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Pediatric Nerve Conduction Studies and EMG

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Summary

A variety of neuromuscular conditions affect children, ranging from severe, usually fatal disorders, such as spinal muscular atrophy type I (Werdnig–Hoffman syndrome) to relatively mild problems, such as benign congenital hypotonia. The evaluation of children in the EMG laboratory requires special care because of the discomfort of the tests. Moreover, other considerations, such as slower baseline nerve conduction velocities and conditions that generally do not present in adulthood, such as congenital myasthenic syndromes, can make the pediatric neurophysiological examination especially challenging. This chapter reviews both the common pediatric neuromuscular conditions and their assessment in the EMG laboratory.

Key Words: Electromyography; Guillain–Barré syndrome; hereditary neuropathies; pediatrics; root avulsion; spinal muscular atrophy.

1. INTRODUCTION

EMG is a useful diagnostic tool in children suspected of having acquired or inherited neuromuscular disease and complements the advances in molecular genetic testing during the past decade. The technical limitations of performing EMG in children require that the electromyographer be selective in deciding how to approach the study. In evaluating for generalized processes, examination of one or two extremities is often adequate to narrow the differential diagnosis or, in some cases, suggest a specific disorder. EMG may be used to evaluate hypotonia in infants, assess the severity and localization of perinatal brachial plexus injuries, and distinguish between different possible causes of gait difficulties in older children. In children, EMG may contribute to the diagnosis of many disorders, including spinal muscular atrophy (SMA), brachial plexus injury, hereditary polyneuropathy, acquired polyneuropathy, disorders of neuromuscular transmission, myopathy, muscular dystrophy, and myotonic disorders.

2. TECHNICAL ISSUES

2.1. General Approach

Electrodiagnostic studies are highly dependent on technique in adults, but are even more dependent on technique in children. Infants and toddlers do not understand the purpose of the study, have a limited tolerance for discomfort, and often squirm and withdraw during testing. In selected cases, sedation or general anesthesia is required to obtain adequate data. Topical anesthetic creams are used in some pediatric EMG laboratories, which can help reduce pain,

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but they require that the electromyographer either guess which sites are most likely candidates for needle EMG before the nerve conduction studies, or impose a stressful half-hour wait between the nerve conduction studies and needle EMG on the patient. Despite these limitations, it is usually possible to perform a successful and informative study.

As in adults, a brief history and focused physical examination is critical in directing the tests performed, especially because they may be terminated prematurely. If the child is old enough to understand the procedure, it is important to explain it to the child as well as the parent. It is sometimes possible to convince younger children that the EMG machine is a "tickling" machine for nerve conduction studies—the power of suggestion being quite effective. Infants may be more comfortable sitting in a parent's lap on the examination table or in a chair. Pacifiers and toys are often effective in calming infants and toddlers. Even school-age children and adolescents may feel more comfortable if a parent sits or stands near them during the study.

2.2. Nerve Conduction Studies

The temperature of the extremity during nerve conduction studies is as important in children as in adults. An excessively cold extremity will yield falsely slow conduction velocities and increased amplitudes. Upper extremity skin temperatures, measured at first dorsal interosseous, should be at least 32°C. Lower extremity skin temperatures, measured at lower gastrocnemius, should be at least 30°C. Warming may be achieved with the use of towels dampened with hot water and wrapped around the distal extremity for approx 5 min. Care must be taken not to overheat the towels; a child's skin is more delicate than that of an adult, and is more susceptible to scalding injury. It is also important to wring out the towel before applying to the skin and to dry the skin after warming; a wet extremity will rapidly cool. Some electromyographers use disposable hot packs that produce heat via a chemical reaction. If used in children, these packs should be wrapped in towels to prevent burns, for they may become very hot. The temperature should be measured again after warming to confirm that it is in the acceptable range.

Surface active and reference recording electrodes may need to be trimmed or even cut in half for infants. Full-sized ground electrodes should be used whenever possible, however, because their placement is more flexible. Pediatric stimulators with small cathodes and anodes are useful for this age group. Standard adult distal distances cannot be used in infants and young children because of the small size of the extremities involved; thus, evaluation of motor distal latencies must take the patient's age into account (*see* Table 1). The initial stimulation should always be less than 10 mA. It can be very reassuring to a child when the first stimulation is barely perceptible.

Questions of generalized processes, such as polyneuropathies and myopathies, are common in pediatric EMG laboratories. Limited patient tolerance often makes it practical to perform only a motor and sensory study in an upper and lower extremity. In such cases, the electromyographer may perform median motor and sensory studies in an upper extremity, and peroneal motor and a sural or medial plantar (depending on the age) sensory study in a lower extremity. In a child who is uncooperative or anxious, it may be best to perform the sensory studies first, because those require less stimulation intensity and are, thus, better tolerated.

The median motor study is performed as in an adult, with the E1 recording electrode placed over the abductor pollicis brevis and stimulation occurring at the wrist and cubital

Table 1
Motor Nerve Conduction Studies, Suggested Values

Age	*A (mV)*	*CV (m/s)*	*DL (ms)*
Median nerve			
Preterm (33–39 wk)	—	≥18	—
0–1 mo	≥2.5	≥20	≤3.5
1–6 mo	≥3.5	≥25	≤3.0
7–12 mo	≥2.5	≥30	≤3.0
1–2 yr	≥3.5	≥35	≤2.5
2–3 yr	—	≥40	≤2.5
3–4 yr	—	≥45	≤2.5
4+ yr	—	≥50	≤3.0
Adult	≥4.0	≥50	≤4.0
Ulnar nerve			
Preterm (33–39 wk)	—	≥18	≤3.3
0–1 mo	≥1.5	≥20	≤3.0
1–6 mo	≥2.5	≥25	≤3.3
7–12 mo	≥3.0	≥35	≤2.5
1–2 yr	≥2.5	≥40	≤2.5
2–3 yr	—	≥40	—
3–4 yr	—	≥45	—
4+ yr	—	≥50	—
Adult	≥6.0	≥50	≤3.3
Peroneal nerve			
0–1 mo	≥1.5	≥20	≤3.0
1–6 mo	≥1.5	≥25	≤2.5
7–12 mo	≥2.0	≥30	≤3.5
1–2 yr	≥1.5	≥35	≤3.5
2–3 yr	—	≥40	—
3–4 yr	—	≥40	—
4+ yr	—	≥40	—
Adult	≥2.0	≥40	≤6.5
Tibial nerve			
Preterm (33–36 wk)	—	≥14	—
Preterm (37–39 wk)	—	≥18	—
0–1 mo	—	≥20	≤4.5
1–6 mo	—	≥20	≤4.0
7–12 mo	—	≥25	≤3.5
1–2 yr	—	≥30	≤3.0
2–3 yr	—	≥35	≤4.0
3–4 yr	—	≥40	≤4.0
4–6 yr	—	≥40	≤4.5
6+ yr	—	≥40	≤5.0
Adult	≥4.0	≥40	≤5.8

A, suggested amplitude; CV, suggested conduction velocity; DL, suggested distal latency; —, data not available.

