International Journal of Contemporary Pediatrics
Bainade K et al. Int J Contemp Pediatr. 2024 Nov;11(11):1662-1664
http://www.ijpediatrics.com
pISSN 2349-3283 | eISSN 2349-3291

DOI: https://dx.doi.org/10.18203/2349-3291.ijcp20243097

Case Report

Septo-optic dysplasia: a rare case report

Kapil Bainade, Veeranna Kotrashetti, Karan J. Bhoomraddi*, Neelu Elon

Department of Pediatrics, D Y Patil Medical College and Hospital, Nerul, Navi Mumbai, Maharashtra, India

Received: 31 August 2024
Accepted: 02 October 2024

***Correspondence:**
Dr. Karan J. Bhoomraddi,
E-mail: karan.jb.22@gmail.com

ABSTRACT

Septo-optic dysplasia (SOD), earlier referred to as de Morsier syndrome, is an uncommon disorder of development wherein the septum pellucidum—a thin membrane located between the two cerebral hemispheres-as well as the eyes and pituitary gland, are affected. It is a spectrum of disorders that causes optic nerve defects and also affects the optic disc. It is also associated with midline brain anomalies and those of the pituitary gland. Thus, the disease may present as visual problems, reduced tone of the muscles, delays in development, and hormonal imbalances. Clinical examination identifies features related to these anomalies. The diagnosis is confirmed by neuroimaging, or a genetic study. Managing such a case requires a multi-disciplinary approach. Here we present an 8-year-old boy who was diagnosed with septo-optic dysplasia and was appropriately managed.

Keywords: Septo-optic dysplasia, De morsier syndrome, Optic nerve hypoplasia

INTRODUCTION

Septo-optic dysplasia, also referred to as the optic nerve hypoplasia syndrome, or de Morsier syndrome, may be detected due to a clinical observation of nystagmus and visual impairment in infancy. It is a rare condition affecting around 1 in every 10,000 births, with an equal incidence among boys and girls.[1] SOD is diagnosed in the presence of two or more of the following problems: optic nerve hypoplasia, midline brain abnormalities, and pituitary gland abnormalities.[2] Only 33% of cases have all of these 3 features. Due to the proximity of these deformities to the optic chiasm, ocular manifestations are common.[5] These can be unilateral or bilateral. Neuroimaging demonstrates optic nerve anomalies and structural brain anomalies.[3] It can be associated with anterior and/or posterior pituitary hormone deficiencies in up to 75% of the cases. Most of these patients show the triad of a small anterior pituitary gland, an attenuated pituitary stalk, and an ectopic posterior pituitary bright spot. However, hypothalamic dysfunction is considered to be the primary cause of hypopituitarism in this condition.[3] The hormone deficiencies could be either an isolated growth hormone deficiency (IGHD) or multiple pituitary hormone deficiencies (MPHD). IGHD is the more common of the two.[3] The disease has a multifactorial etiology, with interactions between genetic and environmental factors. A few people with SOD may have variations in the HESX1 OTX2, SOX2 or SOX3 genes.[4] However, in most cases, a single gene defect has not been identified and thus it is considered mostly non-inheritable. It has a highly variable phenotypic penetration. SOD can have varied presentations. The affected child may have delayed attainment of developmental milestones. They may also have movement and coordination difficulties due to the midline brain anomaly. The most commonly efficient hormone is the Growth Hormone (GH), which can cause short stature or hypoglycemic episodes. 5% of children develop diabetes insipidus.[1] Reduced cortisol levels may be life-threatening. Low levels of thyroid hormones can have the associated symptoms. Puberty is usually delayed. The affected boys might have micropenis and/or undescended testes. Some cases do have a precocious puberty.[1] There is visual impairment, which could also be accompanied by nystagmus or strabismus. Most cases

have muscle tone abnormalities.[6] In neonates and infants, it can present as hypoglycemia and seizures. Severe cases manifest as birth asphyxia, persistent neonatal jaundice, cryptorchidism, lethargy, poor feeding, and irritability.[1] These tend to progress to cerebral palsy. Sensorineural hearing defects have been reported. Anomalies such as cleft palate and esophageal atresia can also be associated.[4] Many children have sleep disturbances, autistic behaviors, and a tendency to gain weight.[1]

CASE REPORT

Here we present an 8-year-old boy who was brought with complaints of delayed attainment of developmental milestones since, 1 year of age, abnormal gait, and abnormal eye movements since, 4 years of age. He had a speech impediment. There were also complaints of farsightedness. He had a plagiocephalic skull. On physical examination, the child had reduced tone and power of the muscles on the right lower limb. This led to a hemiplegic gait. There was also a weakness in the right upper limb. He had hypermetropia and a nystagmus in both eyes. The child was short in stature, which on further evaluation, was determined to be Familial Short Stature.

