children below nine years of age; it should have been the 10th cause. Intracranial injuries excluding skull fractures are the 6th and skull fractures excluding vault and base fractures are the 12th most common causes.

(3) Dr Lantz first presented the case in 2002 (Lantz P, Sinal S. Perimacular retinal folds in non-abusive head trauma. *Fourth National Conference on Shaken Baby Syndrome*. Salt Lake City, UT, 2002). One of our authors (AVL) had the privilege of attending this presentation after which Dr Lantz kindly sent him images from the original histological preparations including images, which did not appear in the published description.[2]

(4) There is copious evidence to support the observation that retinal haemorrhage is uncommon in accidental head trauma. This evidence is well beyond the scope of this reply and the reader is referred elsewhere.[3,4]

(5) Three of the four children with retinal haemorrhage had few, small haemorrhages largely confined to the posterior pole. The fourth child also had microscopic (ie, not clinically detectable) haemorrhage and a few that were visible on gross inspection at the ora serrata. That child also had significant neck injuries consistent with atlanto-occipital disruption. It would be improper to suggest that this high percentage (4/9, 44.4%) should be used in any way as a reflection of the potential rate of haemorrhages in other types of accidental head trauma. The rate is entirely inconsistent with a literature that examines tens of thousands of eyes in victims of non-abusive injury. Although the high percentage may simply be a reflection of small sample size, the higher prevalence may simply reflect, if confirmed on a larger sample size, the severe fatal crush injury mechanism (head out of window when car rolled, car rolled over head, unrestrained back seat passenger, and pedestrian struck by car), and even then it is remarkable how mild the retinopathy is and how it differs from that seen in two-thirds of shaken babies.[5]

Although we certainly value the examination of new data and outlier reports,[6] we regret that Drs Lantz and Stanton reject the large body of medical literature, which not only supports the current understanding of the pathophysiology of retinal haemorrhage in abusive head trauma but also weighs very heavily against the paucity of evidence to the contrary.

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Sir,

A novel peripherin/RDS mutation resulting in a retinal dystrophy with phenotypic variation

Peripherin/RDS is a structural transmembrane glycoprotein that contributes to the formation and stabilisation of rod and cone photoreceptor outer segment discs. Mutations in the peripherin/RDS gene can result in generalised retinal dystrophies or macular dystrophies[1,2] and are known to cause variable manifestations within families. We describe a novel mutation in exon 2 of the peripherin/RDS gene resulting in a three amino-acid deletion and causing a variable retinal phenotype within a three-generation family.

Case reports

Case 1

A 30-year-old man presented with a history of nyctalopia and reduced-peripheral visual fields. His best-corrected visual acuities were 6/6 in each eye. Anterior segments were normal on slit-lamp examination. Fundus examination revealed punctate hyperpigmentation in the peripheral retina (Figure 1). Colour vision was normal. Electroretinograms showed a reduction in light-adapted

