Dedication

To my wife Barbara, with love, on the occasion of 47 years together.
Neurology is an ever-changing field. Standard safety precautions must be followed, but as new research and clinical experience broaden our knowledge, changes in treatment and drug therapy may become necessary or appropriate. Readers are advised to check the most current product information provided by the manufacturer of each drug to be administered to verify the recommended dose, the method and duration of administration, and contraindications. It is the responsibility of the licensed prescriber, relying on experience and knowledge of the patient, to determine dosages and the best treatment for each individual patient. Neither the publisher nor the author assumes any liability for any injury and/or damage to persons or property arising from this publication.
Preface

It is now almost 20 years since I began work on the first edition. Several people have asked how I came to use the format of a *signs and symptoms approach*. The idea was not mine. The suggestion came from Dana Dreibelbis, then an editor for W. B. Saunders publishing company. It was probably the third edition before I finally got the organization correct.

With each new edition, the text evolved into a personal statement reflecting my approach to the practice of pediatric neurology. If I were not actively engaged in practice, I could not continue writing the text. The text is a practical approach to diagnosis and management, and it expresses my own biases in situations in which a single standard of practice is not established.

Modern neuroimaging, which has greatly improved diagnosis, has also dampened the neurologist's skill in clinical diagnosis. A well-taken history and directed examination remain the starting points for diagnosis. More often than not, they are the only requirements for diagnosis. Invest most of your time in listening to the history. If you have no idea what is wrong after hearing the history, it is unlikely that diagnostic tests will enlighten you. A differential diagnosis is a list that contains one correct answer and several wrong answers. There is little to commend a list of wrong answers. Their exclusion is both time consuming and expensive. Better to think through the possibilities at the onset and zero in on the right answer.

In prior editions, I have recommended two web-based resources for genetic disorders: Online Mendelian Inheritance in Man (http://micro189.lib3.hawaii.edu:2200/omim) and GeneClinics (http://www.geneclinics.org). GeneReviews, now part of GeneClinics, contains a wealth of up-to-date information on neurological disorders. I have referenced it throughout the text and rarely go to the clinic without logging on.

The intent of this book is to provide a sensible approach to the common presenting problems of children with disorders of the nervous system. The general organization remains the same because the presenting clinical features are unchanged. Some chapters have required more revision than others, but the reader will find new information in all.

I wish to express my gratitude to Susan F. Pioli, Publishing Director, Elsevier Health Sciences, for her friendship and assistance in developing several textbooks.

Gerald M. Fenichel, MD
THE SUDDEN ONSET of neurological dysfunction characterizes paroxysmal disorders. In children, such events often clear completely. Disturbance of ion channels (channelopathies) is often the underlying cause. Examples of channelopathies are genetic epilepsies, migraine, periodic paralysis, and paroxysmal movement disorders.

Approach to Paroxysmal Disorders

The diagnosing physician almost never witnesses the paroxysmal event. The nature of the event requires surmising what transpired by listening to the eyewitness description offered by a family member or, worse, to the secondhand description that the parent heard from a teacher. Never accept a secondhand description. Most “spells” are not seizures, and epilepsy is not a diagnosis of exclusion. The most common confusion is between seizures and syncope. Most people stiffen and tremble at the end of a faint.

Spells seldom remain unexplained when viewed. Because observation of the spell is crucial to diagnosis, ask the family to videotape the spell. Most families either own or can borrow a video camera. Even when a purchase is required, it is more cost-effective than brain imaging studies, and the family has something useful to show for the expenditure. Always ask the following two questions: Has this ever happened before? Does anyone else in the family have similar episodes? Often, no one offers this important information until requested. Episodic symptoms that last only seconds and cause no abnormal signs usually remain unexplained and do not warrant laboratory investigation. The differential diagnosis of paroxysmal disorders is different in neonates, infants, children, and adolescents and presented best by age groups.
The terms used in the text to describe states of decreased consciousness are provided in Table 2–1. With the exception of coma, these definitions are not standard. They are more precise, however, and more useful than such terms as semicomatose and semistuporous. The term encephalopathy describes a diffuse disorder of the brain in which at least two of the following symptoms are present: (1) altered states of consciousness, (2) altered cognition or personality, and (3) seizures. Encephalitis is an encephalopathy accompanied by cerebrospinal fluid (CSF) pleocytosis.

**Table 2-1 -- States of Decreased Consciousness**

<table>
<thead>
<tr>
<th>Term</th>
<th>Definition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lethargy</td>
<td>Difficult to maintain the aroused state</td>
</tr>
<tr>
<td>Obtundation</td>
<td>Responsive to stimulation other than pain *</td>
</tr>
<tr>
<td>Stupor</td>
<td>Responsive only to pain *</td>
</tr>
<tr>
<td>Coma</td>
<td>Unresponsive to pain</td>
</tr>
</tbody>
</table>

* Responsive indicates cerebral alerting, not just reflex withdrawal.

Lack of responsiveness is not always lack of consciousness. Infants with botulism (see Chapter 6) may have such severe hypotonia and ptosis that they cannot move their limbs or eyelids in response to stimulation. They appear to be in a coma or stupor but are actually alert. The locked-in syndrome (a brainstem disorder in which the individual can process information but cannot respond) and catatonia are other examples of diminished responsiveness in the alert state.

Either increased or decreased neuronal excitability may characterize the progression from consciousness to
coma. Patients with increased neuronal excitability (the high road) become restless and then confused; next, tremor, hallucinations, and delirium (an agitated confusional state) develop. Myoclonic jerks may occur. Seizures herald the end of delirium, and stupor or coma follow. Table 2–2 summarizes the differential diagnosis of the high road to coma. Tumors and other mass lesions are not expected causes. Instead, metabolic, toxic, and inflammatory disorders are likely.

Table 2-2  -- Causes of Agitation and Confusion

<table>
<thead>
<tr>
<th>Category</th>
<th>Causes</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Epileptic</strong></td>
<td></td>
</tr>
<tr>
<td>Absence status (see Chapter 1)</td>
<td></td>
</tr>
<tr>
<td>Complex partial seizure (see Chapter 1)</td>
<td></td>
</tr>
<tr>
<td><strong>Infectious Disorders</strong></td>
<td></td>
</tr>
<tr>
<td>Bacterial infections</td>
<td></td>
</tr>
<tr>
<td>Cat-scratch disease</td>
<td></td>
</tr>
<tr>
<td>Meningitis (see Chapter 4)</td>
<td></td>
</tr>
<tr>
<td>Rickettsial infections</td>
<td></td>
</tr>
<tr>
<td>Lyme disease</td>
<td></td>
</tr>
<tr>
<td>Rocky Mountain spotted fever</td>
<td></td>
</tr>
<tr>
<td>Viral infections</td>
<td></td>
</tr>
<tr>
<td>Aseptic meningitis</td>
<td></td>
</tr>
<tr>
<td>Arboviruses</td>
<td></td>
</tr>
<tr>
<td>Herpes simplex encephalitis</td>
<td></td>
</tr>
<tr>
<td>Measles encephalitis</td>
<td></td>
</tr>
<tr>
<td>Postinfectious encephalomyelitis</td>
<td></td>
</tr>
<tr>
<td>Reye syndrome</td>
<td></td>
</tr>
<tr>
<td><strong>Metabolic and Systemic Disorders</strong></td>
<td></td>
</tr>
<tr>
<td>Disorders of osmolality</td>
<td></td>
</tr>
<tr>
<td>Hypoglycemia</td>
<td></td>
</tr>
<tr>
<td>Hyponatremia</td>
<td></td>
</tr>
<tr>
<td>Endocrine disorders</td>
<td></td>
</tr>
<tr>
<td>Adrenal insufficiency</td>
<td></td>
</tr>
<tr>
<td>Hypoparathyroidism</td>
<td></td>
</tr>
<tr>
<td>Thyroid disorders</td>
<td></td>
</tr>
<tr>
<td>Hepatic encephalopathy</td>
<td></td>
</tr>
<tr>
<td>Inborn errors of metabolism</td>
<td></td>
</tr>
<tr>
<td>Disorders of pyruvate metabolism (see Chapter 5)</td>
<td></td>
</tr>
<tr>
<td>Medium-chain acyl-CoA dehydrogenase deficiency</td>
<td></td>
</tr>
<tr>
<td>Respiratory chain disorders (see Chapters 5, 6, 8, 10)</td>
<td></td>
</tr>
<tr>
<td>Urea cycle disorder, heterozygote (see Chapter 1)</td>
<td></td>
</tr>
<tr>
<td><strong>Renal disease</strong></td>
<td></td>
</tr>
<tr>
<td>Hypertensive encephalopathy</td>
<td></td>
</tr>
<tr>
<td>Uremic encephalopathy</td>
<td></td>
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<tr>
<td><strong>Migraine</strong></td>
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</tbody>
</table>
Decreased neuronal excitability (the low road) lacks an agitated stage. Instead, awareness progressively deteriorates from lethargy to obtundation, stupor, and coma. The differential diagnosis is considerably larger than that with the high road and includes mass lesions and other causes of increased intracranial pressure. Table 2–3. Table 2–4 lists conditions that cause recurrent encephalopathies. A comparison of Tables 2–2 and 3 shows considerable overlap between conditions whose initial features are agitation and confusion and conditions that begin with lethargy and coma; the disorders responsible for each are described together to prevent repetition.

