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Schizencephaly with septo-optic dysplasia

KEYWORDS: Schizencephaly, septo-optic dysplasia.

Dr Abhijit Pawar	Brig Dr Hariqbal Singh	Dr Manisha Hadgaonkar
Lecturer, Department of Radiology, Shrimati Kashibai Navale Medical College, Pune 411041	Professor and Head of Department, Shrimati Kashibai Navale Medical College, Pune 411041	Lecturer, Department of Radiology, Shrimati Kashibai Navale Medical College, Pune

ABSTRACT *Schizencephaly is an uncommon congenital disorder of cerebral cortical development, defined as a gray matter lined cleft extending from the pial surface to the ventricle. It is known that Schizencephaly and septo-optic dysplasia frequently coexist and that both are associated with absence of the cavum septum pellucidum. This case had findings of both Schizencephaly and septo-optic dysplasia.*

Introduction:
Schizencephaly is an uncommon congenital disorder of cerebral cortical development, defined as a gray matter lined cleft extending from the pial surface to the ventricle. It is known that Schizencephaly and septo-optic dysplasia frequently coexist and that both are associated with absence of the cavum septum pellucidum. This case had findings of both Schizencephaly and septo-optic dysplasia.

Case report:
An eight months old girl child presented with history of no neck holding, no rolling over, generalized tonic clonic convulsions and blindness. Ophthalmic examination revealed bilateral optic nerve atrophy. There was no significant birth history. A clinical diagnosis of cerebral palsy, microcephaly and seizure disorder was made and the patient was subjected to magnetic resonance imaging (MRI) of brain. MRI revealed large, bilateral, fronto-parietal, wedge shaped cerebro-spinal fluid (CSF) clefts (Fig. 1a). These clefts were lined with gray matter (arrows). This was confirmed with TIR (T1 inversion recovery) images (Fig. 1b). The cerebral parenchyma in these regions was deficient. Both lateral ventricles were dilated. The thalami (black arrows) were separated by normal appearing third ventricle (white arrow) (Fig. 2 a). Fourth ventricle, brain stem and cerebellum were normal. A small portion of frontal and temporal lobes was seen bilaterally. Corpus callosum and cavum septum pellucidum were absent (Fig. 2 b). CSF spaces around the bilateral optic nerves (arrows) were prominent indicating optic nerve atrophy (Fig. 5). Based on these MRI findings a diagnosis of open lip schizencephaly with septo-optic dyplasia was made.

Discussion:
Schizencephaly is an uncommon congenital disorder of cerebral cortical development, defined as a gray matter lined cleft extending form the pial surface to the ventricle.[1] Pathologically these clefts are characterized by an infolding of gray matter along the cleft from the cortex into the ventricles and a fusion of the cortical pia and ventricular ependyma within the cleft. [2]

The pathogenesis of schizencephaly has not been firmly established. Barkovich and Norman have proposed an ischemic episode occurring during the seventh week of gestation as the underlying cause of these anomalies. They have hypothesized that a gray-matter-lined cleft can develop secondary to an episode of hypotension, causing infarction of the watershed area. Areas of polymicrogyria and heterotopias commonly seen surrounding the cleft could be secondary to ischemic changes in the less severely affected surrounding areas of germinal matrix.[2] The association of optic nerve hypoplasia with schizencephaly[3] could also be explained in this schema, since the retinal layers and optic nerve fibers also form during the seventh week of gestation. The frequent absence of the septum pellucidum, in both schizencephaly and optic nerve hypoplasia can also be explained on an ischemic basis.[3] The developing septum pellucidum is also a watershed area that is supplied by tenuous transcallosal branches of median artery of the corpus callosum. In summary Schizencephaly represents a disorder of cerebral cortical development that occurs during the time of neuronal migration, mainly due to vascular insult. [1] Other etiologies like genetic, toxic, infective and metabolic have also been proposed. [1]

