

## CHAPTER 13

### Neurologic Disorders of Childhood

Neurologic evaluation of children is important because 25% of hospitalized children have some type of neurologic disease. The neurologic evaluation of a child begins with a complete and accurate history from the parents or guardian of the child. Hospital birth records and perinatal information are essential. Developmental events should be compared with standard tables (e.g., Denver Developmental Screening Test). In older children records of performance in school or day-care centers can provide information concerning intellectual and emotional characteristics of the child. Other prior medical problems or a family history of neurologic disease can give a clue to the ongoing neurologic process.

To understand the disease processes and the pathogenesis of pediatric neurologic diseases, the reader should be familiar with normal development of the brain as is described in embryology textbooks. The cerebrum that begins as a smooth round structure evolves into a complex convoluted bihemispheric structure through a well-organized and predictable pattern. At the same time the neurons proliferate and differentiate and are organized through proliferation and enlargement of synaptic connections. Growth of the brain continues after birth, and, although myelination occurs mainly in the first year of life and is almost completed by the second year, it continues until the third decade of life.

The neurologic examination of children varies according to patient age. In infants, observation of the patient on the lap of a parent or guardian is least threatening and reveals the patient's spontaneous activities. The ability to grasp objects from the floor or from above the head and transfer them from hand to hand demonstrates motor function and coordination. Using small objects for this test facilitates evaluation of vision. Hearing can be tested by snapping fingers, shaking keys, or creating a loud noise to startle the infant. Head control can be evaluated by simple observation or by pulling the patient to a sitting position by his or her hands to reveal head lag. Sensation can be tested by pinching or tickling the skin surfaces of extremities and trunk. In addition to testing the stretch reflexes, one should test infants with the following maneuvers to obtain certain postures and movements that can indicate whether there are abnormalities of motor function: The *Moro reflex* is elicited by making a loud noise, sudden withdrawal of head support, and slapping the bed or examining table. The resulting startle response consists of an elevation, abduction, and claspings of the arms. This reflex disappears at 4 or 5 months of age. The *tonic neck reflex* is elicited by passively turning the head to either side. The response consists of extension of the limbs on the side the head is rotated toward and flexion of the limbs on the opposite side. This reflex should disappear after 6 months, and a normal child usually breaks the response in 15 to 30 seconds. The *parachute response* is elicited by holding the infant at the trunk and then dropping the infant forward with the head downward. The normal 7-month-old infant will thrust the arms forward for support or protection. The *Landau maneuver* is elicited by suspending the infant in a prone horizontal position. An infant who is older than 6 months will respond by extending the neck and trunk. The presence or absence of these reflexes indicate the degree of maturation of the nervous system and will give an indication of focal deficits when the response is a symmetrical or of more diffuse neurologic deficit in a symmetrically absent response. No neurologic examination is complete without measuring the patient's head circumference to detect microcephaly or macrocephaly and visualizing eye

grounds to look for cherry-red spots and splinter hemorrhages. Hepatosplenomegaly can indicate some type of storage disease. The following questions about a child with neurologic disease need to be answered:

- Is the disease process acute or chronic?
- Is the disease static or progressive ?
- If it is static, when did it happen?
- Is it a primary nervous system process or part of a systemic disease ?
- If it is a primary disease process, is it confined to the central nervous system, the peripheral nervous system, or both ?
- If it is a CNS process, does it affect cortex, white matter, or both ?

These questions may not have an answer in every case, but asking the questions helps the examiner arrive at a diagnosis.

## DISORDERS OF MOTOR EXECUTION

Children who have disorders of motor execution are clumsy and show poor performance of sequential motor acts but with preservation of motility and no alterations of muscle strength, tone, or posture. These motor deficits have been called, incorrectly in the past, "soft neurologic signs." In most of these children there are associated learning disorders and hyperactive behavior. The presence of these disorders can cause long-term educational and social disadvantages. The physician needs also to remember that in a few cases these findings can be the early manifestations of more serious and progressive neurologic disorders. *Dyspraxia* is the inability to perform sequential motor tasks in the presence of good motor strength, sensation, and cooperation. In children with dyspraxia, motor milestones have usually been normal, but the children have difficulty buttoning, zippering, and tying shoes and using combs, toothbrushes, and scissors. The best single office test that is most likely abnormal for manual dyspraxia is the finger-to-nose test. When the examiner asks the child to touch the tip of his nose with his index finger, the child is able to touch his nose with his finger, but then he may use the other hand's finger to touch the examiner's finger. Although this test is very reliable, a battery of tests are needed to make a definite diagnosis. Failure of other imitation tasks can also help in the diagnosis. In dyspraxia of speech buccal-facial gesturing is not imitated by the patient and can affect speech (dilated speech). The cause of the pure dyspraxia is unknown, but dyspraxia can be part of a more severe neurologic disorder. Clumsiness is a disorder of speed and dexterity of movements in children who have normal strength and muscle tone. In the preschool years parents of clumsy children can think that their children are lazy, slow, or uncooperative. In the early school grades in which the demands are more motor than intellectual, the children usually do poorly. This disorder frequently produces low self-esteem and, later, depression in some of the children. Teachers need to know about the diagnosis to be able to understand, tolerate, and help these children. *Adventitious movements* appear early in childhood and are frequently associated with clumsiness. There are mainly three types of adventitious movements: choreas, tremors, and synkinesis. *Choreas* are more easily detected when the children stand with the legs slightly apart, the arms pronated and extended, and the wrists dorsiflexed. Once in this position, rapid asymmetrical movements of the fingers can appear, and in severe cases there are associated facial and arm movements. The acute onset of chorea in school-age children may alert the physician to a rheumatic or collagen vascular disease etiology. In severely retarded children on neuroleptics, abnormal movements suggest drug induced dyskinesia. *Tremor* is a rhythmic distal involuntary oscillatory movement that can appear at rest or be aggravated by movements

