Multimodal imaging in familial dominant drusen

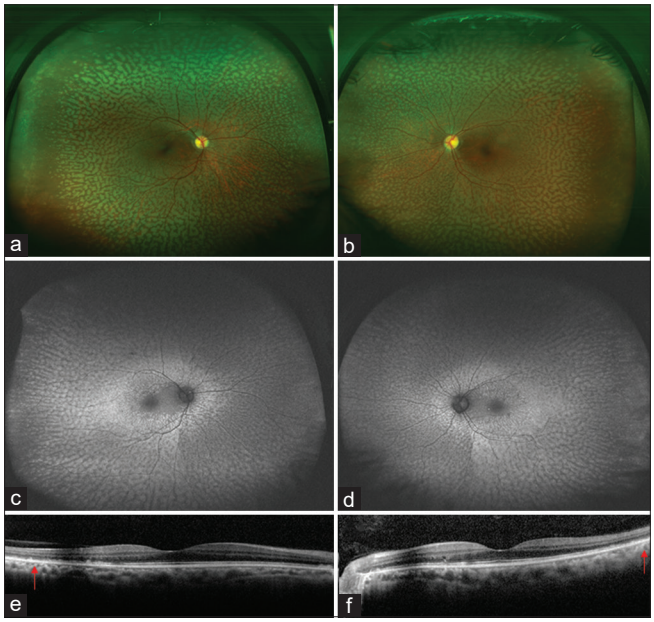

Figure 1: (a-e) Ultra widefield fundus photographs showing bilateral, numerous, yellowish-white, round, closely spaced lesions extending from the vascular arcades till the periphery, sparing a small island of the macula (a and b), Fundus autofluorescence revealed bright hyperautofluorescence of the lesions (c and d). Optical coherence tomography scans of both eyes showing thickening of RPE-Bruchs complex with dome shaped RPE elevations (red arrows) and relative noninvolvement of neurosensory retina corresponding to the drusen (e and f)

A 42-year-old female came for routine checkup with 20/20 vision. Fundus examination revealed bilateral, numerous, yellowish-white, round, and closely spaced lesions extending from the vascular arcades till the periphery, sparing a small island of macula, suggestive of familial dominant drusen [Fig. 1a and b]. Fundus autofluorescence revealed bright hyperautofluorescence of the lesions [Fig. 1c and d]. Optical coherence tomography scans showed thickened retinal pigment epithelium-bruchs membrane complex with localized elevations [Fig. 1e and f]. The disease is caused by mutation in the EFEMP1 gene on short arm of chromosome 2, which encodes Fibulin-3, an extracellular matrix glycoprotein. Most patients are asymptomatic and retain good vision.[1-3]

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 The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.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