthopedic abnormalities such as scoliosis and ankle contractures leading to toe walking.[1,2]

Other important parts of the medical history that could indicate an underlying neuromuscular condition include pregnancy complicated by polyhydramnios, clubfoot, or arthrogryposis identified on prenatal

ultrasounds or decreased fetal movement.[1] Early respiratory distress or a paucity of movements after birth can also indicate an NMD. Developmental milestones and achievement of gross motor skills are also essential to note. Family history can be informative as many of these conditions are inherited, although the method of inheritance can vary. A complete pedigree should be performed.

Neuromuscular examination is crucial to the diagnosis. Specific findings and patterns of weakness or sensory changes can direct the next steps in testing. Table 11.1 highlights the important features of the neuromuscular examination.

DIAGNOSTIC TESTING

History and physical examination findings can direct targeted laboratory, electrodiagnostic, pathologic, and genetic workup. Common studies are highlighted in Figure 11.1. Creatine kinase (CK) level is typically the first test utilized given the ease of obtaining the level and its relatively low cost. Based on these findings and the clinical information, workup typically proceeds with a combination of electrodiagnostic studies, including electromyography/nerve conduction studies/repetitive nerve conduction (EMG/NCS/RNS), genetic testing, and muscle biopsy. The availability and the declining cost of genetic testing and the relatively noninvasive way it is performed have led to earlier utilization.[3] In many cases, genetic testing may eliminate the need for more invasive evaluations such as muscle biopsy.[3]

MEDICAL MANAGEMENT AND SUPPORTIVE CARE

A multidisciplinary approach is the standard of care for pediatric patients with neuromuscular disease given the multiorgan involvement in many peripheral nervous system conditions.[4] The care team for these patients typically involves specialists from neurology, physical medicine and rehabilitation, pulmonology, cardiology, nutrition, physical therapy, and occupational therapy. Referrals are also frequently made to orthopedic surgery, ophthalmology, endocrinology, gastroenterology, orthotists, speech therapy, genetics, psychiatry, sleep medicine, and social work.[4,5]

Neurology

Neuromuscular specialists, typically child neurologists and physiatrists with subspecialty focus, are essential members of this multidisciplinary team and play a central role in coordinating care. These physicians often lead the diagnostic investigation and provide patients and their family with anticipatory guidance, prognosis, and expectations regarding the condition. Neuromuscular specialists will then facilitate appropriate referrals to other specialists and allied health professionals, providing longitudinal care throughout the years. In the case of specific conditions with targeted treatments, neuromuscular specialists will also guide patients in

Table 11.1 Key Features of Examination to Evaluate a Child With Suspected Neuromuscular Disorder

	EXAMINATION PART	FINDINGS	CONDITIONS TO CONSIDER
General examination	HEENT	Dysmorphic features	NMJ disorders such as CMS
		Ptosis	NMJ disorders such as MG, CMS, and myopathy
		High-arched palate	Neuropathy, myopathy
		Macroglossia	Myopathy, common in DMD, Pompe disease
		Tongue fasciculations	SMA
	Chest, including CV/lungs	Bell-shaped chest, paradoxical breathing	SMA
	Back	Scoliosis	Seen in multiple NMDs
		Lordosis	Seen in multiple NMDs, common in DMD
		Scapular winging	Common with limb-girdle pattern of weakness
	Abdomen	Hepatomegaly	Pompe disease
		Palpable stool retention	Seen in multiple NMDs

(*continued*)

Table 11.1 Key Features of Examination to Evaluate a Child With Suspected Neuromuscular Disorder (*Continued*)

	EXAMINATION PART	FINDINGS	CONDITIONS TO CONSIDER
	Extremities	Ankle contractures or equinovarus deformities, pes planus	Seen in multiple NMDs
		Pes cavus	Inherited neuropathy like CMT
		Multiple contractures	Arthrogryposis, collagen myopathy, congenital muscular dystrophy, LGMD, EDMD
		Hyperlaxity	Seen in multiple NMDs
	Skin	Keratosis pilaris	Common in collagen myopathies and DMD
Neurologic examination	Mental status	Autistic features	CNS syndrome, common in DMD and some congenital myopathies
		Hypophonic and nasal speech	Myopathy, NMJ disorders, SMA
	Cranial nerves	Ophthalmoparesis	NMJ disorders such as MG, CMS, myopathy, mitochondrial disorders
		Facial weakness: eyelid closure weakness, tented open mouth, transverse smile	Myopathy, FSHD

	Sensation: light touch, pinprick, vibration, joint position sense, temperature	Diminished sensation distally	Peripheral neuropathy (inherited or acquired)
	Cerebellar	Tremor	Neuropathy, SMA
		Ataxia	Spinocerebellar ataxia, Friedreich ataxia
	Muscle bulk	Pseudohypertrophy of muscles	DMD: most common in calves; myotonia congenita
		Distal atrophy, such as "inverted champagne bottle" appearance	Peripheral neuropathy like CMT or chronic acquired neuropathies
	Muscle tone	Hypotonia	Seen in multiple NM diseases
	Muscle strength: 0: no muscle contraction 1: flicker of tendon movement 2: full range of motion with gravity eliminated 3: full range of motion against gravity 4: full range of motion against gravity with moderate resistance added 5: normal strength	Proximal > distal weakness, i.e., positive Gowers' maneuver	Myopathy, NMJ disorders, immune-mediated neuropathy, i.e., GBS or CIDP
		Distal > proximal weakness	Neuropathy
		Variable and fatigable weakness throughout the day	NMJ disorders
		Significant bulbar weakness	MG, SMA
		Acute onset of weakness	MG, periodic paralysis, GBS, toxin, vasculitis

(*continued*)

Table 11.1 Key Features of Examination to Evaluate a Child With Suspected Neuromuscular Disorder (*Continued*)

	EXAMINATION PART	FINDINGS	CONDITIONS TO CONSIDER
	Hyperexcitability of the muscles	Grip or percussion myotonia: delayed relaxation of muscle following contraction	Myotonic dystrophy, myotonia congenita, periodic paralysis, SchwartzJampel syndrome
		Paradoxical myotonia: myotonia worsens with repeated contraction	Paramyotonia, periodic paralysis
		Rippling muscle: wave-like motion of the muscle	Rippling muscle disease
	Reflexes	Absent or diminished	Seen in multiple NM diseases, particularly neuropathies and myopathies
		Normal	NMJ disorders; can be normal in many NM diseases, especially early on
	Gait	Toe walking	Seen in multiple NM diseases

CIDP, chronic inflammatory demyelinating neuropathy; CMS, congenital myasthenic syndrome; CMT, CharcotMarieTooth; CNS, central nervous system; CV, cardiovascular; DMD, Duchenne muscular dystrophy; EDMD, EmeryDreifuss muscular dystrophy; FSHD, fascio scapular humeral muscular dystrophy; GBS, GuillainBarrè syndrome; HEENT, head/ears/eyes/nose/throat; LGMD, limb girdle muscular dystrophy; MG, myasthenia gravis; NM, neuromuscular; NMD, neuromuscular disorder; NMJ, neuromuscular junction; SMA, spinal muscular atrophy.

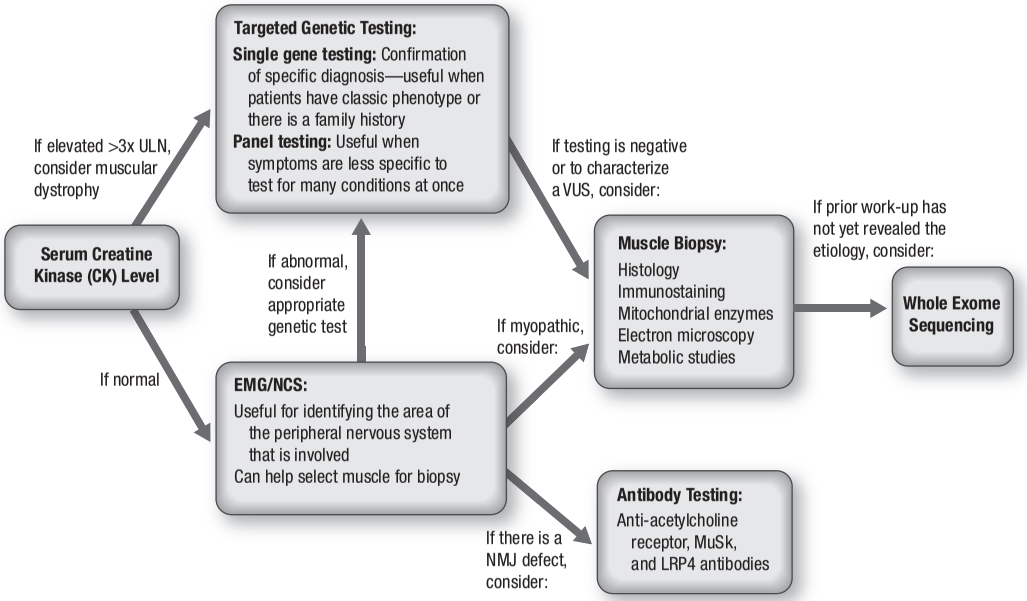

Figure 11.1 Common studies for neuromuscular disorders.

LRP4, low-density lipoprotein receptor-related protein 4; MuSK, muscle specific kinase; ULN, upper limits normal; VUS, variant of undetermined significance

selecting treatment and the expected outcomes and in monitoring for side effects. Many will also participate in research studies to aid in the advancement of treatments. Therefore, knowledge of clinical trials and the ability to direct interested patients to these studies are crucial.

Pulmonology

The role of pulmonology in the care of neuromuscular patients is to aid in the evaluation of lung disease secondary to neuromuscular weakness. Patients with neuromuscular disease will often develop restrictive lung disease and can have respiratory muscle weakness and weak cough leading to poor airway clearance. Pulmonary function testing and chest x-rays are typically performed twice per year as part of their pulmonological evaluation. Based on the results of these tests, patients may be recommended to use invasive or noninvasive ventilation with bilevel positive airway pressure (BiPAP), as well as airway clearance devices such as cough assist machine and chest physiotherapy (CPT) vest.[6] Sleep specialists also have a critical role in evaluating for obstructive sleep apnea (OSA), a common complication of the low airway tone seen in neuromuscular patients. Baseline polysomnography is recommended as part of the initial evaluation of any patient diagnosed with an NMD. Patients diagnosed with OSA require nighttime use of BiPAP titrated by a sleep specialist.[7]

Cardiology

The cardiac team is another critical aspect of neuromuscular patient care as skeletal muscle disease can demonstrate some pathologic overlap with striated heart muscle. Neuromuscular patients can develop cardiomyopathy related to their illness or arrythmias.[8] Screening with echocardiogram and EKG is performed approximately yearly, but can be spaced out further for some patients if studies remain normal. Specifically, for patients with dystrophinopathies such as DMD or Becker muscular dystrophy (BMD), yearly screening is recommended starting at the time of diagnosis. Cardiac MRI is also recommended after 10 years of age or earlier if appropriate. In the most recent consensus statement regarding the care of patients with DMD, it was suggested that patients start treatment with an angiotensin-converting enzyme inhibitor or angiotensin receptor blocker (ARB) around 10 years of age, even if they have normal cardiac evaluations. If there is evidence of cardiac disease or heart failure present before the age of 10, heart failure medications are started at this time.[9]

Physical and Occupational Therapy

Both physical and occupational therapists have a role in the care of neuromuscular patients.[10] Therapist evaluations with standardized scoring are performed twice per year, typically in conjunction with clinic visits. These standardized scales provide an objective measure of patients' weakness and possible progression. This scoring can help guide clinical trials and

determine eligibility for treatments and adequate equipment. Regularly scheduled outpatient physical and occupational therapy is important for most patients to prolong their level of functioning or ambulation and maintain safety.

MANAGEMENT OF OTHER COMMON COMPLICATIONS

Scoliosis

Due to the intrinsic weakness of abdominal and spinal musculature, neuromuscular patients are prone to developing scoliosis. This is often exacerbated by maintaining prolonged seated positions, particularly following loss of ambulation. Involvement of orthopedic surgery is indicated when scoliosis is noted to monitor the progression of spinal curvature and discuss treatments, such as bracing or surgery, when necessary.[11,12]

Contractures

Limb contractures occur in many neuromuscular conditions as muscle weakness limits joint movement. Fixed contractures can cause joint deformity and further limit patient function. Contractures are often managed with orthotic devices, including anklefoot orthoses (AFOs), supramalleolar orthoses (SMOs), and hand splints, to maintain a neutral position.[13] These orthotic devices are often worn only at night to improve joint mobility without contributing extra weight to the limbs when patients are awake and active. The use of orthotics is tailored to individual patients and their degree of weakness.

Nutrition and Feeding Issues

Weakness of pharyngeal muscles and low muscle tone can contribute to significant dysphagia in many neuromuscular patients. Videofluoroscopic swallow studies (VFSS) can evaluate the degree of dysphagia and identify problematic textures or consistencies that may cause aspiration. Careful weight gain assessment is also important as the initial symptom of dysphagia may be failure to thrive.[4] Many neuromuscular patients require adjunctive feeding methods with gastrostomy or gastrojejunostomy tubes for supplemental nutrition.[4,14]

Bone Density

Low bone density is seen in numerous NMDs and is most common in DMD due to the combination of prolonged glucocorticoid therapy and progressive weakness.[15] Early identification of patients with low bone mineral density is crucial as they are at increased risk of fractures following minimal trauma and vertebral fractures. Intermittent spine radiographs and/or dual-energy x-ray absorptiometry (DEXA) scans are recommended to monitor patients at risk.[15] Data on treatment options vary based on the underlying condition, and IV bisphosphonates are

recommended to treat osteoporosis in DMD.[15] Vitamin D is also recommended to those with documented insufficiency.[16]

Anesthesia Concerns

A preoperative consultation with an anesthesiologist with experience in treating patients with NMDs is recommended to patients with planned surgical procedures. There is an increased risk of malignant hyperthermia and rhabdomyolysis in many conditions in response to inhaled anesthetics such as halothane and depolarizing muscle relaxants such as succinylcholine.[16] As such, these medications are contraindications in myopathies and muscular dystrophies such as DMD. Some medications can exacerbate other conditions such as MG.

TARGETED THERAPIES IN PEDIATRIC NEUROMUSCULAR DISEASE

Duchenne Muscular Dystrophy

Glucocorticoids are a mainstay of treatment to slow disease progression and prolong ambulation in patients with DMD. Glucocorticoids, such as prednisone and deflazacort, are hypothesized to slow disease progression by reducing inflammation and increasing repair of weakened muscle cells.[17] Use of glucocorticoids has been found to delay loss of ambulation and secondary complications, including scoliosis and restrictive lung disease, and prolong upper extremity strength.[18]

Newer therapies for DMD have emerged in recent years, including antisense oligonucleotide (ASO)-mediated exon-skipping. This treatment aims to allow the production of a partially functional dystrophin protein to stabilize muscle membranes and reduce muscle breakdown. This allows for a milder phenotype closer to BMD. Due to variability in genetic deletions causing DMD, this treatment is not universally applicable to all DMD patients as all therapies are mutation-specific. The first exon-skipping therapy, eteplirsen, was approved in 2016, targeting exon 51.[19] Currently, exons 51, 53, and 45 have Food and Drug Administration-approved exon-skipping treatments, while other mutation therapies are still being investigated.

Spinal Muscular Atrophy

Three SMN-enhancing treatments for SMA have been approved in the past several years (Table 11.2). In 2016, the first disease-modifying treatment, nusinersen, was approved. Nusinersen is an intrathecally administered ASO targeting SMN2 expression.[20] Several years later, in 2019, gene therapy with Zolgensma (onasemnogene abeparvovec-xioi) was approved for treatment of SMA. This therapy, delivered as a single IV dose, transports a functional SMN gene to motor neuron cells via an AAV9 viral vector.[21] In 2020, risdiplam, an orally administered daily medication, was

Table 11.2 SMN-Enhancing Treatments for SMA

TREATMENT	ADMINISTRATION	TYPE OF THERAPY	PATIENT AGE APPROVED FOR ADMINISTRATION
Nusinersen (Spinraza)	Intrathecal, every 4 months	ASO, SMN-enhancing	All ages
Onasemnogene (Zolgensma)	One-time IV treatment	Gene therapy, SMN-enhancing	Up to 2 years of age
Risdiplam (Evrysdi)	Daily oral/per g-tube medication	SMN-enhancing	2 months and older

ASO, antisense oligonucleotide; SMA, spinal muscular atrophy.

approved. This small molecule increases the expression of functional SMN protein by modulating SMN2 gene splicing.[21]

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Neuroimmunological Disorders

12

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INTRODUCTION

Immune-mediated disorders of the nervous system have increased in prevalence and recognition, along with our understanding of the relevant pathophysiology. The conditions can be categorized based on the target of inflammation, the type of underlying immunologic pathology (e.g., cell-mediated vs. humoral-mediated pathologies), and the time course of the condition (e.g., monophasic vs. recurring conditions). Some immune-mediated conditions target the central nervous system (CNS), some target the peripheral nervous system (PNS), while a few can cause abnormalities in both systems. Additionally, within the CNS and PNS, there are a multitude of well-defined specific targets of a pathologic immune system.

Molecular testing and diagnostics have improved our ability to tailor therapeutic interventions to individual conditions. Improved recognition of immune-mediated neurologic conditions has offered us the opportunity to initiate therapy earlier in the course of the disease and potentially limit tissue damage. The symptoms experienced by the patient depend on the part of the nervous system targeted, and the outcome depends on the severity of damage, timing of therapy, success of therapy, ability to repair, and rehabilitation-induced improvements. This chapter explores the general principles, diagnostic approaches, and therapeutic options for immune-mediated neurologic conditions.

GENERAL PRINCIPLES

Definition

Neuroimmunological disorders include both autoimmune disorders and autoinflammatory disorders. Autoimmune disorders are defined by a pathologic immune system that targets a specific antigen or limited number of antigens, whereas autoinflammatory disorders are caused by a dysregulated immune response against a number of potential antigens. Autoinflammatory conditions are much rarer than autoimmune conditions and will not be covered in this chapter.

Epidemiology

In general, pediatric neuroimmunological disorders are rare. The incidence and prevalence of autoimmune disorders have been steadily increasing over the last 5 decades.[1,2] For pediatric neuroimmunological disorders, there is not as much data as compared with adults. The incidence and prevalence depend on the specific condition being considered. For example, the reported incidence of pediatric multiple sclerosis ranges from .05 to 2.85 per 100,000, while the prevalence ranges from .69 to 26.92 per 100,000.[3] Anti-NMDA receptor antibody autoimmune encephalitis is rarer, with an estimated incidence of 1 to 1.5 per million population.[4] Acute inflammatory demyelinating polyneuropathy (i.e., AIDP or GuillainBarrè syndrome [GBS]) has an estimated incidence of .62 to .75 per 100,000 person-years.[5]

Classification

Classification of neuroimmunological disorders can occur based on a number of factors, including site of inflammation (e.g., CNS vs. PNS), type of immune-mediated pathology (e.g., antibody-mediated vs. cell-mediated), or time course of the condition (e.g., monophasic vs. multiphasic/recurring). For the purposes of this chapter, a classification based on the site of pathology is most helpful. Table 12.1 lists several of the prototypical neurologic autoimmune disorders arranged by site of pathology. Ultimately, the classification of these disorders does have implications for the spectrum of symptoms a patient may experience, the treatments offered acutely, the potential long-term treatments, and the possibility of relapses. Yet, regardless of the underlying cause, the approaches to rehabilitation for brain-based, spinal cord-based, or peripheral nerve-based pathologies will have significant overlap.

Etiology

The exact etiologies for the various autoimmune disorders of the nervous system are not known. Developing an autoreactive immune system requires a breakdown in immunologic tolerance. This is likely the result of:

- Certain genetic risk factors
- Certain exposures (e.g., infectious and/or environmental) at certain times of the patient's life

Thus, there is not a single cause or etiology for any of the disorders covered in this chapter.

Pathophysiology

The pathophysiology of these disorders varies based on the underlying immune system abnormality. For antibody-mediated disorders such as anti-NMDAR antibody-mediated encephalitis, anti-aquaporin-4 (AQP4) antibody-mediated neuromyelitis optica spectrum disorder (NMOSD), anti-myelin oligodendroglial glycoprotein (MOG) antibody-associated

Table 12.1 Examples of Neuroimmunological Disorders

DISORDER	SITE OF INFLAMMATION	IMMUNOLOGIC CAUSE	MONOPHASIC VS. RECURRING
Central nervous system			
Multiple sclerosis	Brain, optic nerve, spinal cord	B and T cell-mediated	Recurring
Neuromyelitis optica spectrum disorder	Brain, optic nerve, spinal cord	Anti-AQP4 antibody	Recurring
Anti-MOG antibody-associated disorder	Brain, optic nerve, spinal cord	Anti-MOG antibody	Both
Autoimmune encephalitis	Brain	Anti-NMDA R antibody	Both
ADEM	Brain, optic nerve, spinal cord	B and T cell-mediated	Monophasic
Idiopathic transverse myelitis	Spinal cord	B and T cell-mediated	Monophasic
Acute flaccid myelitis	Spinal cord gray matter	Infectious, B and T cell-mediated	Monophasic
Peripheral nervous system			
Acute inflammatory demyelinating polyneuropathy	Peripheral nerve myelin	B and T cell-mediated	Monophasic
Chronic inflammatory demyelinating polyneuropathy	Peripheral nerve axons	B and T cell-mediated	Recurring
Myasthenia gravis	Neuromuscular junction	Anti-AchR antibody	Recurring

AchR, acetylcholine receptor; ADEM, acute disseminated encephalomyelitis; AQP4, aquaporin-4; MOG, myelin oligodendroglial glycoprotein.

disorder, or anti-acetylcholine receptor (AchR) antibody-mediated myasthenia gravis, there can be antibody-mediated dysfunction of targeted cells and/or destruction to targeted cells and surrounding tissue. Damage to cells and tissue can occur via antibody-mediated complement pathway activation or antibody-mediated cellular cytotoxic damage.[6–8]

T cell-mediated disorders have a variety of pathologic effects in neuroimmunological conditions. These include:

- B cell activation
- Class switching of antibodies
- Cytotoxic cell-mediated damage

While the most common neurologic autoimmune disease associated with pathologic T cells is multiple sclerosis, there are many others reportedly related to pathologic T cells. For example, Rasmussen disease and narcolepsy have reportedly been associated with T cell-mediated brain inflammation.[9,10]

Mechanism of Injury

Immune-mediated dysfunction or destruction can occur via a variety of mechanisms.

- The binding of an antibody to a target can cause dysfunction of the target cell or downregulation of the bound antigen (e.g., anti-NMDAR antibody encephalitis).[11]
- Binding of the anti-AQP4 antibody to astrocytes causes cellular damage and tissue necrosis primarily through activation of the complement pathway and formation of a membrane attack complex.[7]
- CD8+ T cell-mediated cellular damage can trigger target cells to undergo apoptosis.[12]
- Autoreactive T cells have also been implicated in neurologic disease by inhibiting neurogenesis and repair pathways.[13]

Importantly, the mechanism of injury for neuroimmunological disorders likely explains why certain therapies work in some conditions versus others. Therapies that target circulating antibodies, complement activation, or T cell expansion would likely have different success depending on the underlying cause of the disease.

DIAGNOSIS

Risk Factors

Autoimmune disorders of the nervous system are thought to result from certain individuals experiencing certain exposures at certain times of their life. Genetic risks have been identified for almost all of the conditions reviewed in this chapter. In multiple sclerosis, for example, there have been numerous studies associating multiple sclerosis with the HLA-DRB1*15:01 allele.[14] Additional studies have found an increased prevalence of autoimmune disorders in the families of pediatric patients with multiple sclerosis.[15]

"Exposures" that can trigger autoimmune disorders in certain patients include underlying tumors, infections, nutritional deficiencies, and certain bacterial profiles in the intestinal microbiome. Anti-NMDAR antibody-mediated encephalitis has been described in children with ovarian teratomas.[16] The benign tumor likely acts as a source of antigen that triggers the immune system against the otherwise normal CNS antigen. Infectious triggers of neuroimmunological disorders have also been described. Similar to a teratoma, the infectious agent likely contains an antigen that resembles a self-antigen. In the wake of an infection, a child's immune system can then mistakenly target the normal antigen. For example, NMDAR antibody autoimmune encephalitis has been described in the weeks following herpes simplex virus (HSV) encephalitis, and acute inflammatory demyelinating polyneuropathy (AIDP; also known as GBS) has been linked to infections with *Campylobacter jejuni*.[17,18] Nutritionally, vitamin D_3 deficiency has been implicated in a number of neuroimmunological conditions, including multiple sclerosis, recurrent transverse myelitis, myasthenia gravis, and chronic inflammatory demyelinating polyneuropathy (CIDP).[19–22]

Clinical Presentation and Symptoms

The clinical presentation of these various neuroimmunological disorders is completely dependent on the immune system's target tissue. The disorders can affect cognition, vision, strength, sensation, gait, bowel, and bladder function. Some autoimmune disorders of the nervous system can cause seizures or movement disorders. The kinetics of onset can be acute, subacute, and chronic. Typical clinical presentations of various neuroimmunological disorders are summarized in Table 12.2. Given the variety of clinical presentations, clinicians must remain vigilant when evaluating patients with new-onset neurologic dysfunction.

Physical Examination

Physical examination of a child with suspected neuroimmunological disorder is dictated by the presentation, chief complaint, and differential diagnosis. The goal of physical examination is to simultaneously confirm and qualify the subjective symptoms described while also looking to document evidence of unrecognized neurologic dysfunction. For example, in a patient presenting with weakness, a physical examination would include a confrontational motor examination of the "weak" limbs to determine the severity of weakness and determine if the weakness follows an upper or lower motor neuron pattern. While the classic findings of increased tone and hyperactive reflexes may be absent in the early days of an upper motor neuron injury, their presence essentially rules out an isolated PNS pathology. Neuromuscular junction disorders (e.g., myasthenia gravis, botulism) are classically described as having fatigable weakness, but this may vary at presentation.

Table 12.2 Clinical Presentations and Symptoms of Neuroimmunological Disorders

DISORDER	COMMON SYMPTOMS	TIME COURSE AT ONSET
Central nervous system		
Multiple sclerosis	Blurred vision, numbness, weakness, gait changes	Acute, subacute
Neuromyelitis optica spectrum disorder	Blurred vision, numbness, weakness, gait changes, intractable vomiting	Acute, subacute
Anti-MOG antibody-associated disorder	Blurred vision, numbness, weakness, gait changes, encephalopathy	Acute, subacute
Autoimmune encephalitis	Confusion, personality changes, seizures, movement disorders	Acute, subacute
ADEM	Confusion, encephalopathy, weakness, ataxia	Acute, subacute
Idiopathic transverse myelitis	Weakness, numbness, gait changes, bladder dysfunction	Acute, subacute
Acute flaccid myelitis	Weakness, gait changes	Acute
Peripheral nervous system		
Acute inflammatory demyelinating polyneuropathy	Numbness, weakness, gait changes	Acute, subacute
Chronic inflammatory demyelinating polyneuropathy	Numbness, weakness, gait changes	Chronic
Myasthenia gravis	Weakness, double vision, dysphagia	Acute, subacute

ADEM, acute disseminated encephalomyelitis; MOG, myelin oligodendroglial glycoprotein.

Simultaneously, completing a sensory examination in patients with weakness can provide clinicians with vital information to support the localization of the injury. Patients with a sensory level should be suspected to have a spinal cord pathology until proven otherwise. Similarly, patients with loss of dorsal column function on one side of the body and loss of pain/temperature sensation on the contralateral body should be evaluated for a CNS pathology below the medulla.

A careful physical examination can be critically important in designing appropriate rehabilitation strategies for patients. Documenting cognitive deficits can be critical to ensuring that patients can adequately participate in complex therapeutic endeavors. Furthermore, some patients can have mixed patterns of weakness in different limbs. For example, in patients with acute flaccid myelitis, many will have lower motor neuron patterns of weakness in the upper extremities and upper motor neuron patterns of weakness in the lower extremities.[23] The rehabilitation approaches for a patient such as this would be distinctly different from a patient with diffuse upper motor neuron dysfunction. Additionally, nerve transfer surgeries are a viable option for muscles denervated from an anterior horn cell pathology, but would not be of value in the setting of an isolated corticospinal tract injury.[24,25] Thus, a comprehensive physical examination is critical for both diagnostic purposes and for planning rehabilitation strategies.

DIAGNOSTIC EVALUATION

Diagnostic evaluation of a patient with suspected neuroimmunological condition is dictated by the differential diagnosis and the need to balance etiology evaluations with therapeutic interventions. In general, when possible, treatment should not be delayed to facilitate diagnostic testing. Patients will often be subjected to laboratory testing, imaging, and/or physiologic testing (e.g., nerve conduction studies [NCS] or EEG).

Laboratory Studies

Laboratory studies of patients with suspected neuroimmunological disorders can be classified by the body fluid being tested (i.e., blood vs. cerebrospinal fluid [CSF]) and whether or not the test is a specific or nonspecific test for autoimmune disease. Laboratory tests to consider include:

- Serum autoantibody panels
- CSF autoantibody panels
- CSF white blood cell count
- CSF protein
- CSF and serum glucose
- Oligoclonal bands (CSF and serum)
- Immunoglobulin G (IgG) index
- CSF IgG synthesis rate

Specific tests for autoimmune conditions comprised autoantibody tests where the presence of a known pathologic antibody (in the correct clinical

context) is considered diagnostic of a condition. For example, in a patient who presents with new-onset psychosis, seizures, and chorea, a positive anti-NMDAR antibody is considered diagnostic of anti-NMDAR antibody encephalitis.

Important considerations when interpreting the results of autoantibody tests include the clinical context, the source of fluid (i.e., serum vs. CSF), the titer of the antibody, prior immunotherapies, and the assay used to detect the antibody. Each antibody test has operating characteristics that are important in interpreting the results. For example, the anti-AQP4 antibody, which is specific for NMOSD, can be screened for with a number of assays, including immunofluorescence, ELISA, and cell-based assays. Cell-based assay is considered the gold standard, while enzyme-linked immunosorbent assay can frequently have false positive results (especially when reporting a low titer).[26] Relative to the impact of body fluid source of testing, the anti-NMDAR antibody detected in CSF is more sensitive and specific for active autoimmune encephalitis than a serum-based result.[27]

Nonspecific tests for autoimmune conditions of the nervous system include CSF cell count, CSF protein, CSF IgG index, CSF IgG synthesis rate, and test for unmatched oligoclonal bands within the CSF. The latter three tests (i.e., IgG index, IgG synthesis rate, and oligoclonal bands) require a matched CSF and serum sample to be submitted for analysis at the same time. Elevated white blood cell count, protein, or evidence of intrathecal synthesis of antibodies can be found in patients with autoimmune conditions of the nervous system, but can also occur in infections or neoplastic processes. Thus, these nonspecific tests can be helpful in certain clinical situations.

Imaging Assessments

Autoimmune conditions of the CNS often cause changes on MRI. The location of signal change and the pattern of signal abnormalities can assist in narrowing the differential diagnosis. There are several imaging patterns that are classically associated with certain conditions. Multiple sclerosis patients have white matter changes that are periventricular or juxtacortical. The periventricular lesions are often perpendicularly oriented to the ventricles.[28] Anti-AQP4-associated NMOSD is associated with longitudinally extensive lesions within the spinal cord.[29] In acute flaccid myelitis, half of the patients have isolated signal change in the gray matter of the spinal cords, while half have signal change in both the gray and white matter.[23] Distinct from other autoimmune conditions of the CNS, anti-NMDAR antibody encephalitis patients usually *do not* have distinct lesions on brain MRI. Rather, these patients can suffer from significant brain atrophy.[30]

A burgeoning imaging technology for CNS autoimmune conditions is optical coherence tomography (OCT). This device uses near infrared light to measure the cell layers of the retina and can document damage to the retinal nerve fiber layer (RNFL) that results from optic neuritis.

Documenting prior optic neuritis with this technology can be helpful in the diagnosis and monitoring of multiple sclerosis, NMOSD, and anti-MOG-associated disorder.[31,32]

Neurophysiology Testing

In certain settings, diagnosis and management of patients with neuroimmunological disorders are facilitated by physiologic studies. The three most common are NCS, electromyography (EMG), and EEG. NCS and EMG are routinely done at the same time and are critical in the evaluation of patients with possible or confirmed peripheral nerve disorders. NCS/EMG studies involve the use of electrodes or needles to stimulate a nerve or muscle and measure the velocity and amplitude of responses. NCS and EMG are critical in the diagnosis of AIDP, CIDP, and myasthenia gravis. NCS and EMG can be used for assessment of acute flaccid myelitis (AFM) patients who are being evaluated for possible nerve transfer procedures.

EEG utilizes electrodes placed on the scalp in an arrayed fashion. The electrodes are attached to an amplifier which allows recording of neuronal firing patterns. The data from an EEG can be used to screen for seizures or document cellular dysfunction, which appears as slowing on the EEG. Patients with autoimmune encephalitis will frequently have either ongoing seizures or slowing noted on EEG. A normal EEG significantly reduces the possibility of autoimmune encephalitis.[33]

TREATMENT

Guiding Principles

Treating autoimmune conditions of the nervous system can be broken down into four categories: acute treatment, preventive therapy, symptom management, and rehabilitative programs to improve function. Several of these therapeutic interventions can happen simultaneously. For example, preventive therapy, symptom management, and rehabilitation can happen concurrently. Patients admitted to acute care settings can have rehabilitation services started early in the course of illness. The guiding principle of acute therapy is to "put the fire out" to limit the amount of damage done.

Initial Management

The focus of initial management of patients with neuroimmunological disorders is always on general stabilization. Patients with brainstem inflammation, spinal cord inflammation, peripheral nerve disease, and/or neuromuscular junction dysfunction are at risk of respiratory distress. Proper airway, breathing, and cardiovascular support is needed in all of these patients. Beyond initial stabilization, the focus of initial management is to suppress the immune-mediated inflammation. The most common therapies used to acutely treat neuroimmunological conditions include:

- High-dose corticosteroids
- Plasmapheresis/plasma exchange (PLEX)

- Intravenous immunoglobulin (IVIG)
- Cyclophosphamide

While high-dose corticosteroids are the therapy of choice for most immune-mediated conditions of the nervous system, they are not used in the setting of AIDP.[34] PLEX therapy has increased in use within the pediatric population over the last 10 years. Open-label studies have established its safety and efficacy in children with immune-mediated conditions of the nervous system.[35] The use of PLEX has been shown to improve outcomes in a variety of patients.[23,36,37]

Ongoing Care

Ongoing care includes preventive therapy, symptomatic management, and rehabilitation. During diagnostic evaluation, testing is completed to determine which patients may be at risk of future relapses. These include patients with multiple sclerosis, NMOSD, anti-MOG antibody-associated disorder, some cases of autoimmune encephalitis, CIDP, and myasthenia gravis. Preventing new inflammatory events in these patients uses various medication strategies that are dependent on the diagnosis. For example, in multiple sclerosis, there are numerous Food and Drug Administration-approved therapies for adults that are used in an off-label fashion in children.[38,39] For many of the pediatric autoimmune conditions, immunomodulatory or immunosuppressive therapy is used in an off-label fashion. Patients may require lifelong therapy to prevent relapses.

Depending on the site of inflammation and the severity of damage, patients may suffer from a variety of symptoms that require independent treatment. Some of the most common symptoms requiring therapy are spasticity, neuropathic pain, neurogenic bladder dysfunction, and neurogenic bowel dysfunction. Relative to pain, it is important for practitioners to take a detailed history in order to separate out the various types of pain that patients may suffer from, including neuropathic pain, pain associated with spasticity, orthopedic pain, or tonic phasic spasms.

Ongoing care for pediatric patients with neuroimmunological conditions also requires rehabilitation. This can include cognitive, physical, and occupational therapy. In some settings, activity-based rehabilitation has shown unique benefits and can be combined with functional electrical stimulation.[40,41] Functional recovery in children is dictated by the degree of nervous system repair and the plasticity-based compensation that can occur. Patients often require prolonged time courses of therapy to achieve functional improvement.

Treatment Controversies

Given the rarity of pediatric neuroimmunological conditions, there is a limited number of randomized controlled studies in the literature. Thus, therapeutic decisions are often based on clinician experience, retrospective studies, and open-label, nonrandomized observational trials. In the acute care setting, there remains variability in the use of high-dose

corticosteroids, IVIG, and PLEX. Despite studies verifying the safety and efficacy of PLEX in pediatric patients, many practitioners are uncomfortable with the procedure and its requirement for a central line. Furthermore, among centers that utilize PLEX therapy, some will decide on its initiation based on the relative clinical response to corticosteroids (i.e., using PLEX in patients who fail to show a meaningful response), while others initiate PLEX in all patients with significant deficits. Limited data suggest that delays in PLEX therapy should be avoided in neuromyelitis optica spectrum disorder (NMOSD) patients given the correlation between delays and poor outcomes in these patients.[42] Early PLEX therapy should be considered in all patients with significant deficits from suspected neuroimmunological conditions.

ADDITIONAL CONSIDERATIONS

When evaluating patients for autoimmune conditions of the nervous system, it is important to complete a thorough assessment of the child. Published data have identified a higher than expected frequency of cognitive dysfunction in children with nonbrain-based disorders (e.g., transverse myelitis).[43] This was an unexpected result, given the focus of inflammation in the spinal cord, but reinforced the need for children to receive multidisciplinary care. Furthermore, pediatric patients have dynamic needs over the course of their development that may require intermittent evaluations and interventions even after monophasic immune-mediated attacks on the nervous system.

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Pediatric Electrodiagnosis

13

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GENERAL PRINCIPLES

- Electrodiagnostic (EDX) testing is performed to evaluate the function of the peripheral nervous system. The most common components of the test include nerve conduction studies (NCS) and electromyography (EMG), and in some cases repetitive nerve stimulation (RNS). Single-fiber EMG (SFEMG) or stimulated EMG can be performed to evaluate for neuromuscular junction (NMJ) disorders. Other less commonly performed components of EDX include somatosensory evoked potentials (SSEPs), brainstem auditory evoked responses (BAERs), and sympathetic skin responses.
- Indications for EDX testing in children include localization of lesion in the peripheral nervous system, characterization of pathology (demyelinating vs. axonal vs. myopathic) and its severity, and for prognostic guidance. Indications for EDX have evolved over time as genetic testing has allowed more precise identification of many genetic neuromuscular conditions; however, the test continues to be relevant in children to characterize the neurophysiologic phenotype when genetic testing is nonconclusive.[1,2]
- There are differences in the performance of EDX testing in children and adults.
  - *Reason for referral:* The types of pathology seen in a pediatric EMG lab are different from an adult EMG lab, where degenerative conditions such as carpal tunnel syndrome and radiculopathy are most common. The most common reasons for EDX testing in children in one hospital included polyneuropathy (29%–35% of the requested studies), suspected mononeuropathy (13%–26% of the requests), and various focal neurologic symptoms in one or more extremities (11%–25% of the requests).[1]
  - When choosing stimulation sites, anatomical landmarks are often used rather than the standard adult distances due to the small size of the limbs of pediatric patients. As entrapment neuropathies are less common in children, the distal latency is less important than the velocity and amplitude.[3]
- There are factors to be considered during EDX testing in infants and young children.
  - *NCS:* It is technically difficult to obtain sural or superficial peroneal sensory nerve action potentials (SNAPs) due to their subcutaneous

fat. Medial and lateral plantar responses (orthodromic) are more easily obtainable.[4]
    - It is preferable to use an infant stimulator for children <18 to 24 months old, with an anode-catheter distance of .5 to 1 cm (compared with 3 cm on a standard adult stimulator).[5]
- Limb temperature is an important variable to control due to its effect on latencies and amplitudes. The standard adult temperature goals of >30°C for the lower extremity and >32°C for the upper extremity are recommended for children,[3] although a recent practice guideline from the American Association of Neuromuscular and Electrodiagnostic Medicine (AANEM) recommended warming to 34°C to 36°C for the upper extremities and 32°C to 34°C for the lower extremities in children.[3]
- *Pain and sedation:* EDX testing has been perceived by many medical providers to be a painful test. A recent prospective study on pain perception during EMG found that children >4 years old rated discomfort from EDX testing on average in the same range as venipuncture. More pain was reported by families when more than one muscle or proximal muscles were needled in patients <4 years old.[6] Performing a "focused EMG" in children to answer a clinical question while balancing the child's comfort has been advocated by Pitt.[2] Care in how the test is described can reduce the child's anxiety. Avoid the use of words "needle" and "shock"; instead use "wire" or "special microphone" and "tapping sensation" to describe the test. The presence of a child life specialist is also very helpful for distraction and support. Some centers use topical anesthetics (such as vapocoolant spray or lidocaine cream), and a small surface vibrator may help provide additional distraction during needle insertion.[3]
- Behavioral distress is most frequent in children aged 2 to 6 years old. The use of sedation is controversial. While it allows for easy and accurate performance of NCS and testing of insertional and rest activity on EMG, it limits evaluation of voluntary motor unit analysis (sedation can be lightened for the EMG portion of the test, but only allows limited voluntary motor unit action potential [MUAP] analysis). The risk of sedation must also be considered. Sedation is needed for rapid repetitive stimulation due to its painful nature and is helpful in performing repetitive stimulation and facial studies in children less than 5 years of age.
- Normative data are different in children compared with adults.
    - In a newborn infant, sensory and motor velocities are approximately 50% of the adult values. Axon diameter and myelination gradually increase from birth to adulthood, with adult values reached by 3 to 6 years of age.[7,8]
- *Studies with normative data:* Seven articles with high-quality normative data from preterm infants to older children were recently reviewed by an AANEM taskforce.[4] The most recent study with the largest cohort was by Ryan et al. published in 2019,[9] which had high-quality data but was limited by a small sample size of infants and with a recommendation to compare data with those from earlier studies as needed for these age groups.[4] The normative data from this study are summarized in Exhibits 13.1 and 13.2.

Exhibit 13.1 Table of Normative Data for Motor Nerves

LOWER LIMB MOTOR NCS

Peroneal (Fibular)			
AGE	**ONSET LATENCY (ms)**	**AMPLITUDE (mV)**	**NCV (m/s)**
0–<1 month	2.5 (0.5)	2.1 (1.1)	31 (14)
1–<6 months	2.0 (0.5)	2.8 (1.7)	41 (4)
6–<12 months	2.0 (0.4)	3.4 (1.2)	44 (7)
12–<24 months	2.2 (0.4)	3.7 (1.3)	48 (8)
2–<3 years	2.4 (0.4)	3.7 (1.5)	49 (5)
3–<4 years	2.9 (0.7)	4.4 (1.6)	50 (5)
4–<5 years	3.1 (0.4)	4.3 (1.8)	50 (4)
5–<10 years	3.6 (0.6)	4.7 (1.6)	52 (4)
10–<15 years	4.2 (0.7)	5.4 (2.0)	51 (5)
15–<18 years	4.4 (0.7)	6.4 (2.1)	50 (3)

Tibial			
AGE	**ONSET LATENCY (ms)**	**AMPLITUDE (mV)**	**NCV (m/s)**
0–<1 month	2.7 (0.2)	5.3 (1.6)	24 (3)
1–<6 months	2.3 (0.2)	9.5 (0.9)	40 (5)
6–<12 months	2.7 (0.9)	10.0 (2.8)	41 (5)
12–<24 months	2.4 (0.4)	11.1 (3.0)	46 (4)
2–<3 years	2.5 (0.4)	11.1 (3.1)	51 (5)
3–<5 years	2.8 (0.4)	13.6 (5.2)	50 (6)
5–<10 years	3.3 (0.6)	12.8 (3.8)	52 (5.0)
10–<15 years	4.0 (0.7)	11.8 (3.6)	50 (4)
15–<18 years	4.2 (0.6)	13.2 (3.9)	50 (4)

UPPER LIMB MOTOR NCS

Median			
AGE	**ONSET LATENCY (ms)**	**AMPLITUDE (mV)**	**NCV (m/s)**
0–<1 month	2.2 (0.2)	2.2 (1.6)	25 (3)
1–<6 months	1.7 (0.1)	3.3 (0.8)	37 (9)
6–<12 months	2.1 (0.2)	5.9 (2.5)	45 (13)
12–<24 months	2.2 (0.2)	5.7 (1.9)	47 (5)
2–<5 years	2.3 (0.3)	7.2 (1.7)	51 (6)
5–<10 years	2.9 (0.6)	8.9 (2.8)	56 (7)
10–<15 years	3.3 (0.4)	10.9 (2.7)	58 (4)
15–<18 years	3.3 (0.4)	11.6 (2.9)	59 (3)

Ulnar			
AGE	**ONSET LATENCY (ms)**	**AMPLITUDE (mV)**	**NCV (m/s)**
0–<1 month	2.2 (0.5)	3.8 (1.6)	35 (7)
1–<6 months	1.8 (0.3)	4.5 (1.9)	43 (7)
6–<12 months	1.7 (0.2)	5.4 (1.5)	51 (7)
12–<24 months	1.7 (0.2)	5.8 (1.8)	53 (7)
2–<3 years	1.7 (0.2)	6.2 (1.9)	56 (6)
3–<4 years	1.9 (0.3)	7.8 (1.9)	58 (6)
4–<5 years	1.9 (0.4)	7.2 (1.7)	60 (6)
5–<10 years	2.1 (0.3)	9.2 (2.7)	61 (6)
10–<15 years	2.5 (0.3)	10.7 (2.4)	62 (5)
15–<18 years	2.6 (0.3)	11.9 (2.5)	63 (5)

Notes: The tables report the mean values with the standard deviations in parentheses.
Amplitudes were taken from the proximal stimulation site (if both proximal and distal sites tested). Distal latency was measured from the onset (or rise of the negative deflection) of the compound motor nerve action potential.

NCS, nerve conduction studies; NCV, nerve conduction velocity

Source: Adapted from Ryan CS, Conlee EM, Sharma R, et al. Nerve conduction normal values for electrodiagnosis in pediatric patients. *Muscle Nerve.* 2019;60(2):155–160. doi:10.1002/mus.26499

Exhibit 13.2 Table of Normative Data for Sensory Nerves (Antidromic)

LOWER LIMB SENSORY NCS

Sural			
AGE	**PEAK LATENCY (ms)**	**AMPLITUDE (mcV)**	**NCV (m/s)**
0–<6 months	-	-	-
6–<12 months	2.4 (0.8)	18 (10)	-
12–<24 months	1.9 (0.3)	20 (7)	-
2–<5 years	2.2 (0.3)	21 (9)	57 (5)
5–<10 years	2.9 (0.6)	21 (10)	56 (7)
10–<15 years	3.6 (0.3)	18 (8)	52 (6)
15–<18 years	3.6 (0.3)	21 (9)	51 (5)

Medial Plantar			
AGE	**PEAK LATENCY (ms)**	**AMPLITUDE (mcV)**	**NCV (m/s)**
0–<1 months	3.6 (2.6)	10 (8)	-
1–<6 months	1.8 (0.6)	21 (11)	-
6–<12 months	1.7 (0.2)	34 (15)	-
12–<24 months	1.8 (0.3)	32 (16)	-
2–<3 years	1.9 (0.3)	36 (18)	-
3–<4 years	2.2 (0.3)	38 (17)	-
4–<5 years	2.2 (0.3)	38 (17)	-
5–<10 years	2.5 (0.4)	34 (16)	-
10–<15 years	2.9 (0.4)	27 (13)	-
15–<18 years	3.0 (0.4)	28 (13)	-

UPPER LIMB SENSORY NCS

Ulnar			
AGE	**PEAK LATENCY (ms)**	**AMPLITUDE (mcV)**	**NCV (m/s)**
0–<2 years	–	–	-
2–<5 years	1.8 (0.2)	41 (15)	65 (7)
5–<10 years	2.2 (0.3)	41 (15)	65 (5)
10–<15 years	2.6 (0.3)	41 (12)	67 (5)
15–<18 years	2.6 (0.2)	44 (17)	67 (5)

Median			
AGE	**PEAK LATENCY (ms)**	**AMPLITUDE (mcV)**	**NCV (m/s)**
0–<1 month	2.5 (0.2)	24 (7)	26 (4)
1–<6 months	2.0 (0.3)	36 (12)	38 (9)
6–<12 months	2.1 (0.3)	48 (8)	53 (20)
12–<24 months	2.1 (0.2)	54 (23)	55 (6)
2–<3 years	2.2 (0.3)	62 (24)	59 (6)
3–<5 years	1.8 (0.2)	54 (20)	61 (5)
5–<10 years	2.5 (0.3)	55 (19)	64 (5)
10–<15 years	2.9 (0.3)	50 (15)	64 (4)
15–<18 years	2.9 (0.3)	56 (18)	65 (4)

Notes: The tables report the mean values with the standard deviations in parentheses.
Amplitudes are measured as peak-to-peak amplitude at the distal stimulation site. Sensory distal latencies are recorded as the peak latency of the distal site. Conduction velocities were determined with onset latencies.

NCS, nerve conduction studies; NCV, nerve conduction velocity

Source: Adapted from Ryan CS, Conlee EM, Sharma R, et al. Nerve conduction normal values for electrodiagnosis in pediatric patients. Muscle Nerve. 2019;60(2):155–160. doi:10.1002/mus.26499

TEST COMPONENTS

- *Sensory NCS* including SNAPs, with measurement of amplitude and peak latency
- *Motor NCS* including compound motor nerve action potentials (CMAPs), with measurement of amplitude, onset latency, and nerve conduction velocity (NCV)
- *Late responses:* F-waves and H-reflexes
- *Needle EMG* to evaluate for insertional activity, rest activity, motor unit configuration, polyphasicity, and recruitment
- *MUAP size* (also age-dependent)
- *RNS:* used to evaluate for NMJ disorders; performed at a low rate (2–3 Hz) if there is concern for postsynaptic NMJ disorder and at both low and high rate (20–50 Hz) or after brief exercise simulating high-rate stimulation if there is concern for presynaptic NMJ disorder
- *SFEMG, stimulated SFEMG (SSFEMG), or stimulated potential analysis using concentric needle electrodes (SPACE):* sensitive tests for NMJ disorders but not specific (can also be abnormal in motor neuron disorders)

ROLE OF ELECTRODIAGNOSIS IN THE EVALUATION OF SPECIFIC NEUROMUSCULAR CONDITIONS

Motor Neuron Disorders

Spinal Muscular Atrophy

- Genetic testing is first line, but EDX may be helpful in confirming clinical suspicion while awaiting genetic testing results or in cases with non-5q spinal muscular atrophy (SMA).[3]
- *NCS:* SNAPs are normal. Motor NCS can be normal early in the disease process, with reduced CMAP amplitudes correlating with degree of weakness.[3]
- EMG shows evidence of active denervation/chronic reinnervation, although this is variable depending on SMA subtype and stage in the disease process (may be normal in the early stages of SMA). To improve accuracy, it may be helpful to test multiple muscles in different limbs, including the genioglossus.[2,3]

Juvenile Amyotrophic Lateral Sclerosis

- EDX findings are similar to those in adult with normal SNAPs, and decreased CMAPs with normal NCV. EMG shows signs of active denervation and reinnervation.[3]

Acute Flaccid Myelitis, Including West Nile Virus and Poliomyelitis

- *NCS:* SNAPs are normal, with decreased motor amplitudes after the first 1 to 3 weeks and decreased recruitment and fibrillations on EMG.[10]

Sensory Neuronopathy

Friedreich Ataxia

SNAPs are absent or reduced, with motor NCVs slightly decreased or normal. EMG can show denervation and reinnervation changes.[3,11]

Radiculopathies and Plexopathies

Neonatal Brachial Plexopathy

- EDX establishes the presence of a peripheral nerve injury (vs. central nervous system [CNS] injury of the brain or spinal cord) and localizes the areas of involvement.
- EDX determines if root avulsion is present versus a more distal postganglionic plexus injury. A nerve root avulsion portends a graver prognosis and suggests a more likely need for surgery.[12] Be aware that combined preganglionic and postganglionic lesions can exist.
- EDX clarifies the pathophysiology of the injury, that is, axonal versus demyelinating.
- EDX assesses severity.
    - CMAP amplitudes are helpful in determining the degree of axonal integrity of the involved nerves. To assess the upper trunk, conduction studies can be performed across the brachial plexus, stimulating at the Erb's point and recording from the deltoid (axillary nerve), or stimulating at either the Erb's point or at the axilla and recording from the biceps (musculocutaneous nerve). Technical pitfalls include difficulty obtaining supramaximal stimulation at the Erb's point, which may give a mistaken impression of a conduction block, and costimulation of the adjacent nerves.
    - EMG can provide information on motor continuity, but caution is recommended as continuity by EMG has not correlated well with outcome in infants.[13,14]
    - EDX testing is controversial in the evaluation of neonatal brachial plexopathy and is not always needed to make what most clinicians consider a clinical diagnosis. EMG in plexopathy is commonly used as an adjunct to other modalities such as MRI, CT myelogram imaging, and clinical examination.
- Most surgeons find EDX useful in presurgical planning as it can establish if there is some degree of axonal continuity and can also help identify potential donor nerve options if surgery is a consideration.
- *Limitations of EDX:* EDX can be technically difficult to perform in infants. The optimal timing is not clearly established and there is concern for EDX providing an overly optimistic view of axonal integrity, possibly due to a much smaller fiber diameter (more fibers "seen" in the uptake area of the needle), aberrant innervation, or abnormal agonist/antagonist.[13,15] Thus, MUAPs may be observed despite lack of observable movement clinically.

