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Images in Cardiology

Tendon xanthomas as indicators of atherosclerotic burden on coronary arteries

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ABSTRACT

The presence of tendon xanthomas is an almost certain indicator of familial hypercholesterolemia (FH). They also reflect coronary atherosclerotic burden and therefore must be treated aggressively. Tendon xanthomas also occur in two rare conditions, cerebrotendinous xanthomatosis and sitosterolemia, which are not easily confused with FH, can be easily differentiated with clinical history and biochemical tests.

A 24-year-old gentleman presented with a history of exertional fatigue, probably angina equivalent and multiple swellings all over the body for the past 1 year. Physical examination revealed several different sized swellings (ranging from 1 to 6 cm in diameter) over the extensor aspects of elbow, hands, hips, knees and feet (Fig. 1A and B). They were firm and painless swellings. The resting electrocardiogram, echocardiography were normal. The lipid profile showed a total serum cholesterol of 590 mg/dL, LDL cholesterol of 430 mg/dL, HDL cholesterol of 40 mg/dL, triglycerides of 135 mg/dL. The exercise stress test was done, which was strongly positive. Subsequently, an elective coronary angiogram revealed left main disease with triple vessel disease (Fig. 1C and D). Patient was advised with LDL apheresis, which he refused. Patient opted for medical management and so was treated with high-dose statins and ezetimibe. The patient did not return for follow-up, so we do not know the effect of medical therapy on lipid profile.

Familial hypercholesterolemia (FH) is an autosomal codominant disorder characterized by elevated LDL-C with normal triglycerides, tendon xanthomas and premature coronary artery atherosclerosis. The Food and Drug Administration (FDA) has approved the use of dextran sulfate–cellulose adsorption (DSA) and heparin-induced extracorporeal LDL precipitation (HELP) apheresis system in three categories of patients in the US: functional FH homozygotes, with LDL cholesterol >500 mg/dL; functional FH heterozygotes, with LDL cholesterol >300 mg/dL; and functional FH heterozygotes with documented CHD and LDL cholesterol >200 mg/dL.

Tendon xanthomas also occur in two rare conditions, cerebrotendinous xanthomatosis and sitosterolemia, which can be easily differentiated by clinical history and biochemical tests.

Cerebrotendinous xanthomas is a rare genetic metabolic disorder characterized by infantile-onset diarrhea, childhood-

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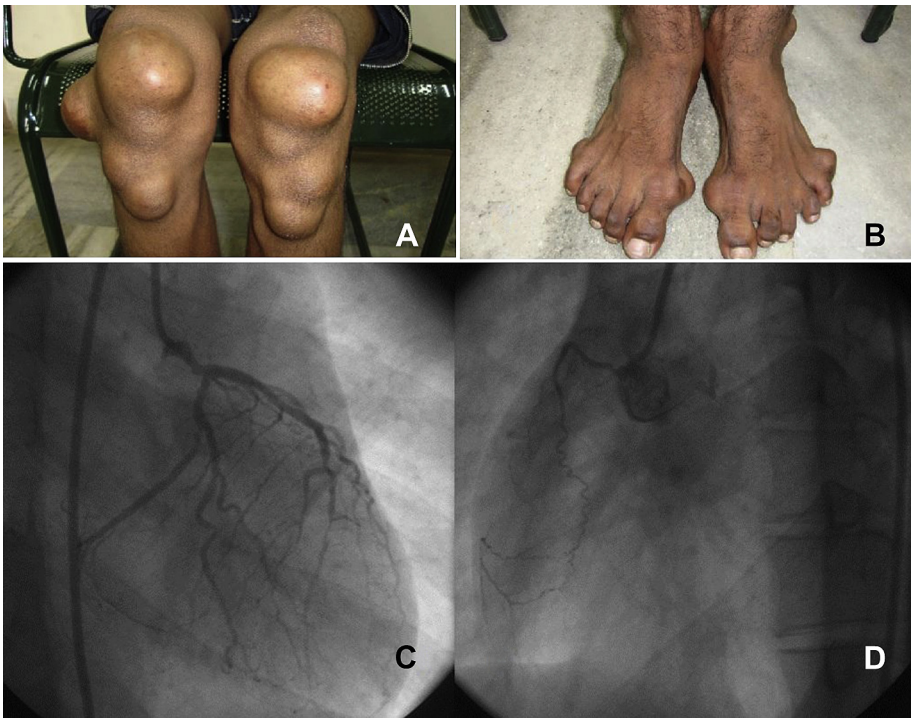

Fig. 1 – A – Multiple tendon xanthomas on the knee joint, B – Tendon xanthomas on the feet, C – Distal left main, ostial circumflex and distal left anterior descending artery shows atherosclerotic stenotic lesions, D – Right coronary artery shows total occlusion.

onset cataract, adult-onset tendon xanthomas with progressive neurological dysfunction. The abnormalities in the blood include high levels of cholesterol, decreased chenodeoxycholic acid, and increased concentrations of certain bile alcohols. Genetic testing for the gene associated with CTX, *CYP27A1* is also available.

Sitosterolemia, is an inherited sterol storage disease characterized by tendon xanthomas and by a strong predisposition to premature atherosclerosis. The serum concentration of plant sterols – sitosterol and campesterol is increased. The tendon xanthomas can regress completely with treatment in some cases.

It is important to treat FH aggressively to prevent acute coronary syndromes and the associated complications and also to prevent progression of atherosclerosis.

Conflicts of interest

All authors have none to declare.