Unusual superior iris and retinochoroidal coloboma

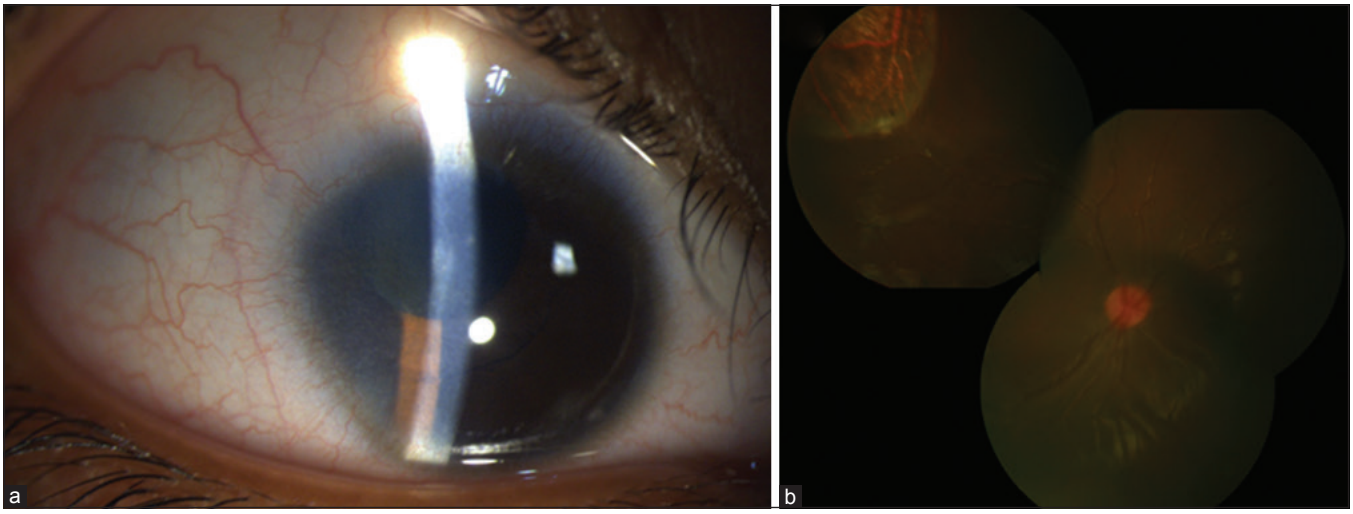

Figure 1: Slit-lamp photograph showing superior iris coloboma with microcornea (a), Montage: fundus photograph of right eye showing isolated choroidal coloboma in superotemporal periphery (b)

A 6-year-old female presented with decreased vision and inward deviation of right eye for five years. Her vision was 2/60 in the right eye. A slit–lamp examination revealed right eye esotropia, microcornea, and superior iris coloboma [Fig. 1a]. The fundus examination showed isolated superior retinochoroidal coloboma [Fig. 1b]. The typical inferior iris and retinochoroidal coloboma occurs due to the failure of closure of choroidal fissures during embryogenesis.[1-3] Variation in genes encoding the type 1 bone morphogenetic protein receptor (BMPR1A) and Tbox transcription factor 2 leads to superior retinochoroidal coloboma due to incomplete closure of superior ocular sulcus.[4]

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 The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.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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