An eye behind an eye: A rare association of bilateral optic disc coloboma with retrobulbar cyst

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Key words: Optic disc coloboma, retrobulbar cysts, visual stimulation exercises

A 2-month-old male child brought by parents with complaints of not following light. Child was born at full-term by LSCS. Antenatal scan revealed cleft lip but no other abnormality. Baby was not able to fix or follow the light. Retinoscopic shadow was normal for that age. Anterior segment examination was normal. Fundus examination [Fig. 1] revealed a well-defined posterior excavation in optic nerve head with a thin rim of neural tissue surrounding the excavation in both eyes suggestive of optic disc coloboma. Ultrasound B-scan [Fig. 2] showed symmetric excavation along the optic disc leading to a cyst in retrobulbar space in both eyes. MRI scan of orbits [Fig. 3] showed a homogeneous intraconal retrobulbar cystic lesion along the optic nerve on both sides.

Systemic evaluation revealed no other abnormalities, but parents denied for genetic evaluation. They were counseled about visual prognosis. The child was 2-month old and fixation has not developed. We advised vision stimulation exercises like running lights, colored balls, as visual acuity is reduced to varying degrees with bilateral disc coloboma. On follow-up at 3 months after presentation, child was able to fix and follow light and hold objects at a distance of 1–1.5 feet.

Discussion

The development of optic disc coloboma was due to incomplete fusion of proximal ends of the optic cup[1] or due to defective migration of neural crest cells and/or due to PAX-2 gene mutation causing abnormal astrocytic differentiation.[2]

Clinically child may present with defective vision, significant refractive error, and anisometropia.[3] Visual outcome depends on foveal involvement by coloboma. Imaging modalities such as B-scan (USG), computed tomography (CT), and magnetic

Access this article online	
Quick Response Code:	Website: www.ijo.in
	DOI: 10.4103/ijo.IJO_1957_19

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Received: 25-Oct-2019 Revision: 26-Dec-2019
Accepted: 02-Mar-2020 Published: 24-Jul-2020

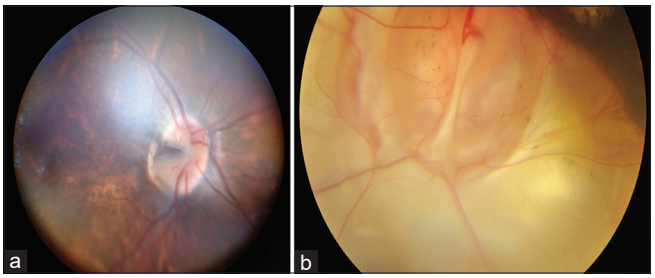

Figure 1: Fundus photograph of right eye (a) and left eye (b) showing a well-defined posterior excavation in optic discs suggestive of optic disc coloboma

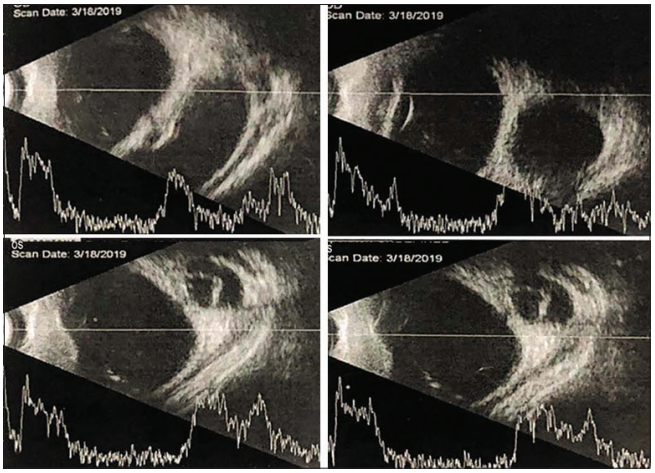

Figure 2: Ultrasound B-scan of both eyes showing abnormal optic nerve head with a retrobulbar hypoechoic lesion communicating with the optic nerve head

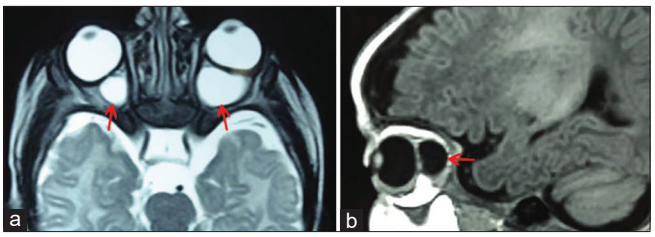

Figure 3: MRI scan of the orbits, axial T2W (a) and sagittal T1 W (b) sequence in parasagittal location, showing bilateral retrobulbar intraconal cystic lesions, isointense to vitreous and probably communicating with the globe. No intracranial communication is seen

Cite this article as: Shankar LG, Khadia A, Gayam K, Priya S, Moutappa F. An eye behind an eye: A rare association of bilateral optic disc coloboma with retrobulbar cyst. Indian J Ophthalmol 2020;68:1656-7.

resonance imaging (MRI) are helpful in supporting the diagnosis, in differentiating it from other cystic lesions or tumors and also for identifying intracranial extension of the cyst.

To conclude, any child presenting with optic disc coloboma and/or orbital cyst should be evaluated to rule out other ocular and systemic abnormalities and giving a trial of visual stimulation exercises will help in improving the visual potential of the child.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

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