Traumatic or Nonneonatal Brachial Plexopathy

- EDX establishes the presence of peripheral injury and localizes lesion as pre/postganglionic. It also helps rule out other diagnoses on the differential (e.g., multifocal mononeuropathy and polyradiculopathy).
- EDX identifies mMyokymia if postradiation plexopathy is a concern.
- *EMG:* Typically, extensive sampling is performed in adults to cover a variety of nerve roots and brachial plexus trunks and divisions, but must be more focused in infants/children.

Lumbosacral Plexopathy

- It is rare in children.
- NCS show decreased sensory and motor amplitudes (help differentiate from radiculopathy).
- EMG shows neurogenic changes in the plexus innervated muscles. It is helpful to test hip musculature (gluteus maximus or gluteus medius) to differentiate lumbosacral plexus versus sciatic neuropathy, and hip adductors to distinguish lumbar plexopathy versus femoral neuropathy.[3]

Radiculopathy

- It is less common in childhood and adolescence.
- *EDX findings are similar as in adults:* SNAPs are normal (even in the setting of sensory loss due to the location of the dorsal root ganglion proximal to the neuroforamen), with intact distal sensory nerve segment. CMAPs may be decreased or normal.
- EMG shows denervation in a myotomal distribution.

Polyneuropathies

- The role of EDX is to confirm the presence of peripheral nerve disease, determine the types of fiber involved (sensory and/or motor), assess the severity, and describe the features of the neuropathy (demyelinating vs. axonal, symmetric vs. asymmetric, uniform vs. nonuniform CMAP configuration). The sensitivity of EDX in confirming a neurogenic diagnosis has been reported to be as high as 99.5%.[16]
- Polyneuropathies can be classified into hereditary and acquired groups. Hereditary neuropathies are more common in the pediatric population than acquired neuropathies.

Hereditary Neuropathies

- CharcotMarieTooth (CMT) disease is the most common cause of hereditary neuropathy and is classified into types on the basis of neurophysiologic features (demyelinating, axonal), genetic mutation, and clinical

features.[2,3,17] CMT type 1 has demyelinating features, with severely decreased NCVs and uniform slowing that is typically symmetric on side-to-side comparison. SNAPs and CMAPs are reduced in lower extremities > upper extremities and have prolonged latencies with uniform CMAP configuration.[2,18] CMT type 2 has axonal features and is associated with multiple genetic mutations. EDX findings include low-amplitude SNAPs and CMAPs with normal NCVs and evidence of denervation/reinnervation on EMG.[19,20]

Acquired Neuropathies

- Acquired polyneuropathies can be due to multiple causes, including immune-mediated inflammation, toxins, metabolic or other systemic disorders, infection, vasculitis, or vitamin deficiency.
  - Guillain–Barrè syndrome (GBS) is an immune-mediated inflammatory neuropathy of acute onset. It is classified by clinical and neurophysiologic findings into several subgroups, including acute inflammatory demyelinating polyneuropathy (AIDP), acute motor axonal neuropathy (AMAN), and Miller Fisher syndrome (MFS). NCS are typically normal in the first few days after symptom onset, with prolonged or absent F-waves the first abnormality noted. As early EDX findings may be minimal, the diagnosis is also based on clinical signs and symptoms, cerebrospinal fluid analysis, and spine MRI.[21] By 2 weeks, 50% of patients with AIDP have prolonged distal latencies, NCV <75% of normal values, and evidence of segmental demyelination, including conduction block and temporal dispersion. Of the patients, 85% have these abnormalities by 3 weeks.[22] Sural sparing (normal sural SNAPs with abnormal median and ulnar SNAPs) is an unusual pattern that is associated with GBS.[22] EDX findings often lag clinical symptoms and recovery.[22]
  - In axonal GBS, sensory and motor NCS show decreased amplitudes, normal or mildly prolonged latencies, and decreased NCV by no more than 75% of normal. It can be difficult to distinguish between distal conduction block and axonal neuropathy in early GBS and serial studies may be needed to determine the correct subtype.
  - Chronic inflammatory demyelinating polyneuropathy (CIDP) is rare in children. Different criteria for diagnosis have been published and are summarized in the references. EDX diagnostic criteria include conduction block or abnormal temporal dispersion in one or more motor nerves not at a site prone to compression.[2]
- *Toxins:* Most neuropathies caused by toxic exposure are axonal neuropathies with decreased SNAP and CMAP amplitudes, normal or mildly prolonged latencies, and decreased NCVs depending on the degree of axonal loss. EMG may be normal early in the process and develop changes consistent with denervation, motor unit remodeling, and decreased recruitment.
- *Chemotherapy:* Vincristine/vinca alkaloids are the most common cause of neuropathy in children with malignancies and cause a

sensory-motor axonal neuropathy. Symptoms usually improve when vincristine treatment is stopped, but some children have persistent significant weakness warranting evaluation for pre-existent hereditary neuropathy.[23]

- *Nutritional deficiencies:* These commonly include thiamine (B_1) deficiency, vitamin E deficiency, and pyridoxine deficit. Sensory involvement is common in all three, with decreased SNAP amplitudes. Axonal motor changes are also present with thiamine deficiency.
- *Mononeuritis multiplex:* This is characterized by sensory/motor abnormalities in two or more peripheral nerves occurring in a stepwise pattern over time. Hereditary neuropathy with liability to palsy (HNPP) is a type of mononeuritis multiplex with history of recurrent multiple mononeuropathies followed by complete recovery. EDX shows sensory-motor demyelinating polyneuropathy with conduction abnormalities at common entrapment sites. Atypical presentations occur with chronic focal neuropathies and polyneuropathy-like abnormalities.[24]

Mononeuropathies

- Mononeuropathies are the second most common reason for referral for pediatric EDX testing.[1] Trauma, with associated fractures or lacerations, is the most common etiology for mononeuropathies in children, with nerve compression the second most common etiology.[19]
- *Most commonly involved nerves:* In a study with 2,100 patients over an 11-year period, the peroneal and ulnar nerves accounted for 50% of diagnosed mononeuropathies, followed by the median (17%), facial (7%), sciatic (9%), radial (8%), tibial (4%), femoral (2%), and sural (2%) nerves.[1]
- Compressive median neuropathy (carpal tunnel syndrome) is relatively rare in children but occurs in the setting of mucopolysaccharidoses, congenital wrist bone abnormalities, HNPP, and other rare conditions.

Cranial Neuropathies

- *Facial palsy:* In facial palsy, EDX testing can be useful during the second to third week after onset of weakness (after Wallerian degeneration has occurred). Full recovery is predicted if there are normal CMAP latency and blink reflexes (BR) in the first weeks after onset. Facial nerve inexcitability, severe decreased CMAP, and absent BR indicate poor outcome. Loss of facial CMAP of <90% in the opposite side is usually followed by complete recovery in 7 to 8 weeks.[3]

Neuromuscular Junction Disorders

- Repetitive stimulation is performed along with NCS and EMG to localize dysfunction of the presynaptic or postsynaptic NMJ and can be helpful to diagnose myasthenia gravis (MG), botulism, and congenital myasthenia syndrome (CMS), in addition to other rare disorders.[3]

- SFEMG is the most sensitive test to diagnose NMJ defects, but it is difficult to perform in children. SSFEMG or SPACE is easier to use in children and the techniques are described in detail in the references.[2,3,25]

Juvenile MG

- *EDX:* Sensory and motor NCS are normal. EMG findings are usually normal, but in the presence of blocking, unstable MUAPs and/or small amplitude, short duration MUAPs (similar to those seen in myopathy) can be seen.
- *Low-rate RNS:* A decrement of >10% is considered abnormal.

Infantile Botulism

- The most sensitive and specific finding for botulism is an incremental response to rapid-rate stimulation (20 or 50 Hz), seen in 92% of patients. However, rapid RNS requires sedation.[3,19,26]
- Concentric needle EMG is also sensitive, but less specific. It is abnormal in many infants, with short-duration, low-amplitude MUAPs and abnormal spontaneous activity.[26]
- SSFEMG has high sensitivity but decreased specificity; jitter studies may be more sensitive than rapid RNS.[19]

Congenital Myasthenic Syndromes

- CMS has multiple different types, classified by presynaptic, postsynaptic, or both.
  - Presynaptic CMS (e.g., LambertEaton myasthenic syndrome [LEMS])
    - *NCS:* normal SNAPs, decreased CMAPs with postexercise facilitation of >25% from baseline; EMG may have small-amplitude/short-duration MUAPs without fibrillations; low-rate RNS: normal or decrement of >10%; rapid-rate RNS (10–50 Hz or brief voluntary exercise, if cooperative): facilitation >60% in LEMS
  - Postsynaptic CMS
    - *NCS:* normal sensory and motor studies; EMG may have small-amplitude/short-duration MUAPs without fibrillations; low-rate RNS with decrement of >10% in many subtypes

Myopathies

- EDX is ordered less frequently due to advances in genetic testing, muscle MRI, muscle ultrasound, and muscle biopsy, but can be helpful if differential diagnosis includes an NMJ condition or myotonic disorder.[27]
- Typical EDX findings include normal SNAPs; CMAPs with normal or decreased amplitudes; EMG with short-amplitude, short-duration MUAPs, with increased (early) recruitment pattern; and changes that may be subtle/partial with mixed motor unit potentials, making diagnosis difficult.[2]

- Typical protocol
  - *NCS:* One sensory and one motor NCS in the upper and lower extremities to rule out neuropathy and EMG of the distal and proximal musculature, paraspinals, and genioglossus are recommended, but often less muscles tested in practice due to patient tolerance. Evaluation of voluntary motor unit configuration and recruitment on needle EMG is best done in a nonsedated patient due to the need for voluntary muscle contractions.[3]
- *EMG sensitivity:* In one study, EMG was 95% sensitive for myopathy diagnosis and 67% specific in children <18 years old when compared with muscle biopsy.[28]
- *Congenital myopathies:* SNAPs are normal, with normal or mildly reduced CMAPs. EMG is often normal in infants <2 years old, with myopathic motor units for older children. In one study, EMG sensitivity for congenital myopathy in infants <2 years old was 36%, with a false negative rate of 25%.[29]
- *Juvenile dermatomyositis:* Small-amplitude, short-duration MUAPs with early recruitment pattern and prominent fibrillations are most prominent in the proximal muscles and paraspinals.[3] EMG is less frequently ordered with the new criteria and increased use of muscle MRI.[28]
- *Myotonic dystrophy:* NCS are usually normal with myopathic MUAPs and myotonia on EMG of the distal more than the proximal muscles. Clinical/electrical myotonia rarely presents at birth and may not develop until later in childhood.[3]

Nondystrophic Myotonic Conditions

- Myotonia congenita (Thomsen and Becker)
  - *NCS:* normal sensory and motor studies
  - EMG with myotonic discharges, normal MUAP configuration and recruitment
  - *RNS:* transient reduction in CMAP or after brief exercise[3]

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PART IV

Musculoskeletal Disorders

AL Grawany

Limb Deficiency

14

PHOEBE SCOTT-WYARD

GENERAL PRINCIPLES

Introduction

Limb deficiency in children occurs in a wide spectrum of presentations, causes, and outcomes. Limb deficiency can be a result of intrauterine or genetic factors (in the case of congenital limb deficiency), cancer, infection, or trauma (in acquired limb deficiency). Not all children benefit from prosthetic fitting, nor do those with multiple limb deficiencies always benefit from prosthetic fitting for all limbs. Each case should be treated individually. However, there are a few guiding principles that should be considered.

Epidemiology

- Across all etiologies, the prevalence of lower limb deficiency was found to be 38.5 cases per 100,000 commercially insured children in the United States, tracked over a 7-year study period. Congenital limb deficiencies accounted for 84% of cases, followed by 13.5% from trauma and .5% from cancer.[1]
- In a recent review of all pediatric traumatic amputations from 2007 to 2011,[2] the trends were consistent with prior studies:
  - 3:1 male to female ratio.
  - The most common amputations were of the fingers (54%) and toes (20%).
  - A caught-between mechanism (16.3%) was the most common, followed by machinery, powered lawnmowers, motor vehicle collisions, firearms, and off-road vehicles.
    - Lawnmower injuries were associated with lower limb amputations in children 5 years and younger.
    - Motor vehicle injuries were the most common cause of adolescent amputations.
    - Firearm-related amputations occurred predominantly in adolescents, whereas off-road vehicle-related amputations occurred in all ages.
- Congenital limb deficiencies[1,3–4]
  - Congenital limb deficiencies occur at a rate of 1 per 1,300 to 2,000 live births.

- Upper limb deficiency occurs at a rate of two to three times that of lower limb deficiency.
- Limb deficiency occurs with other major congenital anomalies in 12% to 33% of cases; therefore, it is important to consider common syndromes.

Classification

- For traumatic amputations, the nomenclature used for children is the same as adults, named for the joint or location of the body (e.g., transtibial, transfemoral).
- For congenital amputations, the limb is characterized as either transverse (absence of all elements distal to a specified level of the limb) or longitudinal (absence or hypoplasia of some elements along the length of a limb). Historical terminology should be avoided due to its confusing nature (e.g., hemimelia; Figure 14.1).

Etiology of Congenital Limb Deficiency

- Of more than 120 clinically defined congenital limb deficiencies, less than 40% have a known molecular origin.[1]

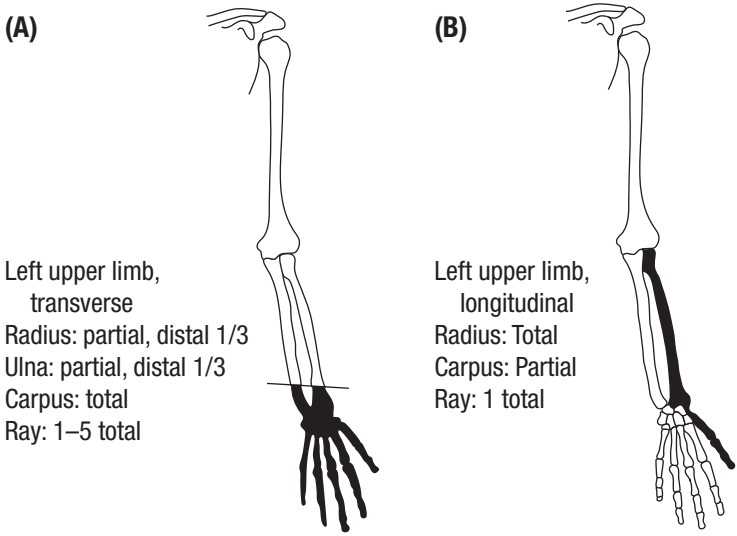

Figure 14.1 Examples using the standard nomenclature for limb deficiencies. (**A**) Transverse loss of the left distal forearm and hand. (**B**) Longitudinal left radial and thumb aplasia.

Source: Reproduced with permission from Wilcox WR, Coulter CP, Schmitz ML. Congenital limb deficiency disorders. *Clin Perinatol.* 2015;42(2):281–300.

- Medications that are known to affect limb development include thalidomide, retinoic acid, and misoprostol.
- Limb development occurs between 4 and 8 weeks of gestation.
- Causes of congenital limb deficiency include vascular disruption, vascular malformation, and genetic factors (spontaneous point mutations).
- Some risk factors for congenital limb deficiency include:
  - Maternal cigarette smoking, which increases the risk of longitudinal deficiencies[5]
  - Poorly controlled maternal diabetes, which is associated with longitudinal deficiencies as well as sacral agenesis with lower extremity hypoplasia[6]
  - Maternal thrombophilia[7]
  - Disruptions in the uterine environment, such as in the case of chorionic villus sampling and amniotic band syndrome[8,9]

DIAGNOSIS

Physical Examination[10]

- In vascular disruption, limbs typically have small, hypoplastic distal elements (e.g., "nubbin" residual fingers; Figure 14.2).
- In amniotic band syndrome, distal elements are typically fully formed with ligated or fused distal elements. Scars consistent with amniotic bands are often seen in multiple areas of the body. Children may benefit from plastic surgery evaluation if banding scars impede circulation.

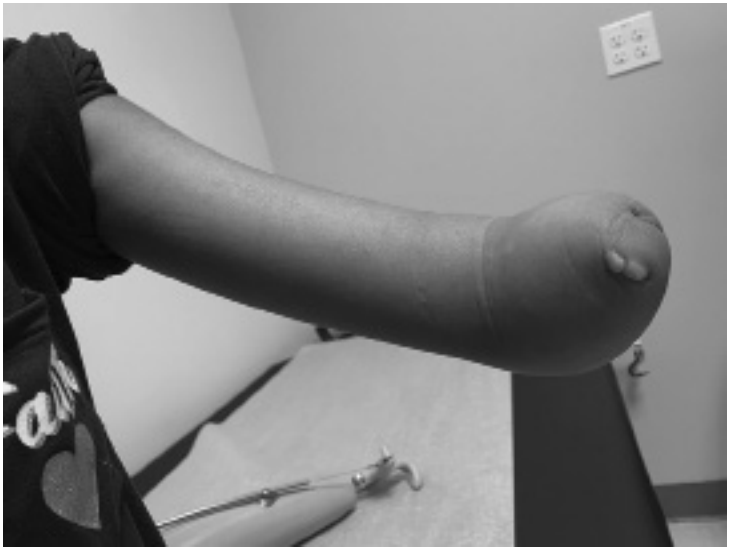

Figure 14.2 Congenital transradial limb deficiency with residual nubbins.

- Fibular deficiency[11]
  - Fibular deficiency is characterized by anterior bowing of the tibia with a dimple at the apex of the bow.
  - There is laxity of anterior cruciate ligament (ACL).
  - There is hypoplasia of the lateral femoral condyle with genu valgus.
  - There is hypoplasia or lack of lateral toe rays.
- Tibial deficiency[12,13]
  - Tibial deficiency may present with knee subluxation.
  - Tibial deficiency may present with hypoplasia or lack of medial toe rays with equinovarus foot.
  - It is important to evaluate for quadriceps function as this has implications for knee function and surgical planning.
  - Evaluate for other organ involvement, for example, cardiac, genitourinary, or gastrointestinal.
  - Evaluate other limbs for bony abnormalities, including the ulna and the femur.
  - Classification is based on the quantity and quality of the tibia present using the Jones criteria.
- Proximal femoral focal deficiency (PFFD)[14]
  - Affected hip is usually in external rotation and flexion, with proximal enlarged, foreshortened thigh (described as a "ships funnel" appearance).
  - On examination, the hip joint may have pistoning.
  - Evaluate all joints for integrity for future surgical planning.
- Radial longitudinal deficiency
  - Radial longitudinal deficiency often occurs with hypoplastic or absent thumb.
  - One-third of cases are associated with complex medical syndromes.
  - Recommend evaluation of hematologic, cardiac, and renal systems.[15]
- Upper limb deficiency
  - Unilateral pectoralis muscle or breast hypoplasia may be associated with Poland syndrome and therefore it is important to evaluate for dextrocardia.[16]
- Multiple limb deficiency
  - Evaluate function and use of all residual limbs, which is important to establish prior to any surgical planning.

Diagnostic Evaluation[3]

- *Transverse deficiency:* radiography of the affected limbs to determine the extent of bony abnormality and consideration of placental pathology if amniotic banding is present
- *Longitudinal deficiency:* more often syndromic and therefore necessitates a more thorough workup, including:
  - Radiography of the affected and contralateral limb
  - Skeletal survey (evaluate for skeletal dysplasia and/or spine abnormalities)

- Brain MRI if abnormal neurological examination
- Examination of the parents for limb abnormalities

- *Radial deficiency:* echocardiogram

Laboratory Studies[3]

- *Congenital longitudinal or multiple limb deficiency:* consider chromosomal microarray and genetic consultation; molecular testing of a single gene if phenotype is clear
- *Radial deficiency:* complete blood count and diepoxybutane (DEB) chromosome breakage assay

TREATMENT

Guiding Principles

Upper Limb

- There are five types of prostheses[17]:
  - Cosmetic or passive
  - Body-powered (uses a cable system)
  - Externally powered (uses myoelectric sensors or switches)
  - Hybrid (combining body and externally powered function)
  - Activity-specific (e.g., used for bike riding or swimming)
- *To fit or not to fit:* Not all children benefit from prosthetic fitting, particularly in unilateral upper limb deficiency. Families should be provided with education and access to fitting and be allowed to make an informed decision.
- *Start young/early:* In an international analysis of rejection, those fit prior to 2 years of age (congenital) or within 6 months of amputation (acquired) were 16 times more likely to continue prosthesis use.[18]
- Simple prosthetic components are more easily assimilated, for example, use of a hook terminal device is much easier for prosthetic training and function than a hand terminal device.[17]
- In order to foster prosthetic embodiment (integration of prosthesis into one's body image), it is important to establish a daily wear pattern.[19]
- Occupational therapy is imperative in successful prosthetic device use and should focus on control training, bimanual functional skill training, and integration into school activities and play.[20]
- *Residual digits or nubbins:* Removal is not recommended as this can promote neuroma formation and they are often functional (e.g., for touchscreen use).[21]
- Hand or orthopedic surgery consultation may be beneficial.
  - Toe-to-hand transfers or pollicization of the second digit can create prehensile function of the hand.
  - Bilateral upper limb deficiency[10]
    - Transradial amputation may benefit from Krukenberg procedure, separating the radius and the ulna to create a "lobster claw"-like functionality.

Figure 14.3 Example of bimanual use of bilateral transhumeral amputations for activities of daily living, such as brushing teeth or applying makeup. Photos courtesy of Bella Tucker.

  - *Transhumeral level:* If unable to touch in the midline, humeral lengthening for improved bimanual function may be beneficial (Figure 14.3).
- *Oral health:* As with all children, regular dental evaluations are important, even more in those who use teeth for prehensile function.
- *Length:* The ideal prosthetic length is "pocket length" (shoulder to pants pocket).[22]

Lower Limb

- Surgical considerations[23]
  - Preserve joints whenever possible to improve function and avoid bony overgrowth. Unlike that of adult amputees, pediatric patients' limb length can be managed later with guided growth or epiphysiodesis.
  - In children who require an ankle-level amputation, Boyd amputation (with calcaneal fusion to the tibia) is preferred over Syme (ankle disarticulation) as it allows for distal end-bearing both within and without the prosthesis.
  - *Van Nes rotationplasty:* This refers to the surgical removal of the knee with 180° rotation of the ankle and foot, creating a "neo-knee" (Figure 14.4), and is useful for:
    - Lower extremity intercalary traumatic amputation
    - Cancerous lesion of the distal femur or proximal tibia
    - PFFD with stable ankle
  - *Lengthening:* When lengthening, a stable proximal joint and >50% length of the contralateral side at baseline are needed (or <20 cm predicted limb length discrepancy at skeletal maturity).
  - Fibular deficiency[11]
    - Patients with a stable ankle joint and three or more toe rays may benefit from bone lengthening (following the guidelines above).
    - Patients with an unstable ankle joint with two or fewer toe rays are recommended to have foot ablation (Syme or Boyd).
    - Some children may require surgical correction of the tibial bow and genu valgus (if it progresses to be painful or causes patellar dislocation).
    - Studies suggest better patient satisfaction and fewer surgical complications with amputation when compared with limb reconstruction in fibular deficiency.[24]
  - *Tibial deficiency:* This is typically distinguished using the Jones criteria. Surgical intervention is typically based on the function and integrity of the knee and ankle joints (Figure 14.5).[10,12]
  - *Femoral deficiency or PFFD:* Treatment depends on femoral length (following the lengthening principles above) and stability of the joints. The Aitken classification system is typically used for the hip joint.[10,14]

Figure 14.4 Example of a prosthesis for a patient with rotationplasty. Note the thigh cuff needed for increased stability of the "neo-knee." Photo courtesy of David Rotter Prosthetics.

- *Stable hip joint:* Consider reconstruction procedures if appropriate (typically involves multiple procedures to lengthen the affected side and shorten the contralateral side).
- *Stable knee joint:* Consider foot ablation surgery and fitting as a below-knee amputee.

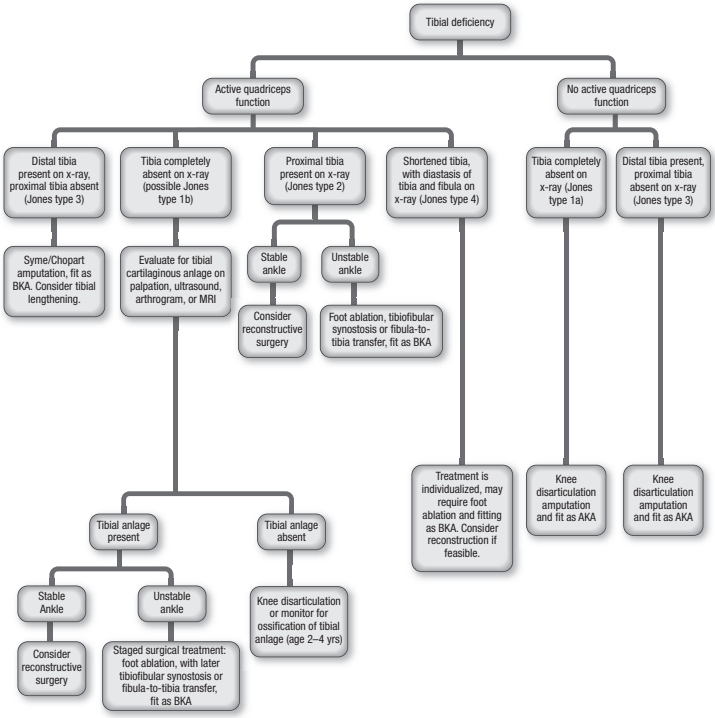

Figure 14.5 Surgical decision-making tree for patients with tibial deficiency.

AKA, above knee amputee; BKA, below knee amputee.

  - Unstable knee joint
    - Consider surgical knee fusion with foot ablation and fitting as an above knee amputee.
    - Van Nes rotationplasty is as above. This option requires an experienced surgeon, physical therapy, and prosthetic team.
- Fit when pulling to stand (typically 10–12 months old).
- In bilateral lower limb amputees:
  - Start with bilateral locking knee units and unlock one at a time.
  - Use arm span to estimate height.
  - Gait aid may be needed for balance.
- For congenital amputations, physical therapy is typically needed during the early years and when changing components. For acquired amputations, physical therapy can be helpful with initial prosthetic fitting.
- Many families are not ready for surgical intervention when the child is starting to be ambulatory. These patients can be fit with a "prosthosis"

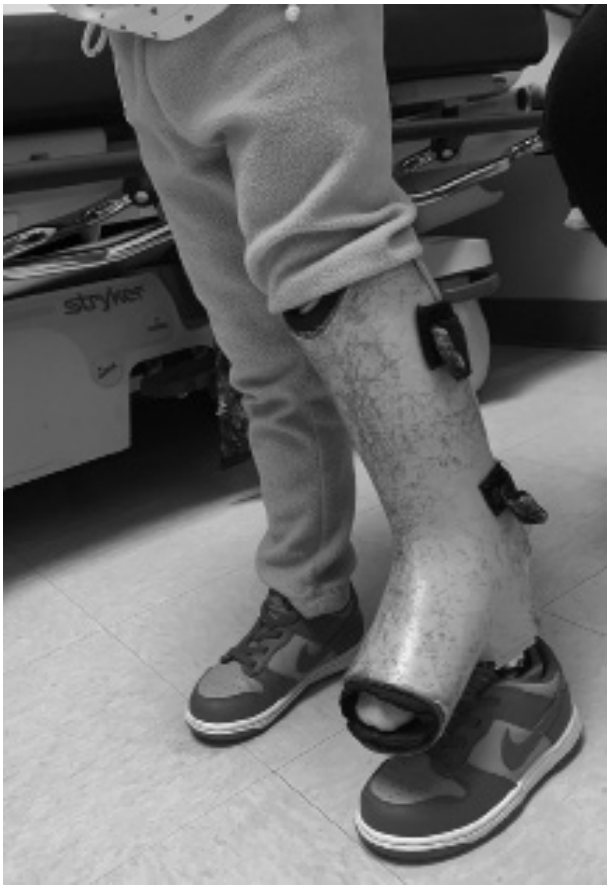

Figure 14.6 Example of "prosthosis" used for a patient with proximal femoral focal deficiency.

or extension prosthesis that includes the deformity to promote ambulation and independence with mobility[23] (Figure 14.6).

Multiple Limbs

- It is not always necessary or practical to fit all affected limbs.
- Occupational therapy evaluation can be extremely helpful as it is important to consider possible adaptive functional use of all limbs prior to any surgical intervention.
- Adaptive equipment is recommended, regardless of prosthetic fitting. Examples include wall-mounted sponge, dressing stick, universal cuff, bidet, Dycem, and adapted clothing.[17]

Initial Management

- Management includes consultation in combination with orthopedics if surgery is being considered.
- It also includes consultation with social work for parental/child emotional support, connection with support groups and social network, and counseling if needed.
- After surgical intervention, fit with stump shrinker socks once the incision is healed. They should be worn 23 hours per day for at least 2 weeks prior to prosthetic socket molding.
- *Durable medical equipment:* Consider prescribing wheelchair or crutches for children with lower limb deficiency for mobility in case of prosthetic complications.

Ongoing Care

- Children should be seen in follow-up every 6 months. Team-based care with occupational/physical therapy, prosthetist, social work, and rehabilitation physician is ideal, if available. They should see their prosthetist every 4 months for assessment of prosthetic fit, not just when having complications.
- Residual limb should be closely inspected at each visit for:
  - *Bony overgrowth:* A sharp, painful bony prominence at the end of the stump, with skin breakdown, requiring urgent surgical consultation and consideration of stump capping.
  - *Joint deformity:* Frequently, growth plates can be affected in children with limb deficiency, causing joint deformity over time (e.g., fibular deficiency and genu valgus).
  - *Pain in the limb:* Any patient with traumatic amputation is at risk of neuroma formation.
  - *Phantom pain:* While not as common in children, phantom pain can still be debilitating and all patients should be screened and treated.
  - *Skin breakdown:* For those who wear prosthesis, skin issues abound, such as fungal or bacterial infections, chronic choke syndrome, and contact dermatitis. Treatment should include suspending prosthetic wear until completely healed, evaluating prosthetic fit, and reviewing hygiene of the prostheticskin interface.
- Promote independence with care of prosthesis and regular skin checks.
- Monitor for signs and symptoms of overuse or detrimental compensatory adaptations, such as carpal tunnel syndrome and back pain.

Treatment Controversies

- Limb reconstruction versus amputation for treatment of affected limbs in osteosarcoma patients
  - Presence of an endoprosthesis after limb reconstruction precludes participation in high-impact activities or sports.[25]

- In multiple studies comparing the long-term outcomes of osteosarcoma survivors, there was no difference in functional or quality of life outcomes between patients who underwent amputation and those with limb reconstruction, although in one report 8% of survivors later underwent secondary amputation due to endoprosthesis failure.[26,27]
- A retrospective review of the National Cancer Database compared amputation and limb salvage in patients with osteosarcoma and found that more patients had undergone limb salvage in recent years, and amputations were associated with larger tumors, greater comorbidities, lower income, and advanced age and stage. Limb salvage was associated with improved survival, although these findings do not imply causation.[28]

- Limb lengthening/reconstruction versus amputation in congenital limb deficiency
  - In a retrospective study comparing amputation versus lengthening in patients with fibular deficiency, it was found that those who had undergone lengthening had more residual fibula and more toe rays, as well as more surgical interventions.[29] Numerous studies comparing outcomes across multiple quality of life and functional measures, including physical performance, found these treatments for fibular deficiency to be equivalent.[30,31]
  - In general, the aforementioned guidelines for reconstruction (following fibular length and number of toe rays present) are recommended for best clinical outcome.
- Hand transplantation
  - In 2015, the first successful pediatric bilateral hand transplantation occurred in a child with bilateral transradial amputations due to sepsis. After 18 months of intensive rehabilitation, the child was able to surpass previous upper extremity function. The child was a candidate due to a preexisting immunosuppressive regimen for renal transplantation.[32]
  - A decision model analysis of prosthetic device fitting versus bilateral hand transplantation in a child showed only marginal benefit to transplantation, with a risk of losing life years due to immunosuppressive complications.[33]
- Timing of articulating knee in prosthetic prescriptions
  - Historically, children with above-knee amputations were not fit with articulating prosthetic knees until after the age of 5 years. However, more recent evidence has shown that for children with knee disarticulation or above-knee amputations, early use of a knee unit in the prosthesis encourages more normal gait patterns.[34]
- Fitting with myoelectric prosthesis in pediatric upper limb deficiency
  - While not common in the United States, children have been successfully fit with myoelectric prostheses as young as 2 years of age in Scandinavian countries and 6 months old in Canada.[35]
  - Some studies have suggested that earlier fitting (prior to 2 years of age) with myoelectric prostheses can improve acceptance and continuation of use.[36]

- In a retrospective study of patients fitted with multiple prostheses (body-powered, myoelectric, and cosmetic), only 15% of those fitted with myoelectric prostheses continued use long term. It was concluded that the function of the prosthesis frequently dictated acceptance, and sometimes the simplest design was the most functional.[37]

- Osseointegration
  - At the time of this writing, there have been no pediatric patients who had undergone surgical implantation of osseointegrated prostheses. This field offers exciting advancements in the prosthetic–limb interface with improved proprioception, but carries risks such as infection and soft tissue stoma irritation.[38]
- Three-dimensional (3D) printing
  - The ability to customize and the inexpensive nature of 3D printing have caused an explosion of its use in pediatric upper limb prostheses. However, there is lack of randomized controlled clinical trials assessing the durability, functionality, and long-term effects on quality of life of children who have been provided with these devices.[39]

ADDITIONAL CONSIDERATIONS

- Social stigma/bullying
  - Children with physical disabilities, including limb deficiency, are at high risk of bullying and should be screened at each visit.[40]
- Adaptive sports
  - People with disabilities can benefit in numerous ways from participation in adaptive sports, such as improvement in quality of life, mood, life satisfaction, community participation, and employment; however, a multitude of barriers exist, both environmental and attitudinal. The pediatric physiatrist should actively educate and encourage patients to participate in adaptive sports.[41,42]
- Targeted muscle reinnervation (TMR)
  - TMR is a surgical technique utilized for more intuitive control of a prosthesis, wherein multiple peripheral nerve transfers to a target muscle allow for increased EMG control signals for a multifunctional myoelectric prosthesis. It is most suitable for upper extremity amputees with significantly impaired function who have previously been fitted with a myoelectric prosthesis.[43]
  - When compared with neurectomy, TMR has also proven to both treat chronic limb and phantom pain in amputees and prevent its occurrence when done at the time of major limb amputation.[44,45]
- Crossover prosthetic feet
  - A new hybrid style of prosthetic foot, described as a "crossover foot," is an energy-storing foot that is designed to be an everyday walking foot with stability and energy return to allow users to run comfortably. Given the lack of insurance coverage for running-specific prosthetic feet, this option may improve access to energy-storing feet for children and allow them to participate in sports.[10]

- There is some evidence that this style of feet reduces energy expenditure in walking compared with running blades, making it a better all-around foot for those who participate in higher level activity.[46]

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Rheumatologic Disorders

15

TRACEY B. WRIGHT

GENERAL PRINCIPLES

Rheumatic diseases are characterized by chronic inflammation frequently in the joints but also in other tissues and organs, with periods of disease activity and remission. When considering an inflammatory cause of joint symptoms, key elements of the history provide important clues to inform the diagnosis.

- Inflammatory joint pain is worse in the morning or after prolonged inactivity.
- Stiffness, described as slow movements of the involved joint, is worse in the morning or after prolonged inactivity.
- A joint effusion noted on examination may be associated with pain and limitation, but erythema is not expected.

While inflammatory arthritis is the primary manifestation of juvenile idiopathic arthritis (JIA), other rheumatic diseases involve multiple organs beyond the joints, and thus a detailed, comprehensive history and physical examination are critical.

Without appropriate treatment, chronic inflammation of rheumatic disease may negatively affect growth and development. With early diagnosis and improved treatment, children with rheumatic diseases are more likely to survive into adulthood and thus attention to the long-term impact of chronic disease and its therapies is essential.

- The broad goals of treatment are to control inflammation, prevent treatment complications, lessen the burden of morbidity from the consequences of chronic diseases, and improve quality of life.

JUVENILE IDIOPATHIC ARTHRITIS

JIA is a heterogeneous group of diseases with the hallmark symptoms of persistent arthritis in a joint or set of joints lasting for at least 6 weeks and with onset before 16 years of age.[1] The subtypes of JIA are currently defined according to the International League of Associations for Rheumatology (ILAR) classification criteria, which is based on a naming system rather than the underlying biology.[1] The epidemiology of JIA varies by gender, race/ethnicity, geographic location, and subtype. In the United

States, there is an estimated incidence rate of 11.9 per 100,000 person-years and a prevalence rate of 44.7 per 100,000 persons.[2]

- Because JIA is a diagnosis of exclusion, other causes of arthritis should be considered, including infection, malignancy, trauma, and other connective tissue diseases.
  - Severe pain or joint erythema is uncommon in JIA and should raise concern for septic arthritis, postinfectious arthritis, or malignancy.
- Each subtype of JIA has a variable presentation, with a unique pattern of arthritis, associated symptoms, trajectory of disease, and scope of outcomes and complications.[3]
- A detailed history should include an assessment of joint pain, swelling, morning stiffness, fever, rashes, and change in activity level.
- JIA-associated uveitis, the most common extraarticular manifestation for most subtypes, is characterized by chronic inflammation affecting the anterior chamber. Uveitis is asymptomatic at presentation in all JIA subtypes, except for enthesitis-related arthritis (ERA), where patients have acute iritis.

Although the exact cause is unknown, these diseases likely occur in a genetically susceptible host and are brought on by an environmental trigger. Pathogenesis is multifactorial and relates to loss of immunologic self-tolerance.

- Abnormal T cell responses help activate autoreactive B cells and increase the production of proinflammatory cytokines,[4] including interleukin-6 and interleukin-17, which may vary among the JIA subtypes.[5]
- Anticitrullinated protein antibodies (ACPA), occurring in 60% of patients with rheumatoid factor (RF)-positive polyarticular JIA, are present prior to overt clinical manifestations, contribute to immune complex formation and complement activation,[6] and are associated with more severe, erosive disease.

Oligoarticular Juvenile Idiopathic Arthritis

Oligoarticular JIA is the predominant subtype occurring in 50% to 80% of children with JIA, affecting young children between the ages of 1 and 3, and is more common in girls.[7]

- This is subdivided into persistent, with no more than four affected joints throughout the disease course; and extended, with more than four affected joints after the initial 6 months of disease.[1]
- Arthritis is most common in the lower extremities, frequently manifesting as a monoarticular knee or ankle arthritis.[8]
- Leg length discrepancy, an example of a localized growth disturbance, muscle atrophy, and joint contracture result from long-standing arthritis.
  - Increased blood supply to the swollen joint brings growth factors and proinflammatory cytokines directly affecting the growth plate, resulting in increased length on the affected side.[9]

- Antinuclear antibody (ANA) is frequently positive, especially in girls, and is associated with increased risk of uveitis.[10]
- Aspirated synovial fluid shows moderate inflammation with 2,000 to 50,000 cells/mm^3.[11]
- Intraarticular glucocorticoids for monoarticular arthritis are the first-line treatment[12] and are required when joint contractures are present.
- While nonsteroidal anti-inflammatory drugs are commonly prescribed, inadequate response to joint injection requires escalation of therapy to disease-modifying antirheumatic drugs (DMARDs) or biologics.
- Physical therapy is indicated for joint contractures to preserve joint range of motion.
- Risk factors associated with poor prognosis include arthritis of the hip, cervical spine, wrist, or ankle, elevated inflammatory markers, joint damage detected on radiographs, and the extended oligoarticular JIA phenotype.[13]

Polyarticular Juvenile Idiopathic Arthritis

Polyarticular JIA, comprising 15% to 20% of all JIAs, is further subdivided into RF-negative and RF-positive subtypes based on the presence or absence of this antibody.[14] Both subtypes are more common in girls.

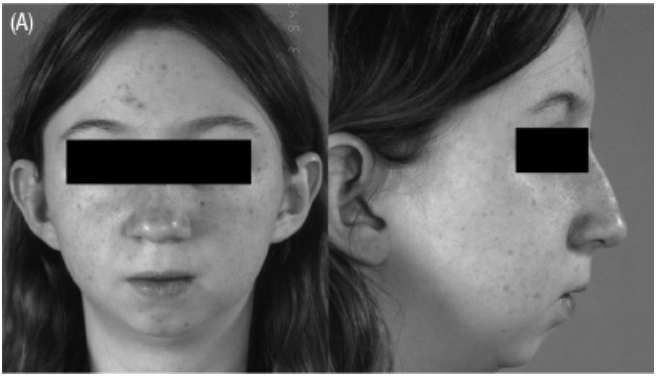
RF-negative polyarticular JIA has a bimodal distribution in younger children between 1 and 3 years of age and older children between 9 and 14 years of age.[15]

- Arthritis is asymmetric, affecting the knees, ankles, wrists, and small joints of the hands and feet.
- Temporomandibular joint (TMJ) arthritis is common and is highly susceptible to growth disturbances and severe damage.[16]
  - Localized growth disturbance of the TMJ results in facial asymmetry, limited mouth opening, and micrognathia[17] (Figure 15.1A). TMJ replacement is indicated for severe damage.
- Arthritis of the hip and cervical spine, considerable number of affected joints, and elevated inflammatory markers warrant early, more aggressive treatment.[13]

RF-positive polyarticular JIA is like rheumatoid arthritis in adults occurring in older children starting between 10 and 13 years of age.[15]

- Arthritis is symmetric, involving the wrists and the small joints of the hands and feet.
- Joint deformities include boutonniere (Figure 15.1B) and swan-neck deformities of the fingers and ulnar drift or subluxation of the wrist.[18]
- Fatigue and weight loss are common.
- Subcutaneous nodules (Figure 15.1C) on the bony prominences are the most frequent extraarticular manifestation.
- The presence of ACPAs denotes more aggressive disease and risk of joint damage.[19]

- This subtype carries the greatest risk of developing cartilage loss and bony erosions[20] and atlantoaxial subluxation of the cervical spine.[21]
- Early and aggressive treatment increases the potential for durable clinically inactive disease,[22] but the risk of future disability remains.[23]
- DMARDs, such as subcutaneous methotrexate, are the initial treatment.
- Persistent arthritis warrants escalation to a biologic, typically with tumor necrosis factor inhibitors.[24]



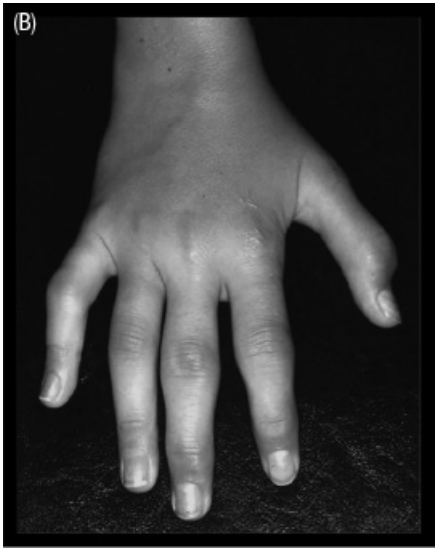
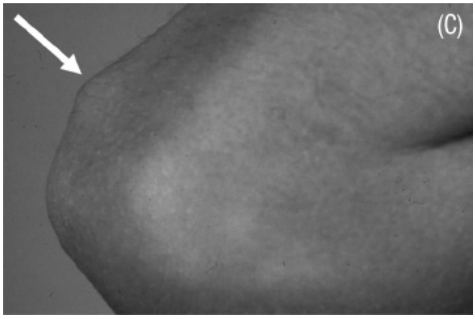


Figure 15.1 (**A**) Micrognathia from temporomandibular joint arthritis. (**B**) Arthritis of the interphalangeal and second and third proximal interphalangeal joints, and boutonniere deformity of the fifth digit. (**C**) Subcutaneous nodule (arrow). (**D**) Dactylitis. (*Continued on the next page.*)



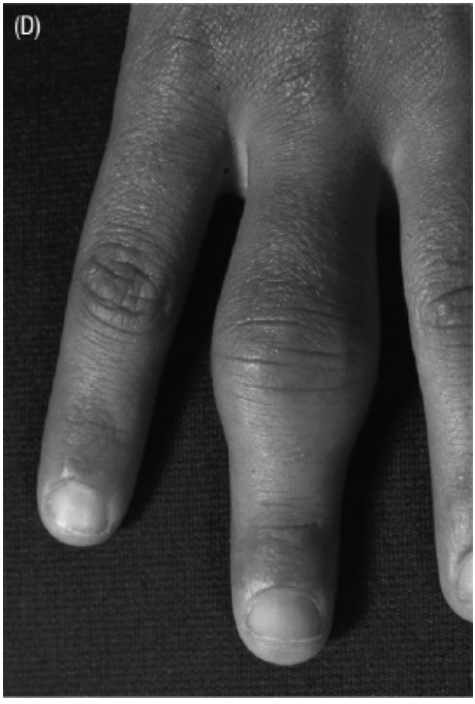


Figure 15.1 (*continued*)

- Chronic oral glucocorticoids are not recommended due to adverse effects.[24]
- Adjunctive physical and occupational therapy can address functional limitations.[24] Weight-bearing exercise may improve endurance and decrease joint symptoms.[25]
- Severe joint damage may result in joint replacement.[23]

Enthesitis-Related Arthritis

ERA is diagnosed in middle childhood and is more common in boys.

- Lower extremity arthritis is the predominant phenotype.
- Enthesitis, inflammation at the site of the bony attachments of the tendons, ligaments, and fascia, is a key differentiating feature.[26]
- There are two patterns of arthritis: peripheral versus axial.
  - Peripheral arthritis
    - Peripheral arthritis is asymmetric, affecting the large lower extremity joints.
    - It affects the small joints of the feet and toes.
    - Tarsitis, or inflammation of the midfoot, is a distinct manifestation.[27]
  - Axial arthritis
    - Arthritis affecting the sacroiliac joints or spine is less common at disease onset.[28]
    - Inflammatory back or buttock pain, worse in the morning or after prolonged sitting, is a typical symptom.
- Direct palpation over the sacroiliac joints is a less sensitive and specific method to detect sacroiliitis and thus MRI is preferred.[29]
- Acute, anterior uveitis characterized by a red, painful eye sensitive to light is unique.
- Weight loss, crampy abdominal pain, diarrhea, or bloody stool should prompt evaluation for inflammatory bowel disease.
- Human leukocyte antigen B27, considered a genetic risk factor for the disease, is present in 60%.[28]
- Targeted biologic therapy has demonstrated greater clinical improvement compared with DMARDs[30] and is recommended for axial disease.[24]
- Risk factors for a poor prognosis include ankle and hip arthritis in the first 6 months and sacroiliitis.[31]

Psoriatic Arthritis

- Psoriatic arthritis may have variable presentation, with 50% of children developing arthritis before the onset of psoriasis.[32] Age of presentation includes young children between 2 and 3 years of age and those in middle to late childhood.
- Oligoarthritis, frequently involving the knee, ankle, hip, wrist, and fingers, occurs at presentation, while polyarthritis develops without treatment.
- Older children may develop sacroiliitis or enthesitis.[33]
- Dactylitis (Figure 15.1D), also known as a sausage digit, manifests as a more diffuse swelling of the joint or may be fusiform swelling localized around the proximal interphalangeal joint.
  - Dactylitis tends to occur in one or two digits and may not always be painful.[34]

- Arthritis of the distal interphalangeal joints and flexor or extensor tenosynovitis, defined as inflammation of the tendon sheaths, are more frequent.
- Psoriasis may occur in 40% to 60% of children with psoriatic arthritis, and associated nail changes may occur in 40% to 80% of psoriatic arthritis.[35]

Systemic Juvenile Idiopathic Arthritis

Systemic JIA is quite distinct from other JIA subtypes and may be more appropriately classified as an autoinflammatory disease.

- Fever and rash are predominant and arthritis is not always present at diagnosis.[36]
- A persistent chronic polyarthritis may develop even after systemic symptoms resolve.
- Laboratory studies demonstrate marked systemic inflammation.
- Most have prompt response to targeted biologic therapy.

Imaging is critically important for evaluation and management of inflammatory arthritis.

- Radiographs are of limited value in defining the extent of arthritis in a growing child who still has a significant amount of cartilage overlying the joints.
  - Early changes include soft tissue swelling, osteopenia, and effusions.
  - Joint space narrowing and bony erosions tend to occur later, except in patients with more aggressive arthritis.[37]
  - Ankylosis may occur when joint space narrowing affects the spine.
- Musculoskeletal ultrasound is increasingly used to evaluate disease activity and response to treatment. It may detect subclinical synovitis[38] and erosions.[39]
- MRI is highly effective in the evaluation of bone marrow edema and structural damage including bony erosions. It is preferred to detect early inflammatory changes of axial disease in ERA.[40]

Early, aggressive treatment is essential as many children present at an early age, and chronic arthritis has significant effects on overall growth and development and the health of individual joints.

- Systemic glucocorticoids should only be considered for short-term treatment of severe cases.
- With biologics, there is increased likelihood of achieving inactive disease and low disease activity states and decrease in disease damage indices.[41]
- The American College of Rheumatology (ACR) guidelines and the Childhood Arthritis and Rheumatology Research Alliance (CARRA) consensus treatment plans are beneficial to inform therapy decisions.[24,42,43]
- Physical and occupational therapy improves function and prevents loss of range of motion in an affected joint.