(intention tremor). A cerebellar lesion needs to be excluded. If the tremor is severe, medication should be used. *Synkinesis* is an involuntary movement that occurs during voluntary action such as opening of the mouth when the child opens his eyes and the other way around or the presence of mirror image movements. Mirror movements are more frequently seen in association with agenesis of the corpus callosum. No treatment is available for this condition.

## CEREBRAL PALSY

Cerebral palsy is a clinical description of a heterogeneous group of disorders that affect control of movement or posture (motor dysfunction). The symptoms appear early in life, can change in the first two years (hypotonia can turn into spasticity or dyskinesias), are of a fixed nature, and are caused by a nonprogressive brain lesion or dysfunction. In 55% to 60% of the cases, the cause cannot be established. *Cerebral palsy does not mean mental retardation and is not always an outcome of difficult labor.* In spite of spasticity, Babinski signs are not constant features. Associated features include seizures, language disorders, dystonia, ataxia, speech impairments, and occasionally mental retardation. The classification of cerebral palsy is based on clinical features and includes the following:

- *Spastic diplegia.* Bilateral spasticity that is worse in the legs, with scissors posturing and gait. It is commonly associated with prematurity. Associated features can include seizures and a variable degree of mental impairment.
- *Spastic quadriplegia.* Severe bilateral spasticity that is worse in the legs, with few voluntary movements, pseudobulbar palsy, frequent aspiration, and commonly severe intellectual impairment. Seizures and optic atrophy can be seen.
- *Spastic hemiplegia.* Unilateral spastic hemiparesis is present at birth but is not usually recognized until sometime in the first 2 years of life. The paresis can be associated with athetotic posturing or other abnormal movements. Although seizures are frequent, mental impairment is less severe.
- *Spastic double hemiplegia.* Bilateral spasticity that is worse in the arms. Mental retardation and seizures are frequent.
- *Extrapyramidal cerebral palsy.* Diffuse hypotonia occurs at birth and changes into progressive choreoathetosis in the first 2 years of life. Dysarthric speech is frequent and can be associated with drooling as a result of impaired swallowing. Seizures and mental impairment are rare.
- *Mixed forms of cerebral palsy.* These are manifested by combinations of spasticity with abnormal movements.

Treatment of cerebral palsy needs to be individualized for every patient according to the patient's needs. The treatment should be directed toward early and intensive physical therapy, bracing when indicated, and, in selected patients, surgical intervention for release of contractures or elongation of Achilles tendons. For most CP patients learning some way of communication is more important than ambulation. Muscle relaxants including diazepam, dantrolene sodium, and baclofen can help temporarily in some of the spastic types.

## MENTAL RETARDATION

Intelligence has been defined as the capacity to obtain new knowledge and solve problems. It consists of a series of special and natural abilities: to have curiosity, observe, comprehend, think, and remember and use these qualities in a rational way during social interactions. Intelligence

can be modified by schooling and training but is predominantly innate. Intelligence has been measured by different tests designed to measure special abilities and to predict scholastic achievement. Mental retardation is an "incomplete or insufficient general development of mental capacities" as defined by the World Health Organization. Adams and Victor differentiate two groups of mental retardation: the *severely impaired* (IQ less than 20), or pathologic mentally retarded and the *less severely impaired* (IQ is 45 to 70), or subcultural, physiologic, or familial mentally retarded. The severely impaired patients often have neurologic impairment and are more frequently males; most need to be institutionalized. The less severely impaired show no neurologic impairment, are frequently females, and do not require institutionalization; the parents and siblings are often subnormal. The brains of the less severely impaired look normal with conventional histologic techniques, but a sparsity of dendritic arborizations and abnormal dendritic spines have been found with Golgi-Cox preparations, suggesting a synaptic transmission problem. Management should be directed toward the planning of the patient's special education and training to achieve independent living when possible.