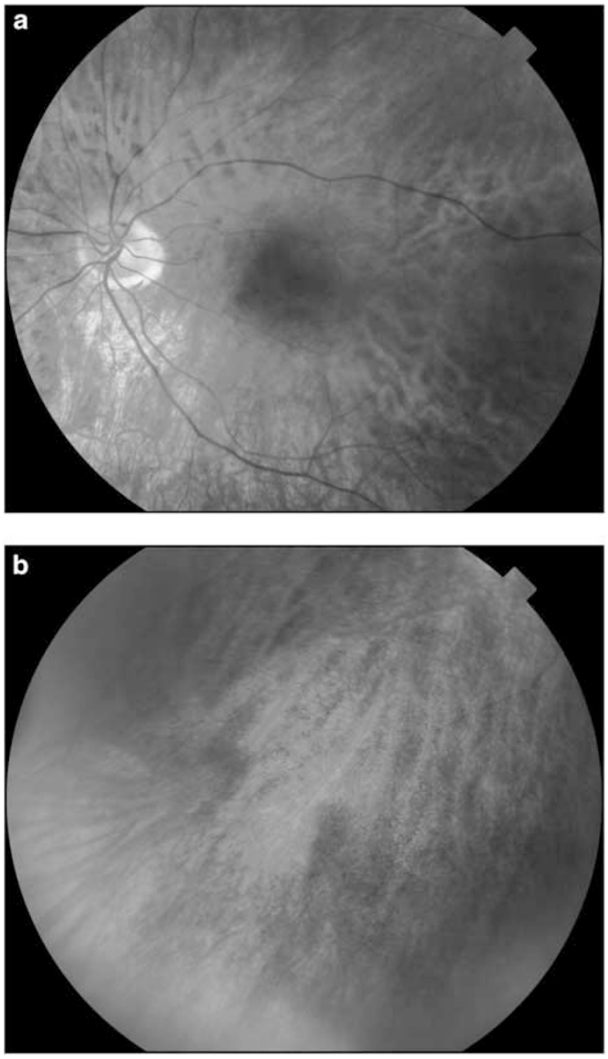

Figure 1 Posterior pole (a) and inferior nasal retinal view (b) of case 1.

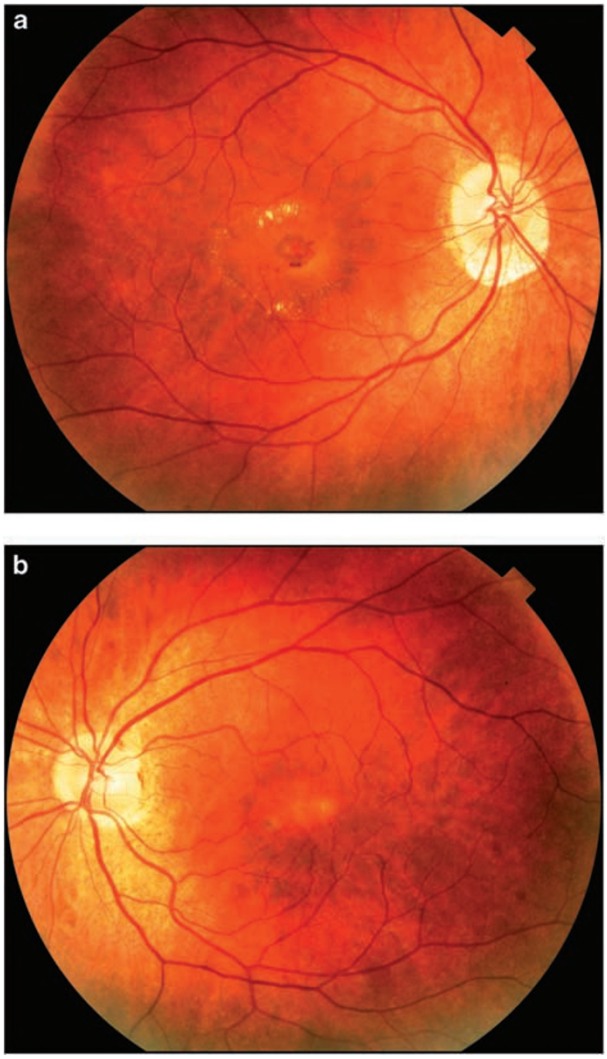

Figure 2 Posterior pole of case 2 (a) Right eye showing haemorrhage in the right macula with fine exudates and possible subretinal fluid. (b) Left eye showing a vitelliform lesion with atrophic changes.

responses and markedly attenuated responses on dark adaptation.

Case 2
A 61-year-old woman (mother of case 1) developed visual distortion at the age of 59. Initially when seen, she was found to have bilateral vitelliform macular changes associated with atrophy in the left eye. After 12 months, she developed haemorrhage, exudation and subretinal fluid at the right macula (Figure 2). Fundus fluorescein angiogram demonstrated a classic subretinal neovascular membrane, and photodynamic therapy was commenced (Figure 3). Electroretinogram showed a normal light- and dark-adapted responses and the Electro-oculogram was subnormal.

Case 3
The 86-year-old father of case 2 was diagnosed with retinitis pigmentosa many years ago and described poor vision since his 20s. According to him, his father also had poor vision for many years (Figure 4).

Mutation analysis of the coding region of the RDS/peripherin gene revealed c.618–626 del9 mutation in exon 2 of the RDS/peripherin gene in cases 1 and 2. This mutation deletes three amino acids p.207 Asp, 208 Gly, and 209 Val in frame.

Comment
Mutations involving these three amino acids have previously been reported causing autosomal dominant retinitis pigmentosa.[3,4] To our knowledge, this is the first report of this three amino-acid deletion that interestingly causes a highly variable phenotype within the same family. In addition, it highlights choroidal neovascularisation as a potential complication.

