

Septo-Optic Dysplasia with Pineal Gland Cyst

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Abstract: Septo-optic dysplasia (SOD) is a neurodevelopmental disorder that affects midline brain structures. Magnetic resonance imaging (MRI) documented midline brain defects include absent septum pellucidum, agenesis of corpus callosum, posterior displacement of pituitary gland and cerebellar hypoplasia. We are reporting a case of a 10-year-old boy who presented with blurring of vision of the left eye with features suggestive of optic nerve hypoplasia. MRI revealed absence of septum pellucidum with pineal gland cyst which has never been reported as a spectrum of this disorder. As pineal gland is a midline brain structure that is derived from diencephalon, this finding may indicate another spectrum of the septo-optic dysplasia.

Keywords: Optic nerve hypoplasia, Septo-optic dysplasia, Septum pellucidum, Pineal gland cyst

INTRODUCTION

Septo-optic dysplasia (SOD) was first described by Reeves in 1941 as the absence of septum pellucidum associated with optic nerve disorder [1, 2]. Its incidence is 1 in 10 000 live births [3, 4], equally prevalent between males and females [4], and more common in infants born to younger mothers [5] and areas with higher unemployment rate [3]. It is a rare congenital anomaly which involves defective forebrain development resulting in midline brain structural defects and optic nerve hypoplasia (ONH) [6]. Its involvement may cause dysfunctional pituitary gland leading to symptoms of panhypopituitarism [7].

Diagnosis of SOD involves the presence of 2 out of the 3 classical triad of optic nerve hypoplasia, midline structure defects from neuroimaging studies and hormonal deficiency due to hypopituitarism (with growth hormone deficiency being the most common) [4, 8]. Visual prognosis of ONH generally is poor and a child with SOD may require life-long medical treatment as pituitary hormonal insufficiency may occur later in life [9].

CASE REPORT

A 10 year old boy, with a background history of nephrotic syndrome was presented with painless reduction of left eye vision since 2006. There was no history of ocular trauma or fall previously. Antenatal, intra-partum and post-partum history was uneventful. Developmental was up to his age. There was no history of seizure, headache or neurological symptoms. There

was no delay in gross and fine motor developmental milestones. No other siblings or family members has similar problem. There was no consanguineous marriage.

Ocular examination revealed visual acuity of 6/12 on the right eye and 6/60 on the left, with presence of relative afferent pupillary defect (RAPD) in the left eye. There was no microphthalmia or anophthalmia noted. Anterior segment examination was unremarkable. Intraocular pressures were 16 mmHg bilateral eyes. Fundoscopy of the left eye showed tilted disc with pallor at temporal segment of the optic disc with normal optic disc on the fellow eye. Initial working diagnosis was left eye optic atrophy secondary to unknown cause. Magnetic resonance imaging (MRI) brain was ordered to rule out compressive lesion. MRI brain showed absence of septum pellucidum (Fig. 1) with presence of a pineal gland cyst measuring 8 mm (Fig. 2). There was no agenesis of corpus callosum, ectopic posterior pituitary or cerebellar hypoplasia. There was no other space occupying lesion noted.

On subsequent review, further fundus examination of left eye noted DM (disc to macula) to DD (disc diameter) ratio of more than 3 (Fig. 3), which support the diagnosis of optic nerve hypoplasia. Refraction showed bilateral myopic astigmatism with anisometropia (RE: -1.00DS/-0.75DC x180 [6/7.5] / LE: -5.50 DS/-2.00DC x 180 [6/24]) and left eye amblyopia. There was color vision defect (using Ishihara isochromatic plate) and Bjerrum screening

showed constricted visual field in the left eye. Humphrey visual field 30-2 was attempted but not reliable as it had high fixation loss.

On general examination the child has no dysmorphic features, mental slowness or short stature (growth within 25th to 50th centile). We referred this patient to our paediatric colleagues for endocrinological assessment. Their assessment revealed no neurological

or endocrinological dysfunction except for his nephrotic syndrome in remission.

Clinical diagnosis of SOD was made based on the criteria of presence of optic nerve hypoplasia and neuroradiological findings of absent septum pellucidum although he didn't have any hormonal or endocrinological disturbances at the time of writing.

