Dramatic atherosclerotic vascular burden in a patient with familial lecithin-cholesterol acyltransferase (LCAT) deficiency

Sir,

It was with interest that we read Weber et al.’s manuscript on stability of lipid profile in a patient with familial lecithin-cholesterol acyltransferase (LCAT) deficiency treated by peritoneal dialysis [1].

More than 20 years ago, an 18-year-old man presented at our hospital with proteinuria (about 3 g/day), microhaematuria and normochromic anaemia; a corneal opacification was present.

Total cholesterol (TC) was 243 mg/dl, triglycerides (TG) 530 mg/dl, high-density cholesterol (HDL) 8 mg/dl, low-density cholesterol (LDL) 198 mg/dl, serum creatinine 1.06 mg/dl and urea 40 mg/dl. His father had died at the age of 48 of myocardial infarction.

A kidney biopsy was executed and the diagnosis of LCAT deficiency was done; the histological picture was previously described [2], remarkable larger mesangial deposits and a sieve-like transformation of the basement membrane, the absence of foam cells in the glomeruli and the finding of C3 in the glomerular deposits. Serum LCAT activity was undetectable.

His renal function gradually worsened until 1987, when, aged 24, he started haemodialysis. After 3 years, he received a renal transplant, but it was complicated by acute graft rejection and a cytomegalovirus pulmonary infection, successfully treated. After 6 years, in 1996, because of severe hypertension (more than 210/110 mmHg) with retinal haemorrhages and exudates, a worsening of renal function was likely to occur: an occlusion of the renal artery of the transplanted kidney was found by Doppler sonography. Haemodialysis was resumed and a renal angioplasty plus stenting was done: after that, dialysis treatment was stopped over 4 years because of renal function recovery.

In 2000, due to the graft failure, chronic haemodialysis treatment was resumed; over this period his lipid profile was characterized by elevated levels of TC (>250 mg/dl), very high TG (>1700 mg/dl) and low HDL (<20 mg/dl), despite the use of lipid-lowering drugs (statins and fish oils).

His clinical story in the following years was constellation with severe vascular diseases, namely claudication at the age of 42.

In January 2005, because of the worsening of the LP AO, he was submitted to a right femoral-axillo artery by-pass and, after 1 month, to a thigh amputation for gangrene. In February 2005 he died at the age of 42.

Our experience in a single patient allows us to reply to the Weber’s question: ‘Is there an increased risk of premature atherosclerosis in LCAT deficiency?’: Yes, undoubtedly.

Our case differs from Weber’s patient, because of a dramatic atherosclerotic burden, as also found by Ayyobi et al. [3], albeit less pronounced, in heterozygotes LCAT patients followed over 25 years, with intima-media thickness abnormalities.

In our 30-year experience in peritoneal dialysis (PD) we considered such a dialytic option as a first-line choice in young patients on renal transplant, with some reservations in overweight patients, such as Weber’s patient, with poor dietetic compliance who will with difficulty reach an adequacy target.

Moreover, PD potentially favours a more atherogenic lipoprotein profile, highly compromised in LCAT deficiency, due to the continuous adsorption of glucose from the dialysis bags, the frequent association of impaired tolerance to glucose with peripheral hyperinsulinaemia and increased insulin resistance, and finally protein losses with dialysate, stimulating the hepatic synthesis of Lp(a) as happens in nephrotic syndrome [4].

Conflict of interest statement. None declared.

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Reply

Sir,

We thank Dr Scarpioni for his interest in our paper [1]. While we did not see the manuscript relating the dyslipidemia of peritoneal dialysis to a cardiovascular risk factor, this certainly was a potential concern in our patient.

It is important to remember that a spectrum of phenotypic expression exists in patients with metabolic diseases, largely influenced by environmental and genetic factors. Concordant with the weight of evidence in LCAT deficiency and despite a markedly abnormal lipid profile, our patient did not manifest evidence of atherosclerotic disease. However, case reports have been published that describe a...