### Table 2-3 -- Causes of Lethargy and Coma

<table>
<thead>
<tr>
<th><strong>Epilepsy</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>Postictal state (see Chapter 1)</td>
</tr>
<tr>
<td>Status epilepticus (see Chapter 1)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th><strong>Hypoxia-Ischemia</strong></th>
</tr>
</thead>
<tbody>
<tr>
<td>Cardiac arrest</td>
</tr>
<tr>
<td>Cardiac arrhythmia</td>
</tr>
<tr>
<td>Congestive heart failure</td>
</tr>
<tr>
<td>Hypotension</td>
</tr>
<tr>
<td>Autonomic dysfunction</td>
</tr>
<tr>
<td>Dehydration</td>
</tr>
<tr>
<td>Hemorrhage</td>
</tr>
<tr>
<td>Pulmonary embolism</td>
</tr>
<tr>
<td>Near-drowning</td>
</tr>
<tr>
<td>Neonatal (see Chapter 1)</td>
</tr>
</tbody>
</table>
Increased Intracranial Pressure
Cerebral abscess (see Chapter 4)
Cerebral edema (see Chapter 4)
Cerebral tumor (see Chapters 4 and 10)
Herniation syndromes (see Chapter 4)
Hydrocephalus (see Chapters 4 and 18)
Intracranial hemorrhage
  Spontaneous (see Chapter 4)
  Traumatic

Infectious Disorders
Bacterial infections
  Cat-scratch disease
  Gram-negative sepsis
  Hemorrhagic shock and encephalopathy syndrome
Meningitis (see Chapter 4)
  Toxic shock syndrome
Rickettsial infections
  Lyme disease
  Rocky Mountain spotted fever
Viral infections
  Aseptic meningitis
  Arboviruses
  Herpes simplex encephalitis
  Measles encephalitis
  Postinfectious encephalomyelitis
  Reye syndrome
  Postimmunization encephalopathy

Metabolic and Systemic Disorders
Disorders of osmolality
  Diabetic ketoacidosis (hyperglycemia)
  Hypoglycemia
  Hypernatremia
  Hyponatremia
Endocrine disorders
  Adrenal insufficiency
  Hypoparathyroidism
  Thyroid disorders
Hepatic encephalopathy
Inborn errors of metabolism
Disorders of pyruvate metabolism (see Chapter 5)
Glycogen storage disorders (see Chapter 1)
Medium-chain acyl-CoA dehydrogenase (MCAD) deficiency
Respiratory chain disorders (see Chapter 5, 6, 8, 10)
Urea cycle disorder, heterozygote (see Chapter 1)

Renal disorders
Acute uremic encephalopathy
Chronic uremic encephalopathy
Dialysis encephalopathy
Hypertensive encephalopathy

Other metabolic disorders
Burn encephalopathy
Hypomagnesemia
Parenteral hyperalimentation
Vitamin B complex deficiency

Migraine Coma
Toxic
Immunosuppressive drugs
Prescription drugs
Substance abuse
Toxins

Trauma
Concussion
Contusion
Intracranial hemorrhage
Epidural hematoma
Subdural hematoma
Intracerebral hemorrhage
Neonatal (see Chapter 1)

Vascular
Hypertensive encephalopathy
Intracranial hemorrhage, nontraumatic (see Chapter 4)
Lupus erythematosus (see Chapter 11)
Neonatal idiopathic cerebral venous thrombosis (see Chapter 1)
Vasculitis (see Chapter 11)

Table 2-4  Causes of Recurrent Encephalopathy
Assume that any child with the acute behavioral changes of delirium (agitation, confusion, delusions, or hallucinations) has an organic encephalopathy until proved otherwise. The usual causes of delirium are toxic or metabolic disorders diffusely affecting both cerebral hemispheres. Schizophrenia should not be a consideration in a prepubertal child with acute delirium. Fixed beliefs, unalterable by reason, are delusions. The paranoid delusions of schizophrenia are logical to the patient and frequently part of an elaborate system of irrational thinking in which the patient feels menaced. Delusions associated with organic encephalopathy are less logical, are not systematized, and tend to be stereotyped.

A hallucination is the perception of sensory stimuli that are not present. Organic encephalopathies usually cause visual hallucinations, whereas psychiatric illness usually causes auditory hallucinations, especially if the voices are accusatory. Stereotyped auditory hallucinations that represent a recurring memory are an exception and suggest temporal lobe seizures.

History and Physical Examination

Delirious children, even with stable vital function, require rapid assessment because the potential for deterioration to a state of diminished consciousness is real. Obtain a careful history of the following: (1) the events leading to the behavioral change; (2) drug or toxic exposure (prescription drugs are more often at fault than substances of abuse, and a medicine cabinet inspection should be ordered in every home the child has visited); (3) a personal or family history of migraine or epilepsy; (4) recent or concurrent fever, infectious disease, or systemic illness; and (5) a previous personal or family history of encephalopathy.

Examination of the eyes, in addition to determining the presence or absence of papilledema, provides other etiological clues. Small or large pupils that respond poorly to light, nystagmus, or impaired eye movements suggest a drug or toxic exposure. Fixed deviation of the eyes in one lateral direction may indicate that (1) the encephalopathy has focal features, (2) seizures are a cause of the confusional state, or (3) seizures are part of the encephalopathy. The general and neurological examinations should specifically include a search for evidence of trauma, needle marks on the limbs, meningismus, and cardiac disease.
Approach to Headache

Most children referred to a neurologist for “headache” have already had a normal brain imaging study. The distinction between painful and harmful headaches occurs before consultation. Nevertheless, families seek medical attention for a child with headache not only hoping to relieve pain, but also seeking assurance that the child does not have a serious intracranial disease. Not every headache is explicable, and the term *psychogenic* is not a synonym for idiopathic.

In my practice, I see two headache patterns among most children. One is chronic low-grade headache, and the other is an intermittent disabling headache. The cause of the former is either caffeine or analgesic abuse, and the latter is migraine. Ten percent of children 5 to 15 years old have migraine. Children with migraine average twice as many days lost from school as children without migraine.

An initial headache classification proposed by the International Headache Society in 1988 has undergone several revisions with the newest published in 2004 (International Headache Society, 2004). As a rule, only poorly understood disorders require classification by a committee.

Sources of Pain

*Table 3–1* lists pain-sensitive structures of the head and neck. The main pain-sensitive structures inside the skull are blood vessels. Mechanisms that stimulate pain from blood vessels are vasodilation, inflammation, and traction-displacement. Increased intracranial pressure causes pain mainly by the traction and displacement of intracranial arteries (see Chapter 4). The brain parenchyma, its ependymal lining, and the meninges, other than the basal dura, are insensitive to pain.

*Table 3–1 -- Sources of Headache Pain*
Intracranial
- Cerebral and dural arteries
- Dura mater at base of brain
- Large veins and venous sinuses

Extracranial
- Cervical roots
- Cranial nerves
- Extracranial arteries
- Muscles attached to skull
- Periosteum/sinuses

Pain transmission from supratentorial intracranial vessels is by the trigeminal nerve, whereas pain transmission from infratentorial vessels is by the first three cervical nerves. The ophthalmic division of the trigeminal nerve innervates the arteries in the superficial portion of the dura and refers pain to the eye and forehead. The second and third divisions of the trigeminal nerve innervate the middle meningeal artery and refer pain to the temple. All three divisions of the trigeminal nerve innervate the cerebral arteries and refer pain to the eye, forehead, and temple. In contrast, referred pain from all structures in the posterior fossa is to the occiput and neck.

Several extracranial structures are pain sensitive. Major scalp arteries are present around the eye, forehead, and temple and produce pain when dilating or stretched. Cranial bones are insensitive, but periosteum, especially in the sinuses and near the teeth, is painful when inflamed. The inflamed periosteum is usually tender to palpation or other forms of physical stimulation. Muscles attached to the skull are a possible source of pain. The largest muscle groups are the neck extensors, which attach to the occipital ridge; the masseter muscles; and the frontalis muscle. The mechanism of muscle pain is incompletely understood but probably involves prolonged contraction. The extraocular muscles are a source of muscle contraction pain in patients with heterophoria. When an imbalance exists, especially in convergence, long periods of close work cause difficulty in maintaining conjugate gaze, and pain localizes to the orbit and forehead.

Pain from the cervical roots and cranial nerves is generally due to mechanical traction from injury or malformation. Pain follows this nerve distribution: the neck and back of the head up to the vertex for the cervical roots and the face for the cranial nerves.
Increased Intracranial Pressure is not a presenting complaint (Table 4–1). Most often, headache, vomiting, personality change, and alterations in states of consciousness bring it to medical attention. Less frequently, diplopia or the observation that one or both eyes are turning in is the initial complaint. The basis of referral for some children is the diagnosis of papilledema by another physician. Some conditions causing increased intracranial pressure are discussed elsewhere in this book (see Chapters 2, 3, 10, and 15). This chapter is restricted to conditions in which symptoms of increased intracranial pressure are initial and prominent features.

### Table 4-1 -- Features of Increased Intracranial Pressure

<table>
<thead>
<tr>
<th>In Infants</th>
<th>In Children</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bulging fontanelle</td>
<td>Diplopia (see Chapter 15)</td>
</tr>
<tr>
<td>Failure to thrive</td>
<td>Headache (see Chapter 3 )</td>
</tr>
<tr>
<td>Impaired upward gaze (setting sun sign)</td>
<td>Mental changes</td>
</tr>
<tr>
<td>Large head (see Chapter 18 )</td>
<td>Nausea and vomiting</td>
</tr>
<tr>
<td>Shri1l cry</td>
<td>Papilledema</td>
</tr>
</tbody>
</table>

**Pathophysiology**
Normal intracranial pressure in the resting state is approximately 10 mm Hg (136 mm H2O). Pressures greater than 20 mm Hg are abnormal. When the cranial bones fuse during childhood, the skull is a rigid box enveloping its content. Intracranial pressure is then the sum of the individual pressures exerted by the brain, blood, and cerebrospinal fluid (CSF). An increase in the volume of any one component requires an equivalent decrease in the size of one or both of the other compartments if intracranial pressure is to remain constant. Because the provision of oxygen and nutrients to the brain requires relatively constant cerebral blood flow, the major adaptive mechanisms available to relieve pressure are the compressibility of the brain and the rapid reabsorption of CSF by arachnoid villi. Infants and young children, in whom the cranial bones are still unfused, have the additional adaptive mechanism of spreading the cranial bones apart to increase cranial volume.

