Photo Essay

Nasal involvement in X-linked retinoschisis

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Key words: Swept source optical coherence tomography, ultra-wide field imaging, X-linked retinoschisis

An 11-years-old male presented with steadily decreasing vision over 5 years. The best-corrected visual acuity was 20/100 right eye (OD) and 20/200 left eye (OS). Family history and anterior segment examination in both eyes (OU) were unremarkable. Dilated fundus examination OU revealed foveal schisis with typical cartwheel appearance at macula and inferotemporal retinal schisis reaching up to the inferotemporal vascular arcades [Fig. 1a and b]. The macula was displaced superiorly in OD. Swept-source optical coherence tomography (OCT, Topcon Inc.,) confirmed the presence of cystoid spaces separated by vertical pillars of retinal tissue suggestive of retinoschisis in the inner nuclear layer [Fig. 2a and b] in the macular area OU. The height of foveal schisis was greater in OS, explaining the worse visual acuity in that eye. Due to poor visual acuity, the patient had eccentric fixation OU in the area nasal to the fovea. The 12 mm long radial scans (covering almost 50° of retina) thus captured the area nasal to the optic disc as well. Interestingly, the retinoschisis extended to the area nasal to the disc as well [Fig. 2a and b]. A full-field electroretinogram showed selectively reduced b-wave amplitude, confirming the diagnosis of X-linked retinoschisis (XLRS). The patient was advised low vision aids and regular follow-up.

Discussion

The macular schisis in XLRS involves deeper and multiple layers, the most commonly affected being inner nuclear layer.[1] In contrast, the inferotemporal schisis usually occurs in the superficial retina. The macular schisis in XLRS is more extensive in its distribution when assessed with OCT as compared to clinical examination and may extend up to the vascular arcades or even beyond.[2,3] Gregori *et al*. recently used montage technique for spectral domain OCT to create wide-field OCT and reported that schisis may extend outside of macula near the arcades.[3] Two out of 11 eyes in their series had small cystoid spaces in the inner and outer nuclear layer nasal to optic disc. The present case,

however, had prominent retinoschisis involving the inner nuclear layer in the area nasal to optic disc. Although nasal and peripapillary retinoschisis have been demonstrated in patients with glaucoma, involvement of nasal retina is not well known in XLRS.[4,5]

XLRS is caused by mutations in *RS1* gene, which encodes a protein complex – retinoschisin. Retinoschisin functions as a cell adhesion protein.[6] Mutations of *RS1* disrupt subunit assembly of protein structure and cause XLRS. The reason for selective involvement of macula and inferotemporal retina, however, is not known. This case highlights that retinal involvement may not be restricted to one sector and may be more diffuse.

Ultra-wide-field imaging (Optos Inc.) is a useful tool in the assessment and documentation of pediatric peripheral retinal disorders.[7] It is an excellent tool for documenting the progression of peripheral changes since such patients frequently have poor vision and are unable to cooperate in the generation of montage images.

To conclude, this case highlights the utility of wide-field imaging in better phenotypic characterization of XLRS and may help us gain a better understanding of the disease in the future.

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Conflicts of interest

There are no conflicts of interest.

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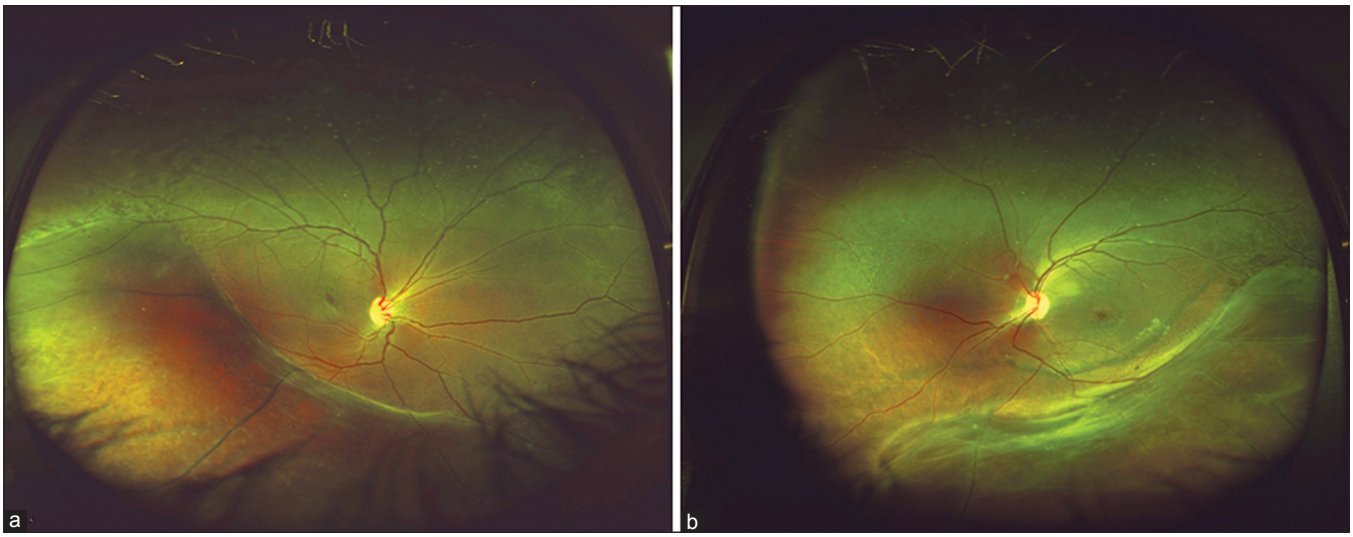