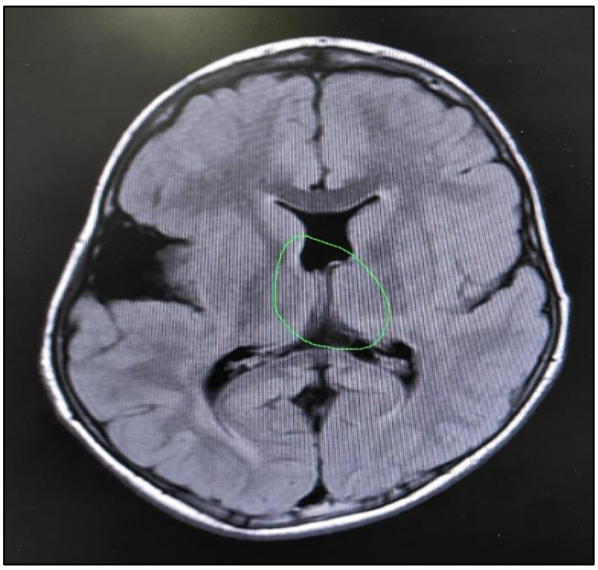


Figure 1: MRI Brain shows the presence of an interhemispheric fissure along the entire midline (encircled), and an arachnoid cyst in the right anterior temporal fossa.

Ophthalmic evaluation also revealed optic disc hypoplasia, as depicted in figures 3 (A and B). Given the above findings, a magnetic resonance imaging (MRI) of the brain was obtained, which revealed an absent septum pellucidum, and the presence of an interhemispheric fissure along the entire midline with complete separation of the thalami. These findings were consistent with septo-optic dysplasia. The MRI also revealed an arachnoid cyst in the right anterior temporal fossa (Figures 1 and 2).

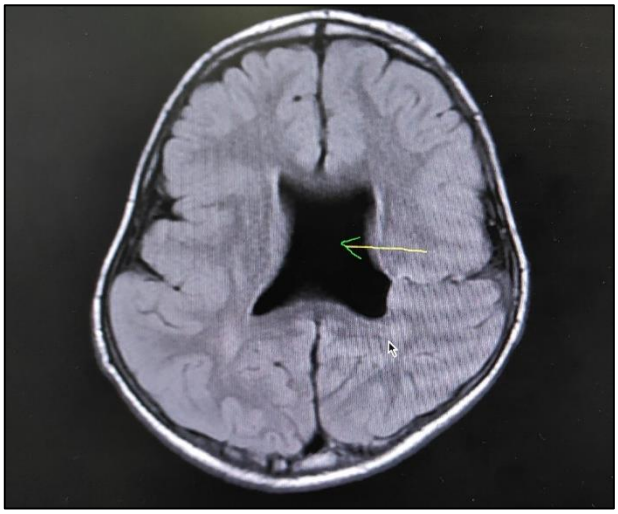
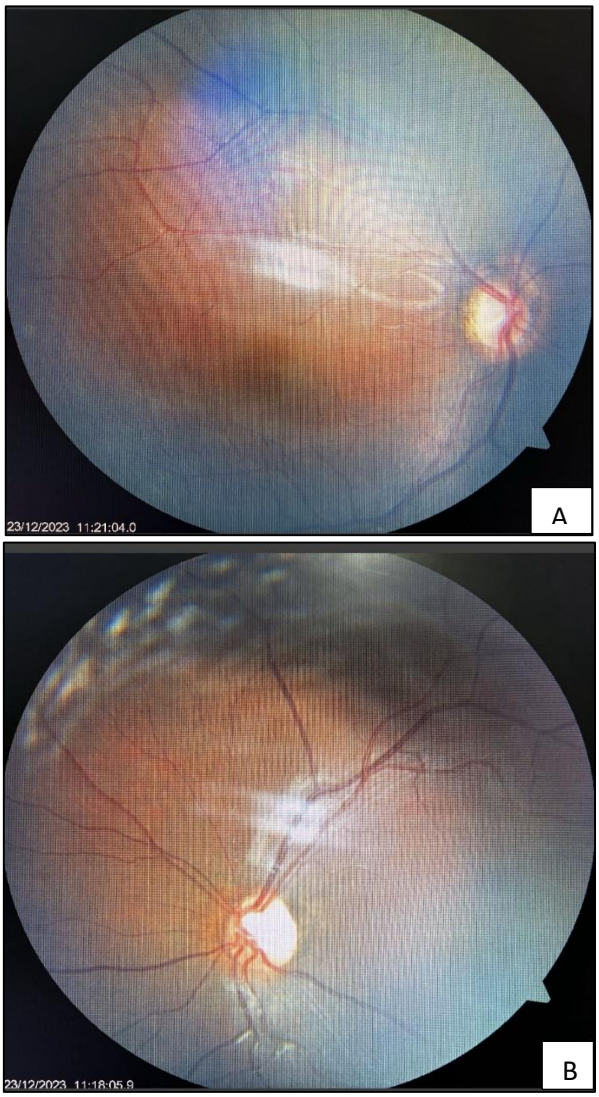


Figure 2: MRI Brain reveals an absent septum pellucidum (arrow).



Figures 3 (A and B): Optic nerve hypoplasia.

