ORIGINAL RESEARCH PAPER

Radio-Diagnosis

SEPTO-OPTIC DYSPLASIA PLUS SYNDROME : A CASE REPORT

KEY WORDS: septo-optic dysplasia, septum pellucidum, schizencephaly

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ABSTRACT

Septo-optic dysplasia (SOD) is a neurodevelopmental disorder characterised by optic nerve and septum pellucidum dysgenesis as cardinal features. When septo-optic dysplasia occurs with other anomalies such as schizencephaly or callosal dysgenesis, it is called Septo-optic dysplasia plus syndrome. We report a case of 3 year old male patient who presented with global development delay and left sided weakness. Neurological examination revealed left hemiparesis and ophtalmological evaluation revealed right optic disc hypoplasia. The magnetic resonance imaging (MRI) showed absence of the septum pellucidum with right optic nerve and optic chiasma hypotrophy, bilateral open lip schizencephaly and polymicrogyria. On the basis of typical imaging findings, septooptic dysplasia-plus syndrome was implied as diagnosis. Despite major advances in diagnostic modalities including genetic studies, SOD still represent diagnostic challenges due to multifactorial and heterogeneous nature of the disorder. A life long multidisciplinary approach is helpful in management of these patients to optimise their growth and development.

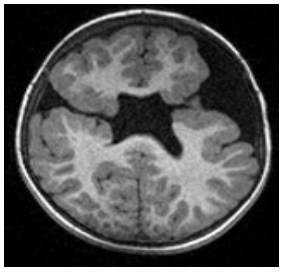
INTRODUCTION

Septo-optic dysplasia (SOD) syndrome is a rare embryological developmental disorder of brain related to midline malformations occuring usually in 6th-7th week of embryogenesis. It was first described by de Morsier in 1956, hence, also known as de Morsier disease. It is also considered as a very well-differentiated form of lobar holoprosencephaly by some authors. Risk factors include maternal diabetes mellitus, alcohol consumption, Cytomegalovirus infection, drug intake such as antiepileptics and substance abuse. As the name suggests, two characteristic findings include absence of septum pellucidum and optic nerve dysplasia. Hypothalamic-pituitary dysfunction and schizencephaly may also be associated. When SOD occurs with schizencephaly or other anomalies like callosal dysgenesis , the syndrome is called SOD plus.

CASE PRESENTATION

A 3 years old male child presented in department of pediatrics of our hospital with features of Global Developmental Delay including delayed milestones and inability to walk and talk. Weakness of left upper and left lower limb was observed by parents from the age of 3 months. No bowel or bladder abnormality was mentioned. Birth history included normal term vaginal delivery at home with normal cry at birth. No antenatal investigations were done. Maternal history included episodes of fever during first trimester, however, no records were made available about the same. Physical examination showed normal head circumference. Neurological examination revealed left hemiparesis and non-photoreactive right pupil. Right optic disc atrophy was observed on ophthalmological examination. Endocrinological tests revealed no significant abnormality. MRI brain (with gadolinium) was performed using "Siemens Magnetom Symphony" 1.5 Tesla Helium cooled superconducting MR scanner, using dedicated protocol in various orthogonal planes, and following observations were made:

- Complete absence of septum pellucidum in midline. (Figure1A)
- Bilateral open lip schizencephaly extending as CSF cleft reaching upto body of lateral ventricle in bilateral frontal region. (Figure1A)
- Vertically oriented bilateral hippocampi, however, showing normal signal intensity. (Figure1B)
- Polymicrogyria in bilateral frontal region and left frontal lobe volume loss. (Figure2A)
- Focal thinning of Corpus callosum in body region. (Figure2B)
- Hypoplastic right optic nerve and optic chiasma. (Figure3)



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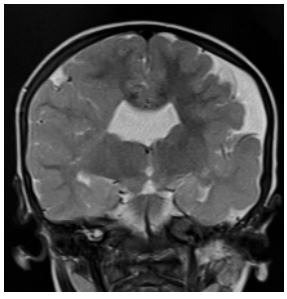

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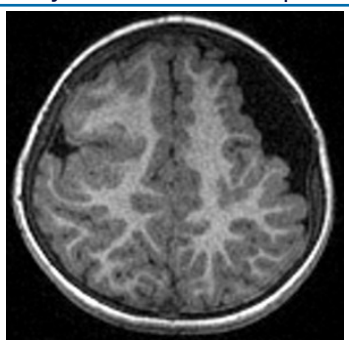
FIGURE 1: AXIAL MPRAGE IMAGE(A) SHOWING ABSENT SEPTUM PELLUCIDUM IN MIDLINE AND B/L OPEN LIP SCHIZENCEPHALY. CORONAL T2 WEIGHTED IMAGE (B) SHOWING VERTICALLY ORIENTED BILATERAL HIPPOCAMPI

A.