[a]Adapted from refs. *1, 2, 4–6, 23–25.*

fossa. The peroneal motor study is performed as in an adult, with the E1 recording electrode placed over the extensor digitorum brevis and stimulation occurring at the ankle, fibular head, and popliteal fossa. Recording of tibialis anterior may be used when no response or a very small response is obtained from extensor digitorum brevis. One of the proximal stimulation sites may be omitted in children, unless, of course, there is a question of a peroneal neuropathy at the fibular head, such neuropathy being quite rare in children.

In infants, the most difficult aspect of the median antidromic sensory study involves the placement of the active and reference electrodes. The active electrode may be placed on the second digit and the reference on the third, or the electrodes can be cut in half and both placed on the second or third digit. If this is unsuccessful, ring electrodes may be used instead. The stimulation site is, as usual, at the wrist. In neonates and some infants, surface recordings of sural nerve sensory action potentials are often obscured by artifact caused by high skin impedance and short interelectrode distances. In this age group, the most technically reliable sensory study to obtain in the lower extremity is the orthodromic medial plantar study, recording the tibial nerve at the ankle and stimulating the medial plantar region of the sole.

Full-term newborns typically have nerve conduction velocities that are approximately half those expected in an adult. Nerve conduction velocities increase steadily during the first 3 to 5 yr of life, because of growth in axon diameter and thickening of myelin. Thus, adult nerve conduction values cannot be reliably expected until 3 to 5 yr of age, although some children's responses reach those values earlier. Normal amplitudes are also diminished in children compared with adults, especially in the first year of life. In a child younger than 3 to 5 yr, it is important to consult age-matched reference values in interpreting the results of nerve conduction studies. Tables 1 and 2 list suggested normal values adapted and summarized from published data on surface recordings, and also include adult normal values for comparison. Most studies seeking to establish normal values in children focus on motor nerve conduction studies, thus, Table 1 is more comprehensive than Table 2.

Elicitation of late responses may produce significant discomfort in a child, and should not be performed unless they can help answer a specific question, for example, when Guillain–Barré syndrome is a possible diagnosis. If a number of late response studies are necessary, sedation or anesthesia should be considered. Despite incomplete myelination, short stature results in F-response latencies that are shorter in children than in adults. Age-matched normal values, such as the ones listed in Table 3 should be used. H-reflexes may be obtained in the upper as well as the lower extremities in the first year of life, but are rarely necessary in the pediatric population.

2.3. Needle EMG

For the needle examination, a 30-gauge, 25-mm disposable concentric needle (the smallest commercially available) is almost always the best choice. School-age children often ask if "it will hurt." One can reply that it will "pinch" or say that it will be like a blood draw, while avoiding the term "needle." The needle itself can be described as a "microphone." Older children and adolescents may often be examined as thoroughly as adults, but it is still advisable to examine the fewest muscles necessary to answer the question at hand. These patients sometimes reach a limit of tolerance unexpectedly during the needle examination and become upset with little warning.

Except in cases of neonatal brachial plexus injury or trauma, extensive root screens are rarely required in children. In many cases, the question revolves around a generalized process,

Table 2
Sensory Nerve Action Potentials, Suggested Values

Age	*A (mV)*	*CV (m/s)*
Median nerve (antidromic)		
0–1 mo	≥5	≥25
1–6 mo	≥10	≥35
7–12 mo	≥15	≥30
1–2 yr	≥15	≥40
2–3 yr	—	—
3–4 yr	—	—
4+ yr	—	—
Adult	≥20	≥50
Sural nerve (antidromic)		
0–1 mo	≥6	≥20
1–6 mo	≥6	≥20
7–12 mo	≥6	≥25
1–2 yr	≥6	≥30
2–3 yr	≥6	≥35
3–4 yr	≥6	≥40
4+ yr	≥6	≥40
Adult	≥6	≥40
Medial plantar nerve orthodromic in children		
0–1 mo	≥10	—
1–6 mo	≥15	≥35
7–12 mo	≥15	≥35
1–2 yr	≥15	≥35
2–4 yr	—	—
4+ yr	—	—
Adult (antidromic)	≥2	≥35

A, suggested amplitude; CV, suggested conduction velocity; —, data not available. Sural sensory responses may be obscured by subcutaneous tissue in neonates and infants; medial plantar studies may be more accurate in this age group.

[a]Adapted from refs. *1–3, 6, 25.*

therefore, a limited needle examination often yields the necessary data. In infants and toddlers, poor cooperation often makes it impossible to evaluate both insertional activity and voluntary activity in the same muscle. In the upper extremities, insertional activity is most easily observed in triceps and first dorsal interosseous, whereas motor unit activity is better assessed in biceps and flexor carpi radialis. In the lower extremities, medial gastrocnemius and vastus lateralis are better for insertional activity, whereas tibialis anterior and iliopsoas are preferred for motor unit potential (MUP) analysis.

As in adults, if there is a question of a polyneuropathy, it is important to examine distal muscles, although it is rarely necessary to study intrinsic foot muscles in young children. Children with possible myopathy should have several proximal muscles evaluated, but one extremity (preferably lower) should be spared so that histological examination of a muscle biopsy performed at a later time will be free of potential artifacts from needle injury.

Table 3
F-Wave Responses, Suggested Values

Age	*Latency (ms)*
Median nerve	
0–1 mo	≤20
1–6 mo	≤20
7–12 mo	≤21
1–2 yr	≤21
2–4 yr	≤21
4–6 yr	≤23
6–14 yr	≤29
Adult	≤31
Ulnar nerve	
0–1 mo	—
1–6 mo	≤17
7–12 mo	≤17
1–2 yr	≤17
2–4 yr	—
4–6 yr	—
6–14 yr	—
Adult	≤32
Peroneal nerve	
0–1 mo	≤26
1–6 mo	≤27
7–12 mo	≤29
1–2 yr	≤30
2–4 yr	≤34
4–6 yr	≤36
6–14 yr	≤43
Adult	≤56
Tibial nerve	
0–1 mo	—
1–6 mo	—
7–12 mo	≤24
1–2 yr	≤26
2–4 yr	—
4–6 yr	—
6–14 yr	—
Adult	≤56

—, data not available.
[a]Adapted from refs. *1, 3, 25, 26.*

3. COMMON REFERRALS TO THE PEDIATRIC EMG LABORATORY

Deciding whether to request or perform an EMG in a particular case is sometimes difficult, because the number of available diagnostic modalities has increased dramatically in recent years. Genetic testing is available for many neuropathies and muscular dystrophies, and may make EMG unnecessary in certain patients. However, there are many children with

acquired conditions and unusual presentations of inherited disorders in whom EMG is an essential test, and the volume of EMG referrals, in our experience, has remained steady.

