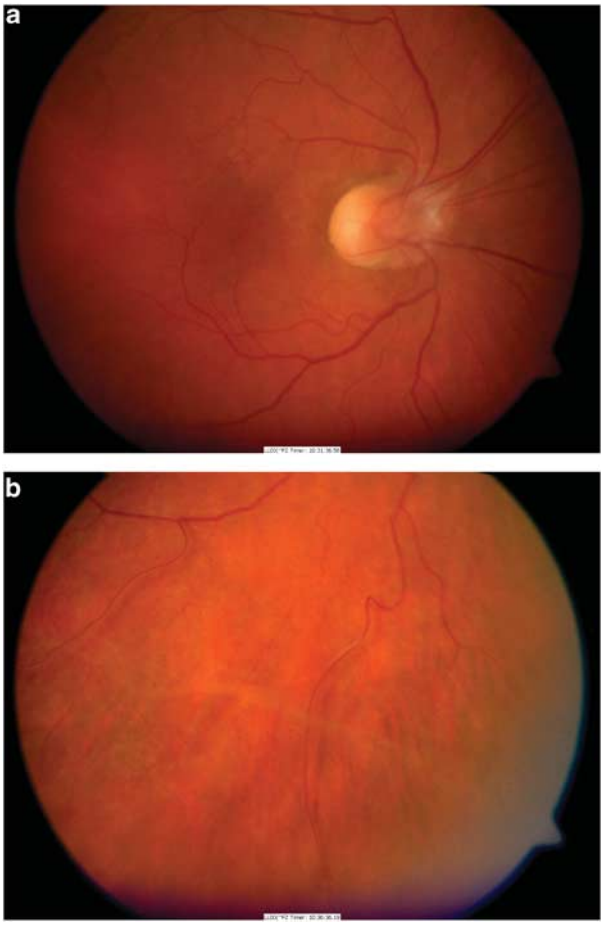

Figure 1 (a) A nasally dragged disc and retinal vessels. (b) Intact avulsed retinal vessels floating freely in the vitreous cavity.

avulsed retinal vessels floating freely in the vitreous cavity was present (Figure 1b). She had no retinal tear or detachment. OCT demonstrated areas of retinoschisis. Following review by the vitreoretinal team, a policy of observation was adopted. To date her visual acuity is 6/9, with no retinal detachment.

Comment

Various vitreoretinal complications have been described in the 'Boomer ROP' generation (born between 1940 and 1980).[4] Premature newborn survival improved during this time, but no defined treatment protocol was available for ROP giving rise to adult patients with various late-onset fundus findings. These include dragging of the retina, retinal detachment, retinal folds, lattice-like degeneration,[4] non-neovascular vitreous haemorrhage,[5] and myopia.[4]

Occasionally elevated blood vessels have been demonstrated secondary to vitreous traction.[6] We postulate that in this case separation of intact retinal vascular arcades from the retina occurred because of antero–posterior mechanical forces on already dragged retinal vessels under tension when PVD occurred. Kingham also advocated conservative management for such cases.

Conflict of interest

The authors declare no conflict of interest.

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Eye (2011) **25,** 1097–1098; doi:10.1038/eye.2011.86; published online 29 April 2011

Supplementary Information accompanies the paper on Eye website (http://www.nature.com/eye)

Sir,

Mizuo-Nakamura phenomenon in Oguchi disease due to a homozygous nonsense mutation in the *SAG* gene