**Cerebrospinal Fluid**

The choroid plexus accounts for at least 70% of CSF production, and the transependymal movement of fluid from the brain to the ventricular system accounts for the remainder. The average volumes of CSF are 90 mL in children 4 to 13 years old and 150 mL in adults. The rate of formation is approximately 0.35 mL/min or 500 mL/day. Approximately 14% of total volume turns over every hour. The rate at which CSF forms remains relatively constant and declines only slightly as CSF pressure increases. In contrast, the rate of absorption increases linearly as CSF pressure exceeds 7 mm Hg. At a pressure of 20 mm Hg, the rate of absorption is three times the rate of formation.

Impaired absorption, not increased formation, is the usual mechanism of progressive hydrocephalus. Choroid plexus papilloma is the only pathological process in which formation sometimes can overwhelm absorption. When absorption is impaired, efforts to decrease the formation of CSF are not likely to have a significant effect on volume.
Chapter 5 – Psychomotor Retardation and Regression

Developmental Delay
Progressive Encephalopathies with Onset Before Age 2
Progressive Encephalopathies with Onset After Age 2
References

The differential diagnosis of psychomotor retardation (developmental delay) is quite different from that of psychomotor regression. Slow progress in the attainment of developmental milestones may be caused by either static (Table 5–1) or progressive (Table 5–2) encephalopathies. In contrast, the loss of developmental milestones previously attained usually indicates a progressive disease of the nervous system, but also may result from parental misperception of attained milestones or by the development of new clinical features from an established static disorder as the brain matures (Table 5–3).

Table 5-1 -- Diagnosis of Developmental Delay: No Regression

<table>
<thead>
<tr>
<th>Predominant Speech Delay</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bilateral hippocampal sclerosis</td>
</tr>
<tr>
<td>Congenital bilateral perisylvian syndrome (see Chapter 17)</td>
</tr>
<tr>
<td>Hearing impairment (see Chapter 17)</td>
</tr>
<tr>
<td>Infantile autism</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Predominant Motor Delay</th>
</tr>
</thead>
<tbody>
<tr>
<td>Ataxia (see Chapter 10)</td>
</tr>
<tr>
<td>Hemiplegia (see Chapter 11)</td>
</tr>
<tr>
<td>Hypotonia (see Chapter 6)</td>
</tr>
<tr>
<td>Neuromuscular disorders (see Chapter 7)</td>
</tr>
<tr>
<td>Paraplegia (see Chapter 12)</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Global Developmental Delay</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cerebral malformations</td>
</tr>
<tr>
<td>Chromosomal disturbances</td>
</tr>
<tr>
<td>Intrauterine infection</td>
</tr>
<tr>
<td>Perinatal disorders</td>
</tr>
<tr>
<td>Progressive encephalopathies (see Table 5–2)</td>
</tr>
</tbody>
</table>

Table 5-2 -- Progressive Encephalopathy: Onset Before Age 2
Acquired immunodeficiency syndrome encephalopathy
Disorders of amino acid metabolism
Homocystinuria (21q22)
Maple syrup urine disease
Intermediate form
Thiamine-responsive form
Phenylketonuria
Disorders of lysosomal enzymes
Ganglioside storage disorders
GM₁ gangliosidosis
GM₂ gangliosidosis (Tay-Sachs disease, Sandhoff disease)
Gaucher disease type II (glucosylceramide lipidosis)
Globoid cell leukodystrophy (Krabbe disease)
Glycoprotein degradation disorders
I-cell disease
Mucopolysaccharidoses
Type I (Hurler syndrome)
Type III (Sanfilippo disease)
Niemann Pick disease type A (sphingomyelin lipidosis)
Sulfatase deficiency disorders
Metachromatic leukodystrophy (sulfatide lipidoses)
Multiple sulfatase deficiency
Carbohydrate-deficient glycoprotein syndromes
Hypothyroidism
Mitochondrial disorders
Alexander disease
Mitochondrial myopathy, encephalopathy, lactic acidosis, and stroke (see Chapter 11)
Progressive infantile poliodystrophy (Alpers disease)
Subacute necrotizing encephalomyelopathy (Leigh disease)
Trichopoliodystrophy (Menkes syndrome)
Neurocutaneous syndromes
Chédiak-Higashi syndrome
Neurofibromatosis
Tuberous sclerosis
Other disorders of gray matter
Infantile ceroid lipofuscinosis (Santavuori-Haltia disease)
Infantile neuroaxonal dystrophy
Lesch-Nyhan disease
Progressive neuronal degeneration with liver disease
Rett syndrome
Other disorders of white matter

Aspartoacylase deficiency (Canavan disease)
Galactosemia: transferase deficiency
Neonatal adrenoleukodystrophy (see Chapter 6)
Pelizaeus-Merzbacher disease
Progressive hydrocephalus

<table>
<thead>
<tr>
<th>Table 5-3  -- Causes of Apparent Regression in Static Encephalopathy</th>
</tr>
</thead>
<tbody>
<tr>
<td>Increasing spasticity (usually during the first year)</td>
</tr>
<tr>
<td>New-onset movement disorders (usually during the second year)</td>
</tr>
<tr>
<td>New-onset seizures</td>
</tr>
<tr>
<td>Parental misperception of attained milestones</td>
</tr>
<tr>
<td>Progressive hydrocephalus</td>
</tr>
</tbody>
</table>

Developmental Delay

Delayed achievement of developmental milestones is a common problem evaluated by child neurologists. Two important questions require answers: (1) Is developmental delay restricted to specific areas, or is it global? (2) Is development delayed or regressing?

In infants, the second question is often difficult to answer. Even in static encephalopathies, new symptoms, such as involuntary movements and seizures, may occur as the child gets older, and delayed acquisition of milestones without other neurological deficits is sometimes the initial feature of progressive disorders. When it is clear that milestones previously achieved are lost or that focal neurological deficits are evolving, however, a progressive disease of the nervous system is a consideration.

The Denver Developmental Screening Test is an efficient and reliable method for assessing development in the physician’s office. It rapidly assesses four different components of development: personal-social, fine motor adaptive, language, and gross motor. Several psychometric tests amplify the results, but the Denver Developmental Screening Test in combination with neurological assessment provides sufficient information to initiate further diagnostic studies.

Language Delay

Normal infants and children have a remarkable facility for acquiring language during the first decade of life. Children exposed to two languages concurrently learn both. Vocalization of vowels occurs in the first month, and by 5 months, laughing and squealing are established. At 6 months, infants begin articulating consonants, usually M, D, and B. Parents translate these to mean “mama,” “dada,” and “bottle” or “baby,” although this is not the infant’s intention. These first attempts at vowels and consonants are automatic and sometimes occur even in deaf children. In the months that follow, the infant imitates many speech sounds, babbles and coos, and finally learns the specific use of “mama” and “dada” by 1 year of age. Receptive skills are always more highly developed than expressive skills because language must be decoded before it is encoded. By 2 years of age, children have learned to combine at least 2 words, understand more than 250 words, and follow many
Developmental disturbances in the language cortex of the left hemisphere that occur before 5 years of age displace language to the right hemisphere. This displacement does not occur in older children.

**Autistic Spectrum Disorders**

Infantile autism is a developmental disorder of brain function defined by behavioral characteristics. The terms *autistic spectrum disorders* (ASD) and *pervasive developmental disorders* classify the spectrum of behavioral consequences. *Asperger disorder* represents the high-functioning end of autistic spectrum disorders. The high concordance in monozygotic twins, a 4.5% increased risk for recurrence in siblings, and autistic behaviors in children with several genetic disorders support a hereditary basis.

Autism has become an increasingly popular diagnosis. An apparent increasing incidence of diagnosis suggests to some investigators an environmental factor. Data do not confirm the notion, however, of an autism epidemic or causation by any environmental factor. Most biological studies suggest prenatal factors.

**Clinical Features.**

Major diagnostic criteria are impaired sociability, impaired verbal and nonverbal communication skills, and restricted activities and interests (Rapin, 2002). Failure of language development is the feature most likely to bring autistic infants to medical attention and correlates best with the outcome; children who fail to develop language before age 5 have the worst outcome. The IQ is less than 70 in most children with autism. Some autistic children show no affection to their parents or other care providers, whereas others are affectionate on their own terms. Autistic children do not show normal play activity; some display a morbid preoccupation with spinning objects, stereotyped behaviors such as rocking and spinning, and relative insensitivity to pain. An increased incidence of epilepsy in autistic children is probable.

**Diagnosis.**

Infantile autism is a clinical diagnosis and not confirmable by laboratory tests. Infants with profound hearing impairment may display autistic behavior, and tests of hearing are diagnostic. An electroencephalogram (EEG) is indicated when seizures are suspected.

**Management.**

Autism is not curable, but several drugs may be useful to control specific behavioral disturbances. Behavior modification techniques improve some aspects of the severely aberrant behavior. Despite the best program of treatment, however, these children function in a moderately to severely retarded range, although some individuals have islands of normal or extraordinary ability (*idiot savant*).

**Bilateral Hippocampal Sclerosis**

Bilateral hippocampal sclerosis and the *congenital bilateral perisylvian syndrome* cause a profound impairment of language development. The former also causes failure of cognitive capacity that mimics infantile autism, whereas the latter causes a pseudobulbar palsy (see Chapter 17). Infants with medial bilateral hippocampal sclerosis generally come to medical attention for refractory seizures. The syndrome emphasizes, however, that the integrity of one medial hippocampal gyrus is imperative for language development.
Hearing Impairment

The major cause of isolated delay in speech development is hearing impairment (see Chapter 17). Hearing loss may occur concomitantly with global developmental retardation, as in rubella embryopathy, cytomegalic inclusion disease, neonatal meningitis, kernicterus, and several genetic disorders. Hearing loss need not be profound; it can be insidious, yet delay speech development. The loss of high-frequency tones, inherent in telephone conversation, prevents the clear distinction of many consonants that humans learn to fill in through experience; infants do not have experience in supplying missing sounds.

