Lipaemia retinalis in a 29-day-old infant with type 1 hyperlipoproteinaemia

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SUMMARY A 29-day-old infant born of consanguineous parents had type 1 hyperlipoproteinaemia associated with lipaemia retinalis. Biochemical analyses disclosed hypertriglyceridaemia, hyperchylomicronaemia, and lipoprotein lipase deficiency. Ophthalmoscopically all retinal vessels had the creamy appearance of lipaemia retinalis. The plasma lipoprotein levels and fundus condition became normal after her ingestion of fat was limited. To our knowledge this is the youngest patient reported with type 1 hyperlipoproteinaemia associated with lipaemia retinalis.

Type 1 hyperlipoproteinaemia is an autosomal recessively inherited disorder characterised by hyperchylomicronaemia and hypertriglyceridaemia. To date fewer than 100 cases of the disorder have been reported. Lipaemia retinalis is peculiar in this disorder. However, the fundus picture has been rarely shown. We describe lipaemia retinalis in a 29-day-old infant with type 1 hyperlipoproteinaemia.

Case report

An apparently normal baby was delivered on 12 July 1983 after a full-term uneventful pregnancy. Her birth weight was 3280 g. The parents, who were first cousins, and a 2-year-old brother were in good health (Fig. 1). The infant was breast-fed. When a blood sample was obtained from the cubital vein for newborn screening at 5 days of age, the patient's serum was observed to be milky. On 4 August she was referred to the Tohoku University Hospital for further examination.

Biochemical analyses disclosed a slightly elevated serum cholesterol level (400 mg/dl (10·4 mmol/l); normal, 130–250 mg/dl (3·4–6·5 mmol/l)) and a markedly increased serum triglyceride concentration (3000 mg/dl (33·9 mmol/l); normal, 35–130 mg/dl (0·4–1·5 mmol/l)). During the examination serum triglyceride level sometimes increased to 15000 mg/dl (169·5 mmol/l). The serum phospholipid level was within normal range. The presence of hyperchylomicronaemia was demonstrated by agar-gel electrophoresis. Apolipoprotein CII assayed by immunodiffusion was normal. Lipoprotein lipase activity in postheparin plasma examined by the method of Murase and associates, based on immunoprecipitation of hepatic triglyceride lipase, was negligible. The deficiency of lipoprotein lipase was thus confirmed in this case. Other laboratory data, including blood cell counts, electrolytes, liver function tests, estimations of glucose, ammonia, and amylase, and immuno-electrophoresis, were normal. The liver and spleen were not enlarged.

At 29 days of age an ophthalmic examination was performed. A few lipid depositions, or eruptive xanthoma, were noted in the upper eye lids. Ocular tension was normal to palpation. Pupils were isocoric.

Pedigree

Fig. 1 Pedigree.
Lipaemia retinalis in a 29-day-old infant with type I hyperlipoproteinaemia

Lipaemia retinalis is a manifestation of excess triglyceride in the bloodstream: the retinal vessels are found to be milky or creamy when triglyceride levels rise to 2000 to 2500 mg/dl (22.6 to 28.2 mmol/l) or greater. Lipaemia is seen not only in retinal vessels but also in limbal vessels. Lipaemia retinalis has been graded 0 to III by Vinger and Sachs. Fundus changes in our case were grade III according to this classification. Increased cholesterol levels alone do not cause the milky plasma.

Lipaemia retinalis is reported in cases of types I, II, IV, and V hyperlipoproteinaemia. Type I hyperlipoproteinaemia is characterised by hyperchylomicronaemia and hypertriglyceridaemia and confirmed by lipoprotein lipase deficiency. The plasma lipoprotein levels and fundus condition usually respond to treatment with a low-fat diet, as in our case.

Fundus photographs of lipaemia retinalis have been shown mostly for type V hyperlipoproteinaemia secondary to diabetes. The fundus picture is not found in cases of idiopathic or retention (former name of type I) hyperlipaemia, and has been found only in 18- and 28-year-old men with type I hyperlipoproteinaemia. Type I hyperlipoproteinaemia in an 8-day-old infant has been reported, but she lacked eruptive xanthoma and lipaemia retinalis.

To our knowledge our present case is the youngest patient with type I hyperlipoproteinaemia associated with lipaemia retinalis.

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