Hearing evaluation revealed no abnormalities. A Growth Hormone panel was sent to evaluate pituitary function, which turned out normal. The Thyroid Panel also came out normal. Hence there was no evidence of pituitary dysfunction, which is unusual for a case of Septo-optic Dysplasia. As the child had complaints of paroxysmal events a few months prior resembling a seizure, an electroencephalogram was done, which showed no epileptic spikes/ waves. A whole exome sequencing was done to identify genetic mutations; however, the report revealed no abnormalities. The child was prescribed appropriate spectacles and advised to sit in the front row in his classes at school. He was started on physiotherapy to help with his gait. He was referred to a speech therapist because there were no hearing abnormalities. The parents were also counselled about good nutrition practices to ensure proper growth and development.

DISCUSSION

Even though most cases of SOD present with symptoms of optic nerve hypoplasia (ONH), a review of the literature suggests that visual impairment accounts for just 23% of overall reported symptoms in these patients, which includes very mild cases of astigmatism.[2] Due to variations in presenting complaints, SOD is difficult to diagnose without neuroimaging.[5] Radiological confirmation of ONH necessitates high-resolution imaging and interpretation by an experienced neuro-radiologist.[8] Currently, SOD is not curable; however, many of its symptoms can be managed through a tailored approach, consisting of hormonal replacement, corrective ophthalmological intervention, and neuropsychological support.

Occasionally septo-optic dysplasia is diagnosed during routine prenatal ultrasound scanning, but it is most commonly diagnosed during childhood. It is suspected early in childhood if the child has small male genitalia, poor growth, low blood sugar levels and is prone to infections.[1] If septo-optic dysplasia is suspected, various tests and scans will be needed to confirm or rule out the diagnosis. MRI scans of the brain are used to show the presence and severity of brain abnormalities. Blood tests are used to measure hormone levels. Vision testing is used to measure the severity of the optic nerve hypoplasia. Development assessments are needed to measure developmental delay. MRI is preferred for evaluating CNS abnormalities in patients with optic nerve hypoplasia (ONH). The goal of treatment for septo-optic dysplasia is to address associated symptoms.[7] Because the symptoms can vary greatly from child to child with this condition, treatment is tailored to each patient. Treatment may involve many different specialists, including ophthalmologists, neurologists, endocrinologists and therapists to help the child build strength and ability in areas of weakness.[7] Some children with septo-optic dysplasia have normal intelligence, while others have learning disabilities and developmental delays. Learning problems can be helped with therapy. Vision loss from underdeveloped optic nerves cannot be restored. But in many children, vision may improve somewhat during early childhood. Vision therapy and low vision services should always be considered in patients with reduced acuity who struggle with activities of daily living.[7] Endocrine function should be watched closely in patients with optic nerve hypoplasia. Hormone replacement therapy is effective in addressing endocrine deficiencies caused by an underdeveloped pituitary gland.[7]

CONCLUSION

Septo-optic dysplasia can have a variable presentation. The etiology is multi-factorial. Most cases present during early childhood. Neuroimaging plays a vital role in diagnosis. Even though it is currently incurable, the symptoms can be managed through a multi-disciplinary approach.

Funding: No funding sources
Conflict of interest: None declared
Ethical approval: Not required

REFERENCES

1. Great Ormond Street Hospital. Septo-optic dysplasia. GOSH hospital site. Available at: https://www.gosh.nhs.uk/conditions-and-treat. Accessed on 20th March 2024.
2. Ganau M, Huet S, Syrmos N, Meloni M, Jayamohan J. Neuro-ophthalmological manifestations of septo-optic dysplasia: Current Perspectives</p> Eye And Brain. 2019;11:37-47.
3. Kliegman RM, St Geme JW III. Nelson Textbook of Pediatrics E-Book. Elsevier Health Sciences. 2019. Available at: https://shop.elsevier.com.
4. Septo-optic dysplasia spectrum. About the disease. GARD. 2020. Available at: https://rarediseases.info.nih.gov/diseases. Accessed on 17th March 2024.
5. Cappelli ODR, Tucker CT, Taylor VD. Ocular manifestations of septo-optic dysplasia. Optometric Clinical Practice. 2023;5(1):41.
6. Septo-optic dysplasia. National institute of neurological disorders and stroke. 2021. Available at: https://www.ninds.nih.gov/health-information. Accessed on 17th March 2023.
7. Philadelphia CHO. Septo-optic dysplasia. children's hospital of philadelphia. Available at: https://www.chop.edu/conditions-diseases/septo. Accessed on 28th March 2024.
8. Al-Senawi R, Al-Jabri B, Al-Zuhaibi S, Al-Azri F, Al-Yarubi S, Harikrishna B, et al. Septo-optic dysplasia complex: Clinical and radiological manifestations in Omani children. Oman J Ophthalmol. 2013;6(3):193-8.

Cite this article as: Bainade K, Kotrashetti V, Bhoomraddi KJ, Elon N. Septo-optic dysplasia: a rare case report. Int J Contemp Pediatr 2024;11:1662-4.