## **AUTISM**

*Autism* is a heterogeneous syndrome characterized by development of excellent motor skills and retentive memory but failure to use language for communication (lack of speech, abnormal speech content, or prosody), profound withdrawal from contact with people, and an obsessive desire to preserve sameness (Kanner). Autistic children frequently show repetitive stereotyped movements (whirling or rocking) and demonstrate selectivity of attention such as ignoring noises or painful stimuli yet concentrating on spinning objects or self-induced sounds. They ignore other persons' feelings or existence and make no eye contact. Imitation is poor, and they show abnormal social play. In most cases there is no family history of the same syndrome. A good number of neurologic disorders, including those caused by prenatal infections, perinatal anoxia or trauma, or metabolic or chromosomal abnormalities, can have the same clinical phenotype, but none provide clues as to the cause of autism. The screening of these disorders depends on the clinical information obtained by the physician. In autistic cases that show no other neurologic deficit, neuropathologic studies have described subtle cytoarchitectonic abnormalities in the temporal lobes, neostriatum, and thalamus suggestive of a dopaminergic disturbance. However, a definite cause is still elusive. The EEG is frequently normal, and the imaging studies can show nonspecific subtle abnormalities. The prognosis is usually poor with only one third of the cases developing some form of speech (communication). Treatment consists of behavior modification and the use of some antipsychotic medications. Fenflurazine, a serotonin-depleting agent, has shown mixed results. There is poor response to CNS stimulants.

## **HYPERACTIVITY-ATTENTION DEFICIT DISORDER**

The normal level of activity varies considerably among children from the time of birth. Some normal children are very placid, and others are extremely active. Activity is normally accentuated when a child begins to run and walk. Males are generally more active than females, and cerebral defects can accentuate hyperactivity. Hyperactive behavior can become apparent when a child cannot stay in one position for more than a few seconds; is very inattentive, distractible, or impulsive; and is unable to maintain adequate concentration in school and often other situations. This problem is occasionally seen as the sole impediment to adequate school performance, and teachers may demand a medical consultation. To make a diagnosis of

hyperactivity in a child, the physician needs to obtain information from different sources, including the child's teachers, and the diagnosis should be made after observation for at least 12 months. Parental marital problems and parental depression, anxiety, and substance abuse need to be investigated. The cause of the disorder is not known but resembles the postencephalitic syndrome. Some cases are familial. Some patients can show "soft neurologic signs" but not more than what is seen in the normal pediatric population. Imaging studies have revealed asymmetry in the anterior part of the cerebral hemispheres. The prognosis is variable, but nearly half of the patients improve around adolescence. However, as a group the patients have a higher incidence of truancy, alcohol abuse, and juvenile delinquency. The treatment should be individualized and should include counseling for the patient and the family, adjustments in the school programs, and use of stimulants when needed. Three medications are most frequently used. Methylphenidate (Ritalin) takes 30 minutes to work, and its action lasts 4 to 6 hours; thus, the patients need to take one 5 mg tablet in the morning and one at noon. Some patients need a third dose in the afternoon. The undesirable side effects include anorexia, insomnia, headache, abdominal pain, and hypertension. A 20 mg sustained-release tablet can be used for one dose a day in the mornings. Dextroamphetamine (Dexedrine) has a longer half-life than Ritalin and can be used in patients who are unresponsive to Ritalin. Pemoline (Cylert) is the third choice of medication. It has a 12-hour half-life, but the medication needs several days or weeks to reach the maximum effect.

## DEVELOPMENTAL DYSLEXIA

*Developmental dyslexia*, or congenital word blindness, is a specific visuoperceptual defect of reading in an otherwise intelligent person. Children with this disorder have extraordinary difficulty identifying printed words and also have difficulty spelling and writing despite the ability to recognize individual letters. However, dyslexics are able to recognize the meaning of diagrams, objects, and pictures. There are several levels of reading performance, from minor faulty letter sound associations to an almost complete inability to read. The disorder is highly familial, and left-handedness is statistically higher in patients and relatives. Hyperactivity and attention deficit disorders are frequently associated with dyslexia. Neuropathologic studies have revealed cytoarchitectonic dysplasias in the perisylvian regions of the left hemisphere. The significance of these changes is undetermined. Imaging studies can detect subtle changes in the same regions. Treatment should be started after a complete evaluation of the deficit is made and should emphasize the needs of the child, including phonetics and using better learning strategies. Most dyslexic children finish high school, and most live independent successful lives. Some variations of disorders of higher cortical functions include *dysphasia*, *dysgraphia*, and *dyscalculia*. These disorders also are most likely associated with other brain pathology, can be identified by special neuropsychologic testing, and should be treated accordingly.