The hearing of any infant with isolated delay in speech development requires audiometric testing. Crude testing in the office by slamming objects and ringing bells is inadequate. Hearing loss is suspected in children with global retardation caused by disorders ordinarily associated with hearing loss or in retarded children who fail to imitate sounds. Other clues to hearing loss in children are excessive gesturing and staring at the lips of people who are talking.
Chapter 6 – The Hypotonic Infant

**Appearance of Hypotonia**

Clinicians test two kinds of tone: phasic and postural. Phasic tone is a rapid contraction in response to a high-intensity stretch. The tendon reflex response tests phasic tone. Striking the patellar tendon briefly stretches the quadriceps muscle. The spindle apparatus, sensing the stretch, sends an impulse through the sensory nerve to the spinal cord. This information is transmitted to the alpha motor neuron, and the quadriceps muscle contracts (monosynaptic reflex). Postural tone is the prolonged contraction of antigravity muscles in response to the low-intensity stretch of gravity. When postural tone is depressed, the trunk and limbs cannot maintain themselves against gravity, and the infant appears hypotonic.

The maintenance of normal tone requires intact central and peripheral nervous systems. Hypotonia is a common symptom of neurological dysfunction and occurs in diseases of the brain, spinal cord, nerves, and muscles ([Table 6–1](#)). One anterior horn cell and all the muscle fibers that it innervates compose a motor unit. The motor unit is the unit of force. Weakness is a symptom of all motor unit disorders. A primary disorder of the anterior horn cell body is a neuronopathy, a primary disorder of the axon or its myelin covering is a neuropathy, and a primary disorder of the muscle fiber is a myopathy. In infancy and childhood, cerebral disorders far outnumber motor unit disorders. The term cerebral hypotonia encompasses all causes of postural hypotonia caused by a cerebral disease or defect.

**Table 6–1 -- Differential Diagnosis of Infantile Hypotonia**

<table>
<thead>
<tr>
<th>Cerebral hypotonia</th>
</tr>
</thead>
<tbody>
<tr>
<td>Benign congenital hypotonia</td>
</tr>
<tr>
<td>Chromosome disorders</td>
</tr>
<tr>
<td>Prader-Willi syndrome</td>
</tr>
<tr>
<td>Trisomy</td>
</tr>
<tr>
<td>Chronic nonprogressive encephalopathy</td>
</tr>
<tr>
<td>Cerebral malformation</td>
</tr>
<tr>
<td>Perinatal distress</td>
</tr>
<tr>
<td>Postnatal disorders</td>
</tr>
<tr>
<td>Peroxisomal disorders</td>
</tr>
<tr>
<td>Cerebrohepatorenal syndrome (Zellweger syndrome)</td>
</tr>
</tbody>
</table>
Neonatal adrenoleukodystrophy
Other genetic defects
Familial dysautonomia
Oculocerebrorenal syndrome (Lowe syndrome)
Other metabolic defects
Acid maltase deficiency (see “Metabolic Myopathies”)
Infantile GM1 gangliosidosis (see Chapter 5)
Spinal cord disorders
Spinal muscular atrophies
Acute infantile
Autosomal dominant
Autosomal recessive
Cytochrome-c oxidase deficiency
X-linked
Chronic infantile
Autosomal dominant
Autosomal recessive
Congenital cervical spinal muscular atrophy
Infantile neuronal degeneration
Neurogenic arthrogryposis
Polyneuropathies
Congenital hypomyelinating neuropathy
Giant axonal neuropathy (see Chapter 7)
Hereditary motor-sensory neuropathies (see Chapter 7)
Disorders of neuromuscular transmission
Familial infantile myasthenia
Infantile botulism
Transitory myasthenia gravis
Fiber-type disproportion myopathies
Central core disease
Congenital fiber-type disproportion myopathy
Myotubular (centronuclear) myopathy
Acute
Chronic
Nemaline (rod) myopathy
Autosomal dominant
Autosomal recessive
Metabolic myopathies
Acid maltase deficiency
Cytochrome-c oxidase deficiency
Muscular dystrophies
Bethlem myopathy (see Chapter 7)
Congenital dystrophinopathy (see Chapter 7)
Congenital muscular dystrophy
Merosin deficiency primary
Merosin deficiency secondary
Merosin positive
Congenital myotonic dystrophy

**Appearance of Hypotonia**

When lying supine, all hypotonic infants look similar, regardless of the underlying cause or location of the abnormality within the nervous system. Spontaneous movement is lacking, full abduction of the legs places the lateral surface of the thighs against the examining table, and the arms lie either extended at the sides of the body or flexed at the elbow with the hands beside the head. Pectus excavatum is present when the infant has long-standing weakness in the chest wall muscles. Infants who lie motionless eventually develop flattening of the occiput and loss of hair on the portion of the scalp that is in constant contact with the crib sheet. When placed in a sitting posture, the head falls forward, the shoulders droop, and the limbs hang limply.

Newborns who are hypotonic in utero may be born with hip dislocation, multiple joint contractures (arthrogryposis), or both. Hip dislocation is a common feature of intrauterine hypotonia. The forceful contraction of muscles pulling the femoral head into the acetabulum is a requirement of normal hip joint formation. Arthrogryposis varies in severity from clubfoot, the most common manifestation, to symmetrical flexion deformities of all limb joints. Joint contractures are a nonspecific consequence of intrauterine immobilization. Among the several disorders that equally decrease fetal movement, however, some commonly produce arthrogryposis and others never do. Table 6–2 summarizes the differential diagnosis of arthrogryposis. As a rule, newborns with arthrogryposis who require respiratory assistance do not survive extubation unless the underlying disorder is myasthenia. The traction response, vertical suspension, and horizontal suspension further evaluate tone in infants who appear hypotonic at rest.

### Table 6-2 -- Differential Diagnosis of Arthrogryposis

<table>
<thead>
<tr>
<th>Cerebral malformations</th>
</tr>
</thead>
<tbody>
<tr>
<td>Cerebrohepatorenal syndrome</td>
</tr>
<tr>
<td>Chromosomal disorders</td>
</tr>
<tr>
<td>Fetal, non–nervous system causes</td>
</tr>
<tr>
<td>Motor unit disorders</td>
</tr>
<tr>
<td>Congenital benign spinal muscular atrophy</td>
</tr>
<tr>
<td>Congenital cervical spinal muscular atrophy</td>
</tr>
<tr>
<td>Congenital fiber-type disproportion myopathy</td>
</tr>
<tr>
<td>Congenital hypomyelinating neuropathy</td>
</tr>
<tr>
<td>Congenital muscular dystrophy</td>
</tr>
<tr>
<td>Genetic myasthenic syndromes</td>
</tr>
<tr>
<td>Infantile neuronal degeneration</td>
</tr>
<tr>
<td>Myotonic dystrophy</td>
</tr>
<tr>
<td>Neurogenic arthrogryposis</td>
</tr>
<tr>
<td>Phosphofructokinase deficiency</td>
</tr>
</tbody>
</table>
Traction Response

The traction response is the most sensitive measure of postural tone and is testable in premature newborns within an isolette. Grasping the hands and pulling the infant toward a sitting position initiates the response. A normal term infant lifts the head from the surface immediately with the body. When attaining the sitting position, the head is erect in the midline for a few seconds. During traction, the examiner should feel the infant pulling back against traction and observe flexion at the elbow, knee, and ankle. The traction response is not present in premature newborns of less than 33 weeks’ gestation. After 33 weeks, the neck flexors show increasing success in lifting the head. At term, only minimal head lag is present; after attaining the sitting posture, the head may continue to lag or may be erect briefly, then fall forward. The presence of more than minimal head lag and of failure to counter traction by flexion of the limbs in the term newborn is abnormal and indicates hypotonia.
Chapter 7 – Flaccid Limb Weakness in Childhood

Clinical Features of Neuromuscular Disease
- Progressive Proximal Weakness
- Progressive Distal Weakness
- Acute Generalized Weakness
- Periodic Paralysis

References

MOST CHILDREN WITH acute or chronic flaccid limb weakness have a disorder of the motor unit. Flaccid leg weakness may be the initial feature of disturbances in the lumbosacral region, but other symptoms of spinal cord dysfunction are usually present. Also, consult Table 12–1 when considering the differential diagnosis of flaccid leg weakness without arm impairment. Cerebral disorders may cause flaccid weakness, but dementia (see Chapter 5) or seizures (see Chapter 1) are usually a concomitant feature.

Clinical Features of Neuromuscular Disease

Weakness is decreased strength, as measured by the force of a maximal contraction. Fatigue is inability to maintain a less than maximal contraction, as measured by exercise tolerance. Weak muscles are always fatigued more easily than normal muscles, but fatigue may occur in the absence of weakness. Conditions in which strength is normal at rest, but fatigue or cramps occur with exercise are discussed in Chapter 8.

Initial Complaint

Limb weakness in children usually is noted first in the legs and then in the arms (Table 7–1). This is because many neuromuscular disorders affect the legs before the arms, and walking is impaired. Delayed development of motor skills is often an initial or prominent feature in the history of children with neuromuscular disorders. Marginal motor delay in children with otherwise normal development rarely raises concern and often is considered part of the spectrum of normal development. Prompts for neurological consultation in older children with neuromuscular disorders are failure to keep up with peers or easy fatigability.

