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XANTHOMA WITH OCULAR AND CARDIOVASCULAR INVOLVEMENT IN A BOY

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ABSTRACT

In this report, a 10 year old boy with both tendinous and tuberous xanthoma is presented. Premature corneal arcus and aortic stensosis were associated findings in this case. Tuberous and tendinous xanthomas are two clinical varieties of one disease, both of which are seen in type II hyperlipoproteinemia. Increased serum cholesterol with pathological findings of skin lesions with oculocardiac involvement are all in favour of a homozygous condition. This is a rare disorder, and occurs in one per million of the general population, but a combination of two kinds of clinical skin lesions with cardiac involvement and corneal arcus is very rare. Therapy with cholestyramine 2 grams daily orally was prescribed. After six months of therapy, there was no improvement in his problem.

Keywords: Xanthoma, hyperlipoproteinemia

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INTRODUCTION

Xanthomas are localized infiltrates of lipid-containing cells usually found within the dermis or tendons, but occasionally in other tissues of the body.[3] This group of diseases is an uncommon presentation of disorders of lipid metabolism, and may be associated with an increased risk of arteriosclerotic vascular disease.[2] The homozygous condition is seen in only one per million of the general population.[11] The very rare homozygous condition usually presents in the first year of life.[10] This is a particular case of homozygous hypercholesterolemia whose skin lesions appeared in the second year of life.

CASE REPORT

A 10 year old boy with multiple soft deep skin tumors was referred to our dermatology clinic. Some tumors were only subcutaneous and some were adjacent to extensor tendons, especially near the distal joints such as the achilles and interphalangeal tendons (Fig. 1). Others were superfi-

Fig. 1. Tuberous and tendinous xanthomata, two clinical variations of xanthoma seen in this case.

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Fig. 2. Bilateral corneal arcus (senile arci) is seen in a 10 year old boy.

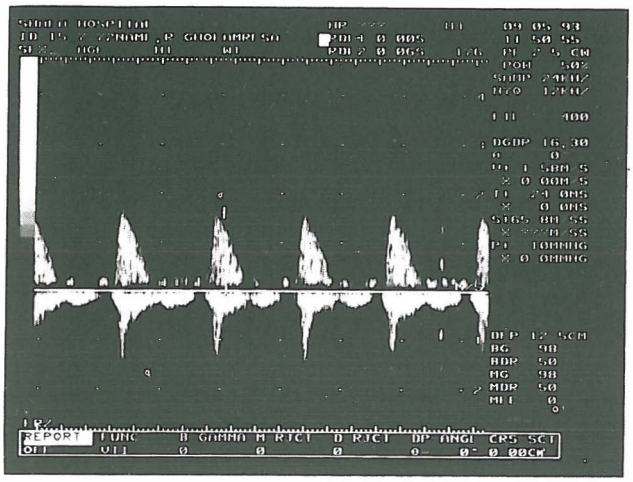

Fig. 3. Doppler echo study of aortic valve flow.

Fig. 4. Tendinous xanthomas in feet.

Fig. 5. Tuberous xanthomas.

cial, hard, round nodules with a yellowish flat sharp border adjacent to the epidermis, without pain or other symptoms. Development of skin lesions began when he was only two years old. At birth he had no skin lesions or other organ disorders. Regarding familial history, his parents are cousins, with no history of skin lesions, chest pain or myocardial infarction. His two brothers and five sisters all are healthy on clinical examination. Ophthalmic examination of our patient showed symmetric bilateral yellowish corneal arcus that was seen bilaterally at the superior margins of the ophthalmic corneum (Fig. 2). A mild anterior right uveitis was seen but fundus examination including discus and retinal vasculature was with no abnormalities. On cardiovascular examination, peripheral pulses were full, symmetric with no thrill or heave. Blood pressure and heart rate were within normal range. There were normal heart sounds with a systolic ejection type (Grade III/VI) murmur at the aortic area with radiation to the carotid arteries. Echocardiographic findings revealed no abnormality in function or ventricular size, and septal and posterior wall thickness was also normal. Doppler echocardiogram revealed about 10 mmHg transaortic valve gradient (Figs. 3,4). Mitral valve flow was within normal range. Laboratory tests showed hypercholesterolemia of 540 mg/100 mL last year and 680 mg/100mL presently, serum triglyceride level was 248 mg/mL last year and recently was 230 mg/mL. C-reactive protein was one-plus positive, and blood urine, stool analysis, liver function tests and metabolic examinations were all normal. Pathological study of an early tendinous lesion showed many abnormal intracellular sudanophilic materials that were in accordance with the clinical diagnosis, and excisional biopsy of the tuberous skin lesions was also in support of the diagnosis (Figs. 5,6).

DISCUSSION

Xanthomatosis is a cutaneous manifestation of lipidosis in which plasma lipoproteins and free fatty acids are changed quantitatively.[1] Although tendinous xanthomas are uncommon among new dermatological patients, they usually are seen in xanthomatous conditions, but overlap of tuberous and tendinous lesions with premature corneal arcus is rare. Homozygous type xanthomas are seen in about one per one

Fig. 6. Histologic view of xanthoma (Giemsa stain, × 40).

million general population.[9] Beginning of skin lesions in our case was when he was 2 years old, although juvenile xanthogranuloma has been reported in a four month old infant.[7] Aortic stenosis is also a cardiovascular complication of constant hyperlipidemia in this case at 10 years old.[6] Homozygous xanthoma is usually seen before 20 years of age, and there is a report of a case, who was 18 months old with premature cardiac infarction in a homozygous patient.[4] If the physician is to prevent the disastrous vascular consequence of type II disease, it is obvious that the disorder must be detected early and the appropriate therapy should be initiated.[5] Although he does not complain of any myocardial dysfunction, but a systolic ejection is heard in the aortic area by auscultation. Corneal arcus is seen mostly in patients with serum cholesterol levels more than 350 mg/mL.[6] The cause of mild anterior right uveitis and its relationship with hypercholesterolemia is unknown.

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