



SKIN MANIFESTATIONS OF INTERNAL DISEASE

## A RARE CASE OF XANTHOMATOSES WITH TYPE IIA FAMILIAL HYPERLIPIDEMIA

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**BACKGROUND:** Cutaneous Xanthomas, characterized by a yellow hue, are due to deposition of intracellular and dermal abnormal lipids. They do not represent a disease but rather are signs of different lipoprotein disorders. The major forms of xanthomas associated with hyperlipidemia are eruptive, tuberous, tendinous and plane xanthoma. To the best of our knowledge, we are unable to find previous reports of plane xanthomas co-existing with both tuberous and tendinous xanthoma with type IIA hypercholesterolemia.

**OBSERVATION:** A 26 year old non obese female presented with multiple, asymptomatic, soft to firm nodular lesions of size varying from 1X1 to 8X10 cm over buttocks, both elbows, knees, eyelids, Achilles tendons, interphalangeal and intertarsal joints since 20 years. Patient had a traumatic non healing ulceration over one of the lesions on buttock since three months. Her past, medical and drug history were unremarkable. No any other significant systemic complaints seen. No similar lesions in any of her family members were seen. Complete hemogram, biochemistry, sugar levels, uric acid levels, RA factor, thyroid profile, 2d echo, Fundus examination were normal. Fasting lipid profile showed increased total cholesterol- 506, and LDL -444 mg/dl, while triglycerides- 60mg/dl, HDL- 48, and VLDL- 14 were normal. Histopathology examination showed collections of foam cells and lipid-laden macrophages with cholesterol clefts suggestive of Xanthoma and showed no signs of malignancy from ulcerative lesion. A diagnosis of tuberous and tendinous xanthoma with xanthelasma palpebrarum due to Familial hypercholesterolemia was made. Excision was done for larger lesions over buttocks as they affected her daily activities. After nine months of atorvastatin and fenofibrate therapy patient is attaining normolipidemia, and reports generalized global reduction of size of xanthomas with no new lesions.

**KEY MESSAGE:** Early diagnosis, treatment, long term follow up helps in preventing the development of early coronary artery disease, atherosclerosis and pancreatitis.

