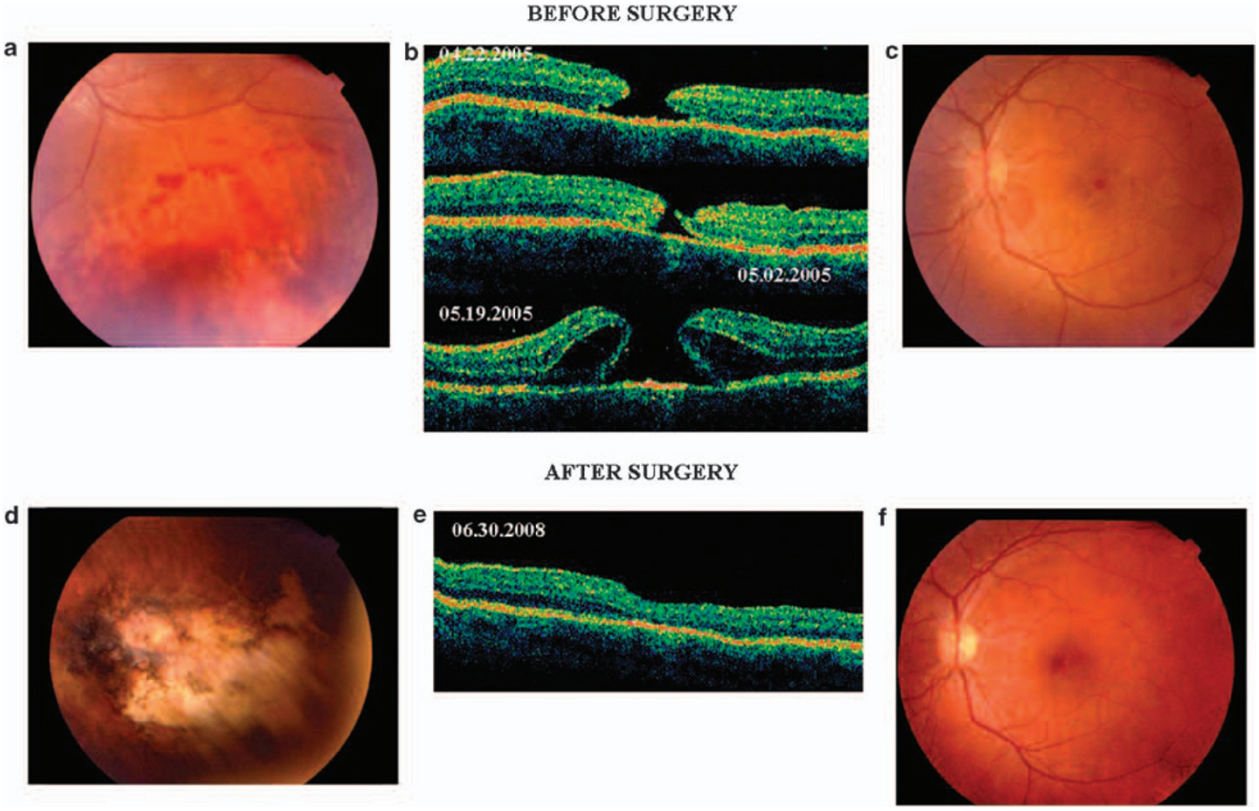

Figure 1 (a) The colour fundus photograph before surgery shows retinal and choroidal haemorrhages; (b) OCT scans show the development of a full-thickness macular hole; (c) the colour fundus photograph shows a full-thickness macular hole; (d) the colour fundus photograph after surgery shows that the chorioretinal peripheral lesion healed with the development of white, fibrous scar tissue with pigment at its edges; (e and f) colour fundus photograph and OCT scans show the closure of macular hole after the vitrectomy and ILM peeling.