Table 7-1 -- Symptoms of Neuromuscular Disease
An abnormal gait can be the initial symptom of either proximal or distal leg weakness. With proximal weakness, the pelvis fails to stabilize and waddles from side to side as the child walks. Running is especially difficult and accentuates the hip waddle. Descending stairs is particularly difficult in children with quadriceps weakness; the knee cannot lock and stiffen. Difficulty with ascending stairs suggests hip extensor weakness. Rising from the floor or a deep chair is difficult, and the hands help to push off.

Stumbling is an early complaint when there is distal leg weakness, especially weakness of the evictors and dorsiflexors of the foot. Falling is first noted when the child walks on uneven surfaces. The child is thought to be clumsy, but after a while parents realize that the child is “tripping on nothing at all.” Repeated ankle spraining occurs because of lateral instability. Children with footdrop tend to lift the knee high in the air so that the foot clears the ground. The weak foot then comes down with a slapping motion (steppage gait).

Toe walking is commonplace in Duchenne muscular dystrophy (DMD) because the pelvis thrusts forward to shift the center of gravity, and the gastrocnemius muscle is stronger than the peroneal muscles. Toe walking also occurs in upper motor neuron disorders that cause spasticity and in children who have tight heel cords but no identifiable neurological disease. Muscular dystrophy usually is associated with hyporeflexia, and spasticity is associated with hyperreflexia. The ankle tendon reflex may be difficult to elicit, however, when the tendon is tight for any reason.

Adolescents, but usually not children, with weakness complain of specific disabilities. A young woman with proximal weakness may have difficulty keeping her arms elevated to groom her hair or rotating the shoulder to get into and out of garments that have a zipper or hook in the back. Weakness of hand muscles often comes to attention because of difficulty with handwriting. Adolescents may notice difficulty in unscrewing jar tops or working with tools. Teachers report to parents when children are slower than classmates in climbing stairs, getting up from the floor, and skipping and jumping. Parents may report a specific complaint to the physician, but more often, they define the problem as inability to keep up with peers.

A child whose limbs are weak also may have weakness in the muscles of the head and neck. Specific questions should be asked about double vision, drooping eyelids, difficulty chewing and swallowing, change of facial expression and strength (whistling, sucking, chewing, blowing), and clarity and tone of speech. Weakness of neck muscles frequently is noticed when the child is a passenger in a car that suddenly accelerates or decelerates. The neck muscles are unable to stabilize the head, which snaps backward or forward.
Cramps, Muscle Stiffness, and Exercise Intolerance

**Abnormal Muscle Activity**
- Decreased Muscle Energy
- Myopathic Stiffness and Cramps

**References**

**A CRAMP IS** an involuntary painful contraction of a muscle or part of a muscle. Cramps can occur in normal children during and after vigorous exercise and after excessive loss of fluid or electrolytes. The characteristic electromyogram (EMG) finding for such cramps is the repetitive firing of normal motor unit potentials. Stretching the muscle relieves the cramp. Partially denervated muscle is particularly susceptible to cramping not only during exercise, but also during sleep. Night cramps may awaken patients with neuronopathies, neuropathies, or root compression. Cramps during exercise occur also in patients with several different disorders of muscle energy metabolism. The EMG characteristic of these cramps is electrical silence.

Muscle stiffness and spasms are not cramps, but are prolonged contractions of several muscles that are able to impose postures. Such contractions may or may not be painful. When painful, they lack the explosive character of cramps. Prolonged contractions occur when muscles fail to relax (myotonia) or when motor unit activity is continuous (Table 8–1). Prolonged, painless muscle contractions occur also in dystonia and in other movement disorders (see Chapter 14).

**Table 8-1 -- Diseases with Abnormal Muscle Activity**

<table>
<thead>
<tr>
<th>Continuous motor unit activity</th>
<th></th>
</tr>
</thead>
<tbody>
<tr>
<td>Neuromyotonia</td>
<td></td>
</tr>
<tr>
<td>Paroxysmal ataxia and myokymia (see Chapter 10)</td>
<td></td>
</tr>
<tr>
<td>Schwartz-Jampel syndrome</td>
<td></td>
</tr>
<tr>
<td>Stiff-man syndrome</td>
<td></td>
</tr>
<tr>
<td>Thyrotoxicosis</td>
<td></td>
</tr>
<tr>
<td>Cramps-fasciculation syndrome</td>
<td></td>
</tr>
<tr>
<td>Myotonia</td>
<td></td>
</tr>
<tr>
<td>Myotonia congenita</td>
<td></td>
</tr>
<tr>
<td>Myotonia fluctuans</td>
<td></td>
</tr>
<tr>
<td>Systemic disorders</td>
<td></td>
</tr>
<tr>
<td>Hypoadrenalism</td>
<td></td>
</tr>
<tr>
<td>Hypocalcemia (tetany)</td>
<td></td>
</tr>
<tr>
<td>Strychnine poisoning</td>
<td></td>
</tr>
</tbody>
</table>
Many normal children, especially preadolescent boys, complain of pain in their legs at night and sometimes during the day, especially after a period of increased activity. These pains are not true cramps. The muscle is not in spasm, the pain is diffuse and aching in quality, and the discomfort lasts for an hour or longer. Stretching the muscle does not relieve the pain. These pains are not a symptom of neuromuscular disease and are called growing pains, for want of better understanding. Mild analgesics or heat relieves symptoms.

Exercise intolerance is a relative term for an inability to maintain exercise at an expected level. The causes of exercise intolerance considered in this chapter are fatigue and muscle pain. Fatigue is a normal consequence of exercise and occurs in everyone at some level of activity. In general, weak children become fatigued more quickly than children who have normal strength. Many children with exercise intolerance and cramps, but no permanent weakness, have a defect in an enzyme needed to produce energy for muscular contraction (Table 8–2). A known mechanism underlies several such inborn errors of metabolism. Even when the full spectrum of biochemical tests is available, however, identification of a metabolic defect is not possible in some children with cramps during exercise.

### Table 8-2 -- Diseases with Decreased Muscle Energy

<table>
<thead>
<tr>
<th>Defects of carbohydrate utilization</th>
</tr>
</thead>
<tbody>
<tr>
<td>Lactate dehydrogenase deficiency</td>
</tr>
<tr>
<td>Myophosphorylase deficiency</td>
</tr>
<tr>
<td>Phosphofructokinase deficiency</td>
</tr>
<tr>
<td>Phosphoglycerate kinase deficiency</td>
</tr>
<tr>
<td>Phosphoglycerate mutase deficiency</td>
</tr>
<tr>
<td>Defects of fatty acid oxidation</td>
</tr>
<tr>
<td>Carnitine O-palmitoyltransferase 2 deficiency</td>
</tr>
<tr>
<td>Very-long-chain acyl coenzyme A dehydrogenase deficiency</td>
</tr>
<tr>
<td>Mitochondrial (respiratory chain) myopathies</td>
</tr>
<tr>
<td>Myoadenylate deaminase deficiency</td>
</tr>
</tbody>
</table>

Myasthenia gravis is a disorder characterized by exercise intolerance, but it is not covered in this chapter because the usual initial symptoms are either isolated cranial nerve disturbances (see Chapter 15) or limb weakness (see Chapters 6 and 7). Conditions that produce some combination of cramps and exercise intolerance are divisible into three groups: (1) diseases with abnormal muscle activity, (2) diseases with decreased energy for muscle contraction, and (3) myopathies. As a rule, the first and third groups are symptomatic at all times, whereas the second group is symptomatic only with exercise. The first group usually requires an EMG for diagnosis. EMG is the initial diagnostic test in children with muscle stiffness that is not due to spasticity or rigidity. It usually leads to the correct diagnosis (Table 8–3).

### Table 8-3 -- Electromyography in Muscle Stiffness
Normal Between Cramps

- Brody myopathy
- Defects of carbohydrate metabolism
- Defects of lipid metabolism
- Mitochondrial myopathies
- Myoadenylate deaminase deficiency
- Rippling muscle disease
- Tubular aggregates

Silent Cramps

- Brody disease
- Defects of carbohydrate metabolism
- Rippling muscle disease
- Tubular aggregates

Continuous Motor Activity

- Neuromyotonia
- Schwartz-Jampel syndrome
- Stiff-man syndrome

Myotonia

- Myotonia congenita
- Myotonic dystrophy
- Schwartz-Jampel syndrome

Myopathy

- Emery-Dreifuss muscular dystrophy
- Rigid spine syndrome
- X-linked myalgia

* Or may be myopathic.

Abnormal Muscle Activity

Continuous Motor Unit Activity

The cause of continuous motor unit activity (CMUA) is the uncontrolled release of acetylcholine packets at the neuromuscular junction. The EMG features of CMUA are repetitive muscle action potentials in response to a single nerve stimulus; high-frequency bursts of motor unit potentials of normal morphology abruptly start and stop. Rhythmic firing of doublets, triplets, and multiplets occur. During long bursts, the potentials decline in amplitude. This activity is difficult to distinguish from normal voluntary activity. CMUA occurs in a heterogeneous group of disorders characterized clinically by some combination of muscle pain, fasciculations, myokymia, contractures, and cramps (Table 8–4).

Table 8–4  -- Abnormal Muscle Activity
Fasciculations—Spontaneous, random twitching of a group of muscle fibers

Fibrillation—Spontaneous contraction of a single muscle fiber, not visible through the skin

Myotonia—Disturbance in muscle relaxation after voluntary contraction or percussion

Myokymia—Repetitive fasciculations causing a quivering or undulating twitch

Neuromyotonia—Continuous muscle activity characterized by muscle rippling, muscle stiffness, and myotonia

The primary defect in CMUA disorder may reside within the spinal cord (stiff-man syndrome) or the peripheral nerve (neuromyotonia). The original name for neuromyotonia is Isaac syndrome. These disorders may be sporadic or familial in occurrence. When familial, the usual mode of transmission is autosomal dominant inheritance.

