Magnetic resonance imaging (MRI) is the imaging modality of choice for the study of congenital brain disease. Here, T1-weighted scans play a dominant role. T1-weighted scans provide excellent information regarding structural lesions and disorders of white matter. For structural lesions, acquisition of images in multiple planes is important. T1-weighted and T2-weighted scans both chronicle the normal myelination process, making possible early diagnosis of the leukodystrophies. The role of T2-weighted scans is limited, as is the role of contrast enhancement. These only occasionally contribute information about structural defects and significant associated sequelae. Contrast enhancement is, however, important in the evaluation of associated tumors, as may occur with the neurocutaneous syndromes.

NORMAL VARIANTS
(VENTRICULAR SYSTEM)
Cavum Septum Pellucidum
The septum pellucidum is a thin, translucent plate consisting of two laminae (leaves) lying in the midline between the frontal horns of the lateral ventricles. The septum pellucidum links the hippocampus to the hypothalamus. Abnormalities of the septum pellucidum may have subtle associated neuropsychiatric symptoms. The cavum septum pellucidum is a normal embryologic space. It is present in all fetuses and premature infants. By 3 months of age, it is seen in only 15%. Persistence into adulthood can occur and is considered a normal variant (Fig. 5–1). When the distance between the leaves is large (greater than 1 cm), obstruction can occur to cerebrospinal fluid (CSF) flow at the foramen of Monro.

Cavum Septum Vergae
The cavum septum vergae is a normal embryologic cavity, like the cavum septum pellucidum. It is essentially a posterior extension of the cavum septum pellucidum. The cavum septum vergae is that part of the midline cavity posterior to the columns of the fornix. It ends at the splenium of the corpus callosum. The cavum septum vergae begins to disappear at 6 months gestational age. In the adult, it is considered a normal variant.

Cavum Velum Interpositum
Cavum velum interpositum are much less common than cavum septum pellucidum or vergae. In this variant, there is separation of the crura of the fornix between the thalami and above the third ventricle. A cavum septum vergae lies superior to the internal cerebral veins. The latter lie within a cavum velum interpositum.

CRANIOVERTEBRAL ANOMALIES
Basilar Invagination (Impression)
In this bony craniovertebral junction anomaly, the odontoid is high in position relative to the foramen magnum. A more specific definition is that the tip of the odontoid lies more than 5 mm above Chamberlain's line. The latter extends from the posterior edge of hard palate to the posterior lip of the foramen magnum. The dens can compress or displace the medulla (Fig. 5–2).

Basilar invagination can be the result of a primary bone anomaly. In this circumstance, it is often associated with assimilation of the posterior arch of Cl to the occiput. Basilar invagination can also be secondary to other diseases. These include osteoporosis, osteomalacia, Paget's disease, fibrous dysplasia, achondroplasia, and osteogenesis imperfecta. Platybasia may also be present. In platybasia, there is a flattened relationship between the anterior and middle cranial fossae, with the angle formed greater than 140 degrees. Patients with basilar invagination can present clinically with headache, neurologic deficits, and symptoms from vertebrobasilar artery compression.

CHIARI MALFORMATIONS
Chiari type I and II malformations are commonly encountered in clinical practice. The anatomic features of these conditions are well delineated by MRI. Because Chiari type I malformations may present with clinical symptoms suggesting demyelination or neoplastic disease, MRI is the usual mode of examination. Chiari type I malformations are also one of the more common congenital abnormalities encountered in asymptomatic patients. Requests for imaging are frequent with Chiari type II malformations because of the associated spinal anomalies and their sequelae. For the Chiari malformations, MRI is the easiest and most accurate method of diagnosis because of the anatomic detail visualized within the posterior fossa and upper cervical canal.

Chiari Type I Malformation
The primary feature of Chiari type I malformation is the abnormal cerebellar tonsil position and morphology.
There is downward and posterior herniation of the cervicomedullary junction with a variable amount of tissue below the foramen magnum (Fig. 5–3). The tonsils are wedge shaped. The cisterna magna is small or absent. Two thirds of cases with Chiari type I have downward displacement of the tonsils inferior to C1. In one fourth, the herniation reaches the C3 level. Associated anomalies include hydrosyringomyelia (typically cervical), with variable extent, and hydrocephalus. The primary features of Chiari type I that separate it from Chiari type II are the normal position of the fourth ventricle, absence of supratentorial structural anomalies, and lack of an associated myelomeningocele.

Chiari type I may also be associated with bony anomalies involving the skull base and cervical spine. These anomalies include basilar impression, fusion of C1 to the occiput, fusion of C2 and C3, Klippel-Feil deformity, and spine bifida occulta.

