Septo-Optic Dysplasia with Bilateral Schizencephaly

CASE:
5 month old girl with nystagmus.

FINDINGS:
Septo-optic dysplasia with bilateral schizencephaly (left sided open-lip and right sided closed-lip types).

DISCUSSION:
Septo-optic dysplasia is the term when optic nerve hypoplasia is seen in association with absence of the septum pellucidum. 2/3 of patients have hypothalamic-pituitary dysfunction and ½ have schizencephaly.

Schizencephaly is a rare central nervous system malformation and often presents after birth during an evaluation for developmental delay or seizures. With more widespread use of prenatal ultrasound and MR, the diagnosis is now occasionally being made in utero. There are two main types of Schizencephaly; closed-lip and open-lip:

Closed-lip: characterized by grey matter lined lips that are in contact with each other (type 1).

Open-lip: has separated lips and a cleft of CSF extending to the underlying ventricle (type 2).

Schizencephaly is frequently associated with other migrational anomalies like heterotopic grey matter or cortical dysplasias.

ETIOLOGY:
The association between schizencephaly and septo-optic dysplasia is well known, however the exact etiology is unclear. The embryologic basis for the association is thought to be due to an insult during the late 7th or 8th week of gestation when the germinal matrix, septum, and optic nerve are forming (CMV infection, prenatal drug abuse or abdominal trauma have all been proposed as possible mechanisms).

Schizencephaly is thought to be a primary malformation due to a neuronal migrational anomaly. Familial cases have been reported, as well as associations with heterozygous mutations of the EMX2 gene (although found in only a minority of patients).

IMAGING FINDINGS:
The diagnosis of septo-optic dysplasia is difficult with imaging alone; often times the finding of optic nerve hypoplasia is difficult on MR because of chemical shift artifact obscuring visualization of the nerve – even if apparently normal on MR imaging, clinically apparent optic nerve hypoplasia can be present on ophthalmologic exam.
Schizencephaly is more easily diagnosed on MR and is characterized by a defect lined with gray matter extending from the pial surface to the ventricle wall. The cavum septi pellucidi is absent in 2/3 of patients and the corpus callosum can be focally thinned or absent.

CLINICAL MANIFESTATIONS:
Patients with septo-optic dysplasia may present with seizures, hypothalamic-pituitary dysfunction, bilateral or unilateral blindness or nystagmus.

The clinical manifestations of schizencephaly are extremely variable, and usually consist of motor deficits and mental retardation. Severity of symptoms is based on the size and location of the clefts and associated malformations. Epilepsy is present in ½ of patients and drug resistant in 1/3.

Generally with unilateral defects, there are fewer CNS deficits if the defect is smaller. There can be late onset seizure disorders, and drug resistant epilepsy, but these are usually compatible with long life and most patients do well (most graduated from high school in a small series).

Patients with bilateral defects usually have severe CNS impairment.

REFERENCES:

