Focal choroidal excavation in Stargardt's dystrophy

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DESCRIPTION

A 35-year-old woman presented with symptom of gradually progressive diminution of vision in both eyes since childhood. Patient gave no history of defective night vision. Family history was unremarkable. On examination best corrected visual acuity was 1/60 in both eyes (OU). Anterior segment was within normal limits in either eye. On funduscopy, scattered yellowish flecks were seen at posterior pole around the vascular arcades, fundus autoflourescence revealed hypoautofluorescence at posterior pole indicating retinal pigment epithelium (RPE) and photoreceptor loss and swept source optical coherence tomography (OCT) revealed bilateral macular atrophy (right eye: figure 1; left eye: figure 2). Additionally, a dark red lesion above fovea was noted in left eye (OS) (figure 2A: white arrow). OCT through this lesion revealed a focal choroidal excavation (FCE) (figure 2C: white arrow). A diagnosis of Stargardt's disease (SD) OU with FCE OS was made. Patient was counselled regarding poor visual prognosis and conservatively followed.

SD is one the most commonly inherited macular dystrophies causing vision loss in the young. Genetic basis involves mutation in ABCA4 gene on chromosome 1 or rarely a PROM1 mutation on chromosome 4.[1] Vision loss is attributed to accumulation of lipofuschin in the RPE with subsequent damage of photoreceptors. FCE is described as a localised area of choroidal excavation without any evidence of posterior staphyloma or scleral ectasia.[2] FCE is broadly divided in two types—*conforming type* in which the photoreceptor tips are in direct contact with the RPE; and the *non-conforming type* in which the two are separated by a hyporeflective cleft.[2 3] Initially it was thought to be a congenital malformation; however, newer insights suggest an acquired aetiology. It is hypothesised that FCE could coexist with macular dystrophies because of focal degeneration of RPE and choroid.[4] FCE alone do not warrant treatment, still, its association with choroidal neovascularisation mandates meticulous follow-up. FCE in our case was of conforming type (figure 2C). Battaglia Parodi *et al* in their study on macular dystrophies, found only a single eye to have FCE with SD.[4] Likewise, Braimah *et al* in a large series of patients with SD, found no eye with coexisting FCE.[5] FCE coexisting with macular dystrophies have been reported mainly in Best disease.

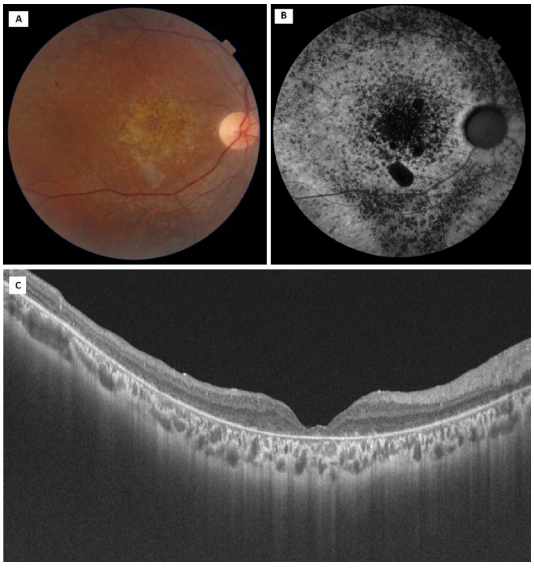

Figure 1 (A) Right eye fundus photograph with posterior pole mottling. (B) Hypoautofluorescence at posterior pole. (C) Swept source optical coherence tomography horizontal scan with macular atrophy.

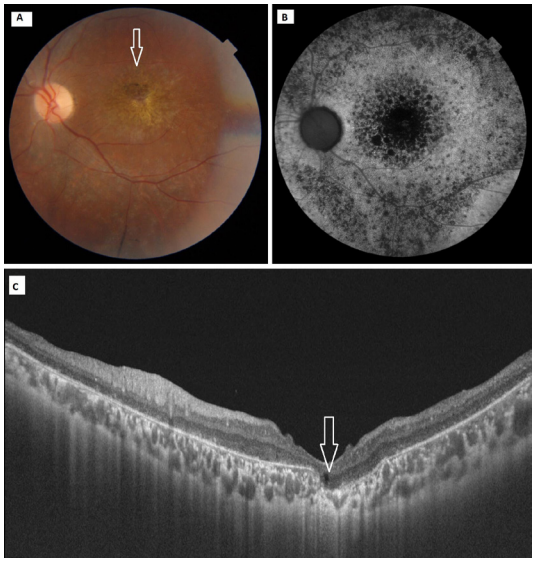

Figure 2 Similar findings in left eye. (A) Red patch with increased choroidal show—white arrow. (C) Focal choroidal excavation—white arrow.

Learning points

- Focal choroidal excavation (FCE) is defined as an area of concavity in choroid and should be differentiated from posterior staphyloma or scleral ectasia.
- FCE can coexist with Stargardt's disease apart from other macular dystrophies.
- FCE alone does not warrant treatment, but its association with choroidal neovascularisation mandates meticulous follow-up.

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FCE in SD is limited to a single case until now. To the best of our knowledge, our case is only the second in literature reporting such an association. We propose that degenerative process in macular dystrophies might be contributory to the FCE.[5–7]

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