**Chiari Type II Malformation**

Chiari type II refers to a cerebral dysgenesis associated with a neural tube closure abnormality, specifically a myelomeningocele. There is a myelomeningocele in nearly 100% of cases. Myelomeningoceles are the extreme form of spinal dysraphism. There is a midline defect of the posterior bony elements of the vertebral body, usually in the lumbosacral region. Although the muscle, fascia, and skin are split along the midline, the meninges typically remain intact. The incidence of this sporadically appearing syndrome is 0.3%. Chiari type III refers to a very rare dysgenesis, with Chiari type II features and a low occipital or high cervical encephalocele.

Common infratentorial structural changes in Chiari type II include the following (Fig. 5–4). There may be downward displacement of the cervical spinal cord. The brainstem may be elongated, with the medulla inferiorly displaced into the cervical canal. In extreme cases, the displaced medulla may fold over on itself behind the cervical spinal cord, forming a kink. The fourth ventricle is typically inferiorly displaced and may be at or below the foramen magnum. The cerebellum may protrude through the foramen magnum to create a cerebellar peg with compression of the inferior vermis. There may
be forward displacement of the cerebellar hemispheres, enveloping the brainstem. These may touch anteriorly, in front of the pons, in rare cases. In many cases, the folia of the superior cerebellum have an abnormal configuration, with an exaggerated craniocaudal orientation. The tentorial incisura is wide, creating the visual effect of a towering cerebellum with enlarged supracerebellar CSF spaces. The clivus and Petroc ridges may have an altered contour.

There are multiple common midbrain and supratentorial structural changes in Chiari type II. The colliculi are typically fused, creating a beaked or bulbous tectum. The massa intermedia may be large. Hydrocephalus is common, with inferiorly pointing frontal horns, large atria, and a prominent suprapineal recess of the third ventricle. Interdigitating gyri are common, accompanying fenestration or partial absence of the falx. The gyri are often thin and numerous, an appearance termed stenogyria. This is not to be confused with polymicrogyria, a finding not seen in Chiari type II and one in which the gross appearance is that of a smooth brain. There is agenesis of the corpus callosum in about one third of all cases. Also common is an abnormal interhemispheric CSF space, of variable size and configuration. Hydrosep- ringomyelia, which may be cervical or lumbar in location, is seen in about half of all cases.

**OTHER ANOMALIES OF THE POSTERIOR FOSSA**

**Dandy-Walker Malformation**

The Dandy-Walker malformation is characterized by absence of the inferior vermis (Fig. 5–5). The fourth ventricle is large and communicates freely with a large cystlike structure, posterior in location. The posterior fossa is usually expanded, with elevation of the torcular. CSF flow dynamics may be abnormal because of obstruction at the foramen of Magendie or Luschka. However, not all cases have obstruction at autopsy, and in vivo demonstration of foraminal patency is difficult.

These structural anomalies are thought to be due to an embryologic dysgenesis and not to a permanent obstructive process. Hydrocephalus is usually present and is highly variable in severity. The severity of hydrocephalus is the most important prognostic factor. Other cerebral anomalies associated with Dandy-Walker malformation are agenesis of the corpus callosum, cortical heterotopias, polymicrogyria, and brainstem lipomas.

In a few patients, the CSF collection is smaller, without posterior fossa expansion. In this Dandy-Walker variant, the torcular is in normal position. The foramen of Magendie is patent, and there are normal CSF dynamics.

**Arachnoid Cyst**

With a retrocerebellar arachnoid cyst, the inferior vermis is intact and the CSF space anterior to the brainstem small because of mass effect. These features differentiate a retrocerebellar cyst from the Dandy-Walker malformation. However, as with the latter, the torcular may be elevated. The presence of mass effect is used to differentiate a retrocerebellar arachnoid cyst from a prominent cisterna magna.

Other characteristic locations for arachnoid cysts include the middle cranial fossa (the most common), brain convexity (Fig. 5–6), and perimesencephalic cistern. Hypogenesis of the temporal lobe is a common finding in middle cranial fossa arachnoid cysts. Arachnoid cysts are benign CSF-filled lesions. They should be CSF signal intensity on all pulse sequences and lack contrast enhancement. Most arachnoid cysts are congenital in origin. An arachnoid cyst may also form after head trauma, leptomenigitis, and subarachnoid hemorrhage. Although frequently asymptomatic, arachnoid cysts can be symptomatic as a result of mass effect.

**Cerebellar Hypoplasia**

Cerebellar hypoplasia is characterized by absence of cerebellar or vermian tissue. In its place is a passive CSF
FIGURE 5–4. Chiari type II malformation. A. The midline sagittal T₁-weighted image demonstrates elongation and inferior displacement of the fourth ventricle, a beak-shaped tectum, and a large massa intermedia. The corpus callosum is thin anteriorly and absent posteriorly. At the level of the fourth ventricle on the axial T₁-weighted scan (B), the cerebellar hemispheres are displaced anteriorly, partially surrounding the pons. C. The coronal T₁-weighted scan demonstrates abnormally wide margins of the tentorial incisura, a towering cerebellum, abnormal orientation of the cerebellar folia, an enlarged interhemispheric fissure, and interdigitation of gyri (with absence of the falx).

