Sir,

Case report: an 8-year-old boy with unusual retinal hyperpigmentation

History

An 8-year-old child was referred by a local optometrist with an unusual pigmentary appearance of both fundi.

He had been born premature at 34 weeks with a collapsed right lung for which he had to be ventilated. No retinopathy of prematurity had been reported. He was medically fit and healthy.

Examination

Visual acuities (VA) were OD $6/6^{-2}$ and OS $6/9^{+2}$. He had severe blepharitis, quiet anterior segments, and a significant speckled retinal appearance 'like black pepper' (Figure 1). This did not appear to be of the bony spicule type. He was recommended lid hygiene and given a course of tetracycline, cortisone, and nystatin combination ointment twice daily for 1 month only, to treat the severe blepharitis.

He had Goldmann visual fields (Figure 2) and electrodiagnostic tests (EDTs). He was reviewed a month later and the visual fields and EDT results were found to be substantially normal with VA OD 6/6 pt and OS $6/6^{-1}$. A follow-up appointment 1 year later showed VA OD 6/6 pt and OD 6/6, with normal colour vision and normal intraocular pressures (right 16 mmHg and left 18 mmHg). The anterior segments were quiet, lenses clear, no sign of blepharitis, and unchanged fundal appearance. Repeat Goldmann visual fields (Figure 3) were normal and he did not have any systemic problems.

Investigations

Goldmann visual field tests

The Goldmann visual field tests were performed on initial presentation, which showed full fields but slightly constricted. This was thought to be due to a lack of concentration by the patient. This was then repeated a year later; this showed normal full fields which were not constricted.

Electrodiagnostic tests

Retinal studies showed that the pattern ERG was normal. Flash ERG responses were substantially normal in photopic and particularly scotopic conditions, considering the conditions of testing. The only abnormality identified was a delay in the implicit times of the red flicker ERG, particularly on the left.

Pattern reversal visual evoked response studies showed a normal response to right eye stimulation. Left eye stimulation appeared to show mild delay to 40-min checks, but corrected to 80-min checks.

Conclusions of EDT

The studies showed minor irregularities, which in conjunction with his age and conditions of testing were not clinically significant. There was no evidence of an abnormality of the peripheral rod-dominated retina, and hence no supportive evidence of retinitis pigmentosa.

Treatment

The child required no treatment and was reviewed periodically.

Diagnosis

Idiopathic retinal hyperpigmentation.

Differential diagnosis and discussion

Fundal hyperpigmentation has been reported in association with rare conditions like Kearns–Sayre syndrome.[1,2] This usually presents with ptosis, limited eye movement, and 'salt and pepper'-like retinal pigmentary changes. There are also systemic associations like ataxia, dementia, diabetes, and hyperaldosteronism. Similar fundal hyperpigmentation has also been noted in Cockayne's syndrome,[3] congenital Rubella,[4] congenital syphilis,[5] Alstrom's disease,[6] and with pigmentary glaucoma.[7]

Our patient was different in that there were no other ophthalmic or systemic associations and the fundal hyperpigmentation was more diffuse rather than of the 'salt and pepper' type. This might bear some resemblance to the autosomal dominant dystrophy of retinal pigment epithelium,[8] but we did not get an opportunity to examine any other members of the family. He also did not have any systemic abnormalities.

Fundal hyperpigmentation may be associated with other abnormalities and usually warrants a thorough investigation. Our case report highlights that it might be a normal occurrence. As this is a rare condition, it would be interesting to know of any further observations in future. This report suggests that it might be a normal variant. As this is unusual, it would be interesting to watch for delayed development of any further signs or symptoms.

