Temporal iridofundal coloboma with persistent pupillary membranes with persistent fetal vasculature

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Fundal coloboma and persistent fetal vasculature are congenital vitreoretinal disorders that can severely hamper vision. Typically, iridofundal colobomas are located in inferonasal quadrant.

A 15-year-old boy presented with painless nonprogressive diminution of vision in right eye since childhood.

Best-corrected visual acuity (BCVA) in right eye was finger counting close to face (FCCF), PR accurate, whereas left eye had 6/6 vision. On slit-lamp examination, right eye had microphthalmia, temporal iris coloboma involving the pupillary margin and persistent pupillary membranes [Fig. 1]. There was temporal lens coloboma with posterior subcapsular cataract. On posterior segment evaluation with indirect ophthalmoscopy, he was found to have persistent fetal vasculature and temporal chorioretinal coloboma [Fig. 2] sparing the macula. Posterior segment OCT revealed fluid in the sub-intercalary membrane space [Fig. 3]. The left eye was unremarkable.

Nature of the disease and poor prognosis was explained to the patient and he was kept on regular follow-up.

Discussion

Coloboma is derived from the Greek word “koloboma” which means defect or curtailed. Ocular coloboma due to failed embryonic fissure closure occurs in 0.5 to 2.2 cases per 10,000 live births.[1]

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Iris coloboma is a full-thickness defect of iris involving both the stroma and pigment epithelium. It occurs due to failure of closure of embryonic fissure anteriorly, whereas posteriorly it leads to choroidal coloboma. Thus, in cases where iris and choroidal coloboma occur simultaneously, they are generally located in the same quadrant.

Small fine strands of iris can be seen bridging the pupil or as webs with attachments to the lens.[2] These are persistent pupillary membranes, remnants of tunica vasculosa lentis, a vascular structure which surrounds the lens during development which usually disappears completely by 8th month of gestation. These are seen in 95% of neonates and 20% of adult population.[3]

Ocular coloboma may cause defective or absence of zonules in a segment which causes flattening of the lens equator due to lack of tension on the lens capsule. This is called lens coloboma but it is a misnomer as there is no lens tissue loss.

Persistence of hyaloid vessel is called persistent fetal vasculature or persistent hyperplastic primary vitreous (PHPV), which causes cataract and is usually associated with microphthalmia.[2] Posterior PHPV is usually associated with developmental anomalies of the surrounding retina and optic nerve, hampering vision significantly.

In contrast to typical colobomas, in atypical colobomas, iris and choroidal colobomas are rarely located in the same quadrant. To date, very few atypical colobomas have been reported. Ward[6] reported a case with temporal iris and lens coloboma with cataract. Azad and Mittal[5] reported a case with a combination of typical inferonasal coloboma involving optic disc and a temporal atypical coloboma in both eyes. Kumar et al.[6] reported a case of 21-year-old female with 2 fundal colobomas—one in inferior hemisphere and the other in superior hemisphere of the globe resembling an hourglass in right eye and typical type-3 fundal coloboma in left eye.

Atypical temporal location of coloboma may be explained by the presence of “accessory embryonic fissures” in developing eye which may be located in any quadrant. Therefore, failure of fusion of temporal embryonic fissure with normal closure of inferonasal embryonic fissure could lead to temporal iridofundal coloboma. This is further substantiated by the observation of accessory fissures in sheep and chick embryo but it has not been documented in humans yet.

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.

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Conflicts of interest
There are no conflicts of interest.

References
1. Warburg M. Classification of microphthalmos and coloboma. J Med Genet 1993;30:664-9.
2. MacDonald AE. Causes of blindness in Canada: An analysis of 24,605 cases registered with the Canadian National Institute for the Blind. Can Med Assoc J 1965;92:264-79.
3. Tasman W, Jaeger E. Duane’s Ophthalmology 2007. Philadelphia: Lippincott Williams and Wilkins; 2007. p. 242-58.
4. Ward M. Temporal iris and lens coloboma associated with cataract. EyeRounds.org Online Ophthalmic Atlas. Webeye.opth.uiowa.edu 2012. Available from: https://webeye.opth.uiowa.edu/eyeforum/atlas/pages/coloboma-temporal-iris-lens-cataract.html. [Last accessed on 2020 Mar 17].
5. Azad S, Mittal K. Atypical choroidal coloboma. Ophthalmol Retina 2018;2:1142.
6. Kumar M, Shanmugam M, Sagar P, Kumar D, Konana VK. “HOURGLASS COLOBOMA”: A case report and review of etiopathogenesis. Retinal Cases Brief Rep 2018. doi: 10.1097/ ICB.0000000000000810.