FIGURE 5–5. Dandy-Walker malformation. On the midline sagittal T₁-weighted scan (A), the posterior fossa is noted to be enlarged, containing principally cerebrospinal fluid. The inferior cerebellar vermis is absent and the torcular herophili elevated. There is scalloping of the inner table of the occipital bone. Communication between the fluid posteriorly and the fourth ventricle is confirmed on the axial T₂-weighted scan (B).

FIGURE 5–6. Arachnoid cyst. A large mass with cerebrospinal fluid (CSF) signal intensity is noted over the right brain convexity on T₂- (A) and T₁-weighted (B) images. There was no abnormal contrast enhancement (not shown). Long-standing mass effect is evident, with scalloping of the adjacent calvarium.
space. Usually only the most anterior portions of the cerebellar hemispheres are present. The cerebellar hemispheric remnants may be asymmetric. The cerebellar peduncles and brainstem are hypoplastic. The posterior fossa is small, with a low torcular.

**DISORDERS OF CEREBRAL HEMISPHERIC ORGANIZATION**

**Agenesis of the Corpus Callosum**

Agenesis of the corpus callosum is a relatively common congenital anomaly. Agenesis may be partial or complete. The hippocampal and anterior commissures may also be absent. The posterior commissure is typically intact. The corpus callosum is composed of four parts. Progressing anteriorly to posteriorly, the rostrum, genu, body, and splenium can be identified. These structures form embryologically from anterior to posterior. This temporal sequence of formation explains the consistent absence of the more posterior elements in partial agenesis.

Multiple features are associated with agenesis of the corpus callosum (Fig. 5–7). The third ventricle is large and high in location. In the coronal plane, the frontal horns are concave medially. White matter bundles (of Probst) run along the medial wall of the lateral ventricle. The lateral ventricles are widely separated and parallel. The gyri radiate in a medial direction because of the absent cingulate gyrus. The anterior commissure, if present, may be enlarged and dysplastic. The ventricular atria may be rounded because of the absence of the splenium and portions of the forceps major. There may be a wandering anterior cerebral artery. Heterotopic gray matter may be present.

Agenesis of the corpus callosum is easy to identify on MRI scans after the age of 2 years. In neonates, however, myelination is not complete, and agenesis is difficult to diagnose. At this age, the normal corpus callosum is very thin and difficult to visualize. However, if the sulci on the medial surface of the brain radiate directly from the lateral ventricle, the diagnosis may be suggested even in the neonate. Eighty percent of cases of agenesis have other associated findings. The most common anomalies associated with agenesis are Chiari type II malformation, Dandy-Walker malformation, holoprosencephaly, neuronal migration abnormalities, encephaloceles, and interhemispheric cysts.

**Lipoma**

The incidence of intracranial lipomas is 0.1%. The most common location is midline within the interhemispheric fissure near the corpus callosum (Fig. 5–8). Other common sites include the quadrigeminal plate cistern, tuber cinereum, and cerebellopontine angle. Less common sites include the base of the cerebrum, the cerebellum, the brainstem, cranial nerve roots, ventral aspect of the midbrain, and choroid plexus of the lateral ventricles. Intracranial lipomas arise from the pia and envelop adjacent neural structures. MRI will demonstrate a fat intensity mass. If associated with agenesis of the corpus callosum, the lesion is most often situated in the midline where the genu of the absent callosum would lie. Approximately 50% of interhemispheric lipomas have associated hypoplasia of the corpus callosum. Calcifications may be present but are not readily identified on MRI.

**Holoprosencephaly**

In utero failure of hemispheric or thalamic separation leads to holoprosencephaly. Half of all cases have chromosomal abnormalities. The incidence is 0.01%. Holoprosencephaly is classically divided into three types by grade of severity. From most to least severe, these are alobar, semilobar, and lobar holoprosencephaly.

Absence of the falx and interhemispheric fissure, fused thalami (with absence of the third ventricle), and a horseshoe-shaped monoventricle characterize the alobar form. The superior sagittal, inferior sagittal, and straight sinuses are absent along with the internal cerebral veins. The roof of the third ventricle may balloon out posteriorly, giving the appearance of a large dorsal “cyst.”