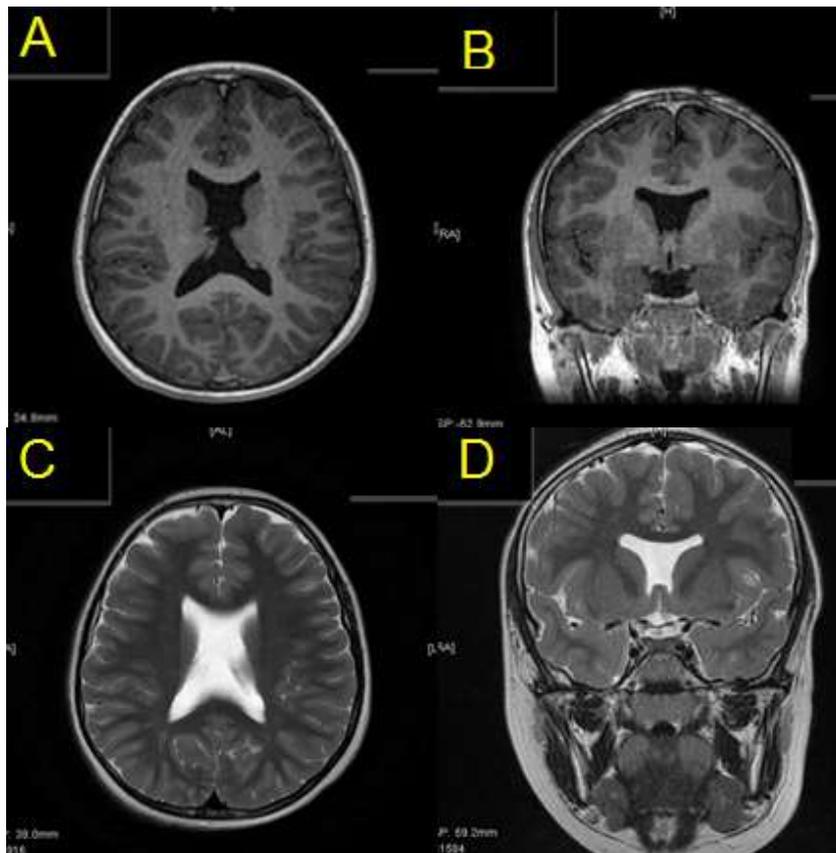


Fig. 1: T1-weighted (A,B) and T2-weighted (C,D) MRI images show absent septum pellucidum



Fig. 2: Sagittal midline T1-weighted MRI images show pineal cyst measuring 8mm (white arrow)

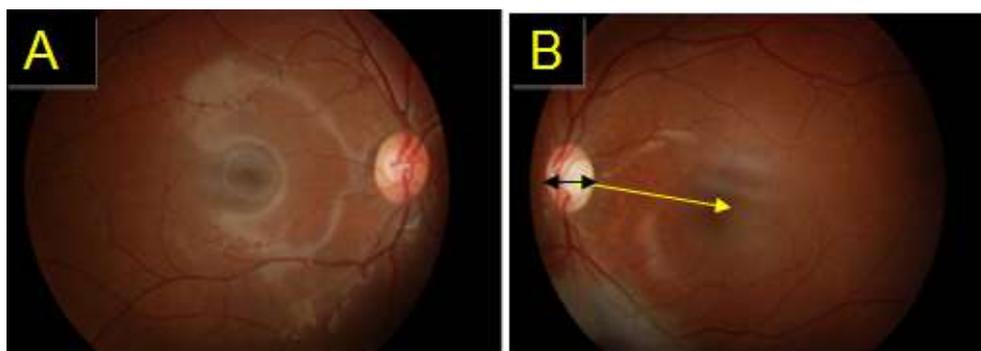


Fig. 3: Left fundus shows pallor optic disc temporally with disc to macula (DM; yellow arrow) to disc diameter (DD; black arrow) ratio more than 3 (B), Normal fundus of the right eye (A)

DISCUSSION

SOD is a heterogenous and highly variable phenotypical disorder [4]. The diagnosis of SOD is based on the following criteria; there is presence of any 2 combination of (a) optic nerve hypoplasia, (b) neuroradiological imaging of midline brain defects (absent septum pellucidum, corpus callusom agenesis) and (c) hormonal deficiency due to hypopituitarism [4, 8]. Morishima and Aranoff in 1986 reported that about 30% of SOD patients had evidence of all three components of the diagnostic criteria, whereas 60% had absent septum pellucidum and 62% had features suggestive of hypopituitarism [10]. Besides the classical features of SOD, Polizzi *et al.* [11] had reported cases of SOD associated with dysmorphic features (not only involving midline facial structures) and a spectrum of additional clinical and imaging features including autism, facial haemangioma and holoprosencephaly, which made SOD more phenotypically diversified.

Thus, it has been suggested that SOD be considered as a complex syndrome rather than a single defined entity [12], giving rise to a new terminology of heterogenous malformation syndrome or septo-optic dysplasia complex [11].

The common reported neuroradiological findings found in SOD are absent septum pellucidum, agenesis of corpus callosum, undescended posterior pituitary, ectopic posterior pituitary and cerebellar atrophy [13-15].

Individuals with SOD often have problems with their pituitary gland [16]. There has been no reported case of associated pineal gland cyst. The possibility of pineal gland cyst as seen in this patient can occur simultaneously during abnormal developments of the posterior pituitary, as the pineal gland arises from epithelial thickening of caudal root plate of the diencephalon opposite to the infundibulum (which attaches to the hypothalamus and grows distally to form the infundibular stalk and pars nervosa of the pituitary gland). As the origin of these two structures is

derived from diencephalon, pineal gland cyst might share similar pathogenesis pathway as posterior pituitary pathology in SOD; therefore the presence of pineal gland cyst might be due to the manifestation of SOD itself. Thus, pineal gland pathology (cyst) might represent another spectrum of SOD which has never been reported before.

CONCLUSION

This case reveals absence of septum pellucidum with pineal gland cyst. This further supports the notion that SOD is a heterogenous syndrome rather than a single entity and further study will be needed to ascertain its correlation for a better diagnosis and treatment strategy in the future.

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