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Images in Medicine

Tendinous xanthoma

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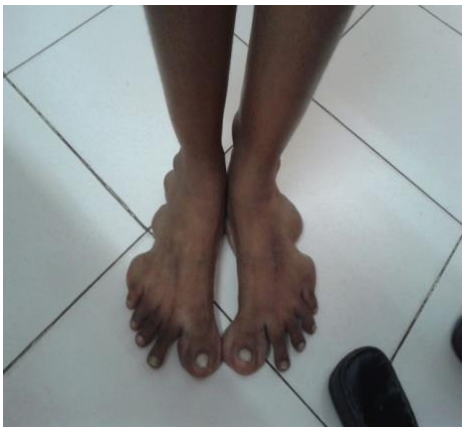

Figure 1: Tendinous xanthoma feet.

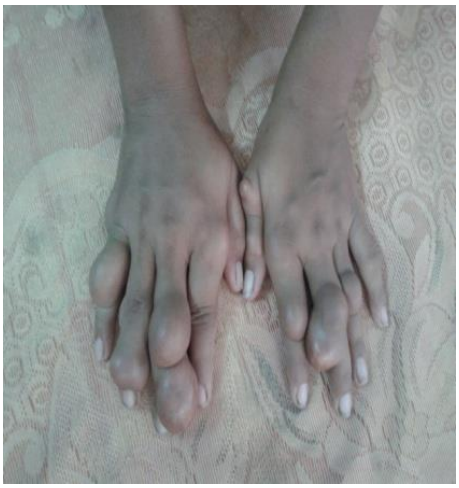

Figure 2: Tendinous xanthoma hands.

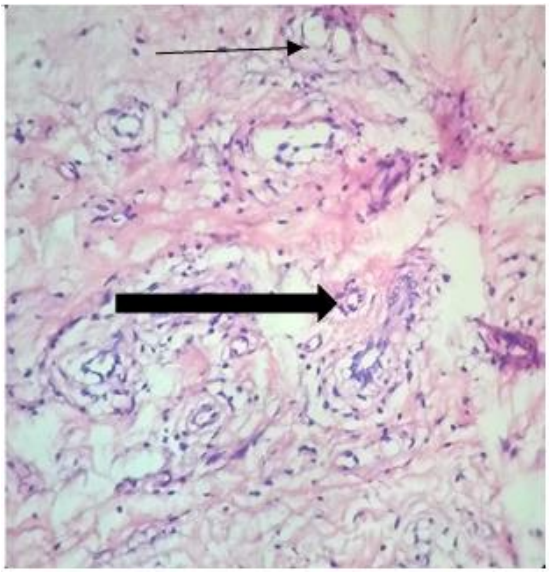

Figure 3: Xanthoma - showing lipid laden clear cells (small arrow) and giant cell (black arrow) H&E 40x10x.

A 31 years old female presented with multiple nodular, progressive swellings over feet since her age of 16 years & similar type of swellings both hands (Figure 1 & 2). She was non diabetic, non-hypertensive & not on any Thyroid replacement therapy. And no past history of coronary heart disease. She had no family history of sudden cardiac death except her 19yrs brother was having similar kind of swellings. Her physical examination revealed multiple subcutaneous, firm, non-tender swellings involving feet &hands (Figure 1 & 2).

Rest of the clinical exam was normal. Investigations showed normal hemogram, renal function, liver functions, thyroid function & blood sugar. Electrocardiogram, X-ray chest, 2D echo and abdominal ultrasound were normal. Her lipid profile revealed serum cholesterol 498 mg/dl, triglyceride 172 mg/dl, high density lipoprotein 38.57 mg/dl, low density lipoprotein cholesterol 345 mg/dl, and very low density lipoprotein 45 mg/dl. Histopathological findings of the swelling showed fibrocollagenous tissue with minimal mononuclear inflammation along with foamy cells with extra cellular lipid collection & giant cells suggestive of xanthomas (Figure 3).

The presence of tendinous xanthomas, dyslipidemia & family history of tendinous xanthomas diagnosis of primary familial hypercholesterolemia (Frederickson's type IIa) was entertained. She was managed with Atorvastatin 40mg daily& supportive therapy (diet, regular exercises) and is on regular follow up. Her brother is also on similar kind of treatment & under our follow up.

Xanthomas are accumulation of lipid laden macrophages which develop due to altered cell metabolism in conjunction with elevated lipids. Primary hyperlipoproteinemia is due to genetic mutations that yield defective apolipoproteins and secondary hyperlipoproteinemias results from various disorders, such as diabetes mellitus, hypothyroidism, nephrotic syndrome & certain drugs like beta blockers, thiazides & anti-depressants.[1] Hyperlipidemias have been classified in to 6 types by Frederickson based on electrophoretic patterns of lipoprotein. Cutaneous xanthomas associated with hyperlipidemia are clinically subdivided into xanthelesma palpebrarum, tuberous xanthoma, tendinous xanthoma, eruptive xanthoma, and planar xanthoma. Tendinous xanthomas are seen in majority of adults with familial hypercholesterolemia. They are also seen in familial defective apoB100 type 3 hyperlipoprotenemia and sitosterolaemia.[2] Lipid lowering therapies usually does not lead to resolution of tendinous xanthomas. Surgical & locally destructive options should be explored in resistant cases.[3]

Our patient had tendinous xanthomas with underlying familial primary hypercholesterolaemia.

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