Polidistrectual vascular involvement in Familial Hyperchilomicronemia

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A 72-year-old man was referred to our clinic with a lipid profile, under combination therapy with a statin and ezetimibe, characterized by severe hypertriglyceridemia (7230 mg/dl), hypercholesterolemia (374 mg/dl), low HDL-cholesterol (17 mg/dl), and normal circulating Lp"a". The patient had undergone post-traumatic splenectomy and presented a history of systemic hypertension treated medically with well-controlled blood pressure. The patient had also presented in the past abdominal pain with subsequent diagnosis of chronic pancreatitis complicated by diabetes mellitus, well compensated when he came to our attention.

Following a pathological exercise test, a coronary angiogram was performed which showed a significant stenosis of the right coronary artery, which was treated successfully with percutaneous coronary intervention. Clinical work-up revealed ectasia of the abdominal aorta (28 mm), non significant bilateral carotid artery disease, and peripheral artery disease of the femoral-popliteal axis symptomatic for intermittent claudication.

A lipidogram was also performed and electrophoretic lipoprotein patterns did not vary 2 hours after heparin infusion, pointing to the existence of lipoprotein lipase deficit. Electrophoresis also showed a broadband of chylomicrons at baseline, at the beginning, and at the end of heparin infusion.

Hyperchilomicronemia is a rare genetic disorder with an incidence of 1 per 1000000. Following diagnosis, our patient began plasma exchange therapy with subsequent improvement of his lipid profile. At the present time, he is regularly followed up at our clinic and non invasive imaging has excluded any significant progression of atherosclerosis after 2 years of therapy.

Key words: hyperlipidemia, lipoprotein lipase deficiency, atherosclerosis, plasmapheresis.