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Sir,

An unusual fundus phenotype of inner retinal sheen in X-linked retinoschisis

X-linked retinoschisis (XLRS) is the leading cause of juvenile macular degeneration in men.[1,2] The disorder is associated with mutations in the *RS1* gene.[3] The usual clinical presentation is with reduced visual acuity, strabismus, or less commonly vitreous haemorrhage. The macula typically has a stellate appearance resulting from microcystic spaces radiating from the fovea and peripheral schisis is present in about 50% of cases. Macular atrophy often develops with age. Abnormal pigmentation, subretinal fibrosis, peripheral white flecks and white dots at the macula have also been described.[2,4] Ocular coherence tomography (OCT) usually reveals cyst formation and intraretinal cleavage.[2] Full-field electroretinograms (ERGs) typically show evidence of generalised inner retinal dysfunction.[1,2,4] In this report, we describe a child with an *RS1* mutation and an unusual inner retinal sheen with none of the typical fundus features or OCT findings associated with XLRS.

Case report

A 10-year-old male child born to parents who were first cousins was found to have reduced vision at a routine

visit to the optician. There was no relevant family history. When examined in the clinic, best-corrected visual acuity was 6/9 in each eye. Fundus examination showed an unusual inner retinal sheen (Figure 1a). The macula and fovea appeared normal. Fundus autofluorescence (AF; Figure 1b) and optical coherence tomography (OCT; Figure 1c) were normal. International standard full-field ERGs[5] showed dark-adapted bright-flash ERG b-waves that were subnormal with minimal a-wave reduction (Figure 2). Light-adapted 30 Hz flicker ERGs were mildly delayed and reduced, and photopic transient ERGs had a low b : a-wave ratio. The ERG findings were consistent with generalised retinal dysfunction of both rod and cone systems with a locus of dysfunction that was postphototransduction and raised the possibility of XLRS. Molecular genetic analysis revealed the c304C > T (pArg102Trp) mutation in exon 4 of the *RS1* gene;[3,6] genetic testing of the mother was negative, consistent