Semilobar holoprosencephaly is characterized by partial formation of the interhemispheric fissure and falx (Fig. 5–9). There is rudimentary differentiation of the occipital and temporal lobes and respective ventricular

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**Figure 5–7.** Agenesis of the corpus callosum. A, The midline sagittal T1-weighted image shows a small, dysplastic genu. The remainder of the corpus callosum is absent. Adjacent to the genu, the cingulate gyrus has a normal orientation. Posteriorly, where the corpus callosum is absent, the cerebral gyri have an abnormal radiating appearance. B, The axial T1-weighted image reveals the lateral ventricles to be widely separated and oriented parallel to each other. On the T1-weighted coronal image (C), the lateral ventricles have an abnormal crescent-like appearance (with indentation medially).
FIGURE 5–8. Interhemispheric lipoma. Axial (A) and coronal (B) T₁-weighted images reveal a midline high-signal-intensity mass immediately superior to the corpus callosum. The lesion was isointense with fat on all pulse sequences, and there was no abnormal contrast enhancement.

FIGURE 5–9. Semilobar holoprosencephaly. A 9-month-old infant presented with microcephaly and developmental delay. Axial T₁-weighted images demonstrate hypotelorism (A), rudimentary formation of the temporal horns (B), a small third ventricle with fusion of more anterior structures (C), and preservation of the corpus callosum posteriorly (D) (arrow). The falx is absent anteriorly. There are no identifiable frontal horns. Incidentally noted is a large retrocerebellar cyst.
horns. A rudimentary corpus callosum is present. There is cleavage of the thalami to form a third ventricle.

A nearly complete interhemispheric fissure and falx characterize lobar holoprosencephaly. Only a small area of the frontal lobe is fused. There are well-formed occipital and temporal lobes. The thalami, third ventricle, and corpus callosum appear normal or nearly normal.

**Septo-Optic Dysplasia**

In septo-optic dysplasia, the septum pellucidum is abnormal and there is hypoplasia of the optic nerves (Fig. 5–10). The abnormality of the septum varies from mild dysplasia to complete absence. Half of all patients with septo-optic dysplasia also have schizencephaly. Septo-optic dysplasia is not a single homogeneous entity. Clinical symptoms include blindness, seizures, hypothalamic-pituitary dysfunction, developmental delay, and growth retardation.

**NEURONAL MIGRATION ANOMALIES**

The neuronal migration anomalies are a varied group of entities caused by abnormal migration of the embryologic neuroblasts, which are arrested along their normal course from the ventricular germinal matrix to the cortical periphery. The primitive neuroblasts normally ascend along radial glial cells during the third and fifth gestational months. If this cellular migration is prevented, abnormal cortical structures result. The entities included in this group are agyria and pachygyria, polymicrogyria, schizencephaly, gray matter heterotopia, and unilateral megalencephaly.

**Agyria and Pachygyria**

Agyria and pachygyria represent a spectrum of cortical malformations, with agyria being the most severe form.
Lissencephalia is a term often used for agyria when gyral formation is very rudimentary, creating the appearance of a “smooth brain.” Agyria and pachygyria describe regions of cortical brain that have diminished gyral formation, creating thick, broad gyri (Fig. 5–11). The ratio of the gray matter to white matter is reversed. Thus, the gray matter cortex is broad. The corpus callosum is thin. The brainstem is small because of failure of corticospinal tract formation. On T₂-weighted images, there is a band of increased signal intensity in the peripheral cortex. It is theorized that normal migration is impaired here at the cell-sparse layer.

**Polymicrogyria**
On imaging in polymicrogyria, the cortex is thick, gyri are not detectable, and the underlying white matter is decreased in quantity (Fig. 5–12). The number of cerebral convolutions is much greater than normal; however, this feature can only be identified histologically. Polymicrogyria is also characterized by an abnormal cellular histology, with the cortex composed of four layers instead of the normal six. Polymicrogyria is to be differentiated from stenogyria. In the latter, the gyri are thin and too numerous but visible on imaging. The cortex in stenogyria has a normal number of cellular layers.

**Schizencephaly**
Schizencephaly is a disorder of cell migration within a segment of the brain, creating a cleft lined by gray matter traversing the hemisphere from the cortex to the ventricles (Fig. 5–13). The cleft may be unilateral or bilateral. Bilateral clefts are associated with severe clini-
BRAIN: CONGENITAL DISEASE

Figure 5–12. Polymicrogyria. On axial T1-weighted images (A–C), the gyri in the distribution of the left middle cerebral artery are noted to be abnormally broad and diminished in number. The cortical gray matter is also too thick. The left lateral ventricle is mildly enlarged and the quantity of white matter on the left diminished. Anomalous venous drainage of the abnormal cortex is also often seen in polymicrogyria. This can be noted in the present case by comparing A and D, the latter a scan with intermediate T2-weighting, which together show a network of vessels feeding a large abnormal vein within a deep sulcus.

Cal impairment. Covering the cleft is a “pial-ependymal” seam, representing fusion of the pial lining of the brain and the ventricular ependyma. The clefts are situated at the precentral or postcentral gyri. Abnormal gyral patterns surround the clefts, including stenogyria and gray matter heterotopia. The clefts may be gaping or “open” or the clefts may be “closed,” with only a gray matter seam extending from the peripheral cortex to the ventricular level. The identification of gray matter lining the cleft permits differentiation from porencephaly and other acquired destructive lesions. This differentiation is important because siblings of patients with schizencephaly have an increased incidence of brain anomalies.