## EPILEPSIES

The epilepsies have been reviewed in Chapter 11. Only the most frequent types that may be seen by primary care doctors will be emphasized here.

### Febrile Seizures

*Febrile seizures* occur at the onset of an acute febrile illness in a child who is usually between 3 months and 5 years of age and who most likely has a family history of febrile seizures. The

seizure occurs early in the illness when the temperature is rising rapidly. The most frequent seizure is the generalized tonic-clonic type, but other types of seizures can occur including focal motor seizures and seizures manifested by staring, stiffening of the limbs, or just limpness. The febrile illness is usually a tonsillitis, upper respiratory infection, or otitis media. Recurrence after the first febrile seizure occurs in one third of the patients. The younger the patient is when a febrile seizure occurs, the more likely it is that there will be a recurrence. The risk factors for later epilepsy include developmental abnormality or neurologic deficit before the seizure, a family history of epilepsy in one parent, a history of a febrile seizure that was focal or that lasted more than 15 minutes, and an abnormal EEG. The differential diagnosis includes bacterial or viral meningitis or encephalitis, epidural or subdural empyema, hypernatremic dehydration, septic embolization, Reye's syndrome, and any other infection. Management consists of controlling the seizure by lowering the patient's temperature and treating the seizure with anticonvulsants. The cause of the fever needs to be investigated and treated. Treatment with anticonvulsants has not been shown to prevent the development of epilepsy. Parents of children with the first seizure need to be reassured and counseled regarding the favorable prognosis in most of the patients.

### **Benign Rolandic Epilepsy**

*Benign Rolandic epilepsy* is an important form of autosomal dominant childhood epilepsy that remits spontaneously. The seizures occur more frequently between the ages of 5 and 10 (range 2 to 12) and are characterized by a single nocturnal generalized seizure of focal onset and diurnal focal motor seizures. The nocturnal seizure starts with brief twitches of the mouth with salivation and gurgling sounds followed by generalization of the seizure. Unfortunately most parents see only the generalized part. The daytime seizures are focal in onset. In most cases there is no loss of consciousness and no amnesia or postictal confusion. The seizure starts in the face and is associated with speech arrest, salivation, and guttural sound. Involvement of the arm and leg on the same side can occur. There are no automatisms, hallucinations, or auras associated with the seizures. The seizures are sporadic, and the interictal neurologic examination is normal. The interictal EEG shows high-amplitude spikes followed by slow waves that can be seen isolated or in groups and that originate in the midtemporal and Rolandic (central) region. The EEG pattern is seen in 50% of patients' close relatives, but only 12% of the people with this EEG abnormality develop seizures. When this stereotype seizure occurs along with the EEG pattern, the use of costly imaging studies should be avoided. Treatment may not be necessary after the first or even the second seizure. Monotherapy with phenytoin or carbamazepine easily controls the seizure.

### **Lennox-Gastaut Syndrome**

This syndrome is characterized by medically intractable recurrent seizures of various types that frequently begin in the first 6 years of life (rarely after 10), are frequently associated with severe mental retardation, and show a slow spike-wave or slow spike-wave discharges on the EEG during a portion of the awake state. The type of seizure varies. *Tonic* contractions of muscle groups occur commonly during sleep and are associated with impaired consciousness that lasts from a few seconds to 1 minute. Atonic (astatic) seizures are also known as drop attacks. There is no warning, and they are sudden in onset and are associated with loss of tone and momentary loss of consciousness. There is no postictal confusion. Facial injuries can occur because most children fall forward. Myoclonic jerks frequently precede the atonic seizure. Nocturnal generalized tonic-clonic seizures occur in 60% to 70% of the patients. Seizure frequency is

variable from day to day in the same child. Seizures occur more frequently during inactivity or drowsiness and can be as frequent as 50 or more per day. The prognosis of this syndrome is poor, and the response to anticonvulsants is dismal. Most patients need polytherapy. Treatment should be individualized for each patient.

### **Infantile Spasms**

Infantile spasms are also known as salaam seizures, flexion spasms, massive spasms, jack-knife seizures, massive myoclonic jerks, and infantile myoclonic seizures. All these terms describe the type of seizure that is seen in these infants, which varies from brief head nods to violent flexion of the trunk and limbs. The minor forms can be misdiagnosed as colics or other medical problems. The onset of the seizures is confined to the first 2 years of life. The seizures usually occur in clusters, during sleep or after awakening. The EEG is highly abnormal and frequently shows diffuse high-voltage random slow waves and multifocal spikes, called hypsarrhythmia. Infantile spasms can occur without a clear cause (idiopathic) or can be found in infants with severe hypoxia, congenital malformations, congenital infections, neonatal intracranial hemorrhage, and metabolic disorders such as phenylketonuria. Tuberous sclerosis is frequently associated with infantile spasms. The prognosis is poor for most patients but is worse in those with associated neurologic disease. In the idiopathic cases the patients are left with significant neurologic impairment. The treatment consists of adrenocorticotrophic hormone (ACTH) or corticosteroids. Clonazepam and valproic acid have also been used with moderate success.

