Pediatric Tendinous Xanthomas

An 8-year-old non-obese girl, born of non-consanguineous marriage, presented with skin-colored papules and nodules on the hands for two years. The lesions were firm, non-tender and were localized to the extensor aspect of interphalangeal and metacarpophalangeal joints (Fig. 1). There was no joint pain or restriction of mobility. Complete blood count, serum uric acid, renal, liver and thyroid function tests were normal. The patient had elevated total cholesterol (630 mg/dL) and low density lipoprotein cholesterol levels (528 mg/dL). The corresponding levels in her father were 324 mg/dL and 210 mg/dL, respectively. Triglyceride and high density lipoprotein levels were normal in both the patient and her father. Skin biopsy showed diffuse dense nodular infiltrate of foamy histiocytes in the dermis consistent with the diagnosis of xanthoma. Genetic mutation analysis for familial hypercholesterolemia could not be done.

Tendinous xanthomas are caused by altered lipid metabolism, which results in cholesterol deposits in ligaments, tendons or periosteum. These are frequently seen in familial hypercholesterolemia due to mutation in low density lipoprotein receptor, apolipoprotein B or proprotein convertase subtilisin/kexin type 9. Tendinous xanthomas localized to the hands should be differentiated from rheumatoid nodules, multicentric reticulate histiocytosis, knuckle pads, pachydermodactyly, tophaeous gout and subcutaneous granuloma annulare. Early diagnosis and treatment of hyperlipidemia by dietary modification and lipid lowering drugs can reduce the risk of cardiovascular morbidity.

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