Tendinous xanthoma with familial hypercholesterolemia

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A 10-year-old non-obese boy presented with a painful nodular eruptions over the back of ankles for the last 5 years. On examination smooth, skin colored painful nodules were noted over the lower part of each tendo-achilles [Figure 1] and left elbow [Figure 2]. The skin overlying the nodules was freely movable. Arcus juvenilis was present. His parents and two siblings did not show any such findings. A diagnosis of tendinous xanthoma was considered and the patient was subjected to investigations. The lipid profile and the lipoprotein electrophoresis revealed elevated levels of low-density lipoprotein (LDL) cholesterol (453 mg/dl) and total cholesterol (600 mg/dl). Blood sugar, liver function tests, thyroid profile, chest radiograph, electrocardiogram, glucose tolerance test and ultrasonography of the abdomen were normal. There was no evidence of any coronary artery disease. Biopsy from the nodule revealed infiltration of the dermis with admixture of foam cells, histiocytes and lymphocytes [Figure 3]. The lipid profile of the parents showed a rise in the LDL cholesterol. A final diagnosis of familial hypercholesterolemia with tendinous xanthoma was made.

DISCUSSION

Xanthomas are commonly caused by a disturbance of lipoprotein metabolism.[1] Tendon xanthomas are cholesterol deposits in tendons. They appear as slowly enlarging papules or subcutaneous nodules attached to tendons, ligaments, fascia and periosteum and commonly affect the tendons of the dorsal surface of the hands and the achilles tendon. Their presence is a clinical sign of familial hypercholesterolemia, an autosomal dominant inherited disorder characterized by high LDL cholesterol levels and premature cardiovascular disease.[2] Triglycerides are generally normal and high-density lipoprotein cholesterol either normal or reduced.[3] Xanthomas are tumor-like collections of foamy histiocytes within the dermis. Histopathologically, xanthomas are characterized

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Figure 1: Nodules over tendo-achillis
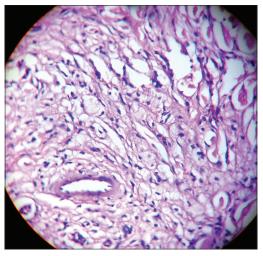

Figure 2: Nodules over elbow

Figure 3: Histopathology from lesion showing foam cells, histiocytes and lymphocytes (H and E, x400)

by the presence of vacuolated macrophages in dermis. These macrophages are filled with lipid droplets that are dissolved and removed during tissue processing.

Dietary modification and treatment with the statin group of drugs is recommended. Gemfibrozil is effective in patients with Type II (both a and b) and IV hyperlipidemia.[4]

This case highlights the importance of the dermatologist in diagnosing hypercholesterolemia, as children may first present to them with xanthomas. Early diagnosis and treatment may help in preventing the development of early coronary artery disease.

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