SYSTEMIC LUPUS ERYTHEMATOSUS

Systemic lupus erythematosus (SLE) is a multisystemic inflammatory disease characterized by autoantibody production. Of the cases, 20% are diagnosed before the age of 16. The estimated incidence is .3 to .9 per 100,000 children-years[44] and is more common in females. Underrepresented racial/ethnic minorities are disproportionately affected with more severe disease. The underlying pathophysiology is extraordinarily complex and involves loss of tolerance to self-antigens in a genetically susceptible host.

- Dysregulation of type I interferons, a proinflammatory cytokine, and an inability to appropriately clear intracellular materials from apoptosis are key abnormalities in the innate immune system.[45]
- Abnormalities in B and T lymphocytes result in autoantibody production, causing direct cellular injury or facilitating immune complex formation, ultimately resulting in more tissue damage.[46]

Children commonly manifest major organ involvement and more frequently accrue damage than adults with SLE.[47]

- Mucocutaneous manifestations, including malar rash and oral ulcers, are common.[48]
- Arthritis affects the small joints, although rarely develop to chronic arthritis as seen in JIA.[49]
- Lupus nephritis is prevalent, occurring in 40% to 70% of pediatric SLE, and is a common cause of morbidity and mortality,[50] with varied presentations including hypertension, nephrotic syndrome, and abnormal urinary sediment.
- The spectrum of neuropsychiatric disease is diverse, including stroke, cerebritis, psychosis, and neuropathy, frequently occurring at presentation, although it may manifest several years after diagnosis.[51]
- Cognitive dysfunction may be a primary neuropsychiatric manifestation but may also result from chronic steroid exposure.[52]

Initial laboratory studies should include complete blood count to evaluate for cytopenias, erythrocyte sedimentation rate to assess the degree of systemic inflammation, and creatinine, albumin, and urinalysis to assess for nephritis.

Detection of autoantibodies is a hallmark of SLE. While the ANA is frequently positive, it is nonspecific, whereas double-stranded DNA and Smith antibodies are more specific.

With few drugs for SLE approved by the Food and Drug Administration, many therapeutic options are used off-label.

- Most are prescribed hydroxychloroquine, an antimalarial drug with immunomodulatory effects.
- Glucocorticoids effectively treat inflammation but their use warrants calcium and vitamin D supplementation to promote bone health.
- Other immunomodulatory treatments are chosen according to clinical manifestations.
- Photoprotection, including consistent use of sunscreen, is critical due to risk of cutaneous and other disease flare with UV light exposure.

Because survival has improved, with an estimated 10-year survival rate of at least 90%, attention to other aspects of management is critically important to reduce long-term morbidity.[53]

- Monitoring bone health is essential as patients who develop SLE in childhood have a much greater risk of steroid-related damage (e.g., osteoporosis with fracture, avascular necrosis, cataracts, and diabetes) compared with adult-onset disease, notably when there is longer disease duration.[54]

JUVENILE DERMATOMYOSITIS

Juvenile dermatomyositis (JDM) is the most common idiopathic inflammatory myopathy in childhood,[55] with an estimated annual incidence of 1.9 to 3.2 cases per million[56,57] and a mean age of onset of 7 years.[58]

- Environmental factors, including sun exposure and infection, trigger immune dysregulation of both the innate and adaptive immune system in genetically susceptible individuals.[59,60]
- Current diagnostic criteria include (a) muscle weakness, (b) muscle biopsy findings, (c) elevated skeletal muscle enzymes, (d) electromyography (EMG) changes, and (e) dermatologic features.[61]
- Proximal, symmetric muscle weakness is the hallmark, most notably in the lower extremities, followed by the upper extremities, although the neck and trunk muscles may be affected.[59]
  - A positive Gowers's sign or head lag may be noted on examination.
  - Muscles may be tender and edematous.
  - Patients have progressive difficulty with basic activities of daily living and inability to exercise.[59]
  - Oropharyngeal muscle weakness results in dysphagia and risk of aspiration.[62]
  - Shortness of breath warrants evaluation for respiratory failure.
  - Joint contractures may develop from muscle inflammation and arthritis may occur.[63]
  - Objective characterization of muscle strength can be tracked over time using the Childhood Myositis Assessment Scale (CMAS) and the manual muscle testing of eight muscle groups (MMT8).[64,65]
- Prototypical rashes include heliotrope rash, facial erythema, and Gottron papules (Figure 15.2A and 15.2B).
- Abnormal nailfold capillaries (Figure 15.2C), visualized proximal to the nail, are associated with disease activity and appear as irregular or enlarged vessels or areas of capillary dropout.[66]
- Calcinosis (Figure 15.2D) refers to deposits of dystrophic calcium in the skin and soft tissue and is associated with delayed diagnosis and treatment, more severe disease, and prolonged disease duration.[67]
- Elevated muscle enzymes (creatinine kinase [CK], lactate dehydrogenase [LDH], aspartate aminotransferase [AST], alanine aminotransferase [ALT], and aldolase) occur in the majority, although the extent and timing of elevation are variable.[58]

- Myositis-specific antibodies are associated with specific clinical phenotypes,[68] risk of disease complications,[69] and histology patterns on muscle biopsy.[70]
- Although EMG changes are part of the diagnostic criteria, this procedure is not used in clinical practice due to the associated pain.
- MRI using fat-suppressed T2-weighted images and short-tau inversion recovery (STIR) sequences detects muscle edema and can clarify the best site for muscle biopsy.[70]
- Radiographs are suitable to determine the extent of calcinosis.
- Muscle biopsy has characteristic findings including perifascicular atrophy, perivascular inflammation, muscle fiber degeneration/regeneration, and tubuloreticular inclusion bodies.[59]
- Glucocorticoids are a mainstay of treatment, while immunosuppressants treat inflammation and help minimize long-term exposure to glucocorticoids.

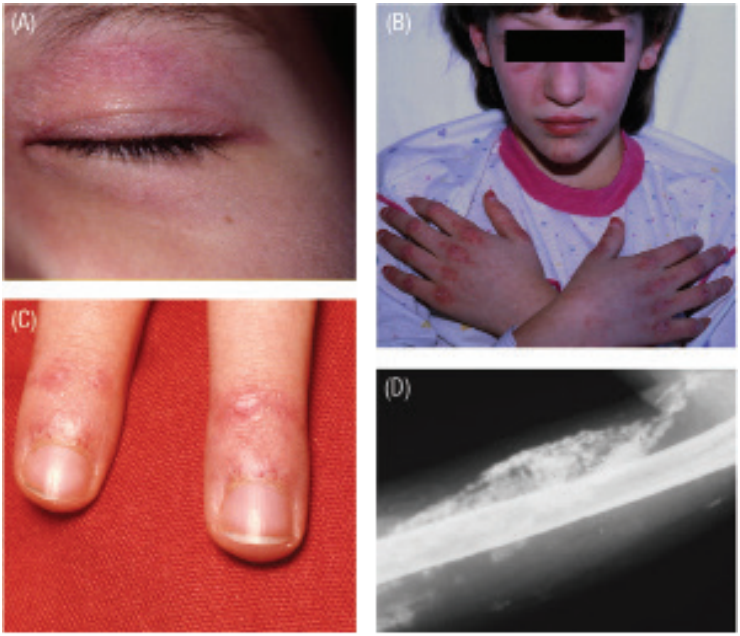


Figure 15.2 (**A**) Heliotrope rash: purple discoloration above the eyelids sometimes associated with periorbital edema. (**B**) Facial erythema may occur in a malar distribution. Gottron papules: erythematous papules and patches overlying the finger joints and other extensor surfaces, including the elbows and knees. (**C**) Nailfold capillaropathy. (**D**) Calcinosis as seen on x-ray.

- The CARRA published a consensus-based recommendation to guide JDM management.[72,73]
- Photoprotection, including consistent use of sunscreen, is a critical component of therapy due to risk of disease flare with UV light exposure.
- Implementation of a home exercise program improves muscle function and functional ability,[74] but the application of structured exercise as therapy is variable.
- The development of calcinosis should prioritize altering treatment to improve disease control.
  - Consider surgical removal for significant lesions, although there is risk of reaccumulation.[75]
- Disease course can be monocyclic, polycyclic, or persistent/recalcitrant.[76]
- While the mortality rate has significantly improved to less than 5%,[77] death may occur from gastrointestinal vasculitis, with severe bleeding or intestinal perforation, or progressive cardiopulmonary disease.[78]
- Active disease may persist, resulting in long-term damage including cutaneous scar, muscle atrophy and dysfunction, lipodystrophy, and joint contractures.[77]

JUVENILE SCLERODERMA

Juvenile scleroderma is a rare inflammatory connective tissue disease with two forms: systemic and localized. Juvenile systemic sclerosis (jSS) involves multiple organs, whereas juvenile localized scleroderma (jLS) is generally limited to the skin and subcutaneous tissues.

Juvenile Systemic Sclerosis

jSS comprises only approximately 10% of scleroderma cases overall and accordingly is very rare, with an estimated prevalence of three per million children in the United States.[79] The average age of onset is between 8 and 10 years and is more common in girls.[80]

- Inflammatory changes define the early stages, while the later stages are characterized by progressive deposition of fibrous collagen in the skin and end organs.[81–83]
- Raynaud phenomenon, a key diagnostic feature, especially in a younger child, reflects underlying vasculopathy.
  - Triphasic color changes (purple, pallor, erythema) occur in the digits, frequently triggered by cold exposure or other factors.
  - Persistent ischemia, ulcers, and ultimately necrosis may develop.[84]
- Skin involvement is a predominant feature that evolves over time with generalized edema, then sclerosis, followed by atrophy.[84]
  - Skin changes in the hands result in tapering fingers, known as sclerodactyly.[84]
  - Fingers lose extension and flexion as tendons become shorter, leading to significant hand deformities with resulting dysfunction.

- A small mouth opening reflects facial changes.[84]

- Interstitial lung disease (ILD) and pulmonary artery hypertension are the most common cardiopulmonary manifestations.
  - The inflammatory phase of ILD may evolve to progressive pulmonary fibrosis, resulting in severe restrictive lung disease.
- Tenosynovitis, myopathy, and myositis occur, whereas joint contractures with limitations in range of motion result from cutaneous changes.
- Baseline and regular screening for cardiopulmonary disease with EKG, echocardiogram, pulmonary function test, and chest imaging are essential.

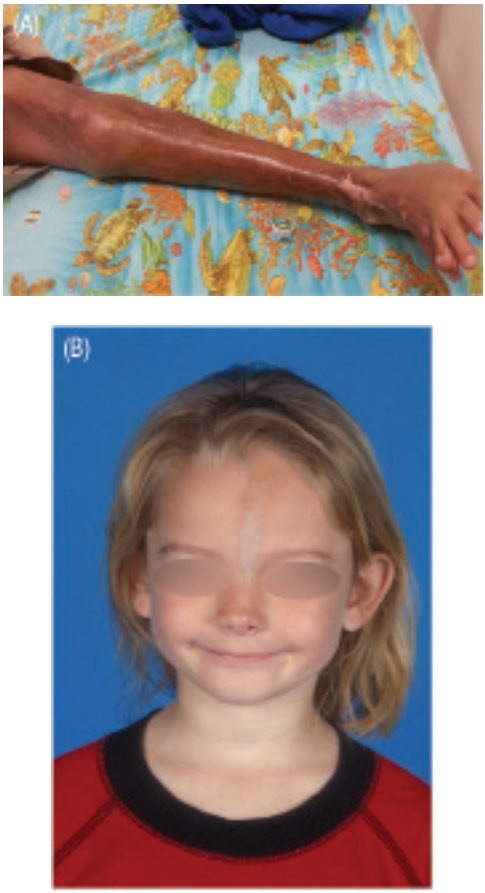


Figure 15.3 (**A**) Linear scleroderma. (**B**) En coup de sabre.

Treatment recommendations, derived from adult consensus recommendations, are tailored according to the organs impacted by the disease.[85]

- The primary approach to treating Raynaud phenomenon is to avoid triggers, most commonly cold exposure, by using warming techniques.
  - Calcium channel blockers promote vasodilation and are an effective treatment for digital ulcers, with prostanoids used for more severe disease.[85]
  - Digital botox injections and digital sympathectomy treat severe cases with digital ulcers when there is concern for progression to digital ischemia.[86,87]
- Nonbiologic disease-modifying therapy, mycophenolate mofetil, and cyclophosphamide treat ILD.
- Severe cardiac disease and respiratory failure are the most common causes of death.[80]

Juvenile Localized Scleroderma

jLS is the form of scleroderma that is 6 to 10 times more common in children.[88] Localized scleroderma is rare, with an incidence of .34 to 2.7 cases per 100,000 per year,[89,90] and is more common in females, with a mean age of disease onset of 8.3 years.[89] The mechanism of evolution of inflammatory to fibrotic skin changes in jLS is similar to what occurs in jSS.[89,91]

- The five subtypes are based on the descriptions and location of the skin lesions: circumscribed morphea, linear scleroderma, generalized morphea, pan sclerotic morphea, and mixed subtype.[92]
- Linear scleroderma (Figure 15.3A) is the most common, manifesting as a "longitudinal band lesion" on the extremities and face.[93]
- When a localized scleroderma lesion crosses a joint, there is increased risk of developing joint contractures.
- En coup de sabre (Figure 15.3B) refers to a linear lesion of the face and scalp, while ParryRomberg syndrome is a form of linear scleroderma manifested by facial hemiatrophy.
- Active lesions are erythematous with violaceous color change, warm, and abnormal skin texture (smooth, shiny, and/or waxy appearance), whereas chronic changes include postinflammatory pigmentation, dermal and subcutaneous atrophy, and progressive skin thickening.[94]
- Joint contractures and arthritis are examples of extracutaneous manifestations.[95]
- Extensive lesions on an extremity may result in significant atrophy and marked leg length discrepancy (Figure 15.2D).
- MRI can define the extent of involvement of deeper tissues beyond the skin, including the subcutaneous tissues and fascia.[96]
- The general principle of treatment is to reduce tissue inflammation in an attempt to prevent damage.
  - While topical therapies are available for mild, isolated lesions, methotrexate is frequently prescribed as first-line treatment.

- While many have good response to methotrexate and glucocorticoids mycophenolate mofetil and biologic therapy are used if response is inadequate.[97,98]
- Botox injections can treat abnormal movements and muscle spasms.[99]
- When joint contractures occur, physical therapy is critical and surgical intervention may be needed in those with severe contractures.[100]
- Musculoskeletal disease is a major cause of disability due to impaired function from muscle atrophy, joint contracture, and extremity shortening.[95,101]

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Pediatric Sports Medicine and Adaptive Sports

16

STEPHANIE TOW, AMY E. RABATIN, and MARY DUBON

SPORTS PARTICIPATION AND PHYSICAL ACTIVITY

- The U.S. Department of Health and Human Services "Physical Activity Guidelines for Americans" recommends the following:
  - Preschool-age children (3–5 years old): physical activity daily
  - Children and adolescents (6–17 years old): moderate- to vigorous-intensity physical activity at least 1 hour daily
    - *Aerobic exercises* that raise heart rate (e.g., swimming, running, jumping)
      - At least 3 days per week
    - *Muscle-strengthening exercises* through strengthening/resistance exercises of the muscles (e.g., push-ups, tug-of-war, resistance bands, weights)
      - At least 3 days per week
    - *Bone-strengthening exercises* through weight-bearing exercises (e.g., running, jumping)
      - At least 3 days per week[1]
- Sports participation for individuals without disabilities
  - *Prepandemic:* While sports participation is common among children and adolescents in the United States, sports participation decreases from elementary school to middle school years.[2,3]
- Sports participation for individuals with disabilities
  - *Prepandemic:* Sports participation is less for children and adolescents with disabilities compared with children and adolescents without disabilities.
    - Sports participation is lower in children with functional and mobility impairments compared with children with sensory and cognitive impairments.
    - Sports participation is lower in adolescents with mobility impairments compared with those with functional, cognitive, or intellectual impairments.
    - Lower likelihood of participating in physical activity correlates with lower sports participation in children and adolescents with disabilities.[4]

- Current terminology and definitions in adaptive and para sports
  - While adaptive/adapted sports and para sports are terms often used interchangeably, the following are the official definitions:
    - *Adaptive/adapted sports:* sports modified from mainstream sports (e.g., wheelchair basketball, which is adapted from basketball)
    - *Para sports:* umbrella term for sports of individuals with impairment, whether or not an equivalent exists in individuals without impairment (e.g., goalball is a para sport but not an adaptive sport as it is not modified from a mainstream sport)[5]
      - *"Para":* refers to being in parallel to sports for individuals without impairments[5,6]
      - Often refers to types of sports in the Paralympics[6]
- Paralympics
  - These are elite games/movements for athletes with physical disabilities, blindness/low vision, or intellectual disabilities that are in parallel to the Olympics.[6]
    - General eligible impairment categories are physical, visual, and intellectual.[7]
  - The Team USA Paralympic Sport Development is a program that introduces individuals with disabilities to Paralympic-style sports as a pathway to learn these sports and possibly train for elite Paralympic Games.[8]
- The classification process is a sport-specific process that aims to level the playing field so that athletes are judged in competition by their athletic ability and not by their impairment/disability.[9]
- Special Olympics[10]
  - Special Olympics are sports training and competitions for athletes with intellectual disabilities aged 8 and older, from novice to elite.[10,11]
  - Special Olympics are also programs for young athletes 2 to 7 years old with and without intellectual disabilities.[12]
- Elite USA and World Games[10]
  - Divisioning for athletes is similar to the classification, as mentioned in the "Paralympics" section.[10,11]
- Deaflympics
  - Deaflympics are elite sporting games for deaf athletes.
  - Deaflympics are hosted by the International Committee of Sports for the Deaf.[13]
- Sports participation guidelines should be considered for athletes with medical conditions (e.g., seizure disorders, bleeding disorders, cardiovascular disease, etc.).[14]
  - If athletes are on medications, the medication side effects/impact on sports performance or safety also need to be considered.[15]
    - Medications can cause side effects or have safety implications; therefore, it is important to have a baseline medical history form with a medication list on each athlete.[16]

    - Sports competitions may have antidoping screening and regulations. If an athlete requires medications for a medical condition, a Therapeutic Use Exemption (TUE) may need to be submitted by a deadline for the athlete.[17,18]
    - The World Anti-Doping Agency (WADA) and the Global Drug Reference Online (Global DRO) are useful resources to check the status of medications.[19,20]
- Barriers and facilitators of physical activity participation for children/adolescents with disabilities
    - Barriers include the following (those with an asterisk are similar between individuals with disabilities and those without disabilities):
        - Cost*
        - Transportation availability*
        - Lack of time*
        - Lack of motivation*
        - Accessible program/facility/equipment availability
        - Physical limitations
        - Pain
        - Self-confidence[21,22]
    - Facilitators include the following:
        - Accessible program/facility/equipment availability
        - Knowledgeable and supportive coaches/instructors/teammates[21,22]
- Pediatric physicians should include discussions about sports and physical activity as an essential part of an evaluation with every child.[23]

SPORTS MEDICINE PRIMER FOR PHYSICIANS

- While some sports medicine injuries are similar in adult and pediatric patients, many injuries are quite different due to growth and growth-related injuries.
- This chapter presents a primer on pediatric sports medicine conditions and is not all-encompassing.
    - This primer focuses on some of the main sports medicine conditions unique to the pediatric/adolescent population.
    - Given the unique nature of pediatric/adolescent musculoskeletal injuries, referral to a pediatric sports medicine or pediatric orthopedic surgeon is recommended for pediatric-onset sports injuries.
    - When assessing pediatric musculoskeletal conditions, child abuse, bone tumors, pediatric orthopedic conditions (such as Freiberg disease), and rheumatologic conditions (such as juvenile idiopathic arthritis) should remain on the differential.[24–28]
    - Sports injuries such as sprains, strains, stress fractures, and joint internal derangement (e.g., anterior cruciate ligament tear, meniscus tear) are commonly seen in pediatric athletes, but are out of the scope of this chapter.

Pediatric Fractures

- Fractures are common in the pediatric population.[26,29,30]
- Due to the nature of immature growing bone, fractures may not break all the way through and may bend (plastic deformation), buckle (torus/buckle fracture), or be incomplete (greenstick fracture).[31]
- *SalterHarris (SH) fractures:* These are fractures through the epiphyseal growth plate (physis), most commonly classified using the SH classification system.
  - When using the SALTER mnemonic, be sure to draw the image in the correct orientation, as demonstrated in Figure 16.1.
    - **S**—type I: **S**traight across through the physis
    - **A**—type II: **A**bove the physis, extending through the physis and into the metaphysis
    - **L**—type III: **L**ower to the physis, extending through the physis and into the epiphysis
    - **T**—type IV: **T**hrough (extends through the physis, metaphysis, and epiphysis)
    - **ER**—type V: **ER**ased severe crush or compression injury of the physis[32–34]
  - Fractures should be managed by providers who are formally trained in pediatric fracture diagnosis and management, which is beyond the scope of this chapter.[32]

Effect of Growth on Muscle Tightness/Injuries

- Peak height velocity is typically reached at the age of 12 for girls and at the age of 14 for boys.[35,36]
- Muscular imbalances occur during peak height velocity, which may predispose youth athletes to injury.[36,37]
  - The length of the muscle tendon complex lags behind the growth of the long bones.[37]
  - With increased limb length, increased muscle forces must be generated to perform a movement task, which can increase stress to tendons and apophyses.[38]
- Children/adolescents with spasticity may also have increased muscle tension during peak times of growth.

Apophysitis

- Characterized by irritation at the apophysis (minor growth center, which is usually adjacent to the attachment site of a tendon to the bone; Table 16.1).[39]
- Symptoms include pain at the site of the apophysis in a growing child with an open apophysis.
- Diagnosis
  - Apophysitis is typically diagnosed clinically with pain and tenderness to palpation over the apophysis; however, in some cases, plain film x-rays can be useful.

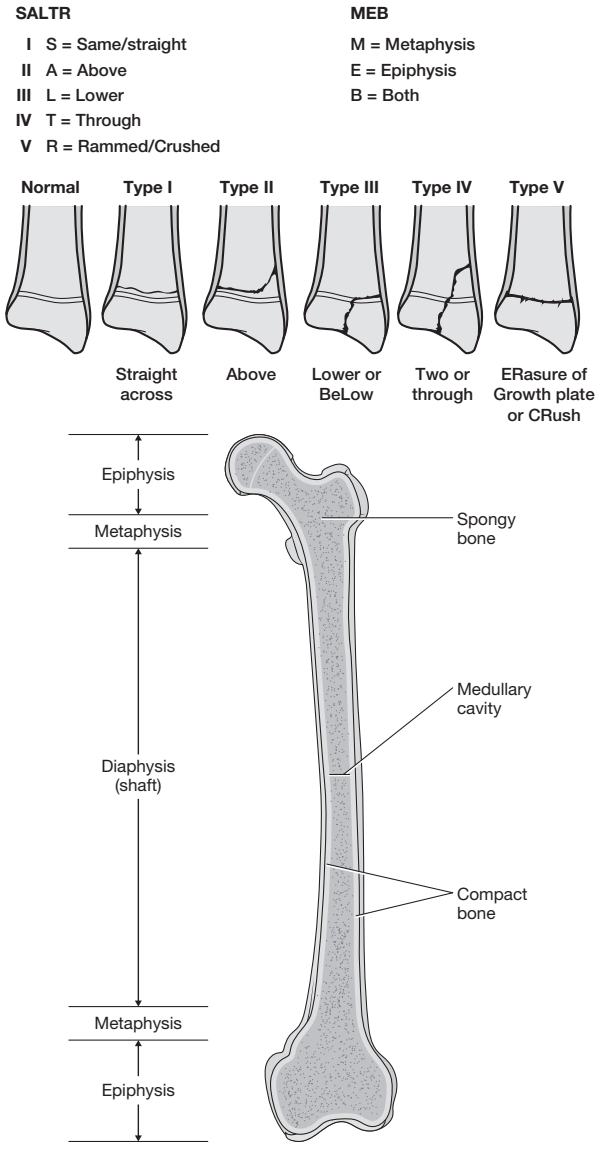

Figure 16.1 SalterHarris classification system for epiphyseal growth plate (physis) fractures.

Source: Kyle T. *Primary Care Pediatrics for the Nurse Practitioner: A Practical Approach*. Springer Publishing Company, 2022; Tkacs NC, Herrmann, LL, Johnson RL. *Advanced Physiology and Pathophysiology: Essentials for Clinical Practice*. Springer Publishing Company, 2020.

Table 16.1 Examples of Apophyseal Injuries in Children/Adolescents

APOPHYSITIS	LOCATION	AGE	DIAGNOSIS	TREATMENT
OsgoodSchlatter disease	Apophysis at the tibial tuberosity (distal patellar tendon attachment)	8–13 years old (female) 10–15 years old (male)	Pain and tenderness at the tibial tuberosity +/– mild swelling +/– tibial tuberosity prominence Painful resisted knee extension	Activity modification/rest Patellar tendon strap Padding over the tibial tuberosity Stretching of the quadriceps and hamstrings Ice to ease discomfort (after activity, not before activity)[40]
Sinding-LarsenJohansson syndrome	Apophysis at the inferior pole of the patella (proximal patellar tendon attachment)	8–13 years old Mean ages: • 8 years old (female) • 12 years old (male)	Pain and tenderness at the inferior pole of the patella	Activity modification/rest Patellar tendon strap Stretching of the quadriceps and hamstrings Ice to ease discomfort (after activity, not before activity)[40]
Sever disease	Calcaneal apophysis (Achilles tendon attachment)	7–15 years old Mean ages: • 9 years old (female) • 12 years old (male)	Pain and tenderness at the calcaneal apophysis (+ calcaneal squeeze test)	Activity modification/rest Calf stretching Heel cups Ice to ease discomfort (after activity, not before activity)[40]

Medial epicondyle apophysitis (little league elbow, thrower's elbow)[28,40]	Medial epicondyle apophysis (common elbow flexor tendon attachment)[40,41]	6–15 years old Peak age range: 11–12 years old[42]	Pain and tenderness at the medial epicondyle Plain film x-rays, and in some cases MRI, may be helpful to evaluate for widening or for other causes of elbow pain (such as osteochondritis dissecans)[40]	Rest from throwing until asymptomatic (typically 4–6 weeks) Physical therapy once asymptomatic (range of motion → core/proximal strengthening → sports-specific → gradual return to throwing) Typical time from diagnosis to return to throwing is 8–12 weeks Good prognosis with appropriate treatment; however, if throws through pain may have complications Widening of the apophysis 5 mm: refer to pediatric orthopedic surgery[28,40]
Iselin disease	Apophysis of the base of the fifth metatarsal (peroneus brevis attachment)	Most common ages: 12–13 years old Can be as young as: • 8 years old (female) • 10 years old (male)	Pain and tenderness at the base of the fifth metatarsal Pain with resisted ankle eversion and passive ankle inversion Plain x-ray images or MRI may be helpful to rule out other causes of pain	Activity modification/rest Peroneal and calf stretching Midfoot taping Ice to ease discomfort (after activity, not before activity)[40]

- Treatment
  - Rest/activity modification
  - Consideration of physical therapy
    - Stretching about the joint and targeted strengthening to decrease stress on the apophysis
  - Gradual return to sport[28,40]

Osteochondritis Dissecans

- Osteonecrosis of the subchondral bone
  - Can lead to injury to the cartilage
  - Can lead to loose body fragment
  - Joint pain, swelling, and limitations in range of motion
    - Common locations
      - Medial femoral condyle (knee)
      - Talar dome (ankle)
      - Capitellum (elbow)
  - Imaging
    - Plain film x-rays
    - May require MRI[43,44]
  - Treatment
    - Stable lesions (osteonecrotic fragment still attached)
      - Rest, may require nonweight-bearing, bracing, or crutches
      - Strongly recommend referral to a pediatric sports medicine or orthopedic specialist
    - Unstable lesions (fragment is detached)
      - Referral to pediatric orthopedic surgeon for surgical evaluation[44]

Spondylolysis

- Stress injury of the pars interarticularis
- Pain with lumbar extension
  - Commonly seen in sports that involve lumbar extension/loading (e.g., gymnastics, football)
- Imaging
  - Plain film x-rays do not rule out spondylolysis.
  - If x-rays are obtained, two-view (anteroposterior and lateral) x-rays are advised rather than four-view (anteroposterior, lateral, obliques) x-rays, which have higher radiation exposure without much added value.
  - If there is high suspicion, consider MRI (best for acute) or CT imaging (best for chronic).[45,46]
- Treatment
  - Rest from sports/impact/running for at least 10 to 12 weeks
    - Physical therapy
    - +/− bracing
  - After at least 10 to 12 weeks of rest, if asymptomatic, can start a gradual return to sport[46]

- *Bilateral spondylolysis:* can result in spondylolisthesis, or a slippage of one vertebra on another
  - Referral to pediatric orthopedics recommended if seen[46]

Preparticipation Physical Evaluations

- History and physical examination performed prior to sports participation to ensure safety in sports
- Screens for life-threatening medical conditions
- Healthcare touch point for athletes[16,47]
- Standard preparticipation physical forms available at the American Academy of Pediatrics (AAP) website
  - Medical history form
  - Physical evaluation form
  - Medical eligibility form
  - Athletes with a disability supplemental history form[16,48]
- A different preparticipation physical evaluation (PPE) form required for Special Olympics, which can be found on the website of both the Special Olympics and the AAP[48,49]
- *Preparticipation Physical Evaluation Monograph:* a reference guide that reviews the key components of the PPE and their medical relevance[16]

Athletes With Disabilities

- There is lack of research on injuries, illnesses, and sports medicine related to pediatric/adolescent athletes with disabilities.[10,17]
- Care of pediatric/adolescent athletes with disabilities requires specialized knowledge of pediatric sports medicine and pediatric disability care.
- Upper extremity injuries are more common in para athletes in wheelchair or seated equipment for their sports.[50]
- Lower extremity injuries are more common among ambulatory para athletes.[50]
- If athletes have spasticity at baseline, sudden increases in spasticity should raise suspicion for an underlying injury or medical illness.[51]
- For athletes with epilepsy or seizures, the International League Against Epilepsy established a consensus statement determining the risk of participation in various sports activities.[52,53]
- The following are medical considerations for athletes with various disabilities and medical conditions:
  - Athletes with spinal cord injuries
    - Athletes with spinal cord injuries at the T6 level or above are at increased risk of autonomic dysreflexia (AD).[17,51]
      - Boosting is the practice of intentionally inducing AD to enhance sports performance. It is a form of doping and is very dangerous.
      - The incidence of boosting is unclear and adaptive/para sports organizations have been working on the education of athletes, families, sports organizations, and staff on the risks and dangers of boosting.[54]

- There is risk of pressure sore due to impaired skin sensation and impaired motor function.[17,51]
  - Appropriate athletic equipment fit is important.[17]
- There is risk of hypothermia due to impaired thermoregulation.[17,51]
  - It is important to implement safety measures such as precooling measures prior to exercise and dressing for the weather.[55]
- Decreased bone mineral density could lead to increased risk of fractures, which may not present with pain given impaired sensation.
  - This may present with AD, increased spasms, bruising, swelling, and so forth.[17]

- Athletes with spina bifida
  - There is impaired sensation/risk of pressure ulcers.[17]
    - Appropriate athletic equipment fit is important.
  - There is increased risk of fracture with decreased bone mineral density.[17]
    - It may not present with pain, but may present with bruising, swelling, deformity, and so forth.
  - Neurogenic and bowel/and or bladder management[17]
    - Catheterizing and bowel programs should be accounted for during tournaments/competitions.[17]
  - There is risk of latex allergy.
    - It is important to have latex-free equipment.
  - There may also be hydrocephalus (requiring ventriculoperitoneal shunt) and/or Chiari malformation.
    - Monitor for neurologic changes concerning for shunt malfunction, Chiari malformation herniation, or tethered cord syndrome.[17]
- Athletes with limb deficiency/amputation
  - The residual limb may have skin conditions which may be related to prosthesis fit and/or terminal overgrowth in growing athletes with amputation through the long bone.[17]
    - Proper prosthetic fit is key.
  - Athletes may have neuromas that can be painful.
    - Treatment options include prosthetic adjustments, medications, injections, and in some cases surgery.[51]
- Athletes with cerebral palsy or brain injury
  - Decreased bone mineral density increases the risk of fractures.[17,51]
  - Impaired mobility leads to increased risk of pressure ulcers.[51]
    - Equipment fit is important.
  - Seizure disorder may be present in some.[17,51]
    - It is important for the medical team to be trained in seizure first aid and cardiopulmonary resuscitation techniques.[17]
- Athletes with blindness/low vision
  - Athletes with a single eye should wear protective eyewear.[16,53]

- In some sports, athletes must wear eyeshades to level the playing field between athletes with partial sight and blind athletes.[53,56]

- Athletes with intellectual disability
  - The cause of intellectual disability may or may not be known.[57]
    - If the cause is known, it is important to consider known associated medical conditions (e.g., atlantoaxial instability, congenital heart disease, seizures).[51]
    - Whether the cause is known or not, a thorough medical history and evaluation is important.
  - Clear examiner communication is key during PPE and medical care.[10]
  - Athletes with Down syndrome should be screened for atlantoaxial instability.
    - Special Olympics medical forms and PPE evaluations include assessment for signs and symptoms of spinal cord compression that could represent symptomatic atlantoaxial instability.
      - Athletes with symptoms of spinal cord compression require additional workup prior to consideration of athletic clearance.
      - Athletes without symptoms of spinal cord compression do not require screening x-rays.[10,58]
- Concussion in para sport
  - The first consensus statement on concussion in para sports was published by an international expert group in 2021.
    - There is still lack of research in this area.
    - Obtaining baseline examination is important.
    - Para sports follows the same principles as in mainstream sports population.
    - Adjustments are based on disability/impairment.
    - Table of recommended adjustments is based on impairment.[59]

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Musculoskeletal Conditions

17

LANE WIMBERLY, EDWIN PORTALATIN, and EMMANOUIL GRIGORIOU

GENERAL PRINCIPLES

The practice of pediatric orthopedic surgery encompasses a wide variety of musculoskeletal conditions, including genetic, developmental, metabolic, acquired, posttraumatic, and neurologic diagnoses. Hip dysplasia is a generic term indicating a clinically or radiographically abnormal relationship of the femoral head and the acetabulum of the pelvis. While the cause of the dysplasia can vary, the goal of treatment is to improve the joint mechanics and prolong the health and function of the hip, with a concentric and congruent hip reduction.

Adolescent idiopathic scoliosis (AIS) is a coronal plane curvature of the spine measuring greater than 10° when measured on an appropriate upright radiograph and by definition has no known etiology. Scoliosis can also be found in neurologic patients and in patients with congenital bone anomalies. Treatment may be nonoperative or surgical depending on the etiology, the magnitude of the curve, and the patient's maturity. Maintaining a healthy and well-balanced spine is the optimal outcome.

HIP DYSPLASIA

General Principles

Definition

The American Academy of Pediatrics defines dysplasia of the hip as a condition in which the femoral head has an abnormal relationship with the acetabulum. This represents a spectrum of disorders with various presentations, ranging from a mere ultrasonographic acetabular deficiency in a newborn to a hip dislocation with pain and osteoarthritis in an adult.[1] By definition, developmental dysplasia of the hip (DDH) refers to an instability noted in a newborn or a young child. This also has been known as congenital dysplasia of the hip, but the term *developmental dysplasia* is preferred as it is more inclusive and recognizes the evolution of the insufficiency. In syndromic or neuromuscular patients, the presentation, natural history, and prognosis can be highly variable but follow a similar spectrum of pathology from radiographic changes, instability, and eventual dislocation of the femoral head. A teratologic dislocation of the hip is usually associated with neuromuscular syndromes, especially those related to muscle paralysis (e.g., myelodysplasia and arthrogryposis), and the hips

are dislocated before birth and are rarely mobile during clinical examination and require surgical care to achieve reduction.

Epidemiology

- 1 in 1,000 to 3 in 100 depending on the population studied
- Dislocation reported at 1.0 to 1.5 per 1,000 live births
- More common in female infants

The incidence of DDH has been variably reported in the literature secondary to disparities in the definition of the condition, the population being studied, and the degree of screening that is in place in different countries. It is more common in females (6:1) as well as in Native Americans and Laplanders.[2]

Etiology

- Predisposing risk factors
  - Firstborn
  - Female infants
  - Breech presentation
  - Positive family history

Although no single cause of DDH has been identified, many predisposing risk factors are well-described. Other variables often found in association are overall ligamentous laxity, intrauterine positioning difficulties, extremes of postnatal positioning (use of swaddling boards), and racial predilection.[3]

- Associated orthopedic pathology
  - Clubfoot
  - Torticollis

In normal embryology, the hip joint develops at approximately seventh week of gestation and the concave shape of the hip socket is strongly influenced by the round femoral head abutting against the acetabular cartilage of the acetabulum. At birth, the acetabulum and femoral head are completely cartilaginous. The cartilaginous femoral head begins to ossify at the center of the epiphysis, usually before 9 months of age, and grows until late adolescence, ultimately giving the femoral head its characteristic round shape with a thin layer of articular cartilage. In a dislocated hip, delayed ossification is expected. If the proximal femoral epiphysis is not resting within the acetabulum during development, the socket will be shallow and the femoral head ultimately will lose roundness. The majority of acetabular development is completed by 8 years. If the hip is left unreduced, the shape of the acetabulum and the femoral epiphysis are no longer congruous and a mismatch is expected if there is an attempt at reduction. This lack of concentricity will lead to altered forces within the joint and eventually changes consistent with premature osteoarthritis.

The femur is also sensitive to muscle imbalances that can also significantly affect the growth and morphology of the proximal femur. For example, excessive adductor tone or inadequate abductor muscle function

results in a valgus deformity of the upper femur that is commonly seen in the spastic cerebral palsy patient. A failure of expected remodeling of infantile femoral anteversion also contributes to hip instability.

Diagnosis

Clinical Presentation

In neonates, physical examination findings may be subtle and an effective evaluation requires a calm infant. A quick assessment of hip abduction is appropriate and asymmetry may be a sign of a unilateral hip dislocation. Another physical examination known as the Klisic sign can identify a dislocated hip and may be particularly useful when both hips are dislocated. To assess this finding, an imaginary line is drawn from the palpable greater trochanter of the femur to the anterior superior iliac spine of the pelvis. If the line is below the umbilicus, it implies the greater trochanter is higher than normal and a sign of potential hip dislocation. It is also common to examine a child for excessive or asymmetric thigh folds as a sign of dislocation. The soft tissues are usually normal but will accordion as the hip dislocates and effectively shortens. This bunching of the normal soft tissue creates the excessive folds found in DDH.

In babies with true hip instability, the examiner may feel the hip dislocate from the acetabulum (Barlow-positive) or note the dislocated hip may be reduced into the socket (Ortolani-positive). Occasionally, the hip merely slides excessively in the socket without a frank dislocation (subluxation). The terminology of the examination can be confusing, and as such it is recommended the physician note their examination findings in plain language describing the relative stability and reducibility of the hip. A simple hip click often represents a normal sliding of a tendon over a bony prominence and may be misinterpreted as a hip dislocation.[4]

- Physical examination findings
  - Hip instability
    - Hip able to dislocate from the acetabulum (Barlow-positive)
    - Hip can be reduced into the acetabulum (Ortolani-positive)
    - Often a combination of the two findings
  - Limb length difference
    - Foreshortened dislocated hip
    - Increased thigh soft tissue folds
  - Limp
    - Limb length difference
    - Hip instability with stance
    - Increased lumbar lordosis, especially in bilateral dislocations
  - Klisic sign
    - Indicates palpably elevated greater trochanter that implies hip dislocation

If a hip dislocation is unrecognized at infancy, parents may notice a leg length discrepancy or a limp during early walking. If both hips are

dislocated, an increased lumbar lordosis and potential Trendelenburg gait may be identified.[5]

Radiographic Assessment

Within the spectrum of DDH, some infants merely have deficient acetabular development that is recognized only with radiographic assessment. It is recommended that an ultrasound be obtained in infants at 6 weeks of age when risk factors are present, regardless of examination findings (Figure 17.1). Although some mild acetabular dysplasia may improve without intervention, other patients may slowly progress to frank dislocation if not treated. For patients less than 6 to 12 months of age, the imaging modality of choice is a dynamic ultrasound. After approximately 9 to 12 months, the normal femoral head ossific nucleus is present and plain anteroposterior pelvic radiographs are the preferred method for evaluating DDH and monitoring appropriate remodeling of the acetabulum (Figure 17.2).[1,6,7]

Treatment

Initial Management

The goal of treatment of DDH is to achieve and maintain a concentric reduction of the femoral head in the acetabulum that will allow for appropriate continued acetabular development and a normal hip joint. There is much variability in the clinical management of DDH in infants. In general, infants under the age of 4 to 6 months are managed with nonoperative splinting to maintain the hips in a flexed and abducted position to

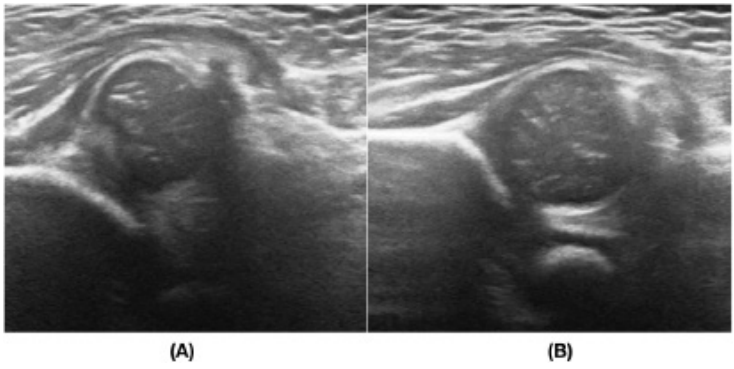

Figure 17.1 (**A**) An ultrasound demonstrating hip dislocation. The unossified femoral head is resting out of the bony acetabulum within the abductor muscle. This is the preferred imaging modality for children under 6 to 9 months of age. (**B**) By comparison, a reduced hip rests concentrically within the acetabulum.

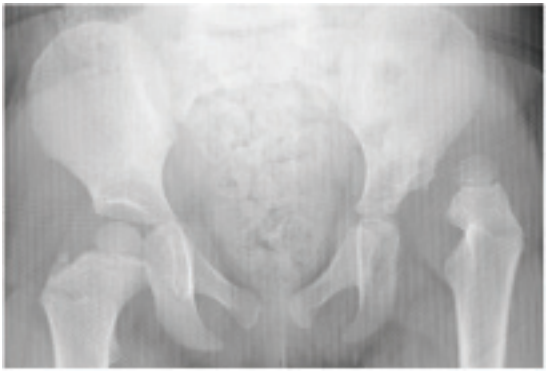

Figure 17.2 A plain radiograph demonstrating a dislocated hip in a 2-year-old. Note the smaller ossification center of the proximal femur and the upsloping, shallow acetabulum indicating existing dysplasia.

encourage stability. The most common device used is a Pavlik harness. The harness is relatively safe; however, the possibility of femoral nerve palsy from excessive flexion and avascular necrosis from forced abduction are described complications of its use.[1,5,7,8]

In patients who fail to reduce with nonoperative care, consideration for an attempted closed reduction and casting is often the next line of treatment. A prolonged course of casting may be needed to achieve a stable reduction and acetabular dysplasia may persist and eventually require correction, despite adequate reduction.

In patients who are persistently unstable despite attempts at closed reduction and casting, surgical care is recommended. The surgical approach and treatment algorithms are highly individualized and surgeon-dependent. In general, the hip joint capsule is opened and common impediments to reduction are removed to allow the proximal femur to medialize within the acetabulum. In patients over 12 months of age, an anterior approach to the hip is commonly preferred and also allows the surgeon to perform a capsulorrhaphy to tighten the expected redundant soft tissue of the hip capsule and aid in maintenance of reduction. An assessment of the tension required to reduce the hip may lead to femoral shortening and, depending on age and acetabular morphology, acetabular osteotomy may be needed to improve stability and reduce residual dysplasia. Complications of open reduction include growth disturbance of the proximal femur, residual dysplasia, and the possibility of repeat subluxation, especially in nonidiopathic conditions (Table 17.1).

- DDH treatment
  - Nonsurgical
    - Hip abduction bracing
    - Best for infants under 6 to 12 months of age

Table 17.1 Suggested Guidelines for Management of Developmental Dysplasia of the Hip

AGE	TREATMENT	COMMENTS
0–6 mo	Abduction orthosis	Commonly Pavlik harness
6–18 mo	Closed hip reduction and hip spica casting	For failed presentation or failed abduction orthosis use
>12–18 mo	Open hip reduction with possible femoral shortening osteotomy and hip spica casting	If closed reduction fails or is unstable
>2–8 y	Open hip reduction with possible femoral and pelvic osteotomies and hip spica casting	Pelvic osteotomy to address acetabular dysplasia
>8 y	Consider attempted surgical reduction	More difficult to obtain an acceptable concentric reduction

- Surgical
  - When nonoperative care fails to adequately reduce the hip
  - Closed reduction and casting
  - Open reductions with or without femoral and pelvic osteotomy

Ongoing Care

Most treated patients need to be followed clinically and radiographically until skeletal maturity to observe for residual, clinically relevant dysplasia that may require additional care. Some patients with significant risk factors but radiographically normal hips may also be followed during development.[5]

SCOLIOSIS

General Principles

Definition

Scoliosis is defined as a lateral curvature of the spine greater than 10° when measured on a radiograph.[9,10] The spine deformity is more

accurately described as a three-dimensional deformity with deviations in the coronal (lateral deviation), sagittal (kyphosis or lordosis), and axial (rotation) planes.[9,11] In the United States, approximately 1% to 3% of adolescents are affected.[12,13] Of scoliosis patients, 80% are considered idiopathic in etiology as an identifiable cause is never discovered.[9,10] Neuromuscular curves may be found in spastic, paralytic, and myotonic patients. A congenital scoliosis is found when there is improper development of the spinal column resulting in alterations of growth from failures of segmentation or formation. Many syndromic diagnoses are also associated with the development of spinal deformity.[10,13] Regardless of etiology, spine curvature tends to worsen with growth, and as such most treatments are targeted to the pediatric and adolescent populations.[13] As spinal deformity progresses, structural changes in the vertebrae and thoracic cage may result in pulmonary compromise, challenges with seating, and potential painful osteoarthritic changes. Thoracic curves that are >100° may affect the lung function of the patient secondary to restrictive lung disease.[9,11]

- Definition of scoliosis
  - Lateral curvature of greater than 10°
  - Smaller measurements considered spinal asymmetry
- Common types of scoliosis
  - Idiopathic
    - No identifiable cause, the most common
  - Congenital
    - Failure of normal development of the spinal column
    - Failure of segmentation
    - Failure of formation
  - Neuromuscular

Spine Development

Normal spine growth is a complex process consisting of more than 130 growth plates working in perfect synchronization.[14] Spine growth can be described in three phases: birth to age 5, age 5 to 10, and beyond 10 years.13–15 During the first and third phases, the spine experiences the most rapid growth.[15] As scoliosis worsens with spinal growth, the younger the child develops scoliosis, the higher the risk of progression.[9,14]

Adolescent Idiopathic Scoliosis

AIS is the most common type of scoliosis, diagnosed in a patient over 10 years of age at presentation, with a lateral curve of greater than 10° and without a known cause of deformity.[10,11] The incidence of a spinal curvature greater than 40° is .4% among adolescents.[16] AIS is a diagnosis of exclusion and can only be properly assigned after performing a thorough physical, neurologic, and radiographic examination.[10] During adolescent growth, full-spine standing radiographs are regularly obtained to monitor progression of the deformity. The most common curve pattern in AIS is a right thoracic left lumbar curve.

The natural history of idiopathic scoliosis depends on several factors, including skeletal maturity, sex, curve type, and curve magnitude.[11,16] Younger age, female sex, family history of scoliosis, and skeletal immaturity are risk factors for progression of the curve.[16] Even with the commencement of puberty, adolescents have about 13% of their growth left and are at risk of significant curve progression.[14] Once a curve magnitude is greater than 50°, it is very likely progression will occur, even beyond skeletal maturity.

Significant back pain is not associated with the presence of scoliosis; however, many patients do indicate some discomfort and the incidence of overall adult-type back pain is reportedly greater than the general population. When the patient's chief complaint is back pain in the presence of a curve, the examiner would be prudent to consider an alternate diagnosis that may cause the deformity, including tumor, infection, or inflammation.[16]

- AIS
  - No identifiable cause
  - Progression risks
    - Skeletal immaturity
    - Larger initial curve magnitude

Diagnosis and Radiographic Assessment

The presence of a curve is often identified by parents or patients who notice prominence of the ribs of the back, breast asymmetry, shoulders that appear to be of different heights, or waist asymmetry. The three-dimensional nature of the scoliotic changes leads to these findings as the rib cage will rotate into the curve. School screening programs often identify patients with spinal deformity by performing two common tests: the forward bend test and scoliometer measurement.[10,16]

Adam's Forward Bend Test

In the examination room, the examiner asks the patient to face away and bend forward at the waist until the spine levels with the ground. Preferably, the shirt is removed, although the bra may remain in place. The examiner proceeds to evaluate the back, ribs, and shoulders for any signs of asymmetry. Any findings on clinical examination suggestive of asymmetry may suggest rotation of the thorax, which could be related to scoliosis (Figure 17.3).[17]

Scoliometer

A scoliometer is an instrument like a carpenter's level which can be used in combination with the forward bend test to quantify the degree of scoliosis. The instrument is placed at the point of maximal asymmetry on the patient's back while performing the forward bend test. The cutoffs at which radiography is ordered vary from 5° to 7° of the trunk rotation via the scoliometer. In a patient of normal weight, a 7° measure at the scoliometer correlates with a 20° Cobb angle spine curve.[9,16,17]

During the initial assessment of the scoliosis, plain radiographs on posteroanterior and lateral views are obtained to measure the magnitude of

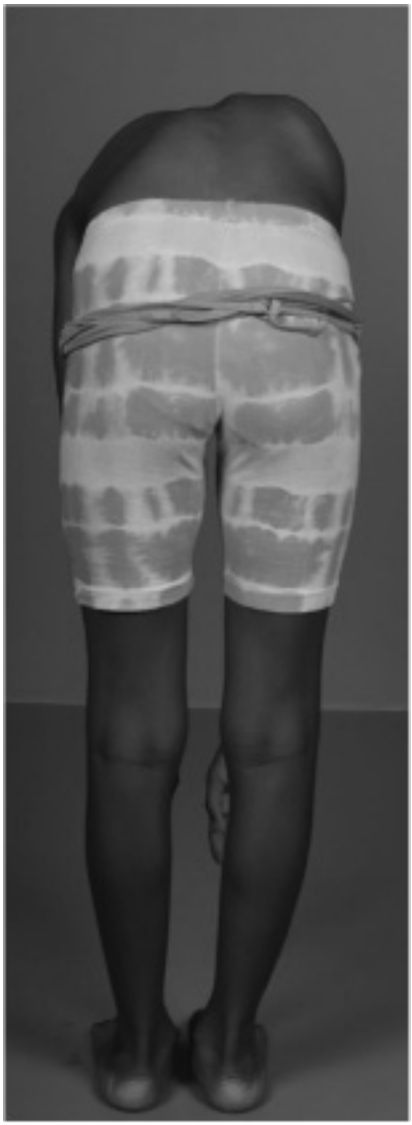

Figure 17.3 Adam's forward bend test. The prominence of the ribs is due to the rotatory deformity in adolescent idiopathic scoliosis. When this asymmetry is noted, formal evaluation for scoliosis is indicated.

the curve, to assess for congenital anomalies, and to ensure the radiographic characteristics of the curve are consistent with an idiopathic diagnosis. It is unnecessary to routinely obtain lateral views for continued observation of curve progression. Cobb angle measurement, the main standard for radiographic evaluation of scoliosis, is defined by the angle formed by the intersection of two lines parallel to the upper and lower end vertebrae most tilted from the midline. A curve is considered to have progressed if there is an increase of 5° to 7° between consecutive radiographic examinations, as measured changes of less than 5° to 7° may be within measurement error. Traditionally, skeletal maturity is estimated by reviewing the Risser sign, which assesses the ossification of the apophysis of the ilium on a standard posteroanterior view of the spine (Figure 17.4).[10]

- Radiographic assessment
  - Measure the coronal and sagittal curve.
  - Observe for markers of rotation, including rib prominence and pedicle asymmetry.
  - Review skeletal maturity.

