CARDIOVASCULAR DISEASE IN A PAEDIATRIC 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. At present in Spain there are 7 children diagnosed. It is caused by mutations in the gene that codes for the LDL-C receptor, producing its total or almost total lack of activity. It can be diagnosed at birth, showing a total cholesterol (TC) of 700-1000 mg/dl, at the expense of LDL-C. In the first decade it is possible to detect xanthomas, corneal arch, atherosclerosis and, from the second decade on, an increase of mortality.

CASE REPORT

An 8-year-old boy with multiple tuberous xanthomas and bilateral corneal arch is referred to our center because his parents are affected by heterozygous familial hypercholesterolemia. His lipid profile at presentation showed cLDL blood levels of 672 mg/dl, without other biochemical abnormalities. Genetical analysis identified a homozygous rLDL mutation (p.Glu228-stop), confirming the diagnosis. At first, his clinical manifestations comprehended:

- Ergometry and myocardial perfusion studies were normal. He was started on rosuvastatin and ezetimibe, and also received acetylsalicylic acid prophylaxis, as well as biweekly sessions of lipid-apheresis, observing a decrease in LDL-C of 50%. Finally lomitapide was added to the treatment, with this an LDL decrease of 67% was achieved, being able to pass lipid-apheresis to monthly sessions.

CONCLUSIONS

The initial treatment of HFHo includes dietary recommendations, lifestyle modification and lipid-lowering drugs, but the effectiveness is partial, so the LDL-apheresis is necessary to decrease the cardiovascular risk. Our case confirm that LDL-apheresis can be efficient by reducing atheromatosis and carotid disease, as well as the skin lesions. Coronary plaques potential evolution needs more time to be evaluated.