Irrespective of the cause, schizencephaly presents with CSF containing cleavage in the cerebral hemisphere. The spectrum of cleavage ranges from a thread of CSF connecting the subarachnoid space to the ventricle (closed lip type) to a wide communication between the subarachnoid space and the ventricle (open lip type). The clefts may be unilateral or bilateral and associated with other central nervous system anomalies. The clinical manifestations include varying degrees of developmental delay, motor impairment and seizure disorder. The severity of motor and mental deficiencies depends on the extent of the anatomic defect. Patients with bilateral open lip defects are more severely affected as compared to unilateral closed lip cleft. [4]

On MRI, the clefts are isointense to CSF on all sequences. Other differentials of CSF containing lesions (Table 1) should be considered before making a diagnosis of schizencephaly. The diagnosis of schizencephaly can be made on identifying a gray matter lined CSF cleft. Specific MRI sequences like spoiled gradient (SPGR) and TIR can be employed for confirmation of gray matter lining. Other MRI features of schizencephaly are enlisted in Table 2.

Other important finding in this case was absent cavum septum pellucidum (CSP). Cavum septum pellucidum is an important marker for the normal fetal development.[3] Absence of CSP has been associated with a wide variety of outcomes ranging from devastating to incidental (Table 3). Absence of CSP with bilateral optic nerve atrophy is suggestive of septo-optic dysplasia.

This case had all features of schizencephaly and septo-optic dysplasia. It is important to understand that these two conditions often co-exist.

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Conflict of Interest: None identified.

