CASE REPORT

TENDON AND TUBEROUS XANTHOMAS, A CLINICAL SIGN OF FAMILIAL HYPERCHOLESTEROLEMIA

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ABSTRACT: We report a case of a 23 year old male patient who was admitted with congestive cardiac failure secondary to ischemic dilated cardiomyopathy. On examination, he had multiple tendon and tuberous xanthomas. His lipid profile revealed high LDL and total cholesterol and normal triglyceride levels. He was diagnosed as a case of familial hyperlipoproteinemia type II A. Awareness about the manifestations of familial hypercholesterolemias would help in the early diagnosis and treatment and thus delay or prevent the morbidity and mortality associated with the disease.

CASE REPORT: A 23 year old male was admitted with breathlessness since 8 months which was insidious in onset and gradually progressive and was associated with episodes of paroxysmal nocturnal dyspnoea. He later developed orthopnea. He also had complaints of left side chest pain and cough. Since 2 months he had started noticing ankle oedema. He had no history of palpitations, cyanosis or syncopal attacks. Patient also gave a history of multiple swellings over the extermities since childhood which were painless and not increasing in size. He gave no history suggestive of rheumatic fever, infective endocarditis, recurrent lower respiratory tract infections in childhood. He was not a known diabetic, hypertensive, asthmatic. He had no history of intake of drugs. Father is a known case of ischemic heart disease since the age of 46 years. No other family members suffered from any cardiac illness. No history of consanguinity in parents.

On examination, patient was dyspnoeic, had tachycardia and BP -110/ 80 mmHg. His JVP was raised. He had multiple, subcutaneous, firm, nontender swellings involving ankles, elbows and knees. Respiratory system revealed bilateral basal crepitations. Cardiovascular system examination showed a hyperdynamic apical impulse, gallop rhythm and pansystolic murmur in mitral area.

With the suspicion of familial hypercholesterolemia, his lipid profile was done which showed total cholesterol value of 381 mg/dL, LDL -353mg/dL, HDL- 18.4 mg/dL and triglyceride levels – 111mg/dL. His 2DECHO revealed dilated cardiomyopathy with an ejection fraction 10%. ECG showed left bundle branch block, chest Xray showed cardiomegaly. His blood sugar values and renal function tests were normal. His Hemoglobin was 9.6 gm/dL, total leukocyte count and platelet count were normal. LFT was normal except for hypoalbuminemia. Ultrasound abdomen showed bilateral renal calculi with hydroureteronephrosis.

He was diagnosed as a case of familial hyperlipoproteienmia type II A due to the presence of tendon and tuberous xanthomas, ischemic heart disease and high total and LDL cholesterol values in the presence of normal triglyceride levels.

DISCUSSION: Tendon xanthoma is a clinical sign of familial hypercholesterolaemia (FH), an inherited disorder characterized by high lowdensity lipoprotein (LDL) cholesterol levels and premature cardiovascular disease. These are nodular deposits of cholesterol that accumulate in tissue macrophages in the Achilles, patellar tendon, elbow, and hand tendons. The tendons involved with those fatty deposits are biomechanically weaker and tend to be more susceptible to tendon injuries and ruptures. Tuberous xanthomas develop in areas that are susceptible to trauma such as elbows and knees.[1] They are seen in familial hypercholesterolemia, familial defective apo B 100 and sometimes in those with dysbetalipoproteinemias.[2,3]

The presence of tendon xanthomas is associated with a three-fold higher risk of CVD in patients with FH. This finding suggests that xanthomas and atherosclerosis share pathophysiological mechanisms. The composition of xanthomas and atherosclerotic plaques show similarities as both lesions consist of connective tissue matrix containing macrophages transformed into foam cells. In addition, both xanthomas and plaques can be prevented or reduced by the use of cholesterol lowering drugs. Variants in genes of the RCT pathway and the LDL oxidation pathway are associated with the presence of tendon xanthomas in FH patients.[4]

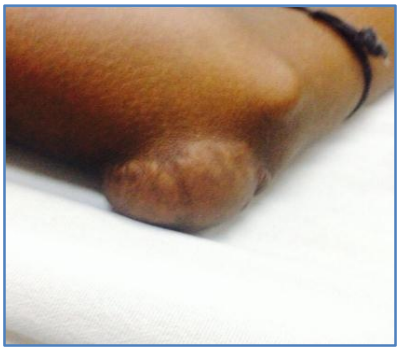
Because the Achilles tendon is the most common location for xanthomas to develop, ultrasonography of theAchilles tendon has been demonstrated to increase sensitivityup to 75%.[5] A speckled on reticulated appearance on axial MR images, with or without enlargement, is a characteristic, if not pathognomonic, feature of xanthomatous Achilles tendons.[6] Because musculoskeletal symptoms (xanthomas) often precede the diagnosis of hyperlipidemia, it is important to be familiar with FH because patients with FH are at high risk for premature coronary atherosclerosis.[1]

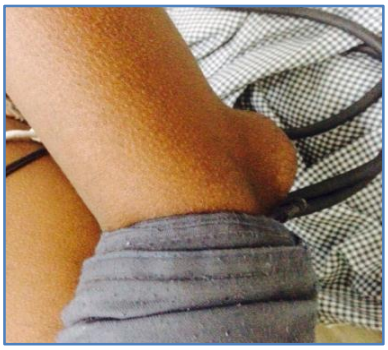
CONCLUSION: Familial hypercholesterolemia is an inherited disorder characterized by high lowdensity lipoprotein (LDL) cholesterol levels,normal triglycerides, premature cardiovascular disease, tendon and tuberous xanthomas. The presence of tendon xanthomas is associated with a three-fold higher risk of CVD in patients with FH.As xanthomas precede the onset of cardiovascular disease, awareness about it would help in the early diagnosis and treatment of familial hypercholestrolemia.

REFERENCES:

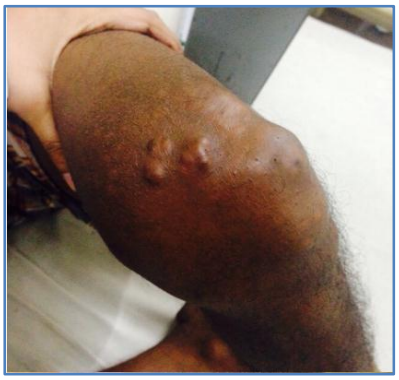
1. Huri G, Joachim N. An Unusual Case of Hand Xanthomatosis. Case reports in Orthopaedics. Hindawi Publishing Corporation. 2013: Article ID 183018; 1-3.
2. Kumar KVSH, Ghosh AK, Kumar KK, Prusty P. Tendinous Xanthoma. JAPI: 2012(60); 46.
3. SK Jain, Nagina Agarwal. Tendon Xanthomas. JIACM 2003; 4(1): 69.
4. MO Daniella et al. The risk of tendon xanthomas in familial hypercholesterolaemia is influenced by variation in genes of the reverse cholesterol transport pathway and the low-density lipoprotein oxidation pathway. European Heart Journal (2010) 31, 1007–1012.
5. Civeira F. et al. Tendon Xanthomas in familial hypercholesterolemia are associated with cardiovascular risk Independently of the low-density lipoprotein receptor gene mutation. Arterioscler Thromb Vasc Biol. 2005; 25: 1960-1965.
6. Bude R 0, Adler RS, Bassett DR. Diagnosis of Achilles Tendon Xanthoma in Patients with Heterozygous Familial Hypercholesterolemia: MR vs Sonography. AJR 1994; 162: 913-917.

CASE REPORT





Tendon Xanthoma



Tuberous Xanthoma

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