XANTHOMA WITH OCULAR AND CARDIOVASCULAR INVOLVEMENT IN A BOY

S. SHAMSADINI, M.D., M. MOAZENZADEH,* M.D., S. DABIRI,** M.D., AND M. MESHKAT,*** M.D.

From the Department of Dermatology, *Cardiology, **Pathology and ***Ophthalmology, Kerman Darman Hospital, Kerman, Islamic Republic of Iran.

ABSTRACT

In this report, a 10 year old boy with both tendinous and tuberous xanthoma is presented. Premature corneal arcus and aortic stenosis 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


INTRODUCTION

Xanthomas are localized infiltrates of lipid-containing cells usually found within the dermis or tendons, but occasionally in other tissues of the body. 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. The homozygous condition is seen in only one per million of the general population. The very rare homozygous condition usually presents in the first year of life. 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-
Xanthomatosis is a cutaneous manifestation of lipidosis in which plasma lipoproteins and free fatty acids are changed quantitatively. 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...
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. Aortic stenosis is also a cardiovascular complication of constant hyperlipidemia in this case at 10 years old. 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. 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. 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. The cause of mild anterior right uveitis and its relationship with hypercholesterolemia is unknown.

REFERENCES
