Atypical superior iris and chorioretinal coloboma

Shweta Parakh, Shrutanjoy Das, Shrey Maheshwari, Gaurav Luthra, Saurabh Luthra

Key words: Atypical coloboma, chorioretinal coloboma, iris coloboma, superior coloboma

An 8-year-old male presented with diminution of vision in the left eye since childhood. Best corrected visual acuity was 6/6 in the right eye (OD) and 6/24 in the left eye (OS). Refractive error of -0.50 DC at 180 degrees OD and +5.50 DS/-2.50 DC at 160 degrees OS was noted. Anterior segment and fundus were normal OD. The left eye showed a superotemporal coloboma in the iris [Fig. 1a]. Superotemporal zonules were absent [Fig. 1b]. Disc and macula were normal OS. Fundus examination with scleral indentation revealed an atypical chorioretinal coloboma in the superotemporal quadrant OS [Fig. 2].

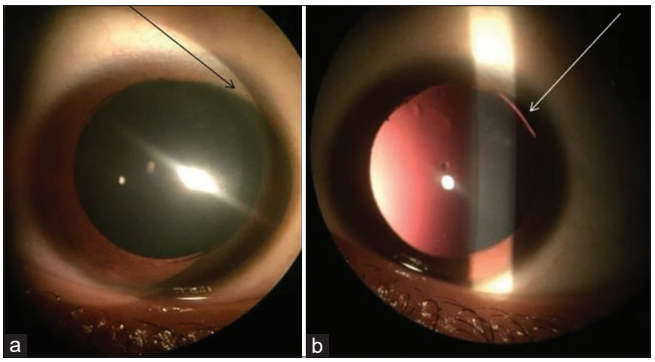

Figure 1: (a) Anterior segment photograph of the left eye showing superotemporal iris coloboma (black arrow); (b) Absent zonules in superotemporal quadrant of the left eye (white arrow)

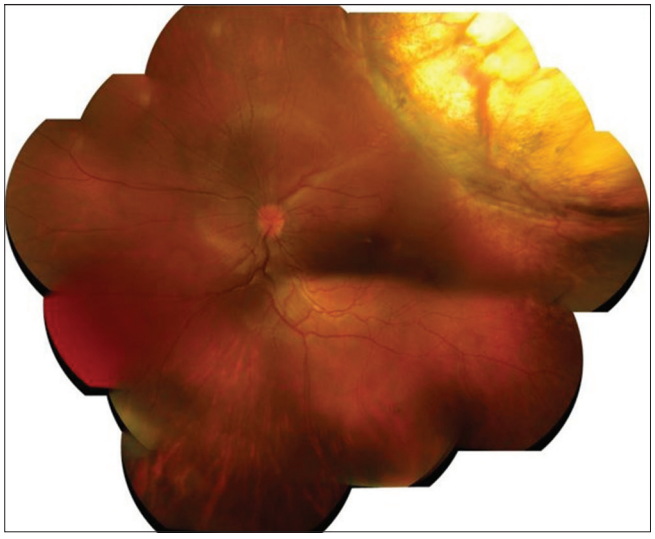

Figure 2: Mosaic fundus photograph of the left eye showing an atypical superotemporal chorioretinal coloboma

Discussion

Atypical coloboma involving the superior quadrants of iris, retina, and choroid is a rare anomaly with very few cases being reported so far.[1] Typical inferonasal ocular coloboma occurs due to defective closure of inferior embryonic fissure between 6th and 7th week of intrauterine life and may extend from the iris to the optic nerve head, involving one or more defects along the fusional lines.[2]

Atypical colobomas are found outside the inferonasal area and do not originate in a defect of embryonic fissure closure. They are thought to be either pathogenic or inflammatory in origin or develop due to faulty differentiation of ocular structures or rotation of the fetal fissure. They may also be due to persistence of embryonic mesodermal tissue blocking the forward growth of the neuroectoderm, thereby causing a defect in the ciliary body and iris.[3,4]

Hocking *et al*.,[5] proposed that atypical superior chorioretinal coloboma results from defective closure of superior ocular sulcus: a structure not conventionally defined in ocular embryology. This may result from variation in genes encoding type 1 bone morphogenetic protein receptor (BMPR1A) and T-box transcription factor 2 (TBX2).

A detailed retina examination is an essential part of ophthalmic evaluation to rule out disorders such as atypical coloboma.

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

Access this article online

Quick Response Code:	Website: www.ijo.in
	DOI: 10.4103/ijo.IJO_164_22

Drishti Eye Institute, Dehradun, Uttarakhand, India

Correspondence to: Dr. Saurabh Luthra, Drishti Eye Institute, 16, Subhash Road, Astley Hall, Dehradun - 248 001, Uttarakhand, India. E-mail: drsaurabhluthra@gmail.com

Received: 25-Jan-2022 Revision: 04-Mar-2022
Accepted: 14-Mar-2022 Published: 30-Jun-2022

This is an open access journal, and articles are distributed under the terms of the Creative Commons Attribution-NonCommercial-ShareAlike 4.0 License, which allows others to remix, tweak, and build upon the work non-commercially, as long as appropriate credit is given and the new creations are licensed under the identical terms.

For reprints contact: WKHLRPMedknow_reprints@wolterskluwer.com

Cite this article as: Parakh S, Das S, Maheshwari S, Luthra G, Luthra S. Atypical superior iris and chorioretinal coloboma. Indian J Ophthalmol 2022;70:2665-6.

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.

References

1. Jain AM, Ranjan R, Manayath GJ. Atypical superior iris and retinochoroidal coloboma. Indian J Ophthalmol 2018;66:1474-5.
2. Pagon RA. Ocular coloboma. Surv Ophthalmol 1981;25:223-36.
3. Onwochei BC, Simon JW, Bateman JB, Couture KC, Mir E. Ocular colobomata. Surv Ophthalmol 2000;45:175-94.
4. Klien BA. The pathogenesis of some atypical colobomas of the choroid. Am J Ophthalmol. 1959;48:597-607.
5. Hocking JC, Famulski JK, Yoon KH, Widen SA, Bernstein CS, Koch S, *et al*. Morphogenetic defects underlie Superior Coloboma, a newly identified closure disorder of the dorsal eye. PLoS Genet 2018;14:e1007246.