Case 16993

Septo-optic dysplasia plus syndrome with associated arachnoid cyst in a toddler presenting with ocular and motor dysfunction

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Imaging Technique: MR
Special Focus: Congenital Case Type: Clinical Cases
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Patient: 1 year, female

Clinical History:

A one-year-old female presented with rotatory nystagmus, generalised hypotonia, and developmental delay. Ophthalmologic examination revealed bilaterally small optic discs, while physical examination showed hyperreflexia in the right lower extremity. A past history of febrile seizure was elicited. Her mother reported of alcohol intake during the first month of pregnancy.

Imaging Findings:

MRI revealed hypoplastic optic nerves and optic chiasm, with the optic nerve diameter measuring 1 mm bilaterally (Figure 1). The pituitary gland and pituitary stalk were also hypoplastic (Figure 2). The septum pellucidum was absent (Figure 3).

A CSF-filled cleft lined by gray matter was seen involving the parasagittal region of the left frontal lobe and body of the corpus callosum and communicating with the body of the left lateral ventricle. This is compatible with an open-lip schizencephaly. (Figure 4)

Heterotopic grey matter nodules were noted along the walls of the lateral ventricles. A linear band of heterotopic grey matter tissues was also seen in the left centrum semiovale. (Figure 5)

An arachnoid cyst was visualized in the anterior aspect of the left middle cranial fossa, causing mild compression of the adjacent left temporal lobe (Figure 6).

Discussion:

Septo-optic dysplasia (SOD), also called de Morsier syndrome, is a rare, clinically heterogeneous condition that consists of the triad of optic nerve hypoplasia, pituitary hypoplasia, and midline brain abnormalities. When concomitant cortical dysplasias are present, the term “septo-optic dysplasia plus syndrome” is used. The most common cortical malformation associated with SOD is schizencephaly [1,2].
SOD has an incidence of 1 in 10,000 births and affects males and females equally [3]. Although the exact cause of septo-optic dysplasia is unknown, frequently cited predisposing factors include embryonic vascular insult, bleeding during the first trimester of pregnancy, primiparity, young maternal age, maternal alcoholism, and drug abuse during pregnancy [1]. Most cases are sporadic, but familial cases have been reported and linked to mutations in developmental genes [1,4].

MRI is considered the imaging modality of choice for diagnosing SOD and SOD plus syndrome. It has been reported to identify optic nerve and optic chiasm hypoplasia with near 100% sensitivity [4]. It is also capable of demonstrating other relevant brain abnormalities, including the absence of septum pellucidum, pituitary hypoplasia, corpus callosum agenesis, schizencephaly, and abnormal grey matter development, as well as possible alternative diagnoses.

In this case, we report a patient with the complete triad of SOD findings on MRI examination. Cortical malformations, particularly schizencephaly and grey matter heterotopia, were also identified. Ocular dysfunction (eg. nystagmus and inability to fixate), developmental delay (eg. delayed motor milestones), and endocrine abnormalities (eg. overweight) were noted on clinical examination. This constellation of findings is compatible with a diagnosis of SOD plus syndrome.

In addition to the classic findings of SOD plus syndrome, an arachnoid cyst was discovered on MRI of our patient. An association with large arachnoid cysts has been reported in up to 12.5% of cases of optic nerve hypoplasia [5], but arachnoid cysts in conjunction with SOD are limited to a few case reports [6,7], even though they are a common associated finding in a variety of other brain malformations.

Managing cases of SOD and SOD plus requires a multidisciplinary team. The radiologist plays a key role in their diagnosis and prognostication by identifying specific abnormalities and their extent on imaging studies. Although there is no cure for the disorder, early diagnosis allows clinicians to promptly address medical issues on development, vision, and endocrine function.

Written informed consent for publication was obtained from the patient’s parent.

**Differential Diagnosis List:** Septo-optic dysplasia plus syndrome, Septo-optic dysplasia, Lobar holoprosencephaly, Corpus callosal dysgenesis

**Final Diagnosis:** Septo-optic dysplasia plus syndrome

**References:**


Figure 1

Description: Hypoplastic optic nerves and chiasm. (a) Axial T2-weighted MR image shows hypoplastic optic nerves, each measuring approximately 1 mm. (b) Axial heavily T2-weighted MR image shows hypoplastic optic chiasm (arrow). Origin: © Department of Radiology, University of the Philippines - Philippine General Hospital / Manila, Philippines 2018.
Description: Hypoplastic optic nerves and chiasm. (a) Axial T2-weighted MR image shows hypoplastic optic nerves, each measuring approximately 1 mm. (b) Axial heavily T2-weighted MR image shows hypoplastic optic chiasm (arrow). Origin: © Department of Radiology, University of the Philippines - Philippine General Hospital / Manila, Philippines 2018.
Figure 2

Description: Hypoplastic pituitary gland. (a) Sagittal T1-weighted MR image shows a hypoplastic pituitary gland, with height of 2.7 mm. (b) Axial heavily T2-weighted MR image demonstrates a hypoplastic pituitary stalk (arrow). Origin: © Department of Radiology, University of the Philippines - Philippine General Hospital / Manila, Philippines 2018.
Description: Hypoplastic pituitary gland. (a) Sagittal T1-weighted MR image shows a hypoplastic pituitary gland, with height of 2.7 mm. (b) Axial heavily T2-weighted MR image demonstrates a hypoplastic pituitary stalk (arrow). Origin: © Department of Radiology, University of the Philippines - Philippine General Hospital / Manila, Philippines 2018.
Description: Absent septum pellucidum. (a) Axial T2-weighted MR image demonstrates absence of the septum pellucidum, with fusion of the lateral ventricles across the midline. (b) Coronal T1-weighted MR image shows “squared-off” appearance of the frontal horns, typically seen with absent septum pellucidum. Origin: © Department of Radiology, University of the Philippines - Philippine General Hospital / Manila, Philippines 2018.
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Description: Open-lip schizencephaly. (a) Coronal T2-weight MR image shows a gray matter-lined cleft involving the parasagittal region of the left frontal lobe (arrow), and communicating with the body of the left lateral ventricle. (b) Sagittal T1-weighted MR image reveals involvement of the body of the corpus callosum by the CSF-filled cleft (arrow). Origin: © Department of Radiology, University of the Philippines - Philippine General Hospital / Manila, Philippines 2018.
**Description:** Open-lip schizencephaly. (a) Coronal T2-weight MR image shows a gray matter-lined cleft involving the parasagittal region of the left frontal lobe (arrow), and communicating with the body of the left lateral ventricle. (b) Sagittal T1-weighted MR image reveals involvement of the body of the corpus callosum by the CSF-filled cleft (arrow). **Origin:** © Department of Radiology, University of the Philippines - Philippine General Hospital / Manila, Philippines 2018.
Description: Subependymal and periventricular gray matter heterotopia. Axial T1-weighted MR image shows a small nodule along the wall of the right lateral ventricle (white arrow). It exhibits same signal intensity as gray matter. In the same image, a linear band of gray matter tissues is also seen in the left frontal and parietal white matter region (black arrow). Origin: © Department of Radiology, University of the Philippines - Philippine General Hospital / Manila, Philippines 2018.
Description: Arachnoid cyst. (a) Axial T2-weighted MR image reveals an extraaxial cystic lesion in the left middle cranial fossa, compressing the adjacent left temporal lobe parenchyma. (b) Axial FLAIR image shows suppression of the lesion’s signal intensity, following CSF signal pattern. Origin: © Department of Radiology, University of the Philippines - Philippine General Hospital / Manila, Philippines 2018.
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