ejpmr, 2016,3(8), 562-564

SJIF Impact Factor 3.628

EUROPEAN JOURNAL OF PHARMACEUTICAL AND MEDICAL RESEARCH

www.ejpmr.com

Case Report

ISSN 2394-3211

EJPMR

TUBEROUS XANTHOMA IN AN AMPUTATED STUMP

Meena Rinoo Banwarilal, Kumar Prateek*, Shyam Sundar Chaudhary, Sagarika Chatterjee, Raj Kishor, Anu Garg

Rajendra Institute of Medical Sciences, Ranchi-834003, Jharkhand, India.

Corresponding Author: Kumar Prateek

Rajendra Institute of Medical Sciences, Ranchi-834003, Jharkhand, India.

Article Received on 14/06/2016 **Article Revised on 05/07/2016** **Article Accepted on 26/07/2016**

ABSTRACT

Xanthomas are focal infiltrates of lipid-containing histiocytic foam cells that are usually found within the dermis or tendons. It is a clinical manifestation of lipoprotein metabolic disorders. We report a case of tuberous xanthoma in an 45 year old woman who presented with multiple firm papules and nodules involving multiple sites of the body including right hand amputated with elevated serum cholesterol, triglycerides, LDL and VLDL.

KEYWORDS: It is a clinical manifestation of lipoprotein metabolic disorders.

INTRODUCTION

Xanthoma is a deposition of yellowish cholesterol rich material that can appear anywhere in the body in various disease states. They are cutaneous manifestations of lipidosis in which lipids accumulate in large foam cells within the skin. Xanthomas are plaques or nodules consisting of abnormal lipid deposition and foam cells in skin or in tendons. They do not represent a disease but rather are signs of a variety of lipoprotein disorder. Clinically, xanthomas can be classified [Table 1] as eruptive, tubo-eruptive or tuberous, tendinous or planar. Tubo-eruptive xanthoma are nodules that are frequently localized to the extensor surface of the elbow, knees, knuckles and buttocks.

CASE REPORT

A 45 year old female patient presented to us with asymptomatic multiple nodules over left elbow, left hand over dorsal aspect on kunckles, amputated stump of right forearm, both the legs on knees, soles and bottocks since 5 years. There was no complain of fever, itching. No history of similar lesion in family. No history of diabetesmellites, hypertension, tuberculosis and asthma. History of snake bite 20 years back on right hand which became gangrenous and eventually amputated.

On examination, multiple yellowish shiny, smooth nodules are present over left elbow(Figure1).Multiple small papules are also present around it which coalesce forming a nodule. Similar lesions are present over palmar aspect of left fingers, dorsum of the fingers over knuckles of left hand(Figure4),right arm over elbow joint and stump of amputated right hand(Figure3),both legs over knees, around ankle joint over medial and lateral aspect,over both sole and buttocks.

Figure1 Multiple yellowish shiny nodules on left elbow. A few papules also noted.

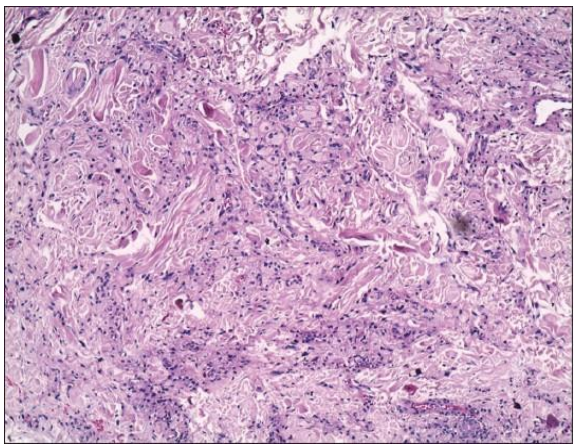

Figure2 Diffuse dense infiltrate of foamy histiocytes involving whole of dermis. H & E 40x (Biopsy from nodule over thigh).

Figure: 3 Similar lesions on amputated right hand.

Figure: 4 Multiple shiny nodules on dorsum of fingers of left hand.

Table 1

Clinical Presentations of Xanthomas

Type of Xanthoma	Genetic Disorders	Secondary Disorders
Eruptive	Familial lipoprotein lipase deficiency ApoC-II deficiency ApoA-I and apoA-I/C-III deficiency Familial hypertriglyceridemia Familial hypertriglyceridemia with chylomicronemia	Obesity Cholestasis Diabetes Medications: retinoids, estrogen therapy, protease inhibitors
Tuberous	Familial hypercholesterolemia Familial dysbetalipoproteinemia Phytosterolemia	Monoclonal gammopathies Multiple myeloma Leukemia
Tendinous	Familial hypercholesterolemia Familial defective apoB Familial dysbetalipoproteinemia Phytosterolemia Cerebrotendinous xanthomatosis	
Planar		
Palmar	Familial dysbetalipoproteinemia Homozygous apoA-I deficiency	
Intertriginous Diffuse	Familial homozygous hypercholesterolemia Familial hypercholesterolemia	Cholestasis Monoclonal gammopathies, cholestasis
Xanthelasma	Familial dysbetalipoproteinemia	Monoclonal gammopathies
Other		
Corneal arcus Tonsillar	Familial hypercholesterolemia Tangier disease	

On investigation complete blood count,renal function test,liver function test,thyroid function test are within normal limits.Only her alkaline phosphates enzyme level are slightly raised [146 U/L]. Routine urine within normal limits.Her blood sugar, fasting=98 mgm%, postprandial 153 mgm%.Her lipid profile revealed, Serum cholesterol- 686 mgm%, Serum triglycerides- 743 mgm%, HDL cholesterol- 37 mgm%, LDL cholesterol- 342 mgm%, VLDL Cholesterol- 149 mgm%.

