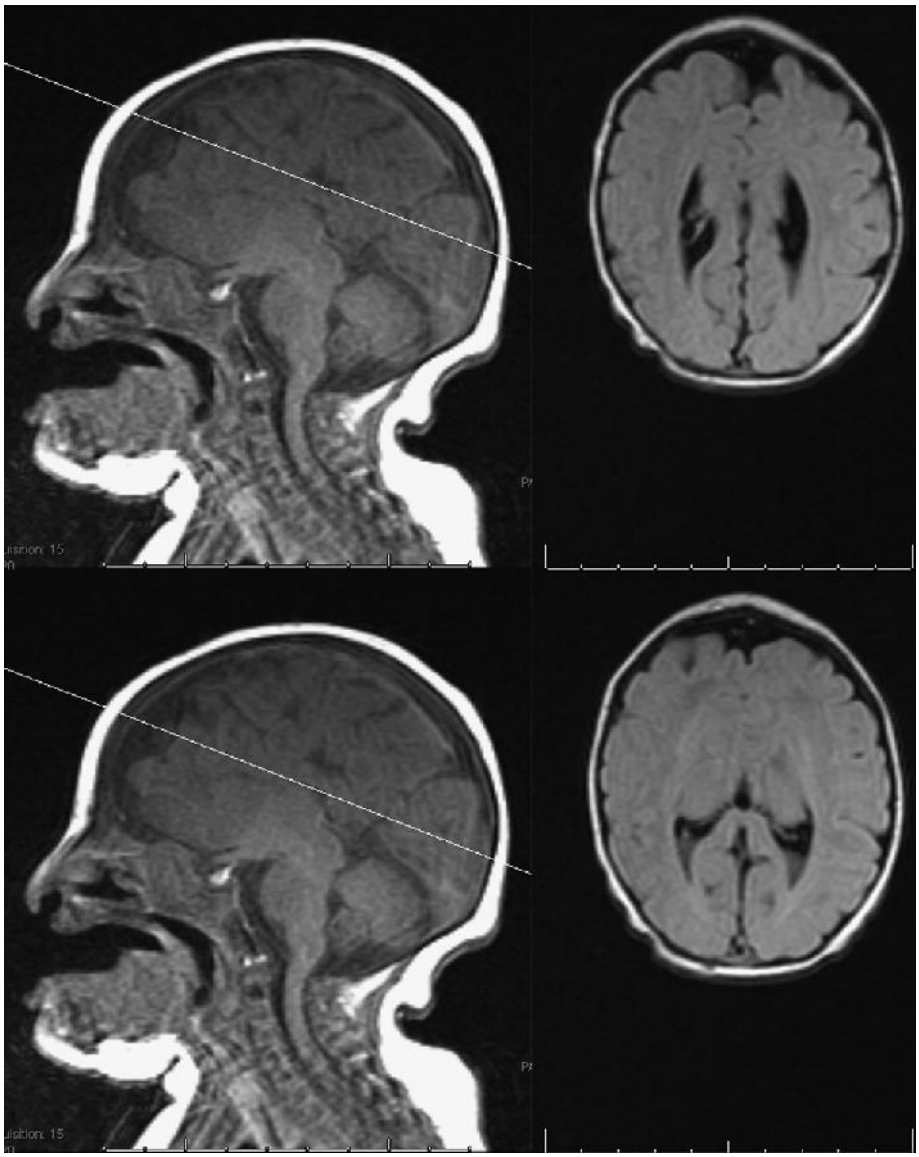

FIGURE 115-1 Semilobar HPE. MRI (sagittal T1 image taken at midline paired with and the axial FLAIR image whose location is indicated by the scout line) of 13 day old with hypotelorism and microcephaly. There is presence of partial fusion of the frontal lobes with lack of interhemispheric fissure/falx and septum pellucidum. The body and genu of corpus callosum is likewise poorly formed. There is appropriate separation of the thalami.

Diagnosis/Differential

Neuroimaging is the primary method of diagnosing HPE, ACC, and SOD. MRI allows for specific anatomic diagnoses and for delineating the full extent of brain malformations. Ophthalmology examination can also detect hypoplastic optic nerves. HPE, ACC, and SOD are associated with a number of genetic syndromes, including trisomies and familial disorders.

Treatment

Surgical treatment can improve associated craniofacial anomalies (e.g., cleft lip) and hormone replacement is needed for pituitary dysfunction (seen in SOD). These patients can present with a wide range of medical problems related to their underlying disabilities, including the development of joint contractures, hip dislocations, impaired swallowing, and respiratory insufficiency.

Prognosis

Survival has improved for these children thanks to aggressive treatment and management of associated medical problems. Long-term outcome depends on the degree of neurologic impairment and associated medical comorbidities.

Disorders of Neuronal Migration and Organization

Definition/Epidemiology

These disorders, including lissencephaly, schizencephaly, polymicrogyria and pachygyria, are caused by disrupted migration of neuronal progenitor cells, resulting in the abnormal appearance of cortical sulci and gyri. The more severe forms of lissencephaly occur in approximately 1/100,000 births. Other migrational disorders have a more variable presentation and, while their true incidence is unknown, they are more common than lissencephaly.