Sir,
Fatal thrombotic thrombocytopenia purpura

This is an interesting case which highlights one of the many presentations of Thrombotic thrombocytopenic purpura which also include retinal haemorrhages, oculomotor nerve palsies, pupil abnormalities as well as our case of retinal vascular occlusion and the case of choroidal vasculopathy as described by the authors. It is important to recognise this spectrum of presentations, as Thrombotic thrombocytopenic purpura can be fatal if not diagnosed and treated promptly.

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Sir,
Ten year follow up of pigmentary retinopathy associated with 3-hydroxyacyl-CoA dehydrogenase deficiency

Mitochondrial β oxidation is the principal pathway by which energy is obtained from fatty acids. Defects in this

pathway are associated with hypoketotic hypoglycaemia, hepatic steatosis, rhabdomyolysis, cardiomyopathy, hypocarnitaemia, and abnormal urinary metabolites. Long-chain 3-hydroxy-acyl-CoA dehydrogenase (LCHAD) deficiency disrupts this pathway and was first described in 1989.^{1–4} Treatment is with a low-fat high-carbohydrate diet, carnitine and docosahexanoic acid (DHA) supplementation. Pigmentary retinopathy has been associated with this abnormality.^{5–9} We report such a case with follow-up over a 10 year period.

Case report

A 5-year-old emmetropic girl with a history of LCHAD deficiency was referred to the ophthalmology department for evaluation. She was on vitamin supplementation, carnitine and a low-fat high-carbohydrate diet. At first presentation, her visual acuity was recorded at 6/6 in each eye. Fundal examination was documented as showing a bilateral pigmentary retinopathy. During the first 6 years of follow-up her visual acuity remained at a similar level. At the age of 11 a myopic prescription was prescribed, right -0.75 DS/ $-1.0@180$, left -0.5 DS/ $-0.75@180$, visual acuity remaining 6/6 in each eye. At the age of 15 the patient complained of visual deterioration. Visual acuity in the right eye had deteriorated to 6/24 while the left eye retained 6/6 vision. The degree of myopia had increased to right -4.0 DS/ $-1.75@10$, left -3.25 DS/ $-1.0@170$. Ophthalmoscopy showed extensive posterior pole atrophy (Figure 1) in each eye with an 'island' of retinal pigment epithelium retained in the left eye (Figure 1b). A small hyper-pigmented spot was also noted at the fovea of each eye. The periphery of each eye showed granular RPE changes. Retinal vasculature was of normal appearance. Anterior segments were normal.

Figure 1 Endoscopic findings showing the well-demarcated atrophic posterior pole in each eye (a) right eye (b) left eye. Note the hyperpigmented spots at each macula.

Electrophysiology has been carried out at intervals over a 10-year period. The electroretinogram has remained within normal limits, however, the amplitude of the scotopic a-wave and photopic b-wave have fallen to the lower limit of normal range. The electro-oculogram has been normal with an Arden index of over 210%.

Comment

The ocular findings associated with this rare metabolic disorder include developmental cataracts, progressive myopia, visual field defects, and colour vision abnormalities.⁸ Tyni *et al*⁸ classified the chorioretinopathy into four stages. At stage 1, the fundus appears normal to diffusely hypo pigmented. Stage 2, pigment clumping in the RPE gives rise to pigmentary changes. At stage 3, the granular fundus pigmentation disappears centrifugally with concomitant atrophy of the choroid and the posterior pole, leaving bare sclera.

At stage 4, the central island of the remaining choroids and photoreceptors is lost and posterior pole becomes atrophic and devoid of RPE. At the periphery choroidal vessels and RPE remain relatively normal.

As with all β oxidation defects treatment is aimed at ensuring sufficient caloric intake during periods of metabolic stress and fasting. Owing to secondary carnitine deficiency carnitine supplementation is prescribed to all patients with this condition. A low-fat high-carbohydrate diet is also regarded as beneficial. However, one of the most beneficial forms of the treatment is to avoid fasting.

We describe the striking fundal findings and progression of this retinal dystrophy in a patient with a 10-year follow-up period. One noticeable additional feature seen in this patient's fundal findings was the hyperpigmented spot at the centre of the macula in the right and the left eye. ERG findings show only subtle changes in implicit times and amplitudes. These do not appear to mirror visual acuity but may be representative of the gradual but ultimately progressive nature of this condition. Progressive myopia has been noted to coincide with the development of the staphylomatous like posterior pole changes and may be a better indicator of disease progression in this dystrophy.

In conclusion, LCHAD deficiency is associated with a progressive retinal dystrophy, which maybe slowed by careful dietary management but ultimately over time progression appears to occur.

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