Figure 1: Ultra-wide-field color photograph of the right (a) and the left eye (b) showing macular cartwheel appearance and inferotemporal retinoschisis. The macula is displaced superiorly in the right eye

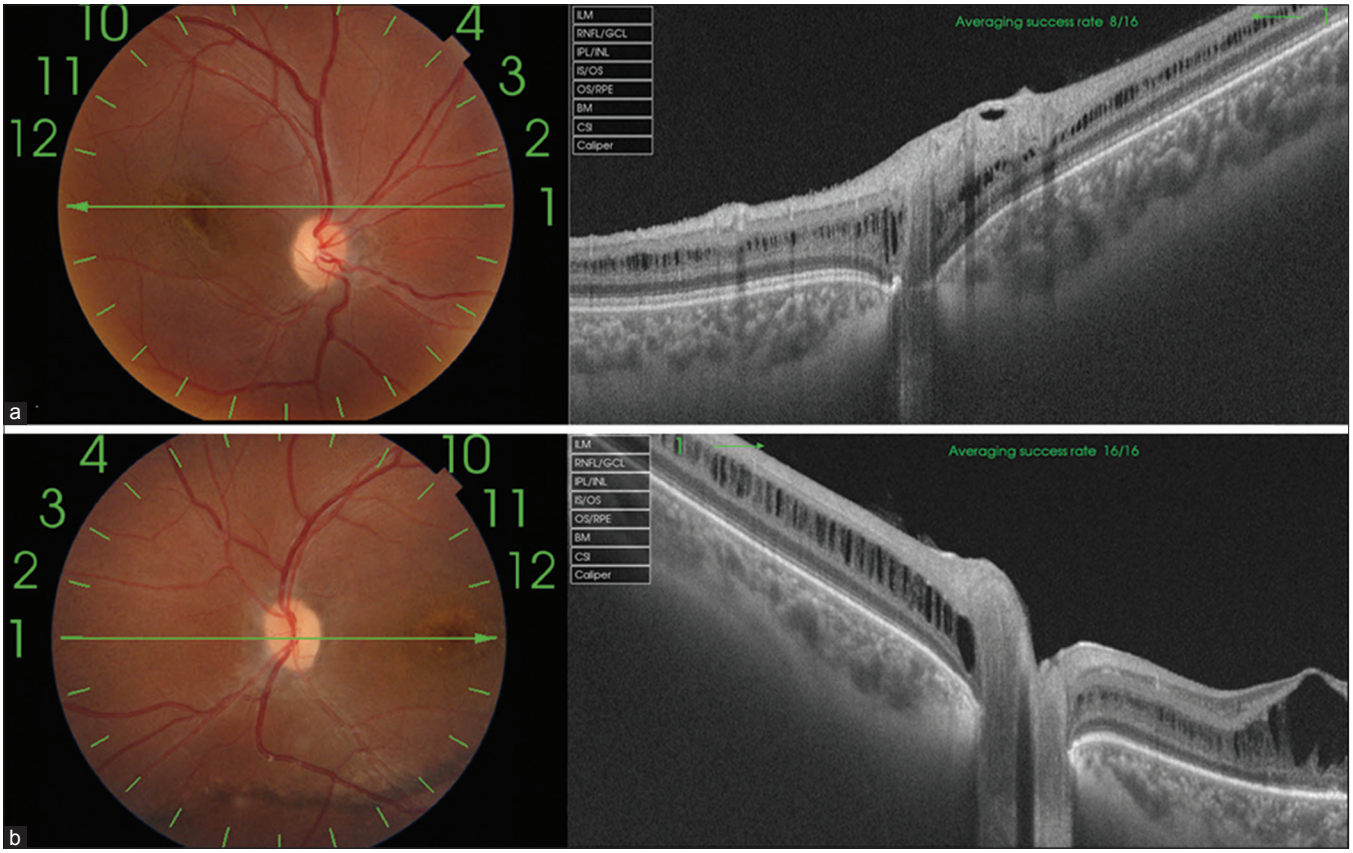

Figure 2: The 12 mm long radial swept-source optical coherence tomography scans of the right (a) and the left eye (b) show prominent inner nuclear layer schisis in the area nasal to the optic disc in addition to the foveal retinoschisis

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