### **Benign Juvenile Myoclonic Epilepsy of Janz**

This disorder is characterized by the presence of mild myoclonic jerks (see myoclonic epilepsy, p. 337) that affect predominantly the neck and upper limbs in a patient who is alert. The jerks are more frequent in the mornings and are aggravated by sleep deprivation. Some patients can have absence or generalized tonic-clonic seizures. The seizures affect predominantly males with an onset in the second decade (age 8 to 24) and a family history of the disorder. The EEG is characterized by 3.5 to 6 Hz spike-wave or multiple spike-wave complexes. The seizures are easily controlled with long-term use of valproic acid.

## **MALFORMATIONS OF THE CENTRAL NERVOUS SYSTEM**

Most major deviations from normal embryonic development of the brain occur early in the first trimester of embryonic life and can be produced by a variety of genetic, infectious, metabolic, toxic, and irradiation insults. Although the nature of the insult may be obscure, the time of the insult can be determined. This group accounts for 75% of fetal deaths and 30% to 40% of deaths during the first year of life. Surviving children can have intellectual impairment or various somatic anomalies.

### **Spina Bifida**

Spina bifida, myelomeningocele, and myelodysplasia are failures of closure of the vertebral arches with variable involvement of the spinal cord and meninges. These defects are more frequently found in the lumbar areas. *Spina bifida occulta* is not accompanied by protrusions of meningeal or spinal cord elements. Symptoms are absent or appear late in life. It can be associated with dimples of overlying skin, dermal sinuses, lipomas, diastematomyelia (division of spinal cord by fibrous tissue, cartilage, or bony spicules), or syringomyelia. The defects can be

detected only by x-rays or imaging studies of the spine. *Meningomyelocele* is evident at birth, is most frequently found in the lumbosacral region, and can rupture during birth causing leakage of cerebrospinal fluid. Neurologic deficits vary according to the level of the lesion and consist of weak legs with inverted feet held in equinovarus position, depressed stretch reflexes, sensory loss (in the feet and saddle or perirectal region), and sphincteric disturbances. In 90% of patients with lumbar meningomyelocele, there is an associated Chiari type II malformation usually with hydrocephalus. The exact cause of the defect is not known, but in families with one affected child there is a 10% chance of another child having a similar defect. Treatment consists of immediate surgical closure of the defect. Folic acid, 4 mg a day, supplementation in subsequent pregnancy decreases incidence of neural tube defects in women who have given birth to affected infants. Folic acid, 0.4 mg, is recommended to decrease incidence of birth defects in all pregnancies.

### **Chiari Type I Malformations**

Chiari type I malformation consists of cerebellar tonsillar displacement through the foramen magnum down into the cervical spine but without brain stem displacement. The malformation is usually asymptomatic until later in life, when brain stem and upper cervical cord symptoms of a progressive nature can occur as a result of adhesions between the cerebellum, lower brain stem, and spinal cord. MRI is used to diagnose the lesion. Treatment consists of surgical decompression through a laminectomy and occipital craniectomy.

### **Chiari Type II Malformation**

This malformation consists of elongation of the pons, medulla, and fourth ventricle and downward displacement of the brain stem along with the cerebellar tonsils through the foramen magnum into the cervical spinal canal (Figure 13-1). Very frequently there is an association of other malformations such as aqueductal stenosis, microgyria, gray matter heterotopia, and syringomyelia. The malformation should be suspected in all newborns who have meningomyelocele and are born with or develop hydrocephalus early after birth. The diagnosis is confirmed by MRI. Treatment consists of relief of hydrocephalus.

### **Dandy-Walker Syndrome**

This syndrome consists of a cyst dilatation of the fourth ventricle, hypoplasia of the cerebellum, and hydrocephalus (Figure 13-2). Frequently there are other anomalies such as agenesis of the corpus callosum, facial clefts, and cardiac abnormalities. The expanded posterior fossa produces upward displacement of the tentorium along with the torcular and lateral venous sinuses. The large posterior fossa causes dolichocephaly. CT scanning and MRI confirm the presence of the lesion.

## **NEURO CUTANEOUS SYNDROMES**

Neurocutaneous syndromes are genetically determined disorders that affect the CNS, skin, and other viscera. They can be present at birth or appear later in life.

*Neurofibromatosis* is an autosomal-dominant inherited disease with marked variability of expression and a slight preponderance in males. There are two different syndromes. Neurofibromatosis type I is the most frequently found syndrome and is linked to chromosome 17(17q11.2). Neurofibromatosis type II is linked to chromosome 22(22q11-q12).