The differential diagnosis of hypotonia in infancy and early childhood is vast, and includes a number of peripheral nervous system processes. However, it is important to remember that the majority of cases of infant hypotonia arise from central causes. An extensive peripheral nervous system evaluation should be pursued only if supported by the history and examination. Peripheral nervous system lesions may localize to the anterior horn cell, peripheral nerve, neuromuscular junction, or muscle. If a congenital myopathy or muscular dystrophy lies in the differential diagnosis, it is often useful to obtain a set of muscle enzymes (creatine kinase, aldolase, alanine aminotransferase, aspartate aminotransferase, and lactate dehydrogenase) before requesting or performing an EMG or muscle biopsy. It is important to remember that muscle enzymes may be artifactually elevated immediately after needle EMG is performed. A significant elevation of one or more enzymes (the elevated enzymes should include either creatine kinase or aldolase, because these are the ones that are most specific to muscle) will justify a muscle biopsy as the next step. Mild abnormalities or normal levels of these enzymes do not always exclude a congenital myopathy; in those cases, an EMG may be helpful in directing further studies. The precise sensitivity of EMG in the evaluation of the hypotonic infant is unclear. One study reported an 80% overall sensitivity for the correct diagnosis, whereas another recorded a 65% sensitivity for SMA and 10% for myopathy.

A neonate with a birth injury to the brachial plexus may present with a weak or flaccid upper extremity. It is rare for alternative diagnoses to be plausible, therefore, the question for the electromyographer usually revolves around issues of severity and prognosis. Except in the most severe cases, an EMG is most helpful after the first few months of life, when the presence or absence of chronic reinnervation and axonal continuity may best be assessed.

Toddlers and older children may present with delayed motor milestones or difficulty walking. As in the evaluation of hypotonia, the differential diagnosis is broad, and may include almost any segment of the peripheral nervous system. Proximal weakness, often detected by the presence of Gowers sign, Trendelenburg gait, or both, typically suggests the possibility of Duchenne, Becker, or limb–girdle muscular dystrophy. If such a patient has significantly elevated muscle enzyme levels, either molecular genetic testing or muscle biopsy will yield a diagnosis in most cases. When such an evaluation is unrevealing, an EMG may be useful in assessing the child for a neuropathic process, such as SMA type III (also known as Kugelberg–Welander disease). SMA type III may resemble a muscular dystrophy with a proximal distribution of weakness and a mild elevation of the creatine kinase level.

Abrupt onset gait difficulty may be caused by acute inflammatory demyelinating polyneuropathy (AIDP; Guillain–Barré syndrome), an inflammatory myopathy, tick paralysis, spinal cord conditions (mass lesion or transverse myelitis), or central causes. Although poliomyelitis, fortunately, has almost been eradicated in much of the world, West Nile virus has also been found to produce a similar syndrome of acute motor paralysis. A detailed skin and scalp examination is important in such acutely weak children, because tick paralysis is readily cured by removing the tick. Because ataxia is often observed in AIDP, it may be difficult in some cases to distinguish between that condition and other causes of ataxia, such as acute cerebellitis. If a particular diagnosis cannot be supported by other findings, such as areflexia, cytoalbuminological dissociation, rash, elevated creatine kinase level, or nerve root enhancement on MRI imaging of the spine, EMG may be helpful in distinguishing between the possibilities.

Chronic gait difficulty associated with areflexia, sensory loss, pes cavus, distal weakness, or a combination of these suggests the presence of an isolated inherited polyneuropathy, such as Charcot–Marie–Tooth (CMT) disease, also called hereditary motor sensory neuropathy. In some instances, a demyelinating polyneuropathy may be associated with a more generalized neurodegenerative disorder, such as metachromatic leukodystrophy, especially if there is an associated cognitive decline.

4. IMPORTANT FINDINGS IN THE PEDIATRIC EMG LABORATORY

4.1. Motor Neuron Disease

Because of the development of the polio vaccine, SMA has become the dominant anterior horn cell disease affecting children in developed countries. The onset of the most common variant, type I (Werdnig–Hoffman disease), is typically in the first 6 mo of life. The infants present with hypotonia, weakness, and delayed motor milestones. Mothers may report decreased fetal movements. Examination is notable for areflexia or hyporeflexia, hypotonia, weakness, and preserved extraocular movements. Tongue fasciculations may sometimes be observed, but are not a reliable finding, especially in young infants, and the lack of tongue fasciculations should never be used to exclude SMA. These children never sit independently and never walk. The majority do not survive the second year of life.

The onset of SMA type II is typically from 6 to 18 mo, with some cases beginning as early as the neonatal period. These children often present with motor delays. They eventually sit independently but never walk, and survive only to adolescence or early adulthood. The gait difficulties that are often the first signs of SMA type III usually begin after 18 mo. These patients walk independently for most of their lives. Life expectancy is normal in most cases of SMA type III, although some patients require wheelchair assistance as adults.

SMA is caused by a deletion in the survival motor neuron (*SMN*) gene on chromosome 5q13. Until this discovery, EMG and muscle biopsy were the principal means of confirming the diagnosis during life. Genetic testing for deletions in *SMN* is now readily available, but EMG is still helpful in many cases, especially if the presentation is mild or otherwise unusual, or if an infant develops respiratory failure before the results of genetic testing become available.

As in adult motor neuron disease, it is critical to document normal sensory responses on nerve conduction studies; if these are abnormal, the diagnosis should remain in serious doubt. Compound motor action potential (CMAP) amplitudes are often diminished in SMA type I, but may be minimally reduced or normal in SMA types II and III. During the needle examination, at least three limbs should be studied. Cranial muscles may be substituted for one limb, especially if tongue fasciculations are observed. Examination of paraspinal muscles is not generally helpful and should be avoided. Both ongoing denervation and chronic reinnervation may be observed. In adults, chronic reinnervation should be demonstrated in most muscles of at least three limbs to meet criteria for motor neuron disease, but in children it is not always possible to do this extensive a needle examination, and any findings should be confirmed by genetic testing. Fasciculation potentials are observed more rarely in SMA as compared with amyotrophic lateral sclerosis, and the MUP abnormalities are generally symmetrical in SMA.

4.2. Sensory Neuropathy and Neuronopathy

It is rare to find an isolated sensory neuropathy or sensory neuronopathy in childhood. In infants, the sural and superficial peroneal sensory responses are often difficult to record. Thus, in children of that age, the medial plantar sensory responses and upper extremity

sensory responses must be checked before concluding that the patient has a true loss of sensory responses. Age-specific normal values must be used in children younger than 3 to 5 yr.

If a true sensory neuropathy or sensory neuronopathy is present, the most likely causes are Friedreich's ataxia and the hereditary sensory and autonomic neuropathies. Because genetic testing for the triplet-repeat expansion in Friedreich's ataxia is available, patients with the typical clinical presentation are often diagnosed without the assistance of an EMG. Associated findings include ataxia, absent lower extremity reflexes, extensor plantar responses, thinning of the spinal cord on spine MRI, normal brain MRI early in the course, onset before 20 yr, pes cavus, dysarthria, distal sensory loss, optic atrophy, diabetes, and cardiomyopathy.

4.3. Brachial Plexus

Brachial plexus injuries in children most commonly occur at birth, as a result of shoulder dystocia (difficulty in extracting the shoulder from the birth canal). Risk factors include difficult delivery and large birthweight. As might be expected from the mechanism of injury, upper plexus injuries (Erb's palsy) predominate. Lower brachial plexus lesions (Klumpke's palsy) and total plexus injuries (Erb-Klumpke paralysis) are less common. The neonate will have flaccid weakness of one upper extremity.

