glial in origin, indicating massive retinal gliosis. There was calcification in the retina with some osseous metaplasia at the RPE level and abnormal retinal blood vessels (Figure 1).

The anterior choroid contained a plaque-like proliferation of variably pigmented spindle cell melanocytes. Although the lesion was generally well demarcated, there was local infiltration of adjacent structures (Figure 1). The cells were immunoreactive for HMB45. The melanocytes showed anisonucleosis and occasional prominent nucleoli. These features were consistent with a spindle cell malignant melanoma. No evidence of extraocular extension was seen.

Comments

It has been long known that a phthisical eye may harbour an occult malignant melanoma, though very few such cases have been reported. Sarma *et al*[3] reported a case of malignant melanoma in the left eye of a 62-year-old man who had been blind due to trauma for 35 years. Perry *et al*[4] reported a case of occult choroidal malignant melanoma in an eye with spontaneous expulsive choroidal haemorrhage and suggested that this association may be more than a chance occurrence, inasmuch as both are associated with necrosis of the posterior ciliary arteries.

Undiagnosed malignant melanoma of choroid is uncommon in phthisical eyes. Massive retinal gliosis is also an uncommon finding seen in eyes enucleated for phthisis bulbi. The case reported here shows both these rare features. It has been reported earlier that retinal gliosis may simulate a choroidal malignant melanoma[5,6] and also that ultrasound reflectivity and magnetic resonance imaging findings of massive retinal gliosis may resemble those of choroidal malignant melanoma. However, to the best of the authors' knowledge, the presented case is the first one to exhibit histological evidence of both malignant melanoma of choroid and massive retinal gliosis. It is possible that factors that lead to massive retinal gliosis in this phthisical eye, also lead to atypical proliferation of melanocytes giving rise to malignant melanoma. One may speculate that necrosis of posterior ciliary arteries was a precipitating factor for suprachoroidal haemorrhage, retinal gliosis and malignant melanoma formation in our case. Whatever the cause, this case once again emphasises the importance of subjecting enucleated eyes to a histopathological examination since the discovery of a malignant melanoma would influence the clinical management of such patients.

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Sir,

Cataract surgery in Senior–Loken syndrome is beneficial despite severe retinopathy

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A common problem in patients with retinopathy is whether they will benefit from surgical correction of co-existent cataracts.[1] The timing of surgery is dependent upon a clinical assessment of the degree of cataract compared to severity of retinopathy. The case presented here illustrates such a dilemma where cataract surgery exceeded all expectations of predicted benefit in the context of severe, widespread retinopathy.

Case report

A 28-year-old female was referred to our ophthalmic department in 1995. Her optometrist had noted reduced acuity and cataracts. Other medical history of

note was chronic renal failure, due to nephronopthisis, that had required kidney transplantation in 1979. The patient reported no skeletal abnormalities and normal hearing. The patient's identical sibling had also suffered from chronic renal failure that had resulted in death in infancy. The patient's brother was also affected with renal failure and suffered from nyctalopia. There was no other family member affected, the parents of the patient were reported to have normal vision and their union was non-consanguineous.

At presentation in 1995, right visual acuity was 6/18 with a +0.75/−2.25 at 5° lens and 6/18 left with a +2.0/−2.0 at 177° lens. No nystagmus was present. Pupil responses were normal. Anterior segment examination revealed marked, bilateral, posterior subcapsular cataracts. Retina examination was difficult due to the lens opacities, no definite retinopathy was identified but both optic nerve heads were described as 'abnormal'.

At that time, the cataracts were presumed secondary to the oral steroid immunosuppression and cataract surgery was offered. The patient underwent uneventful phacoemulsification cataract surgery in the right eye in 1995 and in the left eye 4 months later. The patient subsequently required bilateral YAG capsulotomy resulting in visual acuities of 6/9 right and left with appropriate spectacle correction.

By December 2000, the patient's main complaints were of a significant deterioration in her night vision. Visual acuity was still 6/9 with spectacles. Retina examination revealed pale, non-cupped optic discs, attenuated blood vessels and featureless maculae (Figure 1). Visual field assessment revealed a mild abnormality in the right eye and an upper nasal defect in the left (Figure 2). This asymmetric field deficit did not correspond with the degree or extent of retinal abnormality seen clinically. Such field defects, especially those localised to the upper field are however commonly seen in moderately severe, panretinal, outer retinal dystrophy.[2] Surprisingly, electrophysiological investigation (using ISCEV standard protocols and Goldfoil electrodes) reported pattern and full field electroretinograms (ERGs) as undetectable.[3] Visual evoked potentials (VEPs) were delayed. It was concluded that these findings were indicative of severe, generalised, retinal dysfunction and VEP abnormalities were secondary to the retinal changes. The electrophysiological investigations, kidney disease and family history suggested a diagnosis of Senior–Loken syndrome.