Heterotopic Gray Matter

Heterotopic gray matter represents collections of neurons of varying size in aberrant locations (Fig. 5–14). These collections may be found anywhere between the ependyma and the cortex. Heterotopic gray matter may be an isolated asymptomatic lesion or may be associated with other anomalies, such as Chiari type II or neuronal migration anomalies. Sequences that provide high gray-white matter contrast, such as heavily T1-weighted inversion recovery scans, best delineate these lesions.

Unilateral Megalencephaly

Unilateral megalencephaly is a rare anomaly of the brain characterized by overgrowth of part or all of a cerebral hemisphere, with distorted, thickened cortex and ipsilateral ventricular dilatation. Abnormal signal intensity within the centrum semiovale represents areas of decreased myelination. Because of the intractable seizures associated with this disorder, all areas of abnormal tissue should be delineated to permit surgical resection if possible.

NEUROCUTANEOUS SYNDROMES (PHAKOMATOSES)

The neurocutaneous syndromes refer to a group of disorders that are dysplasias of tissues primarily derived
Figure 5–13. Schizencephaly. Bilateral cerebrospinal fluid-filled clefts are noted on sagittal (A), axial (B), and coronal T₁-weighted images (C). The clefts are lined by gray matter and traverse the brain from the cortex to the ventricular system. Schizencephaly is associated with absence of the septum pellucidum, also noted in this case. Clinical symptoms in this 9-month-old infant included decreased movement of the left arm and leg and a generalized increase in limb tone; the latter suggests bilateral brain involvement.

Figure 5–14. Heterotopic gray matter. Abnormal soft tissue, isointense with gray matter on all pulse sequences, is seen adjacent to the posterior right lateral ventricle on proton density (A), T₁-weighted (B), and T₂-weighted (C) images. The lesions project into the ventricle as small nodules. There is no abnormal contrast enhancement (C). Most patients present clinically with seizures, as this one did. Late-onset, mild symptoms are characteristic for isolated anomalies.
from the embryonic ectoderm. These congenital disorders may also affect the embryonic mesoderm and endoderm. The more common syndromes in this group are neurofibromatosis (von Recklinghausen’s disease), tuberous sclerosis, von Hippel-Lindau disease, and Sturge-Weber syndrome.

**Neurofibromatosis**

Neurofibromatosis (NF) is an autosomal-dominant disorder of neuroectodermal and mesodermal tissues in which the Schwann cell is the primary abnormal element. The incidence of NF is approximately 1 in 3000 births. Two main subtypes exist. NF1 is the classic von Recklinghausen’s neurofibromatosis with multiple central nervous system (CNS), cutaneous, and osseous lesions. Most patients with NF1 have high signal intensity lesions in the brain on T2-weighted imaging. These abnormalities are most often seen in the basal ganglia (specifically the globus pallidus), brainstem, and cerebellar white matter. The pathologic basis and clinical consequence of such abnormalities are unknown, although these lesions most likely represent hamartomas or heterotopias. Abnormal hyperintense foci on T1-weighted scans involving the globus pallidus and internal capsule bilaterally (usually symmetrically) with extension across the anterior commissure have also been described. The same lesions appear smaller and less prominent on T2-weighted scans. There is typically no associated mass effect or abnormal contrast enhancement (Fig. 5–15). Optic nerve gliomas are the most frequent intracranial tumor associated with NF1. Less common, but seen in 10% to 15% of patients, is a primary glioma. NF2, which is much less common than NF1, is characterized by bilateral acoustic neuromas (Fig. 5–16). Cranial nerve tumors, cranial and spinal meningiomas, paraspinal neu-

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**Figure 5–15.** Neurofibromatosis 1. There is symmetric abnormal hyperintensity in the globus pallidus and posterior limb of the internal capsule on both T2- (A) and T1-weighted (B) axial images. There is no mass effect. This patient also had abnormal hyperintensity in the cerebellar white matter bilaterally (not shown).

**Figure 5–16.** Neurofibromatosis 2. Bilateral acoustic neuromas, with prominent contrast enhancement, are identified on T2- (A), precontrast T1- (B), and postcontrast T1-weighted (C) images. Two additional enhancing lesions are seen postcontrast, both adjacent to the dura, compatible with meningiomas. Dense calcification is the cause of the central hypointensity of several lesions.
rofibromas, and spinal cord ependymomas are often seen in NF2.