The history and examination are, in most cases, diagnostic of perinatal brachial plexus injury. A careful examination will suggest the root levels involved. In upper plexus lesions, the posture known as the "waiter's tip" is usually found, consisting of arm adduction, elbow extension, forearm pronation, and wrist flexion. In lower plexus lesions, wrist and finger flexors and intrinsic hand muscles are weak. A Horner's syndrome is typically present. A combination of these findings or complete upper extremity paralysis suggests total plexus involvement.

Initial management includes gentle immobilization of the affected extremity against the abdomen and range of motion exercises, preferably under the direction of a trained physical therapist. In almost all cases, observation and conservative management are recommended until 3 to 6 mo of age. If there is no improvement at that point, evaluation for possible microsurgical repair may be indicated, including EMG and MRI.

In this setting, the purpose of performing an EMG study is to determine the localization and severity of the injury. The severity is largely dependent on the localization, because discontinuity of the axon, most commonly caused by avulsion of the nerve root, is associated with a much poorer prognosis than "stretching" of the brachial plexus structures.

If brachial plexus injury and root avulsion were mutually exclusive conditions, the study would be relatively simple, because, in the former condition, sensory responses would be absent, whereas, in the latter, they would be preserved. However, in many cases, the two lesions coexist, making abnormal sensory nerve action potentials (SNAPs) less reassuring than they would be under other circumstances.

The needle study is the key to determining prognosis. Because examination of the cervical paraspinal muscle is impractical in most infants, the presence of MUPs on needle EMG at various root levels confirms continuity of the axon and, thus, the likelihood of at least partial recovery of function. This assessment can only be made several months after birth, after reinnervation has begun to occur, because a premature examination may yield an inaccurately grim prognosis.

Precise localization among the root, trunk, division, cord, and nerve structures requires the evaluation of a large number of muscles, but unless the infant is anesthetized, this is

not usually possible. Thus, because of the high likelihood that the study will be terminated prematurely, the choice of muscles is critical. If an infant has a typical upper plexus presentation, the deltoid, biceps, and triceps muscles should be studied first. Comparing deltoid and biceps can help distinguish between root and plexus involvement; both muscles will be abnormal when a C5–C6 root lesion is present, but only one may be abnormal in lesions of the brachial plexus. Unfortunately, examination of the rhomboids and serratus anterior is rarely practical in infants, limiting the precision of localization within the brachial plexus.

When a Horner's syndrome or other signs of lower plexus involvement are present, a distal muscle, such as the first dorsal interosseous, should be among the first muscles examined. Depending on the clinical picture and the tolerance of the patient, other muscles that may be studied include the supraspinatus, extensor digitorum communis, flexor digitorum superficialis, and abductor digiti quinti. If the triceps has been successfully examined, needle EMG of extensor digitorum communis may not be necessary. As for median-innervated nerve muscles, examination of the flexor digitorum superficialis is usually more informative than that of abductor pollicis brevis, because the infant is more likely to activate the former.

Muscle activation in infants is often fleeting. For flexor muscles, tickling or gentle pinching of the fingers and hand may stimulate withdrawal when the needle itself fails to induce this response. The infant is less likely to activate extensor muscles under these circumstances. It is sometimes difficult to decide how long to examine a muscle, especially if the child is crying and it seems that the time remaining in the study is rapidly diminishing. When no MUPs are present, tickling or pinching to activate muscle contractions should be performed at least once for each major level (C5–C6, C7, and C8–T1), and preferably in multiple muscles. Because a decision whether or not to perform surgery may hinge partly on the study results, any examination that is too truncated to be reliable should be repeated under anesthesia.

4.4. Polyneuropathies

AIDP (Guillain–Barré syndrome) may occur at almost any age in childhood, from 1 mo to adulthood. The classic clinical presentation is the same as in adults, with an acute onset of ascending paralysis, areflexia, and cytoalbuminological dissociation in the cerebrospinal fluid, with protein values often ranging from 80 to 200. An EMG may be very helpful in confirming or casting doubt on the diagnosis. The use of spine MRI with gadolinium enhancement to detect nerve root enhancement is also becoming more widespread. The initial symptoms may be atypical in some patients, consisting primarily of numbness, paresthesias, or pain.

Nerve conduction studies are most useful in the evaluation of possible AIDP. In the classic demyelinating form of AIDP, the diagnostic findings are temporal dispersion and conduction block in the setting of slow conduction velocities and prolonged distal latencies. The criteria for these findings are generally the same in children as in adults: at least 15% prolongation of CMAP duration on proximal vs distal stimulation for temporal dispersion, at least 50% diminution of CMAP amplitude on proximal vs distal stimulation for partial conduction block, and an absent CMAP on proximal stimulation for complete conduction block. The presence of temporal dispersion excludes partial or complete conduction block. Another criterion for nonuniform slowing suggesting an acquired demyelinating process is a significant

disparity in nerve conduction velocities between different nerves, usually at least 10 m/s difference within the upper or lower extremities, and at least 15 m/s difference between an upper and a lower extremity. The usual adult standards stipulating the number of nerves involved may not always be practical to apply in children, because the study may be terminated early. Evidence for axonal loss may be present, but unless severe, this is usually a secondary phenomenon.

In some early or mild cases, temporal dispersion, conduction block, and conduction velocity slowing may not be present. Prolonged or absent F-responses and H-reflexes may be early signs of AIDP, as may be reduced recruitment on an otherwise normal needle EMG study. If tolerated, F-waves should be performed on several nerves in multiple extremities. H-reflexes are extremely uncomfortable, especially for a child, and, thus, should be avoided. These early electrophysiological signs of AIDP may be subtle and nonspecific; if the diagnosis cannot clearly be supported by other data, a repeat study later in the course may provide more definite evidence.

The less common, primarily axonal form of Guillain–Barré, known as acute motor axonal neuropathy, was first recognized in China, and most commonly occurs there, but has also been described in the United States, Europe, and other parts of Asia. In adults, the prognosis is clearly worse in axonal variants. Children with acute motor axonal neuropathy have a worse course acutely (many require assisted ventilation) and recover more slowly than those with AIDP, but the long-term prognosis is generally good if they survive the acute phase. Conduction velocities are normal or mildly slow. There is evidence for axonal loss both on nerve conduction studies and needle EMG.

A generalized sensorimotor polyneuropathy in the setting of long-standing symptoms may be caused by chronic inflammatory demyelinating polyradiculoneuropathy (CIDP) or an inherited polyneuropathy, such as CMT (also known as hereditary sensory and motor neuropathy), metachromatic leukodystrophy, Cockayne syndrome, Pelizaeus-Merzbacher disease, or adrenomyeloneuropathy. An EMG can determine whether a polyneuropathy is present, and may, in some cases, help distinguish between acquired and genetic causes. Traditionally, signs of nonuniform slowing of nerve conduction, such as temporal dispersion, conduction block, and significant disparities in nerve conduction velocities between different nerves have indicated the presence of an acquired polyneuropathy, whereas uniform slowing has been associated with an inherited polyneuropathy. However, numerous reports have demonstrated nonuniform slowing in some instances of inherited polyneuropathies, including metachromatic leukodystrophy, hereditary neuropathy with liability to pressure palsies, the X-linked variant of CMT (CMTX), and adrenomyeloneuropathy.