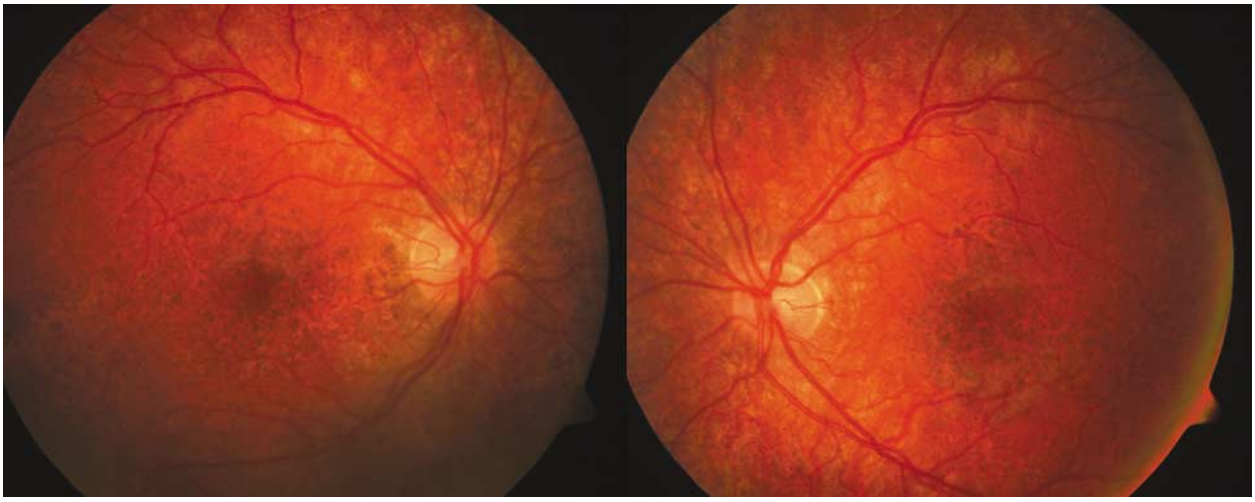

Figure 1 Peppered fundal hyperpigmentation.

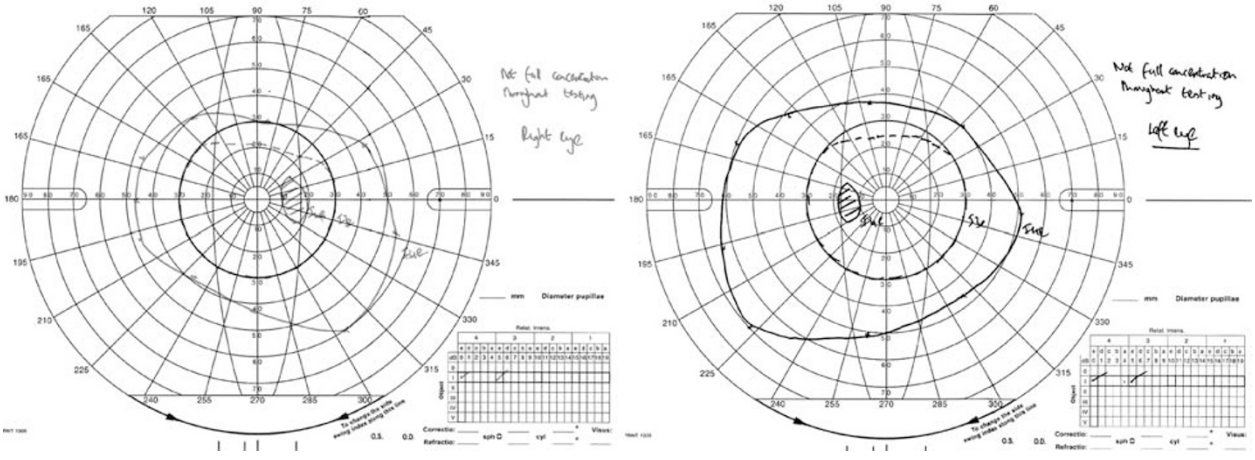

Figure 2 Initial Goldmann visual fields.

