CASE REPORT

Lamellar macular hole in X linked retinoschisis

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SUMMARY

X linked retinoschisis (XLRs) is the most common juvenile onset retinal degeneration. The disorder leads to poor vision in old age. Complications, however, can lead to earlier loss of vision in this condition. This report describes two patients of XLRs, who had presented with poor vision because of having had a lamellar macular hole at a young age. Lamellar macular holes are rare and have never been reported to cause early onset poor vision in XLRs.

BACKGROUND

X linked retinoschisis (XLRs) is a bilateral progressive retinal disorder and is one of the most common causes of retinal degeneration and decreased vision in young males. Stellate foveal schisis is pathognomonic of the disorder, while peripheral retinoschisis is seen in up to 50% of cases. The decrease in visual acuity in XLRs could be due to the presence of retinal detachment, vitreous haemorrhage, progression of peripheral schisis towards macula and formation of a full thickness macular hole. A lamellar macular hole is a rare occurrence in patients with XLRs and may cause decreased vision at a young age in these patients. We report two patients of XLRs who presented with decrease of vision due to formation of a lamellar macular hole.

CASE PRESENTATION

Case 1

A 10-year-old otherwise healthy boy presented with decrease of vision in his right eye for the past 3 months. There was no history of similar symptoms in his other family members. The best-corrected visual acuity (BCVA) was 3/60 and 6/24 in right and left eyes, respectively, without any refractive error. Anterior segments were unremarkable. A dilated fundus examination showed a macular hole in the right eye and typical cartwheel appearance in the left eye (figure 1A, B). Dendritic lesions were seen in both eyes; however, no peripheral schisis was noted in either eye. Spectral domain optical coherence tomography (SD-OCT) showed the presence of a lamellar macular hole along with a large schitic cavity in the right eye (figure 1C). The left eye SD-OCT showed large schitic spaces in the outer plexiform layer, separated by pillars of retinal tissue (figure 1D). Electroretinogram (ERG) showed selectively reduced b-wave amplitude confirming the diagnosis of XLRs. The patient was offered pars plana vitrectomy, which the parents refused. The patient was advised regular follow-up.

Case 2

A 13-year-old boy presented with decrease of vision in his right eye for the past 6 months. He had a history of subnormal vision in both eyes since childhood. BCVA was 5/60 and 6/24 in right and left eyes, respectively, with -1.5D sphere. Anterior segments in both eyes were normal. A dilated fundus examination showed a macular hole in the right eye and a typical cartwheel appearance in the left eye (figure 2A, B). SD-OCT showed schitic spaces in the outer plexiform layer in both eyes (figure 2C, D). In addition, a lamellar macular hole was noted in the right eye (figure 2C). ERG showed negative waveform confirming the diagnosis of XLRs. The patient was advised to follow-up regularly.

DIFFERENTIAL DIAGNOSIS

Foveal schisis of Goldmann-Favre vitreoretinal degeneration can closely mimic the foveoschisis in XLRs. The absence of nyctalopia and pigment clumping, and presence of better visual acuity, vitreous veils and relatively preserved a-wave in ERG point towards the diagnosis of XLRs in this report.

In the presence of myopia, myopic foveoschisis can imitate the foveoschisis of XLRs, however, associated features of XLRs are not seen in myopia. In our cases, refraction ruled out myopia. Other differential diagnoses include cystoid macular oedema and degenerative retinoschisis.

DISCUSSION

XLRs is an inherited retinal disorder stemming from a mutation of the RS1 gene on X chromosome.¹ Most patients present with decreased vision, due to macular changes, in the first or second decade. Visual acuity subsequently tends to remain stable until the fifth to sixth decade, when progression of macular changes to macular atrophy may occur. This may lead to legal blindness by the sixth to seventh decade.¹ Complications such as tractional or rhegmatogenous retinal detachment, vitreous haemorrhage, haemorrhage within a large schitic cavity and intraretinal splitting, can lead to an abrupt fall in vision in some cases.²

Rarely, a full thickness macular hole (FTMH) can occur in a patient of XLRs, which can contribute towards decrease of vision of these patients. Only a handful of reports of FTMH in XLRs exist in the literature.³⁻⁶ Each of these patients with a FTMH had visual acuity below 6/60. Deroofing of the retinal cyst formed by coalescence of schitic cavities is proposed to be the cause of macular hole formation in these eyes. Vitreomacular traction may also play a role in the formation of macular holes.⁴

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Figure 1 Colour fundus photograph of the right eye (A) showing a large macular hole and left eye (B) showing stellate foveoschisis. Horizontal spectral domain optical coherence tomography (SD-OCT) through the fovea showing the lamellar macular hole in the right eye (C) with wide separation of inner and outer retinal layers. SD-OCT of the left eye (D) showing typical foveoschisis with tissue pillars connecting the inner and outer retinal layers.

Lamellar macular holes have been reported very rarely in XLRS.^{7,8} The patient described by Leng⁷ had very poor vision (20/120). In a series reported by Wang *et al*,⁸ two eyes had lamellar macular holes (one had vision of 20/200 while vision was not mentioned for the other). Both our patients also had poor vision. The occurrence of lamellar macular holes in XLRS gives further credit to the derroofing theory for the formation of FTMH. This may also support the use of medical therapy to

reduce the macular schisis. This will prevent schisis from derroofing, that can lead to visual loss at a young age in these patients. Long-term follow-up of these cases may provide further insights into the formation of FTMH in patients with XLRS.

To conclude, lamellar macular holes are rare in patients with XLRS and may cause decreased vision at a young age in these patients.

Figure 2 Colour fundus photographs of right eyes (A) showing lamellar macular holes and of left eyes (B) showing stellate foveoschisis. Spectral domain optical coherence tomography (SD-OCT) of right macula (C) showing a lamellar macula hole and schisis involving the macula in the rest of the scan. Left eye SD-OCT showing foveoschisis typical of X linked retinoschisis.

Learning points

- ▶ Increasing height of foveal schisis in patients with X linked retinoschisis (XLRs) can cause deroofing, leading to lamellar/full thickness macular holes.
- ▶ Macular holes may lead to visual loss at an earlier age in patients with XLRs.
- ▶ Medical therapy in the form of topical carbonic anhydrase inhibitors may thus be justified for patients with XLRs to decrease the height of foveal schisis thereby reducing the chances of deroofing.

Competing interests None declared.

Patient consent Obtained.

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