Children with CIDP generally have manifestations of motor delay or impairment. The weakness may show a distal predominance or may be diffusely distributed. Other findings include sensory loss in some patients, normal cranial nerve function, and hyporeflexia or areflexia. CIDP may develop at any age, and has even been reported to be present at birth.

The electrophysiological findings in CIDP are similar to those in fully developed AIDP. Nerve conduction velocities may be as slow as 2.3 m/s. Temporal dispersion and conduction block are classic findings. Needle EMG reveals evidence for secondary axonal loss. The chronicity or relapsing nature of the course distinguishes CIDP from AIDP. It is sometimes difficult to confirm CIDP based on electrophysiology alone, especially in light of the exceptions to the uniform vs nonuniform slowing rule that have recently been described. Palpable enlarged nerves and pes cavus may be present in CIDP as well as in CMT. Elevated protein in cerebrospinal fluid

is found in CIDP, but also in inherited disorders such as metachromatic leukodystrophy and CMT. The absence of cognitive involvement, MRI abnormalities, and long-tract signs is suggestive of CIDP rather than an inherited leukodystrophy, but nerve biopsy is still required to dispel remaining doubts, especially if there is a question of CMT.

CMT type I is the most common inherited polyneuropathy. It is characterized by a predominantly demyelinating pattern on EMG, with nerve conduction velocities typically in the teens. The pattern of inheritance is autosomal dominant. Initial symptoms may include distal lower extremity weakness and wasting, with slow progression over several decades. Hyporeflexia, palpable enlarged nerves, pes cavus, distal weakness and wasting, and distal sensory loss are classic physical findings.

Primary axonal loss is found in CMT type II. Nerve conduction velocities are normal or mildly slow. Onset is typically in early adulthood, and the course is mild. Inheritance is usually autosomal dominant, although autosomal recessive kindreds with moderately slow nerve conduction velocities have been described.

CMTX is characterized by primary axonal loss combined with secondary demyelination. The most prominent EMG findings are reduced CMAP and SNAP amplitudes, but nerve conduction velocities are moderately slow, which helps distinguish CMTX from CMT type II electrophysiologically, and there may be heterogeneity of conduction parameters, including conduction block, giving it an "acquired" appearance. Heterozygote females have mild symptoms, whereas affected males have the full spectrum of manifestations, including pes cavus, distal muscle weakness and atrophy, and distal sensory loss.

Type III (Dejerine–Sottas disease) is associated with severe demyelination. EMG demonstrates profound slowing of nerve conduction velocities, typically in the single digits, with temporal dispersion of CMAPs. Onset may occur anytime between birth and 2 yr. Delayed motor milestones, weakness, areflexia, and hypotonia typically occur. Elevated cerebrospinal fluid protein may be present. Palpably enlarged nerves are a classic but inconsistent finding.

4.5. Mononeuropathies

Mononeuropathies are rare in children compared with adults. Median neuropathies at the wrist may be associated with idiopathic carpal tunnel syndrome, but may also occur in the setting of acute trauma, chronic trauma via sports, mucopolysaccharidosis (Hurler/Scheie, Hunter, and Maroteux–Lamy), mucolipidosis (types II or III), or scleroderma. In some cases, the median neuropathy may be the first specific finding that indicates a systemic condition. Thus, the possibility of a mucopolysaccharidosis or mucolipidosis should always be considered if a child is diagnosed with a distal median neuropathy. Proximal median neuropathies seem to occur at least as often as median neuropathies at the wrist, and are typically caused by trauma, such as a fracture of the humerus or a laceration. The classic clinical picture of a median neuropathy occurs in some of these patients but is not universal. A careful needle EMG is important in cases of median neuropathy to investigate the possibility of a proximal lesion.

An isolated ulnar neuropathy may occur at the level of the elbow, forearm, wrist, or hand. It is most commonly caused by trauma, including fracture in the supracondylar area or forearm, laceration anywhere along the course of the nerve, acute compression during surgery, and chronic compression during activity (e.g., pressure from bicycle handlebars and wheelchair armrests). Cubital tunnel syndrome occurs occasionally. Because direct trauma is more

common in children than compression, axonal pathophysiology is correspondingly observed more often in these patients.

In contrast to median and ulnar mononeuropathies, radial neuropathy may be observed at any age, from newborns to 17-yr-old children, and generally has a good prognosis. Because of the location of the radial nerve, it is susceptible to prenatal or perinatal intrauterine compression. Other causes of compression and entrapment, as well as direct trauma from fractures and lacerations have also been identified. Weakness of extensor muscles and wristdrop are the most common signs. Radial sensory nerve conduction studies should be performed in such cases. If normal values are not available for children younger than 3 to 5 yr and the symptoms are unilateral, a study in the contralateral arm may be used for comparison. Radial motor studies are difficult because of the technical challenges in young children and their limited tolerance for the strong stimuli that may be required. The needle EMG study is critical to localize the lesion. With adequate sedation or anesthesia, detailed needle examination is possible even in neonates. Unless symptoms progress or there is evidence for entrapment, surgical exploration is generally unnecessary, and most patients may be managed conservatively.

Peroneal neuropathy is the most common lower extremity mononeuropathy. It may be caused by direct trauma during sports activities, compression from casts and other devices attached to the leg, or entrapment from fibrous bands. Patients whose lesions were predominantly demyelinating had a better outcome than those with significant axonal loss, as indicated by a low-amplitude or absent peroneal CMAP.

6. Neuromuscular Junction

Disorders of neuromuscular transmission are caused by impaired transmission of acetylcholine across the neuromuscular junction. In children, the most common causes of these disorders are myasthenia gravis (of which there are three types: neonatal, congenital, and juvenile) and botulism. Lambert–Easton myasthenic syndrome is rare in this age group.

In neonatal myasthenia gravis, maternal antibodies to the acetylcholine receptor cross the placenta and enter the fetal circulation. This occurs in 12% of infants born to mothers with symptomatic or quiescent autoimmune myasthenia gravis. Soon after birth, an affected neonate develops respiratory distress, feeding difficulties, and weakness. In one-third of cases, ventilatory support and nasogastric tube feedings are required until the baby clears the antibodies from the circulation, which usually takes several weeks. Oral pyridostigmine may be helpful in severe cases.

Congenital myasthenic syndrome is caused by genetic mutations affecting the neuromuscular junction. The most common mutations are in genes coding for the acetylcholine receptor, but presynaptic lesions may also occur. The inheritance is usually autosomal recessive. These patients do not have antibodies to the acetylcholine receptor. Onset is typically in the first year. Compared with juvenile myasthenia gravis, there are fewer fluctuations in weakness and more prominent ocular features. Episodes of apnea may be present in some subtypes such as choline acetyltransferase deficiency and rapsyn deficiency. Cholinesterase inhibitors may alleviate symptoms, but immune modulating therapies are ineffective, as would be expected from the pathophysiology.