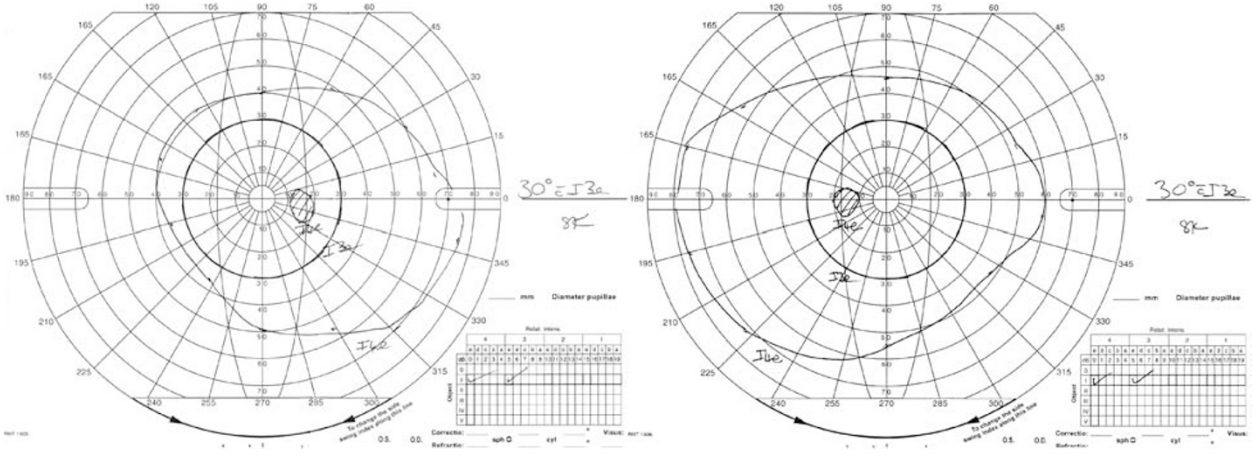

Figure 3 Goldmann visual fields 1 year later.

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RN Syed, Y Ghosh, A Berry-Brincat, ME Gregory and R Brown

Ophthalmology, University Hospital of North Staffordshire, Staffordshire, UK

Correspondence: R Syed,
Ophthalmology,
University Hospital of North
Staffordshire NHS Trust,
Princes Road,
Hartshill,
Stoke-on-Trent,
Staffordshire ST4 7LN, UK
Tel: +44 786 661 4595;
Fax: +44 178 255 4297.
E-mail: Reshma@easy.com or
ReshmaSyed@hotmail.com

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Sir,

Gaze-evoked amaurosis in optic neuropathy due to probable sarcoidosis

Gaze-evoked amaurosis refers to a transient visual loss provoked by eccentric gaze. It is rare and classically associated with orbital mass lesions.[1] Other causes, however, have been described, for example, idiopathic intracranial hypertension,[2] fractures,[3] and thyroid eye disease.[4] We present a novel cause of gaze-evoked amaurosis.

Case report

A previously healthy 33-year-old Caucasian male was reviewed after an initial diagnosis of episcleritis. He gave a 3-week history of left eye pain, worse on eye movement, particularly left gaze. There was no past ophthalmic or medical history of note, and he took no regular medications or recreational drugs. He was an ex-smoker and consumed a moderate amount of alcohol.

On examination, visual acuities were 6/5 and N4.5 in both eyes. He had a left relative afferent pupillary defect (RAPD) in the primary position. Left red desaturation was noted, with the left eye only reading 11/17 Ishihara plates compared to 16/17 plates by the right eye. Goldmann manual perimetry revealed superonasal constriction of the left field with an enlarged blindspot. Anterior segment examination and intraocular pressures were normal. Posterior segment examination was normal on the right and on the left revealed a quiet vitreous and swollen optic disc. Systemic examination was unremarkable and investigations were arranged.

One month later he had subjectively improved, except that he reported recurrent transient loss of vision in his left eye on looking to the left side, which recovered on returning to the primary position. On examination, his visual acuities were 6/5 bilaterally and there was no RAPD in primary position nor was there any loss of colour vision on Ishihara test plates. The left optic disc had become less swollen (Figure 1). However, on gaze to the left, a clear RAPD was apparent. Visual acuity, colour plate testing, disc perfusion, etc were not recorded in laevoversion due to fear of prolonged optic nerve compromise.

Full blood count, renal and liver profiles including calcium, ESR, C-reactive protein, vitamin B12 and folate, clotting and electrophoresis were within the normal range. Treponemal serology, autoantibody screen, rheumatoid factor, and anti-neutrophil antibodies were negative, serum angiotensin-converting enzyme level was not raised, and a chest plain film was unremarkable.

MRI brain and orbits revealed enlargement of the intraorbital and intracranial portions of the left optic