CASE REPORT http://doi.org/10.3126/njr.v14i2.75732

Imaging of Septo-Optic Dysplasia with Focal Pachygyria: A Case Report

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Received: September 7, 2024 **Accepted**: February 16, 2025 **Published**: March 7, 2025

Cite this paper:

Kayastha P, Paudel S, Khatiwada A, KC S. Imaging of Septo-Optic Dysplasia with Focal Pachygyria: A Case Report. *Nepalese Journal of Radiology* 2025;14(2):16-9. **http://doi.org/10.3126/njr.v14i2.75732**

ABSTRACT

Septo-optic dysplasia (SOD) is a rare congenital disorder involving the brain and optic pathways. The syndrome is characterized by optic nerve hypoplasia, hypothalamic-pituitary dysfunction, and midline brain defects such as the absence of the septum pellucidum and/or thinning or agenesis of the corpus callosum. Here, we present a case of a 19-year-old female who underwent brain imaging for diminished vision in both eyes since childhood and was diagnosed with SOD. Early clinical suspicion and proper radiological imaging are crucial for the timely diagnosis and management of these patients.

Keywords: *Corpus Callosum; Optic Nerve Hypoplasia; Septum Pellucidum; Septo-Optic Dysplasia*

INTRODUCTION

Septo-optic dysplasia (SOD), also known as de Morsier syndrome, is a rare congenital disorder affecting the brain and optic pathways, with an estimated prevalence of ~1:10,000. The syndrome is defined by at least two of the following three criteria: optic nerve hypoplasia, hypothalamic-pituitary dysfunction, and midline brain defects such as the absence of the septum pellucidum or thinning/agenesis of the corpus callosum. It results from abnormalities in forebrain development during the 4th to 6th weeks of gestation. Although the exact etiology is unknown, both genetic and environmental factors are believed to play a role. Here, we present the case of a 19-year-old female diagnosed with SOD after undergoing an MRI for diminished vision in both eyes since childhood. This report emphasizes the imaging findings and associated conditions with this syndrome.[1,2,3]

CASE REPORT

A 19-year-old female presented with diminished vision in both eyes since childhood. Her visual acuity was 4/60 in both eyes. Fundus examination revealed bilateral optic disc atrophy. There was no history of seizures or nystagmus, and her stature and clinical evaluation for hypopituitarism were normal. She had no significant family history, and laboratory tests were unremarkable.

A computed tomography (CT) scan of the brain (Figure 1) revealed an absent septum pellucidum with hypoplastic optic nerves. There was inferior pointing of bilateral lateral ventricular frontal horns in the coronal images. Magnetic resonance

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imaging (MRI) (Figure 2) confirmed these findings, showing a right optic nerve measuring 1.9 mm and a left optic nerve measuring 1.8 mm in diameter at the mid-intraorbital segments. Additionally, it revealed focal gyral thickening with reduced sulci in the bilateral frontal cortices, suggestive of focal pachygyria. The optic chiasma was slightly atrophied measuring a width of 10 mm. The pituitary gland, stalk, and corpus callosum were normal. Bilateral globes were normal in size, morphology, and signal intensity. Based on the imaging findings, a diagnosis of SOD with focal pachygyria was made.