Treatment

Proper management of AIS requires an understanding of the patient's growth remaining, the magnitude of the curve, and the likelihood of the progression of the deformity.[15] In general, curves are considered nonsurgical when the Cobb angle is <50°.

In most skeletally immature adolescent patients presenting with a curve with an initial Cobb angle less than 20°, vigilant observation is recommended, with regular scheduled clinical and radiographic follow-up.[10,16] For patients who present with curves from 20° to 40°, the recommendations for treatment are determined by the relative skeletal maturity of the patient. In most premenarchal or skeletally immature patients, who have not yet experienced the peak height growth velocity of adolescence, brace treatment should be considered. The goal of bracing is to halt the progression of the curve, not to correct the existing deformity. Brace prescriptions should recommend the wear exceed 12 hours a day to achieve the most benefit and bracing should be continued until skeletal maturity.[12] If the patient is skeletally mature with a curve magnitude of 20° to 40°, periodic observation is recommended as this curve has limited potential to progress to surgical magnitude. Other nonsurgical treatments including chiropractic care, massage therapy, and corrective spinal exercises have failed to demonstrate reproducible and lasting curve improvement, although research continues in these alternate modalities.

When a patient has developed a curve of greater than 45°, discussion for surgery is appropriate. The current standard of care and treatment is a posterior spinal fusion with instrumentation involving the spinal vertebrae that compose the curve. The goals of surgery should be to correct the deformity, prevent progression of the curve, restore trunk symmetry and balance, and minimize potential for future pain and morbidity (Figure 17.5).[11]

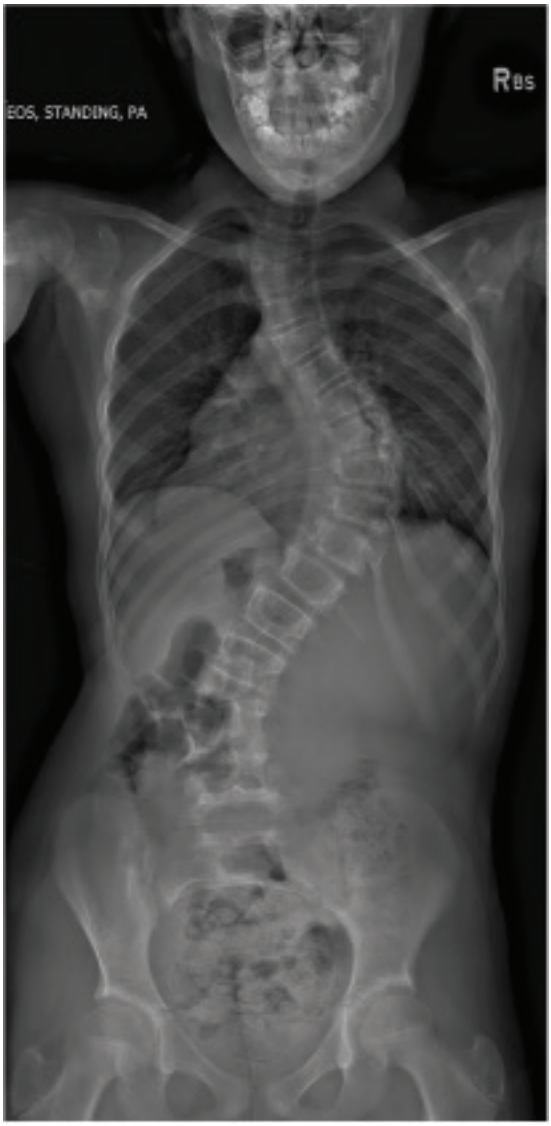

Figure 17.4 Plain posteroanterior radiograph of the entire spine demonstrating a surgical magnitude curve of the thoracic spine.

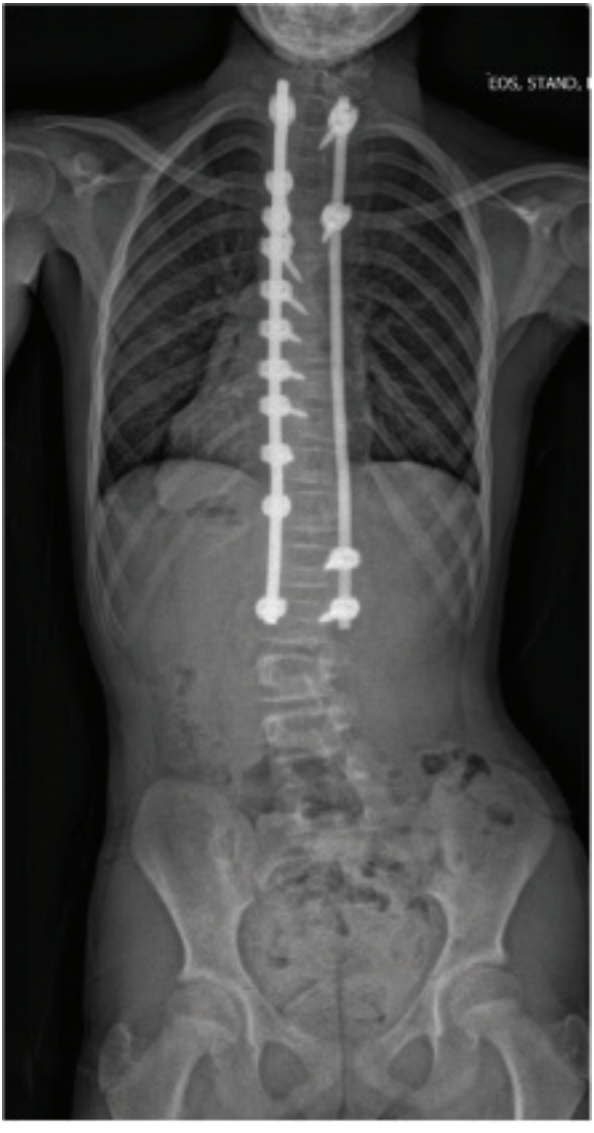

Figure 17.5 Postoperative radiograph of the spine after a posterior spinal fusion with instrumentation and grafting. Note the improved spinal balance.

- AIS treatment
  - Nonsurgical
    - Skeletally immature
    - Curve magnitude typically between 20° and 45°
    - Varied brace prescriptions
  - Surgical
    - Halt curve progression with spinal fusion with instrumentation
    - Achieve some curve correction
    - Goal to improve spinal balance and preserve motion in uninvolved segments

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PART V

Evaluation and Treatment Modalities

Pediatric Orthotics

18

SUPREET DESHPANDE, MARK E. GORMLEY JR., and ALYSSA DAHLHEIMER

GENERAL PRINCIPLES

Orthoses are an integral part of management of children with musculoskeletal or neuromuscular disorders. The International Organization for Standardization defines orthosis as "[a]n externally applied device used to modify the structural and functional characteristics of the neuromuscular and skeletal systems by applying forces to the body."[1] The objective of using an orthosis can be to protect a body segment, prevent contractures, increase stability, or optimize function.[2,3] In this chapter, we will review various orthoses and their role in improving function and participation.

The World Health Organization developed the International Classification of Functioning, Disability and Health (ICF) for assessing health and disability of both the individual and the population.[4] It takes into account psychosocial factors, in addition to anatomical and physiological factors, and the effects on the activity and participation of individuals with disabilities.[4,5] Incorporating these tenets with the assistance of an interdisciplinary team[6] may help facilitate a more personalized approach to orthotic care.[5,7]

Currently,[5] orthoses are generally made of low-temperature or high-temperature thermoplastics and have the benefit of ease of modification with heating. Low-temperature thermoplastics can be easily molded to the body after softening in hot water, tend to be less strong, and are generally used in upper extremity orthosis and postoperative orthosis. High-temperature thermoplastics need to be heated to 200°C and molded to plaster models. The most common high-temperature thermoplastic is polypropylene, which comes in various thicknesses and colors. Orthoses can be modified by adding foams and rubbers, steel or aluminum metal stays, and hinges.[8]

In general, the accepted nomenclature for an orthosis describes the joints that it encloses.[6] So an ankle–foot orthosis (AFO) incorporates the ankle and the foot, and a knee–ankle–foot orthosis (KAFO) will, in addition, include the knee. Less frequently, the name used may describe the function of the orthosis, as in a "cranial remodeling orthosis,"[6] or may take the

name of a person or place, for example, University of California Berkeley Laboratories Shoe Insert (UCBL).[6]

UPPER EXTREMITY ORTHOSES

Upper limb orthoses primarily can serve two purposes:

- *Nonfunctional* hand splints are designed for the primary purpose of improving outcomes in the body function and structure domains of the ICF. For example, "resting hand splints" are used to prevent or correct muscle contracture.[9]
- *Functional* hand splints are designed to improve outcomes in the activity and participation domains of the ICF, such as handwriting or use of utensils at mealtime[9] (Table 18.1).

Upper limb orthoses can be custom-made or prefabricated. To fabricate custom upper limb orthoses, materials with low elasticity, low drapeability, and high rigidity are preferred, and firm handling is often required to obtain the desired position. Prefabricated orthoses are commonly made from neoprene with plastic or metal stays, or a microwavable thermoplastic stay with a neoprene liner. Some of these can be customized for patient size and fit.

Wrist–hand–finger orthoses (WHFO): These orthoses provide a composite stretch of the wrist and fingers.

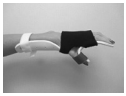
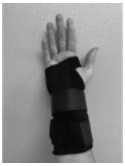
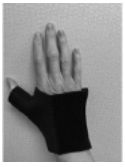
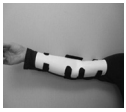
- These are indicated in a child who lacks full active or passive wrist and finger range of motion (ROM) and are typically worn at night so that the hand can be used as much as possible during the day.
- Patients with wrist, finger, or thumb contractures will need a custom orthosis.

Wrist–hand orthoses (WHO): These orthoses extend from the forearm and include part of the hand.

- These orthoses hold the wrist in extension, allowing for functional use of the hand, provided the long finger flexors are not contracted.
- These do not allow for tenodesis and they also cover the hand, reducing sensory input to these areas.

Hand finger orthoses: Hand finger orthoses are hand-based splints that extend to the thumb or one or more fingers. A commonly used hand finger orthosis in children is a thumb abduction splint. For patients who require rigid thumb support, a custom thumb spica can be fabricated to hold the thumb in the desired position and can be worn at night to stretch the thumb into radial or palmar abduction, or during the day to position the thumb for use. Neoprene thumb abduction orthoses are an option for less rigid support. Glove-style orthoses can provide more support for thumb positioning; however, they cover more of the hand, reducing sensory input. Styles with smaller straps across the hand and palm are nice for increasing sensory input but provide less support.

Table 18.1 Upper Extremity Orthoses

PREFABRICATED	CUSTOM-MADE	PURPOSE	MATERIALS
Wristhand–finger orthosis 		Provides a composite stretch of the wrist and fingers Typically worn at night	Custom with thermoplastic or prefabricated with plastic or metal stay and liner
Wristhand orthosis 		Positions the wrist in extension for hand use during the day May also include the thumb if needed	Custom with thermoplastic or prefabricated with neoprene and metal stay, or neoprene and microwavable thermoplastic
Handfinger orthosis 		Supports the hand, thumb, and finger or fingers Can be worn daytime for positioning or at night for a stretch	Custom with thermoplastic or prefabricated with neoprene, with or without thermoplastic stays
Elbow extension orthosis 		Positions and stretches the elbow into extension	Custom with thermoplastic or prefabricated with padded material with metal or plastic stays, or neoprene with microwavable thermoplastic stay

Elbow orthoses (EO): Elbow orthoses encompass the upper arm, elbow, and forearm.

- These are often used for management of an elbow flexion contracture.
- These are typically worn at night, leaving the arm free for functional use during the day.
  - A custom EO provides rigid, more contoured support to the elbow in extension and tends to work best when trying to achieve the last 10° of extension. Those with a microwavable thermoplastic stay can be better fitted to the patient to maintain elbow extension and can be more comfortable.

LOWER EXTREMITY ORTHOSES

Lower limb orthoses in children should meet several goals: improve functional mobility or stance stability, reduce the risk of deformity, and need to be comfortable. When picking a brace, multiple factors need to be considered, including the activities when the brace will be used, the child's strength, flexibility, and muscle tone, and the tolerance in wearing the brace. These factors can change as a child grows and develops, thus bracing needs can change over time.

Lower extremity orthoses are designed to support the joints of the lower limb. These include foot orthoses (FO), supramalleolar orthoses (SMO), AFO, knee orthoses (KO), KAFO, hip–knee–ankle–foot orthoses (HKAFO), reciprocating gait orthoses (RGO), and other specialty braces. (An RGO is an HKAFO with a cable system at the hip that facilitates hip extension when the contralateral hip is flexed, thus facilitating a reciprocal gait pattern.) Materials can vary from plastic, silicone liners, carbon fiber, metal, and leather. Most current orthoses in children are fabricated out of plastic and are custom-molded due to the varying size and shape of children's lower limbs. Recently, fabrication techniques have allowed the inclusion of silicone liners, which can improve tolerance in children who are sensitive to rigid plastics. Shoes designed to accommodate a brace are now available online in various styles and colors and these shoes have improved fit over the braces.

Orthoses can improve the ground reaction forces which support a child in stance. For example, a child with poor plantarflexion strength and excess dorsiflexion in stance can crouch in stance or when walking because their poor plantarflexion force decreases the knee extension moment.[10] A solid AFO can help improve the knee extension moment in stance by providing plantarflexion stability in stance and can reduce crouch.[11,12] Similarly, a KAFO can support the knee and ankle in a child with poor knee extension and plantarflexion strength.

ORTHOTIC USE IN COMMON CONDITIONS

Cerebral Palsy

Children with cerebral palsy (CP) can have varying degrees of impairments, from very functional ambulation to being nonambulatory with no

means of independent mobility. These impairments can be complicated by weakness, spasticity, contractures, and poor selective motor control.[13–15] Orthoses can improve gait and deformity in children with CP.[6] The most common brace used in children with CP is the AFO.

- About 50% of children with CP use AFOs, peaking at 67% at 5 years old and decreases to 20% by adulthood.
- AFO use increases with increasing functional disability in children with CP as measured by the Gross Motor Function Classification System (GMFCS), from 34% use in GMFCS I to 70% in GMFCS IV–V.[15]
- AFOs can be fabricated out of various materials with varying designs. The materials used should be strong and durable enough to control the foot and ankle position and minimize the consequences of spasticity.
- AFOs can be articulated and allow free dorsiflexion, but commonly have a plantarflexion stop at about 90° to prevent excess plantarflexion and toe walking. AFOs can also be solid with no significant plantarflexion or dorsiflexion movement. Posterior leaf spring (PLS) AFOs have trimmed posterior lines around the ankle to allow some flexibility at the ankle while also providing some stiffness and resistance to excess plantarflexion and dorsiflexion. PLS AFOs can be made from plastic, but prefabricated PLS AFOs are often made from carbon fiber. Flexible carbon fiber PLS AFOs may not be strong enough to control plantarflexion in a child with spasticity and may be restricted to children with CP who have minimal hypertonia.
- AFOs have been shown to improve gait efficiency, stance stability, and walking speed and to prevent plantarflexion contractures in children with CP.[16–18]
- Children with CP often have excess plantarflexion tone and contractures which can lead to an excess knee extension moment and genu recurvatum. AFOs that prevent plantarflexion can decrease this excessive knee extension moment, thus improving genu recurvatum and hip extension in stance.[19,20]
- Solid AFOs in children with CP who are nonambulatory, with GMFCS IV and V, can reduce contractures and improve stability in a stander or during stand pivot transfers.[6]
- Both solid AFOs and articulated AFOs improve gait speed, step length, and cadence versus no AFO.[21,22] Children at GMFCS level III who walk with an assistive device, such as a walker or crutches, have better hip and knee stability if they wear solid AFOs.
- Solid AFOs improve crouched gait,[10,12] but more so in children with more severe gait impairments (GMFCS III–IV) than in children with a more functional gait (GMFCS I–II).[23]

Ground reaction AFOs (GRAFOs) are solid AFOs with an incorporated anterior plastic shelf at the upper front of the shin to hypothetically provide more ground reaction forces and increased knee extension moment in stance than a standard solid AFO. In children with CP, while both solid

AFOs and GRAFOs improve crouched gait, there is not a significant advantage seen between the braces on computerized gait analysis.[11]

Children with a higher level of mobility, with GMFCS I or II, or walk without an assistive device have a more functional gait with articulated AFOs than solid AFOs if they have adequate plantarflexion strength.[24] Articulated AFOs can improve ankle dorsiflexion and fluidity of walking if ankle spasticity and contractures are not severe.[25–27] Articulated AFOs work more effectively than solid or PLS AFOs on stairs, during sit-to-stand transitions, and other tasks, even if no significant differences between the style of brace are seen in walking on computerized gait analysis.[28]

AFOs can improve gait and help preserve the positive changes after orthopedic surgery.[29] AFOs also improve step length, gait speed, and ankle kinematics more substantially in children with hemiplegic CP versus diplegic CP.[30]

Patients with CP rarely use KAFOs or HKAFOs. These more extensive braces can increase energy needs and not improve functional mobility.[6,10]

Twister cables have been used to improve intoeing commonly seen in children with CP due to femoral anteversion. Twister cables can be difficult to keep properly aligned, and although they can improve the cosmesis of gait by improving the foot line of progression twister cables usually do not improve stability and walking efficiency, thus not routinely recommended in children with CP.[6]

In the past, specially contoured footplates have been incorporated in AFOs to reduce spasticity. Although contoured footplates can improve foot positioning, they have not been shown to reduce spasticity.[31]

In summary, children with CP can have improvements in gait by wearing AFOs, but their use diminishes as they get older. Both articulated and solid AFOs can improve gait, but articulated AFOs allow more ankle mobility and may be better for higher functioning children with CP, with GMFCS I or II, and who participate in a variety of activities. If a child with CP has adequate knee and ankle ROM and enough strength to walk in plantarflexion or "on their toes" across a room, they may do best with an articulated AFO. Solid or PLS AFOs may be the best choice for a child with a crouched gait and poor plantarflexion strength. Solid AFOs may more effectively improve knee and ankle stability in children functioning at the GMFCS III level and maintain ankle ROM in nonambulatory children functioning at the GMFCS IVV level. Twister cables, HKAFOs, KAFOs, and contoured footplates to reduce spasticity have little utility in children with CP.

Myelomeningocele

Myelomeningocele or spina bifida is a congenital neural tube defect, second only to CP as the most common childhood disability.[31] Unlike CP, which is usually characterized by hypertonia with functional sensation present, myelomeningocele is a lower motor neuron spinal cord impairment with varying levels of weakness, insensate skin, and hypotonia.[32]

These characteristics affect the orthoses used in children with myelomeningocele. The weakness and low muscle tone do not provide stance support so orthoses must be designed to accommodate these deficits, which usually means solid orthoses. Articulated AFOs do not provide enough stance stability in children with ankle weakness and low muscle tone. Most patients with myelomeningocele will not use HKAFOs or KAFOs for long-term mobility because they are cumbersome and less efficient than wheelchairs.[32] Insensate skin carries a higher risk of pressure injuries so custom-made orthoses and frequent skin checks are needed.[33] Patients with myelomeningocele who are ambulatory and have pressure injury have a 24% chance of requiring an amputation.[34] The level of motor impairment influences the likelihood of ambulation and the type of orthoses that will most benefit mobility.

Children with deficits in the lower thoracic area

- These children have some abdominal strength but poor lower extremity movement.
- Children and adults with lower thoracic level impairments use wheelchair for mobility.[32]

Children with high lumbar level function

- These children may have some hip flexion strength but weak quadriceps.
- They may use an RGO as a child but a wheelchair as an adult. By 18 months old, these children usually prefer a wheelchair for mobility.[35]
- If the quadriceps are weak, community ambulation is unlikely, but some patients may ambulate as adults for short distances using KAFOs.[36]

Children with midlumbar level function

- These children may have good quadricep strength but poor plantarflexion, dorsiflexion, knee flexion, and hip extension strength.
- They will usually ambulate household distance and may ambulate in the community with the use of KAFOs or AFOs, but commonly use a wheelchair in the community.
- If a child has fixed knee flexion contractures >20°, KAFOs will usually not be sufficient to support ambulation.[37]

Children with lower lumbar level function

- These children have good quadricep, medial hamstring, and dorsiflexion strength.
- They are likely able to ambulate household and short community distances with bracing and crutches.[35] Due to poor plantarflexion and hip extension strength, these patients benefit most from custom-made solid AFOs to minimize dorsiflexion in stance and rocker bottom shoes to help with weight progression forward.[32,35]
- They will commonly use a swing through gait, as opposed to a reciprocating gait, because it is quicker and more efficient.[38]

- Children with high sacral level function and some plantarflexion strength may be able to walk with SMOs or PLS AFOs. These children usually walk in the community and do not require assistive devices.[35,37]

In summary, children with myelomeningocele require solid orthoses due to low muscle tone and weakness. Mid and lower lumbar level involved children will often ambulate with solid AFOs and crutches. Children with sacral level involvement generally can ambulate with SMOs for ankle support.

Muscular Dystrophies and Neuromuscular Disorders

Most common childhood neuromuscular disorders, Duchenne muscular dystrophy (DMD), spinal muscular atrophy (SMA), hereditary motor and sensory neuropathies (HMSN), and other similar muscular dystrophies have low muscle tone and weakness.[39] Orthoses can help stance stability and minimize musculoskeletal deformities. However, most muscular dystrophies are slowly progressive and orthotic needs can change over time.

Boys with DMD have diffuse weakness which can adversely affect their gait. These boys develop compensatory postures to position their center of gravity in front of their knees and behind their hips.[40,41] Toe walking helps position the center of gravity appropriately and AFOs may negate this compensatory posture.[42] However, some are more secure walking with AFOs. Historically, KAFOs and tendon lengthening surgery have been used to prolong ambulation in boys with DMD.[43] However, these gait improvements can be minimal and require a lot of rehabilitation and commitment from the boys and their families and are currently used much less frequently.[42] Boys with DMD will universally develop severe plantarflexion contractures as their disease progresses, particularly once they have become nonambulatory. Nighttime AFOs can prevent contractures if routinely used and initiated when the ankle cannot be dorsiflexed past a neutral position.[44,45]

Similarly, children with SMA may benefit from AFOs for stance stability and contracture prevention. With the advent of genetic treatments for SMA, standing and walking have become more prevalent and ankle stabilization with solid AFOs may be helpful. Children with SMA rarely benefit from full KAFOs.

HMSN and CharcotMarieTooth (CMT) disease are hereditary neuropathies which are usually slowly progressive and can lead to distal lower extremity weakness (especially weak dorsiflexion), hypotonia, sensory loss, steppage gait pattern, and progressive cavus foot deformities.[46] Early in the course of the disease, most patients do not wish to wear an AFO, but as weakness worsens, especially plantarflexion strength, AFOs are more widely accepted.[47] If adequate ROM is present at the ankle, a carbon fiber AFO can provide assistance in dorsiflexion during the swing phase and some plantarflexion forces at push-off. Shoe insert to help support the cavus foot deformity may be helpful especially if it helps relieve discomfort.[48]

Pes Planus Foot Deformities

"Flat feet," pes planus, or pes planovalgus foot deformities are a common worry of parents, especially in children with mild developmental concerns. Pes planus foot position is typical in children during stance up to 4 years of age, and the medial longitudinal foot arch usually does not fully form until 8 years old.[49]

- *Flexible asymptomatic flat feet:* Foot orthoses do not change a child's foot integrity long term and bracing is not needed.[50]
- *Hypotonia and hyperflexible feet:* Children may develop significant planovalgus foot deformities and respond best to SMOs, which can improve functional activities.[51,52]
- *Symptomatic pes planovalgus foot deformities:* These are commonly characterized by foot pain, especially after walking long distances or on uneven surfaces. An FO or a shoe insert or a UCBL brace may help.

Pes planovalgus foot deformities in a child may be a consequence of restricted ankle dorsiflexion. If a child's ankle cannot be dorsiflexed past 90° with their foot in subtalar neutral and knee fully extended, improved ankle ROM may help improve their foot position.

If a child has a rigid hindfoot, a tarsal coalition may be present. This occurs when the midfoot and hindfoot bones do not fully separate during growth and the foot develops positional deformities, which can become painful, especially after 10 years old. A rigid FO (UCBL or SMO) may help improve symptoms, but if pain persists surgery may be necessary.[53]

SPINAL ORTHOTICS

Spinal orthoses can be prefabricated or custom-made and have three main broad applications: (a) to manage pain by curtailing motion, (b) to stabilize the spine after an injury, and (c) to allow correction or halt progression of a spinal deformity. They are classified not only by the region of the spine that they encompass, such as a cervical orthosis, but also by their stiffness, into rigid, semirigid, or flexible. They all function by utilizing a three-point pressure system to improve alignment of the spine, with the pressure being imparted on bony prominences.

Cervical orthoses (CO): These orthoses are most often prefabricated and can control motion but are not able to entirely immobilize the cervical spine.[54]

- *Foam soft collar:* This collar provides a proprioceptive reminder to decrease motion.[54,55] It is contraindicated in injuries that are likely to cause instability of the cervical spine.[56]
- *Semirigid, prefabricated collars:* These collars decrease flexion/extension movements but not lateral bending or rotational movements.[56]
- *Four-poster style orthosis:* This orthosis provides better cervical motion restriction as it has an occipital, mandibular, sternal, and thoracic pad.

Cervicothoracic orthosis (CTO): These orthoses are similar to a cervical orthosis but have distal extensions on to the chest and are able to provide more stabilization to the cervical spine.[54,56,57]

- *Sternal occipital mandibular immobilizer (SOMI):* This has a sternal plate to which a mandibular and an occipital pad are connected. It provides acceptable flexion control of the lower cervical segments but allows some extension and lateral motion, and can easily be donned on a supine patient.[58]
- *Miami J collar with an extension that snaps on to a chest plate (Miami JTO):* This curtails flexion, extension, and rotation of the lower cervical and upper thoracic spine.[58] The absence of posterior struts allows proper alignment of the spine in all positions without increasing forces on the injured spine with changes in position.[58]
- *Minerva brace:* This has a two-shell design with mandibular and occipital support. It provides good intersegmental stabilization between the C2 and T3 spinal segments and is a feasible option for stable fractures in this area.[59–61] It may be safer and better tolerated than a halo.[61]
- *Noninvasive halo (NIH):* This has a chest piece connected to a chin support, a silicone "face mask," and an occipital support. It is better tolerated and has fewer complications than a halo,[62] and is indicated for use in postoperative stabilization after sternocleidomastoid release for congenital muscular torticollis, C1C2 subluxation, and odontoid fractures in children.[62,63]
- *Halo CTO:* This has a metal halo ring anchored to the skull by pins and fastened to a chest piece.[58] It is thought to be the best option for occiputC1 or C1C2 fractures or injuries, but is less desirable if the injury is between C2 and T3 as it allows for "intersegmental snaking."[58] The most common complications encountered with a halo are pin loosening and pin site infections.[64]

Thoracolumbar orthoses: These can be used for halting or slowing the progression of spinal deformities in skeletally immature children. The basic principles to achieve this are by a combined effect of endpoint control (mechanical constraints to the spine), transversely directed load using pads with the force vector being in an anteromedial direction in the general area of the apex of the curve, and reducing the curvature using a three-point pressure system.[58] Compliance though can be challenging as the conventional recommendation for adequate improvement of scoliosis is for 18 to 23 hours a day of brace wearing.[65]

- *Cervicothoracolumbosacral orthosis (CTLSO):* The Milwaukee brace is the only CTLSO. It has a pelvic girdle connected to a neck ring and has an anterior throat mold and two occipital pads.[58,65] It is indicated in single or double spinal curves, with the apex of the thoracic curve being above T8.[65]
- *Thoracolumbosacral orthoses (TLSO):* Some examples are the Boston brace and Miami orthosis, which are both a one-piece, rigid brace that opens

posteriorly, and Rosenberger, Lyonnaise, and Wilmington, which have anterior opening. TLSOs are preferable for curves between T6 and L4.[58,65]

- *Charleston bending brace and the Providence nocturnal scoliosis system:* These were developed to improve acceptance as the psychological factors of daytime use are eliminated.[58,65] They are recommended in children who have single, effortlessly reducible curves of 25° to 35° in the thoracolumbar or lumbar region.[58,65]
- *Thoracolumbar hyperextension orthoses:* These are designed to unload the anterior column and are suitable for stable compression fractures from T10 to L2.[66] A rigid lumbosacral orthosis (LSO) with a unilateral thigh extension successfully immobilizes L4 through S1 segments.[66]

Cranial orthoses (helmets): Cranial orthoses can be protective or corrective, prefabricated or custom-made. Protective helmets are recommended in children who are at high risk of falls, postoperatively after a craniectomy, or in children with self-injurious behaviors. A soft-shell helmet would be appropriate in a child who is nonambulatory and a hard-shell design in an independent ambulator.[67]

- Corrective or cranial remodeling orthoses (CROs) are recommended in children with positional plagiocephaly or brachycephaly. The purpose of a CRO is to impede growth in some areas and enhance growth in other areas to achieve cranial symmetry. The helmet is made with a close fit to the skull on the side that does not need to grow and with room for the skull to grow on the side with deformation.[68] The amount of correction achieved is proportional to skull growth during treatment and compliance of helmet wear. Early initiation of therapy, between 4 and 11 months, when the skull growth is rapid results in the greatest improvement in shape.[68,69] The duration of treatment is also reduced when CRO use is instituted early.[68,70] CROs are contraindicated in children with craniosynostosis and untreated hydrocephalus.[71] They do need to be cleaned with antibacterial soap three times a day.[67]

Flexible orthoses: These orthoses are an alternative to rigid orthoses. Some of the available varieties include the dynamic movement orthoses (DMO), the Benik orthoses, and the Stabilizing Pressure Input Orthosis (SPIO).

- *DMOs:* These are custom-fitted and provide correct alignment by adding support to specific areas. Their snug fit also provides proprioceptive feedback. They are designed to increase stability and decrease involuntary movements without loss of flexibility.[72] Wearing them even for 2 hours a day is effective.[72] The aim is to have persistent effects even after removal of the orthosis. An entire gamut of DMOs, from gloves to full body suits, are available.
- *SPIO:* These are similar to DMO flexible orthoses and they also have a wide range of orthoses, from WHO to lower body orthosis. They function much the same as the DMOs but can get quite warm for the user.

An SPIO vest can help improve kyphosis but not scoliosis or hip lateralization.[73]

- *Benik orthoses:* These are neoprene orthoses that provide mild dynamic support. They also come with optional malleable aluminum stays and moldable plastic panels, which can be used to increase support.

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Seating Evaluation

19

DEANNA LUSTY and ANN MODRCIN

GENERAL PRINCIPLES

Seating evaluation is intended to evaluate, prescribe, and fit a client for a wheelchair and is best performed in the context of a team approach. This chapter will help understand the role of team members and how to conduct a mat examination, provide an overview of the process of obtaining an equipment, and provide basic information on wheelchair bases and seating components.

TEAM MEMBERS AND ROLES

An integrated team provides an ideal approach to a seating system and wheelchair evaluation. All team members bring essential knowledge to the evaluation, and it is imperative to engage the client and/or the client's caregiver in the ultimate decision-making. Team members and their respective roles may include the following:

- *Client:* Clients bring their mobility history and share the expectations and needs they have related to mobility equipment.
- *Caregiver/family:* It is important to note that a client may have more than one caregiver and that the caregiver's needs may differ due to physical capabilities or settings. All environmental considerations and needs should be explored, with the team's focus being on where the client spends majority of their time.
- *Evaluating physical therapist (PT)/occupational therapist (OT):* It is the role of the evaluating PT/OT to understand and present how the client's combined pathophysiology, including diagnosis, muscle tone, reflexes, range of motion (ROM), strength, postural control, and functional mobility, will interplay in the decision-making.
- *Physician:* The physician will assess for longitudinal care needs associated with a known diagnosis in order to anticipate future postural needs in the life span of the wheelchair being prescribed.
- *Supplier:* It is the supplier's role to provide information about the different products on the market that are appropriate for the client, verify the measurements, and process the orders.
- *Manufacturing representative:* In some cases, a manufacturer's representative may be called in to help with a clinical trial or delivery of

specialized equipment. They can also provide expertise in more technical fittings and product use.
- *Funding source:* A request for the seating system and wheelchair, along with the reasons for medical necessity, is sent to the funding source for approval, and coverage for payment of the mobility equipment.

PROCESS OF OBTAINING EQUIPMENT

The World Health Organization has set forth the *Guidelines on the Provision of Manual Wheelchairs in Less Resourced Settings*[1]; however, this eight-step process is appropriate no matter where the equipment evaluation takes place to ensure a sustainable, ethical, and client-centered approach is maintained.

- *Need determined/referral:* A need for equipment can be identified by anyone and relayed to a medical professional to initiate referral to the assessment team.
- *Team assessment:* The multidisciplinary team assessment can consist of one or more appointments to optimize the recommendations and substantiate the medical necessity and appropriateness of the wheelchair prescription.
- *Prescription/documentation:* Team members complete their respective documentation of the client's examination, measurements, and recommendation needed for funding. Medical professionals compile the evaluation forms and submit a letter of medical necessity, including medical justification related to the patient's diagnosis, presentation, and mobility impairment. The supplier fills out the ordering forms and their company attains additional payor-specific documentation, and then submits all required information to the funding source.
- *Funding:* The payor or funding source reviews the documentation and determines if medical justification is adequate to approve the recommended equipment and if it is a covered benefit in the client's plan.
- *Ordering/product preparation:* Once the equipment is approved by the funding source, the supplier will order all the parts and arrange for shipping. At the supplier's facility, builders combine the parts sent from various manufacturers and set up the chair based on the measurements taken during the assessment.
- *Delivery/fitting:* At the delivery and fitting, it is ideal to have one of the evaluating medical professionals, supplier, client, and/or caregiver all present together. Fine-tuning adjustments are made to the seating system and wheelchair to ensure proper fit and function. Photo documentation is often helpful to include in the medical record.
- *Training:* Initial training begins at the time of delivery and fitting, but training often does not end here. The client/caregiver/family is taught how to fold and use the wheelchair at delivery; however, additional visits may be needed with the treating therapist to ensure the client is using the chair appropriately, with attention to proper positioning, transfers in/out of the device, manual wheelchair skills, or power wheelchair driving skills.

- *Follow-up maintenance/repairs:* Most follow-up maintenance and repairs are completed by the supplier in the client's setting or their office. If a significant medical or structural change has occurred that cannot be addressed in a straightforward service, a new referral may be needed for a seating team assessment.

EVALUATION

Evaluation may be completed in one or more sessions. History, functional review, and mat examination are usually completed together. If time allows and the equipment is available, clinical trial of the equipment and determining the goals for the seating and mobility device will also be completed.

History and Review

- Obtain medical history, including past, present, and planned medical interventions.
- Obtain history of assistive technology, including what is needed and what the client has already used.
- With current equipment, determine what is working well and what is not.
- Determine details of the environment where the mobility device will be used, including home, school, and community environment.
- Accessibility of the areas of use, how the device will be transported, assistance required, barriers, and considerations of emergency evacuation should be explored.

Mat Examination

During the mat examination, tone, reflexes, ROM, flexibility, strength, skin integrity, postural control, postural assessment, and functional assessment will be quantified. It is imperative to conduct the mat examination both in supine and sitting positions, beginning with the client positioned supine. By completing the examination first in a gravity-eliminated position, the therapist can identify concerning tone or postural anomalies that can result in harm and additional postural deviations when sitting.

Pelvis

- The base of all seating and its stability, or lack thereof, affect the structures above and below.
- In supine, palpate the pelvis and note if pelvic obliquity, rotation, or tilt exists in the frontal, transverse, or sagittal plane, respectively.
- With an abnormal finding, gently move the pelvis to see if the abnormality is flexible or fixed.
- For flexible abnormalities, determine whether these can be partially, fully, or overcorrected passively.

- Fixed deformities require accommodation, rather than correction in the sitting position.
- The ideal sitting pelvic posture is a supported active or slight anterior pelvic tilt, that allows for occasional rest in a slight posterior pelvic tilt.
- A posteriorly rotated pelvis will lead to a cascade of postural deviations.

Hips

- Proper seat-to-back angle of a seating system is crucial to achieving optimal postural control and may vary depending on the client's bony anatomy and tone.
- Hip ROM can be affected by a subluxed or dislocated hip or by muscle tone or contracture.
- It is important to assess both hip flexion and extension to determine the optimal position of rest.
- Take note that if there are any asymmetries from side to side these should be addressed separately.
- A seat-to-back angle plays an important role in managing dystonia and spasticity, especially when coupled with poor core strength and tone.
- An open seat-to-back angle in excess can elicit an extensor response.
- A closed seat-to-back angle or aggressive "anti-thrust" seat, causing excessive hip flexion, can trigger neck flexion and loss of trunk righting response.
- Evaluate hip abduction tone in a sitting position to determine if additional support is needed at the knee.

Knee

- The lower legs must be positioned at an angle that does not put tension on the hamstrings.
- Multiple leg hanger angle options (70°, 80°, 90°) and footplate adjustments allow the knees to be flexed greater than 90° so the hamstrings cannot pull the pelvis into a posterior tilt.

Foot

- It is important to know if the client wears orthotic devices and what type of shoe is typically worn.
- Footplate angles should accommodate the most foot-flat position attainable while sitting or match the braced angle of the foot/ankle complex.
- Footrest height should be set so that the thighs are well-supported by the seat cushion and the popliteal fossa is not in close contact with the seat cushion.

Trunk

- Assess full trunk mobility, tonal patterns, and strength of the abdominal muscles in supine.

- When assessing the trunk in sitting position, correct or accommodate the pelvis and hips first as much as possible as this will change the severity of spinal deformities and trunk support needed.
- Spinal curves need, at minimum, three points of contact to provide proper support.
- Deformities found in the thoracic spine always involve the ribs and may cause rib humps that can be accommodated within the back construct.
- Clients with low trunk tone may need full contact and support to improve posture.
- Clients with weak abdominals may present with increased lumbar lordosis and therefore need abdominal support not only for postural control, but also for maintaining skin integrity and enhancing breath support.

Upper Extremities

- ROM and strength in the shoulder, elbow, and wrist/hand help determine how a client will function most independently in a mobility device.
- If full upper extremity (UE) strength is present, the client may have the possibility to self-propel.
- With limited strength or active ROM, power assist or power mobility may need to be considered.
- A client with hemiparesis may be a candidate for a one-arm drive wheelchair.
- Clients who have cognitive ability but not the UE requirements to use a joystick may use alternate drive controls for a power chair.
- For clients with abnormal strength, control, or movement disorders, a tray/upper extremity support orthosis or more supportive arm rests should be considered as relative containment may improve the client's own volitional control.

Head and Neck

- Head and neck support should be determined when sitting only after the pelvis, hips, and trunk positioning has been optimized.
- Orient the client facing forward in supporting and positioning the head and neck.
- Weak trunk and neck musculature may require tilt to reduce the effects of gravity.
- When tilt and/or recline are needed for positioning, weight shifts, or for daily care (suctioning, tube feeding, diapering, or transfers), a head support is required when using these features.

MOBILITY AND SEATING PRESCRIPTION

The mobility base prescribed must meet the functional needs of the client and the documentation will describe various external needs, such as how it will be transported and used in their environment. The seating system

prescription will furnish the components needed for optimal postural alignment, maintenance of skin integrity, and how the seating system will position the client with the stability, functional independence, and ability to interact in the world at large. Accessories such as positioning straps, support accessories for extremities, and adjunct equipment to hold medical supplies will complete the mobility and seating prescription.

MOBILITY BASE CONSIDERATIONS

When prescribing the mobility base, it is best to start with asking overarching questions about the goals of mobility and then move toward narrower questions based on the function assessed during the mat examination and clinical trials.

The answers to these questions will help the team narrow down toward a particular category of wheelchair base. Figure 19.1 can help with the flow of questions to assist in narrowing down to a wheelchair category. The following are key features of each of the categories of wheelchairs or wheelchair functions.

Lightweight Manual Wheelchair

- Weighs 31 to 36 pounds
- Adjustable seat width, depth, and/or height[2]

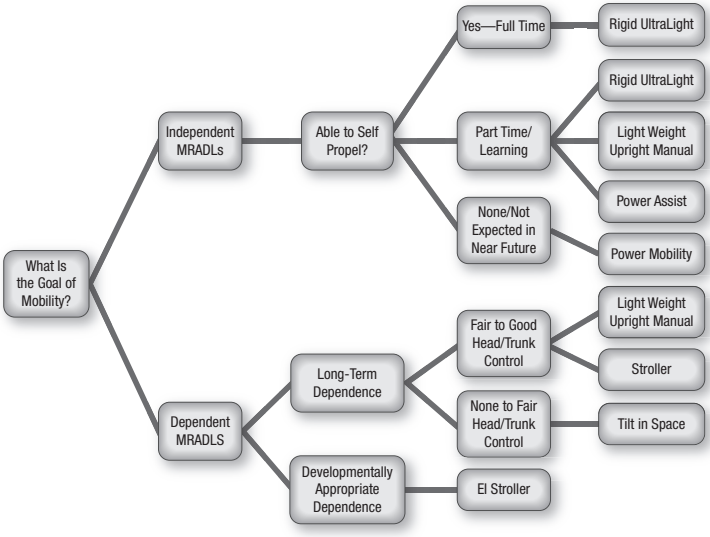

Figure 19.1 Flowchart to help guide selection of wheelchair category.

EI, early intervention; MRADL, mobility-related activities of daily living

- Cross frame that folds
- Used by the client at least 2 hours a day[2]

Ultralightweight Manual Wheelchair

- Fully customizable
- Primary mobility device
- Does not include tilt or recline features
- Defined by various sources as weighing under 25 to 30 pounds, and with advances in technology many on the market weighing under 20 pounds[3]
- Materials used to fabricate with high strength to weight ratios and thus more durable
- Can be rigid or folding
- Adjustable axle location, affecting the relative "tippiness" of the wheelchair relative to higher level function, such as navigation of curbs
- Growth potential for pediatric population

Tilt-in-Space Wheelchair

- *Tilt: defined* as change in seat angle orientation in relation to the ground while maintaining a constant seat-to-back angle and seat-to-leg rest angle[4]
- Greater than 25° of tilt needed to achieve pressure relief and/or allow tissue perfusion at the ischial tuberosities[4]
- *Tilt:* used for pressure relief, transfers, repositioning, added postural control, and to accommodate for limited endurance
- *Pivot tilt:* rotation of tilt a single point located behind and below the seat back and seat cushion
  - Causes distance of movement for the client relative to the wheelbase, which may trigger tonal response
  - Requires longer wheelbase for stability
- *Center-of-gravity tilt:* rotation of tilt sliding forward with increasing tilting position
  - Smaller distance of movement of client that stays within the wheelbase
  - Can have shorter wheelbase that allows for easier maneuverability

Early Intervention Stroller Bases

- Usually allow for both tilt and recline
- Have additional seating and position support
- Can hold medical equipment such as feeding pump, oxygen, vent, and so forth
- Hi-low base useful for activities of daily living, peer interaction, and feeding

Power Assist

- Measures the force of the propeller and amplifies the push through an electromechanical system[5]

- Two possible modes[5]
  - *Intermittent:* push amplified for each individual push
  - *Continuous:* allows for continuous movement with activation of switch or Bluetooth device
- Makes it easier for self-propellers to travel longer distances and on more difficult terrains[5]
- Decreases physical exertion and upper extremity pain and overuse injury[5]
- Must be trialed with the client
- *Power assist function located in the wheels:* may make the chair heavier to push if the power assist feature is not being used and may also affect how the chair is taken apart for transportation
- *Power assist function located as the fifth wheel:* may interfere with wheelchair skills such as wheelies

Power Mobility

Power bases are available in three different configurations: front-wheel, mid-wheel, and rear-wheel drive. Each drive wheelbase differs with respect to maneuverability, location of center of rotation, how it feels to the human driver, and different capabilities on various terrains.[6]

- Front-wheel drive
  - There is longer turning radius compared with mid-wheel drive, but usually shorter than rear-wheel drive.
  - The center of rotation at the front of the wheelchair base allows for turning on the inside corner and good indoor maneuverability.[7]
  - Clients with shorter seat depths may find this center of rotation more intuitive since their center of mass is over the center of rotation.[6]
  - With most of wheelchair base being behind the client, this makes it easier for them to pull up to counter or to be able to reach forward easier.
  - It is optimal for outdoor use due to the ability of the front wheels to pull the rest of the chair up and over obstacles from any angle and on softer terrains.[8]
- Mid-wheel drive
  - The mid-wheel drive has the smallest turning radius.
  - Equal parts of the chair are on either side of the axis of rotation.[7]
  - Clients with longer seat depths will sit over the drive wheels, and with the center of mass over the center of rotation driving can feel more intuitive.[8]
- Rear-wheel drive
  - The rear-wheel drive has the largest turning radius.[7]
  - The center of rotation is behind the driver.[7]
  - Older adults may find it intuitive since it is similar to driving a car.[8]
  - With the majority of the weight of the driver and chair being at the back of the wheelchair, it may be challenging to use certain

power features, especially tilt, and this can feel unbalanced for the driver.[8]

Any time a drive wheelbase is changed, there will be a learning curve for even the most experienced client.

SEATING CONSIDERATIONS

Findings from the mat examination that guide seating decisions include skin integrity and risk of skin breakdown, ability to perform pressure relief, posture, and endurance. When a client has a flexible posture, it is important that the pelvis is adjusted and supported in the most neutral alignment as possible to improve trunk and head alignment. If the client's posture presents with fixed deformities, the trunk and head alignment should be oriented to face forward as much as possible, allowing the pelvis to be accommodated and supported, even if this means that a windswept position of the legs is maintained.

Planar Seating

- Planar seating is essentially flat with no curves in the cushion or back.
- This is utilized when the client does not require much support or need for pressure relief and is not a full-time wheelchair user.

Contoured Seating

- Contoured seating has shaping built into the seating surface.
- This is for clients who spend most of their time in a seating system but may have no to mild postural deformities or support needs.
- Contoured seats can be customized without moving to a custom-molded seating system. This is vitally important in a growing child so modification can easily be made with growth.

Custom-Molded

- This is used mostly for moderate to severe postural or orthopedic deformities or for clients who need maximum support due to significant abnormal tone.
- This is very individualized and thus it is important to have a client or a caregiver that is aware of the proper way to position the client in the seating system and recognize that minimal growth modification or customization is possible after fabrication.

SEATING MATERIALS

Both the shape of the seating system and the materials used for construction can affect the client's seating experience. The inherent stability and pressure relief of the cushion materials must be matched with the client's needs to determine the best product or combination of

Table 19.1 Cushion Materials and Their Relation to Stability and Pressure Relief

CUSHION MATERIAL	STABILITY	PRESSURE RELIEF
Foam/plastics	Most	Least
Hybrids: foam with gel or air insert	Moderate	Moderate
Air	Least	Most

materials. The presence of tone or bony prominences plays a role in the comfort and the effectiveness of the material in meeting the client's goals. Table 19.1 compares the cushion materials and their relationship to seating stability and pressure relief.

ACCESSORY CONSIDERATIONS

In addition to the cushion and back support of a wheelchair, many clients will also require additional accessories to properly support and position them in their seating system.

Pelvis

- Pelvic belt
  - Comes with two or four attachment points
  - Placement of the belt at 60° or 90° to prevent pulling the client into posterior pelvic tilt
  - Strategic belt placement beneficial to patients with excessive tone
  - Many types of buckle options depending on the client's hand dexterity, including buckle guard when elopement is a risk
- Hip guides
  - Can be built into the seating or can be a secondary attachment
  - Used to support the pelvis and/or the trunk

Knees

- *Knee adductors:* used to help support the leg from significant abduction
- *Knee abductor:* used to help separate the knees in cases of significant hip adductor tone causing the knees to touch or cross

Lower Leg/Feet

- *Calf strap/panel:* helps support the lower legs from sliding off the footplate
- *Ankle huggers:* allow the feet some movement on the footplate but keep the feet from being kicked out or from falling off in all directions
- *Shoe holders:* provide sturdier support to the lower leg and feet on the footplates and may help in tone reduction

Trunk

- Trunk laterals
  - Can be built into the seating or as secondary support
  - Various widths, depths, and contour options
- Chest vests/abdominal support
  - Can have two to four attachment options
  - Made out of many types of materials depending on client needs
  - Various shape cutouts and buckle types depending on need

Head/Neck Support

- Should support available free-controlled movement when possible
- Should not obstruct the client's view

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Physical Modalities

20

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INTRODUCTION

Physical therapists utilize modalities in conjunction with therapeutic exercises, therapeutic activities, and gait training throughout a patient's plan of care. A modality is a therapeutic agent applied to a patient with the goal of improving pain, mobility, and function. The goal of this chapter is to introduce some of the more common modalities utilized in pediatric rehabilitation.

KINESIO TAPING

General Principles: What Is It?

Kinesio tape was developed originally by Dr. Kenzo Kase in 1973 with the thought of reducing pain.[1] After years of research, Kinesio Taping has been found to move lymphatic fluids, assist in correcting movement patterns, and change muscle tone and postural alignment.[2] The original Kinesio tape is a latex-free, hypoallergenic cotton fiber tape that is heat-activated. On average, the tape can stay on the skin for 3 to 5 days.[3]

Treatment Guiding Principles: Body Functions and Structures

Kinesio tape works by pulling on the skin to create a space between the layers of the skin and the muscles, allowing for increased lymphatic flow of fluid.[2] Various structures, including the epidermis, fascia, muscle, ligaments, and tendons, can be affected by the Kinesio tape depending on the direction of the placement of the tape, the level of tension, the type of tape, and the cut of the tape. There are six basic corrections to Kinesio Taping: mechanical, fascial, space, tendon/ligament, sensory stimulation (neurologic system), and circulatory/lymphatic.[2] Kinesio tape can also be used to increase support to joints while allowing for movement, stimulate weak muscles, and relax the soft tissues to improve lymphatic circulation.[3]

Indications

Kinesio tape can be used during all phases of injury, including acute, subacute, and chronic, and throughout the rehabilitative stages in adjunct to physical therapy.[1]

- Kinesio tape is beneficial in controlling pain, reducing edema, supporting joint structures, stimulating underactive muscles and relaxing overactive muscles, improving postural alignment, and reducing muscle fatigue.[3]
- Common conditions that benefit from taping include torticollis, cerebral palsy, orthopedic injuries and disorders, and pain control.

Patients should always be tested with a test patch to ensure there is no sensitivity to the adhesive. It can be used safely in most pediatric populations, including small infants. Kinesio Taping should be applied by a certified Kinesio taping practitioner.

