Schizencephaly and Septo optic dysplasia
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History
Newborn with prenatal diagnosis of schizencephaly

Diagnosis
Left open lip schizencephaly, right perisylvian gray matter dysplasia, Septo optic dysplasia

Discussion
Septo optic dysplasia consists of absence of the septum pellucidum and hypoplasia of the optic nerves. Approximately two-thirds will have pituitary dysfunction. Likely the end result of a combination of genetic abnormalities and in utero injuries. The clinical presentation is variable. Detection of optic nerve hypoplasia on MRI can be difficult and is usually best done on sagittal and coronal images. There is a high association of septo optic dysplasia with malformations of cortical development. As in this case, with a schizencephaly and associated contralateral dysplastic, likely polymicrogyric, gray matter.

Schizencephaly is also likely related to a combination of etiologic factors. Imaging with MRI is ideal. Clefts are typically pre and post central in distribution similar to that of polymicrogyria. Open lip type is easier to identify and can be large or small. Closed lip type may be subtle and observation of a slight ventricular outpouching may be a clue that you are dealing with a schizencephaly rather than a transmantle heterotopia. In cases of unilateral schizencephaly, dysplastic cerebral cortex may be seen in a mirror image location in the hemisphere contralateral. Optic nerve hypoplasia is seen in up to one third of patients, as is the case in this patient.

Findings
Large left open lip schizencephaly lined by dysplasic gray matter. Dysplastic gray matter in the adjacent left parietal lobe and in the right perisylvian region. Absence of the septum pellucidum and small optic nerves. Abnormal appearing pituitary gland with absence of the T1 pituitary bright spot.

Reference
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