Atypical and extensive combined irido-retinochoroidal coloboma with microcornea

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DESCRIPTION
A preteenage girl presented with vision in her right eye reduced to counting fingers. Examination of the right eye showed 20 prism dioptres of exotropia. The anterior segment showed microcornea (diameter: 8 mm vertical and 9 mm horizontal) and a temporal focal defect of the iris and the ciliary body (figure 1a). Dilated fundus examination revealed a normal-looking disc with a large temporal and superotemporal retinochoroidal coloboma encroaching the temporal half of the macula. The large areas of colobomatous defect showed intervening pigmentary flash giving the appearance of a notch (figure 1b). Temporal retinal vessels that emanate from the disc traverse through the coloboma (figure 1b, black arrowhead). An independent vessel emanating from the bed of the coloboma appears to be traversing towards the temporal end of the coloboma (figure 1b, white arrowhead).

The eye had astigmatism of −4.25 dioptres at 180° and an axial length of 20 mm. Vision in the right eye did not improve owing to meridional amblyopia. The left eye was emmetropic with a vision of 6/6, an axial length of 22.60 mm, and normal anterior and posterior segments.

Ocular colobomas result from incomplete closure of the embryonic fissure at around 5–8 weeks of gestation. They may be associated with microcornea, myopia and microphthalmia.1 Classic defects occur in the inferonasal quadrants of the iris, choroid and retina. Typical colobomas are those that are present in the inferonasal quadrant, which corresponds to the situation of embryonic fissure during embryogenesis. Atypical colobomas may occur nasally,2 superi-orly3 or temporally4 and be of varying severity. These abnormal locations result from incomplete closure of the superior ocular sulcus,4 persistence of multiple aberrant clefts or rotation of the choroidal fissure.5 Blood vessels that traverse colobomatous defects may either be a retinal vessel emanating from the disc or a retinal/cilioretinal vessel emerging from the colobomatous bed. Vision may be poor in such eyes owing to colobomatous involvement of the disc and macula, cataract, retinal detachment, and amblyopia secondary to uncorrected refractive errors. The poor vision in the right eye is secondary to coloboma involving the macula and meridional amblyopia. The present case highlights a rare coexistence of microcornea with total (combined iris, ciliary body and retinochoroidal colobomas) and arypical coloboma.

Learning points

► Atypical ocular colobomas are rare congenital defects seen in quadrants other than the inferonasal quadrant of the eye.
► They may occur due to persistent multiple aberrant embryonic clefts or rotation of choroidal fissure.
► They may present with poor vision owing to macular involvement, amblyopia or associated anterior segment structural abnormalities.

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Case reports provide a valuable learning resource for the scientific community and can indicate areas of interest for future research. They should not be used in isolation to guide treatment choices or public health policy.

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