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Magda Gharbiya ✉
Francesco Bozzoni Pantaleoni
Massimo Accorinti
Paola Pivetti Pezzi
Institute of Ophthalmology
University 'La Sapienza' of Rome
Viale del Policlinico, 155
I-00161 Rome, Italy
Tel: +39 6 490296
Fax: +39 6 4457706

Sir,

Stable pigmentary retinopathy in a child with 3-hydroxyacyl-CoA dehydrogenase deficiency

Deficiency of long chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD),[1,2] an enzyme involved in mitochondrial beta-oxidation of fatty acid, has been shown to be associated with hypoketotic hypoglycaemia, hepatic steatosis, cardiomyopathy, rhabdomyolysis, peripheral neuropathy and retinopathy.[1–3] Prognosis of life and vision in these patients is poor. The natural course of the disease can be alleviated by a low-fat high-carbohydrate diet along with carnitine and docozahexanoic acid (DHA) supplementation.[4,5] We present a case of stable pigmentary retinopathy in an 8-year-old child with LCHAD therapy on dietary substitution of carnitine and DHA therapy.

Case report

A 3-year-old emmetropic boy was referred to the ophthalmology unit for fundus evaluation following detection of pigmentary retinopathy. The child, a known case of LCHAD deficiency, was on dietary therapy with DHA (65 mg/day) and carnitine supplementation.

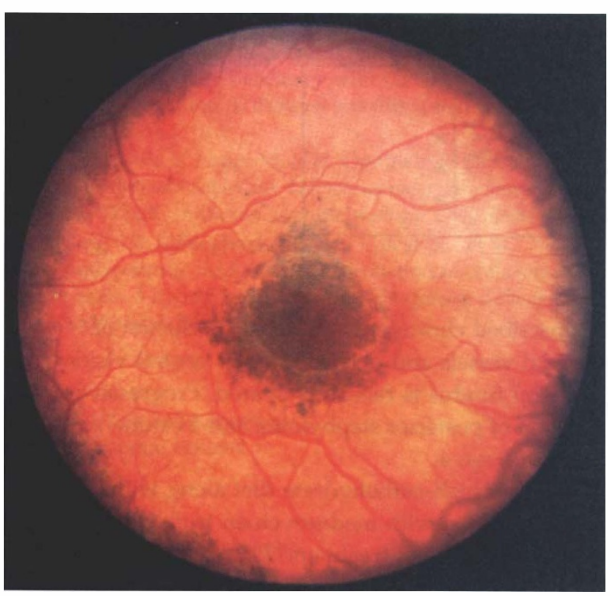


Fig. 1. *Fundus photograph of the right eye showing pigmentary maculopathy.*

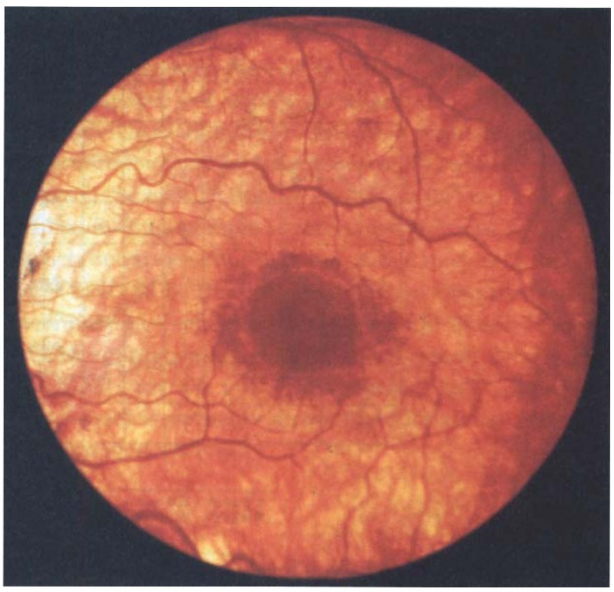


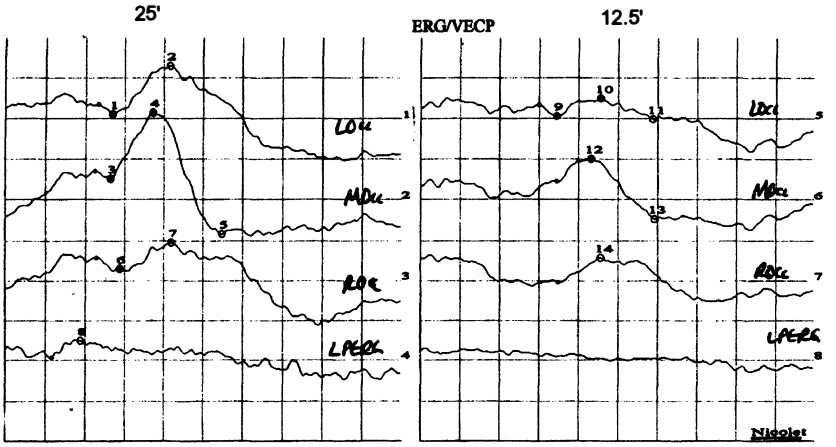
Fig. 2. *Fundus photograph of the left eye showing pigmentary maculopathy.*