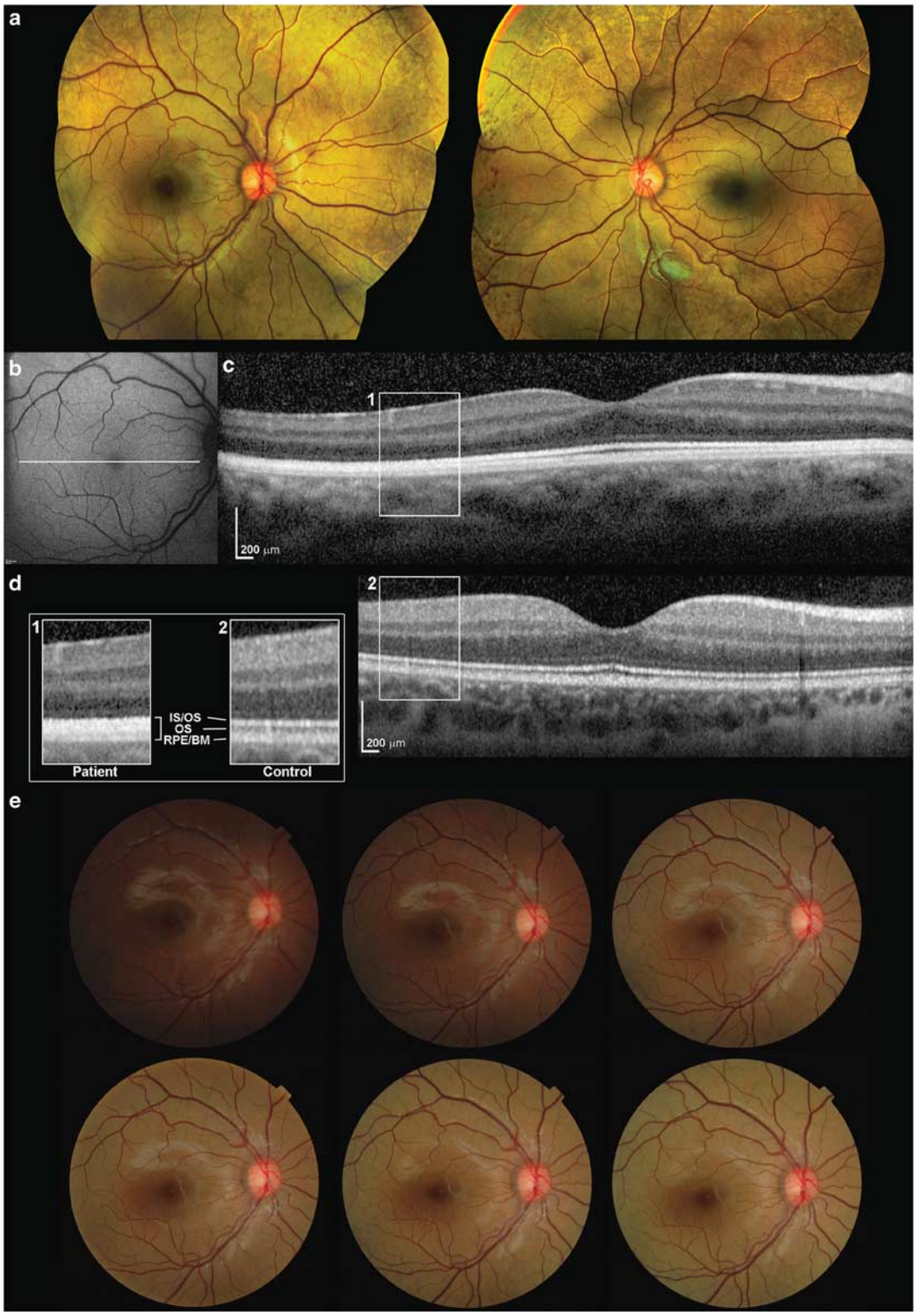
We report the clinical and electrophysiological findings in a case of Oguchi disease carrying a homozygous nonsense mutation in *SAG* (c.874C>T, p.Arg292X).

Figure 1 (a) Colour fundus photography of both eyes showing typical golden fundus reflex and pigment mottling in the far periphery. Discolouration is less profound in the macular area. (b) Fundus autofluorescence imaging of the right eye demonstrating no abnormality. (c) Axial cross-sectional image of the proband's right macula obtained using SD-OCT. In the parafovea, SD-OCT failed to detect the hyporeflective band (outer segments; OS) that is observed between the hyper-reflective layers associated with the inner/outer segment junction (IS/OS), and the RPE/Bruch's membrane complex (RPE/BM). (d) SD-OCT of the right eye of a 9-year-old control individual. Scans in c and d are to scale and acquired using the same SD-OCT protocol. Panel a with enlarged images of boxed regions (1, patient; 2, control) shows outer retina in detail. (e) Colour fundus photography of the posterior pole of the right eye using a non-mydriatic camera. After overnight (12-h) dark adaptation, a series of images were obtained over a 20-min interval. Disappearance of the golden reflex can be seen in the first image taken (top left). The golden colour gradually reappears after 10–15 flashes. Bottom right image is taken after 20 min and 32 flashes.