Comment

Nephronopthisis is a severe kidney disease due to renal tubular fibrosis, renal atrophy or dilatation and thickening of the tubular basement membrane. It is an important cause of renal failure in childhood. Senior–Loken syndrome accounts for an estimated 16% of nephronopthisis cases when a retinopathy and family history suggestive of autosomal recessive inheritance is seen.[4] Loken *et al*[5] first described this combination in a brother and sister in 1961. Senior *et al*[6] and Fairley *et al*[7] also reported the same combination in other families.

The retinopathy in Senior–Loken syndrome, on the basis of retinal appearance and electrophysiology, has been reported as severe, leading to blindness in childhood. This has been described as either a childhood-onset retinitis pigmentosa or a Leber's

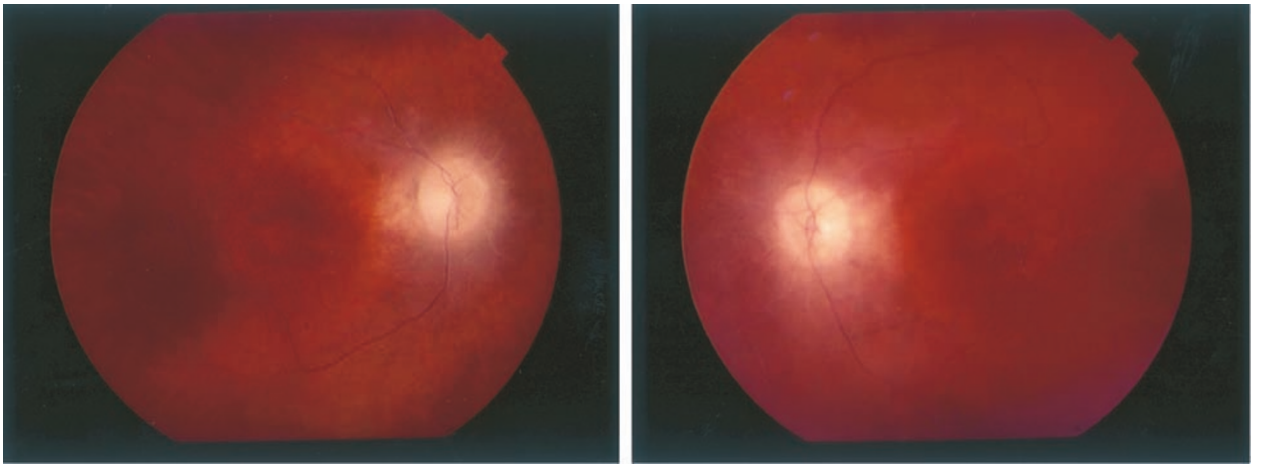

Figure 1 Retinal photographs of our patient with Senior–Loken syndrome demonstrating widespread pigmentary abnormalities, vascular attenuation and secondary optic atrophy.

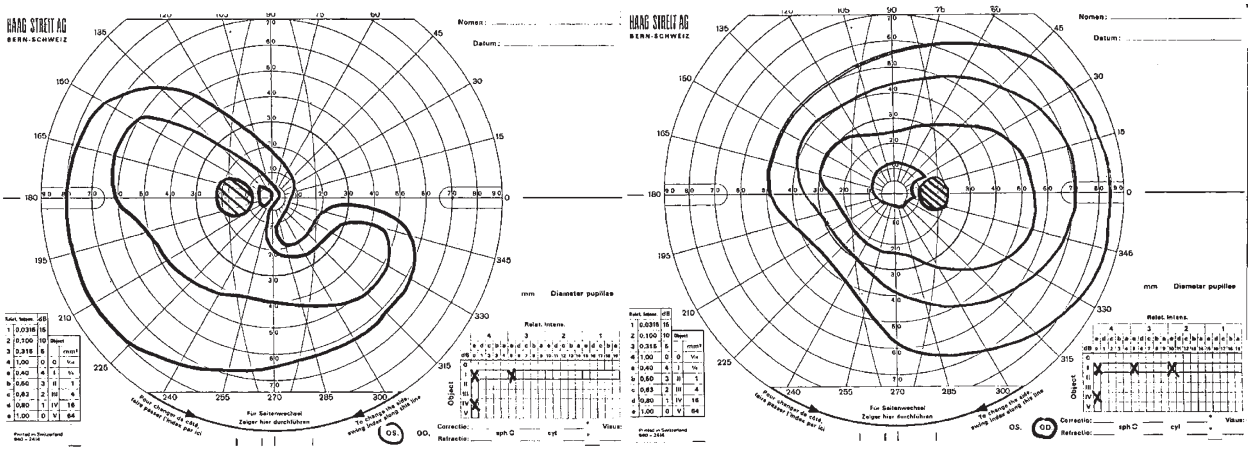

Figure 2 Goldmann visual fields of our patient with Senior–Loken syndrome showing an essentially normal right visual field and an upper nasal defect in the left visual field.

congenital amaurosis type of abnormality.[8] A single case report describes a sector retinitis pigmentosa with subnormal but detectable ERG.[9] Recently, a patchy retinopathy without visual symptoms has been described with nephronopthisis in families with deletions of the *NPH1* gene.[10] ERG abnormalities have also been reported in asymptomatic heterozygotes.[11] Other associated ocular abnormalities include congenital cataracts, Coat's disease and keratoconus.[12]

The case presented here is notable for several reasons. Relatively good visual acuity and significant retention of useful visual field has not previously been described in adult patients with Senior–Loken syndrome. This was despite almost undetectable electrical responses on electrophysiological investigations. This serves to remind us that undetectable electrical responses in clinical practice do not necessarily correlate with functional blindness.