HISTOPATHOLOGICAL examination showed a diffuse, dense, nodular infiltrate of foamy histiocytes involving whole of the dermis. The epidermis above is flattened and surrounding dermis shows mild fibroplasias. The foamy histiocytes show abundant pale foamy cytoplasm and a small centrally located nucleus. Biopsy thus confirmed diagnosis of tuberous xanthoma. Electrophoresis was not done as this facility is not available at our institute.

DISCUSSION

The term XANTHOMA derived from the greek word 'XANTHOS' meaning yellow and is used to describe a variety of subcutaneous lipid deposits,evn those that do not appear yellow.[1] Xanthomas are common presentation of a focal or generalized disorder of lipid metabolism. They are often linked with high risk of arteriosclerotic vascular diseases, pancreatitis, etc. During childhood, Type I and Type IIa are seen and are hereditary.[2] They are characterized by accumulation of

lipid laden macrophages. Pathogenesis involves a complex process by means of dysregulation of macrophage sterol flux. Xanthomas are predisposed by increased levels of cholesterol rich LDL and VLDL remnants. Under normal circumstances, around 80% of LDL clolesterol are taken up by LDL receptor mediated endocytosis. The residual LDL is cleared by scavenger receptor pathways of macrophages. In familial hypercholesterolemia, the accumulation LDL and VLDL remnants are primarily scavenged by macrophages without feedback regulation, resulting in continuous cellular lipid accumulation and foamy cells. Clinically tuberous xanthomas are characterized by yellow to skin colored nodules that are firm, painless, indurated with an erythematous halo and are generally localized to extensor surfaces of buttocks, knees, elbows and knuckles. The lesions can be seen in inguinal and axillary folds and in the sites of trauma and they may be fissured, pedunculated and suppurative.

Frequently associated with primary hypercholesterolemia which includes familial hypercholesterolemia (Friederickson type II) and dyslipoproteinemia (Friederickson type III, an autosomal dominant genetic disorder of lipid metabolism) and in secondary hypercholesterolemia with biliary disease, monoclonal gammopathy and hypothyroidism. In familial hypercholesterolemia, raised LDL levels are due to increased production and decreased resorption of LDL secondary to dysfunctional LDL receptors. As a result of altered endothelial function, elevated serum total cholesterol and LDL with normal triglycerides are found and manifests as atherosclerosis and coronary artery disease.[3] Histopathological examination reveals large and small aggregates of foamy cells, which are macrophages engulfing lipid droplets. Xanthoma cells are mononuclear and may also show Touton giant cells with a wreath of nuclei. Early lesions show a mixture of non foamy cells, neutrophils, lymphocytes and macrophages and well developed lesions show infiltrates consisting mainly of foamy cells which are later replaced by collagen bundles. Cholesterol clefts are also seen.[4] Treatment options include lifestyle modification with dietary changes and pharmacotherapy including statins, eczema, niacin and bile acid sequestrants like cholestyramine. This condition responds well to a combined therapy involving statins, cholesterol absorption inhibitors and a bile acid sequestrant, if needed.[5] Invasive procedures namely lifelong lipid apheresis and liver transplantation can also be considered.[6] Tuberous xanthoma shows slower rate of regression after appropriate therapy.

REFERENCES

1. Kumar B, Dogra S. Metabolic disorders. In: Valia RG, Valia AR, eds. IADVL Textbook of dermatology. 3rd edition. Mumbai: Bhalani Publishing House, 2008; 1298-37.
2. Mohan KK, Kumar KD, Ramachandra BV. Tuberous Xanthomas in type IIA hyperlipoproteinemia. Indian J Deramtol Veneral Leprol, 2002; 68: 105-106.
3. Austin MA, Hutter CM, Zimmern RL, Humphries SE. Familial hypercholesterolemia and coronary artery disease: A huge association review. Am J Epidemiol, 2004; 160: 421-9.
4. Walter H.C Burgdorf, Berhard Zelger. The histiocytosis. Lever histopathology of skin, 10th edition, chap, 2008; 26: 680-686.
5. Avis HJ, Vissers MN, Stein EA, Wijburg FA, Trip MD, Kastelein JJ et al. A systematic review and meta-analysis of statin therapy in children with familial hypercholesterolemia. Arterioscler Thromb Vasc Biol, 2007; 27: 1803-10.
6. Nemati MH, Astaneh B. Optimal management of familial hypercholesterolemia: Treatment and management strategies. Vasc Health Risk Manag, 2010; 6: 1079-88.