## **Neurofibromatosis Type I**

This is a clinically and genetically heterogeneous syndrome with almost 100% penetrance and a high rate of spontaneous mutations. The diagnosis of neurofibromatosis has to be based on finding two of the following features: family history of neurofibromatosis type I, café au lait spots, neurofibromas, axillary or inguinal freckling, optic glioma, Iris (Lisch) nodules, or one of the distinctive osseous lesions (thoracic scoliosis, anterolateral bowing of the tibias, pseudoarthrosis, sphenoid wing dysplasia). Age is an important factor in the appearance of some of these lesions. Café au lait spots are better recognized after the first year of life, the Lisch nodules are seen in all affected patients only after age 20, and neurofibromas grow with age. These last three findings are seen in all adult patients with neurofibromatosis type I. Five café au lait spots measuring more than 5 mm in diameter before puberty and six measuring more than 15 mm after puberty are required to suggest or confirm the diagnosis. The location in the sun-protected areas and the trauma- or rubbing-inducing factor are very characteristic of these freckles. The tumors can occur distally, can be cutaneous or subcutaneous, and are usually localized or nodular (neurofibromas) or can be diffuse or less localized (plexiform neurofibromas). Proximal tumors are usually schwannomas. Acoustic neuromas do not occur in this type of neurofibromatosis. The symptoms associated with these tumors include neuropathic pain and esthetic problems. The CNS problems in neurofibromatosis I can include developmental delays, mental retardation, seizures, loss of vision, and headaches. Some of the CNS symptoms are caused by brain or blood vessel dysplasia. CNS tumors include multiple meningiomas and optic nerve and brain stem gliomas. Spinal cord lesions include syringomyelia, hydromyelia, and compressive myelopathies caused by tumors or bone deformities. Imaging studies are helpful in diagnosing these conditions. The treatment consists predominantly of symptomatic management of the lesion by different specialists, including neurosurgeons and orthopedic doctors. Excision of symptomatic intracranial tumors is mandatory. Mental retardation and seizures should be treated accordingly.

## **Neurofibromatosis Type II**

Also known as bilateral acoustic neuroma or central neurofibromatosis, this condition is a genetically distinct entity from neurofibromatosis type I. Acoustic neuromas, usually bilateral and seen before the age of 30, are always present. Café au lait spots, posterior lens opacities, absence of Lisch nodules, and a family history of the disease are confirmatory of neurofibromatosis type II. Other CNS tumors including meningiomas and gliomas (except for optic nerve gliomas) are more frequent than in type I. Mental retardation, seizures, and macrocephaly do not occur. Clinical symptoms of acoustic neuromas consist of tinnitus, deafness, headaches, and brain-stem dysfunction. Syringomyelia and hydromyelia may occur. MRI studies confirm the diagnosis and can detect small resectable acoustic tumors before they produce permanent deafness. Genetic counseling is very important.

## **Tuberous Sclerosis**

Tuberous sclerosis is inherited as an autosomal dominant disease with markedly variable expression, high penetrance, and a high spontaneous mutation rate. The disease is also genetically heterogeneous and has been linked to chromosome 9(9q34) in 35% of the families and to chromosome 11(11q21-23) in other families. The diagnosis is based on finding one of the characteristic malformations such as the focal cortical dysplasias (tubers), subependymal

nodules, giant cell subependymal tumors, retinal astrocytic hamartomas, facial angiofibroma (misnamed adenoma sebaceum) or unguis fibromas. The diagnosis can also be based on finding several less specific lesions, such as cardiac rhabdomyomas, renal angiomyolipomas, pulmonary angiomyolipomas, or rectal polyps. The clinical neurologic manifestations include a high frequency of seizures that vary from infantile spasms in infancy to posterior focal (partial) seizures before 2 years of age to temporal and frontal seizures later on. Mental retardation is found in half of the patients of all ages. All mentally retarded patients with tuberous sclerosis have seizures, but one third of the tuberous sclerosis patients have seizures without mental retardation. Visceral lesions include involvement of the kidneys and the heart. The renal lesions include multiple cysts or angiomyolipomas. The cardiac rhabdomyomas are usually asymptomatic. Benign rectal polyps and vascular dysplasia have also been described. Treatment consists of control of the seizures with the appropriate anticonvulsant medication, surgical resection of intraventricular tumors when hydrocephalus develops, and resection of renal and other visceral lesions when indicated.