Juvenile myasthenia gravis has the same autoimmune origins as adult myasthenia gravis, but the onset is during childhood or adolescence. Symptoms such as ophthalmoplegia, ptosis (bilateral or unilateral), orbicularis oculi or facial weakness, dysphagia, dysarthria, and dyspnea

in a fluctuating pattern may suggest the diagnosis. Isolated generalized weakness may present a challenge, because the differential diagnosis is broad. The evaluation is essentially the same as in adult myasthenia gravis. Diagnosis may be made through acetylcholine receptor antibody testing, EMG, the edrophonium test, or a combination of these. Once the diagnosis is confirmed, a chest CT study should be performed, although the incidence of thymoma is lower in children than in adults. Symptomatic therapy with acetylcholinesterase inhibitors and immune modulating medications may be used, as in adults. Medications known to exacerbate myasthenia gravis should be avoided.

The most common form of botulism in childhood is infant botulism, caused by ingestion of *Clostridium botulinum* spores from the soil. Endemic states include Pennsylvania, Utah, and California. Botulinum toxin inhibits the release of acetylcholine from the presynaptic terminal. Infants typically present with the acute onset of constipation, extremity weakness, bulbar weakness, sluggish pupillary responses, and oculomotor palsies. Reflexes may be either preserved or diminished. Because the infants ingest spores rather than preformed toxin as in adult botulism, the illness is caused by low levels of subacute toxin production rather than an overwhelming single dose. Thus, stool samples rather than blood samples should be sent for botulinum toxin testing in patients suspected of having infant botulism.

Routine nerve conduction studies are an important component of an evaluation for disorders of neuromuscular transmission. Not only must a generalized polyneuropathy be excluded, but certain findings may be suggestive of particular neuromuscular transmission defects, such as low-amplitude CMAPs in presynaptic disorders and repetitive CMAPs on single supramaximal stimulation in slow channel syndrome (a form of congenital myasthenic syndrome). During repetitive stimulation, the accessory nerve should be the first one studied, unless there is focal or maximal weakness in the distribution of another nerve, such as the ulnar or median nerve. Accessory nerve studies are more sensitive than ulnar studies, and in children, the study may need to be terminated at any time because of patient discomfort. In infants, repetitive nerve stimulation should only be performed with the patient under sedation or anesthesia.

In postsynaptic disorders, such as neonatal, juvenile, or postsynaptic congenital myasthenia, the characteristic finding on low-frequency (2–3 Hz) repetitive stimulation is a greater than 10% decrement in the CMAP amplitude. The nadir typically occurs at the fourth or fifth stimulus. If the nadir occurs earlier or later, the decrement is most likely artifactual. In presynaptic disorders, such as botulism and the presynaptic forms of congenital myasthenia, low-frequency repetitive stimulation may also produce a decremental response, but it is not observed as consistently as in postsynaptic disorders. The more sensitive test in suspected disorders of presynaptic transmission is high-frequency repetitive stimulation at 20 to 50 Hz, which yields an incremental response in nearly all such patients. This pattern may also be observed occasionally in patients with severe postsynaptic defects. The threshold for determining abnormal facilitation of the CMAP amplitude on high frequency repetitive stimulation is typically considered a 100% increment. On the rare occasion when an older child is suspected of having a presynaptic disorder, such as adult botulism or Lambert–Eaton myasthenic syndrome, a single supramaximal stimulus pre- and then again post- 10 seconds of maximal contraction of the muscle being tested may substitute for high-frequency repetitive stimulation. Because high-frequency stimulation is even more uncomfortable than low-frequency stimulation, this procedure is generally performed under anesthesia in any age group.

Abnormalities in at least two nerves should be identified before diagnosing a child with a disorder of neuromuscular transmission. This may be difficult in a child who is uncomfortable or uncooperative. Single-fiber EMG, a technique used often in adults, is impractical in most children. If the study is technically inadequate, other diagnostic modalities (acetylcholine receptor antibody levels and edrophonium testing) may be considered, or the study may be repeated under anesthesia.

Low-amplitude, short-duration MUPs suggestive of a myopathy are often observed on needle EMG in patients with disorders of neuromuscular transmission. Fibrillation potentials may be observed in cases of botulism and severe cases of autoimmune myasthenia gravis. Some investigators have found neurogenic findings in patients with botulism, which may be explained by the functional denervation that can occur in this disorder. In a child with myopathic EMG findings of unclear etiology, it is important to consider the possibility of a disorder of neuromuscular transmission and, if indicated, perform repetitive stimulation studies.

4.7. Muscle

Before the discovery of dystrophin and the subsequent development of genetic testing for Duchenne and Becker muscular dystrophy, EMGs were commonly used in the diagnostic evaluation of boys who had gait difficulties. The question of possible myopathy or muscular dystrophy does still arise in the pediatric EMG laboratory, but the patients who are referred often have atypical presentations or rare conditions. Congenital myopathies include centronuclear myopathy, nemaline myopathy, and central core disease. Muscular dystrophies include congenital muscular dystrophy, Emery–Dreifuss dystrophy, facioscapulohumeral muscular dystrophy, limb–girdle muscular dystrophy, Duchenne muscular dystrophy, and Becker muscular dystrophy. Children may also present with symptoms suggestive of a metabolic myopathy, categorized as glycogenosis or fatty acid oxidation disorder. Mitochondrial myopathies are also considered a form of metabolic myopathy. Glycogenoses include acid maltase deficiency (glycogenosis type II, Pompe's disease), myophosphorylase deficiency (glycogenosis type V, McArdle's disease), and phosphofructokinase deficiency (glycogenosis type VII). Inflammatory myopathies include dermatomyositis and polymyositis.

The evaluation of a possible primary muscle disorder in the EMG laboratory can be difficult. Because normal MUPs are smaller in infants and children than in adults, the threshold for defining a unit as myopathic is different. In infants, MUPs are typically biphasic, with durations of 1 to 4 ms and amplitudes less than 100 μV. In addition, inconsistent muscle activation in some children may make it difficult to detect short-duration MUPs and early recruitment with confidence. When in doubt, it is always more prudent to conclude that the results of a study are normal rather than overemphasize potentially false positive findings. The EMG report should indicate that normal study results do not exclude a myopathic process.

In cases of possible myopathy or muscular dystrophy, it is important to examine proximal muscles and any other muscles that are weak. Iliopsoas should be studied whenever possible—in infants and younger children, it is crucial to locate the femoral pulse and ensure that the needle is not placed in the femoral artery. If the patient is so agitated that this cannot be guaranteed, it is advisable to defer study of that muscle. Other proximal muscles in the upper and lower extremities should also be studied.

In many cases of noninflammatory myopathy and muscular dystrophy, needle EMG will demonstrate low-amplitude, short-duration MUPs, sometimes accompanied by fibrillation potentials and positive sharp waves. These findings may also be observed in disorders of

neuromuscular transmission. Abnormal spontaneous activity will typically be more prominent in cases of inflammatory myopathy. Early recruitment, in which multiple MUPs are activated despite minimal force generation, is another sign of myopathy, but is very difficult to assess in infants and children. Myopathic findings do not generally help to distinguish between the various myopathic disorders, unless there is very prominent spontaneous activity, myotonia, or both.

