

Cutaneous Xanthomas in a Young Child: Familial Hypercholesterolemia

Sir,

Xanthomas are non-neoplastic lesions characterized by yellowish papules, plaques, or nodules due to accumulation of fat-laden macrophages in the dermis and subcutis.^[1]

A 6-year-old, non-obese girl presented to the paediatrics outpatient department with a history of multiple, yellowish plaques, papules, and nodules over the body for past 1 year with a gradual increase in size. These lesions involved the bilateral elbow, knee, knuckle, lateral malleolus, gluteal region, and left upper eyelid and were associated with itching [Figure 1a-d]. The nodules over the knuckles were cystic in consistency. There was absence of similar lesions and cardiovascular disease in other family members.

Complete blood count, liver, renal, and thyroid function tests were normal. Serum lipid profile showed markedly elevated levels of total cholesterol and low density lipoprotein cholesterol level (LDL-C) viz. 803 mg/dl and 732 mg/dl, respectively. However, high density lipoprotein cholesterol (HDL), very low density lipoprotein cholesterol (VLDL), and triglycerides levels were normal. Chest X-ray, electrocardiogram (ECG), two-dimensional echocardiography, and fundus examination of both eyes were normal.

Fine-needle aspiration cytology (FNAC) from nodular cystic swellings over the interphalangeal joint (knuckle) revealed predominantly foamy macrophages in a hemorrhagic background [Figure 2a]. Skin biopsy from the elbow lesion showed pan dermal infiltration by foamy macrophages [Figure 2b].

Lipid profile of her father revealed elevated serum total cholesterol (385 mg/dL) and LDL-C (271 mg/dL) levels, however, lacked clinical manifestations and other

associated systemic disease. Her mother refused to visit the hospital.

Other tests like computed tomography (CT) angiography, LDL receptor activity and genetic mutational analysis were not done due to financial constraint.

A diagnosis of homozygous familial hypercholesterolemia (HoFH) was made and medical treatment was started with statins and resins. She was recommended low fat and high fibre diet. However, follow-up after 6 months revealed no improvement in the clinical lesions and serum total cholesterol levels.

Xanthomas occur due to alteration in the lipid metabolism. Sometimes it can be the only presentation of a serious underlying lipid abnormality. In children, they are mostly associated with abnormalities of cholesterol metabolism with hypercholesterolemia and high LDL-C (Fredrickson, type II).^[2]

Familial hypercholesterolemia (FH) is an autosomal dominant genetic disorder caused by mutation in the gene encoding the receptor for LDL located on chromosome 19.^[3] FH most commonly presents as heterozygous form, becomes symptomatic in the 3rd and 6th decades, and responds well to treatment. Whereas homozygous state is rare and presents in early childhood with poor response to therapy.^[3] Early presentation in HoFH is due to persistently raised serum total cholesterol and LDL-C (>500 mg/dL) levels since birth. Children with HoFH are at risk of early onset atherosclerosis, aortic valve disease, and premature coronary death.^[4] The diagnosis is usually made by biochemical and clinical evaluation. However, LDL receptor activity and genetic mutational analysis can also be done for further confirmation.

Genetic counselling of the family is of utmost important. Therapeutic measures include diet modification and drugs



Figure 1: Cutaneous xanthomas on (a) interphalangeal joints, (b) knee joint, (c) eyelid, and (d) gluteal region

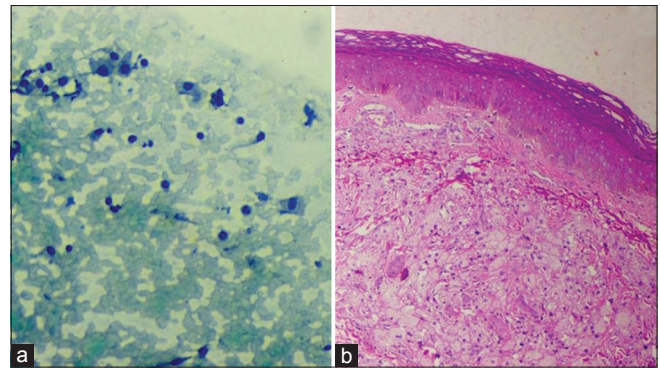


Figure 2: (a) Fine needle aspiration cytology shows many foamy macrophages (Pap, $\times 200$) (b) Skin biopsy shows pan dermal infiltration by foamy macrophages (H and E, $\times 100$)

such as statins used alone or in combination with bile acid sequestrant, fibrates, and Ezetimibe.^[5] LDL apheresis and orthotopic liver transplantation are other available alternatives.^[4]

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Conflicts of interest

There are no conflicts of interest.

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