**Tuberous Sclerosis**

Tuberous sclerosis is an autosomal-dominant disorder with hamartomatous lesions of multiple organs. Seizures, mental retardation, and facial adenoma sebaceum define the classic clinical triad. On MRI, the combination of parenchymal lesions and subependymal nodules is pathognomonic. Rarely, a subependymal nodule may form a giant cell astrocytoma. These usually arise at the foramen of Monro and can be identified as an enlarging, enhancing mass. The subependymal nodules lie along the ventricular wall and have decreased signal intensity on T2-weighted scans. The parenchymal lesions involve both gray and white matter and have increased signal intensity on T1-weighted scans (Fig. 5–17). In some instances, involvement is limited to the subcortical white matter of an expanded gyrus, a “gyral core.” Involvement of two adjacent gyri in this fashion may spare the intervening cortex lining the sulcus, forming a “sulcal island.” The classic renal lesion is an angiomyolipoma, diagnosed by identification of fat within a renal mass.

**Von Hippel-Lindau Disease**

Von Hippel-Lindau disease or retinocerebral angiomatosis is an autosomal-dominant disorder of the vascular elements within multiple organ systems. Hemangiomas and hemangioblastomas are found in the CNS, primarily in the cerebellum. The most common presentation is that of a cystic lesion with a highly vascularized mural tumor nodule. Noncystic, solid lesions do occur but are rare.

On angiography, characteristic findings include a densely staining nodule or an abnormal tangle of vessels. Both may have enlarged feeding arteries and draining veins. Malignant tumors may also involve the retina, kidney, adrenal gland, and pancreas. The retinal lesions

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**Figure 5–17.** Tuberous sclerosis. A 19-month-old infant presented with seizures and mental retardation. Multiple high-signal-intensity lesions are seen on the T2-weighted images (A and B). These involve both gray and white matter. In several instances, the abnormality appears confined to the subcortical white matter core of an expanded gyrus (a “gyral core” lesion). Involvement of two adjacent gyri in this fashion, with sparing of normal intervening cortex lining a sulcus (a “sulcal island”), can also be seen. The parenchymal lesions are of low signal intensity, but in general less well-seen, on T1-weighted images (C and D). There are multiple subependymal nodules, best seen on the T1-weighted scan (C).
are hemangioblastomas, but the malignancies involving the kidney and pancreas are carcinomas. The rare associated adrenal tumors are pheochromocytomas.

**Sturge-Weber Syndrome**

Sturge-Weber syndrome or encephalotrigeminal angiomatosis is a sporadic disorder characterized by a facial cutaneous vascular nevus within the first and second divisions of the trigeminal nerve and an ipsilateral leptomeningeal angiomatosis involving the parietal and occipital lobes. The cutaneous lesion is a capillary angioma. The leptomeningeal lesion contains thin-walled venous structures confined to the pia mater. Diagnosis is readily made on the basis of focal atrophy and prominent leptomeningeal contrast enhancement (Fig. 5–18). Patchy, parenchymal increased signal intensity is also seen on T1-weighted scans. The T2 changes correspond to gliosis and demyelination, presumably caused by ischemic damage from the overlying angiomatous lesion. The gyriform calcifications seen on computed tomography and plain x-ray film are often not well identified on MRI.

**NORMAL MYELINATION**

Of all radiologic modalities, MRI is the best for the assessment of myelination. MRI provides an excellent evaluation of the progression of normal myelination, delays in myelination, and changes caused by the dysmyelinating diseases. Changes of normal myelination follow a well-documented course on both T1-weighted and T2-weighted scans. In the newborn, the signal intensity relationship between gray and white matter is in general reversed compared with the adult because of the lack of myelination. This pattern is seen on both T1- and T2-weighted scans. On T1-weighted scans, peripheral gray matter is higher signal intensity than underlying white matter, the opposite of the adult pattern. These differences, and the changes that occur with age, are important to consider in clinical scan interpretation.

Myelination begins in the brainstem and progresses to the cerebellum and cerebral hemispheres. The order of myelination is central to peripheral, inferior to superior, and posterior to anterior. T1-weighted scans are particularly useful to assess myelination in the first 9 months of life. With normal myelination, white matter becomes higher signal intensity on T1-weighted scans as a result of increasing cholesterol and protein content. T2-weighted scans are more useful to assess myelination after 6 months of age. The time to repetition (TR) for a T1-weighted scan in the infant, however, needs to be longer than that typically used in adults. A TR of 4000 ms is sufficient. On T2-weighted scans, white matter becomes lower signal intensity as it myelinates. This change is due to the myelin becoming progressively hydrophobic, with lower water content, as it matures. Myelination on T1-weighted scans precedes that on T2-
Figure 5-19. Normal myelination in a neonate. A portion of the posterior limb of the internal capsule is low signal intensity on the T1-weighted scan (A) and high signal intensity on the T2-weighted scan (B) consistent with normal myelination. Peripheral white matter (nonmyelinated) is high signal intensity on the T2-weighted scan and low signal intensity on the T1-weighted scan, the reverse of the normal adult pattern.

weighted scans as a result of the different components evaluated.

