Clinical History:

A ten-month-old infant presented with vertical nystagmus. Ophthalmic examination revealed bilateral atrophy of optic nerves. Physical exam showed no focal deficits. Labs revealed no endocrine abnormality.

Imaging Findings:

Brain MRI was performed with acquisition of TSE-T2, FLAIR, TSE-T1, SWI and IR sequences. Imaging yielded an absence of the septum pellucidum with flattening of the roof of the lateral ventricles and caudal projection of the anterior horns. In addition, the optic nerves, chiasm, and pituitary gland (including stalk) were all noted to be atrophic.

Other abnormal findings included an irregular, thick and broad perisylvian and fronto opercular cortex with large perivascular spaces as well as a cleft connecting the lateral ventricle to the subarachnoid space, lined by polymicrogyric gray matter. This constellation of findings was suggestive of cortical development anomaly polymicrogyria - pachygyria and closed lip schizencephaly.

Discussion:

Septo-optic dysplasia (SOD) or De Morsier syndrome is a rare congenital malformation disorder characterized by absence of septum pellucidum and optic nerve/tract hypoplasia [4]. Additionally, up to 60% percent of patients can experience patient pituitary gland dysfunction [3]. It is an early forebrain development abnormality that occurs during the 4th–6th week of gestation. It is believed to be part of the holoprosencephaly spectrum [5].

The aetiology is unknown and a genetic diagnosis can be made in less than 1%. SOD has a reported incidence of 1 in 10.000 live births and is equally prevalent in females and males [3]. SOD is usually associated with other cerebral anomalies such as schizencephaly, polymicrogyria, cortical dysplasia, gray matter heterotopia, Chiari II malformation, aqueductal stenosis and midline malformations. Together, these associated anomalies are termed SOD–plus syndrome [2].

The clinical presentation of SOD is varied and the severity depends on the association with other disorders. Clinical symptoms involve visual impairment and signs of pituitary hypofunction (60–80%). Visual symptoms are decreased visual acuity, colour blindness, visual loss and nystagmus. Pituitary hypoplasia might manifest as variable endocrine deficits ranging from isolated growth hormone deficiency to complete panhypopituitarism. Neonates may present
with hypoglycemic seizures, apnea, cyanosis or hypotonia due to pituitary hypofunction [3].

Prenatal ultrasound is the first image method for diagnose the absence of septum pellucidum. Fetal MRI is a helpful tool to confirm or exclude the features and to assess other associated brain malformation. However, fetal MRI is limited for the evaluation of the optic tracts so the diagnosis must be confirmed after birth.

MRI is the best technique for evaluation of patients with SOD. The most important radiological finding is complete absence of septum pellucidum resulting in a flat roof and downwards pointing of frontal horns with a caudally displaced fornix. Optic nerve/chiasm hypoplasia can be unilateral or bilateral. In addition, MRI often shows a thin pituitary stalk with small anterior lobe which can subsequently be associated with posterior pituitary ectopia [1, 4].

People with SOD must be treated individually and require a multidisciplinary team to assess endocrine, neurological and ophthalmological symptoms. The prognosis varies according to the severity of the clinical presentation.

**Differential Diagnosis List:** Septo – optic dysplasia, Lobar holoprosencephaly, Agenesis of corpus callosum

**Final Diagnosis:** Septo – optic dysplasia

**References:**


Description: Coronal T2-weighted MRI shows complete absence of the septum pellucidum and the shape and flat roof of the frontal horns with inferior pointing of the ventricles. **Origin:** Cruces University Hospital, Vizcaya, Spain
**b**

**Description:** Axial T1-weighted MRI images reveal complete absence of the septum pellucidum. Thick and broad perisylvian and fronto opercular cortex suggestive of pachgyria-polymicrogyria is seen.

**Origin:** Cruces University Hospital, Vizcaya, Spain

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**c**

**Description:** Axial IR sequences show unilateral closed lip schizencephaly. The cleft is lined by gray matter and extends from the pial surface to the lateral ventricle. **Origin:** Cruces University Hospital, Vizcaya, Spain
**Description:** Sagittal T1-weighted image demonstrates a thin pituitary gland including stalk. **Origin:** Cruces University Hospital, Vizcaya, Spain

**Description:** A) Coronal T2-weighted, B) axial T2-weighted and C) axial T1-weighted images reveal bilateral hypoplasia of optic nerves. **Origin:** Cruces University Hospital, Vizcaya, Spain