Also, this is the first reported case of cataract surgery in a patient with Senior–Loken syndrome in which a significant benefit was gained. With marked advances in renal medicine the long-term survival rate in Senior–Loken syndrome is extending and more adult cases are being seen. We conclude that such patients may well benefit from surgery where cataract is present. Severe electroretinographic abnormality should only be considered a relative contraindication to this. Other clinical measures may be more reliable predictors of a successful surgical outcome, such as the presence of nystagmus or information from potential acuity meter investigation.

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Sir,

Visual loss due to cerebral infarcts in pseudoxanthoma elasticum

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Pseudoxanthoma elasticum (PXE) is a rare heritable connective tissue disorder with autosomal dominant or autosomal recessive modes of inheritance. It involves the elastic tissues of the eye, skin and cardiovascular system. Ocular features include angioid streaks due to involvement of the Bruch membrane. In patients with PXE and angioid streaks, visual loss is generally due to development of a choroidal neovascular membrane (CNVM). We report a case of PXE with visual loss due to multiple cerebral infarcts.

Case report

A 36-year-old man with a known diagnosis of PXE presented with a 5-day history of sudden onset right sided blurred vision. His best corrected visual acuity was 6/9 in the right eye and 6/6 in the left eye. Anterior segment examination was normal except for a right-sided relative afferent pupillary defect. Intraocular pressure was normal in both eyes. Fundus examination revealed angioid streaks, pigment mottling of the fundus in both eyes and pallor of the right optic disc (Figure 1). Physical examination revealed cutaneous lesions of PXE in the form of yellowish papules on the neck. Visual field analysis showed a right homonymous visual field defect. Fundus fluorescein angiogram confirmed the absence of CNVM. Magnetic resonance imaging (MRI) of the brain and orbits was performed. Axial fluid attenuated inversion recovery (FLAIR) sequence MRI of the brain revealed multiple ischaemic infarcts in the brain, the largest being in the left parieto-occipital lobe (Figure 2). The orbits and optic nerves were however normal.

The patient was young with no risk factors of cerebrovascular disease. He was a non-smoker. Blood tests revealed no evidence of diabetes mellitus, hyperlipidaemia or coagulopathy. A cardiovascular assessment was sought from a cardiologist. This included an electrocardiogram and transthoracic echocardiogram. There was no evidence of any cardiovascular abnormality apart from borderline hypertension, which did not require treatment.

Comment

Apart from CNVM, visual loss in PXE can also be caused by infarction of the visual pathways, most commonly involving the orbital portion of the optic nerve.[1] Our patient showed features of multiple areas of central nervous system ischaemia. The left parieto-occipital lobe infarct was thought to be the cause for the visual field defects. The presence of a relative afferent pupillary defect and optic disc pallor on the right side suggests involvement of the right optic nerve. Though conjectural, these changes are likely to be due to an old infarct of the optic nerve as the optic disc pallor was noted at acute presentation itself (within 5 days of onset of symptoms).

Cerebral ischaemia in PXE is caused by small vessel

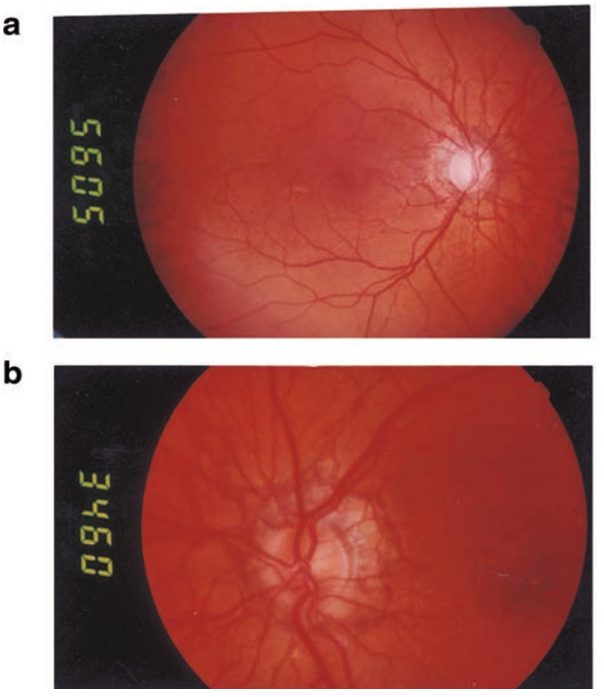

Figure 1 Fundus photographs of right eye (a) and left eye (b) showing prominent angioid streaks and pigment mottling of the posterior pole. Note diffuse optic disc pallor and characteristic peau d'orange appearance temporal to the macula in the right eye.