Fig. 1a

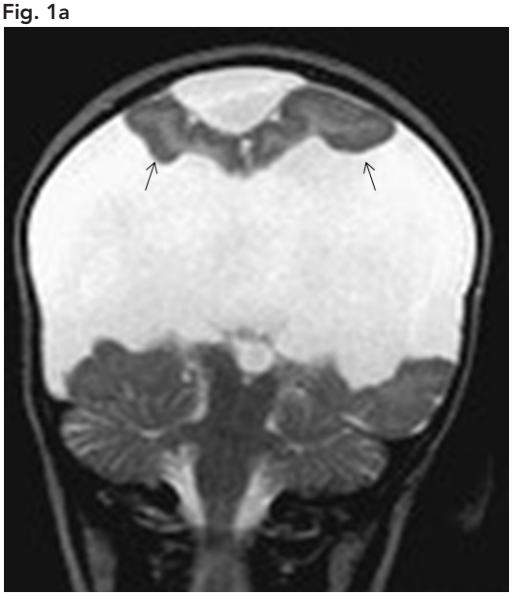


Fig., 1b

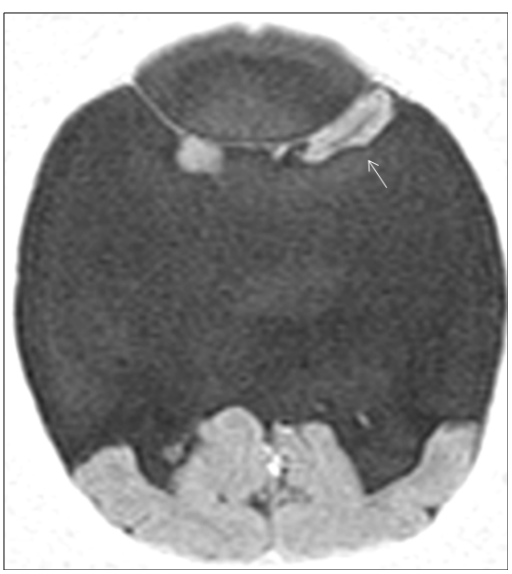


Fig. 2 a

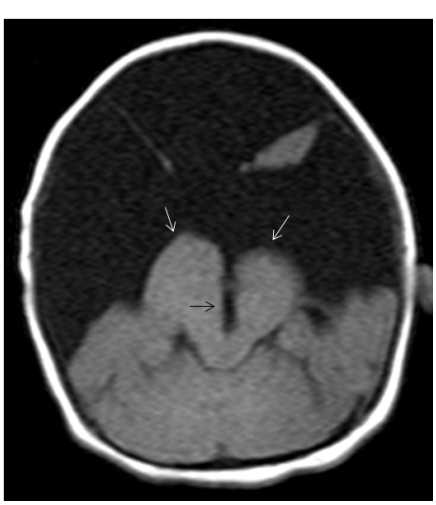


Fig. 2 b

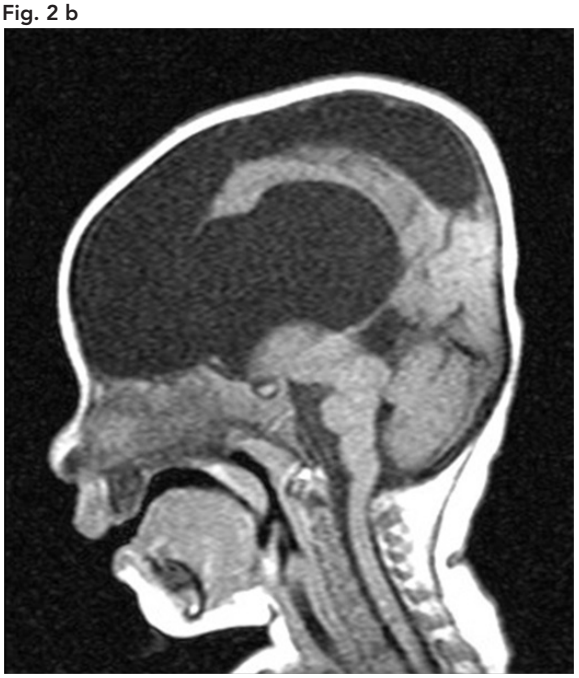
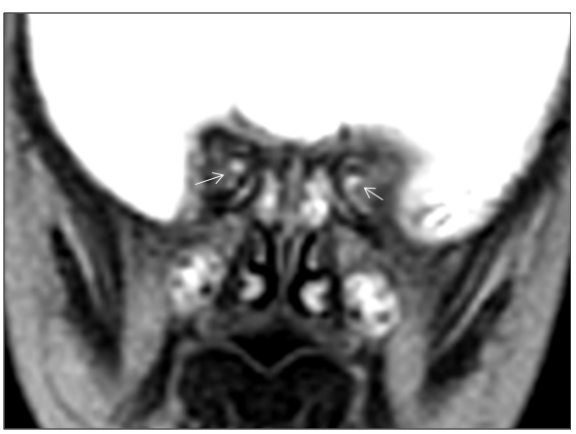


Fig 3



Legends to figures:
Fig 1a: T2 weighted coronal image reveals large bilateral fronto-parietal wedge shaped CSF clefts lined with gray matter (arrow).

Fig 1b: TIR axial image confirms the presence of gray matter lining of clefts (arrow).

Fig 2a: T1 weighted axial image shows bilateral thalami (white arrow) separated by third ventricle (black arrow).

Fig 2b: Corpus callosum is not visualized on this midline sagittal T1 weighted image.

Fig 3: T2 weighted coronal image reveals prominent CSF spaces around the optic nerves indicating optic nerve atrophy.

Table 1:Differental diagnosis of CSF containing abnormalities
Schizencephaly

Ventriculomegaly

Arachnoid cyst

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Holoprosencephaly

Porencephaly

Hydranencephaly

Ventriculomegaly

Table 2 : MRI features in Schizencephaly.
CSF cleft extending from the pial to ependymal surface

Walls of the defect lined with gray matter

Tenting of ventricular wall

Absent cavum septum pellucidum

Absent or hypoplastic corpus callosum

Associated polymicrogyria and heterotopias

Table 3 : Differential diagnosis of absent cavum septum pellucidum (CSP):
Holoprosencephaly

Agenesis of corpus callosum

Aicardi syndrome

Septo-optic dysplasia

Schizencephaly

Porencephaly

Hydranencephaly

Chronic sever hydrocephalus

Basilar encephalocoeles

Isolated septal deficiency

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