Contraindications and Precautions

These include but are not limited to sensitivity to adhesives, fragile and thin skin (in newborns, tape with caution), active malignancy, excessive hair on skin, active cellulitis or infections, open/healing wounds, and deep vein thrombosis.

Tape with precaution when treating a patient with diabetes, kidney disease, congestive heart failure, lymphedema, respiratory conditions, and organ transplants, and those who are pregnant.

Functional Application

Kinesio tape is best used in conjunction with physical therapy. This tool allows for continuous input and stimulation for 3 to 5 days following therapy and can often be taught to patients/caregivers so they can apply the tape at home. The application of the tape should be reassessed and adjusted accordingly to maximize the benefits.

Kinesio tape has shown to improve range of motion (ROM) in patients with varying disorders, allowing for increased mobility.[4] It has also shown to improve postural alignment of the limbs and trunk in children with cerebral palsy, allowing for more efficient movement patterns for mobility and participation in activities of daily living.[5]

MANUAL THERAPY

General Principles: What Is It?

There are a variety of different manual techniques that physical therapists utilize to impact musculoskeletal structures with the goal of improving patient movement and reducing pain. Manual therapy techniques include both hand- and tool-assisted approaches.

Treatment Guiding Principles: Body Functions and Structures

Manual "hands-on" therapy targets soft tissue structures, that is, scar tissue, muscle, fascia, lymphatic channels, neural structures, and occasionally organic structures (abdominal massage). Additionally, it is utilized for

bony structures at specific joints through joint mobilization and manipulation.[6] A number of manual therapy techniques are available for physical therapists:

- *High-velocity thrust versus low-velocity nonthrust:* A manipulation or a thrust technique involves high-velocity, low-amplitude forces to induce cavitation. A nonthrust technique has lower velocity, not necessarily anticipating joint cavitation. The Maitland technique categorizes the intensity of mobilizations into five different grades, progressing from pain relief to manipulation.[7]
- *Kaltenborn technique:* This technique focuses on the kinematics of the joint being mobilized and how the convex surface of the joint rolls or glides on the concave surface. It is classified into three grades of capsule stretch: (a) loosening, (b) tightening, and (c) stretching.[8]
- *McKenzie technique:* This technique involves a patient-driven series of gentle repeated movements in the direction of pain relief, with the goal of enhancing self-management throughout the week.[9]
- *Instrument-assisted soft tissue mobilization (IASTM):* This is a contoured stainless-steel tool that allows for deeper penetration to target soft tissue disorders by creating microtrauma, breaking down scar tissue, and mobilizing the fascia, facilitating a cascade of healing.[10]
- *Cupping:* This is a healing modality, traditionally applied in Asia, where the cups are applied to a prescribed area. They create a partial vacuum, improving microcirculation and relieving muscle spasm.[11]

Indications

- Indications include reducing pain and swelling/edema, increasing mobility of the scar tissue, fascia, joints, and muscles, and increasing circulation.

Contraindications and Precautions

These include avoiding malignancy sites and acutely inflamed joints. Contraindications for joint mobilizations include osteoporosis, spondylolysis, spondylolisthesis, and fractures.

Functional Application

Manual therapy is used in adjunct to other physical therapy treatment techniques to facilitate mobility, flexibility, and ROM, and reduce pain to maximize the potential gains made in therapy and return to prior level of function.

DRY NEEDLING

General Principles: What Is It?

Dry needling is a procedure commonly used by Western-based, healthcare professionals for treatment of neuromusculoskeletal conditions.[12] It involves the insertion of thin monofilament needles into target tissues without the use of an injectate.[13]

Treatment Guiding Principles: Body Functions and Structures

- Physiologic systems affected by dry needling include the muscles, ligaments, tendons, subcutaneous fascia, scar tissue, peripheral nerves, and neurovascular bundles "sensitive bundles" for management of a variety of pain syndromes.[13]
- Goals include relaxation of trigger points, improvement of blood flow, facilitation of healing, realignment of collagen, and reversing the energy deficit that causes trigger points.[13]
- *How it works:* Trigger points are a continuous cycle of localized muscle contraction or muscle overuse and hypertonicity that blocks blood flow, resulting in a shortage of oxygen and nutrients, ischemia, hypoxia, and pain and inflammation.[14] Trigger points are associated with a chronic pain cycle and lead to weakness and restricted ROM. The twitch response occurs by insertion of the needle, followed by pistoning, coning, or fanning to provide a high-pressure mechanical stimulation to sensitive loci, ultimately breaking the trigger point "circuit."[15]

Spinal and neural pathways are targeted through protocols that work by activation of the descending pain modulatory system, resulting in increased opioids, serotonin, and norepinephrine in the spine and decreased norepinephrine in the brain.[16] Providers treat the spine and not just the trigger points.

Indications

- Indications include pain, trigger points, weakness, restricted ROM due to muscle hypertonicity, referred pain, and prolonged healing due to lack of blood supply.

Contraindications and Precautions

- *Absolute contraindications* include needle phobia, no signed/informed consent, patients on high doses of anticoagulant therapy, patients with postsurgical lymphedema distal to the site of lymph node removal, or patients with inability to remain still.
- *Relative contraindications* include bleeding disorders, compromised immune system, vascular disease, diabetes, pregnancy, epilepsy (recent seizure), and possible allergy to the metal found in the needle.

Take precaution using electrotherapy with dry needling in patients who are pregnant or across a pacemaker. Precautions for dry needling with electrical stimulation are similar to precautions for surface-level electrical stimulation as discussed in the Electrical Stimulation section. Depending on the target tissue and patient tolerance, dry needling practitioners will determine the necessary clinical dosing required to successfully treat the neuromusculoskeletal condition. Dosing may be adjusted by the following: number of needles, needle size, location, depth,

mechanical stimulation, electrical stimulation frequency/intensity/duration, and duration of the needles left in situ.[12]

Electrical stimulation and dry needling: Evidence suggests that electrical dry needling results in inhibition of pain, breaking the cycle of chronicity and achieving better analgesia.[17] Stimulation increases blood flow through vasodilation, which contributes to tissue healing and increased opioid-producing immune cells required to reduce inflammatory cytokines.[12]

Functional Application

Acute or chronic pain results in a decrease in functional mobility and quality of life. By reducing pain and improving ROM with use of dry needling followed by therapeutic exercise, practitioners facilitate improved participation with peers. Examples include neck pain or headaches, difficulty with prolonged sitting or concentration, and difficulty with work and school.

ELECTRICAL STIMULATION

General Principles: What Is It?

Electrical stimulation is the application of electrical currents to the body through the skin to achieve a therapeutic goal. In pediatric rehabilitation, this treatment modality is utilized in conjunction with more traditional treatment approaches, including therapeutic exercise, functional activity, and gait training.

Treatment Guiding Principles: Body Functions and Structures

Electrical stimulation produces muscle contractions by stimulating the muscle directly or, more commonly, the peripheral nerve that innervates the target muscle.

- *Direct stimulation of the muscle:* This is typically only performed when the peripheral nerve is not intact. This type of stimulation is often uncomfortable and has a higher risk of burns at the application site.[18]
- *Stimulation of the peripheral nerve:* This tends to be more comfortable and better tolerated. Muscle contraction occurs by applying an electrical current strong enough to depolarize the motor nerve.[18]
    - Electrically stimulated contractions tend to activate all the motor units in the sections of muscles that are being stimulated and tend to activate more easily fatigued fast-twitch muscle fibers first. Patients should be monitored for muscle fatigue during a program eliciting electrically stimulated contractions.[18]

Indications

- Electrical stimulation helps improve neuromuscular deficits, including muscle weakness, spasticity, contractures or deformity, upper extremity dysfunction, gait deficits including foot drop, and poor trunk control.

It also helps manage deconditioning, osteopenia or osteoporosis, and dysphagia.[19]
- Electrical stimulation assists in the rehabilitation after orthopedic injuries to reduce pain and improve muscle recruitment after injury or surgical intervention.[20]

Electrical stimulation may also be used in more medically complex patients, including patients in the ICU with prolonged immobilization and patients with chronic medical conditions to help improve their general condition and strength and prevent muscle atrophy.[20]

Contraindications and Precautions

Electrical stimulation is a relatively low-risk treatment modality that is beneficial to pediatric patients of various ages, abilities, and diagnoses. Adverse reactions are not common with application of electrical stimulation; however, it is important for physicians to perform a thorough review of the patient's medical history to determine if the treatment is contraindicated or if precautions should be taken before and during treatment. Additionally, the treating clinician should carefully monitor the patient throughout the application to assess their response to treatment.[18]

- *Contraindications include placement over cardiac pacemakers or other electronic devices, placement over the anterior or lateral neck near the carotid sinus, placement in areas with active thrombosis or thrombophlebitis, placement over the abdomen or low back of women who are pregnant, placement over areas of malignancy, and placement over reproductive organs.*[18,21]

Precautions include use in patients with impaired sensation or impaired cognition who may not be able to articulate pain or discomfort, use in patients with cardiac disease, and use on areas of the skin that are irritated or have open wounds.[18,21]

Functional Application

Electrodes are placed on the skin over the muscle or muscle groups that elicit the target movement. Electrodes are then connected to leads that connect to the stimulator. Stimulators may be small, portable units or larger mobile devices. Treatment parameters are selected to achieve desired muscular contraction and include the following[18]:

- *Amplitude or intensity:* power of the current
- *Frequency or rate:* how often the current flows
- *Duration or pulse width:* how long the current flows
- *On time:* time that the current will flow, contraction phase
- *Off time:* time when the current is inactive, relaxation phase

- *Ramp time:* how long it takes to reach the peak intensity each on time
- *Treatment time:* how long the patient will receive the stimulation

Once appropriate treatment parameters are selected, electrical stimulation can be performed in the clinic or as a home exercise for families trained in using the device.[18] Studies indicate that electrical stimulation has a greater ability to impact a patient's function when it is paired with activities that are important to the patient.[20]

Many clinics and clinicians now have access to functional electrical stimulation devices that have preprogrammed functional activities that time muscle stimulation with the appropriate muscle activation pattern to complete a functional task. These activities include but are not limited to cycling with the arms or the legs, sit-to-stand transitions, bringing hand to mouth for feeding, reaching for objects of interest, releasing an object from grasp, and bridging.

Electrical stimulation can also be applied and used as an orthotic to improve foot drop during gait.

HEAT AND COLD

General Principles: What Is It?

Heat and cold are used primarily to shorten muscle recovery time post exercise and provide short-term pain relief. Although often used, there is minimal to moderate supporting evidence that heat or cold aids muscle recovery. Stronger evidence exists for use of heat over cold for pain relief.

Treatment Guiding Principles: Body Functions and Structures

The mechanism underlying the use of heat and cold is depicted well by Malanga et al. (Figure 20.1).[22]

Indications

- *Cold:* used to manage inflammatory processes and muscle soreness in acute injuries
- *Heat:* used to manage joint pain and stiffness after the acute phase has passed

Contraindications and Precautions

Application of cold should be avoided over regenerative nerves, impaired sensation, impaired circulation, poor cognition, the anterior neck, and near the site of a deep vein thrombosis. Application of heat should be avoided over an infected tissue, over large body areas that could raise core

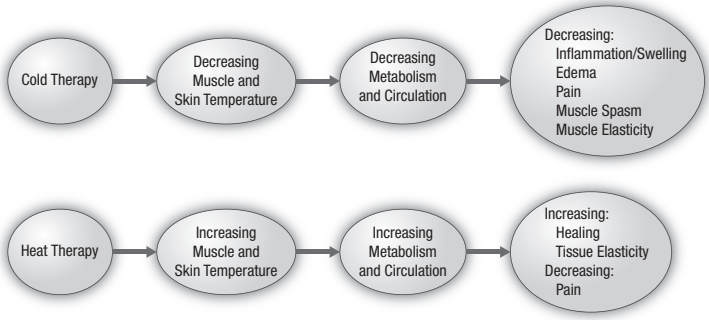

Figure 20.1 Heat and cold therapies.

Source: Adapted from Malanga GA, Yan N, Stark J. Mechanisms and efficacy of heat and cold therapies for musculoskeletal injury. *Postgrad Med.* 2014;127(1):57–65.

temperature, and near the site of a deep vein thrombosis, as well as in patients with suspected malignancy, impaired sensation, and poor cognition.

Functional Application

Cold modalities can be applied via cold water immersion, ice packs, cold gel, ice massages, and cold compression units. Heat modalities can be applied via heat wraps, hot water bottles, ultrasound, diathermy, infrared lamps, and hot baths.[22]

ROBOTIC-AIDED THERAPIES

General Principles: What Is It?

The use of robotics technology in upper and lower limb rehabilitation represents an important opportunity to help people affected by different pathologies. Robot-aided neurorehabilitation devices have an approach similar to a video game, which is appealing to the pediatric population. The goal of this section is to present a number of pieces of rehabilitation technology available to rehabilitation practitioners.

Robotic-Assisted Gait: Exoskeletons

Robotic-assisted overground walking devices (exoskeletons) are different from stationary walking devices and require higher cognition and cardiovascular effort.[23] Exoskeletons require a walker or crutches for the vast majority of users and only result in walking speeds of .3 m/sec, while the average walking speed of a sixth-grade line leader is 1.4 m/sec. In addition, walking distances with an exoskeleton are typically much less than the expectations for community independence.[24,25]

Treatment Guiding Principles: Body Functions and Structures
Exoskeletons are devices used to support impaired body functions and allow limited overground ambulation.

Indications
Exoskeletons are currently being used in patients with spinal cord injury, hemiparesis, and coordination disorders.

Contraindications and Precautions
Exoskeletons should not be used with severe lower extremity contractures, unhealed lower limb skin lesions, surgery within the past 3 months, and severe intellectual impairment resulting in inability to understand verbal instruction.

Functional Application
Exoskeletons are used for limited household mobility with use of walker or crutches.

Robotic-Assisted Gait Training
A robotic-assisted gait training (RAGT) device (e.g., Lokomat) delivers a series of step cycles to a patient's lower limbs while aiding with control on the sagittal plane. The goal of the system is to restore functional walking and improve locomotor ability by increasing selective motor control, walking further distances, improving gross motor skills, and reducing joint contractures.[26]

Indications
These include but are not limited to stroke, hemiparesis, spinal cord paraplegia, cerebral palsy, and abnormal gait patterns.

Contraindications and Precautions
These include high degree of cognitive deficit, recent orthopedic surgery, severe joint contractures, and high degree of osteopenia.

Functional Application
Activity- and participation-based RAGT requires highly repetitive guided movements and for patients to remain cognitively engaged in the activity in order to see substantial improvement. Promising evidence shows that RAGT induces changes in brain plasticity, thus improving the gross motor control of children with cerebral palsy, typically a higher degree in those who actively participated.[27] The benefits of improving standing and walking may include cardiovascular fitness, functional gains, and greater involvement in social roles.[27] There is strong evidence for improved functional outcomes following RAGT in adults with spinal cord injury, but

limited evidence in pediatrics outside of cerebral palsy and conflicting evidence between RAGT and bodyweight-supported treadmill training/traditional functional treatment.[28] The intensity and duration of the treatment plan of care have not yet been established and more evidence is needed.

Robotic-Assisted Bodyweight-Supported Gait Training Devices

Electromechanically assisted bodyweight-supported gait and balance training devices (e.g., Hocoma Andago) support a patient during overground ambulation. The system senses the movement of a patient through the harness straps and automatically follows the patient's movement; the system can also be steered by the therapist.[29]

Indications

Appropriate patients are similar to that for RAGT, but the system may function better in patients with more volitional lower extremity control.

Contraindications and Precautions

These include bone fragility, unstable orthopedic conditions/surgery, lack of head control, skin lesions, uncontrolled seizures, sensory impairments in lower extremities, and any medical condition preventing active use of these systems.

Functional Application

The system serves as a device to bridge the gap between robot-assisted therapy, such as with RAGT, and conventional overground gait training using an assistive device or assistance of a therapist. It is useful in patients who are already able to initiate steps, unlike RAGT which can provide 100% assistance in stepping.[29]

VIRTUAL REALITY

General Principles: What Is It?

Virtual reality (VR) refers to participation in a virtual world through a head-mounted display or a head mount with hand- and foot-mounted trackers to interact with objects in the virtual environment. VR is used for remapping somatosensory processing and has demonstrated significant positive outcomes in pain mitigation and restored movement.[30,31]

Treatment Guiding Principles: Body Functions and Structures

VR offers intense repetitions of meaningful tasks necessary for rehabilitation.[31]

Indications
VR is used as a distraction during procedures that cause pain and/or anxiety. It is also used to aid in chronic pain, neurorehabilitation, cerebral palsy, hemiparesis, brachial plexus injuries, and other conditions.

Contraindications and Precautions
VR may cause visually induced motion sickness and collisions with nearby objects, risk social isolation, and can give false memories in children.

Functional Application
Overall, VR provides an effective distraction therapy and a more rapid gain in motor acquisition versus conventional therapies.[32]

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Management of Sialorrhea

21

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GENERAL PRINCIPLES

Sialorrhea is a common finding in children with congenital or acquired neurologic disorders. Its etiology is multifactorial and requires a multidisciplinary team approach, with the goal of reducing saliva in order to maintain a moist, healthy oral cavity. The stepwise, case-based approach from noninvasive to invasive modalities is the usual practice. In this chapter, the pathophysiology, clinical findings, and the medical and therapeutic interventions for pediatric sialorrhea are discussed, along with a case study presentation.

Definition

Sialorrhea, defined as involuntary escape of saliva from the mouth beyond the margin of the lip due to limitations in a person's ability to control and swallow oral secretions, is a common finding in children with congenital or acquired disabilities. Early identification and intervention by a multidisciplinary team may improve the health outcomes of children with sialorrhea. Clinically there are two presentations of sialorrhea:

- Anterior drooling is when the unintentional loss of saliva from the mouth is clearly visible. It is considered normal up to 18 to 24 months of age and usually resolves by 5 years of age.[1]
- Posterior drooling occurs when saliva spills over the tongue through the faucial isthmus of the oropharynx and into the hypopharynx.[2]

Saliva in humans is produced by exocrine glands with ducts that are located throughout the oral cavity and in front of the ears in the face.

Saliva aids in dental hygiene, swallowing, and speech production,[1] and contains:

- Water
- Electrolytes
- Mucus
- Antibacterial compounds

Three major glands produce 90% of saliva: the parotid, submandibular, and sublingual glands. At rest, the submandibular glands secrete majority of the saliva, with 60% of the total, the parotids with 25%, and with the

sublinguals and the minor glands each secreting 7% to 8% of the total. However, with tactile, olfactory, or gustatory stimulation, there is a fivefold increase in total saliva, mostly produced by increased parotid secretion, to 50% of the total.[3] While the normal rate of saliva secretion in adults is 1.3 L/d (1 mL/min), the normal amount in children is .5 to .6 L/d (.5 mL/min). Based on research by Jongerius et al.[2] and Erasmus et al.[4] in 3- to 7-year-old children with cerebral palsy, this rate is essentially unchanged at .4 mL/min.

The following are the functions of saliva:

- The saliva protects the mucosa and the oropharyngeal tissues from infection and ulceration since its mucin, lipid, immunoglobulin, lysozyme, lactoferrin, and peroxidase content serves as antibacterial, antiviral, and antifungal agents.
- The mucus is a lubricant and as such aids in bolus formation and swallowing, as well as in speech production.
- The serous part of the saliva has alpha-amylase, which facilitates digestion, while calcium, phosphate, and bicarbonate keep the teeth healthy.

Etiology

With a high prevalence, sialorrhea is an important comorbidity of neurologic disorders in all age groups.

In adults:

- One in two patients with motor neuron disease amyotrophic lateral sclerosis (ALS) have it and one in five need continuous management.
- In Parkinson disease, the rate is 70%.[1]

In pediatrics:

- Sialorrhea is seen in 10% to 80% of children with cerebral palsy (40%).[5]
- The American Academy for Cerebral Palsy and Developmental Medicine reports approximately 40% of children and youth with cerebral palsy and 58% of school-age children with cerebral palsy have sialorrhea.

Pathophysiology

Current knowledge on the pathophysiology of sialorrhea shows the cause is multifactorial, with the most common reason found to be impaired oral motor control. It is less commonly due to excess production of saliva. The exception to this is dyskinetic cerebral palsy, which causes increased salivary flow due to hyperkinetic oral movement.[4] Table 21.1 outlines the impairments that may lead to increased sialorrhea.

In 2012, Reid published a population-based study of children with cerebral palsy and provided guidelines for assessment of sialorrhea in this cohort.[5] Significant correlating factors included:

- Gross Motor Function Classification System (GMFCS) level
- Topographical pattern

Table 21.1 Impairments That Lead to Increased Sialorrhea

DYSPHAGIA	DENTAL ISSUES	MEDICATIONS	OTHER FACTORS
Jaw/lip closure	Malocclusion	Antiepileptics	Sensory and cognitive function
Chewing	Oral inflammation	Tranquilizers	Nasal obstruction
Swallowing	Teething		Decreased head/trunk control
	Dental caries		Gastroesophageal reflux
	Tonsillitis		
	Abscess		

- Head posture
- Epilepsy
- Intellectual disability
- Oromotor function
- Eating/speech difficulties

DIAGNOSIS

Risk Factors

The importance of managing impaired saliva flow or secretion control lies in the potential complications it may cause if left untreated. The main concern with untreated posterior drooling is chronic aspiration, which may result in recurrent respiratory infections and progressive lung disease. Other negative outcomes of anterior and posterior drooling include:

- Skin breakdown
- Maceration causing infection
- Dehydration
- Failure to thrive
- Poor dental hygiene
- Foul odor
- Speech disturbance
- Psychosocial burden
- Isolation
- Low self-esteem
- Dependency
- Reduced quality of life

EXHIBIT 21.1 THE DROOLING IMPACT SCALE

OVER THE PAST WEEK

1. How frequently did your child dribble?

Not at all 1 2 3 4 5 6 7 8 9 10 Constantly

2. How severe was the drooling?

Remained dry 1 2 3 4 5 6 7 8 9 10 Profuse

3. How many times a day did you have to change bibs or clothing due to drooling?

Once or not at all 1 2 3 4 5 6 7 8 9 10 10 or more

4. How offensive was the smell of the saliva on your child?

Not offensive 1 2 3 4 5 6 7 8 9 10 Very offensive

5. How much skin irritation has your child had due to drooling?

None 1 2 3 4 5 6 7 8 9 10 Severe rash

6. How frequently did your child's mouth need wiping?

Not at all 1 2 3 4 5 6 7 8 9 10 All the time

7. How embarrassed did your child seem to be about his/her dribbling?

Not at all 1 2 3 4 5 6 7 8 9 10 Very embarrassed

8. How much do you have to wipe or clean saliva from household items, e.g. toys, furniture, computers?

Not at all 1 2 3 4 5 6 7 8 9 10 All the time

9. To what extent did your child's drooling affect his or her life?

Not at all 1 2 3 4 5 6 7 8 9 10 Greatly

10. To what extent did your child's dribbling affect you and your family's life?

Not at all 1 2 3 4 5 6 7 8 9 10 Greatly

Source: Reproduced with permission from Reid SM, Johnson HM, Reddihough DS. The Drooling Impact Scale: A measure of the impact of drooling in children with developmental disabilities. *Dev Med Child Neurol*. 2010;52(2):e23–e28.

Diagnostic Evaluation

Objective and subjective measurements can be used to assess sialorrhea. Changes in drooling and its impact on patients and caregivers can be measured at assessment, reassessment, and discharge using the Drooling Impact Scale.[6]

- The Drooling Impact Scale (Exhibit 21.1) was devised to evaluate longitudinal changes in the impact of drooling in children with neurologic disorders and to quantify the short- to medium-term treatment benefits of interventions, which can help assess satisfaction with the outcome of intervention.
- The scale has been shown to be a reliable and subjective measure that is responsive to change and can reflect the impact on quality of life,

such as decreasing the demands on the caregivers (wiping, bib or clothing changes, frequent suctioning), improving caregivers' perception of their child's needs and appearance, and for some children improved social interaction and self-esteem.

Radiographic Assessment

Instrumental assessment may be recommended to further assess swallow function and posterior secretions.

- A videofluoroscopic swallow study (VFSS) can assess oral and pharyngeal functioning to determine the safety of oral intake and the effectiveness of therapeutic strategies.
- A flexible endoscopic evaluation of swallowing (FEES) can assess and describe posterior drooling. The Secretion Severity Rating Scale[7] may be conducted at FEES assessments to further define the severity of posterior drooling and help track progress of swallow function and saliva management with intervention.

TREATMENT

Ongoing Care

A stepwise, individualized approach is used in the treatment of sialorrhea, with the overall goal of reducing saliva. Table 21.2 offers a summary of medical versus therapeutic intervention options for sialorrhea.

A noninvasive approach is preferred, with more invasive options being considered if the child does not adequately respond to less invasive interventions. Figure 21.1 illustrates the least invasive to the most invasive intervention options.

Pharmacologic options including systemic medications administered orally or through a feeding tube are an appropriate step after a noninvasive therapy.[8] Pharmacologic interventions offer the advantage of a relatively quick onset and peak effect:

- Anticholinergic agents such as glycopyrrolate (Cuvposa, Robinul)
- Transdermal scopolamine

Table 21.2 Medical Versus Therapeutic Intervention Options for Sialorrhea

MEDICAL INTERVENTION OPTIONS	THERAPEUTIC INTERVENTION OPTIONS
Pharmacologic	Oral motor stimulation
Oral appliances	Oral motor exercises
Neurotoxin injections	Therapeutic taping
Surgery	Neuromuscular electrical stimulation
	Behavioral therapy

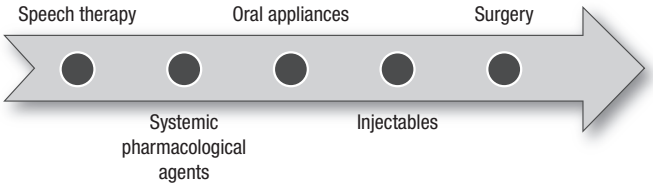

Figure 21.1 Least to most invasive intervention options.

- Trihexyphenidyl, also known as benzhexol (Artane)
- Atropine 1% drop administered sublingually[9,10]

Important reminders:

- Individual dose variation
- Potential side effects due to their nonselective nature
  - Unfortunately, one third of the patients discontinue due to dry mouth, urinary retention, blurred vision, skin flushing, decreased sweating, impaired temperature regulation/fever, sedation, irritability, constipation, and epilepsy.

Oral appliances can be used in a selected group of children with intact cognition when funding is not an issue. Dentists can be consulted, when needed, to assess the patient for an oral prosthetic device. Customized devices are used for individual patients and need patient cooperation. The Innsbruck Sensorimotor Activator and Regulator (ISMAR) worn in short periods daily and then overnight is used in some children with cerebral palsy.[11]

Oral prosthetic devices serve to achieve:

- Oral awareness
- Mandibular stability
- Better lip closure
- Tongue position
- Swallowing

Neurotoxin injections into the salivary glands is the next step before surgery. When patients are refractory to all therapy and pharmacologic interventions, botulinum toxin (BoNT) injection is a reasonable option. There are different types of BoNT injected in pediatric and adult patients that provide dose-dependent reduction in the rate of secretions. The dosage is based on the weight of the child and the number of glands injected, as well as the severity. An international consensus statement in 2010 concluded that the BoNT-A range for submandibular and parotid glands was 10 to 50 U for Ona, 15 to 75 U for Abo, and 250 to 1,000 U for Rima B. More recently, in December 2020, incobotulinum toxin was approved by the Food and Drug Administration for chronic sialorrhea in pediatric patients 2 years and older (>12 kg). The recommended dose is based on body weight, administered in a 3:2 dose ratio into the parotid and submandibular

glands, respectively, no sooner than every 16 weeks; ultrasound guidance is recommended. When injecting BoNT, trained physicians use guided technique to avoid potential side effects such as dysphagia, weakness in the jaw, or respiratory illness.

Intractable cases are referred to ENT (ear, nose, and throat) for submandibular gland excision and/or parotid duct ligation.

A variety of speech therapy approaches can be used to improve outcomes in this patient population, including:

- Oral motor stimulation
- Oral motor exercises
- Therapeutic taping
- Neuromuscular electrical stimulation (NMES)
- Behavioral therapy

Before the intervention can begin, proper positioning and adequate oral care should be addressed. It is vital that the child is in a stable, secure position to support muscle function and breathing. Studies have demonstrated that proper biomechanical alignment is associated with improved swallowing and feeding.[12] Training caregivers on proper oral care strategies can help prevent respiratory illnesses including pneumonia.

Oral stimulation can improve:

- Oral control
- Sensory awareness
- Frequency of swallowing

Oral stimulation can be provided via:

- Icing
- Brushing
- Vibration
- Oral manipulation

Oral motor exercises can improve:

- Jaw stability
- Lip closure
- Strength of the oral musculature

It is imperative both the motor and sensory systems are targeted to obtain an appropriate response for a desired task. Providing tactile, olfactory, or gustatory stimulation results in an increase in saliva, which helps increase bolus volume and can reduce pharyngeal delay and oral transit time.[13]

Therapeutic taping has been shown to improve sensory awareness and motor movement. For example, taping can improve jaw stability and movement of the orbicularis oris, thereby reducing drooling. Pervez et al.[14] and Mederios et al.[15] report improved drooling with use of therapeutic taping.

NMES refers to the use of electrical stimulation to activate the muscles through stimulation of the intact peripheral motor nerves. The major

treatment goals are to strengthen weak muscles and help in the recovery of motor control.[16] A case series by Rice[17] suggests that NMES is an effective intervention in a child diagnosed with pharyngeal phase dysphagia.

Bavikatte et al.[1] describe the use of behavioral therapy to address drooling by increasing awareness of the mouth and drooling and increasing the frequency of wiping and/or swallowing.

ADDITIONAL CONSIDERATIONS

CASE STUDY 21.1:A PEDIATRIC PATIENT WITH VANISHING WHITE MATTER DISEASE

History

A 6-year-old boy with vanishing white matter disease (EIF2B5 mutation) presenting with spastic quadriplegia, dysphagia, and intellectual disability was seen at the Children's Health Saliva Management Clinic by a pediatric rehabilitation medicine specialist and speech pathologist.

Diagnostic Evaluation

An FEES was performed with a Secretion Severity Rating Scale score of 2 reported, indicating deeply pooled secretions. The patient's mother reported he needed 10 bib changes a day. The patient was receiving home health feeding therapy; however, the therapist felt progress was limited. No medication for sialorrhea was recommended. Instead, the patient was referred to the Saliva Management Outpatient Group Therapy program and was seen two times per /week for a total of 27 therapy sessions. The mother completed the Drooling Impact Scale at the initial session and a total score of 82 was recorded.

Treatment

Therapy included NMES with pharyngeal and facial placements, therapeutic taping to the orbicularis oris, oral care, oral stimulation, labial and buccal oral stretches, sensory stimulation with cold and sour tastes by mouth (PO), and home exercise program. The mother completed the Drooling Impact Scale at the conclusion of the program with a total score of 62 recorded. The patient was discharged from the therapy program and was referred to home health therapy for continued therapeutic intervention. The home health speech pathologist was provided with a treatment plan based on the patient's progress and the mother was provided with a detailed home exercise program to help the patient carry over his skills.

Outcome

The patient was seen back at the Saliva Management Clinic 7 months later for a follow-up. A Drooling Impact Scale was completed by the mother

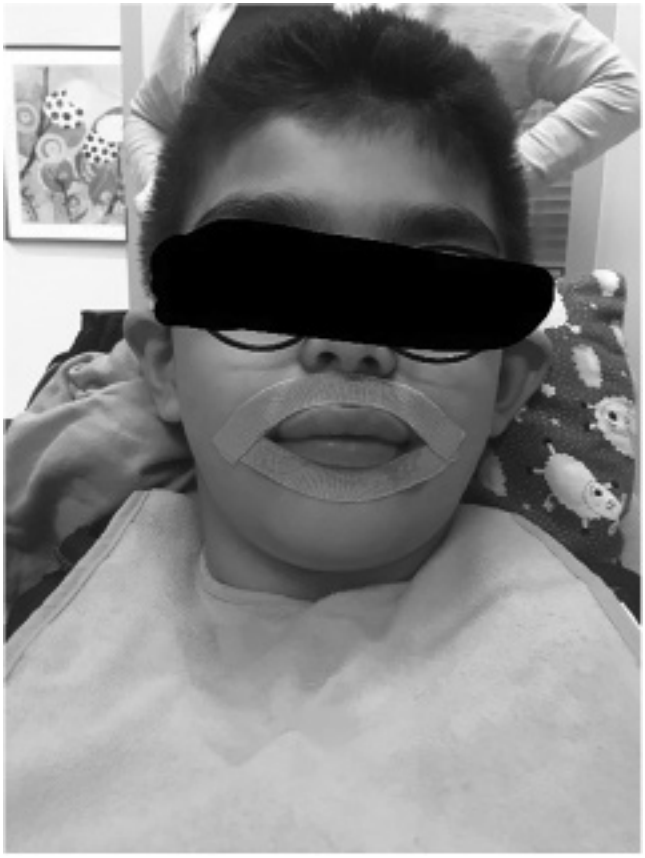

Figure 21.2 A patient with vanishing white matter disease.

with a total score of 53 reported. In addition, the mother reported the patient needed five bib changes a day, which was a 50% decrease from the initial consult (Figure 21.2).

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Speech Disorders 22

MARY FINK, LINDSEY WEISSGARBER, and PATTI WREN

INTRODUCTION

Communication is a process by which information is exchanged between individuals through a common system of symbols, signs, or behaviors.[1] Communication can take a variety of forms, including speaking, writing, and gesturing. An interference with the ability to perform any of these tasks can significantly impact overall communication skills. Oral motor and neurologic impairments can also affect swallowing function.

Speech disorders typically refer to the motor output for production of speech sounds. Language disorders refer to the process by which we encode and process meaning within messages.

APHASIA

Aphasia is an acquired neurogenic language disorder that results from injury to the brain, most often the left hemisphere. Aphasia involves varying degrees of impairment in four primary areas:

- Spoken language expression
- Spoken language comprehension
- Written expression
- Reading comprehension

One of the most common classification systems is based on the pattern of impaired language abilities, where aphasia is categorized as either fluent or nonfluent based on the characteristics of spoken language expression.[2,3] A person with aphasia often experiences both receptive and expressive language difficulties.

There are four common types of aphasia: Broca aphasia is characterized by difficulty finding and saying the right word. The clinical presentation would be severely reduced speech, often limited to short utterances of less than four words, limited vocabulary, clumsy formation of sounds, and difficulty writing. Wernicke aphasia or receptive aphasia is when someone can speak well and use long sentences, but may not make sense. There typically is no awareness that they are using the wrong word and there is often frustration associated with communication attempts. The clinical features of Wernicke aphasia include impaired reading and writing, inability to grasp the meaning of spoken words, inability to produce

sentences that go together, and intrusion of irrelevant words. Anomic aphasia is characterized by an inability to supple the words for things the person wants to talk about, particularly nouns and verbs. Speech is full of vagueness and there is also difficulty finding words in writing as well as in speech. The final type of aphasia is global aphasia, which is both an expressive and a receptive language disorder. The person has deficits in understanding others and in their ability to express themselves.

Aphasia is caused by damage to the language center of the brain, which in most people is on the left hemisphere. Damage to the brain can result from stroke, traumatic brain injury, brain tumors, brain surgery, brain infections, and progressive neurologic conditions.

The speech language pathologist (SLP) is responsible for assessing, diagnosing, and treating children with aphasia. There are many treatment options for individuals with aphasia. Augmentative and alternative communication supplements natural communication modalities with communication boards, signing and/or gesturing, and aided alternative augmentative communication (AAC) devices.

Cognitive-Communication Deficits/Difficulties

A patient may be treated for cognitive-communication deficits following a traumatic brain injury, stroke, encephalitis, brain bleed, or a variety of other diagnoses impacting brain health and function. The location of impact in the brain guides the assessment and treatment principles. Cognitive-communication disorder refers to difficulty communicating wants, needs, or ideas following an injury to the brain, resulting in disordered thinking.[4] Cognitive-communication difficulties can affect a child's ability to participate in a school setting and peer relationships in a social setting, carry out structured and unstructured tasks, plan for the future, and recall novel information, along with a variety of other struggles.[5] It is the role of the SLP, along with a neuropsychologist, to assess their communication needs and make appropriate recommendations for return to a functional setting. Cognitive-communication deficits include:

- Attention
- Awareness of deficits
- Executive function
- Expressive/receptive language
- Memory/recall
- Organization/reasoning
- Problem-solving
- Visual/spatial memory

Initial Management and Assessment

Patients may be in varying stages of recovery and are categorized according to the Rancho Los Amigos Level of Cognitive Functioning Scale.[6] This specific recovery scale takes into account the patient's current state and need for assistance for optimal cognitive function.[7]

Ongoing Care and Treatment

The severity of a patient's brain injury and their Rancho Los Amigos level will determine the appropriate treatment techniques. The goal of therapy will be to determine a functional communication method (low-tech AAC, high-tech AAC, verbal communication, gestures/signs, etc.) for the patient to express their wants and needs, support their return to school, work, and community environment, generalize new skills, and restore lost skills or abilities. External, compensatory strategies can be taught, and a variety of cognitive exercises are practiced to assist in attention, memory, executive function, and other areas of deficit.[4] In the pediatric setting, caregiver education and training are vital given the individual's need for assistance to carry out daily tasks and to monitor one's own safety needs and behavior. Recommendations are often made for classroom modifications, such as seating placement to minimize distractions, a visual schedule to improve planning and organization throughout the day, peer note-taker for recalling novel information, and consistent checks for comprehension of a new material, among other modifications, to ensure optimal return to a school-based setting.[8]

Treatment Controversies

- Compensatory strategies versus restorative therapy versus habilitative approaches
- Gaps in care from medical management to academic management[9]

Additional Considerations

- Baseline cognitive-communication abilities and developmental status
- Caregiver education/training and buy-in
- Respiratory (trach/vent) complications
- School re-entry

DYSARTHRIA

Dysarthria refers to a group of neurogenic speech disorders characterized by "abnormalities in the strength, speed, range, steadiness, tone, or accuracy of movement required for breathing, phonatory, resonatory, articulatory, or prosodic aspects of speech production."[10] There are many types of dysarthria identified by perceptual attributes and their pathophysiology.

- *Flaccid:* associated with disorders of the lower motor neuron system and/or muscle
- *Spastic:* associated with bilateral disorders of the upper motor neuron system
- *Ataxia:* associated with disorders of the cerebellar control circuit
- *Mixed:* various combinations of types of dysarthria (e.g., spastic-ataxic, flaccid-spastic)

Dysarthria can adversely affect intelligibility and naturalness of speech. These abnormalities are due to one or more sensorimotor problems,

including weakness or paralysis, incoordination, involuntary movements, or excessive, reduced, or variable muscle tone.[10]

In the pediatric population, dysarthria may result from many neurologic illnesses, diseases, and disorders, both acquired and congenital. The type of dysarthria is based on the location of the lesion, the nature and course of the underlying condition, and the assessment criteria used. Some of the specific etiologies include cerebral palsy, Chiari malformation, GuillainBarrè and associated autoimmune syndromes, infectious diseases, neoplastic diseases, toxic/metabolic diseases, trauma, and/or vascular diseases.

Signs and symptoms include perceptual speech characteristics and physical signs, which help to identify the various types of dysarthria. Disruptions to speech subsystems include the following:

- *Respiration:* short phrases, reduced loudness, forced expiration/inspiration
- *Phonation:* too high/too low pitch levels, voice tremors, breathiness, hoarse/harsh
- *Articulation:* imprecise consonants, distorted vowels, irregular articulatory breakdown
- *Resonance:* hypernasality, hyponasality, nasal emissions, nasal snorts
- *Prosody:* rate of speech too fast/too slow, rushes of speech, excessive stress

The SLP is responsible for assessing, diagnosing, and treating children with dysarthria. When assessing for dysarthria, the goal is to describe the perceptual characteristics of the speech and identify how each subsystem is affected and the severity of impairment. The impact of dysarthria on speech intelligibility and naturalness, communicative effectiveness, and participation in communication will also be assessed and taken into consideration when determining the plan of care.

APRAXIA

Apraxia of speech (AOS) is a "neurologic speech disorder that reflects an impaired capacity to plan or program sensorimotor commands necessary for directing movements that result in phonetically and prosodically normal speech."[10] The signs and symptoms of AOS include phoneme distortions and distorted substitutions or additions, reduced overall rate of speech, syllable segregation, and equal stress across adjacent syllables. These features are consistent with deficits in planning and programming movements for speech and may increase with higher demands in speech and motor complexity. Acquired AOS is caused by any process or condition that compromised the structures and pathways of the brain responsible for planning and programming motor movements for speech. The most common causes include stroke, traumatic brain injury, tumor, surgical trauma, and/or progressive disease.

The SLP is responsible for assessing, diagnosing, and treating children with acquired AOS. There are standardized and nonstandardized

Table 22.1 Comparison of AOS, Dysarthria, and Aphasia

CHARACTERISTICS	AOS	DYSARTHRIA	APHASIA
Muscle weakness	No	Yes	No
Articulatory deficits	Yes	Yes	No
Prosodic deficits	Yes	Yes	No
Language processing deficits	No	No	Yes
Consistent error patterns	No	Yes	No
Groping articulation	Yes	No	No

AOS, apraxia of speech.

assessment tools which can be used by the SLP. The assessment focuses on the functional aspects of speech production, including intelligibility, or the degree to which the listener understands the acoustic signal; comprehensibility, or the degree to which the listener understands the message; and efficiency, or the rate at which the individual communicates a meaningful utterance. AOS often co-occurs with or presents similarly to other neurogenic communication disorders, specifically dysarthria and aphasia. For this reason, it is important to do a differential diagnosis between these conditions. Table 22.1 provides a basic comparison of AOS, dysarthria, and aphasia.

DYSPHAGIA/FEEDING

Guiding Principles

Dysphagia refers to difficulty in swallowing that occurs during one of the four phases of swallowing. A person may experience dysphagia when they have difficulty managing their own secretions, manipulating foods in the mouth, or unable to safely swallow food or liquids.[11] Difficulty arises during one of the four stages of swallowing:

- *Oral preparatory phase:* The food or liquid mixes with the saliva and is shaped (e.g., sucking, mashing, chewing) in the mouth to form a cohesive bolus.
- *Oral phase:* The oral phase begins with the backward movement of the bolus toward the pharyngeal cavity, ending with initiation of swallow.
- *Pharyngeal phase:* The pharyngeal phase begins with initiation of swallow and continues until the bolus reaches the upper esophageal sphincter entering the esophagus. During this phase, the epiglottis closes to protect the trachea and the larynx to prevent food/liquid from entering the airway.[11]
    - Aspiration refers to the saliva, food, and/or liquid entering the airway before, during, or after the swallow, which can result in overt signs/symptoms such as coughing, choking, watery eyes, throat

clearing, wet vocal quality, color change, increased work of breathing, frequent respiratory infections, or aspiration pneumonia.[12] Aspiration can also be silent, resulting in no overt signs/symptoms of aspiration, and can only be detected by instrumental assessment.

- *Esophageal phase:* The esophageal phase begins as the bolus enters the esophagus and moves to the stomach.

The risks associated with dysphagia include oral aversion, aspiration (a foreign object entering the airway/trachea), compromised respiratory system, gastrointestinal complications, poor weight gain/failure to thrive, dehydration, and prolonged need for tube feedings, along with a variety of sustaining complications impacting the patient's overall quality of life.[13]

Feeding disorders can occur with or without swallowing difficulties. Feeding issues typically consist of a variety of motor complications and/or sensory difficulties that impact the mealtime routine and prevent a child from meeting their nutritional needs by mouth.[14] Common characteristics of feeding disorders include:

- Oral aversion
- Oral hypersensitivity
- Difficulty manipulating bolus in the mouth
- Refusing certain foods or food groups
- Inability to advance to age-appropriate textures or utensils
- Disruptive mealtime behavior or mealtime experiences impacting the family unit
- Poor weight gain/growth over time[13]

Initial Management and Assessment

Both feeding and swallowing disorders require assessment and intervention from an SLP to assess the child's ability to take and receive all nutrition by mouth. An SLP can perform a bedside evaluation and can utilize instrumental assessment when warranted.

- *Bedside evaluation:* This evaluation consists of clinical observation of the patient consuming a variety of food and/or liquid consistencies to monitor for any difficulties during the oral preparatory or the oral phase of swallowing and assess for any overt signs or symptoms of aspiration during the pharyngeal phase of swallowing.[13] The SLP will observe for stress cues from the child or feeder, increased behavior as a form of communication, physiologic changes, and/or other complications.[12]
- *Videofluoroscopic swallow study (VFSS):* This is an instrumental assessment performed under radiographic imaging that provides a lateral or posterior-anterior view of the oral preparatory, oral, pharyngeal, and upper esophageal phases during the swallow.[12] During the study, the patient must consume food/liquid mixed with barium, a substance that appears on imaging, providing a clear picture as to where the bolus is moving during the swallow. An SLP can determine the location of swallow initiation, laryngeal penetration/aspiration (i.e., if/when the

food/liquid is entering the airway), presence of pharyngeal residue, and how these and other anatomical issues may be affecting swallowing. The SLP can trial a variety of consistencies, utensils, or positions to determine the least restrictive diet.[15]

- *Fiberoptic endoscopic evaluation of swallowing (FEES):* This instrumental assessment utilizes a fiberoptic endoscope that is placed through the patient's nasal cavity and passed just above the pharynx to obtain a clear picture of the pharyngeal area, laryngeal vestibule, and pyriform sinuses, and determine functionality during the swallow.[12] An FEES evaluation can determine if food, liquid, or saliva is entering the airway.[16]

Ongoing Care and Treatment

Following bedside and/or instrumental evaluation, an SLP will provide evidence-based therapeutic interventions to address the deficits noted during the assessment. Interventions include but are not limited to:

- Positioning modifications to support stability of the head, neck, and trunk, in collaboration with a physical or occupational therapist[14]
- Oral motor intervention to improve the patient's ability to manipulate food in the mouth and tolerate developmentally appropriate utensils or textures
- Oral sensory intervention to reduce oral aversion and/or hypersensitivity to a variety of flavors and textures
- Therapy techniques which may include use of modalities such as Kinesio Taping, myofascial release, or neuromuscular electrical stimulation (NMES) to improve oral and laryngeal strength and efficiency

The goal of feeding/swallowing therapy is to recommend the safest diet for the patient to receive all nutrition by mouth and/or support tube weaning, support functional oral motor movements to tolerate developmentally or age-appropriate textures, reduce respiratory complications, support mealtime environment, improve the patient's quality of life, and create positive experiences with regard to eating and/or drinking.[13]

Treatment Controversies

- NMES for dysphagia
- Thickened liquids
- Treatment techniques (multidisciplinary approach vs. sensory approach)

Additional Considerations

- Caregiver education/training and buy-in
- Cultural considerations
- Gastrointestinal, respiratory, and cardiac complications
- Oral care
- Quality of life/pleasure feeds
- School accommodations

VITALSTIM

VitalStim is a noninvasive NMES therapy that provides sensory stimulation, improves muscle recruitment to increase muscle strength, and rehabilitates swallowing function.[17] To benefit from this treatment, it is necessary to have intact peripheral nerves which innervate the impaired muscles. VitalStim is used in infants, children,[18] and adults[17] with a broad range of diagnoses and may improve the swallow and allow patients to advance to higher level diets within a shorter time span compared with using traditional methods. Patient populations that have been studied with electrical stimulation include poststroke patients and those with traumatic brain injury, head and neck cancer, neuromuscular disease, progressive neurologic disease, respiratory failure, and other chronic medical conditions.[19]

VitalStim is used to re-educate the swallowing muscles in conjunction with pharyngeal exercises or while swallowing specific textures of food/liquid which challenge the system.

The therapist develops individualized exercise programs to target specific swallowing muscles that have been affected and improve safety and efficiency of swallowing.

EVALUATION AND TREATMENT WITH TRACHEOSTOMIZED AND/OR VENTILATOR-DEPENDENT PATIENTS

In cooperation with a multidisciplinary team, the role of an SLP is to assess, diagnose, and treat communication and swallowing disorders in patients who require an artificial airway due to medical complications. When managed by a multidisciplinary team approach, patient outcomes have been shown to reflect reductions in cannulation times, length of stay in the hospital, adverse events, and cost of care.[20–22]

The role of SLP in these patients includes the following:

- Identifying communication disorders and delays
- Identifying signs and symptoms of dysphagia
- Identifying signs and symptoms of voice disorder
- Conducting comprehensive dysphagia assessment, including noninstrumental and instrumental assessments of swallow function
- Education and training regarding use of augmentative and alternative communication
- Providing treatment for swallowing and communication disorders
- Identifying candidacy for speaking valve trials (multidisciplinary team)

If a patient is a candidate for a one-way speaking valve, the benefits will include the following:

- Restore positive airway pressure
- Improve voice/speech production
- Improve swallow
- Inline ventilator use

- Expedite weaning
- May reduce decannulation time

EVALUATION AND TREATMENT OF VOICE DISORDERS

Tracheostomized or ventilator-dependent patients may also exhibit disorders in vocal characteristics, including vocal quality, pitch, and loudness, which differ or are inappropriate for an individual's age, gender, cultural background, or geographic location.[23,24] The SLP will work with patients to rehabilitate impairments, including vocal quality, loudness, prosody, and phrasing.

In summary, both communication and swallowing are complex skills which require a multisystem coordination. The SLP is highly trained to evaluate, diagnose, and treat acquired disorders, as well as work with families and other professionals to develop a comprehensive plan of care.

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Ultrasound

23

TAMARA ZAGUSTIN

GENERAL PRINCIPLES

In recent years, ultrasound-guided interventions in pediatric rehabilitation medicine (PRM) is more meaningful and used by pediatric physiatrist for treatment of a variety of neuromusculoskeletal conditions that affect children early in life, which make their anatomy unique. With the use of ultrasound, these body structures can be targeted successfully and effectively with less pain, optimizing the outcome of this specific intervention with or without using anatomical landmarks or other nonspecific guidance devices, such as nerve stimulators or electromyography (EMG).

The most common ultrasound-guided structures targeted in PRM targeted by ultrasound are the muscles, peripheral nerves, salivary glands, joint spaces, drug delivery devices, and tendons.

The focus of this chapter is on ultrasound-guided intervention in PRM, not diagnostic ultrasound in PRM, as this would require much more indepth training, knowledge, and expertise. There are many available books, journal articles, and websites that show in full detail how different structures are visually identified and targeted by ultrasound in the adult population and which can be used as a guide for pediatric patients with some differences regarding the identification of muscles/nerve/structures. The main difference is the relatively small size of these structures in younger children, and in some cases some specific differences depending on age and the structure being visualized. For example, when performing an ultrasound-guided knee injection in a 2-year-old child with an acute arthritic knee, the developing bones can have a very different appearance; specifically, the patella of the knee is still not fully ossified (Figure 23.1) and therefore the image of this joint under ultrasound is very different from that of a 15-year-old.