Ocular examination revealed a best corrected Snellen visual acuity of 6/9 in the right and 6/6–4 in the left eye. The child was emmetropic and orthophoric with normal ocular motility. Anterior segment was essentially normal. Fundus evaluation revealed bilateral foveal pigmentary retinopathy and secondary retinal pigment epithelium atrophy (Figs. 1, 2). An impression of bimacular pigmentary retinopathy secondary to LCHAD was made.

The child was followed yearly in the clinic for 5 years to assess the progression of disease. However, his ocular condition and visual acuity remained near stable during this period. At his last follow-up his best-corrected visual acuity was 6/9 in both eyes. Fundus evaluation revealed no progression of retinopathy. The child had full Goldmann fields. Threshold visual fields showed scattered scotomas not conforming to any pattern. Electrophysiology performed at this visit revealed normal visual evoked potentials (VEP); flash electroretinogram (ERG) amplitudes were near the lower limit of normal, and pattern ERG was of poor amplitude (Fig. 3).

Comment

Long chain 3-hydroxyacyl-CoA activity is found in the mitochondrial trifunctional protein (MTP). Its deficiency, a result of G1528C mutation, leads to deficiency in beta-oxidation of fatty acids[2] and is characterised by low activity of LCHAD with normal levels of immunoreactive MTP.[6] Although analysis of serum carnitine[4] can be used as a screening test, LCHAD deficiency can be detected by measuring the enzyme activity or by detection of G1528C mutation in blood or tissue samples. Prenatal diagnosis is also possible by chorionic villous biopsy. Urine analysis in patients with LCHAD deficiency shows increased levels of 3-hydroxylated carboxylic acids.[5]



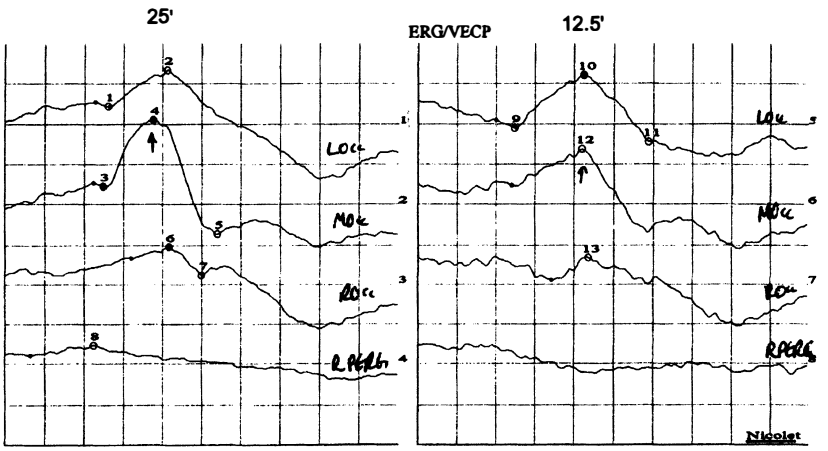
Sensitivity and Sweep Time Per Division

1	9.96 uV	28.5 msec	2	9.96 uV	28.5 msec	3	9.96 uV	28.5 msec	4	9.96 uV	28.5 msec
5	9.96 uV	28.5 msec	6	9.96 uV	28.5 msec	7	9.96 uV	28.5 msec	8	9.96 uV	28.5 msec

MEASUREMENTS

1	2.57uV 61.38ms	6	2.61uV 67.08ms	11	5.54uV 152.58ms	16	
2	12.13uV 103.56ms	7	6.42uV 104.70ms	12	5.45uV 108.12ms	17	
3	1.99uV 60.24ms	8	4.16uV 38.58ms	13	14.92uV 153.72ms	18	
4	16.40uV 91.02ms	9	2.83uV 83.04ms	14	6.27uV 114.96ms	19	
5	30.17uV 141.18ms	10	4.32uV 114.96ms	15		20	

(a)



Sensitivity and Sweep Time Per Division

1	9.96 uV	28.5 msec	2	9.96 uV	28.5 msec	3	9.96 uV	28.5 msec	4	9.96 uV	28.5 msec
5	9.96 uV	28.5 msec	6	9.96 uV	28.5 msec	7	9.96 uV	28.5 msec	8	9.96 uV	28.5 msec