In the newborn, the dorsal pons, superior and inferior cerebellar peduncles, posterior limb of the internal capsule, and ventral lateral thalamus demonstrate partial myelination (Fig. 5–19). These structures will have increased signal intensity on T1-weighted scans and decreased signal intensity on T2-weighted scans. The corpus callosum is not yet myelinated and will be very thin.

At 6 months of age, the cerebellum, posterior limb and genu of the internal capsule, occipital lobe, and posterior centrum semiovale are normally myelinated on T1-weighted scans (Fig. 5–20). The corpus callosum is still thin but now partially myelinated (high signal intensity on T1-weighted scans). The genu myelinates slightly later than the splenium at 8 months of age as opposed to 6 months. On T2-weighted scans, only the posterior limb of the internal capsule demonstrates low signal intensity, indicative of myelination.

At 1 year of age, the adult pattern of myelination (for deep and peripheral white matter) is present on T1-weighted scans. Peripheral arborization continues up to 2 years of age, with visual thinning of the gray matter mantle. At 1 year of age on T2-weighted scans, the deep white matter (internal capsule, corpus callosum, and corona radiata) will appear mature, with low signal intensity (Fig. 5–21). However, the white matter of the frontal, temporal, parietal, and occipital lobes as well as the peripheral (subcortical) white matter will not appear mature. These structures will be isointense to gray matter on T2-weighted scans.

At 2 years of age, deep and superficial white matter of the frontal, temporal, parietal, and occipital lobes are low signal intensity, like the adult, on T2-weighted scans (Fig. 5–22). The signal intensity of these white matter structures may persist up to 10 years of age. Histologically, however, myelination proceeds into late adolescence.

In addition to myelination patterns, other features of immaturity can be observed. The relative ventricular size and width of the extra-axial cerebral spaces may appear differently in the neonate and in the older child. The normal ventricle-to-brain ratio in the neonatal period, as measured at the frontal horns, should be approximately one third of the width of the brain. The extra-axial spaces are less variable. The normal width over the convexities should be 4 mm.

Dysmyelinating Disease

Dysmyelination is defined as the improper laying down or subsequent breakdown of myelin. The dysmyelinating diseases are genetically determined and appear early in life. MRI is extremely sensitive to white matter disease of all types. Thus, it is the imaging modality of choice for the diagnosis and evaluation of the dysmyelinating diseases. Although not indicated in every patient, contrast enhancement provides definition of areas of active demyelination on the basis of focal blood-brain barrier disruption.

Adrenoleukodystrophy

Patients with adrenoleukodystrophy present with adrenal insufficiency and progressive multifocal demyelination. Although there are many subtypes, childhood adrenoleukodystrophy is the most common. For this specific disease, the defective gene is located in the Xq28 region of X chromosome. There is impaired degradation of saturated very long chain fatty acids. The disease onset is from 5 to 14 years of age, with rapid neurologic deterioration. In most instances, at presentation, there will be involvement of the splenium of the corpus callosum, the fornix, and the parieto-occipital white matter. These structures will have abnormal low signal intensity on T1-weighted scans and abnormal high signal intensity on T2-weighted scans (Fig. 5–23). On postcontrast scans,
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**FIGURE 5–20.** Normal myelination at 6 months of age. Although the corpus callosum is thin, it is of increased signal intensity on the sagittal T₁-weighted scan (A), indicating that it is myelinated. The posterior limb of the internal capsule is low signal intensity, consistent with normal myelination, on the axial T₁-weighted scan (B). The genu and posterior limb of the internal capsule are high signal intensity on the axial T₁-weighted scan (C). The periventricular and occipital white matter are also high signal intensity (consistent with normal myelination) on C, whereas the frontal white matter is close in signal intensity to that of gray matter. The periventricular and occipital white matter still have immature signal intensity on the T₂-weighted scan. At a higher level with T₁-weighting (D), it is evident that myelination has progressed further in the posterior portion of the centrum semiovale than the anterior.

**FIGURE 5–21.** Normal myelination at 1 year of age. On the T₂-weighted scan (A), the deep white matter (internal capsule, corpus callosum) appears normally myelinated, with low signal intensity. Peripheral subcortical white matter is not yet mature (as judged by the T₂-weighted scan), having signal intensity isointense to gray matter. The T₁-weighted scan (B) looks much like that of an adult, with high signal intensity in both deep and peripheral white matter.
FIGURE 5–22. Normal myelination at 2 years of age. Deep and peripheral white matter have low signal intensity on T2-weighted scans (A–C) consistent with normal myelinization. The signal intensity of white matter surrounding the ventricular trigones is not as low, reflecting terminal myelinization.