It is important to highlight the fact that no technology or intervention will assist and replace the experience and knowledge of a clinician necessary to understand and best manage:

- Functional anatomy
- Natural history and pathophysiology of underlying neuromusculoskeletal disorders
- Growth and developmental skills of children over time in context with their medical history and overall well-being

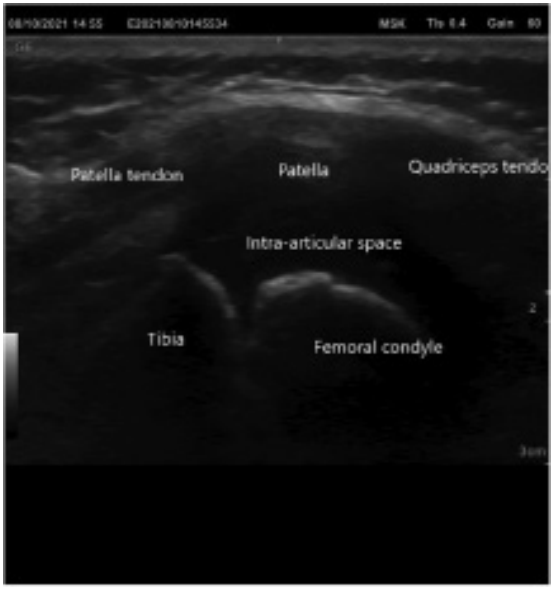

Figure 23.1 Image of a knee joint of a 2-year-old child where the patella bone is not ossified and therefore the femoral condyle can be visualized. This is not visible in older individuals with fully developed and ossified patella bone, which starts between 3 and 5 years of age and completed at 10 or 12 years of age.

Any procedure performed with or without an ultrasound in a child or adult must be done by a trained and knowledgeable physician for it to be meaningful, safe, and effective.

In PRM, ultrasound is considered a helpful tool not only for its clinical use but also for research and education. Many PRM fellowships in North America have identified the need to offer training within their fellowships, and there is greater interest among candidates to pursue training within their fellowships and incorporate it successfully within their practice as specialists.

BASICS

Incorporating ultrasound imaging as a valuable tool in most PRM procedures can maximize precision, minimize complications, limit pain, and optimize patient outcomes.

Ultrasound means "beyond sound" and it is defined as a mechanical energy that is transmitted by longitudinal pressure waves within a medium, which is measured in Hertz (Hz).

Most neuromuscular ultrasound probes have frequencies of between 20 and 23.2 MHz.

The probes have piezoelectric materials which create wave sounds when a voltage is applied, transforming electrical energy into mechanical energy, or sound. At the same time, when sound energy is absorbed into the piezoelectric material, this energy is transformed into electrical energy. This is how the ultrasound image is created.[1]

When the sound wave of a diagnostic ultrasound encounters an area where two different tissues have different acoustic impedances, echoes are created. These echoes increase proportional to the difference between the two impedances. If these echoes are positioned at a perpendicular angle to the probe, these echoes, when they reach the probe, will be recorded as lines and images on the ultrasound. Knowing the location and depth of the tissue creating the echoes, as well as the intensity through the brightness of the returning echoes, can help create the image. This is the image used to help guide interventions with precision.[1]

The higher the frequency of the probe, the higher the resolution of the image and the greater the attenuation of the image as the wave travels through the tissue. Therefore, high-frequency probes are best used for smaller superficial structures (no deeper than 3–4 cm). Most of the structures targeted in PRM are within this depth.

Safe and efficient use of an ultrasound in guided interventional procedures is a skill that is acquired with careful probe and needle alignment, which allows initial good visualization of the needle to facilitate appropriate angle adjustments as it advances toward the target, facilitating accuracy and avoiding structures that could cause complications during the procedure. Acquiring good visual spatial abilities is meaningful when performing ultrasound-guided procedures.

COMMON TERMINOLOGY

- *Echogenicity:* Echogenicity refers to the brightness of an image caused by the reflection of sound waves. It is influenced by tissue density and sound beam characteristics.
  - *Bright areas:* hyperechoic
  - *Dark areas:* hypoechoic
  - *Complete blackness:* anechoic
- *Anisotropy:* Anisotropy is the most common and most important artifact that occurs when the sound wave is not perpendicular to the barrier to allow it to return as an echo and be captured by the probe. The greater the angle, the greater the likelihood of the echoes not reaching the probe. Anisotropy varies among different tissues. Using different manipulations ("rocking" and "heel to toe" maneuvers) on the ultrasound probe to bring out or minimize anisotropy is important as it can confirm the image of, for example, a tendon versus a nerve; the normally bright echoes seen on tendons, for example, will become increasingly dark (hypoechoic) as the probe is tilted away from a 90° angle, while tissues with low anisotropy, such as the nerves, will not change much in terms of brightness as the probe is tilted (Figures 23.2 and 23.3).

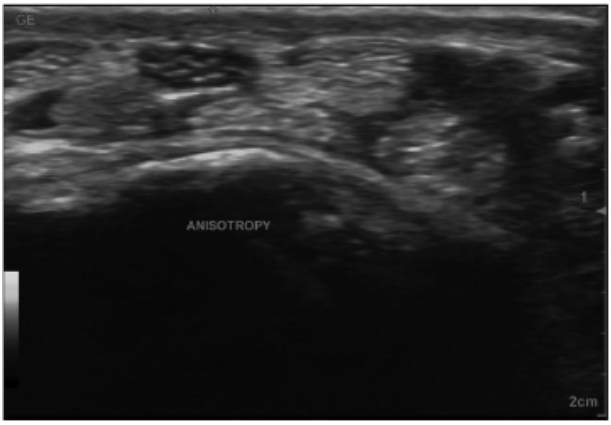

Figure 23.2 Rocking the probe facilitates this image of anisotropy.

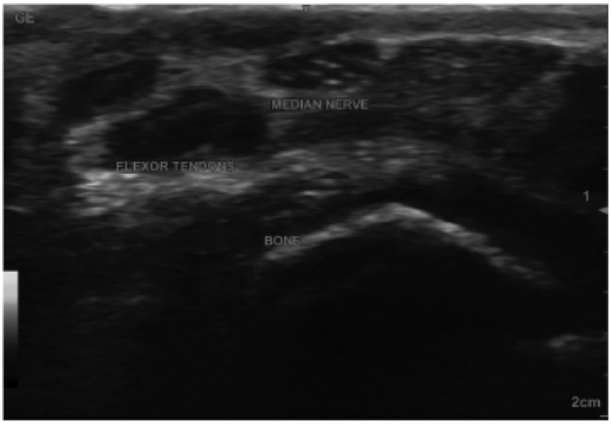

Figure 23.3 Rocking the probe facilitates this image of anisotropy.

- *A-mode:* The amplitude mode is a single line of ultrasound information if the probe only had one piezoelectric element. An image is created by hundreds of piezoelectric elements arranged in a row.
- *B-mode:* The brightness mode is the most common ultrasound image used in PRM. The piezoelectric elements act as both the sender and the receiver.
- *M-mode:* The motion mode combines the A-mode and the B-mode and is most useful when looking at tissue movement, as in cardiac function or diaphragm movement.
- *Doppler mode:* This mode allows evaluation of blood flow with ultrasound (Figure 23.4).
- *Transverse image:* This is a short axis view of the imaged structure (Figure 23.5).

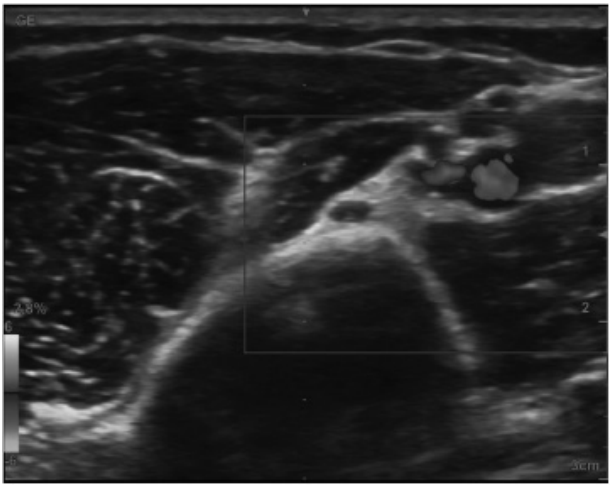

Figure 23.4 Doppler ultrasound facilitates the visualization of the arteries and veins which are often in proximity of larger nerves. The musculocutaneous nerve is to the left of the blood vessels in the arm.

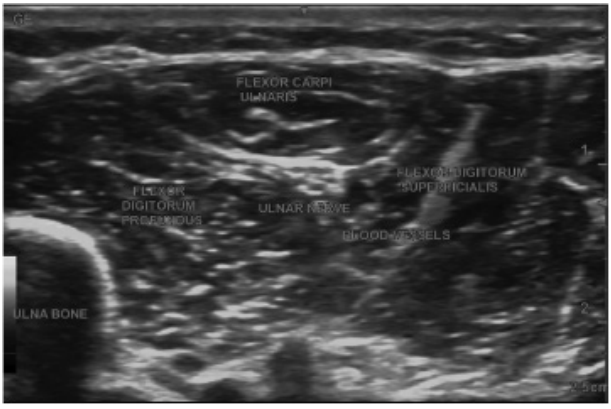

Figure 23.5 Transverse image of the forearm with muscles (starry night appearance), fascia (hyperechoic lines within the muscles that separate one muscle from another), bone (hyperechoic line with black void below), blood vessels (round and oval anechoic structures), and ulnar nerve (honeycomb pattern). The skin and the subcutaneous tissue above are thin hyperechoic linear structures near the surface, and the adipose tissue is hypoechoic with hyperechoic undulating connective tissue.

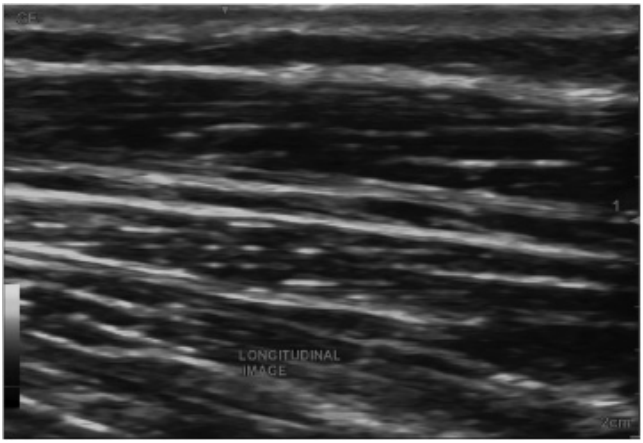

Figure 23.6 Longitudinal image.

- *Longitudinal image:* This is a long-axis view of the imaged structure (Figure 23.6).
- *Oblique image:* The orientation is between transverse and longitudinal images.
- *In-plane approach:* The operator lines up the needle with the long axis of the probe to visualize the needle as it advances to the target as a hyperechoic linear structure (Figure 23.7).

Out-of-plane approach: The operator lines up the probe where the axis of the needle is perpendicular to the long axis of the probe to visualize the needle as it advances to the target as a hyperechoic dot (Figure 23.8).

TYPES OF PROBES

- The linear probe is the most common probe used in PRM as most structures can be easily identified and targeted in children. It creates a rectangular image with the maximal frequencies in the 12 to 16 MHz range.
- The hockey stick probe has a high frequency of 18 to 20 MHz and is best for small superficial structures, where a smaller footprint is needed to optimize visualization of targeted structures.
- The curvilinear probe creates a sector image (piece of a pie) with a 2 to 5 MHz frequency range and is used for deeper structures (nondislocated hip joint, lumbar sympathetic plexus, diaphragm).

TIPS ON HOW TO OPTIMIZE IMAGES

- Choose the best probe depending on the depth and size of the targeted structure, plus body surface size.
- Ensure good contact of the probe with the skin with sufficient gel and light pressure on the probe (avoid anechoic areas and distortion of body structures due to excessive pressure).

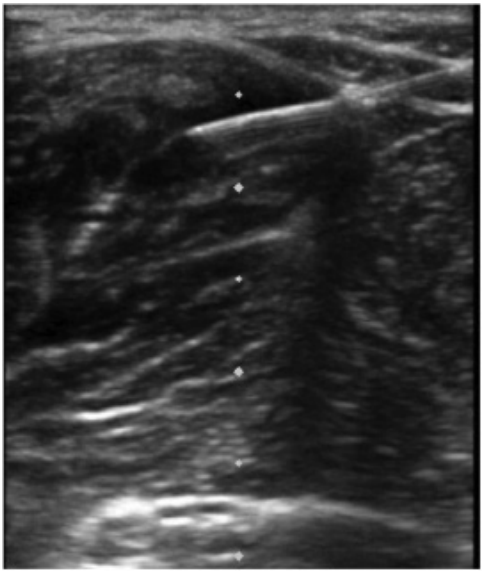

Figure 23.7 In-plane approach of the needle into the muscle (pectineus muscle).

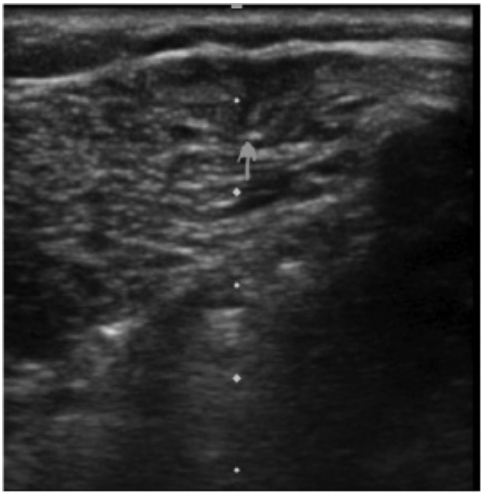

Figure 23.8 Out-of-plane approach of the needle tip into the muscle (arrow pointing at the needle tip). Having good depth perception of the needle is important for patient safety as the needle is not always fully visualized compared with the in-plane approach.

- Adjust brightness, depth, and focus.
- Adjust the gain to amplify the ultrasonic signal in a uniform way to make it easier to see the tissue by making the returning signal stronger.
- Keep probe perpendicular to the structure being imaged.
- Be consistent with the orientation of the probe in relation to the image (standardize the method of providing the image).
  - For longitudinal studies, ensure the left side of the image is cephalad.
  - For axial studies, ensure either the left side of the image is always toward the patient's right side or the left side of the image is always lateral.
- Use color and/or power Doppler to assess for blood vessels, increased vascularity, and inflammation.
- Feel comfortable with moving the probe to visualize well the different structures being examined. Rocking the probe (on its short axis) is more common than the heel to toe maneuver (on its long axis) to create or correct anisotropy. It is reassuring when you can consistently find the image you are looking for as you move the probe on a body area.

ADVANTAGES OF USING ULTRASOUND FOR PEDIATRIC REHABILITATION MEDICINE-GUIDED PROCEDURES

- Ultrasound machines are readily available within clinic and hospital settings.
- Ultrasound machines can be very portable, easy to use, and affordable.
- They have improved software/technology to easily identify neuromusculoskeletal structures, which has allowed its cost-effective use in clinic and hospital settings by nonradiology-trained healthcare professionals.
- Ultrasound allows for real-time evaluation of soft body structures, passively and dynamically.
- It is nonpainful to the patient.
- It is safe with no exposure to radiation.
- It allows structures to be evaluated in many different planes (axial, coronal, sagittal).
- It allows for real-time visualization of needle advancement within the body toward the targeted structures with or without therapeutic interventions (medication, EMG needle, etc.), optimizing procedure outcomes, causing less pain, and minimizing complications by avoiding other nontargeted neighboring structures (blood vessels, lungs, etc.).
- There is no need to rely on body landmarks, which can be imprecise in pediatric patients due to variabilities in patient size due to age, body habitus, prior surgeries altering their anatomy, presence of musculoskeletal deformities, limited mobility/positioning, and presence of anatomical variations due to an underlying diagnosis.
- It allows for good precision when used alone or in combination with a nerve stimulator.

- *Targeting muscles:* It allows the use of smaller gauge needles, causing less pain compared with when targeting muscles with a nerve stimulator.
- *Targeting nerves:* In combination with a nerve stimulator at low intensity, muscle activity can be observed effectively on ultrasound with less pain than if needing to rely on clinically visible or palpable muscle twitch (this would require higher intensity on a nerve stimulator, which would also translate into less precision).
- Targeting anatomical structures blindly in children or adult population using any interventional procedure comes with a high risk of not being on the appropriate targeted structure.

- Ultrasound adds research opportunities/data that could be meaningful to PRM.

HECKMATT SCALE

The Heckmatt Scale (Figure 23.9) is a quantitative muscle scoring scale (Table 23.1) originally described in 1982 in children with neuromuscular disorders to identify subjective changes within the muscles through time.

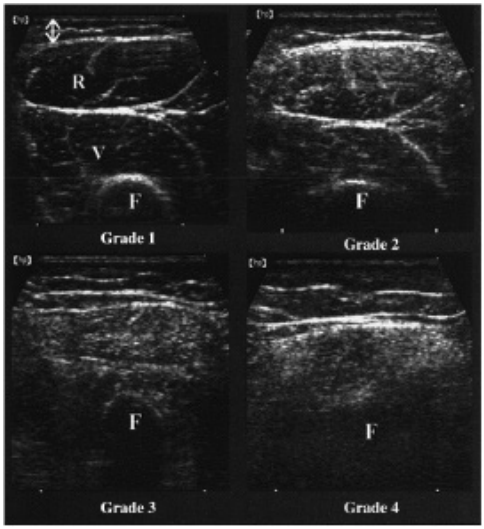

Figure 23.9 The Heckmatt quantitative muscle scoring scale that goes from grade 1 to grade 4 depending on muscle echogenicity and degree of bone reflection.

F, femur; R, rectus femoris; V, vastus medialis.

Source: Heckmatt JZ, Leeman S, Dubowitz V. Ultrasound imaging in the diagnosis of muscle disease. *J Pediatr*. 1982;101(5):656–660. doi:10.1016/s0022-3476(82)80286-2. PMID: 7131136.

Table 23.1 Heckmatt Quantitative Muscle Scoring Scale

Grade 1	Normal; predominantly dark muscle with bright distinct bone reflection
Grade 2	Mildly abnormal; increased muscle echogenicity with normal bone reflection
Grade 3	Moderately abnormal; moderately increased muscle echogenicity with reduced bone reflection
Grade 4	Markedly abnormal; markedly increased muscle echogenicity with absent bone reflection

Source: Heckmatt JZ, Leeman S, Dubowitz V. Ultrasound imaging in the diagnosis of muscle disease. *J Pediatr*. 1982;101(5):656–660. doi:10.1016/s0022-3476(82)80286-2. PMID: 7131136.

This scale has been used to evaluate how these changes could influence muscle flexibility and therapeutic response to certain interventions, such as botulinum toxin injections for spasticity management. Muscles with grades 1 and 2 showed better outcomes than muscles with grades 3 and 4.[2]

Recent studies have combined the echogenicity findings of the Heckmatt Scale with muscle stiffness, which is a mechanical property of the muscles not captured on ultrasound images, and showed that muscle stiffness is not directly proportional to muscle echogenicity.[3]

The Modified Heckmatt Scale has also been validated in individuals with spasticity using a more specific definition for grades 2 and 3 to allow for better differentiation, as a muscle might not be homogenously affected throughout its length, hindering precise distinction between grades 2 and 3.[4] In grade 2 of the Modified Heckmatt Scale, there is 10% to 50% increased muscle echogenicity and distinct bone echo and areas of normal muscle, while in grade 3 there is marked increase in muscle echogenicity between 50% and 90% of tissue, with reduced distinction of the bone echo from the muscle.

Having a more precise definition between the different grades within the Heckmatt Scale and their relationship with muscle stiffness could have clinical implications, as we incorporate this scale consistently in PRM clinical practice and research.

COMMON TISSUE PATTERNS[5]

- *Peripheral nerves:* These are shown with a honeycomb pattern (on transverse image) where the actual nerve fibers are hypoechoic (dark) surrounded by the connective tissue of the perineurium and epineurium, which is hyperechoic (bright). The longitudinal image of a nerve shows bright epineurium on the periphery of the nerve with parallel lines inside, the perineurium. Color Doppler over the nerve shows little or no signal as the blood vessels to nerves is through nondetectable small vessels.
- *Muscles:* These are shown with a "starry night appearance" on axial images. Actual muscle fibers are dark, while the connective tissue of

the muscle is bright. On longitudinal images, the parallel lines of the connective tissue can be seen. The fascia tends to be very hyperechoic.

- *Tendons:* These are densely packed collagen fibers which are hyperechoic and with prominent anisotropy.
- *Bone:* It creates a very hyperechoic line and reflects the ultrasound wave completely, with a black void below the bone image as the ultrasound wave cannot penetrate the bone.
- *Arteries and veins:* These are similar-looking round or oval anechoic structures. Pressure on the probe will cause the vein to shrink or disappear as most veins are easily compressible due to low pressure and slow blood flow, compared with arteries which have greater pressure with blood traveling at higher speeds. Doppler imaging is positive and the arteries are shown in red. The veins are shown in blue on Doppler, but only larger veins are detectable as medium and small veins have slow blood flow.
- *Skin and subcutaneous tissues:* The skin is shown as a thin hyperechoic line near the surface of the ultrasound image followed below by the adipose tissue, which is hypoechoic with prominent hyperechoic undulating connective tissue septa that run through the adipose tissue, which can be mistaken as a muscle but not with the "starry night appearance."
- *Salivary glands:* These are seen as hypoechoic structures with a homogenous echotexture compared with the surrounding tissue. The parotid glands have more fat content, causing attenuation of the signal, compared with the submandibular glands (Figures 23.10 and 23.11).
- *Intraarticular space:* The cortical bone margin appears bright, sharp, and hyperechoic. The synovial fluid within the joint space is anechoic (black; (Figure 23.12).

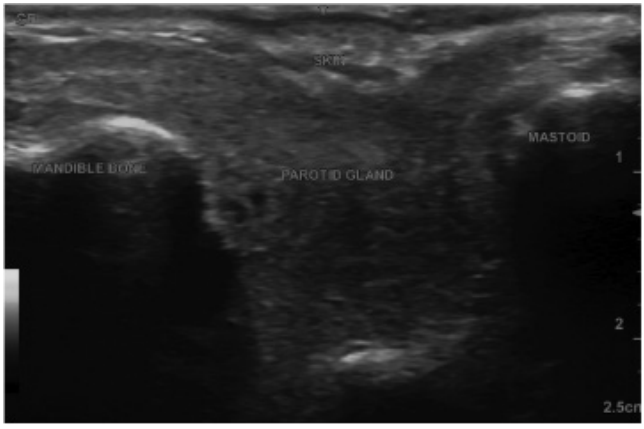

Figure 23.10 Parotid gland (periauricular portion). The facial nerve lies midpoint in depth within the parotid gland, usually not visible on ultrasound yet accompanied by visible vascular structures.

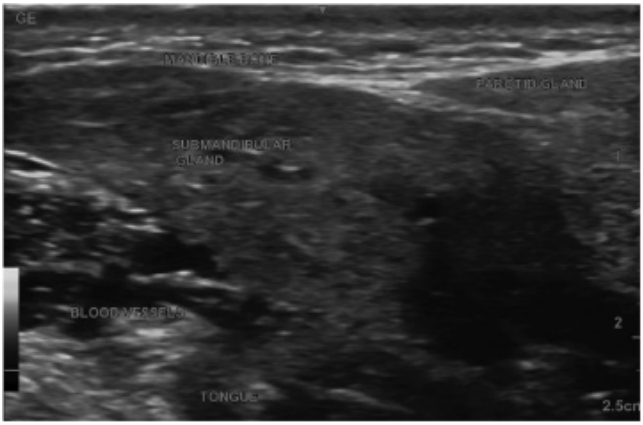

Figure 23.11 Submandibular gland and retromandibular portion of the parotid gland. The parotid gland has a more attenuated signal than the submandibular gland since the parotid gland has higher fatty content. Note that the tongue lies below the submandibular gland so minimize be mindful with injection of large volumes of botulinum toxin. Keep the injection close to the mandible bone.

LIMITATIONS IN USE OF ULTRASOUND

- Structures under the bone, surgical hardware, and medical devices cannot be visualized, but can be easily identified on ultrasound.
- The deeper the structures, the lower the resolution (attenuation of the sound wave happens as it travels through tissues). This is most meaningful in obese patients.
- Scarring tissues can interfere with the ultrasound image.
- Some structures require more experience, time, and skills to master. Overall, it is an art within medicine and its usefulness is physician-dependent; the physician needs to be competent in the skill.
- Ultrasound is an evolving technology that requires update with ongoing continued learning/education.

COMMON PROCEDURES IN PEDIATRIC REHABILITATION MEDICINE WITH ULTRASOUND GUIDANCE

- Needle guidance toward the muscles or nerves for management of hypertonia in children (e.g., diagnostic lidocaine blocks, botulinum toxin muscle injections,[6–8] and neurolysis with phenol/ethanol/cryoablation[9])
- Needle guidance to address acute and chronic pain in children, which can be both diagnostic and therapeutic (e.g., peripheral nerve blocks and/or sympathetic lumbar plexus blocks in cancerous pain such as

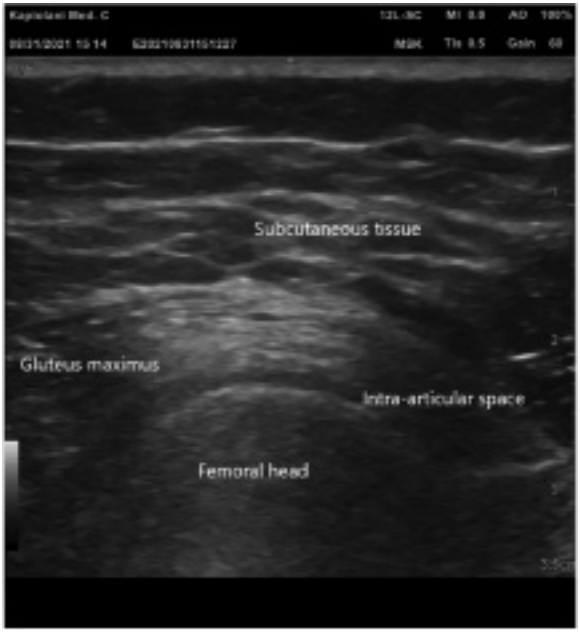

Figure 23.12 Fully dislocated hip joint in a child with spastic quadriplegic cerebral palsy with Gross Motor Function Classification System (GMFCS) 5. The femoral head becomes superficial so a linear transducer could be utilized instead of a low-frequency curvilinear transducer. The hip joint is easy to target with less complications with regard to the large neurovascular structures (femoral vessels) in the vicinity as the hip has migrated proximally and away from these structures. A 1.5- to 2-inch needle is placed in plane with the transducer to access the joint space at the headneck junction of the femur instead. Normally a 3.5-inch needle is necessary as the hip joint is deeper when in joint.

osteosarcoma[10]; greater occipital neuralgia for headaches and neck pain; anterior cutaneous nerve entrapment syndrome in chronic abdominal pain[11]; and intraarticular hip joint injection for chronic hip pain due to osteoarthritis with or without hip dislocation in patients with cerebral palsy[12])

- Needle guidance in difficult-to-access drug delivery devices (Figures 23.13 and 23.14) for medication refill or for troubleshooting pump malfunction in children with hypertonia who require intrathecal baclofen therapy; note ultrasound-guided refills of drug delivery devices compared with template-guided procedures take longer, but with less procedural pain

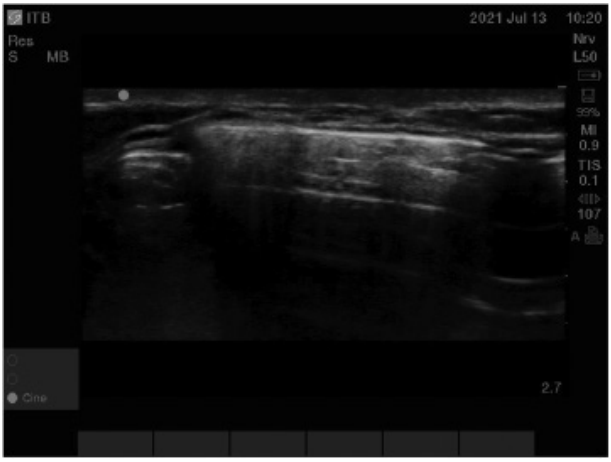

Figure 23.13 Ultrasound view of a drug delivery device, Medtronic II, which is commonly used in pediatric spasticity management in North America with the side port identified on the left side of the screen (smaller and on the edge of the drug delivery device), while in the midportion of the drug delivery device (right side of image) is the access to the drug reservoir.

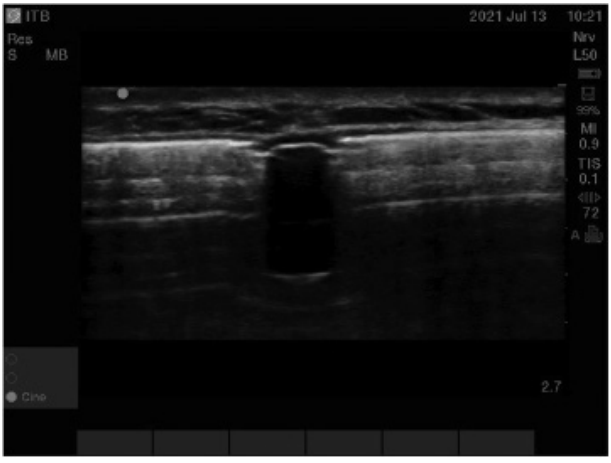

Figure 23.14 Ultrasound view of a drug delivery device, Medtronic II, which is commonly used in pediatric spasticity management in North America with the view of the drug reservoir port in the midportion of the device.

reported by adult patients and with no safety issues observed in either group[13]; instances that require access to drug delivery devices include:
  - Presence of surgical scar tissue formation
  - Challenging body habitus in children, such as obesity, musculoskeletal deformity with neuromuscular scoliosis, and spasticity[14]
  - Suboptimal pump positioning, pump flip, or changes in the position of the drug delivery device throughout time with growth
  - Excessive fluid surrounding the drug delivery device in the presence of seroma or cerebral spinal fluid leak
- Needle guidance for EMG studies, to allow precision, minimize the number of needle sticks, limit discomfort and complications from needle sticks, and therefore improve diagnostic efficiency and accuracy[15]
- Precise access to intraarticular spaces for diagnostic and therapeutic interventions with real-time visualization of the needle tip entering the joint space and injection of therapeutic agent, improving diagnostic and therapeutic outcomes[16]
- Salivary gland injections of the parotid and submandibular glands for effective and safe management of sialorrhea[17]
- Analysis of muscle dynamics/activation, including the diaphragm,[18] and planning of functional surgeries in achondroplasia
- Scout an area where minimally invasive percutaneous needle tenotomy will be performed to address severe/debilitating/complicated/painful muscle contractures and avoid injury to neighboring elements, such as neurovascular bundle or other noncontracted tendons within the vicinity of this otherwise blinded superficial procedure; the tendons and muscles targeted in this procedure usually too superficial for ultrasound guidance to be of any benefit, and palpation of the contracted tendons and muscles is of greater value as the tendons will be outstandingly tight, superficial, and evident to palpation, but no longer evident at the termination of this intervention with some increase in range of motion of the joint in question[19]

TIPS ON ULTRASOUND-GUIDED INTERVENTIONS IN PEDIATRIC REHABILITATION MEDICINE

- Kids are small so not all structures are always as visible and well-defined as in adults, yet ultrasound can provide great guidance and certainty as to where to search for the structure in a more targeted way and within the known anatomical area rather than searching for it blindly.
- Proximal nerve motor branches enter a muscle usually accompanied by vascular structures and therefore using Doppler ultrasound can assist in the identification of vascular structures to then find the targeted motor nerve branch, which is usually not visible especially in children. It is usually in the proximal portion of the muscle within the fascia. Larger nerves in general travel with blood vessels.
- Know the normal neuromusculoskeletal structures well to understand what is abnormal or different.

- Compare with the other side when able to.
- Observe the structures with ultrasound while passively or actively moving the neuromusculoskeletal structure in question. An activated muscle tends to increase its width compared with when it is at rest.
- Passive tendon stretch of a specific muscle with the ultrasound probe on the muscle belly can help with identification of the muscle. This can be helpful when the patient is not able to activate the muscle.
- Teach others how to incorporate ultrasound imaging in their practice. This will increase knowledge and proficiency in the use of ultrasound.
- Be comfortable with in-plane and out-of-plane injections. In children, most targeted anatomical structures are very small, more superficial, and with less surface area available, making out-of-plane injections safer, precise, and less painful at times.
- Relax and do not place pressure on the transducer while scanning as this will alter the image of the structures being visualized.
- Ultrasound can be a distraction for children undergoing a painful procedure and thus could facilitate better tolerance and compliance during the procedure.
- Visualizing how a desired structure is safely targeted during a procedure is reassuring for the health provider, the patient, and the family members present during the ultrasound-guided procedure.
- Set up the procedure including the patient, the ultrasound equipment,[22,19] supplies, medications, and other team members appropriately to facilitate good ergonomics when using the ultrasound to achieve the best procedural outcome, patient comfort, and overall health well-being of the PRM physician.[20,21]

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PART VI

Other Conditions

AL Grawany

Cancer Rehabilitation*

24

DAVID W. PRUITT, PRIYA D. BOLIKAL, ASHLEE K. BOLGER, and KELLI N. CHAVIANO

GENERAL PRINCIPLES

Definition

Impairments in body structures and functions related to a cancer diagnosis and its treatment(s) lead to both activity limitations and participation restrictions that can occur acutely during treatment, subacutely following completion of treatment, and longitudinally into survivorship. Over time, these limitations can accumulate and worsen.[1]

- The high proportion of survivors of childhood and adolescent cancers represents individuals at high risk of experiencing serious, disabling, and life-threatening acute, chronic, and late adverse effects of cancer and its therapy.[1]
- Survivors of childhood cancer require lifelong surveillance and appropriate interventions for physical, cognitive, psychological, and emotional treatment-related toxicities and late effects.[1]
- The optimal timing of rehabilitation input for the patient should begin at the time of diagnosis, even prior to initial surgeries, chemotherapy, radiation, or other treatments.[2]
    - The focus during this prehabilitation time should be optimizing endurance, strength, and nutritional and psychological well-being.
- When initiating and planning for cancer rehabilitative care, two concepts are of utmost importance.
    - First, one needs to consider where the patient is within their timeline of treatment. In 1981, J. Herbert Dietz proposed a model for the rehabilitation care continuum in cancer patients which is used widely today.[3] This model describes four phases of care: preventive, restorative, supportive, and palliative.
    - Considering the appropriate setting in which rehabilitation care is provided is imperative, as an inpatient or an outpatient setting may be appropriate depending on the goals and needs of the individual child, as well as the child's medical status and ongoing treatments.

*This chapter does not contain any studies with human or animal subjects performed by any of the authors.

Epidemiology

- The National Cancer Institute's Surveillance, Epidemiology, and End Results (SEER) Program represents the primary source of population-based data on cancer incidence in the United States.[4]
- Childhood cancers are rare (1% of all cancers diagnosed in the United States each year).[5]
- Cancer rates are higher in adolescents between the ages of 15 and 19 years (annual incidence of 245.4 per million per year) compared with rates in children younger than 15 years of age (annual incidence of 168.9 per million per year).[4]
- In contrast to the adult population, in whom solid tumor malignancies predominate, almost 40% of childhood cancers are hematologic malignancies (leukemia and lymphoma).[6]
- In children aged 0 to 14 years, acute lymphoblastic leukemia (ALL) is the most frequently occurring type of cancer and central nervous system (CNS) cancers are the most common solid tumors.
- In adolescents aged 15 to 19 years, the most frequently diagnosed cancer is Hodgkin lymphoma; however, CNS tumors remain a highly occurring type of tumor in this age group as well.
- Survival rates of children diagnosed with cancer have improved markedly since the 1960s, when the overall 5-year survival rate after a cancer diagnosis was estimated as 28%.[7] Currently, the overall 5-year survival rate of children and adolescents diagnosed with cancer between 2010 and 2016 is 85.4%.[1]
- Despite improvement in survival, cancer continues to be the second most common cause of death among children aged 1 to 14 years in the United States, surpassed only by accidents.[8]

DIAGNOSIS

Risk Factors

Although some genetic syndromes are known to increase the risk of cancer before the age of 18, the etiology of majority of cancers in this age range is largely unknown.

Clinical Presentation

It is important to consider cancer as part of the differential diagnosis when seeing patients with neurologic or musculoskeletal complaints.

- *Systemic symptoms/red flags:* These include unexplained fever, weight loss, emesis, bone pain, fatigue, and gait abnormality.
- *Intracranial tumors:* Pediatric rehabilitation medicine (PRM) specialists are typically consulted following initial tumor resection and prior to potential additional treatments, which may include radiation and/or chemotherapy.
    - Initial functional deficits are primarily related to intracranial tumor location(s) impacted by the tumor (Table 24.1).

- *Leukemia:* Rehabilitation assessment can occur at different phases of treatment and thus clinical presentation is variable depending on the treatment intervention or interval that the patient is receiving.
  - *Induction/Intensification:* generalized weakness, potential focal neurologic deficits, or generalized weakness, potentially related to arachnoiditis
  - *Maintenance:* obesity, gait impairments, peripheral neuropathy, and steroid myopathy
- *Long bone sarcomas (Ewing sarcoma, osteosarcoma):* Interventions by PRM can occur throughout the course of treatment, from initial diagnosis and adjunctive chemotherapy through the postoperative course following definitive surgical intervention.
  - Patients may present in a PRM outpatient clinic before diagnosis with extremity pain, leg swelling, or new-onset gait abnormality.
  - Evaluation during the course of treatment may include new functional deficits related to amputation or limb-sparing surgery, as well as potential weakness related to adjunctive treatments, including peripheral neuropathy or radiation plexopathy.

Symptoms and Physical Examination

- *Intracranial tumors:* varying symptoms based on the location of the tumor (Table 24.1)
  - *Suprasellar:* headache, visual impairments, auditory impairments, behavioral changes, cognitive deficits, weakness including hemiparesis, sensory deficits, seizures, hormonal changes (e.g., pituitary dysfunction), spasticity, and nausea/vomiting
  - *Infrasellar:* headache, ataxia, dysmetria, dysarthria, dysphagia, mutism, behavioral changes, nausea/vomiting, vision changes, and gait abnormality
- Spinal cord tumors
  - Progressive weakness and gait abnormality, and sensory deficits
  - Changes in bowel and bladder
- *Leukemia:* bone pain, gait abnormality, refusal to walk due to proliferation of malignant cells in the bone marrow or other organs,[9] fatigue, and easy bleeding/bruising
- *Bone tumors:* local pain, swelling/mass, gait abnormality, refusal to walk, and pathologic fracture; often mimic musculoskeletal injuries and are mistaken for sports injuries or growing pain[10,11]

Diagnostic Evaluation

Typically, pediatric physiatrists are not involved in the initial diagnosis and treatment of pediatric cancers. However, these diagnoses may be discovered during workup for other neurologic or musculoskeletal complaints that may come to the attention of a PRM specialist.

Table 24.1 Functional Deficits Associated With CNS Tumor Location

TUMOR LOCATION		TUMOR EXAMPLES	NEUROLOGIC/FUNCTIONAL DEFICITS	OTHER CONCERNS
Supratentorial	Central	• Craniopharyngioma • Optic glioma • Pituitary adenoma • Germinoma	• Visual impairments/optic neuropathy/abnormal eye movements • Hydrocephalus • Vertigo • Behavioral changes/school difficulties • Auditory symptoms	• Hormonal deficiencies (e.g., diabetes insipidus, hypothalamic deficiency) • Seizures • Weight loss/weight gain • Headache
	Hemispheric	• Low-/high-grade glioma (e.g., pilocytic astrocytoma, ependymoma)	• Weakness (e.g., hemiparesis) • Sensory changes • Cognitive deficits (e.g., memory/concentration) • Visual concerns • Behavioral changes • Increased tone/spasticity	• Seizures • Headache

Infratentorial	Posterior fossa	• Medulloblastoma • Juvenile pilocytic astrocytoma • Ependymoma • ATRT	• Dysarthria or mutism • Impaired coordination • Visual impairments • Behavioral changes • Abnormal gait • Vertigo • Auditory symptoms	• Nausea/vomiting • Headache • Lethargy
	Brainstem	• Diffuse midline glioma • Low-/high-grade glioma	• Cranial nerve dysfunction • Visual impairments • Pyramidal signs • Impaired coordination • Abnormal gait • Behavioral changes/school difficulties • Increased tone/spasticity	

(*continued*)

Table 24.1 Functional Deficits Associated With CNS Tumor Location (*continued*)

TUMOR LOCATION		TUMOR EXAMPLES	NEUROLOGIC/FUNCTIONAL DEFICITS	OTHER CONCERNS
Spinal cord	Spinal cord	• Extradural (e.g., neuroblastoma, sarcoma) • Intradural, extramedullary (e.g., meningiomas, schwannomas) • Intradural, intramedullary (e.g., gangliomas, PNET, astrocytomas, ependymomas)	• Paralysis or focal motor weakness • Sensory deficits • Increased tone/spasticity	• Back pain • Autonomic dysreflexia • Neurogenic bowel and bladder • Syrinx • Spinal deformities (e.g., scoliosis, kyphosis) • Torticollis

ATRT, atypical teratoid rhabdoid tumor; CNS, central nervous system.

Source: Adapted from Houtrow A. Neurodegenerative and demyelinating diseases and other CNS disorders. In: Alexander MA, Matthews DJ, editors. *Pediatric Rehabilitation, Principles & Practice*. 5th ed. Demos Medical Publishing, LLC; 2015; 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.

TREATMENT

Guiding Principles

The majority of children are enrolled in the National Cancer Institute's Children's Oncology Group (COG) cooperative group treatment protocols.

- These trials are often multimodal and may include surgery, chemotherapy, radiation therapy, molecularly targeted therapies, adoptive cellular immunotherapy, and hematopoietic stem cell transplantation.
- Many of these interventions have associated adverse effects that limit functional activity and participation (Table 24.2).

Initial Management

- *Surgery:* surgical approach specific to each type of cancer
  - For intracranial tumors, gross total resection is attempted prior to adjunctive therapy to minimize morbidity and preserve vital structures.[2]
  - *Surgical timing:* For Ewing sarcoma, osteosarcoma, or other bone tumors, biopsy is obtained first. Patients are then generally treated with adjuvant chemotherapy and/or radiation for resection later.[2]
  - Potential functional risks associated with surgical interventions include the following:
    - Deficits dependent on the location and extent of surgery, age, and tumor type
      - *Posterior fossa syndrome:* constellation of symptoms that can occur with CNS tumor resections localized in the posterior fossa, particularly medulloblastoma
        - *Symptoms:* expressive speech impairment/mutism, emotional lability, motor deficits, ataxia, and hypotonia
        - Exact pathophysiology unknown
- Chemotherapy
  - Treatment protocols with chemotherapy typically include a combination of drugs that have varying mechanisms of action, nonoverlapping toxicities, and synergistic effects which increase the likelihood of achieving remission and decrease the likelihood of relapse.
  - Adjuvant treatment refers to using chemotherapy with local control, such as surgery or radiation.
  - Potential functional risks associated with chemotherapy treatment include the following:
    - *Neurologic:* chemotherapy-induced peripheral neuropathy (CIPN), leukoencephalopathy, neurocognitive deficits, and ototoxicity
    - *Musculoskeletal:* myopathy, impaired bone health and skeletal complications, and decreased exercise tolerance
    - Cardiac and pulmonary toxicity
    - Poor nutrition
    - Fatigue
- Radiation therapy
  - This is a treatment method utilized to attain local control and can be delivered both externally (from a machine outside the body) or internally (temporary or permanent implants placed near the tumor).

Table 24.2 Impact of Anticancer Therapies on Function

TREATMENT	PATHOPHYSIOLOGY	IMPAIRMENT	DISABILITY	QUALIFIERS
Surgical resection Posterior fossa surgery	Unclear	Deficits dependent on location and extent of surgery, age, and tumor type Cerebellar mutism May see high-level linguistic and cognitive deficits	Functional limitations related to areas of deficit Limited verbal communication, may affect social skills and school performance	
Cranial irradiation	Neural/glial degeneration Gliosis Proliferative/sclerosing angiopathy Demyelination Ischemic events related to cerebral vasculopathy Progressive, necrotizing leukoencephalopathy Negative impact on GHRH when the posterior fossa is involved	Cognitive dysfunction Learning disabilities ↓ memory Attention problems Language deficits ↓ executive function ↓ verbal and performance IQ Short stature	↓ academic potential ↓ communication skills to language disorder/delay may impact behavior, social competence, and vocational potential Functional deficits related to location and extent of ischemia Potential for marked functional impairments in all affected areas, including dementia, dysarthria, ataxia and/or spasticity May affect self-image, social competence	Impact related to dose and volume of CNS irradiated and inversely related to age of child at time of exposure Effects potentiated by IT or high-dose IV methotrexate Rehabilitation managed as in stroke Usually seen when treatment has included methotrexate Cosmesis included in the problem list for adolescents

Spinal irradiation	May cause radiation myelitis Failure of vertebral growth	Spastic quadriplegia or paraplegia Neurogenic bowel and bladder Short stature ↑ risk of scoliosis or kyphosis	Functional impairments in mobility and ADLs dependent on level of injury May require special program for evacuation of bladder and bowel Potential altered self-image and ↓ social competence	Management as in spinal cord injury Cosmesis of particular psychosocial impact for adolescents
Mediastinal irradiation	Vascular damage, fibrosis In very young, possible interference with both lung and chest wall growth Fibrosis of the parietal pericardium (most common), intimal proliferation of myofibroblasts, collagen and lipid accumulation	Pulmonary fibrosis Pneumonitis Decrease in lung volume, compliance, and CO_2 diffusing capacity Constrictive pericarditis, myocardial damage (rare), conduction system defects, coronary artery disease	Disability dependent on degree of restrictive lung changes and can significantly limit ADLs, exercise tolerance when severe Functional limitations related to degree of cardiac dysfunction	Decrease in radiation-induced late pulmonary toxicity seen over the last decade due to refinements in radiation therapy

(*continued*)

Table 24.2 Impact of Anticancer Therapies on Function (*continued*)

TREATMENT	PATHOPHYSIOLOGY	IMPAIRMENT	DISABILITY	QUALIFIERS
Methotrexate	Neurotoxicity, including acute, stroke-like encephalopathy, chronic leukoencephalopathy (progressive demyelinating encephalopathy) Osteopathy	Cognitive impairment, developmental delay, learning problems, potential motor impairment with deficits in coordination and high-level skills With intrathecal dosing, can see ascending radiculopathy, similar to GBS Osteoporosis/↑ risk of pathologic fractures, bone pain	May affect school performance, ↓ age-appropriate ADL independence, may limit participation in athletics and team sports, may impact self-esteem and social competence Loss of motor function with consequent mobility and ADL deficits dependent on extent of weakness Limitations in mobility and ADLs related to areas involved	Neurotoxicity potentiated by cranial irradiation Alert parents to monitor for school problems developing in upper grades when ↑ independence and efficiency required Toxicity cumulative
Corticosteroids	Preferential atrophy of type II muscle fibers	Myopathy Osteoporosis Avascular necrosis Growth failure	Decreased mobility related to proximal muscle weakness ↑ risk of pathologic fracture Hip pain, gait abnormality Impacts self-esteem and social competence	Reversible when drug is withdrawn or dose is reduced ↑ risk of osteonecrosis of weight-bearing joints in children when ↑ doses are used

Vincristine/vinblastine	Axonal sensorimotor polyneuropathy Impairment of efferent and afferent pathways from the sacral spinal cord, autonomic neuropathy	Impaired rectal emptying	Paresthesias, neuritic pain, distal weakness which may impair hand function, cause foot drop and walking difficulty Constipation which may alter ADLs, comfort	Neurotoxicity more prominent in the presence of CMT May demonstrate improvement or recovery with end of therapy or ↓ dose Neurotoxicity usually minimal with vinblastine
Anthracycline (doxorubicin, daunorubicin)		Can cause arrhythmias, conduction abnormalities ↓ left ventricular function, chronic cardiomyopathy	Diminished capacity to perform age-appropriate ADLs ↓ endurance, ↓ exercise tolerance, limited ability to participate in sports, may impact self-esteem and social competence	Potentiates radiation reactions Increased toxicity with lower age
Cisplatin	Injury to the hair cells of the organ of Corti	High-frequency sensorineural hearing loss Tinnitus Reversible sensory peripheral neuropathy	Affects communication skills and potentially speech/language development in the young child, may impact social competence Paresthesias/neuritic pain may interfere with ADLs, comfort	Ototoxic and neurotoxic effects are cumulative Symptoms may progress after discontinuation

(*continued*)

Table 24.2 Impact of Anticancer Therapies on Function (*continued*)

TREATMENT	PATHOPHYSIOLOGY	IMPAIRMENT	DISABILITY	QUALIFIERS
Carboplatin	Minor or absent loss of the hair cells of the organ of Corti	High-frequency sensorineural hearing loss	Affects communication skills and potentially speech/language development in the young child, may impact social competence	Effects are cumulative Ototoxicity and neurotoxicity milder than cisplatin
Cyclophosphamide/ ifosfamide	Can cause hemorrhagic cystitis secondary to urotoxic metabolite acrolein	Reversible neurotoxicity with somnolence, disorientation, lethargy, hallucinations Avascular necrosis Potential loss of renal function	Negative impact on ability to perform age-appropriate ADLs Pain may limit ADLs, ambulation	Risk of neurotoxicity ↑ with prior use of high-dose cisplatin Reversible or preventable with methylene blue Occurrence reduced by use of mensa

ADLs, activities of daily living; CMT, Charcot–Marie–Tooth disease; CNS, central nervous system; GBS, Guillain–Barrè syndrome; GHRH, growth hormone-releasing hormone; IT, intrathecal; ↓, decreased; ↑, increased.

Source: Reprinted with permission from Pruitt DW, McMahon MA, Apkon SA. Rehabilitation of the child with cancer. In: Pizzo PA, Poplack DG, eds. *Principles and Practice of Pediatric Oncology.* Wolters Kluwer; 2016:1109–1110.