The *SAG* gene encodes S-antigen, a visual/beta arrestin abundant in rod photoreceptors. S-antigen binds to light-activated rhodopsin preventing further interaction with transducin during the recovery phase of phototransduction. Mutations in *SAG* are primarily associated with Oguchi disease.[1]

Case report

A 7-year-old girl of South Asian origin was referred for evaluation of congenital nyctalopia. Visual acuity was 0.12 logMAR in each eye. Fundus examination revealed widespread golden discolouration and peripheral RPE mottling (Figure 1a). Fundus autofluorescence imaging



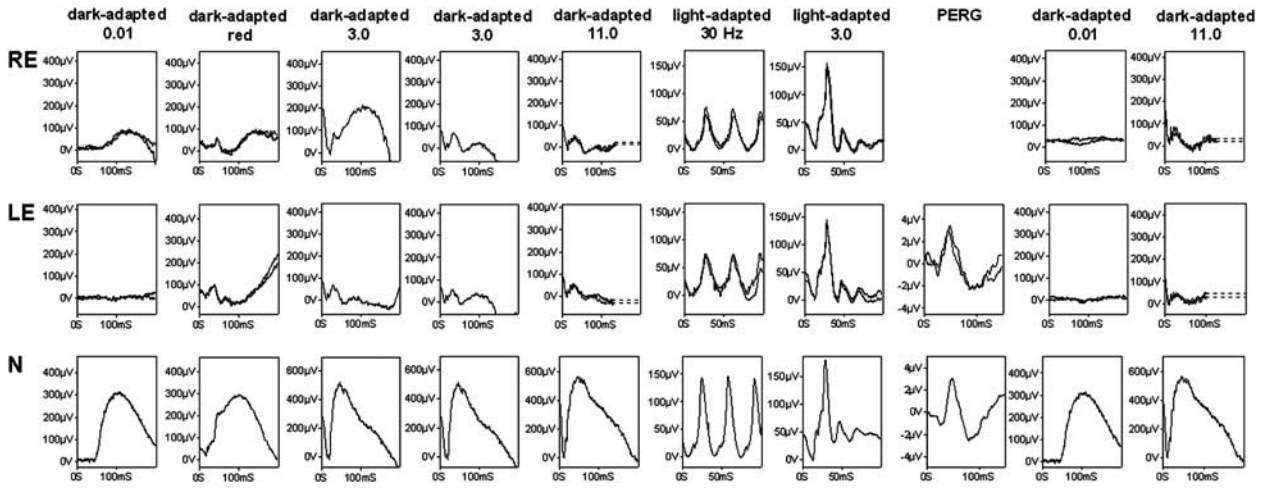


Figure 2 Full-field ERGs from the right (RE) and left (LE) eyes of the patient and normal examples (N) for comparison. After 25-min dark adaptation, left eye rod ERGs (0.01 cd s per m^2) were undetectable and bright flash ERGs (3.0 and 11.0 cd s per m^2) had a waveform that resembled the early component of the red flash ERG, consistent with a dark-adapted cone system origin.[7] After overnight dark adaptation of the right eye, ERGs showed partial recovery but a second bright flash (3.0 cd s per m^2; inter-stimulus interval 60 s) resulted in marked ERG attenuation. Light-adapted ERGs (3.0 cd s per m^2; 30 Hz and 2 Hz) revealed no evidence of generalised cone system dysfunction. The pattern ERG (PERG; left eye only) revealed no evidence of macular dysfunction. After photopic testing ERGs were repeated following an additional 20-min dark adaptation of both eyes.

was normal (Figure 1b). Spectral-domain optical coherence tomography (SD-OCT; Spectralis, Heidelberg Engineering, Heidelberg, Germany) is shown in Figure 1c. In the foveola, three hyper-reflective bands representing the inner segment/outer segment junction of the photoreceptors, the cone outer segment tips, and the RPE/Bruch's membrane complex are visible. Outside the fovea, the hypo-reflective band corresponding to the outer segments is not apparent. The findings were consistent on three different tests over a 1-year interval. SD-OCT of an age-matched control is also shown in Figure 1d.

International-standard full-field electroretinograms (ERGs) were consistent with severe generalised rod photoreceptor dysfunction. Prolonged dark adaptation resulted in partial ERG recovery, in keeping with abnormally slow rod dark adaptation, but with marked desensitisation following a single bright flash (Figure 2). Generalised cone function was normal. Pattern ERG revealed no evidence of macular dysfunction.

To test for the Mizuo-Nakamura phenomenon, the patient's right eye was dark-adapted overnight and images were obtained with a non-mydriatic camera (TRC-NW65, Topcon, Tokyo, Japan). Fundus appearance was initially normal in the dark-adapted eye but after 10–15 flashes, the golden sheen reappeared (Figure 1e).

Comment

Oguchi disease is caused by mutations in either *SAG* or *GRK1*, a gene encoding rhodopsin kinase.[1] Mutated *GRK1* alleles are considered the commonest cause of Oguchi in South Asians and only one Indian family has been reported carrying *SAG* mutation.[2]

SD-OCT findings similar to the proband have been previously described in two non-genetically confirmed Oguchi cases;[3,4] this outer retinal appearence has been attributed to microstructural changes,[3,4] and could indicate increased reflectivity in the light-adapted state. Additional outer retinal attenuation demonstrated in one of these cases, a 31-year-old man,[3] was not evident in our case.

