ERG showed cone–rod dysfunction. OCT demonstrated thinning of the outer retinal layers, in particular the outer nuclear layer and the outer photoreceptor segments. Both patients showed amelogenesis imperfecta (Figure 1). Phenotype details in comparison with the reported patients with *CNNM4* mutations are listed in Table 1. The same homozygous mutation c.1312dup; p.Leu438Pro*fs**9 was found in the affected patients (III:4, III:5). The mother (II:4) was heterozygous for this sequence alteration in *CNNM4* (Figure 2). The mutation within the cystathionine beta-synthase domain most likely results in a premature termination codon and nonsense-mediated mRNA decay of the mutant transcript. No mutation in the *ABCA4* gene was identified.

A similar dental phenotype with the characteristics of AI is described in all publications of patients with *CNNM4* mutations. In his phenotype dissection, Jalili described anterior open bite (AOB) in 2/30 and posterior open bite in 1/30 'type A' patients, whereas AOB was present in all three patients of the 'type B' phenotype. No open bite abnormality was seen on examination in our two patients and in one of the two patients reported by Luder *et al*.[5]

The intra-familial variability presented here is not consistent with a strict phenotype–genotype correlation, and may also argue against a rigid phenotype differentiation.[4] As the patients with 'type B' phenotype were examined at a younger age than the patient with 'type A', it is possible that those patients with 'type B' may have shown minimal macular signs as most of them had a visual impairment and signs of a cone–rod dystrophy.

The proposed strict differentiation between type A and B may not be applicable to all affected patients and families.

Conflict of interest

The authors declare no conflict of interest.

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Sir,
Rupture of Descemet's membrane secondary to presumed non-accidental injury

We present the first case of multiple uniocular breaks in Descemet's membrane secondary to presumed non-accidental injury (NAI).

Case report

The first of twins, born at 35 weeks by spontaneous unassisted vaginal delivery, presented with a 2-week history of unexplained corneal haze in her left eye at the age of 4 months. Topical antibiotics and steroids were

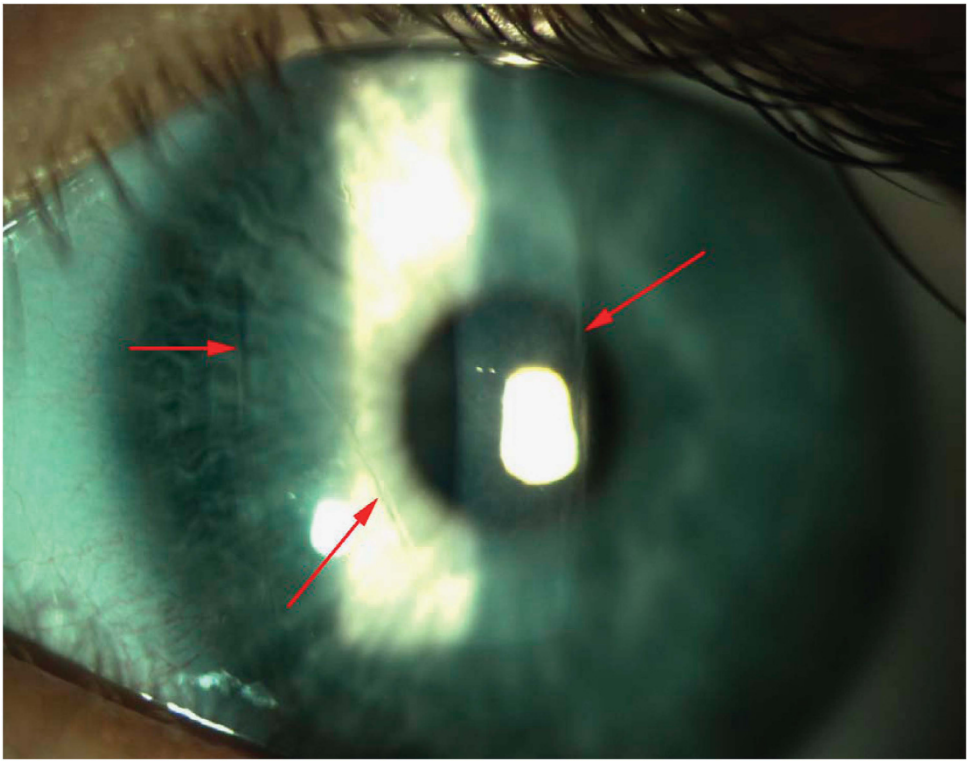

Figure 1 Left cornea on retroillumination demonstrating breaks in Descemet's membrane highlighted with red arrows.

prescribed and she subsequently failed to attend. At 8 months, she was admitted to a second hospital with subdural haemorrhage, rib fractures, retinal haemorrhages and facial bruising. A diagnosis of NAI was made.

At 16 months she attended this unit with left amblyopia, eccentric fixation and esotropia. Her visual acuity was 0.2 logmar in the right eye and 1.0 in the left (Kay pictures, UK). There was no significant refractive error. She had a faint stromal scar medially in the left eye with multiple Descemet's breaks seen on slit-lamp, the largest being vertically in the visual axis (Figure 1). Her cornea was of normal diameter, spherical, with no evidence of keratoconus. Her intraocular pressure was normal. No abnormality was detected in the right eye. Connective tissue disorders were excluded by a paediatrician. Her ocular appearance is unchanged on follow-up.

Discussion

Ocular symptoms and signs are the presenting feature in 1–5% of child abuse cases.[1] It is recognised that 31% of cases are misdiagnosed on their first presentation and many of them go onto suffer repeated and escalating episodes of violence,[2] as in this case.

It is likely that the Descemet's breaks in our case were not seen at presentation due to the overlying corneal haze secondary to acute hydrops, but that they were there from the unexplained injury at age 4 months. Indeed the history of sudden onset of corneal haze would be explained by the Descemet's breaks. Other causes of Descemet's breaks, such as forceps injury at birth,[3] prolonged labour, keratoconus and congenital glaucoma[4] were excluded by history and examination. Posterior polymorphous dystrophy was also considered and excluded.[5]

We recommend increased awareness and the inclusion of non-accidental injury as a differential diagnosis for unexplained Descemet's breaks with corneal haze in a child.

Conflict of interest

The authors declare no conflict of interest.

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