Xanthoma tuberosum in homozygous familial hypercholesterolemia

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ABSTRACT

Familial homozygous hypercholesterolemia is one of the high risk factors that can result in premature coronary arterial disease leading to severe morbidity and premature death in children and young adults. We describe a rare example of extensive xanthoma tuberosum in a case of familial homozygous hypercholesterolemia.

Keywords: Coronary artery disease, hypercholesterolemia, xanthoma tuberosum

INTRODUCTION

Homozygous FH is rare, with symptoms appearing in childhood, and is associated with early death from coronary heart disease. Homozygous FH has an incidence of approximately one case per one million.

A 19-year-old male presented with effort angina of 6-month duration, which has worsened since 1 month associated with rest angina, and postprandial angina. Physical examination revealed multiple tuberous xanthomas over both shoulders, elbows and wrists [Figure 1], which was noticed at the age of 8 years which was gradually progressing in size. His other siblings also had similar xanthomas but they were asymptomatic. His fasting lipid profile is showed total cholesterol of 498 mg/dl, LDL-436, HDL 18.6 mg/dl, VLDL 17.3 g/dl and triglycerides 81 mg/dl. ECG showed non-specific ST-T changes. 2D Echocardiography was normal. He underwent elective coronary angiography that showed critical stenosis of left anterior descending artery and severe disease in the proximal left circumflex [Figure 2a, Video 1] and proximal right coronary arteries [Figure 2b, Video 2]. Later he underwent coronary artery bypass graft surgery and he is asymptomatic during follow-up with intensive statin therapy (Rosuvastatin 40 mg/day) with partial regression in the size of xanthomas. Repeat fasting lipid profile after

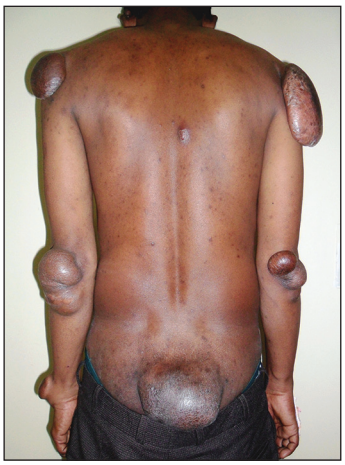

Figure 1: Showing extensive xanthoma tuberosum around shoulders, elbows, wrists, and sacrum

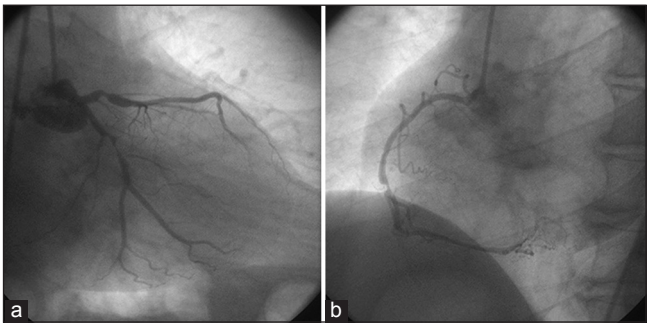

Figure 2: (a) Left coronary angiography showing severe proximal LAD and proximal LCX stenosis (b) Right coronary angiography showing focal severe stenosis in mid-RCA

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3 months of statin therapy showed 40% reduction in LDL and total cholesterol and slight rise in HDL cholesterol.

Familial homozygous hypercholesterolemia is one of the high risk factors that can result in premature coronary arterial disease leading to severe morbidity and premature death in children and young adults. Clinical identification of xanthomas, and knowledge of their association with coronary arterial disease, is essential for every primary care physician and paediatrician, as early diagnosis and treatment can prevent premature deaths due to myocardial infarction.[1]

REFERENCE

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