**Neuromyotonia**

The primary abnormality in neuromyotonia is in the nerve or the nerve terminal. Most childhood cases are sporadic in occurrence, but some show a pattern of autosomal dominant inheritance. An autoimmune process directed against the potassium channel may account for some sporadic cases (Shillito et al, 1995).

**Clinical Features.**

The clinical triad includes involuntary muscle twitching (fasciculations or myokymia), muscle cramps or stiffness, and myotonia. Excessive sweating frequently is associated with the muscle stiffness. The age at onset is anytime from birth to adult life.

The initial features are muscle twitching and cramps brought on by exercise. Later these symptoms also occur at rest and during sleep. The cramps may affect only distal muscles, causing painful posturing of the hands and feet. As a rule, leg weakness is greater than arm weakness. These disorders are not progressive and do not lead to permanent disability. Attacks of cramping are less frequent and severe with age.

In some children, cramps and fasciculations are not as prominent as stiffness, which causes abnormal limb posturing associated with excessive sweating. Leg involvement is more common than arm involvement, and the symptoms suggest dystonia (see Chapter 14). Limb posturing may begin in one foot and remain asymmetrical for months. Most cases are sporadic. Muscle mass, muscle strength, and tendon reflexes are normal. Fasciculations are sporadic and seen only after prolonged observation.

**Diagnosis.**

Some adult-onset cases are associated with malignancy, but this is never the case in children. Muscle fibers fire repetitively at a rate of 100 to 300 Hz, either continuously or in recurring bursts, producing a pinging sound. The discharge continues during sleep and persists after procaine block of the nerve.

**Management.**

Carbamazepine and phenytoin, at usual anticonvulsant doses, are effective in reducing or abolishing symptoms.
Schwartz-Jampel Syndrome

The Schwartz-Jampel syndrome (SJS) is a hereditary disorder, transmitted by autosomal recessive inheritance. The gene locus maps to 1p36.1. Characteristic features include short stature, skeletal abnormalities, and persistent muscular contraction and hypertrophy. Giedion and colleagues (1997) found that some children had mild skeletal changes that may be secondary to CMUA (SJS-1), whereas others had primary bone dysplasia with CMUA (SJS-2). The first group shows linkage to chromosome 1p36.1-p34, whereas the second group does not.

Clinical Features.

SJS-1 corresponds to the original description of Schwartz and Jampel. Bone deformities are not prominent at birth. CMUA of the face is the main feature producing a characteristic triad that includes narrowing of the palpebral fissures (blepharophimosis), pursing of the mouth, and puckering of the chin. Striking or even blowing on the eyelids induces blepharospasm. CMUA in the limbs produces stiffness of gait and exercise intolerance. Motor development during the first year is slow, but intelligence is normal. SJS-2 has prominent bone deformities at birth that suggest the Morquio syndrome (osteochondrodystrophy). Neonatal mortality is high.

Diagnosis.

The EMG shows CMUA. Initial reports suggested incorrectly that the abnormal activity seen on the EMG and expressed clinically was myotonia. Myotonia may be present, but CMUA is responsible for the facial and limb symptoms. The serum concentration of creatine kinase (CK) can be mildly elevated. The histological appearance of the muscle is usually normal but may show variation in fiber size and an increased number of central nuclei.

Management.

Phenytoin or carbamazepine diminishes the muscle stiffness. Early treatment with relief of muscle stiffness reduces the severity of subsequent muscle deformity.
Chapter 9 – Sensory and Autonomic Disturbances

Sensory Symptoms
Painful Arm Syndromes
Central Congenital Insensitivity (Indifference) to Pain
Foramen Magnum Tumors
Hereditary Neuropathies
Spinal Disorders
Thalamic Pain
References

This chapter deals primarily with sensory disturbances of the limbs and trunk. Autonomic dysfunction often is associated with sensory loss but sometimes occurs alone. Chapter 17 discusses sensory disturbances of the face.

Sensory Symptoms

Pain, dysesthesias, and loss of sensibility are the important symptoms of disturbed sensation. Peripheral neuropathy is the most common cause of disturbed sensation at any age. As a rule, hereditary neuropathies are more likely to cause loss of sensibility without discomfort, whereas acquired neuropathies are more likely to be painful. Discomfort is more likely than numbness to bring a patient to medical attention.

Nerve root pain generally follows a dermatomal distribution. Ordinarily, it is described as deep and aching. The pain is more proximal than distal and may be constant or intermittent. When intermittent, the pain may radiate in a dermatomal distribution. The most common cause of root pain in adults is sciatica associated with lumbar disk disease. Disk disease also occurs in adolescents, usually because of trauma. In children, radiculitis is a more common cause of root pain. Examples of radiculitis are the migratory aching of a limb preceding paralysis in Guillain-Barré syndrome (see Chapter 7) and the radiating pain in a C5 distribution that heralds an idiopathic brachial neuritis (see Chapter 13).

Polyneuropathy involving small nerve fibers causes dyesthetic pain. This pain differs from previously experienced discomfort and is described as “pins and needles,” tingling, or burning. It compares with the abnormal sensation felt when dental anesthesia is wearing off. The discomfort is superficial, distal, and usually symmetrical. Dyesthetic pain is never a feature of hereditary neuropathies in children.

Loss of sensibility is the sole initial feature in children with sensory neuropathy. Because clumsiness is the initial feature, delay in establishing a correct diagnosis is common. Strength is normal, as are tests of cerebellar function. Tendon reflexes are absent. The combination of areflexia and clumsiness should suggest a sensory neuropathy. Table 9–1 summarizes the pattern of sensory loss as a guide to the anatomical site of abnormality.
<table>
<thead>
<tr>
<th>Pattern</th>
<th>Site</th>
</tr>
</thead>
<tbody>
<tr>
<td>All limbs</td>
<td>Spinal cord or peripheral nerve</td>
</tr>
<tr>
<td>Both legs</td>
<td>Spinal cord or peripheral nerve</td>
</tr>
<tr>
<td>Glove-and-stocking</td>
<td>Peripheral nerve</td>
</tr>
<tr>
<td>Legs and trunk</td>
<td>Spinal cord</td>
</tr>
<tr>
<td>One arm</td>
<td>Plexus</td>
</tr>
<tr>
<td>One leg</td>
<td>Plexus or spinal cord</td>
</tr>
<tr>
<td>Unilateral arm and leg</td>
<td>Brain or spinal cord</td>
</tr>
</tbody>
</table>
The term **ataxia** denotes disturbances in the fine control of posture and movement. The cerebellum and its major input systems from the frontal lobes and the posterior columns of the spinal cord provide this control. The initial and most prominent feature is usually an abnormal gait. The ataxic gait is wide-based, lurching, and staggering, and it provokes disquiet in an observer for fear that the patient is in danger of falling. One observes a similar gait in people who are attempting to walk in a vehicle that has several directions of motion at once, such as a railroad train.

When an abnormality occurs in the vermis of the cerebellum, the child cannot sit still but constantly moves the body to-and-fro and bobs the head (*titubation*). In contrast, disturbances of the cerebellar hemispheres cause a tendency to veer in the direction of the affected hemisphere, with dysmetria and hypotonia in the ipsilateral limbs. Bifrontal lobe disease may produce symptoms and signs that are indistinguishable from the symptoms and signs of cerebellar disease.

Loss of sensory input to the cerebellum, because of peripheral nerve or posterior column disease, necessitates constant looking at the feet to know their location in space. The gait also is wide-based, but is not so much lurching as careful. The foot raises high with each step and slaps down heavily on the ground. Station and gait are considerably worse with the eyes closed, and the patient may fall to the floor (*Romberg sign*). Sensory ataxia is more likely to cause difficulty with fine finger movements than with reaching for objects. Other features of cerebellar disease are a characteristic speech that varies in volume and has an increased separation of syllables (*scanning speech*), hypotonia, limb and ocular dysmetria, and tremor.

The differential diagnosis of a child with acute ataxia or recurrent attacks of ataxia (*Table 10–1*) is quite different from that of a child with chronic static or progressive ataxia (*Table 10–2*). These two presentations are discussed separately. One “suddenly” may become aware of what had been a slowly progressive ataxia, however, and children with recurrent ataxia may never recover to baseline function after each attack. Progressive ataxia superimposes on the acute attacks.

