

## Case report

# Neuroimaging of septo-optic dysplasia-plus with midbrain hypoplasia and ophthalmoplegia

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## ABSTRACT

Septo-optic dysplasia is a rare brain malformation that can be associated with anomalous cortical development, such as schizencephaly, which is referred to as septo-optic dysplasia plus. This report describes septo-optic dysplasia-plus associated with unilateral atrophy of the midbrain and oculomotor nerve deficiency, which was diagnosed on MRI in a teenage male who presented with ophthalmoplegia.

## 1. Case report

A 14-year-old male presented with strabismus and nystagmus, including right eye esotropia. According to his mother, the patient's vision problems have been present since birth, when he was diagnosed with cerebral palsy and schizencephaly with left hemiplegia. The patient did not have eye patching, but wears glasses when attending school. The patient has no history of seizures.

Ophthalmic examination revealed the visual acuity to be 20/60 in the right eye and 20/40 in the left eye. The visual fields were full to confrontation. The pupils measured 4 mm in dim illumination and constricted briskly and equally to bright light. Fundus examination showed inferior and nasal lattice degeneration in the right eye with no breaks or detachment. Fundus examination results for the left eye were normal. There was severe right upper lid ptosis and minimal lid crease indicating poor levator function. Downward chin and right head tilt positioning was also noted. The patient was able to adduct his right eye on left gaze. Horizontal beating nystagmus that worsened on left gaze was noted in both eyes. The diagnosis of partial right third nerve palsy was made.

MRI of the brain and orbits showed open-lip right cerebral hemisphere schizencephaly and absence of the septum pellucidum, as well as hypoplasia of the right midbrain and the right oculomotor nerve (Fig. 1). Additionally, there was atrophy of the extraocular muscles supplied by the right oculomotor nerve, as well as the right superior oblique muscle (Fig. 2). Furthermore, there was hypoplasia of the bilateral optic nerves, as part of septo-optic dysplasia. Bilateral strabismus correction was recommended for the patient.

## 2. Discussion

Schizencephaly is a rare congenital malformation of the brain in which there are clefts in the cerebral hemisphere lined by abnormal grey matter and commonly presents with seizures, cognitive deficits, and hemiplegia or other motor deficits [1]. There is also an association with septo-optic dysplasia, which can consist of optic nerve hypoplasia, deficiency of the septum pellucidum, and pituitary hypoplasia [2]. Indeed, both schizencephaly and septo-optic dysplasia are believed to be embryological disorders of neuronal migration and organization related to an ischemic event during the seventh week of gestation [3–5]. The presence of septo-optic dysplasia with associated malformation of cortical development is known as septo-optic dysplasia-plus [6].

Septo-optic dysplasia can also be associated with third nerve palsy, as demonstrated in this case and prior reports [7,8]. The presence of oculomotor nerve and trochlear nerve deficiency ipsilateral to the schizencephaly observed in this case is related to ipsilateral midbrain hypoplasia, perhaps as a form of congenital Wallerian degeneration. Along with the other anomalies, the presence of the hemi-midbrain atrophy and oculomotor nerve deficiency may be analogous to the purported vascular etiology reported for congenital third nerve palsy associated with midbrain hypoplasia due to bilateral segmental internal carotid artery agenesis [9].

Although the neuroimaging features of schizencephaly and septo-optic dysplasia have been well-characterized, there is a paucity of literature specifically related to MRI findings of midbrain involvement and associated neuro-ophthalmological deficits in the setting of septo-optic dysplasia plus. MRI is the imaging modality of choice for evaluating congenital malformations of the brain and assessing the cranial

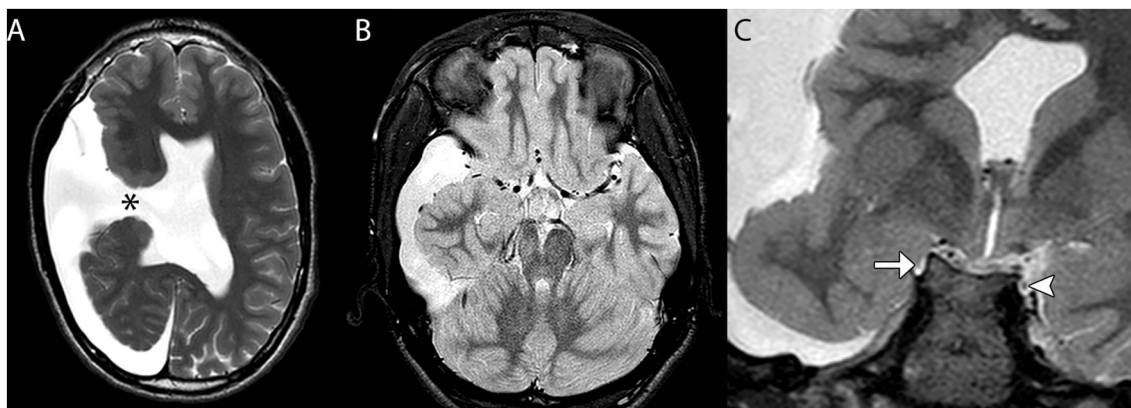
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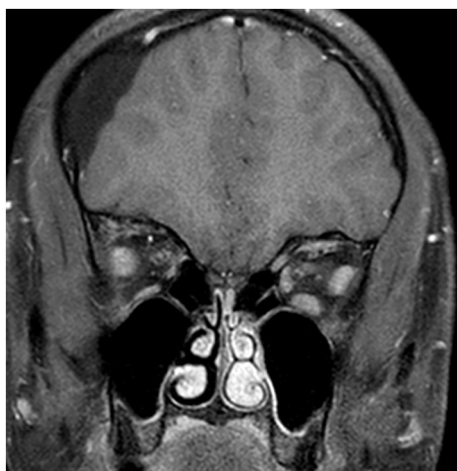
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**Fig. 1.** Axial T2-weighted MRI shows a cleft (\*) in the right cerebral hemisphere lined by abnormal grey-matter, which is consistent with open-lip schizencephaly. There is also absence of the septum pellucidum (A). Axial T2-weighted MRI shows the right midbrain is relatively small (B). Coronal T2-weighted MRI shows the deficient right oculomotor nerve (arrow) and intact left oculomotor nerve (arrowhead) (C).



**Fig. 2.** Coronal fat-suppressed post-contrast T1-weighted MRI shows atrophy of the right extraocular muscles supplied by the oculomotor and trochlear nerves.

nerves, such as in patients with septo-optic dysplasia. In particular, high in plane resolution and small slice thickness MRI protocols are necessary for depicting deficiency of the oculomotor nerve along with the associated atrophy of the extraocular muscles [10], as demonstrated in this case.

### 3. Conclusion

Septo-optic dysplasia associated with schizencephaly can affect the midbrain and nerves supplying the extraocular muscles. The constellation of findings can be readily depicted on MRI.

### Funding

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### Informed consent

Waived.

### Declaration of Competing Interest

The authors declare that they have no conflict of interest.

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