Familial hypercholesterolemia with coarctation of aorta

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Familial hypercholesterolemia (FHC) is a rare genetic disorder affecting 0.2% of the general population and is characterized by increase in low-density lipoprotein (LDL) cholesterol. The incidence of the homozygous form of the disease is about one in one million.\(^1\) Aortic valve and coronary arteries are commonly involved in FHC.\(^2,3\) Coarctation of aorta is a rare association.

A six-year-old female presented with multiple nodular swellings over elbow and wrist which were noticed for last three years [Figures 1, 2]. She had history of frequent episodes of headache and vomiting. Family history was suggestive of sudden death in three siblings [Figure 3]. The exact cause of death was not available but all of them had xanthomas. Parents had an abnormal lipid profile. Father had myocardial infarction at the age of 35 years. On examination, the child had a pulse rate of 98/min. Blood pressure in the lower limbs was less than upper limbs. Examination of cardiovascular system revealed ejection systolic murmur Grade IV over left third intercostal space, which was radiating upwards. Left renal bruit was audible.

Investigations revealed abnormal lipid profile with a total cholesterol of 810 mg/dL, LDL of 724 mg/dL, HDL 18 mg/dL, VLDL 18 mg/dL, and triglycerides of 93 mg/dL. Her chest X-ray revealed cardiomegaly. The ECG revealed left ventricular hypertrophy. Echocardiography revealed bicuspid aortic valve with aortic stenosis and diffuse coarctation of descending aorta. Doppler was suggestive of diffuse left renal artery stenosis. Hypertension was controlled with calcium channel blockers and she was started on lovastatin 4 mg/kg. Her repeat lipid profile after two months of therapy revealed total cholesterol 200 mg/dL, LDL of 118 mg/dL and triglyceride of 74 mg/dL.

Patient was referred to a cardiac centre for further management and was lost to follow-up.

In FHC of homozygous variety LDL-cholesterol levels are between 500-1000 mg/dL as seen in our patient. Pathogenesis in involvement of aortic valve and coronary arteries is an impaired endothelial function in prepubertal children with familial hypercholesterolemia.\(^3\) There are reports of aortic stenosis and coronary artery disease in cases of FHC, though coarctation of aorta as seen in our case has not been reported.

The current recommended management for children with familial hypercholesterolemia includes a cholesterol-lowering diet, and a bile acid binding resin and statins. Available data from limited number of controlled trials suggest that statin treatment is also effective and safe in children.\(^4\) Plasmapheresis is the treatment of choice in the homozygous form but it is expensive and has to be done on a biweekly basis.\(^5\) The outcome is likely to be poor in people with homozygous type of familial hypercholesterolemia because it causes early heart disease and is resistant to treatment.

References


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