**Table 10–1**  -- Acute or Recurrent Ataxia
<table>
<thead>
<tr>
<th>Conversion reaction</th>
<th>Drug ingestion</th>
</tr>
</thead>
<tbody>
<tr>
<td>Encephalitis (brainstem)</td>
<td>Genetic disorders</td>
</tr>
<tr>
<td>Dominant recurrent ataxia</td>
<td>Episodic ataxia type 1</td>
</tr>
<tr>
<td>Episodic ataxia type 2</td>
<td>Hartnup disease</td>
</tr>
<tr>
<td>Maple syrup urine disease</td>
<td>Pyruvate dehydrogenase deficiency</td>
</tr>
<tr>
<td>Migraine</td>
<td>Basilar</td>
</tr>
<tr>
<td>Benign paroxysmal vertigo</td>
<td>Postinfectious/immune</td>
</tr>
<tr>
<td>Acute postinfectious cerebellitis</td>
<td>Miller Fisher syndrome</td>
</tr>
<tr>
<td>Multiple sclerosis</td>
<td>Myoclonic encephalopathy/neuroblastoma</td>
</tr>
<tr>
<td>Pseudoataxia (epileptic)</td>
<td>Trauma</td>
</tr>
<tr>
<td>Hematoma (see Chapter 2)</td>
<td>Postconcussion</td>
</tr>
<tr>
<td>Vertebrobasilar occlusion</td>
<td>Vascular disorders</td>
</tr>
<tr>
<td>Cerebellar hemorrhage</td>
<td>Kawasaki disease</td>
</tr>
</tbody>
</table>

**Table 10-2 -- Chronic or Progressive Ataxia**
### Brain tumors
- Cerebellar astrocytoma
- Cerebellar hemangioblastoma (von Hippel–Lindau disease)
- Ependymoma
- Medulloblastoma
- Supratentorial tumors (see Chapter 4)

### Congenital malformations
- Basilar impression
- Cerebellar aplasias
- Cerebellar hemisphere aplasia
- Dandy-Walker malformation (see Chapter 18)
- Vermal aplasia
- Chiari malformation
- Hereditary ataxias

### Autosomal dominant inheritance (see Table 10–3)
- Autosomal recessive inheritance
- Abetalipoproteinemia
- Ataxia-telangiectasia
- Ataxia without oculomotor apraxia
- Ataxia with episodic dystonia
- Friedreich ataxia
- Hartnup disease
- Juvenile GM₂ gangliosidosis
- Juvenile sulfatide lipidoses
- Maple syrup urine disease
- Marinesco-Sjögren syndrome
- Pyruvate dehydrogenase deficiency
- Ramsay Hunt syndrome
- Refsum disease (HSMN IV) (see Chapter 7)
- Respiratory chain disorders (see Chapter 8)

### X-linked inheritance
- Adrenoleukodystrophy (see Chapter 5)
- Leber optic neuropathy (see Chapter 16)

### Acute or Recurrent Ataxia

The two most common causes of ataxia among children who were previously healthy then suddenly have an ataxic gait are drug ingestion and acute postinfectious cerebellitis. Migraine, brainstem encephalitis, and an
underlying neuroblastoma are the next considerations. Recurrent ataxia is uncommon and usually is caused by hereditary disorders; migraine is the most common cause, and disorders of pyruvate metabolism are second.

**Brain Tumor**

Primary brain tumors ordinarily cause chronic progressive ataxia (see discussion later). Ataxia may be acute, however, if the brain tumor bleeds or causes hydrocephalus. In addition, early clumsiness may not become apparent until it becomes severe enough to cause an obvious gait disturbance. Brain imaging is a recommendation for most children with acute cerebellar ataxia.
Hemiplegic Cerebral Palsy
Acute Hemiplegia
Chronic Progressive Hemiplegia
References

The approach to children with hemiplegia must distinguish between acute hemiplegia, in which weakness develops within a few hours, and chronic progressive hemiplegia, in which weakness evolves over days, weeks, or months. The distinction between an acute and an insidious onset should be easy but can be problematic. In children with a slowly evolving hemiplegia, missing early weakness is possible until an obvious level of functional disability is attained; at this point, the hemiplegia seems new and acute.

An additional presentation of hemiplegia in infants who come to medical attention because of developmental delay is slowness in meeting motor milestones and early establishment of hand preference. Children should not establish a hand preference until the second year. They have a static structural problem from birth (hemiplegic cerebral palsy), but the clinical features are not apparent until the child is old enough to use the affected limbs.

Magnetic resonance imaging (MRI) is the diagnostic modality of choice for investigating all forms of hemiplegia. MRI is especially informative in showing migrational defects in hemiplegic cerebral palsy associated with seizures. Magnetic resonance angiography is sufficiently informative to obviate the need for arteriography in most children.

Hemiplegic Cerebral Palsy

The term hemiplegic cerebral palsy comprises several pathological entities that result in limb weakness on one side of the body. In premature infants, the most common cause is periventricular hemorrhagic infarction (see Chapter 4). In term infants, the underlying causes are often cerebral malformations, cerebral infarction, and intracerebral hemorrhage. Imaging studies of the brain are useful to provide the family with a definitive diagnosis; this often relieves guilt and prevents litigation against the obstetrician.

The usual concern that brings infants with hemiplegia from birth for a neurological evaluation is delayed crawling or walking. Abnormalities of the legs are the focus of attention. An associated but seldom recognized feature is that hand dominance was established during the first year; this is never normal. Unilateral facial weakness is never associated, probably because bilateral corticobulbar innervation of the lower face persists until birth. Epilepsy occurs in half of children with hemiplegic cerebral palsy. Infants with injury to the dominant hemisphere can develop normal speech in the nondominant hemisphere, but it is at the expense of visuoperceptual and spatial skills. Infants with hemiplegia and early-onset seizures are an exception; they show cognitive disturbances of verbal and nonverbal skills.
Congenital Malformations

Migrational defects constitute most congenital malformations causing infantile hemiplegia (Figure 11–1). The affected hemisphere is often small and may show a unilateral perisylvian syndrome in which the sylvian fossa is widened (Sébire et al., 1996). Chapter 17 describes a bilateral perisylvian syndrome with speech disturbances. Seizures and mental retardation are often associated. As a rule, epilepsy is more common when congenital malformations cause infantile hemiplegia than when the cause is stroke.

Figure 11-1  Schizencephaly. The rim of gray matter around the defect in the left hemisphere indicates that the abnormality is a schizencephaly rather than an infarction. The child’s main abnormality was hemiplegia.
Chapter 12 – Paraplegia and Quadriplegia

Approach to Paraplegia

In this text, the term paraplegia denotes partial or complete weakness of both legs, and the term quadriplegia denotes partial or complete weakness of all limbs, obviating need for the terms paraparesis and quadriparesis. Many conditions described fully in this chapter are abnormalities of the spinal cord. The same spinal abnormality can cause paraplegia or quadriplegia, depending on the location of the injury, so these conditions are discussed together in this chapter.

Approach to Paraplegia

Weakness of both legs, without any involvement of the arms, suggests an abnormality of either the spinal cord or the peripheral nerves. Ordinarily a pattern of distal weakness and sensory loss, muscle atrophy, and absent tendon reflexes provides recognition of peripheral neuropathies (see Chapters 7 and 9). In contrast, spinal paraplegia causes spasticity, exaggerated tendon reflexes, and a dermatomal level of sensory loss. Disturbances in the conus medullaris and cauda equina, especially congenital malformation, may produce a complex of signs in which spinal cord or peripheral nerve localization is difficult; both may be involved. Spinal paraplegia may be asymmetrical at first, then the initial feature is monoplegia (see Chapter 13). When anatomical localization between the spinal cord and peripheral nerves is difficult, electromyogram and nerve conduction studies are useful in making the distinction.

Cerebral abnormalities sometimes cause paraplegia. In such a case, the child’s arms and legs are usually weak. Leg weakness is so much greater than arm weakness, however, that paraplegia is the chief complaint. The brain and the spinal cord may be abnormal, and the abnormalities can be in continuity (syringomyelia) or separated (Chiari malformation and myelomeningocele).
Chapter 13 – Monoplegia

**Approach to Monoplegia**

Either pain or weakness may cause refusal to use a limb. The cause of most painful limbs is injuries. Other causes are arthritis, infection, and tumor. A trivial pull on an infant’s arm may dislocate the radial head and cause an apparent monoplegia. Pain and weakness together is a feature of plexitis.

*Table 13-1* summarizes the differential diagnosis of acute monoplegia. Plexopathies and neuropathies are the leading causes of pure monoplegia. Stroke often affects one limb more than others, usually the arm more than the leg. The presentation may suggest monoplegia, but careful examination often reveals increased tendon reflexes and an extensor plantar response in the seemingly unaffected leg. Any suggestion of hemiplegia rather than monoplegia or increased tendon reflexes in the paretic limb should focus attention on the brain and cervical cord as the pathological site.

**Table 13-1**  -- Differential Diagnosis of Acute Monoplegia
Complicated migraine (see Chapter 11)
Dislocation of the radial head
Hemiparetic seizures (see Chapter 11)
Monomelic spinal muscular atrophy
Plexopathy and neuropathy
  Acute neuritis
  Asthmatic plexitis
  Idiopathic plexitis
  Osteomyelitis plexitis
  Poliomyelitis (see Chapter 7)
  Tetanus toxoid plexitis
Hereditary
  Hereditary brachial neuritis
  Hereditary neuropathy with liability to pressure palsy
Injury
  Lacerations
  Pressure injuries
  Traction injuries
Stroke (see Table 11–2)

Chronic progressive brachial monoplegia is uncommon. When it occurs, one should suspect syringomyelia and tumors of the cervical cord or brachial plexus. Chronic progressive weakness of one leg suggests a tumor of the spinal cord or a neurofibroma of the lumbar plexus. A monomelic form of spinal muscular atrophy, affecting only one leg or one arm, should be considered when progressive weakness is unaccompanied by sensory loss.
IN Voluntary MOVEMENTS ARE usually associated with abnormalities of the basal ganglia and their connections, and they occur in several different neurological disorders. Abnormal movements can be the main or initial features of disease, or they can occur as a late manifestation. This chapter discusses the former type.

Approach to the Patient

Movement disorders are not describable; they require visualization. If abnormal movements are not present at the time of examination, instruct the parents to videotape the movements at home. Some relatively common movements are recognizable by description, but the rich variety of abnormal movements and postures that may occur defies classification. The most experienced observer, at times, mistakes one movement for another or has difficulty conceptualizing the nature of an abnormal movement.

