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GIANT PEDUNCULATED TUBEROUS XANTHOMAS- A CASE REPORT

Pathology

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ABSTRACT

Xanthomas are the dermatological manifestations of underlying lipid disorders. The underlying hyperlipidemias may be hereditary or secondary in nature. The familial causes have been well described by Fredrickson into five major categories. The secondary causes of hyperlipidemia range from uncontrolled diabetes mellitus to various dietary factors. We report a case of giant tuberous xanthomas due to secondary hyperlipidemias in an alcoholic patient.

KEYWORDS

xanthoma, pedunculated, hyperlipidemia

INTRODUCTION

Xanthomas are generally the manifestations of abnormal lipid metabolism. They are generally regarded as the dermatologic manifestations of dyslipidemias. Most of the xanthomas are associated with familial disorders in lipid metabolism and the rest are attributed to the secondary causes including diabetes mellitus, hypothyroidism, lymphoproliferative disorders and alcohol abuse or even without any lipid metabolisim abnormalities (1). We report a case of large pedunculated tuberous xanthomas with hyperlipidemia secondary to chronic alcohol abuse.

CASE REPORT

A 55 year old male presented to the outpatient department with multiple lumps over left knee leading to difficulty in walking. The lumps appeared over a period of time starting as small nodules and progressing to the current size over a period of four years. The lesions were progressive in nature and similar lesions had started appearing around waist also. The patient was a habitual smoker and chronic alcoholic since last 25 years. On examination it was found that the lesions were elevated pedunculated disc shaped with crusting on top and measured around 8 to 10 cm in diameter.(Fig 1) They were freely mobile and non tender. Similar lesions of smaller diameter

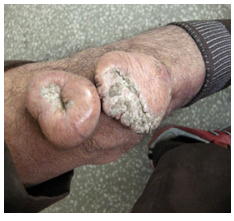

FIG.1: Giant pedunculated tuberous xanthomas over knees

were also found around the waist near the anterior suprior iliac spine and the iliac crest. All the routine examinations including complete blood count, ESR, urinalysis, blood sugar levels, kidney function tests, thyroid profile, chest X-ray, ECG and ultrasonography abdomen were within normal limits. The lipid profile showed raised levels of cholesterol, LDL, and fourfold increase in triglyceride levels. An excision biopsy of the lesion was done and the histopathology revealed large areas of foamy macrophages, admixed with cholesterol clefts and giant cells.(Fig 2) The giant cells were both of Touton as well as foreign body type. Focal areas of epidermal thinning and fibrosis were also noted. Based on these findings, diagnosis of tuberous xanthoma was made.

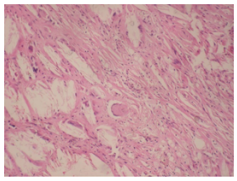

FIG. 2: (H&E-10x) showing large areas of foamy macrophages, cholesterol clefts and giant cells

DISCUSSION

Xanthomas are palpable masses that are typically located within the skin or subcutaneous tissue and consist of cholesterol, cholesterol esters, triglycerides, phospholipids, and numerous lipid-laden foamy macrophages (2). Most of the xanthomas appear due to some hereditary errors in lipid metabolism. Fredrickson in 1967 gave a classification of hyperlipidemias and categorized them from 1 to 5 depending upon the type of raised lipid fragments, ranging from familial hypercholesteronemia as type 1 to mixed hypertriglycer idemia as type 5 (3).

The xanthomas are considered to be the dermatological indication of underlying lipid disorders (4). The xanthomas associated with abnormal lipid metabolism are classified as tendon xanthomas , tuberous xanthomas, eruptive xanthomas, planar xanthomas, some cases of xanthalesma, normolipemic xanthomas, paraneoplastic xanthomas which includes necrobiotic xanthogranuloma and diffuse normolipemic xanthoma, verruciform xanthoma and some miscellaneous types of xanthoma (5). Most of these lesions present themselves as a manifestation of specific lipid abnormality but there is a considerable overlap among the presentations. Eruptive xanthomas are most commonly associated with chylomychronia and are the most common manifestations of secondary hyperlipidemias (6). Eruptive xanthomas consist of small, soft, yellow papules with a predilection for the buttocks and the posterior aspects of the thighs. They come and go with fluctuations in the chylomicron level in the plasma. Tendon xanthomas occur in patients with excessive plasma LDL levels, such as familial hypercholesterolemia and familial apolipoprotein B-100 defect, as well as in phytosterolemia and cholestanolemia (cere brotendinous xanthomatosis). The achilles tendons and the extensor tendons of the fingers are most frequently affected (5). The tuberous xanthomas are associated with hypertriglycerdeimia and cases of familial hypercholesterolemia type II. The associations of xanthomas with familial hypercholesteromia are well described (7). The most common causes of secondary hyperlipidemia are uncontrolled diabetes mellitus, hypothyroidism and dietary factors including chronic alcohol intake (8).

In our case the lipid profile was mildly deranged with mild elevation of total cholesterol, mild elevation of LDL cholesterol and around four time increase in triglyceride levels. These elevated triglyceride levels may be attributed due to alcohol intake over many years. The lesions started as small lumps and then progressed to become like the discoid, pedunculated lesions over a period of four years. The tuberous xanthomas have been generally been described as very small to large tumor like lesions around the knee and waist (9). Their association with raised cholesterol and triglyceride levels is well documented and reported (4,5). Our patient had giant pedunculated discoid lumps which had not been described earlier. This presentation of tuberous xanthomas must be kept in mind as they may be the harbinger of hidden lipid abnormalities.

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