A myotonic discharge is a distinct finding of abnormal spontaneous activity that may best be described as a series of rapidly firing fibrillation potentials or positive sharp waves, with fluctuating amplitude and frequency and a duration of 2 to 20 s. The fluctuations produce a sound that has been described as resembling that of a "dive bomber," but the sound of a motorcycle engine may be a better analogy. Myotonic discharges are often elicited with small insertions of the EMG needle. These discharges may be missed, however, especially in children, if the needle EMG study is technically limited or only performed on a few muscles. The absence of myotonic discharges on a needle EMG study does not entirely exclude that entity.

Classically, myotonic discharges are associated with myotonic dystrophy, myotonia congenita, paramyotonia congenita, and hyperkalemic periodic paralysis. However, because it is, in a sense, a severe manifestation of positive sharp waves and fibrillation potentials, it may also be observed in some myopathic disorders not caused by channelopathies, such as acid maltase deficiency, centronuclear myopathy, polymyositis, and dermatomyositis.

4.8. Myotonic Disorders

There are four classic primary myotonic disorders: myotonic dystrophy, myotonia congenita, paramyotonia congenita, and hyperkalemic periodic paralysis. There are two types of myotonic dystrophy (type 1 and type 2). Type 1 is the most common form in childhood; type 2 is mainly an adult disorder. Onset of symptoms of type 1 myotonic dystrophy is in late childhood or adolescence; there is, however, a variant that is symptomatic at birth. A fully developed case of myotonic dystrophy has many distinctive features, including frontal balding, temporal wasting, distal weakness, and myotonia on examination. The neonatal variant may present with arthrogryposis multiplex congenita. These patients are often diagnosed directly by DNA testing. However, the findings may be more subtle early in the course, therefore, an EMG may be helpful in those cases. Routine motor and sensory nerve conduction studies are usually normal, although some reports demonstrate mild slowing of conduction velocities. Repetitive stimulation produces CMAP decrements. Needle EMG reveals, in addition to myotonic discharges, other abnormal spontaneous activity, and short-duration, low-amplitude MUPs, demonstrative of an underlying myopathic process.

There are autosomal dominant (Thomsen's disease) and recessive (Becker's disease) variants of myotonia congenita, which is caused by mutations in the chloride channel. Muscle stiffness may be present at rest, but is relieved on activity. Muscle hypertrophy occurs in both disorders. Motor strength is normal in Thomsen's disease, but weakness typically occurs in Becker's disease. Routine motor and sensory nerve conduction studies are normal, but repetitive stimulation and short exercise will lead to CMAP decrements. Needle EMG demonstrates myotonic discharges, but there are usually no other abnormal findings.

Paramyotonia congenita and hyperkalemic periodic paralysis are both caused by mutations in the sodium channel. Signs of paramyotonia congenita are often present in infancy. There may be delayed eye opening after sneezing. In later years, the child may experience stiffening of the extremities, weakness, and falling with activity. This pattern of "paradoxical"

myotonia with activity is the reverse of what is typically found in other myotonic disorders, leading to the name paramyotonia. Symptoms may also be triggered by cold weather. Eating hard-to-chew or cold foods, such as bagels or ice cream, may trigger dysphagia. The results of standard motor and sensory nerve conduction studies are normal. The response to repetitive stimulation or exercise is normal in most cases, but some patients may develop CMAP decrements, especially with cooling of the extremity. Myotonic discharges and other abnormal spontaneous activity may be accentuated by briefly cooling the extremity before needle EMG. There may be electrical silence on needle EMG during an episode.

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REVIEW QUESTIONS

1. You are attempting to perform a sural sensory nerve conduction study in a 6-mo-old infant, but despite several attempts cannot record a reproducible SNAP. The best course at this point is to:
   A. Move on to the next patient.
   B. Record a superficial peroneal sensory response.
   C. Record a saphenous sensory response.
   D. Record a medial plantar sensory response.
   E. Proceed to the motor studies.
2. You are performing nerve conduction studies and EMG on a full term neonate with hypotonia and hyporeflexia. The upper extremity nerve conduction study results are as follows:

	Distal latency	*Amplitude*	*Velocity*
Median motor	2.3 ms	3.1 mV	27 m/s
Median sensory		14 μV	30 m/s

   You conclude that:
   A. The study is normal so far.
   B. The patient has a demyelinating neuropathy.
   C. The patient has an axonal neuropathy.
   D. The patient could have either a motor neuron disorder or a myopathy.
   E. Electrode placement is deficient.
3. Nerve conduction velocities are different in infants and children compared with adults mainly because of:
   A. The shorter distances between electrodes.
   B. Persistence of fetal isoforms of ion channels in the axons.
   C. Incomplete myelination of axons.
   D. Smaller surface area of electrodes used in infants and children.
   E. Differences in axon diameters.
4. While performing needle EMG in infants:
   A. It is almost always possible to record information on both spontaneous activity and voluntary activity from each muscle.
   B. Insertional activity is best recorded in extensor muscles, whereas voluntary units are best observed in flexor muscles.
   C. Fibrillation potentials and positive sharp waves may often be regarded as normal findings.
   D. It is important to examine all relevant muscles despite the patient's level of discomfort.
   E. Needle placement is often incorrect because the infant cannot follow directions.

5. A 4-mo-old full term infant whose birthweight was 9 lb, 11 oz has a flaccid, pronated left upper extremity with flexed fingers. He becomes agitated during nerve conduction studies and you anticipate that he will only tolerate needle EMG on one muscle. Which muscle would you choose?
   A. Biceps.
   B. Triceps.
   C. Flexor digitorum superficialis.
   D. First dorsal interosseous.
   E. Abductor pollicis brevis.
6. A 10-yr-old girl has had difficulty walking with frequent falls for 4 yr, and is slowly worsening. On examination, she has proximal weakness with a Gowers sign. Her serum creatine kinase level is normal. Nerve conduction studies reveal low-amplitude CMAPs and normal SNAPs, and needle EMG demonstrates large motor units with significantly decreased recruitment. The next step in the evaluation should be:
   A. Muscle biopsy.
   B. Genetic testing for PMP22 duplication.
   C. Genetic testing for Duchenne muscular dystrophy deletion.
   D. Genetic testing for *SMN* deletion.
   E. Lumbar puncture for protein level.
7. A 15-yr-old boy is teased in school for lifting his legs up high "like a robot" when he walks. His mother reports that he has always walked this way, but that his symptoms have been worsening. He has calf atrophy, high arches, and foot drop on examination, with loss of vibration sensation and proprioception in his toes. His mother also has high arches and uses a cane, and says that "stumbling" runs on her side of the family. Based on the subtype of his suspected disease that is most common in the general population, the most likely findings on nerve conduction studies will be:
   A. Mildly slow conduction velocities, in the 35 to 40 m/s range.
   B. Low amplitude CMAPs.
   C. Low amplitude SNAPs.
   D. Conduction block.
   E. Markedly slow conduction velocities, in the 20 to 30 m/s range.
8. An 18-yr-old woman with a long history of diabetes has developed progressive difficulty walking and numbness in her toes during the past 5 yr. She was initially thought to have diabetic neuropathy and was referred with that presumptive diagnosis, but when you examine her before nerve conduction studies and EMG, you find that she has marked dysarthria, absent lower extremity reflexes, and extensor plantar responses. What electrophysiological findings would you expect?
   A. Low-amplitude motor and sensory responses.
   B. Low-amplitude or absent sensory responses.
   C. Markedly slow conduction velocities.
   D. Myopathic motor units on needle EMG.
   E. Temporal dispersion.
9. A 12-yr-old girl is referred for numbness in her fingers. She has markedly prolonged median motor distal latencies, abnormal median SNAPs, and evidence for active denervation and chronic reinnervation at the abductor pollicis brevis. The remainder of her electrodiagnostic examination is normal, but you notice during the study that she has a large-appearing head with coarse facial features, short hands and fingers, and a protruding abdomen. She wears hearing aids and sometimes has difficulty understanding your directions during the study. Her parents tell you that previous doctors have just said that she is "different." If you were to include a clinical correlation in your report, you would say:
   A. That median neuropathies are quite common in childhood.
   B. She might have CMT disease.