### **Sturge-Weber Syndrome (Encephalotrigeminal Angiomatosis)**

Sturge-Weber syndrome does not have a well-defined inheritance pattern. Although most cases are sporadic, a few are autosomal recessive or autosomal dominant. The disease is characterized by dysplasia of mature capillaries in the distribution of the first division of the trigeminal nerve, ipsilateral occipitoparietal leptomeningeal angiomatosis with degeneration and calcification of the underlying cortex as a result of ischemia, contralateral hemiplegia, and contralateral focal or generalized seizures. Glaucoma in the eye ipsilateral to the vascular lesions can be present. Aspirin and other antithrombotic agents prevent important neurologic sequelae. Anticonvulsant therapy controls the seizures. Hemispherectomy has been performed for many intractable seizures.

## **DEGENERATIVE AND METABOLIC DISEASES**

Degenerative diseases comprise a group of rare neurologic disorders, usually inherited and frequently progressive, that can appear early after birth or later in life with severe developmental neurologic regression. Metabolic diseases must be suspected when these criteria are associated with recurrent unexplained ataxia or spasticity, recurrent episodes of altered consciousness or unexplained vomiting, unexplained acidosis, or mental retardation in the absence of major congenital anomalies. The confirmation of the diagnosis should be made by detection of abnormal substances or their metabolites in the urine or blood and titration of specific enzymes in urine, blood or more accurately from tissue culture of fibroblasts obtained through a skin biopsy. In some of the metabolic disorders in which the exact enzymatic defect has not yet been found but accumulation of abnormal metabolites is deposited in the tissue, biopsy of peripheral nerve, muscle, bone marrow, or brain tissue may be necessary and should be done where adequate facilities are available. Although treatment is available for only a few of these disorders, an exact diagnosis is important for genetic counseling and prognosis. Bone marrow transplantation has been successful in a few cases when done early in the course of the disease. Intrauterine diagnosis by amniocentesis may be possible in pregnancies at risk for certain of these disorders and can be helpful in deciding whether to terminate the pregnancy. Only a treatable and preventable disorder will be described here.

## **Hepatic Encephalopathy and Reye's Syndrome**

Reye's syndrome is an acute encephalopathy of childhood of undetermined cause. The disease is preceded by a viral illness, especially influenza B and varicella. Frequently the affected children have received aspirin, phenothiazines, or trimethobenzamide (Tigan). The disease is characterized by vomiting followed by delirium, lethargy, coma, and seizures. During coma the child can develop decorticate or decerebrate posture or opisthotonos, the pupils dilate, and breathing becomes irregular. Biochemical alterations consist of early hypoglycemia, especially in younger patients, and hyperammonemia, abnormal liver enzymes without elevation of bilirubin, and elevated serum levels of free and short-chain fatty acids. Striking intracranial hypertension produced by severe cerebral edema produces the change in sensorium and eventually results in death. Mortality correlates with the level of consciousness at admission to the hospital. Fatty degeneration of the liver, kidneys, and heart without cell necrosis is a constant feature in postmortem studies. Recovery in nonfatal cases occurs in 2 or 3 days. Treatment consists of intravenous administration of hypertonic glucose solutions in the early stages that may prevent progression of the disease. Control of temperature, early monitoring of intracranial pressure, and reduction of intracranial pressure by hyperventilation and use of mannitol is important. Early correction of metabolic derangements and control of intracranial pressure can lead to recovery in most patients.

### **SUMMARY**

The neurologic evaluation of a child is different from that of an adult, and in a good number of cases it requires the expertise of a pediatric neurologist. Only the most frequent pediatric neurologic problems are reviewed here. The history need to be obtained from the parents or guardian of the child. The hospital records may need to be obtained to review the perinatal history and the birth events. In older children records of performance in school or day-care centers are important to learn about the emotional and intellectual characteristics of the child. Important reasons for pediatric consultation of the neurologist are disorders of motor execution such as dyspraxia, clumsiness, and adventitious movements as well as attention deficit disorders, which are discussed in this chapter. There are important differences between cerebral palsy and mental retardation that are important for the primary care physician to know. There are also special types of seizures, some of them benign, that need to be differentiated from the more malignant types seen in the pediatric population. Finally, the most frequent neurocutaneous syndromes are delineated here.

### **Suggested Readings**

#### **Suggested Readings—General**

- Adams RD, Victor M: Principles of neurology, ed 5, New York, 1993, McGraw-Hill.  
David RB: Pediatric neurology for the clinician, Norwalk, Conn, 1992, Appleton & Lange.  
Schaefer GS, Bodensteiner JB: Evaluation of the child with idiopathic mental retardation, *Pediatr Clin North Am* 39:929, 1992.  
Swaiman KF, Wright FS: The practice of pediatric neurology, ed 2, St Louis, 1982, Mosby.  
Volpe JJ: Neurology of the newborn, ed 2, Philadelphia, 1987, WB Saunders.

## **Birth Injuries**

Allen WC: The intraventricular hemorrhage complex of lesions: cerebrovascular injury in the preterm infant, *Neurol Clin North Am* 8:529, 1990.  
Freeman JM, Nelson KB: Intrapartum asphyxia and cerebral palsy, *Pediatrics* 84:240, 1988.