- The general types of radiation are electromagnetic (traditional photon radiation) and particulate (proton radiation therapy) radiation.
  - Proton therapy allows for more precise targeting of the intended brain tissue than photon therapy, so it is becoming more frequently used in pediatrics to minimize cognitive deficits.[12]
- Potential functional risks associated with radiation therapy include the following:
  - Neurocognitive deficits
    - Neurocognitive deficits are inversely related to age at the time of irradiation exposure to the brain.[13]
      - Supratentorial irradiation has higher risk of deficits than infratentorial irradiation.
    - Full brain radiation of 30 to 36 Gy in infants and young children is associated with severe cognitive deficits.
    - Children below the age of 4 are at the highest risk of neurocognitive impairment with radiation treatment, so radiation therapy is avoided if possible.[12]
    - Common cognitive deficits include impaired verbal and performance IQ, language development, attention, executive function, and perceptual motor skills.[14]
  - Alterations in cerebrovasculature
    - There is an increased risk of cerebrovascular accident (CVA).[15]
    - When radiation is delivered to areas surrounding the circle of Willis, this can cause moyamoya syndrome.[16]
      - This most often occurs when children are irradiated before 5 years old (especially if they have neurofibromatosis type 1).
      - This is most frequently seen at a median of 3 to 4 years post therapy.[15]
  - Radiation necrosis[17]
    - This can occur as soon as 6 months of therapy.[2]
    - Presentation includes nausea/vomiting, new-onset focal neurologic deficits, and headache.
    - Treatment includes surgery, steroids, and bevacizumab.[17]
  - Musculoskeletal issues: pathologic fractures,[2] avascular necrosis,[2] scoliosis, and kyphosis[2]

- Hematopoietic stem cell transplant
  - Stem cell transplants derive from cells within the bloodstream. Bone marrow transplants derive from cells within the bone marrow.
  - There are three types: autologous (the patient is the donor), allogenic (the donor is genetically similar to the patient), and umbilical cord.
  - Functional risks include the following:
    - Graft versus host disease (GVHD)
      - This is more likely to occur with allogenic transplant.[18]
      - Acute GVHD occurs 2 to 5 weeks posttransplant and 40% to 50% of patients' symptoms improve with glucocorticoid treatment.[18]

        - Chronic GVHD develops 3 or more months posttransplant and may require a 3-year treatment course of immunosuppressive agents.[18]
            - Presentation includes but not limited to lichenoid and scleroderma skin lesions, joint contractures, and arthritis.
            - Treatments for skin changes include contract-release techniques, splinting, serial casting, orthotics, and home exercise program.
    - Participation restrictions
    - Modality limitations
- Molecularly targeted therapies
    - These therapies target a genomic alteration that is unique to cancer cells and is not present in normal cells.
        - There is reduced toxicity when compared with chemotherapy agents.[2]
        - Most are still paired with chemotherapy agents.[19]
    - There are fewer options for children compared with adults.
    - Functional risks include the following
        - Impaired bone development, which includes stunted growth, osteonecrosis, and widening growth plates[19]
- Immunotherapies
    - Immunotherapies involve administration of T cells molecularly altered to target antigens unique to cancer cells.[20,21] One example is CAR-T (chimeric antigen receptor T) for B cell ALL.
    - Functional risks include the following
        - Neurocognitive deficits including CAR-T cell-related encephalopathy syndrome (CRES) or immune effector cell-associated neurotoxicity syndrome (ICANS)[22,23]
            - *Symptoms in children:* delirium, headache, tremor, decreased level of consciousness, seizure, hallucinations, visual changes, language disturbance, abnormal movements, ataxia, focal weakness, hydrocephalus, and intracranial hemorrhage[23]

Ongoing Care

Cancer rehabilitation-focused diagnoses encountered during the acute phase:

- Neurologic motor deficits secondary to cortical or motor tract involvement
    - Tumor encroachment and/or surgical disruption of motor centers, or connections among the spinal cord, brainstem, cerebellum, and cerebral hemispheres can be persistent and associated with poor motor performance.
    - Tumor location (Table 24.1) impacts specific neurologic and functional deficits, in addition to tumor size, anatomical comorbidities (hydrocephalus), age of the child, and effects of multimodal treatments.

- Overlapping impairment profiles suggest that rehabilitation interventions for CNS tumors can be approached similarly to other acquired CNS injuries.

- Peripheral neuropathy or CIPN
  - CIPN can be caused by vinca alkaloids (vincristine, vinblastine), taxanes (paclitaxel), and platinum (cisplatin, carboplatin).
  - Symptoms worsen with cumulative exposure.
  - Occurrence is mostly acute; however, symptoms can continue months after treatment ("coasting").[24] In some cases, symptoms can also continue for years or can be permanent.
  - Symptoms include dysesthesias, neuropathic pain, distal weakness, difficulty with fine motor tasks, foot drop, and impaired mobility.
  - Treatment includes splinting/orthotics, desensitization techniques, Kinesio Taping, and medications for neuropathic pain (gabapentin, pregabalin, duloxetine).
- Myopathy
  - Myopathy can be secondary to steroid use (high-dose or prolonged course) or less commonly to chemotherapeutic agents (vincristine).
  - Symptoms include proximal muscle weakness, difficulty transferring from sitting to standing position, difficulty ascending the stairs, and impaired overhead activities.
  - Steroid-induced myopathy is more likely to occur with dexamethasone and is reversible when the dose is reduced or discontinued.[25,26]
- Skeletal complications
  - Acute complications can include avascular necrosis, fracture, and osteopenia.
- Cardiac and pulmonary toxicity
  - This may affect exercise tolerance, endurance, and mobility.
  - Therapy may focus on energy conservation techniques.
- Neurocognitive deficits
  - Speech and language pathology consultations are beneficial when screening for baseline deficits and treating any loss of function.
  - Recommend a neuropsychological evaluation at baseline and at intervals posttreatment to assess for deficits in diagnoses/treatments with increased risk.
  - Assess school performance at baseline to better detect deficits during or after treatments. School services consultation should be completed to address accommodations, such as a 504 plan or an individualized education plan (IEP).
  - Consider pharmacologic interventions for attention deficits.
  - Common cognitive deficits associated with brain irradiation include impaired verbal and performance IQ, language development, attention, executive function, and perceptual motor skills.[14]
- Chemotherapy-induced nausea and vomiting (CINV)
  - CINV is more likely to occur in children, younger adults, and those with history of anxiety and motion sickness.[27]
  - Treatment includes serotonin antagonists, benzodiazepines, corticosteroids, and olanzapine.

- Complementary therapies may include ginger, acupuncture, transcutaneous electrical nerve stimulation (TENS), hypnosis, distraction techniques, and systemic desensitization.[28]

- Nutritional complications
  - Nutritional side effects of treatment include the following: taste change/loss, thick saliva/dry mouth, diarrhea, nausea and vomiting, constipation, stomatitis, mucositis, dysphagia, fatigue, chylous effusion, neutropenia, weight gain, anorexia, and cachexia.[29]
  - Involvement of nutrition and speech and language pathology services is integral to the assessment and treatment.
  - Treatment varies depending on the diagnosis. The following are nonpharmacologic treatment suggestions:
    - *Treatment for changes in or loss of taste:* food with strong flavors, tart or spicy foods, visually appealing meals, and home-cooked or favorite food.[29]
    - *Treatment for nausea/vomiting:* eating frequent, small amounts of food; drinking fluid at least 1 hour before or 1 hour after eating; eating and drinking slowly; avoiding sugary, fried, or fatty food; and avoiding food with strong odors[29]

Rehabilitation Considerations in Acute Care

- Consider the Dietz model of cancer rehabilitation as patients may be in either preventive or restorative phase during their acute treatment phase.
- Many patients will demonstrate impaired participation in therapies.
  - Consider therapy cotreatment with music therapy, pet therapy, or interventions from child life services.
  - Consider pretreatment medication prior to therapies that have multiple treatment effects (e.g., lorazepam and olanzapine can provide benefit for nausea and agitation/anxiety).
- Communication with oncology to discuss treatment roadmap can assist with therapy recommendations with regard to frequency of services offered and timepoints at which an inpatient rehabilitation program might be beneficial.
- Pediatric cancers and their treatments are associated with myelosuppression, and abnormalities (anemia, leukopenia, leukocytosis, and thrombocytopenia) should be monitored so that therapeutic activities can be adjusted appropriately and interventions (e.g., transfusions) can be prescribed as directed.
  - Identifying parameters for transfusion, often in collaboration with the pediatric oncologist, is essential.
- Consider implementing multidisciplinary rounds with the therapy team for a team-based approach and continuity of care.
- Modalities may be beneficial for pain and improvement of functional deficits.
- Recommendations of rehabilitation intervention(s) and frequency may be affected by acute versus chronic functional changes related to treatment(s).

- Osteopathic manipulative treatment may be a supportive treatment option for families and patients.[30]
- Consider referral to complementary services for a multidisciplinary approach.
  - These include palliative care, pain management, psychology, psychiatry, music therapy, massage therapy, pet therapy, osteopathic manipulative therapy, child life therapy, neuropsychology, and ethics.

Survivorship

- While the role of PRM is critical in the acute setting with new-onset deficits, it is simultaneously important to have physiatrist involvement longitudinally in the management of long-term deficits from cancer and its treatment(s).
- Cancer therapy at an early age can cause complications that may not be apparent until years later as the child matures, hence the term "late effect" for late-occurring or chronic outcomes, either physical or psychological, that persist or develop beyond 5 years from the diagnosis of cancer.[31]
  - The Children's Oncology Group (COG) has developed risk-based, exposure-related guidelines (*COG Long-Term Follow-Up Guidelines for Survivors of Childhood, Adolescent, and Young Adult Cancers*[32]; www.survivorshipguidelines.org).[32]
    - The organization of these guidelines is based on the treatment that the child has been exposed to as a result of their diagnosis.
    - A complementary set of patient education materials, known as "Healthy Links," accompany the guidelines in order to enhance patient follow-up visits and broaden their application.
- The volume of potential late and long-term deficits that must be screened for in childhood survivors requires a multidisciplinary approach, including the expertise of a pediatric physiatrist.
- Childhood cancer survivors are at risk of frailty, a physiologic phenotype typically seen in older adults.[33]
  - Poor cardiovascular reserve, muscle weakness, poor nutrition, poor endurance, and slowness in walking constitute this frailty phenotype, and frailty in childhood cancers is associated with subsequent chronic health conditions and mortality.[31]

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Pain Management

25

ANDREW B. COLLINS and KENDRA J. HOMAN

GENERAL PRINCIPLES

Definition

Pain is defined by the International Association for the Study of Pain (IASP) as an "an unpleasant sensory and emotional experience associated with, or resembling that associated with, actual or potential tissue damage."[1] Pain is typically classified as either acute or chronic.

- *Acute pain*: an adaptive biological purpose to alert one to internal or external damage or dysfunction (e.g., injury, disease, or inflammation)[2]
- *Chronic pain:* persistent or reoccurring pain that lasts longer than typical healing times, often 3 to 6 months[3–5]

Chronic pain may arise from no recognizable purpose or last beyond the original associated purpose (e.g., alerting to an injury or disease; preventing ongoing trauma; signaling recovery from surgery).[2] A recent IASP Task Force and the International Classification of Diseases (ICD-11) have both recognized chronic pain as a disease itself, with further classification based on etiology and location.[4,6,7] The ICD-11 definition of chronic primary pain is pain lasting longer than 3 months, associated with emotional and/or physical dysfunction, with symptoms that are not better accounted for by another medical condition.[4]

Epidemiology

- Pediatric chronic pain affects between 11% and 38% of children and adolescents, with approximately 5% suffering from severe chronic pain that interferes with daily functioning.[8,9]
- The prevalence is higher in adolescent females, older adolescents, and patients of lower socioeconomic status (SES).[8] Updated prevalence is currently being calculated based on data published between 2009 and 2020.[10]
- The most common forms of pediatric chronic pain include headache (8%–83%), abdominal pain (4%–53%), back pain (14%–24%), musculoskeletal pain (4%–40%), multiple pains (4%–49%), and other pain (5%–88%).[8]

Classification

Chronic pain conditions are classified in the ICD-11 based on the recommendations from the IASP and the World Health Organization (WHO), first differentiating between chronic primary and chronic secondary pain syndromes. The classification system recognizes some conditions can fall into multiple categories, noting that definitions of such conditions should be consistent throughout the classification system.[6,7,11] An initial differential diagnosis for chronic pain in an anatomical location should include primary and secondary pain conditions. Therefore, an appropriate evaluation should be completed for both types of painful conditions in that anatomical location.

Chronic Primary Pain

Chronic primary pain is a painful condition that persists or recurs for longer than 3 months, is associated with emotional distress or functional disability, and is not better accounted for by another diagnosis.[6] Subtypes of chronic primary pain include[6]:

- Chronic widespread pain
- Complex regional pain syndrome (CRPS)
- Chronic primary headache and orofacial pain
- Chronic primary visceral pain
- Chronic primary musculoskeletal pain

Chronic Secondary Pain

Chronic secondary pain evolves from a symptom of an associated diagnosis to a distinct problem that continues beyond treatment of the initial cause.[7]

- Chronic cancer-related pain
- Chronic postsurgical and posttraumatic pain
- Chronic neuropathic pain
- Chronic secondary headache or orofacial pain
- Chronic secondary visceral pain
- Chronic secondary musculoskeletal pain

Etiology

While the exact mechanism and etiology of chronic pain are not clearly understood, there are multiple theories and models to help understand how it may develop.

- *Gate control theory:* The gate control theory describes nociceptive signal propagation along the peripheral fibers and the ascending spinal fibers to the brainstem, where signals are amplified or diminished before reaching the brain.[12] The brain's role is passive in the gate control theory.

- *Neuromatrix model:* The neuromatrix model is a conceptual framework that describes a complex network of signals from the central and peripheral nervous systems, along with the hypothalamicpituitary axis. This network incorporates peripheral afferents and cognitive aspects of pain, including memory, fear, emotional responses, and associations with suffering or reward. Signals can be influenced by genetics and past pain experiences, helping to explain variability in the peripheral nociceptive system and in pain perception.[13]

The neuromatrix model aligns with the biopsychosocial model of chronic pain and pain-associated disability, supporting the relationship between pain, anxiety, and parental factors.[14,15]

- *Central sensitization:* Stimulating peripheral nociceptors, even briefly, can cause prolonged hyperactivity in the central nervous system, manifesting as reduced pain thresholds, hyperalgesia, and allodynia. When this occurs, there are changes in gene expression in receptors, channels, and signaling pathways causing increased excitability and decreased inhibition.[16]

DIAGNOSIS

Risk Factors

While the pathways from acute pain to chronic pain are not well-understood, there are several risk factors associated with pediatric chronic pain. The most studied patient-related risk factors include comorbid psychopathology, heightened pain-related fear, pain catastrophizing, the subjective experience of stress/stressful life events, maladaptive or passive coping, and poor sleep.[17–21] Additionally, research has also examined several parent and family risk factors such as parental psychopathology, family pain history, parental pain catastrophizing, and poor social functioning outside of the family that may also contribute to increased risk.[22]

The Pediatric Pain Screening Tool (PPST) can be used to stratify risk and guide treatment in children with chronic. This tool includes nine items asking about comorbid pain, ambulation, school, sleep, catastrophizing, fear, anxiety, depression, and how much pain bothers the child.[23]

Clinical Presentation and Diagnostic Evaluation

Evaluation for primary pain disorders includes a comprehensive history and physical examination, as well as evaluation for causes of chronic secondary pain in the same region.

Juvenile Fibromyalgia

Juvenile fibromyalgia is a chronic widespread primary pain disorder characterized by musculoskeletal pain, fatigue, nonrestorative sleep, and cognitive difficulties, as well as associated symptoms of headache, abdominal discomfort, and depressed mood. The diagnosis of juvenile fibromyalgia borrows the diagnostic criteria for the adult population.[24]

The most recent diagnostic criteria for fibromyalgia were developed in 2010 and revised in 2016.[25,26] The 2010 criteria have been validated in an adolescent population.[24]

Fibromyalgia diagnosis can be made regardless of other diagnoses and does not exclude other clinical diagnoses. Specific criteria include[26]:

- Widespread Pain Index (WPI) ≥7 and Symptom Severity Scale (SSS) ≥5, or WPI from 4 to 6 and SSS ≥9
- Pain present in four out of five regions
- Symptoms present for 3 months or more at a similar level

Details of these criteria are shown in Exhibit 25.1.

EXHIBIT 25.1 DIAGNOSTIC CRITERIA FOR JUVENILE FIBROMYALGIA

Exhibit 25.1A General Criteria

The patient must meet all three of the following:

- Widespread Pain Index (WPI) ≥7 and Symptom Severity Scale (SSS) ≥5, or WPI from 4 to 6 and SSS ≥9 (Exhibit 25.1B and Exhibit 25.1C)
- *Generalized pain:* pain present in four out of five regions (left upper, right upper, left lower, right lower, and axial, as noted on the WPI)
    - Symptoms present for 3 months or more at a similar level

Exhibit 25.1B Widespread Pain Index

LEFT UPPER REGION	RIGHT UPPER REGION	AXIAL REGION
Jaw*	Jaw*	Neck
Shoulder girdle	Shoulder girdle	Upper back
Upper arm	Upper arm	Lower back
Lower arm	Lower arm	Chest*
		Abdomen*
LEFT LOWER REGION	**RIGHT LOWER REGION**	
Hip	Hip	
Upper leg	Upper leg	
Lower leg	Lower leg	

One point for each of the areas in the table in which a patient has had pain in the last week (0–19).

*Jaw, chest, and abdomen do not count toward the generalized pain criterion.

Exhibit 25.1C Symptom Severity Scale

Patient Rates Severity of the following Symptoms over the last week from 0 (no problem) to 3 (severe, pervasive problem). This portion is 0 to 9 points.	Patient rates whether they have been bothered by each of the following symptoms in the last 6 months. This portion is 0 to 3 points.
Fatigue	Headaches
Waking unrefreshed	Pain or cramps in lower abdomen
Cognitive symptoms	Depression

The Symptom Severity Scale score is the sum of the two scores in the table, from 0 to 12.

Complex Regional Pain Syndrome

CRPS is a chronic primary pain disorder describing localized or regional neuropathic pain characterized by pain disproportionate to the inciting event, along with some combination of skin and temperature changes, edema, changes in hair or nail growth, or motor dysfunction.[27,28]

CRPS type I and type II are distinguished by absence (I) or presence (II) of an identified nerve injury. Of note, CRPS type II is classified as both a primary and a secondary pain disorder.[6,7]

There are no validated criteria to diagnose CRPS in children and adolescents. The Budapest criteria are preferred for use in adults and have been validated in this population.[28,29] These criteria should not be strictly followed in children, but can be used to guide diagnosis, particularly among providers with less experience in diagnosing or treating pediatric CRPS.[27]

The Budapest criteria are as follows:

- Continuing pain disproportionate in duration and severity to the inciting event
- One symptom reported by the patient in three out of four categories (sensory, vasomotor, sudomotor/edema, and motor/trophic)
- One sign observed by the clinician in two out of four categories (sensory, vasomotor, sudomotor/edema, and motor/trophic)
- No other diagnosis that can better explain the symptoms

Other Primary Pain Disorders

There are diagnostic criteria for other primary pain disorders, including for chronic primary headache[30] and chronic primary abdominal pain.[31,32]

Joint Hypermobility

Joint hypermobility is not a primary pain diagnosis but is associated with chronic pain. Hypermobile joints increase susceptibility to recurrent musculoskeletal pain.[33]

Hypermobility spectrum disorders (HSDs) include joint hypermobility and secondary symptoms limited to the musculoskeletal system, such as pain. These can only be diagnosed in the absence of other conditions associated with hypermobility. HSDs include generalized HSD, peripheral HSD (limited to hands and feet), localized HSD (limited to a single joint or group of joints), and historical HSD (with a negative Beighton score currently).[33]

The diagnostic criteria for hypermobile EhlersDanlos syndrome (hEDS), as well as the overall criteria and classification for Ehlers–Danlos syndrome, were revised in 2017. hEDS is a clinical diagnosis inherited in an autosomal dominant pattern but without specific genetic tests to confirm diagnosis.[34]

The clinical diagnosis of hEDS requires simultaneous presence of three criteria[34]:

- *Criterion 1:* The patient should have generalized joint hypermobility: Beighton score ≥6 for prepubertal children, ≥5 from puberty to age 50, and ≥4 for adults over the age of 50.
- *Criterion 2:* The patient should have two or more of the following three features: nonmusculoskeletal systemic signs, family history of hEDS in a first-degree relative, and musculoskeletal complications (recurrent pain, chronic pain, joint dislocations, or frank joint instability).
- *Criterion 3:* Other appropriate diagnoses have been excluded. For patients with acquired connective tissue disorders, such a rheumatoid arthritis, musculoskeletal complications cannot count toward criterion 2.

Beighton score[34,35]:

- Left and right fifth metacarpal-phalangeal joint hyperextension more than 90° (one per side)
- Left and right thumb passively moved to touch the ipsilateral forearm (one per side)
- Left and right elbow hyperextension more than 10° (one per side)
- Left and right knee hyperextension more than 10° when standing (one per side)
- Ability to bend forward and place total palm of both hands plant on floor in front of the feet (one point)

TREATMENT

Guiding Principles

Traditional biomedical interventions for chronic pain typically do not result in substantial or sustained pain reduction for most patients, lack long-term benefit, and pose a risk of negative side effects.[36–38] Current treatment approaches conceptualize chronic pain as a biopsychosocial problem resulting from the complex interaction of biological, psychological, and sociocultural factors,[38,39] with treatment focusing on increasing adaptive functioning (e.g., functioning needs to improve before pain can improve).[40] One of the most important aspects of treatment is initial feedback with a child and their family. During this feedback, it is important to understand expectations, validate symptoms, provide a diagnosis with education, and emphasize the role of multidisciplinary care.[41]

Ongoing Care

Outpatient Treatment

Outpatient treatment consists of a combination of medication management, psychological treatment, and physical/occupational therapy to address the interplay of factors that contribute to chronic pain.[42]

Inpatient or Day Hospital Treatment

While many pediatric pain patients benefit from comprehensive outpatient treatment, a subset continue to functionally decline.[43–45] These nonresponders can require intensive interdisciplinary pain treatment (IIPT), where the biopsychosocial needs are met simultaneously. Typically, IIPT consists of three or more integrated healthcare disciplines concurrently delivering treatment focused on functional rehabilitation through an inpatient or day hospital setting.[46,47] Evidence for the effectiveness of IIPT programs is growing, with results demonstrating improvements in pain severity, pain-related disability, school attendance, and emotional functioning at posttreatment and short- and long-term follow-up.[48–50]

Medications

Medication use in pediatric pain is typically off-label, with recommendations from adult studies, expert opinion, and dosing for other conditions.[51,52] Despite lack of strong evidence, children with chronic pain have high use of prescription and over-the-counter medications.[51,53–55] To reduce the risk of side effects, medications for chronic pain should be started at a low dose and increased slowly.

See Table 25.1 for a summary of common medications, indications, and side effects.

- Acetaminophen
  - Acetaminophen is used for fever reduction and as a first-line treatment for acute and chronic pain, although the mechanism of action is not fully understood.[51,52,55,56]
  - It carries a risk of hepatic injury, which can be avoided by adhering to recommended maximum daily dosing.[57]
- Nonsteroidal anti-inflammatory drugs (NSAIDs)
  - NSAIDs (e.g., ibuprofen, naproxen, diclofenac, and ketorolac) treat musculoskeletal pain, abortive treatment of migraines, and pain in the setting of juvenile idiopathic arthritis.[51,52]
  - Common side effects include gastrointestinal effects, kidney injury, bleeding disorders, and hypertension. Selective cyclooxygenase-2 inhibitors have decreased gastrointestinal ulcers and decreased platelet inhibition.[51,52]
  - These have equal efficacy and safety to acetaminophen for short-term use.[58] There is insufficient evidence to comment on the efficacy or safety for chronic pain.[59]
- Antiepileptics
  - Gabapentin and pregabalin are commonly prescribed for chronic pain, including widespread and neuropathic pain syndromes.[60–63]

Table 25.1 Medications Commonly Prescribed for Pediatric Pain

CLASS	MEDICATIONS	INDICATIONS	SIDE EFFECTS
Acetaminophen	Acetaminophen	Acute pain, adjunctive treatment	Hepatic injury
NSAID	Ibuprofen, naproxen	Acute pain, inflammatory conditions, abortive headache treatment	GI effects, kidney injury, bleeding disorders, hypertension
NSAID, COX-2 inhibitor	Celecoxib		Same as other NSAIDs with less GI ulcers and less platelet inhibition
Anticonvulsants	Gabapentin, pregabalin	Neuropathic pain, widespread pain, primary pain disorders	Somnolence, altered mood, weight gain
	Topiramate	Headache prophylaxis	Somnolence, altered mood, cognitive effects, weight loss
Antidepressants	Amitriptyline and other TCAs	Neuropathic pain, headache prophylaxis, abdominal pain, widespread pain, primary pain disorders	Somnolence, altered mood, dry mouth, prolonged QTc, risk of serotonin syndrome
	Duloxetine	Neuropathic pain, widespread pain, primary pain disorders	Altered mood, nausea, risk of serotonin syndrome
Muscle relaxants	Methocarbamol	Myofascial pain, spasmodic pain	Sedation, nausea
	Cyclobenzaprine	Myofascial pain, spasmodic pain, widespread pain	Sedation, risk of serotonin syndrome
	Tizanidine	Myofascial pain, spasmodic pain, pain secondary to spasticity	Sedation, dizziness, hepatotoxicity
	Baclofen		Sedation

COX-2, cyclooxygenase-2; GI, gastrointestinal; NSAID, nonsteroidal anti-inflammatory drugs; TCAs, tricyclic antidepressants.

- Common side effects include somnolence, weight gain, and mood alterations.[60–62] Patients should be educated about the small risk of suicidal ideation as well.[64]
- Topiramate is used for pediatric migraine, although its efficacy is recently shown to be similar to placebo.[65]
- Oxcarbazepine, carbamazepine, valproic acid, and levetiracetam are also used for chronic pain.[52]

- Antidepressants
  - Tricyclic antidepressants (TCAs), such as amitriptyline, are prescribed for pediatric pain, including neuropathic pain, widespread pain, abdominal pain, and headache.[52,63,66]
  - TCAs have a risk of prolonging QTc interval; some recommend obtaining an EKG prior to initiating.[67] Other side effects include suicidal ideation, morning sleepiness, and dry mouth.[52]
  - Serotonin and norepinephrine reuptake inhibitors (SNRIs), such as duloxetine, are used to treat neuropathic pain and widespread pain in adults.[68] These may be effective in juvenile fibromyalgia as well.[69]
  - Suicidal ideation is a serious risk in the use of SNRIs in adolescents, including when used for pain.[70] Other side effects include nausea and sleep problems.[52]
- Muscle relaxers
  - Muscle relaxers are a diverse group of medications that include antispasmodic and antispasticity medications.[71]
  - Cyclobenzaprine, methocarbamol, metaxalone, and tizanidine are antispasmodics used to treat painful muscle spasms, with most evidence for back pain.[52,71] Cyclobenzaprine has also been studied for use in fibromyalgia.[72]
  - Tizanidine is both an antispasmodic and an antispasticity medication. Other antispasticity medications, such as baclofen, are sometimes used for painful musculoskeletal conditions, although not as first line.[52,71]
  - Methocarbamol and metaxalone are thought to be less sedating.[52]
  - Cyclobenzaprine has a risk of contributing to serotonin syndrome.[73]
  - Benzodiazepines can be used for acute muscle relaxation, but should typically not be used chronically due to significant risk of sedation, respiratory depression, and abuse.[52]
- Opioids
  - Opioids should typically not be used for chronic primary pain in children.[52,74] In circumstances deemed appropriate, prescription should only occur as part of a structured treatment plan that includes screening and monitoring. Guidelines exist for prescribing opioids to adults, but not children.[75,76]
  - Methadone is a long-acting opioid agonist and N-methyl-D-aspartate receptor (NMDA) antagonist.[77] It is helpful in treating chronic cancer pain and in weaning off of other opioids in children, but has variable pharmacokinetics and significant side effects.[52] It should be prescribed by physicians with appropriate expertise.

- Other medications
  - Additional classes of medications can also be used for specific pain conditions. Low-dose naltrexone has emerging evidence for use in chronic pain.[78] Medications targeting calcitonin gene-related peptide have been introduced for daily control and acute use in headache.[79–81]

Procedures

Evidence for procedural management in pediatric patients with chronic pain is limited.[53,82] Epidural injections for back pain or radiculopathy, joint injections for focal joint pain, celiac plexus blocks for abdominal pain, sympathetic blocks for complex regional pain syndrome, trigger point injections for myofascial pain, and peripheral nerve injections for other regional pain are sometimes incorporated into multidisciplinary care.

There is some controversy over the potential relationship between procedural interventions and IIPT programs. Procedural intervention has been used successfully during IIPT.[83] IIPT without procedural intervention has also been used successfully for patients who have not improved with prior procedural intervention.[84]

Psychology

Psychological treatment has been studied more than other treatment modalities in pediatric pain.[53] The most efficacious psychological treatment for chronic pain is cognitive behavioral therapy (CBT).[85] Typically, CBT begins with psychoeducation regarding pain neurobiology (including the gate control theory of pain, the biopsychosocial model of pain, and the disability cycle[86]) to provide the rationale for therapy, build rapport, and enhance motivation and engagement. CBT then focuses on altering maladaptive behavioral responses to pain, challenging negative cognitions related to pain, and improving pain coping skills through relaxation training, distraction techniques, and activity pacing to increase activity but prevent overexertion.[85] While research is very supportive of CBT for chronic pain, the intractable nature of chronic pain had led to the application of mindfulness-based stress reduction (MBSR) and acceptance and commitment therapy (ACT) to chronic pain, specifically focusing on acceptance of chronic pain while engaging in valued activities when attempts to control pain prove ineffectual.[85]

Physical and Occupational Therapy

Function-focused treatment is important as disability improves before pain intensity.[40] Treatment should emphasize exercise, which has long-standing support in pediatric pain treatment.[87,88] Individual recommendations for physical and/or occupational therapy depend on the diagnosis and needs of the patient.

Physical therapists can emphasize cardiovascular conditioning, aerobic exercise, and biomechanical assessments[42,89–91] to promote normalized movement, posture, and function. Occupational therapy can also be

helpful in functional training, posture, biomechanics, and pacing.[89] There is some evidence for physiologic effects of modalities such as transcutaneous electrical nerve stimulation and dry needling, although these are not pediatric-specific.[92–94] Therapy disciplines should focus on facilitating a self-management approach to promote independence in the presence of pain rather than waiting for the pain to improve before advancing.

Treatment Controversies

It is important to remember that evidence for pharmacologic treatment of chronic pediatric pain is limited for all medications, not just for medications considered controversial.

Ketamine

Ketamine is an intravenous or intranasal NMDA antagonist that has been studied for use in multiple pain conditions, including CRPS, widespread pain, postoperative pain, vaso-occlusive episodes in sickle cell disease, and cancer-related pain.[95–100] Side effects, such as hallucinations, are typically of low frequency at doses for pain. While there is some support for short-term improvements, evidence is lacking for long-term effects.[96,99] One significant limitation of ketamine is the need to administer the medication in a healthcare environment, which shifts focus away from the biopsychosocial model, independent management, and functioning.

Marijuana-Derived Products

Medical use of marijuana has been increasing in the United States, including in areas without legalized use.[101] As of May 2021, medical marijuana is permitted in 36 states and four territories; nonmedical use is permitted in 18 states, two territories, and the District of Columbia.[102] There is limited evidence on relief of neuropathic pain, primarily in adults.[103] Studies on use of medical marijuana in children have focused more on epilepsy and use within oncology, with a need for more evidence globally.[104,105]

Controversies include variable oversight and dosing, federal legal status, cognitive side effects limiting participation in rehabilitative treatment, and potential associations with adverse neuropsychiatric outcomes.[106–109] Neuropsychiatric risks include risk of developing schizophrenia and of neuropsychological decline, although there is debate on the directionality of these associations.[110–112] Adolescent use is particularly controversial, including in the normalization of drug use.[109,113] In animal models, there is also concern that use in adolescents interferes with the role of the endocannabinoid system in neurologic development.[114]

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Functional Neurologic Disorders

26

GADI A. REVIVO and DIANE K. AMSTUTZ

It is much more important to know what sort of a patient has a disease than what sort of a disease a patient has.—Sir William Osler

GENERAL PRINCIPLES

Functional somatic symptom (FSS) disorder is an umbrella term that includes functional symptoms that occur across multiple body systems and involve disturbances in neurophysiologic regulation. These include diagnoses such as functional neurologic symptom disorders (FNSD), functional gastrointestinal disorders, chronic pain syndromes, chronic fatigue syndrome, and dysautonomia.

These disturbances are often the result of stress, either physical, psychological, or both. The brainbody stress systems involved in pain and emotion are interconnected. Activation of any part of this system through physical or psychological threat triggers the body's stress response. When the stress system is activated too much or for too long, functional symptoms may appear. In this chapter, we outline the best ways to communicate with patients and families about these functional disturbances. The role of interdisciplinary treatment, involving physical and occupational therapy, cognitive behavioral therapy, and mindfulness interventions, is integral to successful recovery, with guidelines for delivery of care outlined in this chapter.

Definition

The terminology used to describe physical symptoms for which there is no diagnosable organic pathology has evolved over time. Initially labeled *hysteria*, this term was eliminated from the *Diagnostic and Statistical Manual of Mental Disorders (DSM)* in 1968. Subsequent terms such as *somatization disorder, hypochondriasis*, and *conversion disorder* have also fallen out of favor. In 2013, *functional neurologic symptom disorder* (FNSD) was adopted into the *DSM-5*.[1] By definition, this syndrome involves atypical neurologic symptoms that do not correspond to a known neurologic disorder. Kozlowska et al.[2] and others[3] utilize the nosology *functional somatic symptoms (FSS)* as an umbrella term that includes functional symptoms that occur across multiple physiologic systems and involve disturbances in neurophysiologic regulation. These include diagnoses such as FNSD, functional

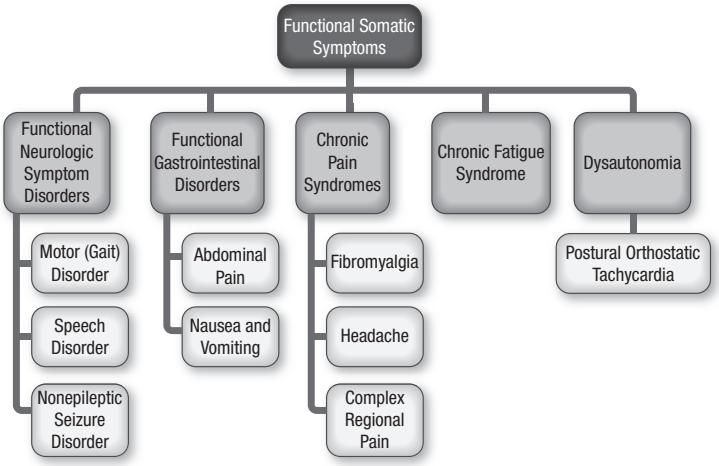

Figure 26.1 Functional somatic symptoms.

gastrointestinal disorders, chronic pain syndromes, chronic fatigue syndrome, and dysautonomia (including postural orthostatic tachycardia syndrome; Figure 26.1).

Epidemiology

The incidence of functional disorders in the pediatric population is uncertain. Estimates suggest:

- 20% of psychiatric and neurologic patients are impacted by functional disorders.[1]
- Functional gastrointestinal disorders are likely the most common (10%87%) in children.[4]
- 20% to 46% of children experience chronic pain.[5]
- .1% to 1% of adolescents report chronic fatigue.[6]
- Postural orthostatic tachycardia syndrome affects .1% to 1% of the U.S. population, mostly impacting females.[7]

Classification

Etiology

Kozlowska has proposed a stress system model to explain FSS.[8] In this model, functional symptoms are a result of disturbances in neurophysiologic regulation. These disturbances of over, under, or aberrant activation are the result of stress, either physical, psychological, or both. The brain-body stress systems—the hypothalamicpituitaryadrenal (HPA) axis, the immune-inflammatory system, the autonomic nervous system, and the brain regions involved in pain and emotionare all interconnected.

Activation of any part of this system through physical or psychological threat triggers the body's stress response and results in stress-induced changes in the brain.[8] When the stress system is activated too much or for too long, functional symptoms may appear.[2] In a study of the impact of adverse childhood experiences (ACE) on medical symptoms, ACE were reported more frequently by adult patients diagnosed with FNSD, and these patients also experienced more alexithymia (reduced perception and expression of negative emotion) than healthy controls.[9] However, the stress that can trigger FSS does not have to be extreme. In fact, the DSM-5 dropped psychological stress as a requirement for diagnosis.[10] Many patients present with common stressors, including perfectionism with school-related tasks, intense athletic demands, family conflict, social pressures, or bullying at school. These events may occur serially and be preceded by viral illness or a minor injury. The cumulative effect of events—the *repeated* experience of stress—is more commonly associated with the development of functional symptoms than the existence of extraordinary stressors.[2]

Pathophysiology

The stress system model of FSS notes that the body does not distinguish between physical and emotional stressors. It responds to all stressors by upregulating the stress system (a condition commonly referred to as *fight or flight*). The HPA axis is activated by stress and produces the stress hormone cortisol. This hormone provides the body with energy to combat stress, but can also disturb sleep. The autonomic nervous system regulates the arousal response of all the organs in the body. It can cause an increase in heart rate (HR), nausea, changes in bowel function, sweatiness, temperature dysregulation, and rapid breathing that can lead to dizziness. When brain stress systems become overactive, this can cause disruption in motor functioning and sensory processing.[2] It can also cause a change in neurochemical balance that results in the brain being overly sensitive to incoming stimuli, by upregulating pain pathways and facilitating an increase in the number of pain messages. This process is called central sensitization.[11] In addition, this upregulation of the nervous system can cause disruption in the circadian clock, which regulates sleep, and can result in lack of restorative sleep. This impacts pain and allows ongoing activation of the stress system. Lastly, the immune-inflammatory system can keep pain nerves activated. Exercise helps this system return to healthy functioning by regulating macrophages that secrete anti-inflammatory molecules modulating pain signals.[2] The goal of treatment is to design interventions that move the stress system from overactivated to regulated, promoting healthier functioning.

DIAGNOSIS

Risk Factors

- Childhood adversity[12]
- Attachment insecurity driven by maternal insensitivity to a child's signals of distress and the inability to set limits

- Patterns of attachment that cause children to avoid awareness of distress or need for comfort[8]
- Use of fear, anger, and helplessness to coerce attention from attachment figures who have seemed unreliable or inconsistent[8]
- Persistent anxiety in the attachment relationship, along with trauma or loss, which may activate the body's arousal system, leading to functional disorders

Clinical Presentation

Functional diagnoses are based on specific, observable symptoms of a patient's presentation and are not a "last resort" conclusion. The earlier the diagnosis is agreed upon and the treatment begins, the better the outcome.[2,11] This can also save costly and often painful medical testing that provides no additional insight into the patient's condition. The criteria for diagnosis of FNSD are found in the *DSM-5*[10]:

- One or more symptoms of changes in voluntary motor or sensory function
- Clinical findings that provide evidence of inconsistency between symptoms and diagnosable medical conditions
- Symptoms not better explained by another disorder that is medical or mental health-related
- Symptoms that cause significant distress or impairment in social, occupational, or other important areas of functioning or that need medical evaluation

The Rome IV guidelines provide the criteria for diagnosis of functional gastrointestinal disorders.[13] The diagnostic Rome IV criteria for functional abdominal pain must be fulfilled for at least 2 months before diagnosis, must be met at least four times per month, and include all of the following:

- Episodic or continuous abdominal pain that does not occur solely during events such as eating or menses
- Insufficient criteria for other functional gastrointestinal diagnoses, including irritable bowel syndrome, functional dyspepsia, or abdominal migraine
- Abdominal pain that cannot be explained by another medical condition, subsequent to appropriate evaluation

Chronic pain diagnoses are made based on the existence of persistent pain, even in the absence of an organic cause, and may be labeled as central sensitization characterized by[14]:

- Widespread pain and hyperalgesia/allodynia
- Fatigue, low mood and cognitive dysfunction, sleep problems, and sensory hypersensitivity
- Previous exposure to psychological or physical stressors and a personal and family history of pain

- Changes in brain gray matter in the pain processing regions, neurochemical imbalances, and altered resting brain network connectivity between the proprioceptive and antinociceptive brain areas evident on neuroimaging studies
- Possible immune system abnormalities

Postural orthostatic tachycardia syndrome diagnosis requires the following criteria in children[15]:

- Presence of chronic symptoms of orthostatic intolerance (≥6 months) accompanied by an increased HR ≥40 beats per minute within 10 minutes of assuming an upright posture and in the absence of orthostatic hypotension (blood pressure [BP] fall >20/10 mmHg)
- Orthostatic tachycardia occurring in the absence of other overt causes of orthostatic tachycardia (e.g., acute blood loss), medications that impair autonomic regulation, or other chronic debilitating disorders that might cause tachycardia (e.g., anemia, diabetes with known autonomic neuropathy, systemic infectious or inflammatory conditions, hyperthyroidism)
- Evaluation that includes a detailed history and a head-up tilt test

The Institute of Medicine criteria for diagnosis of chronic fatigue syndrome include[16]:

- Each of the following three symptoms at least half of the time, to at least a moderately severe degree:
  - Reduced or impaired ability to engage in previous levels of occupational, educational, social, or personal activities that lasts for >6 months and is accompanied by fatigue, which is often profound, is of new or definite onset (not lifelong), is not the result of excessive exertion, and is not alleviated by rest
  - Postexertional malaise
  - Unrefreshing sleep
- Plus at least one of the two following (chronic, severe):
  - Cognitive impairment
  - Orthostatic intolerance

Symptoms

Functional symptoms are often intermittent in nature and can include the following[17]:

- Auditory impairment
- Impaired movement (giveway weakness, paralysis)
- Visual impairment
- Tic or tremor
- Decreased concentration
- Impaired balance (astasia-abasia)
- Decreased memory
- Speech dysfunction (effortful, dysarthria, dysphonia)
- Swallowing dysfunction

- Seizures (nonepileptic, episodic hyporesponsiveness)
- Fatigue
- Pain

Physical Examination

The physical examination of a patient suspected of exhibiting a functional disorder should start during the history taking process, allowing the clinician to observe the patient's behaviors well before drawing attention to any neurologic inconsistencies often exacerbated by physical examination. Note stereotyped movements that heighten or abate during history taking. A complete neurologic assessment will uncover inconsistencies in motor performance and control, strength or range of motion, and movement disorders that are poorly stereotyped. Look for a mismatch in a patient's effort versus motor function when changing position in bed or participating in a transfer during the examination. Findings of functional weakness (difficulty walking, inability to grasp/release with one or both hands, effortful dysfluent speech patterns) that do not fit an organic physiologic pattern are suggestive features of a functional disorder. Patients may state that they cannot walk in spite of normal motor findings in the lower extremities (LEs). Common clinical tests performed in the LEs include the hip abductor sign: hip abduction weakness in an affected limb that resolves with contralateral hip abduction against resistance of the normal leg. The Hoover sign relies on the crossed extensor reflex where when asked to flex the hip the contralateral hip will extend, even in patients with functional weakness. The telltale signs of a nonphysiologic gait pattern seen on examination in patients with a motor functional disorder include excessive effort with giveway weakness, while lacking a consistent pattern, coupled with astasia-abasia.[11,17]

Patients with findings of upper extremity weakness may often demonstrate inconsistencies in functional completion of activities of daily living (ADLs) compared with their motor deficits, including difficulty with writing. They may lack pronation of the forearm, with a functional drift, and exhibit nonphysiologic variability on sensory examination or frequency of tremors and tics. Speech functional disorders include undifferentiated dysarthria and findings of nonsense syllable production, patterns of inconsistent phonological processes that may include deletion of consonant sounds (fricatives and affricates), altered prosody, and/or nasality of normal voice production.[11,17]

Diagnostic Evaluation

Although there exist certain maneuvers, such as the hip abductor sign or the whack-a-mole sign, to name a few, the "specificity and sensitivity of these maneuvers may be biased" (p. 1113) and there is lack of gold standards with which to compare them.[17] During a patient's workup, the most important elements include a thorough history taking and examination, of which atypical inconsistent neurologic findings most accurately point

toward making a diagnosis of FNSD. It is important to note that prior studies reported the frequency of misdiagnosis to be about 4%, which included a follow-up mean of 5 years.[18]

It is imperative that all pediatric specialties involved in the patient's care come to an agreement on a diagnosis based on physical and psychiatric features.[2] Early identification of a functional disorder, in spite of potential overlapping organic symptoms, can prevent ordering costly, painful procedures that further delay diagnosis and treatment. Psychological services are an integral part of the workup, providing much needed evaluation and treatment of psychosocial stressors and maladaptive family dynamics, mood disorders, and dysregulated sleep.[2,11]

TREATMENT

Guiding Principles

First, it must be explained to both the patient and the parents that the presenting symptoms have a name—a medically based diagnosis—and that specific treatment is needed.[2] It is also helpful to convey that full recovery from the condition is possible.[11] Results of medical tests and physical examinations are noted to be normal, so that the body's structure is intact, but function is impaired. Kozlowska et al. note that it is critical to make clear that the child is "medically safe" (p. 22) and that no further testing and medical or surgical intervention are warranted.[2] Establishing these facts allows treatment to proceed and helps eliminate fears that something has been missed and more testing or intervention is required. While it can be tempting to discuss what the symptoms are not (not cardiac-related, not epilepsy, not paralysis), it is more important to validate the existing symptoms, note how they can be related to physical or psychological stress, or a combination of these, and that there is existing treatment for the symptoms. Families may also be confused because they have received a number of different functional diagnoses from different specialists and are left with the notion that their child is suffering from a combination of different disorders. To deal with this confusion, the notion of FSS can be explained to the patient and family, noting the unifying issues that relate these diagnoses.[2]

Initial Management

Successful outcomes for children with functional disorders start with first delivering effective communication about the diagnosis and treatment, provided by the physician and the treatment team. Symptoms should be validated as real and all members of the treatment team need to coordinate their care.[2] It often takes time for families to shift from seeking a structural diagnosis and medical cure to understanding functional diagnoses and the pathway to improvement. Examples of other functional patients who have recovered following treatment can provide a family with a sense of hope.[11] The role an interdisciplinary treatment team plays in recovery must be conveyed to both the patient and the family.[19] Patients

and families should understand that the best way to move forward is through activity, rather than waiting until symptoms improve. The goal is to promote improvements in functioning, both emotional and physical; interventions are designed to help patients better regulate their stress systems.

Treatment can be provided in an inpatient setting on a time-limited basis if patients require significant physical assistance, but children should be transitioned to outpatient care as soon as feasible, reintegrating them back into the home and community.

Ongoing Care

The goal of treatment provided by the interdisciplinary team is to help the patient learn strategies to manage stress and facilitate better physical functioning. In discussing the principles of treatment, Kozlowska et al.[2] believe it is best to start with interventions that target the body and move on to focusing on the mind and the family system.

The most productive, initial intervention is often to regulate sleep.[2] Patients need to understand that good sleep allows the body to rebuild itself and contributes to improvements in mood. Sleep hygiene rules[20] are reviewed and ways to enforce these are discussed with parents. Over-the-counter medications such as melatonin (no more than 3 mg, taken at least 2 hours before bedtime) or prescription for trazodone can often facilitate good sleep when sleep hygiene alone is not enough.[20]

New research also indicates that the microbiome of the gut can have a significant impact on mood and emotional functioning.[21,22] Nutrition counseling can promote improved health, both physically and mentally, and should be included in early intervention strategies.

It is important for parents to understand the role that their attention to symptoms can play.

- Parents are instructed not to ask about pain or symptoms and to focus on function.
- If children refuse to participate in normal activities such as school attendance or household chores, rewards and consequences should be established and reinforcement(s) for positive engagement should be defined.[23,24] Parents need to act as coaches, supporting the child in using cognitive and physical strategies as ways to manage and control symptoms.
- There are factors in the family interactions that can contribute to the child's symptoms. Parents should seek family counseling as needed to improve communication.[2]
- A child who is continually exposed to abuse or bullying (either at home or school) will not be able to get control of their stress system responses. Establishing a sense of safety is necessary for other interventions to succeed.[2]

Treatment approaches that target the body can reduce an overengaged sympathetic nervous system. These interventions can help a child move

Figure 26.2 Steps to success.

forward when the family is not ready to address other factors or when a child's stress level is so high that cognitive strategies cannot work because executive and problem-solving skills are dysfunctional.[25] These strategies may be the only way forward when children or families are opposed to interventions that are more psychologically based. The goal is to use mindbody strategies to interrupt the sequence and stop the functional symptoms from appearing.[26] These strategies include a mindful body scan,[27] focused breathing interventions,[26] progressive muscle relaxation,[2] and grounding techniques[28] to help the child shift awareness away from the body symptoms.

The focus should be on interventions that improve symptom presentation by establishing a plan for steps to success (Figure 26.2). It is important that the therapist and the patient establish overall goals that they agree upon, with treatment milestones that they can reassess as function improves. Children are told that the steps are valued goals they choose to work on. Each step represents a milestone in recovery. Engaging children in distracting activities—games, dancing, making videos, cooking, or hobbies can result in improvement in their abilities and provide a gateway to recovery.

Motor retraining starts with an assessment of sensation, balance, range of motion, strength, and endurance. Psychologically informed physical therapy helps build a therapeutic relationship by changing maladaptive behaviors into functional movement patterns and improving overall motor planning for normalized ambulation.[2] A paced approach for gradually increasing activity and improving the quality of movement is emphasized over the number of steps taken. The use of adaptive devices is encouraged to facilitate greater initial gains, with a stepwise plan to reduce reliance on such devices over time. Setting targets for increasing aerobic activity and capacity can effectively address deconditioning. It is imperative that parents be empowered as coaches in the process of successfully reinforcing completion of the home exercise program in between treatment visits. In tandem, the occupational therapist

should assess the patient's ergonomics in completing ADLs and evaluate how the patient's posture and body mechanics may impact these activities. Pictures of the patient's room and study area(s) can help facilitate appropriate ergonomic setup for successful completion of academic work and hobbies. Awareness of symptom triggers and attention to pacing must be factored into creating a daily activity schedule. Speech therapy should address dysarthria and associated speech impairments (altered prosody, velar sufficiency, noted disfluencies, and phonological substitutions) by focusing on improving breath support while providing speech strategies for improving articulation and speech intelligibility.[17] Finally, cognitive behavioral strategies play a role in managing nervous system arousal by focusing on deliberate efforts to regulate the dysfunctional thoughts that lead to negative feelings and behaviors and reduce healthy coping strategies.[29]

Catastrophizing is a word used to describe active rumination and excessive magnification of negative thoughts and feelings. These thoughts, when verbalized, can lead to excessive attention to symptoms on the part of both the child and the parents. Palermo et al.[30] noted that parental catastrophizing can further maintain the child's symptoms. Imaging studies indicate that catastrophizing leads to activation of brain stress symptoms and increases the connections between those systems and sensory processing regions.[31] Many children may try to put negative thoughts or memories out of their mind. They may even deny feelings of stress or present with signs of alexithymia.[9] Research demonstrates that putting negative feelings, thoughts, and memories out of mind causes increased activation of the brain stress system and can contribute to FSS. These principles need to be explained to the child and the parents, and ways to safely explore these avoided thoughts and feelings can allow them to be acknowledged and give them less power over the individual's life.[29]

Most important to recovery is the communication from the treatment team that this condition can be overcome. Innovative treatment can help patients discover the goals and the next steps necessary to allow their brain to return to normal neurologic functioning. The power of the placebo can be maximized with attitudes that encourage the expectation of recovery based on the treatment provided. Point out what gains the patient has already achieved and reinforce healthy behaviors. Optimism is the key.[11]

ADDITIONAL CONSIDERATIONS

Little data exist on the extent to which pediatric patients experience functional neurologic disorders (FND) relapse. Studies of adults with psychogenic nonepileptic seizures report that 6 months following treatment about half remained seizure-free.[32] Fobian and Elliott suggest that FND responses become conditioned and that treatment does not cause the responses to be unlearned, but rather provides alternative responses.[33]

They note that symptoms can return, depending on the patient's environment and emotional state. In such cases, patients may need to return to interdisciplinary treatment to reinforce appropriate functioning.

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Rehabilitative Nursing

27

AYEHUBIRHAN SHENKUTE, SARAH CLARK, DAWN PENNER, and DULCE IGLE

GENERAL PRINCIPLES

Rehabilitation nursing is a philosophy of nursing practice and an attitude toward caring for people with disabilities and chronic health conditions.[1] The goal of rehabilitation is to restore mental and/or physical abilities lost from injury and/or disease to function in a normal or near-normal manner.[2] As in many areas of nursing, rehabilitation nurses recognize the need to improve the quality of care and provide the most current evidence-based care to their patients and their families. To achieve high levels of competence, rehabilitation nurses need to be aware of existing body of research in this field and increase their knowledge using evidence-based practice.

