FAMILIAL TYPE IIa HYPERLIPOPROTEINEMIA PRESENTING WITH TUBEROUS XANTHOMAS

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ABSTRACT

Xanthomas are localized lipid deposits within organs that may manifest as papules, plaques or nodules in the skin. Xanthomas indicate a more severe form of hyperlipidemia when observed during childhood or adolescence. We report a case of a 3 year old boy who presented to our outpatient department with multiple asymptomatic raised lesions gradually developing since the age of 1 year. Cutaneous examination revealed multiple skin coloured to yellowish smooth surfaced papules, nodules and plaques of variable sizes present bilaterally symmetrically over dorsum of hands and feet, tendo achillis, elbows, knees, posterior aspect of upper thighs and buttocks. Lipid profile revealed total serum cholesterol of 886 mg/dl, triglycerides 156.90 mg/dl, low density lipoproteins of 822.62 mg/dl, high density lipoproteins of 32 mg/dl, VLDL of 31.38 mg/dl. Punch biopsy showed features suggestive of xanthoma. The diagnosis was thus established as a case of tuberous xanthomatosis with Type IIa hyperlipidemia.

INTRODUCTION

Xanthomas are localized lipid deposits within organs that may manifest as papules, plaques or nodules in the skin. The clinical variants of cutaneous xanthomas include a wide array of lesions such as cutaneous xanthomas that can be idiopathic or may present as a sign of an inherited abnormality of lipoprotein metabolism (primary dyslipidemia), hyperlipidemia secondary to systemic disease or medication, or hematologic disease. The subtype of xanthoma provides a clue to the underlying abnormality. Accurate diagnosis of xanthomas is important because it can lead to the identification and treatment of underlying disease\(^\text{(1,2)}\). Xanthomas indicate a more severe form of hyperlipidemia when observed during childhood or adolescence\(^\text{(3)}\). We hereby report a rare case of a 3-year-old male presenting with tuberous xanthomas associated with familial type IIa hyperlipoproteinemia.

Case Report

A 3 year old non obese boy presented to our outpatient department with multiple asymptomatic raised lesions gradually developing since the age of 1 year. The patient was apparently normal 1 year back when his mother noticed a yellowish raised lesion over the posterior aspect of left upper thigh which progressively increased in size over time. The patient developed similar lesions over buttocks, hands, feet, elbows and knees. The lesions were asymptomatic. There was no history suggestive of any systemic involvement. There was history of similar lesions in the mother. There was no history suggestive of myocardial infarction or stroke at a younger age in any of the family members.

On clinical examination, the patient’s general physical examination findings were normal. Systemic examination was unremarkable. Cutaneous examination revealed multiple skin coloured to yellowish smooth surfaced papules, nodules and plaques of variable sizes present bilaterally symmetrically over dorsum of hands and feet, tendo achillis, elbows, knees, posterior aspect of upper thighs and buttocks (Fig 1a,1b,1c). The lesions were non tender and not fixed to the underlying structures. Skin over the lesions was normal.

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Figure 1b. Skin colored linear plaques present over the gluteal folds.

Figure 1c. Well defined raised plaques present bilaterally symmetrically over the tendon achillis.

Routine hematological investigations including complete blood counts, liver and renal function tests and blood sugar were within normal limits. Lipid profile revealed total serum cholesterol of 886 mg/dl, triglycerides 156.90 mg/dl, low density lipoproteins (LDL) of 822.62 mg/dl, high density lipoproteins of 32 mg/dl, VLDL of 31.38 mg/dl. A punch biopsy was taken from a papular lesion present over right buttock. Histopathological examination showed basket weave keratin covered epidermis with flattened rete ridges. Underlying dermis showed large aggregates of foam cells throughout the dermis with admixed lymphocytes (Fig 2a & 2b).

The diagnosis was thus established as a case of tuberous xanthomatosis with Type IIa hyperlipidemia. The patient was advised strict dietary control and started on tablet atorvastatin 20 mg daily at bed time along with chemical cauterization of smaller lesions.

DISCUSSION

The term xanthoma derives from the Greek word ‘xanthos’ meaning yellow and is used to describe a variety of subcutaneous lipid deposits, even those that do not appear particularly yellow. Xanthomas present as nodules or plaques which consist of macrophages loaded with cholesterol and cholesterol esters (‘foam cells’). They do not represent a disease but rather are symptoms of different lipoprotein disorders or arise without an underlying metabolic disease.

Cutaneous xanthomas most often present in adulthood. However, xanthomas associated with familial hypercholesterolemia often begin to develop prior to the age of 10 years. The pathogenic mechanism that leads to cutaneous xanthomas are not fully understood and may differ based upon the etiology and type of xanthoma. For xanthomas occurring in association with hyperlipidemia, it is hypothesized that when serum levels of lipoproteins are substantially elevated, extravasations of lipoproteins through dermal capillary blood vessels with subsequent engulfment by macrophages leads to lipid laden cells found in xanthomas.

The classification of hyperlipidemias is based on clinical assessment, level of plasma fasting lipids, electrophoretic pattern and hydrated density of plasma lipoproteins and is divided in to 6 types (Table 1). Type I and Type IIa are familial and usually present in childhood.

<table>
<thead>
<tr>
<th>Classification</th>
<th>Condition</th>
<th>Type of xanthoma</th>
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<tbody>
<tr>
<td>Type I</td>
<td>Familial chylomicronemia</td>
<td>Eruptive xanthomata</td>
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<tr>
<td>Type IIa</td>
<td>Familial hypercholesterolemia</td>
<td>Tendon, tuberous, plane xanthomas</td>
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<tr>
<td>Type IIb</td>
<td>Familial combined hypercholesterolemia</td>
<td>Usually absent</td>
</tr>
<tr>
<td>Type IIb</td>
<td>Familial dysbeta lipoproteinemia</td>
<td>Palmar, tuberous, tuberuptive xanthoma, xanthelasma</td>
</tr>
<tr>
<td>Type IV</td>
<td>Familial hypertriglyceridemia</td>
<td>Rare eruptive xanthomas</td>
</tr>
<tr>
<td>Type V</td>
<td>Mixed hyperlipidemia</td>
<td>Eruptive xanthomas</td>
</tr>
</tbody>
</table>

Figure 1. Frederickson classification of familial hyperlipidemias.
Clinically, xanthomas can be classified as eruptive, tuberoeruptive or tuberous, tendinous, or planar. Planar xanthomas include xanthelasma palpebrarum, xanthoma striatum palmare, and intertriginous xanthomas. Tuberous xanthomata are nodules that are frequently localized to the extensor surfaces of the elbows, knees, knuckles and buttocks.

The diagnosis of cutaneous xanthoma involves determining the type of xanthoma and the underlying cause through the patient history, physical examination and relevant laboratory findings. Accurate diagnosis of xanthomas is important because it can lead to identification and treatment of underlying disease.

CONCLUSION

Tuberous xanthomas are rare condition and often found to be associated with hyperlipoproteinemia. Hence proper identification and management of underlying disorder should be achieved.

References