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Septo optic dysplasia/De Morsier Syndrome: A Case Report

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Abstract

Septo - Optic Dysplasia (SOD) also known as De Morsier Syndrome, is a rare medical condition, the diagnosis of which is made when there are two or more characteristics of the classic triad. The triad consists of optic nerve hypoplasia, pituitary hormone abnormalities, and midline brain defects. We present the case of a 15-year-old boy with a history of seizures and optic nerve disease that was diagnosed with septo-optic dysplasia with characteristic imaging findings.

Introduction

Septo-optic dysplasia (SOD), also known as "de Morsier's Syndrome," was first reported by George de Morsier, a Swiss neurologist in 1956; described the following triad: hypoplasia/ dysplasia of the optic nerve, hypothalamic-hypophyseal dysfunction, and midline abnormalities (dysgenesis of corpus callosum and/or septum pellucidum) [1]. De Morsier syndrome is rare medical entity, with an estimated incidence of 1 in 10,000 live births [2]. Some authors consider it to be a mild form of lobar holoprosencephaly, although embryologically it may be more appropriately categorized as a disorder of midline development/ congenital dysgenesis of diencephalon with hypoplasia of the optic nerve and hypothalamus- pituitary and midline structural defects [3, 4].

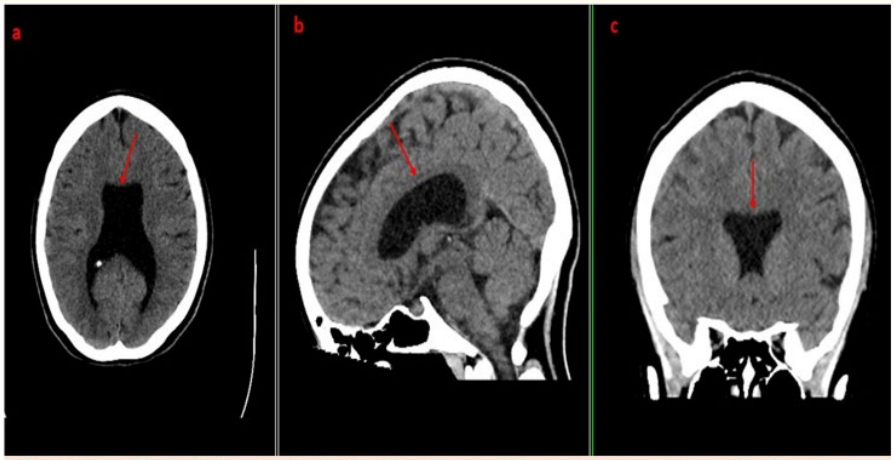
Case Presentation

A 15-year-old male patient was referred to our unit for evaluation of headache and recurrent seizures to rule out space occupying lesion. The patient had generalised tonic-clonic seizures treated initially with valproic acid. Seizures were well-controlled until a year before presentation when the seizure pattern recurred.

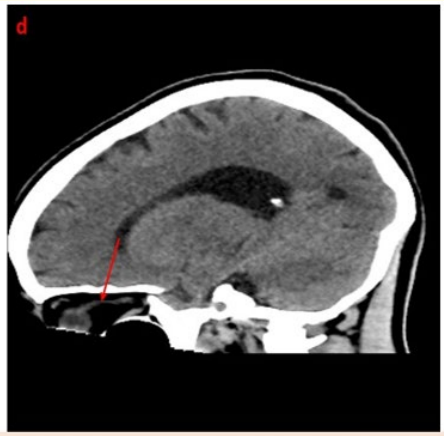
His family history was not significant. His vital signs were within normal limits. He also complained of bilateral diminished visual acuity; ophthalmological examination demonstrating bilateral optic nerve hypoplasia

Non Enhanced CT brain revealed the following findings:

- Absence of septum pellucidum, "mono ventricle appearance" with mild prominence of bilateral lateral ventricles (a, c).
- Hypoplasia of corpus callosum (b).
- Hypoplasia of bilateral optic nerve (d).



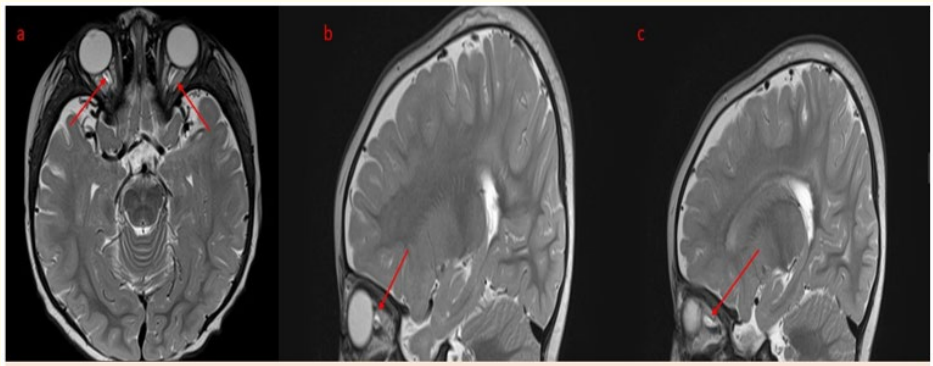
a & c) CT Brain at the level of lateral ventricles {a, axial and c, coronal} demonstrates absent septum pellucidum with "mono ventricle appearance"
b) CT Brain midline sagittal section demonstrates hypoplasia of corpus callosum with mild prominence of lateral ventricle



d) CT brain, Sagittal section at the level of left optic nerve demonstrates hypoplastic left optic nerve (arrow)

Further imaging with MRI Brain and Orbits followed which confirmed the above mentioned CT Findings:

- Absence of septum pellucidum, "mono ventricle appearance" with mild prominence of bilateral lateral ventricles.
- Hypoplasia of corpus callosum.
- Hypoplasia of bilateral optic nerves (involving the intra-orbital, intracanalicular and intracranial segment).



a) T2 weighted MR axial section of the brain at the level of optic nerves demonstrate bilateral optic nerve hypoplasia (arrows)

b & c) T2 weighted MR sagittal section of the brain at the level of right optic nerve demonstrate optic nerve hypoplasia (arrows)

Due to the presence of the characteristic midline abnormalities with hypoplasia of bilateral optic nerves, a diagnosis of SOD/ De Morsiers syndrome was made.

Discussion

While the presentation is variable, the diagnosis of septo-optic dysplasia is defined by two or more of the following features: (i) midline brain abnormalities (including absence of the septum pellucidum and/ or corpus callosum; (ii) hypoplasia of the optic nerves, present in 96% of patients and usually bilateral; and (iii) hypothalamic-pituitary dysfunction, which is seen in up to 60-90% of patients [5, 6]. Our study demonstrated the presence of first two findings.

Conclusion

SOD is a complex disorder with varied clinical manifestations. Differential consideration should include lobar holoprosencephaly [7]. The possibility of septo-optic dysplasia should be raised in children with unilateral or bilateral hypoplasia of the optic nerve [8]. Early diagnosis and multidisciplinary management are crucial for optimizing patient outcomes.

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