FIGURE 5–23. Adrenoleukodystrophy. The patient is a 15-year-old with impaired vision, hearing loss, and intellectual decline. A and B, Sagittal T1-weighted scans reveal abnormal hypointensity in the splenium of the corpus callosum and parieto-occipital white matter. C and D, Axial T2-weighted scans demonstrate abnormal hyperintensity in the corresponding areas.
mild enhancement may be noted along the anterior margin (leading edge) of the involved white matter. If followed temporally, the disease can be seen to progress in anatomic involvement from posterior to anterior. Atypical patterns of white matter disease also occur, with frontal, cerebellar, and asymmetric involvement described.

**Canavan's Disease**

Canavan's disease presents clinically during the first 6 months of life. There is macrocephaly, which is helpful in differential diagnosis. The only other leukodystrophy with macrocephaly is Alexander's disease. Other clinical findings include hypotonia, developmental regression, and cortical blindness. Canavan's disease is autosomal recessive; enzyme tests reveal a deficiency of aspartoacylase. Imaging findings include cortical atrophy, ventriculomegaly, and symmetric abnormal white matter (Fig. 5–24). These findings are not specific for Canavan's disease. Diffuse abnormal white matter, with high signal intensity on T₂-weighted scans, is seen in most of the leukodystrophies.

**Leigh's Disease**

Leigh's disease presents in the first few years of life. Clinical findings include feeding difficulties, psychomotor retardation, and visual disturbances. Leigh's disease is autosomal recessive; tests reveal cerebral inhibition of adenosine triphosphate-thiamine pyrophosphate phosphoryl transferase. Imaging findings include abnormal high signal intensity on T₂-weighted scans in the spinal cord, brainstem, basal ganglia (putamen), and optic pathways (Fig. 5–25).

**Hurler's Disease**

Hurler's disease is the most common of the mucopolysaccharidoses. Hunter's syndrome, or type II, is the second most common. Both are lysosomal storage diseases; this group of congenital enzyme deficiencies includes two main types: the sphingolipidoses and the mucopolysaccharidoses. The sphingolipidoses include the gangliosidoses, including Tay-Sachs disease, as well as Krabbe's disease, Fabry's disease, Gaucher's disease, Niemann-Pick disease, and Farber's disease. The mucopolysaccharidoses include Hurler's disease, Hunter's syndrome, Sanfilippo's syndrome, Morquio's syndrome, Scheie's syndrome, and Maroteaux-Lamy syndrome. The mucopolysaccharidoses all display coarse facial features (“gargoylism”) and have both skeletal and multiple organ involvement.

Patients with Hurler's disease present clinically with mental retardation, deafness, short stature, corneal clouding, and coarse facial features. Death is usually by the teenage years. Enzyme tests in Hurler's disease reveal a deficiency of alpha-L-iduronidase. Imaging findings include ventriculomegaly, cerebral atrophy, a J-shaped sella, cavitated white matter lesions, and diffuse

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**FIGURE 5–24.** Canavan’s disease. The patient is a 4-year-old child who is blind and has macrocephaly, progressive weakness, and severe learning disabilities. Sagittal T₁- (A), axial T₁- (B), and axial T₁-weighted (C) images reveal cortical atrophy and ventriculomegaly. Anteriorly, the central white matter may be of normal signal intensity. However, both posteriorly and peripherally, the signal intensity of white matter is abnormal for age (hyperintense on T₂- and hypointense on T₁-weighted scans relative to gray matter).
Figure 5–25. Leigh’s disease. On T₂-weighted scans, there is abnormal hyperintensity in both the brainstem (A) and the optic radiations (B) (adjacent to the ventricular trigones). The corresponding T₁-weighted scans (not shown) were normal.

Figure 5–26. Hurler’s disease. This 2-year-old child demonstrates diffuse abnormal white matter hyperintensity on the T₂-weighted scan (A). Best demonstrated on the T₁-weighted scan (B) are numerous small holes, principally in white matter, containing cerebrospinal fluid. Moderate ventriculomegaly is also noted.

Figure 5–27. GM₁ gangliosidosis. This 11-month-old infant has diffuse abnormal white matter hyperintensity on the T₂-weighted scan (A). The T₁-weighted scan (B) is also grossly abnormal, with diffuse white matter hypointensity. The pattern of involvement is nonspecific, other than suggesting an inherited metabolic storage disease. There is normal myelination, by signal intensity, of only the posterior limb of the internal capsule.
white matter high signal intensity on T2-weighted scans (Fig. 5–26).

**Other Inherited Metabolic Storage Diseases**
The inherited metabolic storage diseases share a common imaging appearance, particularly in end-stage disease. In most instances, there is cerebral atrophy and diffuse white matter abnormality (Fig. 5–27). The appearance of adrenoleukodystrophy and Leigh’s disease can be distinct. Otherwise, however, MRI is not able to differentiate between the many types of dysmyelinating disease.