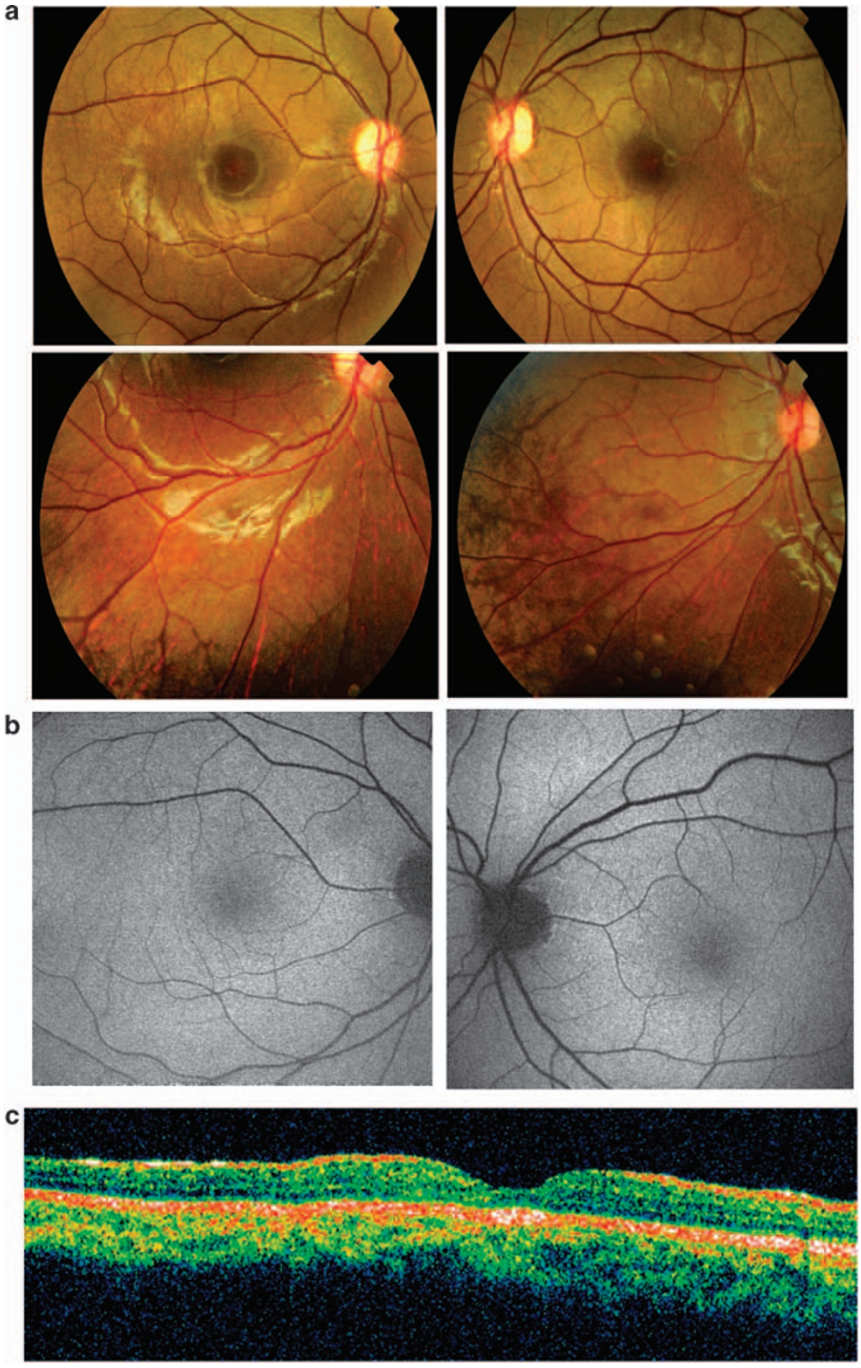

Figure 1 Fundus photographs of the posterior pole (a) showing an inner retinal sheen. Fundus autofluorescence was normal (b). OCT of the macula revealed no evidence of schisis (c).

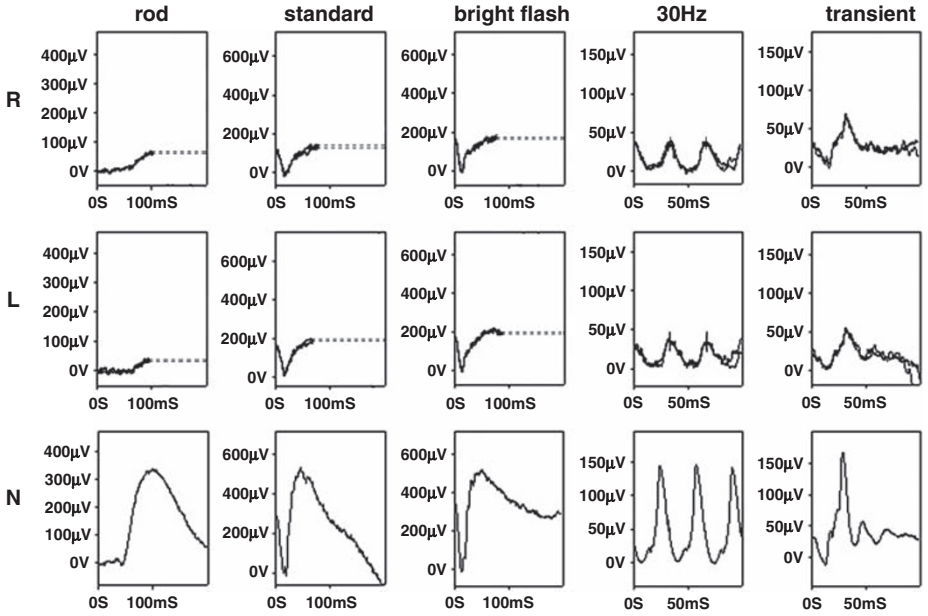

Figure 2 International standard full-field ERGs from the right (R) and from the left (L) eye of the patient, and from a normal subject (N) for comparison. The bright flash ERG was recorded to a flash 0.6 log units stronger than the International standard flash ($3.0\,cd.s.m^{-2}$),[5] to better show the a-wave. Broken lines replace blink artefacts.

with this being a *de novo* mutation. Identical missense changes have been shown to result in absent secretion of mutant RS1,[7] and the phenotype is known to be variable,[6] possibly due to environmental and/or genetic modifiers.

Comment

The majority of children with XLRS show foveal schisis, which is evident clinically and on OCT. Some cases, such as the child reported here, have an atypical fundus appearance. Electroretinography, showing evidence of inner retinal dysfunction, is important for the correct diagnosis. A possible diagnosis of XLRS should be considered in male patients present with an abnormal inner retinal reflex without the evidence of foveal or peripheral schisis.

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