MEASUREMENTS

1	1.09uV 59.10ms	6	2.83uV 103.56ms	11	16.87uV 152.58ms	16	
2	8.98uV 102.42ms	7	7.05uV 127.50ms	12	9.02uV 104.70ms	17	
3	0.81uV 55.68ms	8	2.58uV 48.84ms	13	5.76uV 109.26ms	18	
4	16.57uV 92.16ms	9	2.33uV 55.68ms	14		19	
5	28.60uV 138.90ms	10	13.38uV 105.84ms	15		20	

(b)

Ocular features of this condition include developmental cataracts, progressive myopia, progressive visual field defects (central and paracentral scotomas) and defects in colour vision (initially tritanomaly but later progressing to severe derangement). However, pigmentary retinopathy is characteristic for LCHAD deficiency.[3,7,8] Fundus evaluation shows progressive chorioretinopathy, which can be classified into four stages.[8] It is essentially normal at birth (stage 1). This is followed by the stage of pigment dispersion. Stage 3 is characterised by areas of circumscribed chorioretinal atrophy, occlusion of choroidal vessels and deterioration of central vision; the peripheral fundus is relatively spared. Progressive formation of posterior staphyloma and a central scotoma is seen in stage 4.

A low-fat high-carbohydrate diet along with dietary supplementation with medium-chain triglycerides has been the mainstay of treatment. DHA, an essential fatty acid, the deficiency of which is known to be associated with altered visual electrophysiological parameters has been found to be deficient in patients with LCHAD.[9] It

therefore, when given at a dose of 65 mg/kg (in children less than 20 kg) or 130 mg (in children more than 20 kg), has been shown to cause a small but significant improvement in visual acuity as demonstrated by visual evoked potentials. However, long-term results are awaited.

In the present case, the visual acuity and retinopathy remained clinically stable over a follow-up period of 5 years. However, repeat electrophysiological testing could only exclude the progression. This report thus supports the earlier claims that dietary supplementation along with addition of DHA helps in stabilisation of the retinopathy.

We are very grateful to Mr D. Brosanhan for providing us with the details of this interesting case.

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P. Puri
J. Chan
Department of Ophthalmology
Royal Hallamshire Hospital
Sheffield, UK

Mr J. Chan ✉
Department of Ophthalmology
Royal Hallamshire Hospital
Glossop Road
Sheffield S10 2JF, UK

Sir,

Spontaneous reattachment of retinal detachment in a highly myopic eye with a macular hole

Spontaneous resolution of idiopathic senile macular holes with foveal detachment has rarely been observed in non-myopic eyes.[1,2] Bonnet and Semiglia[3] reported a similar finding in three severely myopic eyes in the French literature. They concluded that such detachments were likely to be tractional rather than rhegmatogenous secondary to a macular hole. We would like to report a case of spontaneous reattachment of retinal detachment in a highly myopic eye with a macular hole. The retinal detachment is probably a combined tractional and rhegmatogenous type.

Case report

A 65-year-old Chinese woman with bilateral severe myopia of –20.00 DS was referred to our vitreoretinal unit with a 5 day history of a sudden increase in a central scotoma of the right eye. Her best-corrected visual acuity was 20/200 and 2/200 over the right and left eye, respectively. Slit-lamp examination revealed aphakia in the right eye and pseudophakia in the left eye. Intraocular pressure and the anterior segment were normal. Fundoscopy showed bilateral myopic chorioretinal degeneration of the macula with the presence of posterior staphyloma, which extended up to the temporal arcades and the nasal edge of the disc. Additionally, there was a convex-shaped retinal detachment over the superior two-thirds of the right macula. Contact lens biomicroscopy of the right macula revealed a full-thickness macular hole of size 200 μm, which was surrounded by a thin epiretinal membrane. Axial length was 32.58 mm and 30.40 mm over the right and left eye, respectively. After detailed explanation, she refused operation at that juncture. Four weeks later, the staphylomatous retinal detachment was flattened and the macular hole was not visible. The epiretinal membrane was separated into the vitreous cavity, with a central round defect corresponding to the original macular hole. Her visual acuity remained at 20/200 but she admitted a decrease in size of the central scotoma. Six months later, her condition remained stable.

Comment

In a study by Morita *et al.*[4] the risk of macular hole causing rhegmatogenous retinal detachment was significantly increased in eyes with high myopia, posterior staphyloma and chorioretinal atrophy. However, contrary to their report, we have illustrated a rare case of spontaneous flattening of macular detachment in an eye with all these risk factors. We hypothesise that the detachment in our patient is a combined tractional and rhegmatogenous type. The rhegmatogenous component is apparent, as the detachment is convex towards the pupil with the presence of a macular hole. Epimacular membrane is responsible for the tractional component. Although the