ABSTRACTS OF CASE REPORTS

CR 09

A Baby with Familial Hypercholesterolaemia

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Introduction

Prevalence of autosomal dominant homozygous familial hypercholesterolaemia (FH) is 1 in 250,000 and heterozygous FH is 1 in 250. Homozygous individuals have severe symptoms with onset of heart disease before the age of 30 years.

Case presentation

A 2 ½-year-old girl presented with yellow coloured patches over the buttock region since the age of eight months. These patches gradually spread to knees, ankles and elbows. Initial lipid profile revealed very high total cholesterol (775 mg/dL) and high LDL (697 mg/dL) with a normal triglyceride level. Tendon xanthomas observed in the Achilles tendon, knee, elbows and buttocks. No xanthelasma was observed. Fundoscopy revealed no arcus cornealis with a normal echocardiogram and ultrasound scan of the abdomen. Younger sister also has tendon xanthomas. There is a strong family history from maternal side (uncles) with two premature deaths due to ischaemic heart disease. Father had a premature myocardial infarction. Index patient and the younger sister were homozygous for a pathogenic variant in the LDLR gene. Parents are heterozygous. Babies were started with simvastatin 10 mg nocte, diet control and vitamin K supplements. They were followed up with lipid profiles and dose was increased accordingly. Now the patient is on simvastatin 70 mg nocte.

Discussion

Diagnosis of familial hypercholesterolemia was made with the genetic studies. Newer treatment options available include PCSK 9 inhibitors, plasmapheresis, partial ileal bypass surgery and liver transplantation which is the treatment of choice.

Keywords

LDLR, PCSK 9, tendon xanthomas, familial hypercholesterolaemia