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A rare case of septo-optic dysplasia

Meenakumari S[1,*], Shaji CV[2], Prasanth SR[3], Nikhil Gladson[4]

[1,3,4]Senior Resident, [2]Professor & HOD, Dept. of Neurology,

***Corresponding Author:**
Email: drmeenakumaris@gmail.com

Introduction

Septo-Optic Dysplasia, also referred to as de Morsier' s syndrome, is a rare condition characterized by Optic nerve hypoplasia, Pituitary abnormalities, and Midline brain defects (involving primarily the septum pellucidum and, at times, the corpus callosum).[(1)] The diagnosis is typically made when two of the three features are present. The term Septo-Optic dysplasia (SOD) was coined in 1956 by the Swiss neurologist, de Morsier, who pointed out the association of optic nerve hypoplasia with an absence of the septum pellucidum.

Case Report

A 11-year-old male child presented to us with recurrent episodes of seizures. He was the first child of a non consanguinous parentage. His mother was 21 years at conception, and had no comorbidities or addictions, and the antenatal period was uneventful. He was born out of a full term vaginal delivery. On the first post natal day, baby developed hypoglycemia and seizures, and was admitted in the Neonatal Care Unit. At discharge, baby was active and had no sucking difficulty. At six months of age, mother noted that his vision was defective and he had jerky movements of both eyes. After Ophthalmology work up, relatives were informed that he had gross defect in vision in both eyes and that it was not rectifiable. At the time of presentation, he was able to communicate well, and could write a few words and could perform simple additions. He was attending regular school, though he had visual impairment and learning difficulty. Over the past six months, he had six episodes of focal onset seizures with secondary generalization. There were no systemic symptoms, or history suggestive of neuroregression. There was no family history of epilepsy, developmental delay or visual impairment.

On examination, he was obese, BMI of 33, with brachycephaly and mild facial dysmorphism in the form of receding forehead. His height was below the third centile for his age. There was hypogonadism and his Sexual maturity score was preadolescent. His cognitive functions were near normal. Vision was grossly defective, with bilateral primary optic atrophy and a bidirectional jerky nystagmus. Intraocular pressure measurements were normal. Visual field charting revealed a marked peripheral constriction in both eyes. Other cranial nerves were normal and there was no motor or sensory deficit.

Routine blood investigations were normal. EEG revealed Epileptiform discharges in right temporoparietal leads. Magnetic resonance Imaging of Brain showed bilateral atrophy of optic nerves, Right > Left, optic chiasma and tracts. Septum Pellucidum was absent.[(6)] The imaging findings were suggestive of Septo- optic dysplasia.

We proceeded with Endocrinology work up. Hypothyroidism and Diabetes Insipidus were ruled out. However, the Growth hormone levels were consistently low (0.1 ng/mL, normal value: 1-16.40 ng/mL), even after estimation following Clonidine Stimulation at 5 µg /kg.

The patient was treated with Carbamazepine, on which seizures were controlled. He was referred to Pediatric Endocrinology Department for further evaluation and consideration of Hormone Replacement therapy.

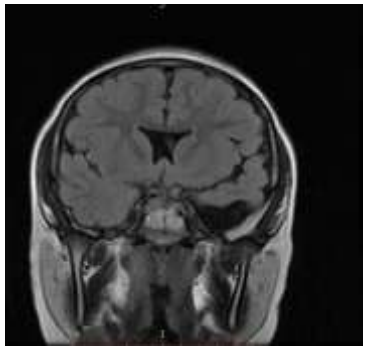

Fig. 1

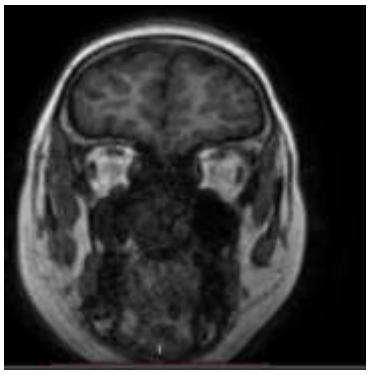

Fig. 2

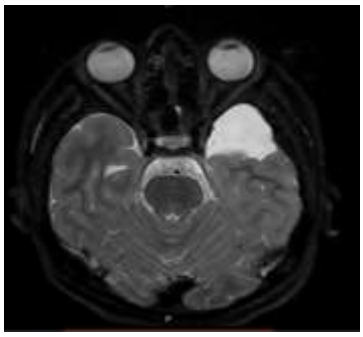

Fig. 3

Discussion

Septo Optic Dysplasia is a rare condition, with an estimated incidence of 10 in 100,000 per year.[2] The causes of this early brain midline dysembryogenesis are unknown, but the most frequently suggested etiologies relate to embryonic vascular insult, usually in the 6th–7th week of embryogenesis, bleeding during the first trimester of pregnancy, primiparity and young maternal age and maternal alcoholism or drug abuse during pregnancy.[3] During fifth and sixth weeks of gestation, ventral specification of neural tube occurs and errors during this stage cause Septo-optic dysplasia and holoprosencephaly.

SOD can present at birth with manifestations of pituitary insufficiency, including hypoglycemia (with seizures), microphallus and undescended testes (from hypogonadotropic hypogonadism), and midline birth defects. Because children are at risk for deficiencies of adrenocorticotropic, thyroid stimulating, and growth hormones, they can present with hypoglycemia, diabetes insipidus, and poor thermoregulation. Multiple case reports of sudden death in SOD patients have described these complications.[5]

Many patients with SOD have additional central nervous system malformations. SOD plus include an assortment of coexisting brain malformations, like polymicrogyria and schizencephaly.[3]

Optic nerve hypoplasia might result in moderate to severe visual loss in children, accounting for about 15 to 25% of infants with serious visual loss.[7] Optic nerve hypoplasia is seen in 25% cases of agenesis of septum pellucidum. Any child presenting with visual impairment and nystagmus should be evaluated for optic nerve involvement,[1] and if ONH is detected, the patient should be assessed for anterior pituitary hormone deficiency. Additionally, many children with SOD can have developmental delay and seizures, and should be evaluated for these possible concerns.[4]

Conclusion

Septo optic dysplasia is a very rare diagnosis. However, it should be kept under consideration in an infant with history of perinatal jaundice, hypoglycemia, seizures, especially when the patient is detected to have poor vision, on subsequent neuro- developmental follow up. The patient reported here had neonatal seizures and hypoglycemia with bilateral optic atrophy and infantile nystagmus. When such is the case scenario, coexistent anterior pituitary malformations and hormone deficiencies should be ruled out. A life-long multidisciplinary approach is crucial to optimize their growth and development and to help them lead as normal a life as possible. Timely hormone replacement can improve cognition and avoid comorbidities like hypothyroidism, Diabetes insipidus, and Growth Hormone deficiency. In some cases, with coexistent adrenal insufficiency, even sudden deaths may thus be prevented.

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