Efficiency of low-density lipoprotein apheresis (LDL-a) in a 6-year-old girl with homozygous familial hypercholesterolemia

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We report here a case of a 6-year-old girl with homozygous FH-Palermo-1 mutation in the LDL-receptor gene and extensive coronary atherosclerosis. This patient developed tendinous and planar xanthomas from age 3. Laboratory evaluation showed LDL-cholesterol level at 22.25 mmol/l. Angiography revealed severe proximal stenosis in the left coronary and ostial stenosis in the right coronary. Weekly LDL-a was initiated with a dextran sulphate cellulose (DSC) column absorption through a jugular central catheter, with a filtred plasma volume at each procedure of 2000 ml. Pre-treatment LDL-cholesterol level is between 7.70-8.10 mmol/L and fall to 1.9-2.50 mmol/L at the end of the apheresis, corresponding to the expected 75% reduction of LDL-cholesterol plasmatic level. During a coronarography done eight months after starting the treatment, she presented a left coronary spasm with severe bradycardia and a stent had to be placed at the origin of the left coronary artery. A second cardiac evaluation done eighteen months after weekly LDL-a procedure and medical treatment (atorvastatin and ezetimibe) showed stabilisation of coronary atherosclerosis. Disappearance of skin xanthomas was observed inbetween.

In conclusion, LDL-a is a safe and efficient procedure which allow to stop progression of atherosclerotic lesions in a child with severe familial hypercholesterolemia. The efficiency of a low risk but life-long procedure needs to be put in balance with the risk of a curative liver transplantation.

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