

Normolipemic multiple skin xanthomas in a child

Xantomes cutanés multiples chez un enfant avec bilan lipidique normal

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ABSTRACT

Xanthomas are a common presentation of disorders of lipid metabolism. In some patients, they may only represent a cosmetic abnormality. We report here the atypical instance of a 12-year-old boy who developed multiple multi-lobulated skin tumours in elbows and knees without any complaints. Histopathology from a lesion showed characteristics of xanthoma (foamy histiocytes with Touton giant cells). No systemic involvement or lipid disorders were evident. This association is a very rare occurrence. Surgical excision of all tumours was done.

RÉSUMÉ

Les xanthomes cutanés se voient fréquemment dans les troubles du métabolisme lipidique. Parfois, ils se présentent chez des sujets normaux avec un statut lipidique normal.

Nous rapportons ici un cas rare de xanthomes cutanée multiples (les deux coudes et les deux genoux) dans leur forme tumorale chez un garçon de 12 ans sans antécédents pathologiques notables particulièrement. Le bilan lipidique était normal. Une exérèse chirurgicale des quatre localisations a été faite en même temps.

Xanthomatosis is a widespread disorder in which lipid depositing in the containing cells appear in the skin and visceral organs. They may represent a localized cutaneous phenomenon or signify a systemic hyperlipidemia with abnormal lipid metabolism [1, 2].

Xanthelasma palpebrum is the most common cutaneous xanthoma [2]. Tuberos xanthoma is rare in both children and adults. It is predominately located on the trunk and extremities. We describe in this paper a boy with normolipemic multiple tuberous cutaneous xanthomas. This case is being reported because of its rare occurrence.

I. CASE REPORT

A 12-year-old boy, born of a consanguineous marriage, presented to the paediatric orthopaedic department with a history of multiple asymptomatic skin tumors since age 2 years. These tumors were located on both elbows and knees and had increased progressively in number and size to become large. No family member had any such lesions. Physical examination revealed multiple yellowish skin tuberous and nodular xanthomas on both elbows and knees. All of these tumors were firm, painless, well delimited and of varying sizes from 2 to 6cm of long axis (Fig 1).



Figure 1: Multiple skin xanthomas on both elbows and knees varying in size

Except for these skin lesions the physical examination was normal. Biological investigations were done in a paediatrics department. Lipid profile was normal and serum protein electrophoresis showed a normal pattern. Biopsy of a tumour of the elbow was consistent with a xanthomatous lesion showing histiocytes with foamy vacuolated cytoplasm (Fig 2). Repeat lipid profile after several months was also normal. The child was operated and surgical excision was done for all xanthomas in the same time. At 3 years follow-up, there were no recurrences.

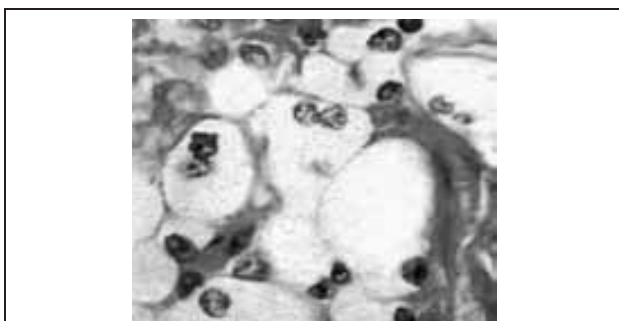


Figure 2: Histiocytes with foamy vacuolated cytoplasm

II. DISCUSSION

Xanthomas are tumours characterised by collections of foamy histiocytes (lipid-laden macrophages). They can be a reflection of altered lipid metabolism or a result of local cell dysfunction. Often secondary to hyperlipoproteinemias, especially hypercholesterolemia, xanthomas result in the accumulation of cholesterol in various tissues [1]. However, normolipemic patients had been rarely described with different types of xanthomas [3]. It has been suggested that 3 pathogenetic processes could be responsible for normolipemic xanthomatosis. The first group includes disorders with accumulation of unusual lipids other than cholesterol such as cholestanol or plant sterols. In the second group planar xanthomas may be seen in patients with lymphoproliferative diseases such as multiple myeloma or lymphomas. Xanthoma formation may be due to cutaneous lymphoreticular hyperplasia with secondary xanthomatization. The third group comprises patients in whom local abnormalities in the skin are thought to play a role. This includes xanthomas following distinct diseases such as erythroderma and epidermolysis bullosa dystrophica [3]. Based on their clinical appearance they are classified into five principal types: eruptive, tuberous, tendinous, planar and xanthelasma [3]. The latter is the most common of the xanthomas and presents as asymptomatic, usually bilaterally symmetric soft, velvety, yellow, flat, polygonal papules around the eyelids. Peripheral tuberous xanthomas are rare nodular, firm, painless subcutaneous swellings that develop in those areas which are subjected to repeated trauma such as elbows, knees, and buttocks. They may arise as single or multiple and vary in size from pea sized to lemon/mango sized. Usually, the lesions evolve for several months and enlarge slowly. Sometimes and like in our case, they may coalesce to form large tumorous swelling. Histologically, they are characterized by foamy cells and Touton giant cells [4]. Some normolipemic xanthomatosis are found to be associated with either a systemic disease or malignancy [5, 6, 7, 8]. Our case showed multiple tuberous xanthomas but without any lipid disorder, associated systemic disease, or malignancy. Although spontaneous involution had been reported in papular xanthomas [1], in our case the tuberous lesions were persistent for 10 years and the aim of surgery was essentially cosmetic and esthetical. There were not functional problems.

III. REFERENCES

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