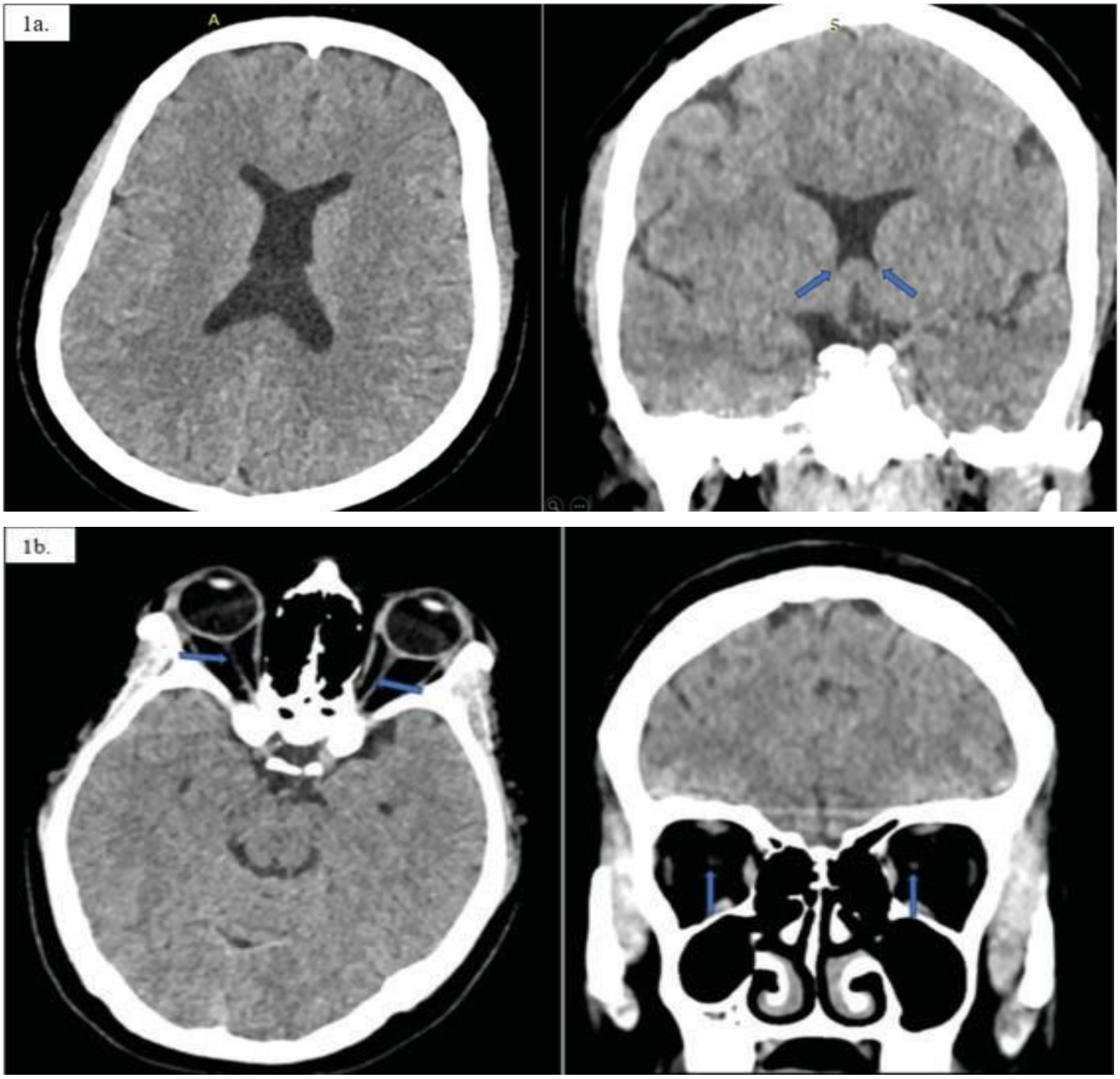

***Figure 1: 1a**. Axial and coronal CT images showing the absence of the septum pellucidum with the inferior pointing of the frontal horns of the lateral ventricle in the coronal view (blue arrows). **1b.** Axial and coronal CT scan images demonstrating hypoplastic bilateral optic nerves (blue arrows)*

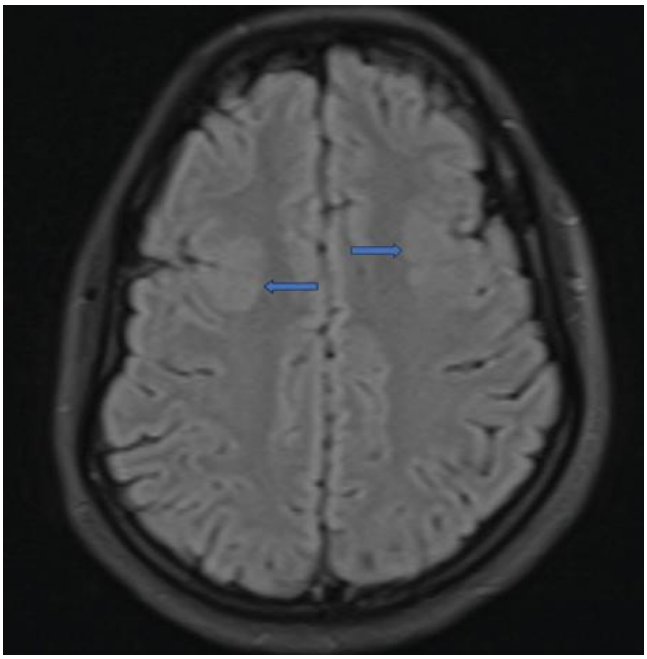

Figure 2: *FLAIR axial MRI image showing focal gyral thickening with a paucity of sulci in the bilateral frontal cortices, suggestive of focal pachygyria (blue arrows)*

DISCUSSION

The clinical presentation of SOD varies widely; some individuals show symptoms at birth, while others develop symptoms during childhood. While optic nerve hypoplasia is common, unilateral optic nerve atrophy may also occur. SOD may present with visual impairment, endocrine dysfunction (including growth hormone deficiency), and neurological issues such as developmental delay, intellectual disability, and seizures. The clinical presentation can be influenced by the presence of schizencephaly, seen in about 50% of cases, referred to as 'Septo-optic dysplasia plus', which is associated with milder optic system impairment and cortical anomalies like polymicrogyria and cortical dysplasia. The precise incidence of pachygyria in individuals with SOD is not well-documented, though studies have reported an association between the two.[1,3,4,5,6,9]

Imaging modalities, including CT and MRI, can detect the absence of the septum pellucidum, optic nerve hypoplasia, enlarged ventricles, and other brain anomalies. There is a 'pointed down' configuration of the lateral ventricular frontal horns in coronal images. CT scan, in addition, can reveal associated small bony optic foramina. MRI is the preferred modality for diagnosing SOD and characterizing optic pathways. It also offers insights into potential associated abnormalities such as corpus callosum hypoplasia, a small pituitary gland with a hypoplastic or absent infundibulum, an ectopic posterior pituitary that appears as a T1 high-signal focus in the hypothalamus' median eminence and cortical malformations like polymicrogyria, cortical dysplasia, and pachygyria. Additionally, MRI can detect the absence of olfactory bulbs, a condition known as arhinencephaly. An optic nerve area of ≤ 4.0 mm² measured on MRI demonstrates moderately high sensitivity and specificity in detecting optic nerve atrophy.[7,10,11]

A multidisciplinary approach is essential in managing SOD, with endocrinologists addressing hormonal imbalances and obesity, ophthalmologists managing vision loss, and neurologists treating neurodevelopmental issues. Prognosis depends on the severity of the condition, and early diagnosis improves outcomes by facilitating prompt management of hormone deficiencies.[3]

CONCLUSION

In children presenting with vision loss, clinicians and radiologists should maintain a high index of suspicion for SOD, especially when other characteristic features are present.

CONFLICT OF INTEREST

None

SOURCES OF FUNDING

None

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