LDL- apheresis as the choice of therapy in the treatment of familiar hypercholesterolemia

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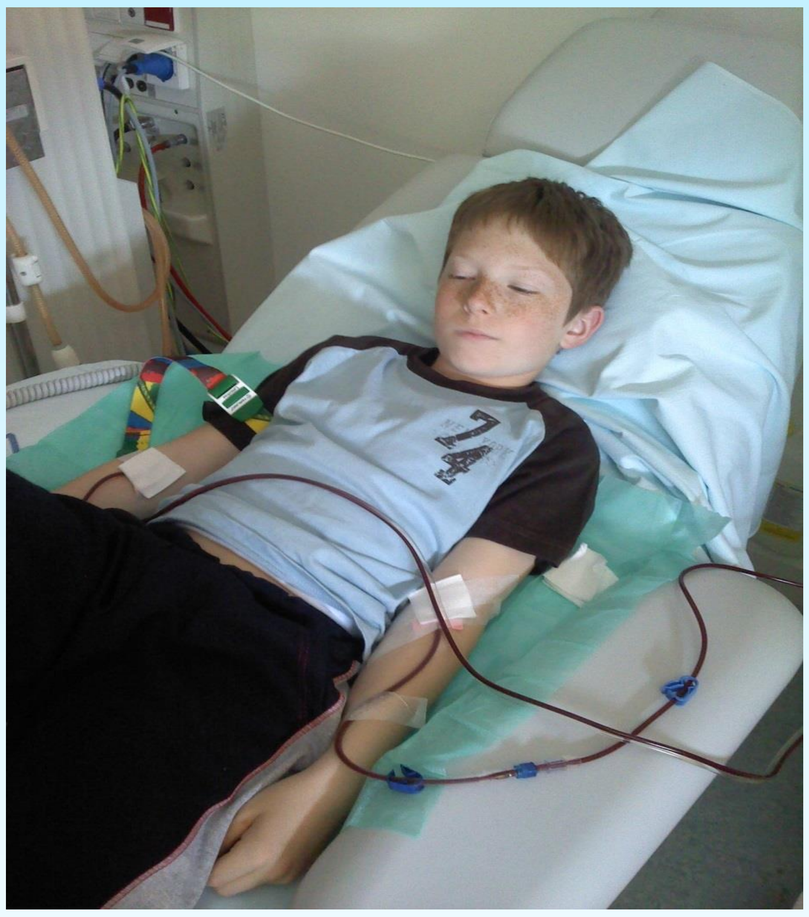
Introduction

Family hypercholesterolemia is an autosomal dominant disorder in which there are mutations in the receptor for apolipoprotein-B in the hepatocytes causing a reduced removal of LDH. Homozygous familiar hypercholesterolemia is a severe aggressive form of the disease that does not respond to traditional forms of treatment. The concentration of LDL cholesterol is usually between 16.8 to 25.9 mmol/l, the occurrence of xanthoma is monitored from the age of four, and ischemic heart disease with possible fatal outcome is diagnosed at the age of 20.

CASE REPORT

At the then six- month boy, born from a regular pregnancy, the appearence of orange skin growths was monitored. We learned from family history that his mother, father as well as his father and mother´s parents have hypercholesterolemia.
Comprehesive diagnostic evaluation detected an extremely high level of total cholesterol (29 mmol/l). The genetic testing was completed and it ultimately diagnosed homozygous familiar hypercholesterolemia.
The amendment process did not reveal atherosclerotic plaques in the vascular system nor the deposit of cholesterol the corneas. The treatment started with a diet and a statin, but without significant effect (total cholesterol 22- 23.3 mmol/l). The LDL- apheresis was imposed as the only acceptable treatment option.



Methods

- The first procedure of LDL- apheresis was carried out by CVC at the age of six, when he weighed 23 kg, with a starting value of LDL 29 mmol/l and the treated blood volume of 810 ml.
- To date 380 LDL- apheresis treatments have been done, in the interval of one therapy per week.

Results

Today the boy is 17 years old and weighs 65 kg.
The treated blood volume is 6 L.Total LDL at the beginning of the procedure amounts to 7 mmol/l, and at the end 3,3 mmol/l. Regular re-evaluations do not find complications of the underlying disease.

Conclusion

LDL- apheresis reduced morbidity and mortality from cardiovascular complications homozygous familial hypercholesterolemia by slowing the development and progression of atherosclerotic plaques. LDL- apheresis reduces serum concentrations of LDL cholesterol and Lp to about 70% by a procedure, which is a 40%- 50% in long- term chronic therapy treatments. LDL- apheresis therapy in patients suffering from severe hyperlipidemia who do not respond to conservative therapy is effective and safe in long- term use.