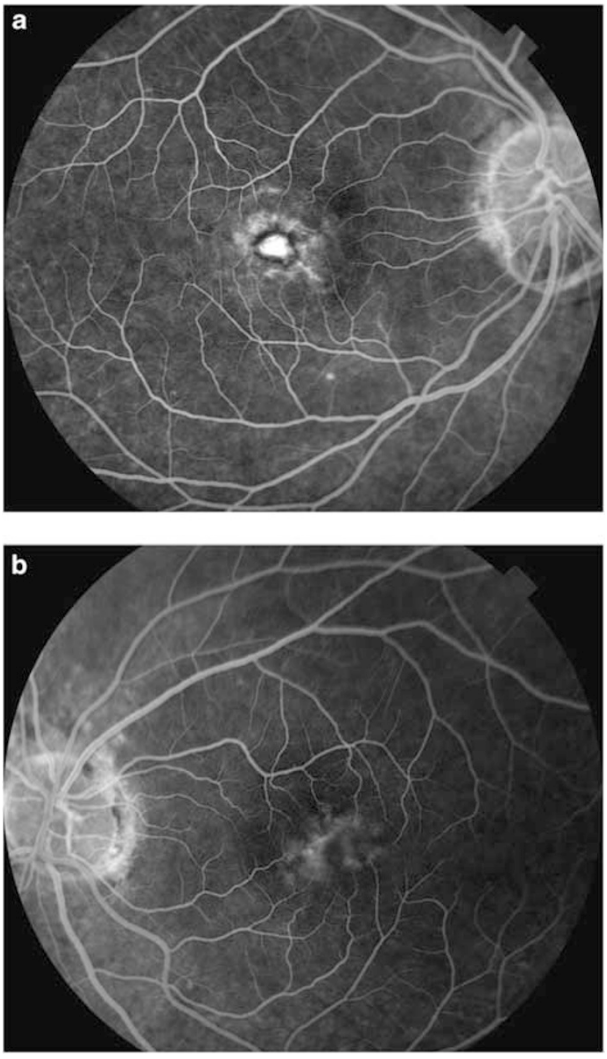

Figure 3 (a) Fundus fluorescein angiogram of case 2 showing the subretinal neovascular membrane in the right macula. (b) Angiogram of left macula.

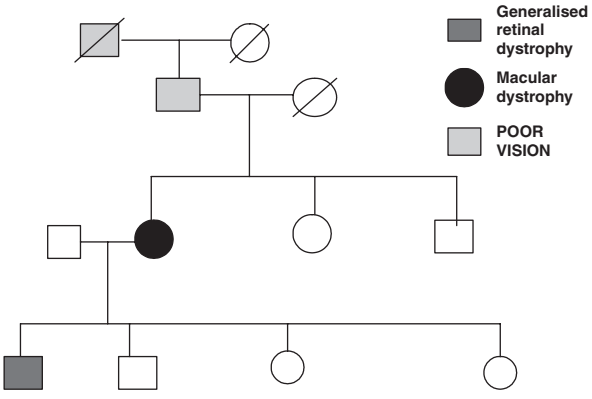

Figure 4 Pedigree of the family with autosomal dominant inheritance with retinal and macular phenotypes.

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Sir,

Necrobiotic xanthogranuloma masquerading as posterior scleritis

Necrobiotic xanthogranuloma is a rare skin disease that may herald eye complications. We report the presentation of posterior scleritis in a patient 2 years after a skin biopsy-proven diagnosis of necrobiotic xanthogranuloma.

Case report

A 47-year-old Caucasian lady presented with a 3-day history of reduced vision in her right eye, redness, watering, photophobia, pain, and headache. She was already under regular follow-up for blepharitis. She had necrobiotic xanthogranuloma skin lesions proven by biopsy on her upper and lower limbs (Figure 1). She also had raised ESR, IgG-κ paraproteinaemia, neutropenia, lymphopenia, and low complement C4.

Visual acuities were 6/18 right eye and 6/5 left eye. She had bilateral anterior blepharitis and anterior segments were otherwise unremarkable. Fundoscopy revealed bilateral choroidal folds and right cystoid macular oedema.

Ultrasonography showed irregular thickening of the sclera in both eyes (Figure 2). Magnetic resonance imaging of the orbits showed bilateral diffuse-enhanced thickening of posterior sclera but no orbital abnormality. A diagnosis of bilateral posterior scleritis associated with