Macular drusenoid deposits in X-linked retinoschisis

Dear Editor,

Though X-linked retinoschisis (XLR) is a common heredomacular degeneration, the diagnosis can be challenging due to high clinical variability,[1] particularly when the patient presents at an advanced age. Drusen-like macular deposits in middle-aged adults are suggestive of age-related maculopathy. We diagnosed XLR in such a patient with the help of electroretinography (ERG), spectral domain optical coherence tomography (SD-OCT), and autofluorescence imaging (AF).

A 48-year-old man presented with poor vision from childhood in both eyes. He gave no history of visual impairment in his family. His visual acuity was 20/200 OD with a correction of + 6.0 + 2.0 × 180, and 20/120 OS with + 3.5 − 2.0 × 165. On ocular examination, anterior segments were unremarkable OU. Fundi showed macular drusen-like deposits OU; OS also had vitreous veils [Fig. 1a, b]. SD-OCT (Cirrus, Carl Zeiss Meditec, Dublin, CA, USA) showed foveal atrophy OD with a few parafoveal cysts, and typical schitic cavities OS involving mainly inner nuclear and outer plexiform layers [Fig. 1c, d]. AF revealed an enlarged area of macular hypo-autofluorescence, more prominent OD, with a ring of hyper-autofluorescence OD, suggestive of metabolic stress and potential for progression of atrophy [Fig. 1e, f]. ERG showed classic reduced b/a wave ratio and reduced amplitude and prolonged implicit time for the 30-Hz flicker stimulation [Fig. 1g, j]. The patient was kept on observation.

OCT has been reported as a consistent diagnostic aid across a variable clinical spectrum of XLR, comparable to ERG.[1,2] increased macular AF signal has also been reported to be of diagnostic value in typical XLR.[2] Tsang *et al*., who first reported drusen-like deposits in XLR, relied mainly on electronegative ERG; OCT displayed only foveal atrophy, substantiated by low-density AF.[3] The discrepancy between AF and OCT in these studies probably reflects the difference in age of presentation and the corresponding changes from juvenile foveal schisis to atrophic thinning toward middle age.[1-3] Atchaneeyasakul *et al*. also described drusenoid deposits in a 30-year-old man with XLR; but OCT and AF findings were not shown.[4] This is the second report of drusenoid deposits in XLR with AF, OCT, and ERG documentation, and the first with SD-OCT findings. The clinical picture initially appeared to suggest age-related macular degeneration; disproportionately

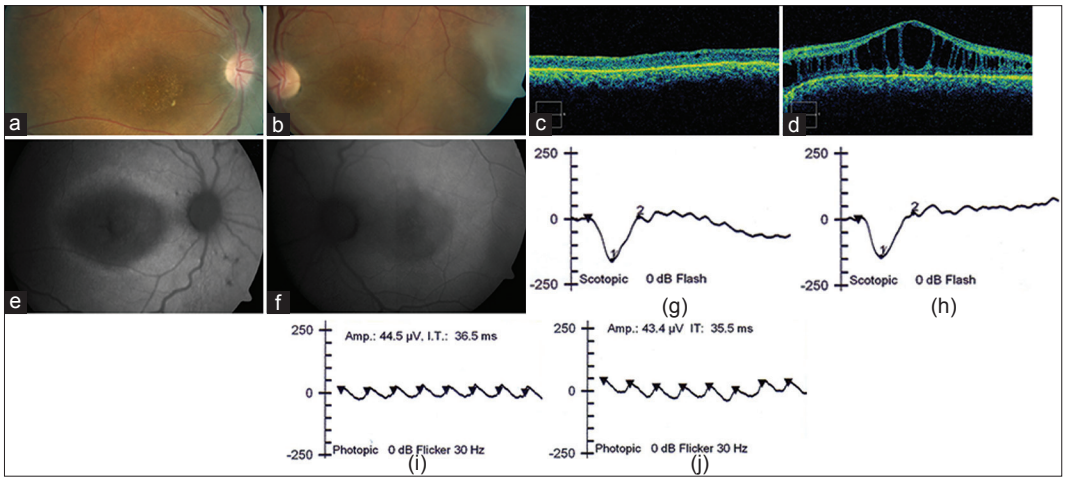

Figure 1: (a, b) Fundi OU show macular drusenoid deposits, more in OD; note the vitreous veil temporally OS. (c, d) Horizontal 6-mm spectral-domain OCT scans reveal generalized foveal atrophy OD, with minimal parafoveal cysts. OS shows diagnostic macular schisis. (e, f) Autofluorescence imaging exaggerated macular hypo-autofluorescence OU, darker and ringed by hyper-autofluorescence OD. (g, h) Electroretinogram shows b-wave suppression with relative sparing of a-wave on single-flash stimulus, and (i, j) a prolonged implicit time in the 30-Hz flicker response

poor vision OU and vitreous veils OS led us to perform OCT, AF, and ERG, which clinched the diagnosis. SD-OCT helped us to detect macular cystoid changes in OD as well, the residua of collapsed schitic cavities. Our case shows that degenerative changes can progress at different paces in the two eyes of a patient, and correlate well with SD-OCT, AF, and functional changes: The eye with greater severity of macular drusenoid deposits had foveal atrophy, more extensive and denser hypo-autofluorescence, and poorer vision. ERG, OCT, and AF complement each other as clinical diagnostic aids in XLR, depending on the severity of the condition and patient's age.

Manish Tandon, Dhananjay Shukla

Retina-Vitreous Service, Aravind Eye Hospital and Postgraduate Institute of Ophthalmology, Madurai, Tamil Nadu, India

Correspondence to: Dr. Dhananjay Shukla, Aravind Eye Hospital, 1 Anna Nagar, Madurai, Tamil Nadu, India.
E-mail: daksh66@gmail.com

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