A retinal sheen similar to Oguchi disease can be associated with *RS1* mutation, and partial or complete ERG recovery following prolonged dark adaption can occur in *RDH5* or *RLBP1*-related disease.[1,5] Rapid ERG attenuation to successive flashes can result from *RGS9*/*R9AP* mutation,[6] but the combination of normal cone function, delayed rod ERG dark adaptation and marked rod desensitisation to a bright flash is distinctive for Oguchi disease.

Conflict of interest

The authors declare no conflict of interest.

Acknowledgements

We acknowledge the following sources of funding: British Retinitis Pigmentosa Society, Fight for Sight, Alexander S Onassis Public Benefit Foundation, Moorfields Eye Hospital Special Trustees, National Institute for Health Research UK (Moorfields Eye Hospital and Institute of Ophthalmology, London, UK), Foundation Fighting Blindness (USA).

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Eye (2011) **25,** 1098–1011; doi:10.1038/eye.2011.88; published online 15 April 2011

Sir,

Development of optic disc drusen in familial pseudopapilloedema: a paediatric case series

Optic disc swelling in a child, with no other features suggestive of raised intracranial pressure is a challenging clinical scenario. These children frequently undergo invasive investigations, such as neuroimaging and lumbar puncture, even though the clinical suspicion of serious pathology is low.

In this study, we report a new clinical presentation of benign optic disc swelling in five siblings of two families, where optic disc drusen were not present at first presentation, but developed many months later as shown on serial B-scan ultrasonography. We have termed this unusual presentation 'familial pseudopapilloedema'.

Five children (two siblings from one family and three siblings from another family) underwent examination and B-scan ultrasonography at first presentation and at all subsequent examinations. Age range at first presentation was 18 months to 12 years with a mean of 6.5 years. The male to female ratio was 4 : 1. All five children had clinically apparent optic disc swelling without other ophthalmoscopic features of papilloedema (retinal nerve fibre layer swelling, surrounding disc haemorrhages, cotton wool spots, hyperemia, venous congestion, Patton's lines, or exudates[1]). None had any symptoms suggestive of raised intracranial pressure, other neurological disease, or systemic upset (Table 1).

Serial B-scan ultrasonography showed no drusen at the first visit. However, all children developed small linear drusen at the optic disc over time (Figure 1). The mean time for development of drusen detectable on B-scan was 2.7 years. At no point was there optic nerve sheath dilation on B-scan. In two children, CT scans were conducted and reported as normal. A CT scan was avoided in three children because of the absence of optic nerve sheath swelling on sonography, the absence of symptoms of raised intracranial pressure and normal visual function.[2,3]

Table 1 Summary table showing each of the five children's age, reason for presentation, vision, refraction, and investigations

	A1	*A2*	*A3*	*B1*	*B2*
Age at presentation	18 months	4 years	6 years	12 years	7 years
Time taken for optic disc drusen to become visible on B-scan	12 months	18 months	4 years	3 years	4 years
Reason for referral to our unit	Family history of optic disc drusen	Optometrist referred for possible drop in visual acuity	Optometrist noted possible swollen optic discs on routine visit	Optometrist referred for 'problems focusing'	Optometrist referred for possible swollen optic discs on routine visit
Refraction at last visit: right eye	+2.50 DS	+2.50 DS	$+3.50/+0.50\times90$	−0.25 DS	$+0.50/-0.25\times180$
Refraction at last visit: left eye	+2.25 DS	+2.50 DS	$+3.50/+0.25\times90$	−0.50 DS	$+0.50/-0.25\times175$
Vision at first presentation	6/7.5 BEO (Cardiff Cards)	R 6/7.5, L 6/7.5 (Snellen)	R 6/6, L 6/6 (Snellen)	R 6/6, L 6/5 (Snellen)	R 6/6, L 6/5 (Snellen)
Visual fields	No	No	Yes—Normal	Yes—Normal	Yes—Enlarged blind spot, otherwise normal
Lumbar puncture	No	No	Yes—Opening pressure 35 cm H_2O	No	No
CT scan	No	No	Yes—Normal	Yes—Normal	No

A and B represent different families. 1, 2 and 3 represent different children within the families.
BEO denotes both eyes open.
Normal VF = No constriction to I4e, I2e, or V4e spot on the Goldmann Kinetic Perimeter.