## **Developmental Abnormalities**

Dunsker SB, Brown D: Craniovertebral anomalies, *Clin Neurosurg* 27:430, 1980.  
Loesser JD, Albord EC: Agenesis of corpus callosum, *Brain* 91:553, 1968.  
Sawaya R, McLaurin RL: Dandy-Walker syndrome, *J Neurosurg* 55:39, 1981.

## **Hydrocephalus**

DeMeyer W: Megalencephaly, *Pediatr Neurol* 2:321, 1986.  
Naidich TP, Schott LH, Baron RL: Computed tomography in the evaluation of hydrocephalus, *Radiol Clin North Am* 20:143, 1982.

## **Neurocutaneous Syndromes**

Riccardi VM, Von Recklinghausen: neurofibromatosis, *N Engl J Med* 305:617, 1981.  
Roach ES: Encephalofacial angiomas, *Pediatr Clin North Am* 39:606, 1992.  
Roach ES: Neurocutaneous syndromes, *Pediatr Clin North Am* 39:600, 1991.  
Roach ES: Tuberous sclerosis, *Pediatr Clin North Am* 39:591, 1991.

## **Metabolic Diseases**

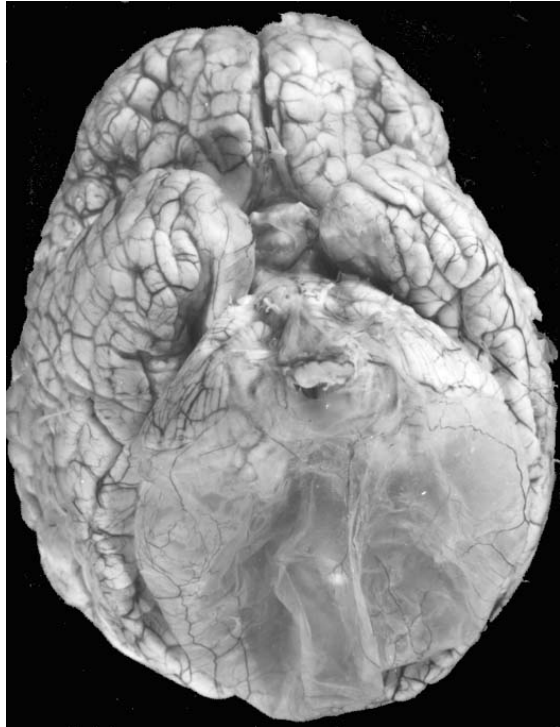
DiMauro S, Moraes CT: Mitochondrial encephalomyopathies, *Arch Neurol* 50:1197, 1993.  
Kolodny EH, Cable WJL: Inborn errors of metabolism, *Ann Neurol* 11:221, 1982.  
Naidu S, Moser HW: Peroxisomal disorders, *Neurol Clin North Am* 8:507, 1990.

## **Hypotonic Infant**

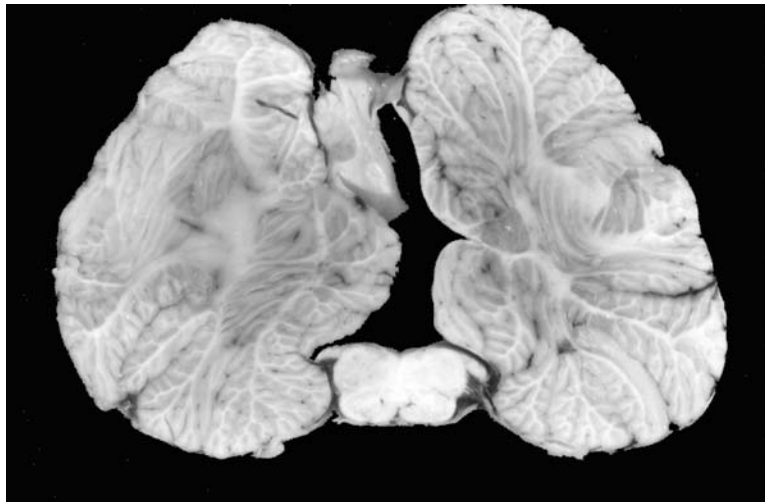
Rabe EF: The hypotonic infant, *J Pediatr* 64:222, 1964.  
Dubowitz V: The floppy infant. London, 1969, Spastics International Medical Publications.



FIGURE 13-1. Sagittal section of brain and spinal cord showing meningocele (lower arrow) and Chiari type II malformation. Upper arrow shows level of foramen magnum. Note hydrocephalus.



A



B

FIGURE 13-2. A, Ventral view of brain showing a dilated fourth ventricle covered by a membrane. B, Same specimen as A. Note the defect of the cerebellar vermis and the attachment of the membrane.