

Atypical superior iris and retinochoroidal coloboma

A 38-year-old female presented with complains of right eye defective vision since childhood with best corrected visual acuity right-eye 6/9 and left-eye 6/6. Right-eye anterior segment showed superior iris coloboma with fundus showing atypical superior retinochoroidal coloboma [Fig. 1]. Presence of typical iris and retinochoroidal coloboma is a relatively common finding resulting from the failure of the choroidal fissure closure during embryogenesis.^[1] Atypical superior coloboma of iris and retinochoroid layer is a rare condition.^[2,3] Recently, a study has

proposed that superior retinochoroidal coloboma results from defective closure of superior ocular sulcus (a structure not defined in conventional model of ocular development) which results from the variation in gene encoding the type 1 bone morphogenetic protein (Bmp) receptor (*BMPRI1A*) and T-box transcription factor 2(*TBX2*).^[3] To our knowledge, this is the first case describing coexistence of superior colobomas of both iris as well as retinochoroid, and hence, supports the role of defective closure of superior ocular sulcus in the pathogenesis.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have

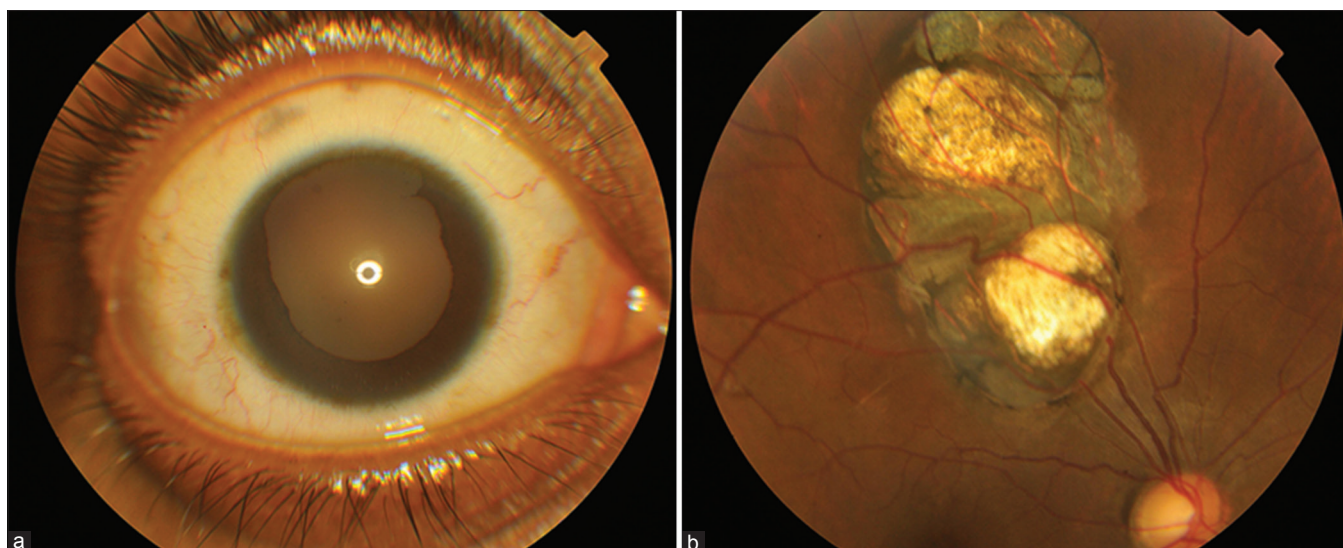


Figure 1: (a) right eye pharmacologically dilated pupil with deficient iris tissue supero-temporally; (b) retino-choroidal coloboma, supero-temporal to optic disc with well demarcated pigmented margins

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 The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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