Tendinous Xanthoma

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A 45 year old lady on levothyroxine replacement for hypothyroidism, presented with progressively worsening angina over past 6 months. She had no past history of coronary artery disease, diabetes, hypertension or family history of sudden cardiac death. On examination, there was arcus lipidicus, xanthelasma, multiple, subcutaneous, firm nontender swellings involving feet (Figure 1), elbows (Figure 2) and yellowish maculopapular lesions over her thighs (Figure 3). Rest of the clinical examination was normal. Investigations revealed normal hemogram and biochemistry screen including sugar, liver, kidney and thyroid function tests. Electrocardiogram revealed ST segment depression and T wave inversion in anterior leads. The patients lipid profile revealed serum cholesterol 473 mg/dL, low density lipoprotein cholesterol - 390 mg/dL, triglycerides - 173 mg/dL, high density lipoprotein - 45 mg/dL and very low density lipoprotein - 62 mg/dL. The presence of tendon xanthomas along with xanthelesma indicates familial hypercholesterolemia (Frederickson’s Type Ha). Her echocardiography revealed anterior wall hypokinesia and coronary angiography showed significant stenosis in left anterior descending branch of left coronary artery. She was managed with coronary stenting, atorvastatin 40 mg daily along with other supportive therapy and is on regular follow up. None of the screened family members had hyperlipidemia. 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 result from various disorders, such as diabetes mellitus, hypothyroidism and nephrotic syndrome. Hyperlipidemias have been classified into 6 types by Fredrickson based on the electrophoretic patterns of lipoproteins. Cutaneous xanthomas associated with hyperlipidemia are clinically subdivided into xanthelasma 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 apo B100, type 3 hyperlipoproteinemia, and sitosterolemia. Lipid lowering therapies usually does not lead to resolution of tendinous xanthomas. Surgical and locally destructive options should be explored in resistant cases. To conclude, our patient had three different clinical types of xanthomas with underlying familial hypercholesterolemia.

References


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