25. Other Congenital Malformations

Malformations of the corpus callosum occur in complete or partial forms. Callosal formation proceeds from anterior to posterior, and thus in partial agenesis the anterior portion is typically preserved, as seen in Figure 25.1 A (although, in this instance, only a very small part of the corpus callosum is present). Note the characteristic radiating sulcal pattern extending to the ventricular margin. In neonates, in whom the corpus callosum can be difficult to visualize, radiating gyri may be diagnostic of agenesis. Mega cisterna magna (Fig. 25.1 A), Chiari type 2, and Dandy-Walker malformations may be seen with agenesis. Parallel lateral ventricles (Fig. 25.1 B) are characteristic, and on coronal images, the lateral ventricles may take a crescent-like shape due to the white matter bundles (of Probst) lying along their medial walls.

Holoprosencephaly is a congenital malformation in which the prosencephalon (forebrain) fails to develop into two hemispheres. Lobar, semilobar, and alobar forms are described, which vary in degree from mild to severe. Alobar (the most severe) forms lack a third ventricle, falx, and interhemispheric fissure. Fused or absent thalami and a horseshoe-shaped monoventricle are characteristic. Figure 25.1 C demonstrates a case of semilobar holoprosencephaly in which the interhemispheric fissure is clearly absent anteriorly (although present posteriorly), and the thalami are mostly fused. A monoventricle divides into atrial but not anterior horns. Lobar holoprosencephaly is least severe and characterized by the absence of the septum pellucidum, a small amount of frontal lobe fusion (inferiorly), and nearly normal thalami, ventricles, and corpus callosum.

Lissencephaly refers to the gross MRI appearance of a “smooth brain” as seen in pachygyria and agyria. Figure 25.1 D demonstrates the rudimentary gyral formation seen in the latter. A cell sparse area (black arrows)—consisting of mainly axons—may be seen as a region of high SI on T2WI between a thin, low SI cortex (representing successfully migrated neurons) and a deep layer of gray matter (where neuronal migration has been disrupted). In Figure 25.1 E smooth, thickened gray matter (white arrows) lines the gyri in a focal case of pachygyria. The sulci in this case are shallow, but more prominent than in agyria. Polymicrogyria may appear grossly similar to pachygyria, but pathologically the cortical gyri are too small and too many. Gray matter lobulation, known as cobblestoning (Fig. 25.1 F, black arrows), is a characteristic MRI finding. The location (involvement of the brain) and appearance are varied.

Schizencephaly is a migrational disorder resulting in a cleft traversing a hemisphere from the cortex to the ventricle. Figure 25.1 G illustrates a case of open lip schizencephaly with wide communication between the ventricle and subarachnoid space. The cleft’s gray matter

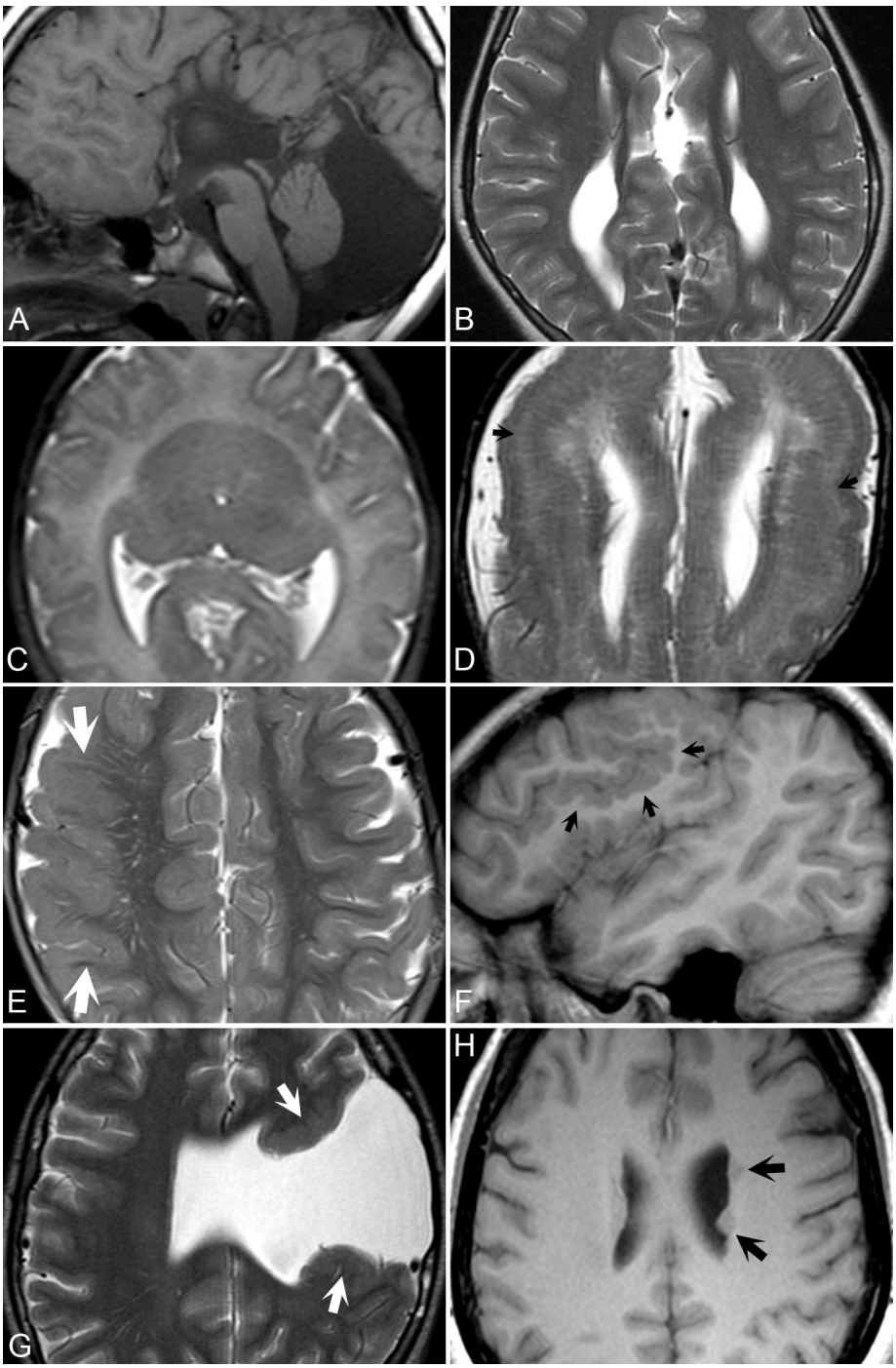

Fig. 25.1

lining (white arrows) defines the abnormality as schizencephaly, rather than porencephaly or other destructive lesions. Closed lip varieties consist of a double layer of cortex between the ventricle and the surface. In a separate patient, two areas of heterotopic gray matter are shown in Figure 25.1 H (black arrows) in their most common location—lining the lateral ventricles. Such foci demonstrate isointensity to gray matter on all sequences, do not

enhance, and are best identified with heavily T1WI where gray and white matter is better differentiated. Septo-optic dysplasia (not illustrated), one last congenital malformation of note, can result in blindness, seizures, hypothalamic-pituitary dysfunction, and growth retardation. The septum pellucidum is dysplastic or absent, and there is optic nerve hypoplasia.