After at least 2 years of rehabilitation nursing experience, a rehabilitation nurse can achieve standardized, nationally recognized certification credential, much like a board certification for physicians, demonstrating their knowledge, dedication, and expertise in the field of rehabilitative nursing. A Certified Rehabilitation Registered Nurse (CRRN) certification is the credential for nurses who care for individuals with disabilities and chronic illnesses to restore, maintain, and promote optimal health. When a rehabilitation nurse earns their CRRN, they are validating their professional standing as a qualified and experienced rehabilitation nurse with a documented level of knowledge, competency, and commitment to patient care.[3]

GUIDING PRINCIPLES

Dorothea Orem, a renowned nursing theorist, created a theory called the Self-Care Deficit Theory.[6] In this theory, Orem surmises that all patients and caregivers want is to care for themselves or their loved ones. The theorist states that patients can recover more quickly by performing as much self-care (or dependent care) as possible.[7] This theory provides a great framework for rehabilitation nursing and influences some of the guiding principles of rehabilitative nursing, including promoting self-care and encouraging independence, preventing complications, restoring quality of life, emphasizing ability, promoting adaptation, and supporting dignity.[8]

Initial Management

According to Wade,[9] all rehabilitation interventions can be broken down into three basic categories:

- Providing support to maintain well-being
- Collecting data
- Giving treatments

Pediatric rehabilitation nurses provide support to maintain the well-being of patients by providing care and assistance to tasks that patients previously would have done themselves (e.g., oral care, other hygiene measures, turning, toileting, taking medications). They also assist in treatment by teaching patients and their families how to care for their current condition and perform multiple assessments which can help provide quantitative and qualitative data.

One of the interventions mentioned by Wade[9] is providing support to maintain well-being. This is what is typically thought of when it comes to rehabilitation nursing and this can include:

- Turning the patient often to prevent skin breakdown and providing wound care if skin breakdown occurs
- Giving pain medication to relieve the patient's pain based on assessment
- Administering enteral and parenteral nutrition if the patient is unable to meet their nutrition needs by mouth or encouraging oral intake to meet their needs
- Administering medication to encourage/elicit adequate bowel movements or implementing a bowel program for patients with spinal cord injuries (SCIs) with neurogenic bowel
- Ensuring the patient is voiding adequately and performing catheterization for those who have neurogenic bladder

Another type of intervention noted by Wade[9] is giving treatments to manage the patient's current condition. During the initial management phase, the pediatric rehabilitation nurse provides treatment by educating the family on how to care for their child's current condition. These treatments can include:

- Encouraging participation of both the caregiver and the patient in physical, occupational, and speech therapies
- Offering guidance to the caregiver and the patient on how to integrate back into the community after their inpatient stay
- Providing education on how to administer medications, the indications and potential side effects of the medications, and enteral feedings if needed, and on the proper care of the tube, how to perform catheterization if needed (see Box 27.1), how to manage the bowel program, how to recognize signs of autonomic dysreflexia, and how to manage complications if they arise (see Box 27.2)

BOX 27.1 NEUROGENIC BLADDER MANAGEMENT PROCEDURE

1. Wash hands. Prepare a sterile field for the procedure.
2. *For females:* Separate the labia with the thumb and index finger of the nondominant hand and cleanse the perineal area. Cleaning from the front to back, spread the labia and hold open with a clean hand while inserting the catheter with a sterile hand.

   For males: If uncircumcised, gently retract the foreskin until the meatus is seen. Hold the penis with the nondominant hand and cleanse the head of the penis in a circular motion, beginning at the meatus and cleaning to the outer edge of the glans. Hold the shaft of the penis with a clean hand while inserting the catheter with the sterile hand.
3. Once complete elimination is achieved, remove the catheter from the urethra and wipe Betadine off from the skin. For males, if uncircumcised, pull the foreskin gently back over the head of the penis.

BOX 27.2 NEUROGENIC BOWEL MANAGEMENT PROCEDURE

1. Gather the equipment (padded shower commode or drop-arm commode over toilet as indicated, suppository, lubrication jelly, gloves). As determined by therapy, suppository inserter and/or digital stimulator may be needed.
2. If in bed, place the patient on the left side with the right leg flexed.
3. Wash hands, lubricate the gloved finger, check the rectum for stool, and gently remove any stool that may be in the rectal vault.
4. Insert well-lubricated suppository or enema into the rectum and wait 10 to 15 minutes to allow the medication to work.
5. If in bed and can transfer, transfer the patient to a padded commode chair.
6. Perform digital stimulation by gently inserting a clean lubricated, gloved finger into the rectum with firm circular motion. Rotate the finger and maintain contact with the rectal wall for 15 to 60 seconds. Do not rotate the finger for any longer than 60 seconds.
7. Repeat step 6 every 5 to 10 minutes to promote prolonged peristalsis while the anus relaxes.

Note: Do not perform digital stimulation more than four times in one bowel program session unless you have spoken with the provider.

An additional intervention is data collection. In the hospital setting, the pediatric rehabilitation nurse's evaluation of vital signs provides quantitative data that can inform the clinician of potential complications in pediatric patients:

- Increased heart rate could be a sign of the following:
  - Autonomic storming in a patient with traumatic brain injury (TBI)
  - Fever in a patient who has been deconditioned after receiving chemotherapy
- Increased blood pressure could mean the following:
  - Increased intracranial pressure (ICP) in a TBI patient
  - Autonomic dysreflexia in a patient with SCI
- Decreased respiratory rate could be a side effect of a muscle relaxer given to a patient with spasticity.

The nurse will also monitor the patient's skin integrity, complete a pain assessment, evaluate intake and output, and make changes to their plan of care based on these metrics.

NEUROGENIC BLADDER AND BOWEL

Neurogenic bladder and neurogenic bowel dysfunction (NBD) are common conditions that a rehabilitation nurse, in collaboration with interdisciplinary team, will work with the patient and their family to optimize elimination pattern. The number of individuals vulnerable to neurogenic bladder and NBD is ever-increasing. SCI, both traumatic and nontraumatic, has an estimated prevalence of over 2.5 million worldwide. Of those with SCI, up to 95% report constipation and 75% have experienced episodes of fecal incontinence. The primary responsibility and focus of the rehabilitation nurse are to teach patients and their caregivers how to manage bladder and bowel training programs and how to incorporate these programs into their daily lives. Treatment for neurogenic bladder and NBD depends on a variety of factors, including patient age, injury or disease process, severity of damage, type of symptoms, and physical medicine and rehabilitation expectations during the course of treatment.

Neurogenic Bladder Management

Bladder training is an essential part of neurogenic bladder management. Several bladder management techniques may be utilized to assist in optimizing bladder function and emptying. Intermittent catheterization (IC) is the gold standard in the management of neurogenic bladder, providing independence, alleviating symptoms, and preventing complications of the urinary tract.

Box 27.1 goes over the step-by-step process of catheterization in the hospital.

It is important to note that while the hospital staff use sterile technique to perform IC to prevent hospital-acquired infections, the caregiver and/or the patient will learn to perform the procedure with clean technique.

Neurogenic Bowel Management

Symptoms of NBD have a substantial negative impact on quality of life, social integration, and personal independence. Only 6% of SCI patients require no intervention to support their bowel function. As many as 65% need to employ intrusive options, such as digital stimulation or evacuation of the anorectum, and one-third require assistance with bowel care. Evidence has shown that it takes over 60 minutes to perform bowel care and most patients need some assistance.[10] Special consideration for the oncology patient should be made as they are prohibited from any invasive options such as suppositories or digital stimulation if they are neutropenic.

Ongoing Care

Individuals with health conditions or injuries may require rehabilitation across their lifespan. The timing and type of intervention that will be selected for each individual depend on several factors. These include the type of injury, prognosis, how the individual will be able to function in the community, and most of all the individual's identified goals.

Rehabilitation services may be delivered in any setting, including hospital, outpatient, and community setting, depending on the need. In the hospital setting, pediatric patients will receive rehabilitation services focused on recovery, maximizing their functional ability to achieve optimal outcomes. Without access to proper rehabilitation and long-term care, such individuals may experience potentially fatal complications, such as pressure injuries and urinary tract infections. These complications can be a secondary source of sepsis, leading to further hospitalizations and potential death.[12] Therefore, the rehabilitation nurse has a significant responsibility in facilitating care during the transition when individuals reenter the community after hospitalization and in the continuum of care from the hospital to home. The Association of Rehabilitation Nurses (ARN) believes that the role of the rehabilitation nurse in the home care setting is essential in the continuum of care. The value of the rehabilitation nurse has been demonstrated by the ARN.[13]

- Improved cost-effectiveness of client care
- Specialized rehabilitation nursing clinical knowledge and skill
- Reduction in the frequency of complications and rehospitalizations experienced by rehabilitation patients
- Increased quality of nursing care
- Reduced costs due to the presence of a resident expert who provides consultation services

In a hospital setting, the rehabilitation nurse initially plays an active role in helping patients function at their best. These include meeting basic needs, activities of daily living, and coordination of care with other members of the team after assessing their needs. The rehabilitation nurse acts as an advocate for pediatric patients and their families during the reentry process from the hospital to home and to the community. After an

individual's reentry to the community, the home care rehabilitation nurse will coordinate the services provided by the interdisciplinary team and enact the plan of care that has been developed by the physician, the rehabilitation team (physical therapy, occupational therapy, speech therapy, neuropsychology), and the family. In this role, the home care rehabilitation nurse functions as a clinical resource, care coordinator, advocate, primary care provider, teacher, consultant, and team member. The home care nurse, using rehabilitation expertise, develops an individualized program for the client and the client's family or caregiver.

Meeting the ongoing needs of these individuals is the primary goal. The essential needs of these individuals focus on hygiene, nutrition, effective communication, gaining strength and mobility, pain management, evaluation of sleep and rest, neurogenic bladder and bowel management, skin integrity/wound care management, and individual and family support. It is also important for the outpatient nurse to evaluate the interventions and provide reeducation if needed. Early inclusion of family members in care interventions will ease the long-term struggle with the disease or condition and create a climate of trust.[14]

TREATMENT CONTROVERSIES

Pain Management

Patients with severe neurologic impairment (whether it be due to birth defects or injuries) are at increased risk of inadequate identification, assessment, and treatment of pain. This may be due to several reasons. The clinician's assumptions and beliefs may interfere with the identification of pain. Agitation, irritability, and increased spasticity are often assumed to be related to the patient's underlying diagnosis and the identification of pain is overlooked. Other times, the parent's report of the patient being in pain is overlooked or may seem out of proportion to the patient's behavior.[15] It is important to remember that the child's parent is usually a good resource and knows their child best. Include the family in the pain identification process by teaching them the signs and symptoms of pain and encourage them to be an advocate for their child.

End-of-Life Care

Decision-making in the pediatric population is sometimes complicated because the patient themselves cannot provide consent. This could be due to their age (too young to consent) or because their diagnosis prevents them from making informed decisions. Sometimes the parent's and the patient's decisions do not align, which can further complicate matters. Other times, the parent's wishes do not align with the medical team's recommendations.[16] In these cases, it is important to involve an interdisciplinary care team, which may include an ethics committee and palliative care. Sometimes, the hospital's legal team may be involved to provide consultation on difficult cases.

ADDITIONAL CONSIDERATIONS

Cultural Considerations

Research has shown that cultural differences can lead to delayed or incorrect diagnosis. An example of this is in the early detection of autism spectrum disorder (ASD), where despite there being protocols and diagnostic materials, ethnic minority children are more likely to be diagnosed later in life than Caucasian children.[17] Different languages and reading difficulties can sometimes lead to difficulties adhering to prescribed medication regimens as well.[18] It is important for the nurse and other members of the healthcare team to receive adequate cultural and diversity training to have better communication with the patient and their parents and ultimately provide the best care. When caring for patients who speak different languages, it is important to provide care and education in their preferred language. This may include utilizing language line services or inperson interpreters when providing updates and education and when going over the plan of care.

Health Literacy and Educating Caregivers

An integral part of the role of a rehabilitation nurse is to educate the caregiver on how to care for their child. People learn differently, so it is important to complete a learning assessment prior to starting education. One area of the learning assessment is an assessment of the caregiver's health literacy (the ability to obtain, understand, and apply health-related knowledge). Low healthcare literacy can be affected by the caregiver's education level, reading ability, and preferred language, among other things.[19] Limited health literacy might lead to adverse health outcomes, including nonadherence to medications, difficulty making informed decisions, increased readmissions, and higher utilization of healthcare resources.[20]

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Occupational Therapy

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INTRODUCTION

Occupational therapy (OT) is the therapeutic use of common occupations to enhance or enable performance and functional independence of a patient or client. An occupational therapist examines the relationship between the individual, the occupation, and the environment to determine functional deficits and develop interventions to address habilitation or rehabilitation to address these needs.

OT addresses, but is not limited to, the following domains: occupations, contexts, and performance skills. These domains interact with one another to positively or negatively affect an individual's functional performance and independence. Occupational therapists must evaluate each of these domains and their impact on the patient or client to assess needs and develop treatment plans within their respective settings.

OCCUPATIONAL THERAPY DOMAINS

Occupations

Occupations are activities that a person or a group of people will do to occupy time or bring meaning or purpose. They range from tasks that one must complete daily, on the job, or as hobbies. There are subsets of occupations that occupational therapists address directly.

- Activities of daily living (ADLs)
- Instrumental activities of daily living (IADLs)
- Health management
- Rest and sleep
- Education
- Work
- Play
- Leisure
- Social participation

Contexts

When evaluating one's occupational performance, an occupational therapist must consider those personal contexts. These are individualized specific factors that influence or affect participation in occupations. These can

be broken down further into factors that are imposed upon the person from the external environment or internal, personal factors.

- Environmental factors are placed upon the person from outside of themselves. These may be natural or human-made, technological factors, relationship-based, or built on a system or policy.
- Personal factors are factors that are internal and person-specific. These include, but are not limited to, chronological age, sexual orientation, gender identity, race/ethnicity, cultural identifications, education level, habits, and fitness status.

Performance Skills

When observing a person complete occupations, the occupational therapist must take note of the skills the individual uses to participate, including motor skills, processing skills, and social interactions.

OCCUPATIONAL THERAPY IN PEDIATRIC ACUTE CARE

Overview

OT in pediatric acute care covers a large span of developmental stages and a wide variety of diagnoses. OT aims to support and promote growth and development, including not only physical changes that occur from infancy to adolescence, but also changes in emotions, personality, behavior, thinking, and communication that children develop as they begin to understand and interact with the world around them. A child's admission to a hospital can be overwhelming for both the child and the family unit. For the child and family admitted to a hospital, there is often emotional intensity, medical complexities, and sometimes uncertainties with the child's diagnosis, prognosis, and length of hospital stay. For children who must be admitted for a lengthy time period and/or have multiple admissions for long periods, there is disruption in their normal development from lack of normal sensory experiences, limited environmental exploration and mobility, and change in roles and typical daily routines.[1,2] Many children who receive acute care OT services have suffered enough loss of function, compared with typically developing peers, that they require skilled intervention to regain some independence in meaningful daily activity. OT takes a holistic view of the child and the family to determine the best interventions for promoting the highest level of independence possible for the child.

OT is a consultative service in the hospital and must be ordered by a physician, physician assistant, or nurse practitioner. Once an OT order is in a patient's chart, the occupational therapist does a chart review of the patient's medical history and developmental history, collecting information also on social history and family dynamics. After reviewing the chart, the occupational therapist completes an assessment of the child and determines the best plan of care to help the child reach their highest level of

independence. The plan of care is set depending on how far off the child is from their normal baseline functioning. If a child has known chronic developmental delays, there may be less expectation of large functional gains during an admission versus a child with an acute injury or new diagnosis that has caused a sudden significant loss of function quickly. The plan of care that is implemented can be altered depending on the child's response to the interventions. If a child's function and engagement in meaningful activity decline further after the original plan is set, then a plan of care can be increased, or if a child makes such significant gains that they do not need to be seen as frequently then the plan of care can be decreased. If a child reaches their functional baseline they had prior to admission to the hospital, they could be discharged from services or monitored peripherally through chart reviews.

Common Diagnoses

- Traumatic brain injury
- Stroke
- Prematurity
- Genetic syndromes
- Congenital cardiac malformations
- Cardiac disease
- Organ transplants
- Orthopedic injuries
- Nerve injuries
- Neurologic/neuromuscular disorders
- Oncologic conditions
- Sickle cell anemia
- Severe respiratory illness
- Septic shock
- Failure to thrive
- Autoimmune disorders
- Postsurgeries

Common Goals and Objectives

The goal of OT is for the child to reach the highest level of independence possible. This varies widely because it will depend on whether the goals are being written for a newborn or an adolescent. The goals are written specifically for the child and may incorporate the family's and the child's hopes and expectations for regaining function. There are usually better outcomes in reaching the goals when the child and the family are involved in writing the goals because then they have some investment. Goals are typically written according to a task analysis of a desired activity. For example, to be able to complete dressing, a child must have the range of motion (ROM) of upper extremities (UEs) and lower extremities (LEs), strength, cognitive ability to problem-solve and sequence, and sitting balance (head and trunk control). Therefore, if a child wishes to be able to put

their own pants on, the goal may start out with a goal for sitting tolerance, or sequencing a three-step task, or increasing the ROM of UEs to be able to reach their foot to thread their legs into the pants. When a goal is reached, a new goal can be written to further advance independence in the area of importance for the child and/or the family.

Splinting

There are many reasons that a child may need a splint, including acute soft tissue injury, neurologic injury, or orthopedic injury. Splinting may be indicated for the following reasons:

- For comfort due to neuropathic pain
- Protection of joint integrity
- Prevention of contractures
- Joint stability
- Stabilization of a fracture

For an acute orthopedic or soft tissue injury, often a very specific type of UE splint is ordered by the orthopedic surgeon or plastic surgeon. For other conditions, it is up to the treating occupational therapist to assess the need for splints and fabricate splints as needed. After a splint is fabricated, the occupational therapist educates the patient, the family, and the nurse what the appropriate wearing schedule should be. Most splints in acute care are made of a thermoplastic material, but there are times when functional casting material is used or even a soft neoprene material, especially for infant use. The goal of splinting is to support ROM and positioning to optimize functional use of the affected part of the UE.

Occupational Therapy in the Neonatal Intensive Care Unit

The NICU is often a very stressful environment for a newborn and an overall decreased normalized extra-utero environment that can be non-supportive for neuromaturation. Often, there is loss of normal parental roles that disrupt the infant and parent dyad when a baby must be hospitalized, and this can have long-term effects on an infant's development.[3]

OT is typically consulted at admission of a baby in the NICU. At initial evaluation, the occupational therapist assesses:

- Reflexes
- Muscle tone
- ROM
- Symmetry/asymmetry
- Behavior
- State regulation
- Vision
- Newbornparent bond
- Environment in the baby's room
- Skin integrity
- Tolerance for touch/handling and positioning

The evaluating occupational therapist sets a plan of care and goals to be addressed. Interventions in the NICU aim to support the neurodevelopment of the hospitalized infant and include:

- Supportive positioning
- Appropriate developmental stimulation through movement and postural changes
- Modifying the environment to reduce overstimulation
- Positive touch
- Nonpharmacologic pain support
- Family education

The infant is typically followed throughout the admission due to the high risk of developing delays from being hospitalized. As the infant and family near discharge, the occupational therapist gives discharge recommendations that fit the baby and family's needs going forward and can typically include referrals to one of the following:

- Home health OT
- Early childhood intervention
- Outpatient OT
- Physical medicine and rehabilitation clinic

OCCUPATIONAL THERAPY IN THE INPATIENT REHABILITATION SETTING

Overview

OT in inpatient rehabilitation focuses on improving functioning, safety, and caregiver education to transition the patient to the home setting. This is accomplished through intensive therapy. Often durable medical equipment (DME) is required to function within the home environment and must be ordered. Family education teaches caregivers how to care for the child after discharge. In this setting, OT often treats patients for at least 1 to 1.5 hours a day, broken up into one or two sessions a day.

Common Diagnoses

- Neurologic disorders including, but not limited to, traumatic brain injuries, cerebrovascular accidents, demyelinating conditions, disorders of consciousness, and spinal cord injuries
- Polytrauma
- Posttransplant conditions
- Deconditioning following prolonged hospitalization or complicated medical history
- Functional changes following surgical intervnetions for acute or chronic medical conditions.

Common Goals and Objectives

- Transition safely to home
- Address ADL deficits

- Improve strength, ROM, or motor coordination deficits
- Improve functional cognition skills
- Address visual and visual-perceptual impairments
- Improve safety during performance of functional skills
- Order equipment to allow for occupational performance at home
- Educate and train caregivers to complete exercises and provide care at home
- Make recommendations for continued therapeutic needs once discharged home

Equipment Considerations

The selection of appropriate DME is key to facilitating completion of daily tasks once the patient returns to their home. Within OT, the therapist often looks at bathroom equipment to assist with toileting and/or bathing. If the equipment is improperly selected, it may either be too difficult to use and thus limit the patient's functional independence or too enabling and hinder their continued progress. Additionally, equipment that is not appropriate may pose a safety risk within the home. DME considerations include:

- Trunk control
- Static and dynamic sitting balance
- Ability to transfer from one surface to another
- Opportunity for growth of the piece of equipment as the child grows
- Parents' ability to manage, care for, and use the equipment with the child
- Size and space in the patient's home where the equipment will be used

Electrical Stimulation

Neuromuscular electrical stimulation (NMES) is a common treatment approach to address a variety of neurologic diagnoses. This is accomplished through placement of a set of electrodes on the skin and delivering a low-level, electrical current to the muscle. This can result in the contraction of the damaged or weakened muscle to strengthen it, reduce spasticity, improve joint mobility, and/or facilitate neuroplasticity. The electrical waveform can be customized to achieve different results, depending on the needs and abilities of the patient and the goal of the treatment. There are a variety of subtypes of electrical stimulation.[4] Within this setting, the most common subtypes are functional electrical stimulation (FES) and transcutaneous spinal cord stimulation (TCSC).

- FES[4]
  - The goal of FES is to facilitate improved motor coordination and functional use of the extremity.
  - NMES is paired with the performance of a specific functional task. This may be motor-based, such as reach and grasp, or activity-based, such as brushing teeth.
  - Electrical stimulation is provided to multiple muscle groups to facilitate completion of the desired activity. This stimulation is provided

asymmetrically to activate the muscles at the desired movement pattern.
    - Research has demonstrated that repetition of activity improved motor coordination and functional use of the extremity or extremities.
- TCSC[5]
    - TCSC is stimulation with a long pulse duration, with the electrodes placed paraspinally and on the abdomen for reference.
    - It is utilized in patients with spinal cord injuries to reduce spasticity and improve volitional activation in the desired extremity or extremities.
    - It can be utilized for both upper and lower extremities.

OCCUPATIONAL THERAPY IN THE OUTPATIENT SETTING

Pediatric OT practitioners work with children from birth to age 21 and their families to develop, maintain, or regain skills necessary for participation in daily occupations or meaningful activities. Occupational therapists utilize occupations or activities that support the health, well-being, and development of an individual,[6] as the means and the end in the rehabilitation process.

Initiation of outpatient services begins with a referral from a physician or advanced practice provider. The child is then evaluated by an occupational therapist. This evaluation consists of a review of the child's medical history, parent/caregiver interview, observation, physical examination, and standardized testing. Based on this evaluation, the occupational therapist determines the plan of care, or the frequency and duration of treatment, and sets client-centered goals based on the deficits identified through the evaluation. If a patient's third-party payor requires prior authorization for treatment, the evaluation will be submitted with the plan of care. Once insurance approves the request for therapy, treatment may begin. The duration of an outpatient session may last for 30 to 60 minutes, and the frequency will depend on the level of acuity and severity of the needs of the patient.

Common Diagnoses

Outpatient pediatric occupational therapists see a wide array of diagnoses, including but not limited to:

- Autism
- Developmental delay
- Sensory processing disorder
- Chromosomal and genetic disorders
- Orthopedic injuries or congenital deformities
- Trauma-related injuries
- Cancer
- Traumatic brain injury, concussion, and vestibular-related impairments
- Brachial plexus

- Cerebral palsy
- Attention deficit disorder/attention deficit hyperactivity disorder
- Epilepsy
- Stroke
- Dysphagia and feeding difficulties
- Fine motor delay
- Muscular dystrophy
- Neurologic diagnoses
- Spinal cord injury
- Spina bifida

Common Goals and Objectives

It is important to note that occupational therapists do not treat the diagnosis but rather the impairments that stem from the diagnosis. The occupational therapist works closely with the patient and the family to determine the goals of therapy. The nature of these client-centered goals changes based on age. The primary occupations for infants, toddlers, and young children are playing, interacting with caregivers and peers, and learning. Occupational therapists facilitate this through work on developmental milestones, which include, but are not limited, rolling, sitting, reaching, grasping, crawling, visual tracking, following instructions, engaging with toys, developing age-appropriate self-care skills, and building skills for social participation. The occupations for older children and adolescents primarily focus on educational pursuits, engaging in social relationships, and working toward the transition to adulthood. Occupational therapists work with this population on identifying modifications and adaptations for participation, navigating social relationships, assisting with vocational planning and transition to work or higher education, and enhancing overall independence.

Handwriting

Occupational therapists are specialists in fine motor skills, including handwriting. The occupational therapist evaluates a child and analyzes their handwriting ability to determine if their difficulty is an issue with the visual component, hand or grip strength, core or upper body strength, sensory processing, grasp pattern, or attention. They are then able to address handwriting with an individualized treatment plan.

Prefabricated Splints

Occupational therapists may utilize prefabricated splints for a number of reasons; however, one of the most common reasons is to prevent or maintain contractures as a result of weakness or increased tone. There are many options when choosing a prefabricated splint, and sizing appropriately is important. Some manufacturers offer stock sizes, while others allow you to take precise measurements and then the splint is created for the

individual. The majority of prefabricated splints are static, but some come with a dynamic component. As with any orthosis, skin integrity is very important and should be monitored closely.

OCCUPATIONAL THERAPY IN THE HOME HEALTH SETTING

Overview

OT services that are delivered in the home setting typically serve those who are at high risk of developmental delays in fine motor skills, coordination, UE dysfunction, oral motor skills, visual-perceptual difficulties, sensory processing differences, or difficulties completing activities related to daily living skills. The plan of care for patients receiving home health OT is typically one to two times per week. Home health OT typically serves those with more chronic developmental delays or those who have difficulty leaving their home for clinic visits.

Common Diagnoses

- Global developmental delays
- Prematurity
- Autism/sensory processing disorder
- Brain injury
- Cerebral palsy
- High risk of developing delays due to diagnosis
- Neuromuscular disease/syndromes
- Neurologic conditions

Common Goals and Objectives

The goals of OT for children in the home setting are written in collaboration with the family. The occupational therapist interviews the family to understand their desired outcomes for the child and combines these with their expectations for the child to create achievable goals. Ultimately, the goal is for the child to reach their highest level of independence as possible so that they can engage in meaningful daily activities alongside their peers.

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Palliative Care

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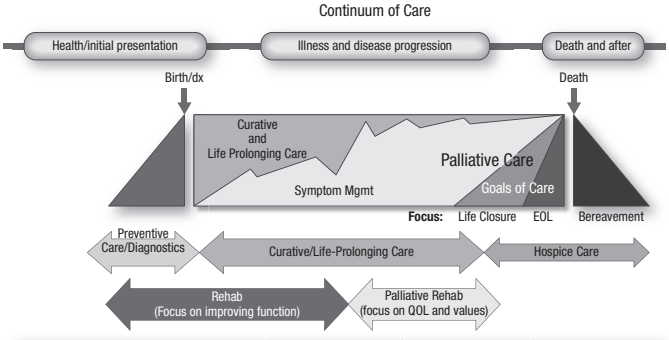
INTRODUCTION

The Center to Advance Palliative Care (CAPC) provides recommended language to use when talking to both patients and colleagues about palliative care services. This is important because the way in which services are introduced to families and to referring providers is important in setting the stage and preparing for a successful first consult and in the ability to build rapport with patients and families.

Full Evidence-Based Definition (Based on the World Health Organization Definition and Revised Consensus-Based Definition By The International Association for Hospice and Palliative Care)[1]

Palliative care is specialized, active holistic medical care for people living with serious health-related suffering from a severe illness. Through a robust multidisciplinary approach including doctors, nurses, social workers, chaplains, and other specialists, palliative care aims to address "the relief of physical, social, emotional, and spiritual suffering in children and their families," with the goal of improving the quality of life (QOL) of both the patient and the family. Palliative care teams enhance patient care by optimizing symptom control, aiding in time-intensive and complex communication with family and specialists, understanding family values to facilitate value-guided decision-making when considering the benefits and the burden of therapeutic options, and providing an extra layer of support for a longitudinal relationship with the patient and their family.

Palliative care is based on the needs of the patient, not on the prognosis. It is appropriate at any age and at any stage of a serious illness and can be provided along with curative treatment. A common misconception is that palliative care is the same as end-of-life care and/or hospice. In fact, palliative care is an approach that supports all forms of treatment, with the sole agenda of providing support to patients and their families as they go through the process from diagnosis to death. This continuum of care is illustrated in Figure 29.1. Interventions are



Focus of care	Cure	Life prolonging	Life closure	Comfort/EOL
Attitude to condition	Beat it	Maximizing life while fighting for it	Prioritizing values	Embrace it/ acceptance
Tolerance of invasive therapy	Very high	High-moderate	Low	Very low

Figure 29.1 Continuum of care.

dx, diagnosis; EOL, end of life; QOL, quality of life.

Source: Adapted from Rita et al. Transition and lifespan care for patients with cerebral palsy. In *Orthopedic Care of Patients with Cerebral Palsy*. Springer, 2020:257–285.

individually tailored to meet the unique goals of the patient and their family. Generally, helping patients to live as well as possible for as long as possible is an important component of structuring treatment plans. Studies consistently show that involvement of palliative care in the treatment of seriously ill and medically complex patients improves the quality of care and lowers healthcare utilization.[2]

PARENTAL EXPERIENCES CARING FOR CHILDREN LIVING WITH SERIOUS ILLNESS

The purpose of this section is to draw attention to the demands of medical caregiving provided at home by parents and the importance of providing thorough and realistic education about the impact of treatment before initiating both invasive and noninvasive therapies. Setting expectations allows parents to thoroughly consider the impact of treatment options on the QOL of the patient and their caregivers. Common interventions within the pediatric rehabilitation medicine (PRM) patient population that should be given pause for thorough consideration regarding potential benefits and burden are tracheostomy and surgery for scoliosis, among others; these will be further discussed in the "Communication Strategies" section. This section also acknowledges the personhood of our patients, their inherent value, and the immense joy they bring their families despite the challenges highlighted.

Children with medical complexity (CMC) are defined as people who "have multiple significant chronic health problems that affect multiple organ systems and result in functional limitations, high health care

utilization, and often the need or use of medical technology."[3] According to Cohen et al., CMC "are increasing in prevalence because of increased survival rates as well as improved treatments for acute illnesses in fields such as intensive care and oncology."[4] Subsequently, more children are living with serious medical challenges requiring complex medical treatments and greater assistance with care. CMC may be totally dependent for care or be entirely reliant on technology or life support. What used to be considered skilled hospital-level care now falls to parents to manage at home and providers cannot dismiss the impact of our patients' disease on their caregivers.

CMC are reliant on medical technology which "compensates for the partial failure or loss of a vital body function." Examples include "assisted ventilation, artificial nutrition [commonly administered via surgically placed feeding tube], intravenous drug therapies, oxygen therapy, renal dialysis and suctioning."[5] In addition to reliance on medical technology, CMC are often entirely dependent on caregivers for all routine care. This could include complex medication administration, respiratory, physical and occupational therapies, and basic hygiene tasks that take many hours a day. Administration tasks, including scheduling appointments, ordering supplies, navigating insurance, and coordinating care, further limit parents' time.

The toll of providing an average of 33 hours of care each week can lead to parents struggling with feeling tired and having too little energy for pleasurable activities, being socially isolated,[6] and at statistically significant increased prevalence for moderate depression and anxiety compared with parents of healthy children.[7] The time demand to care for CMC commonly results in one parent not being able to sustain gainful employment, therefore reducing the overall financial means of the family. Simultaneously, families with CMC are burdened by costly and frequent healthcare utilization. One study concluded that 89% of families with CMC sustained significant financial burden.[8]

The following are the main domains of palliative care[9]:

- *Symptom management* and relief of physical, psychological, psychosocial, and spiritual suffering
- *Communication and coordination* of care among patients, families, and healthcare providers
- *Support in decision-making*, with facilitated discussions regarding *goals of care*
- *Bereavement support*, with ongoing family support post death

Symptom Management

Children with progressive neuromuscular conditions, cancers, severe neurologic impairments, and complex injuries are highly likely to have some difficult-to-treat symptoms, such as fatigue, refractory pain, and secretions. Whether or not cure is possible, relief from symptoms is a vital component of alleviating suffering and improving the QOL of the patient and family unit. Table 29.1 shows examples of common symptoms

encountered by physiatrists in the care of their patients and the strategies used by palliative care specialists to help treat these symptoms.

Some of the most common symptoms that the palliative care team is frequently called upon to help with management are:

Secretions

- Hyperalgesia and central pain
- Persistent feeding intolerance
- Hypertonia

These are addressed in Table 29.1.

Communication Strategies

The Benefits of Good Communication: Why It Matters

A growing body of literature supports the role of effective communication in improving patient outcomes. Improved communication has been associated with the following benefits:

- Improved patient QOL[10]
- Longer patient survival[11]
- Improved caregiver coping, especially in advanced disease[12]
- Decreased provider emotional exhaustion, improved empathy, and decreased burnout and depersonalization[13,14]

Communication is especially important in the medically complex or dying. Ineffective communication has the potential not only to seed confusion during an emotionally tenuous time, but also to tacitly promote aggressive, life-prolonging care that may be unwanted or inconsistent with family goals.

Practical Guidelines for Effective Communication: SPIKES

The SPIKES (Setting, Perception, Invitation, Knowledge, Empathy, Summary) protocol is a commonly used guideline for effective communication (see Table 29.2).[15] A detailed review is outside the purview of this chapter, but two points demand special emphasis:

- *Use concise language when delivering difficult news.* There is evidence that patients retain only 20% of medical information that is provided to them by a physician.[16] Circuitous language and extraneous information risk increasing confusion and uncertainty.
- *Prioritize attention to patient and family emotions.* There is evidence that physicians are poor at engaging in empathetic opportunities when negative emotions arise.[17] In emotional situations, the communicator has an opportunity to offer continued solidarity and support during a period of immense vulnerability. Furthermore, patients and their families will have difficulty making reasoned decisions about the next steps until emotions have been addressed and the emotional tone lowered. The NURSE (Naming, Understanding, Respecting, Supporting, Exploring) statements, outlined in Table 29.3, are a helpful way of effectively responding to emotions.[18]

Table 29.1 Management of Commonly Encountered Symptoms

SYMPTOM	RELEVANCE	MANAGEMENT	OTHER THOUGHTS
Secretions	These are commonly encountered problems in palliative care populations, especially in the setting of children with severe neurologic impairments.	Conservative • Repositioning • High-frequency oscillation vest therapy • Suctioning • Airway clearance (e.g., hypertonic saline, cough assist device) Pharmacologic • Glycopyrrolate 40–100 mcg/kg/dose PO/enterally q4h–q12h • Atropine 10% eye drops, 1–2 drops/dose SL, titrate to effect • Scopolamine ½1 patch transdermally to the skin behind the ear • *Note: All of the above are anticholinergics; beware of side effects of constipation, urinary retention, etc.* Invasive • Botulinum toxin injection • Salivary gland ligation or removal	• Secretions often worsen at end of life due to decreased consciousness, inability to protect the airway, and decreased airway tone. This is often referred to as "death rattle." This is a normal part of the dying process and is not thought to be distressing to patients, although it can be distressing to caregivers. There is little evidence of medication efficacy, though it is reasonable to consider a trial of medication if the symptom is bothersome for family despite education.

(*continued*)

Table 29.1 Management of Commonly Encountered Symptoms (*continued*)

SYMPTOM	RELEVANCE	MANAGEMENT	OTHER THOUGHTS
Hyperalgesia and centralized pain	These are common in children with neurologic impairments, manifesting as recurrent irritability or pain without an obvious source despite extensive assessment.	Conservative • Repositioning, minimize positional discomfort • Decrease stimulation; quiet environment, cool and dark room Pharmacologic • First line: gabapentin ◦ Initial dose: 2 mg/kg/dose TID ◦ Increase by 50%–100% every 2–3 days to effect, maximum dose 40 mg/kg/d • Second line: TCAs • Third line: cyproheptadine, dicyclomine, others	• The physiology is poorly understood, although both visceral hyperalgesia and central pain are thought to be heterogenous processes involving dysregulation of central nervous system perception of noxious stimuli. In the case of visceral hyperalgesia, there may be underlying injury to the bowel as well. • These are diagnoses of exclusion that can only be considered after an extensive history and physical and a reasonable workup. • Differential diagnosis to include constipation, reflux, bed sores, corneal abrasion, occult fracture, UTI, urinary retention, hypertonia, hip dysplasia, etc.

Persistent feeding intolerance	Children with chronic neurologic impairments often progress to feeding intolerance, especially those who are receiving fluid and nutrition through a feeding tube. Feeding intolerance can manifest in a variety of ways, including: • Dysmotility leading to vomiting • Pain and agitation • Diarrhea due to decreased gastrointestinal absorption Persistent feeding intolerance should be viewed as a natural progression of severe neurologic impairment and one which can be managed but not reversed.	• Bowel regimen • Pain medications (e.g., opioids, GI prophylaxis, gabapentin, or other centrally acting agents to manage visceral hyperalgesia) • Antiemetics • If diarrhea or bloating, consider trial of antibiotics for SIBO • If dysmotility a concern, consider prokinetic agents such as erythromycin or metoclopramide • In children fed via gastrostomy tube, may warrant a trial of postpyloric feeds • Consider decreasing volume of feeds to reflect lower caloric needs	• Often, the management options identified show initial benefit that lessens over time. • In children with persistent feeding intolerance despite the above, a goals of care conversation may be warranted. In children who are clearly suffering from feeds, a conversation with the family about feeding for comfort rather than nutrition should be had. • Although concerns are often raised about the ethics of this decision, the position of the AAP and the AAHPM is clear that it is ethically permissible to discontinue medical nutrition and/or hydration when it is prolonging or contributing to suffering.

(*continued*)

Table 29.1 Management of Commonly Encountered Symptoms (*continued*)

SYMPTOM	RELEVANCE	MANAGEMENT	OTHER THOUGHTS
Hypertonia	Hypertonia is a common problem in children with severe brain injuries or spinal cord injuries and is a common cause of either focal or generalized pain. It can also greatly interfere with seating and positioning, as well as make caregiving very difficult.	Conservative • Rehabilitation strategies: positioning, seating, and splinting • Gentle massage • Myofascial release Pharmacologic • Oral baclofen: first line for spasticity and dystonia ◦ Starting dose 2.55 mg/d, with slow titration up to a maximum of 2 mg/kg/d in >2-year-old or 80–100 mg/d in older adolescents and adults divided TID or QID (Lexicomp Online) • Diazepam: useful for spasticity and dystonia; preferred treatment for spasms following orthopedic procedures, ITB withdrawal, and status dystonicus (dystonic storms) ◦ .01–.3 mg/kg/d divided BIDQID, or children >5 years of age can start with 1.25 mg TID and titrate up to 5 mg QID (Lexicomp Online)	• Always address treatable triggers for tone exacerbations first prior to making medication changes, e.g., UTI, pressure sores, GERD, etc. • High doses of oral baclofen can lower seizure threshold. • Rather than having multiple medications from the same class in treating hypertonia, it may be better to have a mix of medications from different classes. • If diazepam is being used as a rescue medication for seizures, refrain from escalating doses of diazepam for hypertonia management unless refractory to other options. • Tizanidine and clonidine have beneficial effects with sleep and lack respiratory depressant side effects.

		• Tizanidine: second line for spasticity ◦ Starting dose .05 mg/kg/d divided BID to QID ◦ Older children and adolescents: can start at 1–2 mg TID and increase every 3–4 days to a maximum adult dose of 36 mg/d • Trihexyphenidyl: second-line treatment for dystonia ◦ 0.1–0.2 mg/kg/day divided TID for the first week, then increase (Lexicomp, Rice and Waugh 2009) ◦ May need to go up as high as 2.6 mg/kg/d divided TID • Carbidopa/levodopa: second-line treatment for dystonia ◦ Starting dose 1 mg/kg/d, maintenance dose 4–5 mg/kg/d TID, and maximum dose not to exceed 10 mg/kg/d or maximum of 600 mg/d	

(*continued*)

Table 29.1 Management of Commonly Encountered Symptoms (*continued*)

SYMPTOM	RELEVANCE	MANAGEMENT	OTHER THOUGHTS
		• Clonidine: may be used as adjunct to diazepam in treatment of refractory status dystonicus ◦ 0.3–0.5 mcg/kg/hr via enteral, IV, or transdermal routes	

AAHPM, American Academy of Hospice and Palliative Medicine; AAP, American Academy of Pediatrics; GERD, gastroesophageal reflux disease; GI, gastrointestinal; ITB, intrathecal baclofen; SIBO, small intestinal bacterial overgrowth; SL, sublingual; TCA, tricyclic antipsychotic medication; UTI, urinary tract infection.

Source: Data from Delgado-Aros S, Camilleri M. Visceral hypersensitivity. *J Clin Gastroenterol*. 2005;39(5 Suppl 3):S194–203; discussion S210. doi:10.1097/01.mcg.0000156114.22598.1b. PMID: 15798485; Hauer JM, Wical BS, Charnas L. Gabapentin successfully manages chronic unexplained irritability in children with severe neurologic impairment. *Pediatrics*. 2007;119(2):e519–e22. doi:10.1542/peds.2006–1609. PMID: 17272610; Dydyk AM, Givler A. Central Pain Syndrome. 2021 Mar 13. In: *StatPearls* [Internet]. StatPearls Publishing; 2021. PMID: 31971703; Zangen T, Ciarla C, Zangen S, et al. Gastrointestinal motility and sensory abnormalities may contribute to food refusal in medically fragile toddlers. *J Pediatr Gastroenterol Nutr*. 2003;37(3):287–293. doi:10.1097/00005176-200309000-00016. PMID: 12960651; Gottrup H, Juhl G, Kristensen AD, et al. Chronic oral gabapentin reduces elements of central sensitization in human experimental hyperalgesia. *Anesthesiology*. 2004;101(6):1400–1408. doi:10.1097/00000542-200412000-00021. PMID: 15564948; Wee B, Hillier R. Interventions for noisy breathing in patients near to death. *Cochrane Database Syst Rev*. 2008;2008(1):CD005177. doi:10.1002/14651858.CD005177.pub2. PMID: 18254072; PMCID: PMC6478131; American Academy of Pediatrics Committee on Bioethics: Guidelines on foregoing life-sustaining medical treatment. *Pediatrics*. 1994;93(3):532–536. PMID: 8115226; American Academy of Hospice and Palliative Medicine: Statement on artificial nutrition and hydration near the end of life. www.aahpm.org/positions/anh; Rice J, Waugh MC. Pilot study on trihexyphenidyl in the treatment of dystonia in children with cerebral palsy. *Journal of Child Neurology*. 2009;24(2):176–182; Mink JW. Dopa-responsive dystonia in children. *Curr Treat Opt Neurol*. 2003;5(4):279–282.

Table 29.2 SPIKES Protocol

STEP	CONTEXT	DESCRIPTION/PHRASING
Setting	Prepare before the meeting.	Review medical information, provide quiet location, minimize distractions, involve significant others, ensure tissues are available, sit down, etc.
Perception	Assess patient/family understanding.	"What have the doctors told you?" "What is your understanding of what is going on?"
Invitation	Ask permission to discuss sensitive topic.	"Would it be ok if we discussed the results of your scan?"
Knowledge	Provide medical information.	Provide a warning shot (e.g., "I have something serious we need to discuss"), followed by concise medical information. Avoid medical jargon.
Empathy	Address patient emotions.	Utilize therapeutic silence and NURSE statements: "I know this isn't what you wanted to hear today."
Summary	Assess understanding and discuss the next steps.	"So I know I explained myself clearly, could you summarize what we just talked about?"

NURSE, Naming, Understanding, Respecting, Supporting, Exploring.

Goals of Care Conversations: REMAP and Value-based Decision-Making

Goals of care conversations are among the most difficult conversations that providers are asked to perform. There are many possible goals of care-curing the disease, prolonging life, decreasing symptoms, and improving QOLand no one goal is more valid than another. The goal that is most worthy of pursuit is the goal most valuable to the child and the family.

"REMAP" (Reframe, Expect, Map, Align, Plan) is a framework for conducting goals of care conversations. It assists in communicating difficult news, eliciting patient/parental values, and making shared medical decisions based on values (see Table 29.4).[19]

Other Helpful Phrases/Strategies

The communication strategies described in Table 29.5 may be helpful in various clinical situations. These strategies should not be viewed as

Table 29.3 NURSE Statements

NURSE STATEMENT TYPE	DESCRIPTION/PHRASING
Naming	"It sounds like you are frustrated." "I can see that this news was hard to hear."
Understanding	"This helps me to understand what you are thinking."
Respecting	"I can see that you have been really trying to follow our instructions." "You have fought for your child for a long time."
Supporting	"I will do my best to make sure you have what you need."
Exploring	"Help me to understand where you are coming from." "Could you say more about what you mean when you say…"

exclusive of each other; rather, they should be used collaboratively even within the same encounter as necessary.

Communicating Prognosis

This is particularly difficult when the prognosis is grim, yet this is where clarity and empathy in communication are particularly important. Box 29.1 provides some important pointers.

BOX 29.1 IMPORTANT POINTERS ON SHARING PROGNOSIS

Why it matters

- There is a moral obligation to discuss prognosis in order to promote fully autonomous and reasoned decisions about care, especially at the end of life.[21]
- Despite this, literature suggests that parents perceive more favorable prognoses than physicians, especially so at end of life.[22]
- Physicians may be reluctant to discuss prognosis for fear of "taking away hope" or creating additional distress, resulting in vague, euphemistic prognoses or even overt deception.[23]

Tips for discussing prognosis	• Ask the patient/family how much information on prognosis they would like. • Utilize clear and direct language (e.g., "death," "dying") rather than milder language or euphemisms (e.g., "pass away," "will no longer be with us"). • Utilize ranges of time ("days to weeks," "weeks to months," etc.) rather than specific estimates to account for uncertainty.

Perinatal Palliative Care

Perinatal palliative care is a medical subspecialty that provides care to families expecting an infant with serious illness. This is uniquely challenging in the perinatal period because expecting parents are making medical decisions for a person whom they have not yet met. There is also a great deal of uncertainty between prenatal predictions and prognosis once a child is born. It can be helpful to think of any diagnosis along a spectrum of possible outcomes to prepare the family to anticipate changes in the plan in accordance with the child's presentation after birth. While talking about the spectrum of possibilities is useful, it is also pertinent to highlight to parents what the most likely outcome is for their child based on prenatal findings. It is important to construct a care plan based on the most likely outcome for a child that agrees with the parents' values and have back-up plans if a child's presentation is different from predicted.

Hospice With Concurrent Care for Children (Birth to ≤21 Years Old)

Children under the age of 21 who are enrolled in Medicaid can concurrently receive the support of a hospice team and continue to receive disease-directed treatment and continue care with their subspecialists. Enrollment in hospice with concurrent care is a shared decision-making process between providers and parents or patients with ability to assent their preferences or consent to treatment preferences. For many patients and their parents, there is no "downside" to enrolling in hospice with concurrent care: patients can continue their current level of treatment *in addition* to the supportive services provided by specialized hospice teams.

Grief/Bereavement

Losing a child to death will always hold some degree of trauma to the parents. It is the job of the hospital staff to support and prepare the families as a form of trauma reduction. Hospital professionals often have little opportunity to follow up with families and attend to their

Table 29.4 REMAP

TASK	DESCRIPTION	LANGUAGE
Reframe the status quo	Communicate that things are not proceeding as initially hoped. Use concise, clear language.	"We're in a different place." "Your child hasn't recovered from his brain injury as we hoped, and he is unlikely to recover further."
Expect emotion	Address emotion empathetically. Use NURSE statements.	"I know this isn't what you wanted to hear." "I can't imagine how difficult this news must be."
Map the future	Explore patient goals/values in the context of the "new" future.	"Given this situation, what is most important to you?" "When you think about the future, what most concerns you?"
Align with values	Reframe the future in the context of patient values.	"As I listen to you, it sounds like being at home and without pain is most important." "As I listen to you, it sounds like getting as much time as possible is most important."
Plan based on values	Provide a recommendation to achieve patient values.	"Here's what I can do now that will help us achieve those important things."

NURSE, Naming, Understanding, Respecting, Supporting, Exploring.

grief after discharge. Therefore, what we do and say and the families' perception of our support or lack thereof are the primary ways that we can exacerbate families' grief or mitigate complicated grief and/or traumatic reactions. Palliative care professionals may also play an important role in providing anticipatory guidance at the end of life that can mitigate future grief.

Reducing trauma/grief from loss of a child will likely take many forms. Helping parents and families anticipate the next steps, guiding them through logistical tasks, offering opportunities for memory making and legacy work, and accommodating unique religious and cultural practices all impact family perception of the dying process. Providers can also

Table 29.5 Other Helpful Communication Strategies

Open-ended questions	Give patients/families autonomy over the visit, encourage full expression of hopes and worries	"What are some of the things that you want to talk about today?" "What have the teams told you will most likely happen?" "What's the hardest thing you've been dealing with?" "What are you most hopeful for?"
Ask–tell–ask[20]	Helpful when sharing information to get permission and assess understanding	Ask: "What have the doctors told you? Would it be ok to tell you what I understand?" Tell: Provide information using concise language and avoiding jargon. Ask: "Can you please summarize your understanding of what we just talked about?"
"Hope and worry" statements	Useful in promoting prognostic awareness while sharing in hope	"I am hopeful that your child recovers enough function to walk again, but I am worried that his injury is severe enough that he will not."
"I wish" statements	Helpful to align with families while reframing or reinforcing a clinical deterioration	"I wish that I had a better way to help the brain recover." "I wish that I could say that your child will recover fully."
Therapeutic silence	Allows for patient and caregiver processing, engenders a sense of remaining present with the patient	Maintain nonverbal presence with eye contact, leaning forward, or holding a hand. Resist the urge to fill the silence.

practice trauma reduction by setting expectations with families regarding their child's dying process. This may involve explaining what is medically and logistically possible to accommodate, compassionately saying "no" when something is not possible, not making false promises, and saying "I don't know, let me get more information about that" instead of giving parents partial or inaccurate information. We have an obligation to find out what people's hopes and expectations are and bridging the gap with what is possible. By not doing so, families will experience additional loses when their expectations are unmet.

A tool to assist with setting expectations of the dying process is outlined in Box 29.2.

BOX 29.2 SETTING EXPECTATIONS FOR THE DYING PROCESS

Who?	Who should be present? Do these people require travel time or have barriers?
When?	Can be influenced by the "who" question Time limited trial of therapy? Important dates or significance?
What?	Do you want choices regarding what is possible or do you prefer recommendations? What cultural practices are important to you? Memory making/legacy practices
Where?	What is the preferred location of death? Is this logistically possible and/or safe?
How?	Do parents wish to hold child as technology is being removed or do they prefer to hold after? "How" should be primarily considered through the lens of what would be most comfortable and peaceful with the least symptom burden to the patient.

Above all else, perhaps more important than what we say is our ability to be present and hold space for the family's grief, fear, questions, and moments of joy and love. Maya Angelou says this best, "People will forget what you said, people will forget what you did, but people will never forget how you made them feel."[24]

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