Many abnormal movements are paroxysmal or at least intermittent. Movement, startle, emotional upset, or sleep induces some movements. The physician should ask what makes the movement worse and if it is action induced. Ask children to perform the action during the examination. Paroxysmal movements raise the question of epilepsy. The concurrent presence of seizures and involuntary movements characterizes many neurological disorders of childhood. Nocturnal paroxysmal dystonia, previously thought to be a movement disorder, is actually a frontal lobe seizure (see Chapter 1). The clinical and conceptual distinction between spinal myoclonus and spinal seizures remains a gray area. The following guidelines are useful to distinguish involuntary movements from seizures:

1. Involuntary movements, with the exception of spinal myoclonus and periodic movements of sleep, abate or disappear during sleep; seizures persist or may worsen.
2. Involuntary movements usually have a more stereotyped appearance and, with the exception of acute drug reactions, are more persistent than seizures.
3. Seizures are often characterized by loss of consciousness or awareness; involuntary movements are not.
4. Seizures are usually accompanied by epileptiform activity on electroencephalogram (EEG); involuntary movements are not.
Involuntary contractions of a muscle that do not move a joint may be fasciculations, focal seizures, or myoclonus. Low-amplitude jerking movements that move a joint or muscles may be focal seizures, chorea, myoclonus, tics, or hemifacial spasm. High-amplitude jerking movements that move a limb or limbs may be seizures, ballismus, or myoclonus. Slow, writhing movements and abnormal posturing may be due to athetosis, dystonia, continuous motor unit activity (see Chapter 8), or seizures. The possible causes of rhythmic movements may be tremor, seizures, or myoclonus.
Chapter 15 – Disorders of Ocular Motility

Nonparalytic Strabismus
Ophthalmoplegia
Nystagmus
Acquired Nystagmus
References

The maintenance of binocular vision by conjugate movement of the eyes is perhaps the most delicate feat of muscular coordination achieved by the nervous system. Disorders of the visual sensory system, ocular muscles, ocular motor nerves, neuromuscular transmission, or gaze centers of the central nervous system may disturb ocular motility. This chapter discusses nonparalytic strabismus, paralytic strabismus (ophthalmoplegia), gaze palsies, ptosis, and nystagmus. Visual and pupillary disorders are discussed in Chapter 16.

Nonparalytic Strabismus

Strabismus (squint), or abnormal ocular alignment, affects 3% to 4% of preschool children. Many individuals have a latent tendency for ocular misalignment, heterophoria, which becomes apparent only under stress or fatigue. During periods of misalignment, a child may have diplopia or headache. Constant ocular misalignment is heterotropia. Children with heterotropia suppress the image from one eye to avoid diplopia. If only one eye fixates continuously, visual acuity may be lost permanently in the other (developmental amblyopia).

In nonparalytic strabismus, the amount of deviation in different directions of gaze is relatively constant (comitant). Each eye moves through a normal range when tested separately (ductions), but the eyes are disconjugate when used together (versions). Many children with chronic brain damage syndromes, such as malformations or perinatal asphyxia, have faulty fusion or faulty control of conjugate gaze mechanisms (nonparalytic strabismus). In neurologically normal children, the most common cause of nonparalytic strabismus is either a genetic influence or an intraocular disorder. Ocular alignment in the newborn is usually poor, with transitory shifts of alignment from convergence to divergence. Constant ocular alignment usually begins after 3 months of age. Approximately 2% of newborns exhibit tonic downward deviation of the eyes during the waking state. The eyes assume a normal position during sleep and are able to move upward reflexively.

Esotropia

Esotropia is an inward deviation (convergence) of the eyes. Early-onset or infantile esotropia is present before 1 year of age. The observation of accommodative esotropia is usually between 2 and 3 years of age and may be undetected until adolescence.

Clinical Features.
Children with infantile esotropia often alternate fixation between eyes and may cross-fixate, that is, look to the left with the right eye and to the right with the left eye. The misalignment is sufficient that family members see that a problem exists. Some children fixate almost entirely with one eye and are at risk for permanent loss of visual acuity (developmental amblyopia) in the other eye.

**Accommodative esotropia** occurs when accommodation compensates for hyperopia. Accommodation more sharply focuses the blurred image. Because convergence accompanies accommodation, one eye turns inward. Some children with accommodative esotropia cross-fixate and use each eye alternatively, while the other maintains fixation. If one eye is more hyperopic than the other eye, however, only the better eye fixates, and the unused eye has a considerable potential for amblyopia.

**Diagnosis.**

An ophthalmologist should examine the eyes to determine whether hyperopia is present.

**Management.**

Eyeglasses correct hyperopic errors. The treatment of early-onset esotropia, in which only one eye fixates, consists of alternate eye patching. Early corrective surgery is required for persistent esotropia.
Chapter 16 – Disorders of the Visual System

Assessment of Visual Acuity
Congenital Blindness
Acute Monocular or Binocular Blindness
Progressive Loss of Vision
Disorders of the Pupil
References

Congenital and acquired visual impairments in children are often associated with neurological disorders. The most common visual disorders are uncorrected refractive errors, amblyopia, strabismus, cataracts, and genetic disorders. Two conditions that are increasing in frequency are retinopathy of prematurity and retinal hemorrhage caused by child abuse.

Assessment of Visual Acuity

The assessment of visual acuity in preverbal children relies mainly on assessing fixation and following and on observing the way an infant or young child interacts with the environment.

Clinical Assessment

The pupillary light reflex is an excellent test of the functional integrity of the subcortical afferent and efferent pathways and is reliably present after 31 weeks’ gestation. A blink response to light develops at about the same time, and the lid may remain closed for as long as light is present (the dazzle reflex). The blink response to threat may not be present until 5 months of age. These responses are integrated in the brainstem and do not provide information on the cognitive (cortical) aspects of vision.

Observing fixation and following behavior is the principal means to assess visual function in newborns and infants. The human face, at a distance of approximately 30 cm, is the best target for fixation. By 9 weeks of age, 90% of infants fixate on faces. After obtaining fixation, the examiner slowly moves from side to side to test the following response. Visually directed grasping is present in normal children by 3 months of age but is difficult to test before 6 months. Absence of visually directed grasping may indicate a motor rather than a visual disturbance.

The refixation reflex evaluates the visual fields in infants and young children by moving an interesting stimulus in the peripheral field. Clues to visual impairment are structural abnormalities (e.g., microphthalmia, cloudy cornea), an absent or asymmetrical pupillary response to light, dysconjugate gaze, nystagmus, and failure to fixate or follow. Staring at a bright light source and oculodigital stimulation indicate severe visual impairment.
Chapter 17 – Lower Brainstem and Cranial Nerve Dysfunction

Facial Weakness and Dysphagia
Hearing Impairment and Deafness
Vertigo
References

This chapter addresses disorders causing dysfunction of cranial nerves VII through XII. Many such disorders also disturb ocular motility (see Chapter 15). The basis for chapter assignment of a disorder is by the most usual initial clinical feature. For example, the discussion of myasthenia gravis is in Chapter 15 because diplopia is a more common initial complaint than dysphagia.

An acute isolated cranial neuropathy, such as facial palsy, is usually a less ominous sign than multiple cranial neuropathies and is likely to have a self-limited course. An isolated cranial neuropathy may be the first sign of progressive cranial nerve dysfunction, however. Conditions causing isolated and multiple cranial neuropathies are discussed together because they may not be separable at onset.

Facial Weakness and Dysphagia

Anatomical Considerations

Facial Movement

The motor nucleus of the facial nerve is a column of cells in the ventrolateral tegmentum of the pons. Nerve fibers leaving the nucleus take a circuitous path in the brainstem before emerging close to the pontomedullary junction. The fibers then enter the internal auditory meatus with the acoustic nerve. Fibers for voluntary and reflexive facial movements separate rostral to the lower pons. After bending forward and downward around the inner ear, the facial nerve traverses the temporal bone in the facial canal and exits the skull at the stylomastoid foramen. Extracranially the facial nerve passes into the parotid gland, where it divides into several branches, which innervate all muscles of facial expression except the levator palpebrae superioris.

Sucking and Swallowing

The sucking reflex requires the integrity of the trigeminal, facial, and hypoglossal nerves. Stimulation of the lips produces coordinated movements of the face, jaw, and tongue. The automatic aspect of the reflex disappears after infancy but returns in bilateral disease of the cerebral hemispheres.

Fibers of the trigeminal and glossopharyngeal nerves ending in the nucleus solitarius form the afferent arc of the swallowing reflex. The motor roots of the trigeminal nerve, the glossopharyngeal and vagus fibers from the nucleus ambiguus, and the hypoglossal nerves form the efferent arc. A swallowing center that coordinates the reflex is located in the lower pons and upper medulla. A bolus of food stimulates the pharyngeal wall or back
of the tongue, and the combined actions of the tongue, palatine arches, soft palate, and pharynx move the food into the esophagus.
The volumes of the three compartments that fill the skull (brain, cerebrospinal fluid, and blood) determine the size of the skull during infancy. Expansion of one compartment comes at the expense of another. In this way, intracranial volume and pressure remain constant (see Chapter 4). The extracerebral spaces (epidural, subdural, and subarachnoid) may expand with blood and significantly affect cranial volume. Less important factors contributing to head size are the thickness of the skull bones and the rate of their fusion.

The skull's content determines its shape, as do external forces acting on the skull. Even more important is the rate at which individual skull bones fuse. The more recent move to have infants sleep supine instead of prone is leading to a generation of children with flat occiputs.

Measuring Head Size

Head circumference is determined by measuring the greatest occipitofrontal circumference. Influencing the accuracy of the measurement is fluid in and beneath the scalp and head shape. After a prolonged and difficult delivery, edema or blood may thicken the scalp and a cephalhematoma may be present as well. Fluid that infiltrates from a scalp infusion can increase head circumference markedly.

A round head has a larger intracranial volume than an oval head of equal circumference. A head with a relatively large occipitofrontal diameter has a larger volume than a head with a relatively large biparietal diameter. Head circumference measurements are most informative when plotted over time. The head sizes of male and female infants are different, and one should not rely on head growth charts that provide median values for both sexes. The rate of head growth in premature infants is considerably faster than in full-term newborns (Figure 18–1). For this reason, the charting of head circumference is always by conceptional age and not by postnatal age.

![Figure 18-1](Image)

Normal growth of head circumference in boys. The rate of growth in premature infants is greater than in full-term infants.