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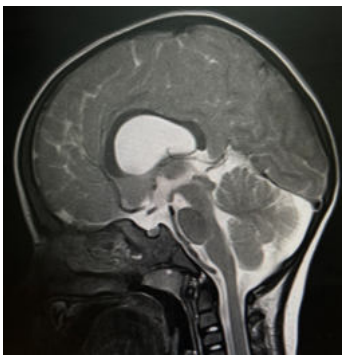
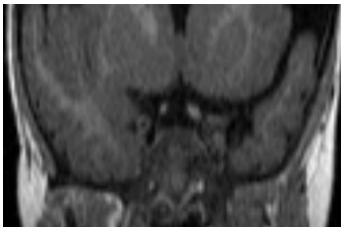


FIGURE 2: A. AXIAL MPRAGE IMAGE SHOWING POLYMICROGYRIA. B.SAGITTAL T2 WEIGHTED IMAGE SHOWING CORPUS CALLOSAL THINNING



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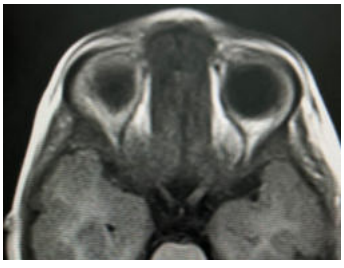

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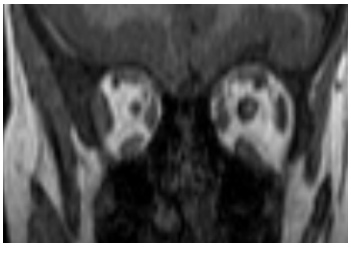

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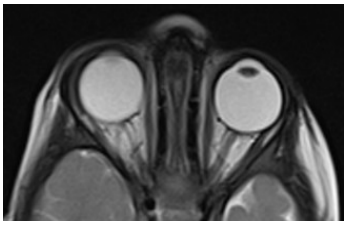

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FIGURE 3. CORONAL T1(A) AND AXIAL T2(B) WEIGHTED IMAGES SHOWING OPTIC CHIASMA HYPOPLASIA. CORONAL T1 WEIGHTED INAGES (C AND D) SHOWING HYPOPLASTIC RIGHT OPTIC NERVE

DISCUSSION

Septo-optic dysplasia (SOD) is a highly heterogeneous syndrome primarily characterised by optic nerve and septum pellucidum dysgenesis. When SOD is associated with other anomalies such as schizencephaly or callosal dysgenesis, the syndrome is called SOD plus. Most cases of SOD are sporadic with no sex predilection. Familial cases are linked to HESX-1 gene mutation, SOX3 duplication and mutations of both SOX2 and SOX3. There are three major constituents forming a triad, septum pellucidum or corpus callosum dysgenesis, optic nerve hypoplasia, and endocrine imbalance secondary to hypothalamic-pituitary axis dysfunction. At least two out of three must be present for formulating the diagnosis. Two out of these three criteria for SOD are evident on imaging and are easy to distinguish.

Clinical presentation may vary depending on the type and extent of associated anomalies. Most common clinical feature of SOD is visual impairment and neurodevelopmental delay. Seizures and intellectual delay are the dominant manifestations. Hormonal dysfuction can occur like pituitary insufficiency, growth hormone deficiency, hypothyroidism and rarely, precocious puberty.

Cross sectional imaging plays a major role in the diagnosis, MRI being the investigation of choice. It is one of those disorders where the role of imaging is essential for a correct diagnosis. The absence of the septum pellucidum is the main finding that directs toward making the precise diagnosis. Other imaging findings include, 'squared off' or box like frontal horns of lateral ventricles with inferior pointing, hypoplastic optic chiasma and optic nerves, cortical development anomalies like heterotopias/ polymicrogyria/ schizencephaly, small pituitary gland with thin or absent stalk, olfactory tract/bulb hypoplasia and incomplete rotation of hippocampus. These MRI findings are diagnostic of the condition.

Differential diagnosis include lobar holoprosencephaly and optic infundibular dysplasia.

CONCLUSION

Septo-optic dysplasia is a rare neurodevelopmental disorder with variable presentation. MRI is the imaging modality of choice which shows dysgenesis of the corpus callosum and/or septum pellucidum and optic nerve hypoplasia as the key features. A lifelong multidisciplinary approach is crucial in management of these patients to optimise their growth and development and help them lead as normal life as possible.

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