Bilateral atypical fundal coloboma with macular drag and abnormal vasculogenesis

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DESCRIPTION

A boy in his middle childhood presented with bilateral diminution of vision since childhood with no history of prematurity, low birth weight or neonatal intensive care unit (NICU) stay. There was no associated systemic morbidity or facial dysmorphism. A best corrected visual acuity of 2/60 OD (right eye), and 6/60 OS (left eye) was noted with pendular nystagmus OU (both eyes). Slit lamp examination revealed microcornea with temporal atypical iris coloboma OD. Prominent persistent pupillary membrane was appreciated OU (figure 1). Fundus examination revealed bilateral symmetrical temporal fundal coloboma approximately 5 clock hours in extent with significant disc macular drag (figure 2A,B). Temporal looping of nasal vessels with peripheral straightening was detected OU. Peripapillary circumscribed retinal pigmentary changes was noted OD, showing a stippled hyperfluorescence on fundus fluorescein angiography (FFA; figure 2C). OS revealed peripapillary neurosensory detachment depicted by a localised hyperfluorescence on FFA (figure 2D). FFA also revealed leakage of dye from vessels in the intercalary membrane in late phases suggestive of abnormal vasculogenesis OU (figure 2E,F). Toxoplasma IgM and IgG antibody titers were within normal limits.

Patient was evaluated to rule out CHARGE syndrome; however, the major or minor criteria required for diagnosis were not met.

Temporal iris coloboma is a rare finding, and it is even rarer to have atypical iris and fundal coloboma in the same quadrant.[1 2] Gulati *et al* has described a case of unilateral temporal fundal coloboma with normal iris and temporal pseudocoloboma.[3] Many theories have been proposed to explain the pathogenesis of atypical colobomas including presence of accessory embryonic fissures, Vossius' theory of rotation of the choroidal fissure and Szily's theory of presence of persistent multiple aberrant clefts.[4–6] Accessory embryonic fissures or rotation of the choroidal fissure with incomplete closure of the choroidal fissure both anteriorly and posteriorly (OD) and complete closure of the fissure anteriorly (normal iris) and incomplete fusion posteriorly (temporal fundal coloboma) OS can explain the findings in our patient.

Temporal disc-macular drag associated with vascular changes is observed in retinopathy of prematurity, familial exudative vitreoretinopathy (FEVR), congenital retinal folds, incontinentia pigmenti and peripheral toxocara granuloma. Association of fundal coloboma with FEVR has only been reported once in the literature.[7] FEVR is a hereditary disorder linked to genes affecting Wingless/Int1 (Wnt) signalling pathway.[8] Wnt signalling pathway plays a pivotal role in vascular morphogenesis in the eye.[9] Genetic deletion of LRP6 (LDL receptor related protein 6) has shown to attenuate Bmp and retinoic acid signalling gene activation in the dorsal optic cup, resulting in the alteration of the expression of dorsal and ventral neuroretinal markers. This can lead to ocular axis defects and ocular coloboma formation.[10] On the other hand,

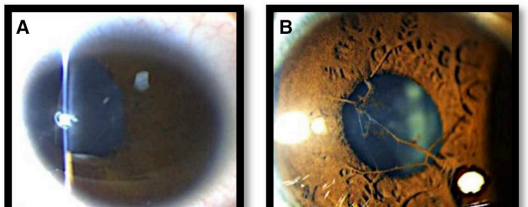

Figure 1 Slit lamp photograph showing microcornea with temporal atypical iris coloboma OD (A) and normal anterior segment findings OS with prominent persistent pupillary membrane OS (B).

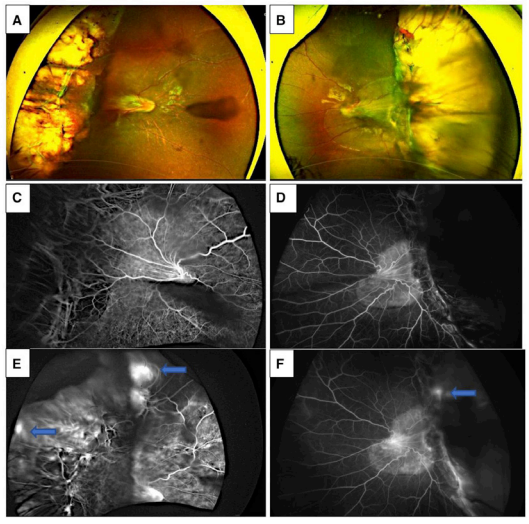

Figure 2 Ultrawide field (UWF) fundus image showing bilateral symmetrical temporal fundal coloboma with significant disc macular drag OD and OS, respectively (A and B). UWF fluorescein angiography showing stippled hyperfluorescence in the posterior pole OD (C) and localised hyperfluorescence corresponding to the area of peripapillary circumscribed neurosensory detachment OS (D). Late phase of fundus fluorescein angiography depicts leakage in temporal periphery OU suggestive of abnormal vasculogenesis (blue arrow; E and F).

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Lrp6 has been implicated to play a critical role in Wnt signalling pathway.[11] Thus, disruption of the Wnt pathway can explain the coexistence of FEVR and fundal colobomain our patient.[12] The presence of vasculopathy in association with the fundal coloboma can also explain the tractional component noted in our patient.

To the best of our knowledge, bilateral symmetrical atypical fundal coloboma with temporal dragging of disc and macula with peripapillary neurosensory detachment with peripheral angiogenesis have not been reported in the literature till date.

Learning points

- Bilateral atypical fundal coloboma with temporal iris coloboma is a rare finding.
- Coexistence of abnormal peripheral vasculogenesis with coloboma is in itself a rare finding with Wnt pathway disruption as a probable link between the two entities.
- We would like to emphasise the importance of ultrawide field angiography in such patients with poor central fixation to evaluate peripheral anatomical changes.

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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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