A rare case of temporal atypical retinochoroidal coloboma associated with posterior embryotoxon

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Key words: Atypical coloboma, lens coloboma, posterior embryotoxon, retinochoroidal coloboma

A 17-year-old female presented to us for routine ophthalmological evaluation. Her best-corrected visual acuity was 20/20, N06 in both the eyes. No previous history of ocular trauma or surgery was noted. Right eye anterior segment examination showed two clock hours (CH) of a prominent white line on the endothelial side just inside the temporal limbus, suggestive of posterior embryotoxon [Fig. 1a]. Gonioscopy showed two CH of angle recession temporally. Temporal 3 CH of lens pseudocoloboma was noted as well [Fig. 1b]. No iris coloboma was seen. The fundus examination showed a healthy disc and macula. Peripheral fundus showed temporal well defined glistening white lesion with minimal excavation and pigmented margins, suggestive of atypical retinochoroidal coloboma [Fig. 1c] and intact retina in inferior quadrant [Fig. 1d]. The left eye ocular examination was unremarkable.

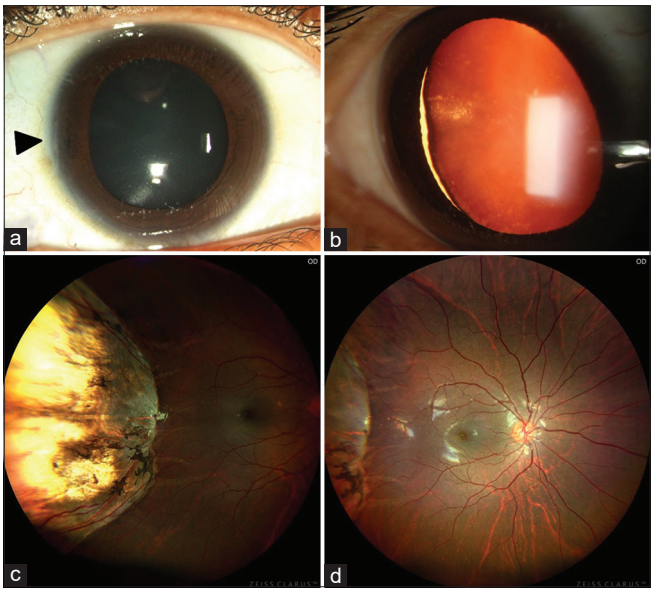

Figure 1: Slit lamp photograph of right eye showing (a) temporal posterior embryotoxon (black arrow head); (b) temporal lens pseudocoloboma; (c) Fundus showing temporal peripheral retinochoroidal atypical coloboma; and (d) healthy optic nerve and intact inferior retina

Discussion

Typical retinochoroidal colobomas are located in the inferonasal quadrant due to the failure of embryonic fissure in the ventral side of the optic cup to close during the fifth week of gestation.[1] Embryological basis of atypical colobomas can be explained by vertebrate eye model that depicts a transient groove in the dorsal side of the optic cup called the superior ocular sulcus (SOS).[2] Hocking *et al*.[2] have shown that failure to close the SOS leads to superior colobomas in adult zebrafish. Several other theories explaining the formation of atypical colobomas are rotation of the fetal fissure, intrauterine inflammatory process, and developmental anomalies of the neural ectoderm, namely, faulty differentiation, and imbalance of growth in various portions of the outer and inner layer of the optic cup.[3,4]

Posterior embryotoxon has been reported in association with retinochoroidal coloboma in Axenfeld–Reigers syndrome[5] and cholestatic disorders.[6]

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.

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