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Cardiovascular disease in a pediatric patient with homozygous familial hypercholesterolemia

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Introduction
Familial homozygous hypercholesterolemia (HFHo) is an autosomal dominant hereditary disease with a prevalence of 1:800.000. To date, 7 are the children diagnosed in Spain. cLDL receptor gene variants are disease causing, resulting in total or almost total lack of enzyme activity. The disease can be diagnosed at birth, showing a total cholesterol (TC) of 700-1000 mg/dl, at the expense of cLDL. Xanthomas, corneal arch, and atherosclerosis are detectable in the first decade of life, while from the second decade an increase of mortality is reported.

Case Report
An 8-year-old boy with multiple tuberous xanthomas and bilateral corneal arch is in actual follow up in our center because his parents are affected by heterozygous familial hypercholesterolemia. His lipid profile at presentation showed TC and cLDL blood levels of 741 mg/dl and 672 mg/dl, respectively, without other biochemical abnormalities. Genetical analysis identified a homozygous rLDL mutation (p.Glu228-stop), confirming the diagnosis. At first, his clinical manifestations comprehended: 1) atheromatous plaques at the carotid vascular US study, 2) a dilated left ventricle (z-score +2.5) at heart US, along with a mild aortic insufficiency and a thickened non-coronary veil, 3) soft atherosclerotic coronary plaques, with no stenosis, and a calcified atheromatous plaque in the ascending aorta at the coronary angioTAC. Ergometry and myocardial perfusion studies were normal. He was started on rosuvastatin and ezetimibe, and also received acetylsalicylic acid prophylaxis. As no improvements where detected, biweekly sessions of LDL-apheresis were also started. A decrease of 50% in baseline cLDL levels, disappearance of tuberous xanthomas, and reduction of carotid atheromatous plaques were observed. Dilatation of the left ventricle persisted. Coronary angioTAC is still pending.

Conclusions / Comments
The initial treatment of HFHo includes dietary recommendations, lifestyle modification and lipid-lowering drugs, but the effectiveness is partial. Our case confirms that LDL-apheresis can be efficient in decrease the cardiovascular risk; we have managed to reduce the skin lesions, as well as atheromatosis and carotid disease. Coronary plaques potential evolution needs more time to be evaluated.