C. It may be helpful to send urine for mucopolysaccharides.
D. A chromosome analysis may be indicated.
E. She should have serum amino acids and urine organic acids checked.

10. You are in practice in Pennsylvania when you are asked to perform nerve conduction studies and EMG on an inpatient at your local hospital, a 4-mo-old infant boy with an acute onset of weakness and hypotonia. On examination, he is intubated and quite flaccid, with ophthalmoparesis, sluggish pupillary responses, and hyporeflexia. His mother tells you that the first sign of difficulty was constipation 4 d ago. His father is not present because he is a foreman on a crew breaking ground for the new hospital outpatient building across the street. The highest yield studies on nerve conduction studies and EMG will be:
A. Median and peroneal motor studies.
B. Sensory studies.
C. 2- to 3-Hz repetitive stimulation.
D. 20- to 50-Hz repetitive stimulation.
E. Needle EMG.

REVIEW ANSWERS

1. The correct answer is D. The medial plantar sensory response is often easier to obtain than the sural sensory response in infants, in part, because of the substantial amount of subcutaneous tissue over the sural nerve at that age.
2. The correct answer is A. Nerve conduction studies and EMG normal values in infants and children differ markedly from those in adults, but follow a consistent age-dependent pattern. A general rule of thumb is that nerve conduction velocities in newborns are half those of adults.
3. The correct answer is C. Myelination of axons continues after birth, often not reaching maturity until the third to fifth year of life. Smaller axon diameters in infants and children also slow conduction velocities to some extent, but have a more modest effect than incomplete myelination.
4. The correct answer is B. An infant's typical reflexive response to a painful stimulus is withdrawal of the extremity, thus, needle insertion in an extensor muscle will usually result in relaxation of that muscle, whereas insertion in a flexor muscle will lead to activation. Fibrillation potentials and positive sharp waves are abnormal findings in infants, just as in adults. The needle study in an infant is often abbreviated compared with an equivalent study in an adult. It is unnecessary to subject an infant to excessive discomfort, and the electromyographer must carefully triage the muscles studied. In the hands of a skilled electromyographer, needle placement is almost always correct. Anatomical landmarks are critical in infant studies, and larger muscles are studied whenever possible.
5. The correct answer is A. The infant most likely has an Erb's palsy affecting the upper trunk of the brachial plexus caused by traction on the neck and shoulders during a difficult delivery, which can be deduced from the large birthweight and clinical presentation. Thus, a muscle innervated by the upper trunk of the brachial plexus would be ideal if only one muscle can be studied. The biceps, innervated by the musculocutaneous nerve, lateral cord, upper trunk, and C5–C6, is ideal. The other options all indicate muscles innervated by lower portions of the brachial plexus. The deltoid is another upper trunk muscle frequently studied in this setting, but is less likely than the biceps to be activated by an uncooperative infant. At the age of 4 mo, the most likely abnormal finding is chronic reinnervation rather than active denervation, making the biceps a better choice. If the infant were 1-mo old, the deltoid might be preferable. In an infant who tolerates the study well, as full a root/plexus screen as possible should be performed, because the extent and severity of injury can vary significantly.
6. The correct answer is D. This patient most likely has SMA type III (Kugelberg–Welander disease), with the proximal weakness, normal creatine kinase levels, and evidence for motor neuron disease on electrophysiological studies. In almost all cases, the patient will have a homozygous deletion in exon 7, exon 8, or both, of the *SMN* gene. A muscle biopsy should demonstrate fiber type grouping suggesting a neurogenic lesion, but is more invasive than

genetic testing on blood lymphocytes and does not reveal a molecular diagnosis. The patient does not fit the picture of hereditary motor and sensory neuropathy (CMT), which is associated with distal weakness, sensory loss, and a duplication in the *PMP22* gene. This presentation is also not consistent with Duchenne or Becker muscular dystrophy, nor with an inflammatory demyelinating polyneuropathy, which is associated with elevations in cerebrospinal fluid protein. If this patient were a severely hypotonic infant with the same creatine kinase levels and electrodiagnostic results, she would most likely have SMA type I (Werdnig–Hoffman disease).

7. The correct answer is E. This patient most likely has CMT disease. Type 1, a demyelinating form, is most common and statistically would be most likely in this patient without further demographic information, especially with the history of autosomal dominant inheritance. Conduction velocities are markedly slow in most cases. Amplitudes are typically normal in this form, although they can be markedly abnormal in type 2, the axonal form, of CMT. Evidence for multifocal slowing, such as conduction block, is classically found with acquired polyneuropathies, although multifocal slowing in inherited polyneuropathies, including some forms of CMT, does occur.
8. The correct answer is B. This patient seems to have Friedreich's ataxia, which is often associated with diabetes and cardiomyopathy. Onset occurs before age 20 yr. A sensory neuronopathy, with severe reduction in sensory response amplitudes is common, and preserved CMAPs and conduction velocities help distinguish it from CMT.
9. The correct answer is C. This girl most likely has Hurler disease, a mucopolysaccharidosis. A urine screen for mucopolysaccharides is indicated, followed by or concurrent with blood lysosomal enzyme testing. Median neuropathies, including carpal tunnel syndrome, are exceedingly rare in children in the absence of a systemic underlying condition. Assuming that she has no lower extremity symptoms, this presentation is inconsistent with CMT. Chromosome analysis, an amino acid panel, and an organic acid panel would not diagnose Hurler disease.
10. The correct answer is D. Pennsylvania, along with Utah and California, are states with high incidences of infant botulism, because of the presence of botulinum spores in the soil. A parent of an affected infant typically works in construction or is otherwise in close and frequent contact with newly disrupted soil. Constipation is a frequent early sign. The most important studies to perform are high-frequency repetitive stimulation studies at 20 to 50 Hz, which should yield an incremental response. It is important to perform such studies with the patient under anesthesia, because the studies are quite painful. Because this infant is already intubated, this should not be difficult. There may also be a decrement on low-frequency repetitive stimulation, but this finding may not always be present. Other electrodiagnostic studies, including routine nerve conduction studies and needle EMG, abnormalities such as diminished CMAP amplitudes will yield normal results or nonspecific abnormalities.

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