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Images in Medicine

Septo-optic dysplasia: Magnetic Resonance Imaging findings

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ABSTRACT

Septo-optic dysplasia is a rare congenital disorder involving brain and optic pathways. We present typical Magnetic Resonance Imaging (MRI) findings of a case of Septo-optic dysplasia in a 19 year old female patient.

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Introduction

Septo-optic dysplasia (SOD) as the name implies means dysplasia involving septum pellucidum and optic nerves. It may also be associated with hypothalamic-pituitary dysfunction and schizencephaly. Most patients of SOD present with impaired vision and seizures. MRI imaging findings are typical and are diagnostic of the condition. We present here a case of septo-optic dysplasia.

Clinical and imaging findings

A 19 year old female patient presented with diminished vision in right eye since childhood. Her visual acuity in right eye was

5/60 and in left eye was 6/6. Fundus examination revealed optic disc atrophy on right side. Relative afferent pupillary defect was noted in the right eye. Exotropia of 30° right eye. Visual evoked potential was suggestive of right anterior visual pathway dysfunction. There was no nystagmus or history of seizure. General examination revealed averagely built and nourished individual with normal stature for age. There was no evidence of hypopituitarism clinically. MRI examination revealed complete absence of septum pellucidum with box like lateral ventricles, bilateral frontal horns appeared “squared off” with distinct inferior pointing (Fig 1a and b). The right optic nerve was hypoplastic with reduced caliber, however the optic chiasm was normal (Fig 2). The pituitary gland and bilateral olfactory bulbs were normal (Fig 3a and b). Based on the MRI findings, diagnosis of septo-optic dysplasia was made.

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Fig. 1 – (a) Axial T2W image showing absent septum pellucidum with box like appearance of lateral ventricles. (b) Coronal T2W image showing squared off frontal horns with inferior pointing of the ventricles (white arrows).

Discussion

Septo optic dysplasia was first described by a Swiss neurologist George de Morsier in 1956.¹ It was Hoyt who described its association with pituitary in 1978.² It has an estimated prevalence of 1 in 50,000 births. Both sexes are equally affected. Most cases are sporadic in occurrence. A

few risk factors like maternal diabetes mellitus, cytomegalovirus infection, drug ingestion such as quinidine, antiepileptics, and substance abuse may have a role in its occurrence.³

The clinical presentation is variable depending upon the type and extent of associated anomalies. Most cases are detected in infancy. However less severely affected cases may be detected in adolescents and adults as happened in our case that had not undergone any imaging since childhood. SOD presents as visual pathway abnormalities and hypothalamic-pituitary dysfunction which may manifest clinically as short stature, visual impairment, nystagmus and endocrine dysfunction.⁴

Cross sectional imaging plays a major role in the diagnosis of SOD. CT scan may show absent septum pellucidum and large ventricles. MRI is the modality of choice as it not only depicts abnormalities of lateral ventricles but also the anomalies of optic pathway and hypothalamic-pituitary axis better than CT scan. Typically MRI shows absent septum pellucidum and box like lateral ventricles which can be seen well on axial image (Fig 1a), bilateral frontal horns appear squared off with inferior pointing on coronal image (Fig. 1b) as were seen in our case. Abnormalities of optic nerve and chiasm are well depicted in axial and coronal image as was seen in this case (Fig 2). SOD may also be associated with other anomalies of the brain such as hypoplastic pituitary stalk/gland or schizencephaly.⁴ If SOD is associated with absent olfactory bulbs, the condition is known as Kallman syndrome. The MRI findings are diagnostic of this condition and hence require no further workup for establishing a diagnosis.

Differential diagnosis of SOD includes optic infundibular dysplasia where the septum pellucidum is present. Lobar holoprosencephaly, also considered by many as a spectrum of

Fig. 2 – Axial T1W image showing hypoplastic right optic nerve (white arrow).

Fig. 3 – (a) T1W sagittal image showing normal pituitary gland and pituitary stalk. (b) T2W Coronal image showing bilateral normal olfactory nerves in olfactory groove (white arrows).

the same disorder⁵ is associated with fusion of the frontal lobes and the fornices.

Conflicts of interest

All authors have none to declare.

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