Multimodal imaging of bilateral macular hole in X-linked retinoschisis

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DESCRIPTION

A 25-year-old man presented with decreased vision in both eyes over the past 10 years. On examination his best corrected visual acuity was 20/100 and N18 in both eyes. Anterior segment examination was unremarkable. Fundus examination of both eyes was remarkable for full thickness macular hole (figure 1A,B). Fundus autofluorescence (FAF) revealed hypoautofluorescence at the centre of the macula and hyperautofluorescence surrounding it (figure 1C,D). Optical coherence tomography (OCT) showed full thickness macular hole with schitic changes at the level of outer nuclear and inner nuclear layers with loss of outer retinal layers (figure 2A,B). The peripheral examination of the fundus did not reveal any other areas of schisis or degenerative changes. Full field electroretinogram (ERG) showed electronegative pattern of b wave in scotopic bright flash ERG (figure 2C,D). Retrospective evaluation of family history revealed macular hole with retinoschisis in younger brother. Based on positive family history, retinoschisis with macular hole and an electronegative ERG, a diagnosis of X-linked retinoschisis (XLRS) with macular hole was made. The patient was advised to use magnification devices for near vision. Surgical intervention for macular hole was not advised as the vision was stable over the last 5 years and OCT showing loss of outer retinal layers, with FAF showing hypoautofluorescence at the centre which is suggestive of low-visual potential even after surgery.

Juvenile XLRS is a congenitally inherited retinal degenerative condition.¹⁻³ It is due to mutation in RS1 gene which codes for protein retinoschisin, responsible for integrity of photoreceptors, bipolar

Figure 2 Optical coherence tomography of both eyes (right eye: A and left eye: B) showed full thickness macular hole with schitic changes at the level of outer nuclear and inner nuclear layers and loss of outer retinal layers. Full field electroretinogram (ERG) showed slight reduction in amplitude of a wave with electronegative pattern of b wave in scotopic bright flash ERG (right eye: C and left eye: D).

cells, and in turn the retinal layers.³⁻⁶ Men are much more commonly affected with women typically being carriers, however both genders can be affected.^{1 2} XLRS is usually present at birth but manifests in the first to second decade. Affected men present with moderate visual loss and schisis at various levels of retinal layers.³ Foveal schisis is present in almost all patients, however the peripheral schisis is present in only about half of them.¹ OCT shows characteristic finding of splitting of retinal layers at various levels.^{7 8} The ERG shows electronegative pattern in which the amplitude of the b wave is less than that of a wave.^{8 9}

Patient with XLRS can present with complications such as vitreous haemorrhage, retinal detachment and rarely macular holes.^{13 10-13} Factors found to be responsible for occurrence of macular hole in XLRS are foveal schisis, degenerative cysts, tangential traction, derroofing of the degenerative schitic cysts.¹⁰⁻¹⁴ Cases with vitreoretinal traction leading to macular hole and retinal detachment have also been reported.¹³

As per our knowledge, only one case of bilateral macular hole in XLRS with poor vision has been reported.¹⁴ Our case was found to have good visual

Figure 1 Fundus of both eyes (right eye: A and left eye: B) showed full thickness macular hole. Fundus autofluorescence of both eyes (right eye: C and left eye: D) showed hypoautofluorescence at the centre of macula and hyperautofluorescence surrounding it.

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Learning points

- ▶ X-linked retinoschisis (XLRS) can rarely manifest as a bilateral macular hole in young patients.
- ▶ Family history and multimodal imaging help in arriving at diagnosis of XLRS with macular hole in young patients.

Images in...

acuity of 20/100, N18 in spite of large holes and complete loss of photoreceptors and retinal pigment epithelium loss in the area of hole documented by OCT and FAF. This good visual acuity could be due to eccentric foveal fixation which has been reported in macular dystrophies.¹⁵

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