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IMAGE ARTICLE

Giant Infantile Xanthomas Revealing Familial Hypercholesterolemia

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Introduction

Xanthomas are benign pseudotumours, most often related to a disorder of lipoprotein metabolism, which require screening. Early diagnosis determines the prognosis and enables the prevention, detection and treatment of premature cardiovascular complications.

Observation

An 8-year-old female child with a history of first degree family consanguinity presented with yellowish-brown, painless, papular lesions on the elbows (Figure 1), knees (Figure 2) and hands (Figure 3) that were progressively increasing in size for about 5 years. A skin biopsy was performed showing infiltration by xanthomatous foamy histiocytic cells and a few multinucleated "Touton" type cells confirming the diagnosis of xanthoma. The biological assessment revealed very high levels of: total cholesterol (9.87 g/l), LDL cholesterol (8.76 g/l) and Apolipoprotein B (6.66 g/l). Apolipoprotein A1 levels were low (0.62 g/l). A work-up including an electrocardiogram, cardiac ultrasound and glycated haemoglobin was performed and was unremarkable. Based on the clinical and biological data, the diagnosis of familial hypercholesterolemia

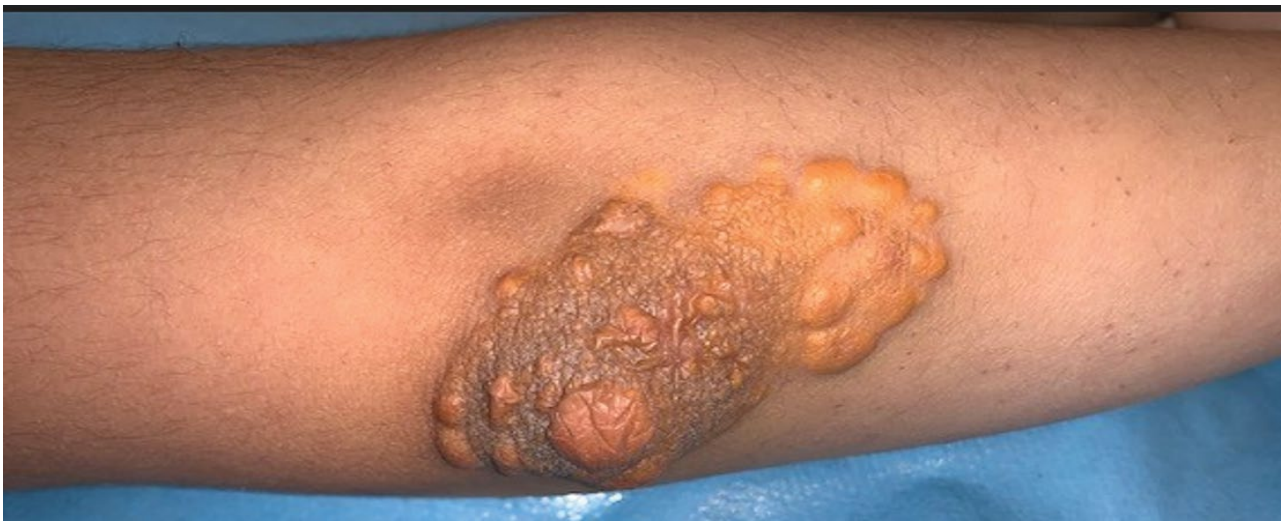

Figure 1: Papular lesions of yellowish brown color on the elbow.

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was made. Medical treatment as well as hygienic and dietary measures were proposed to this child with close monitoring.

Conclusion

Xanthomas are the main skin manifestation of dyslipidaemia. Morbidity and mortality are related to atherosclerosis. Early diagnosis determines the prognosis.

Sources of Support

Nil.

Author Contribution

All authors' contributed equally.

Consent

Written consent has been obtained from the patient.

Conflict of Interest

No conflict of interest.

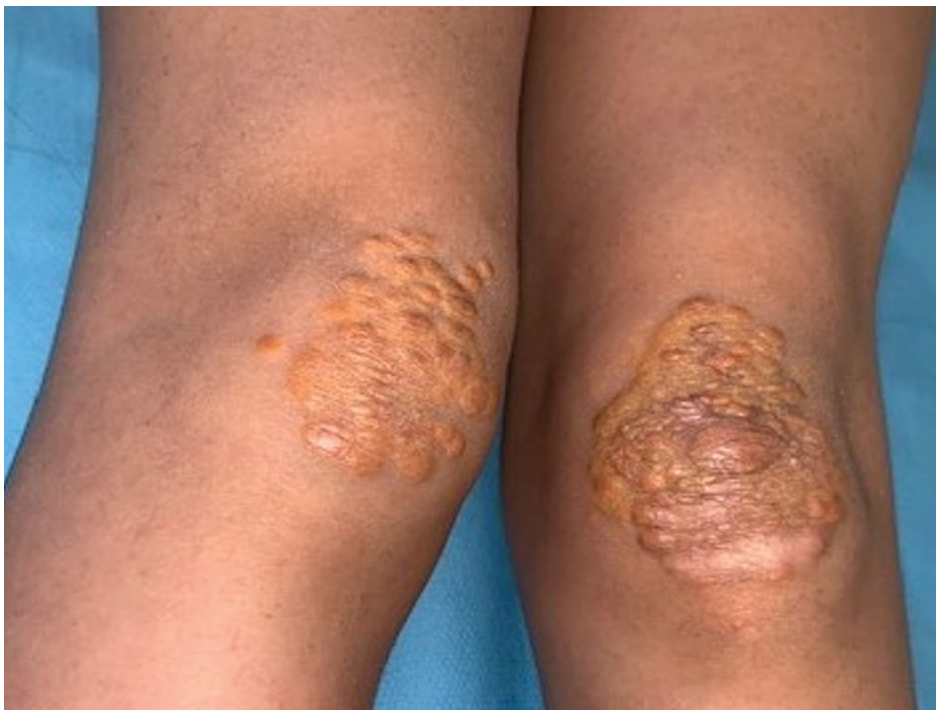

Figure 2: Xanthomas on both knees.

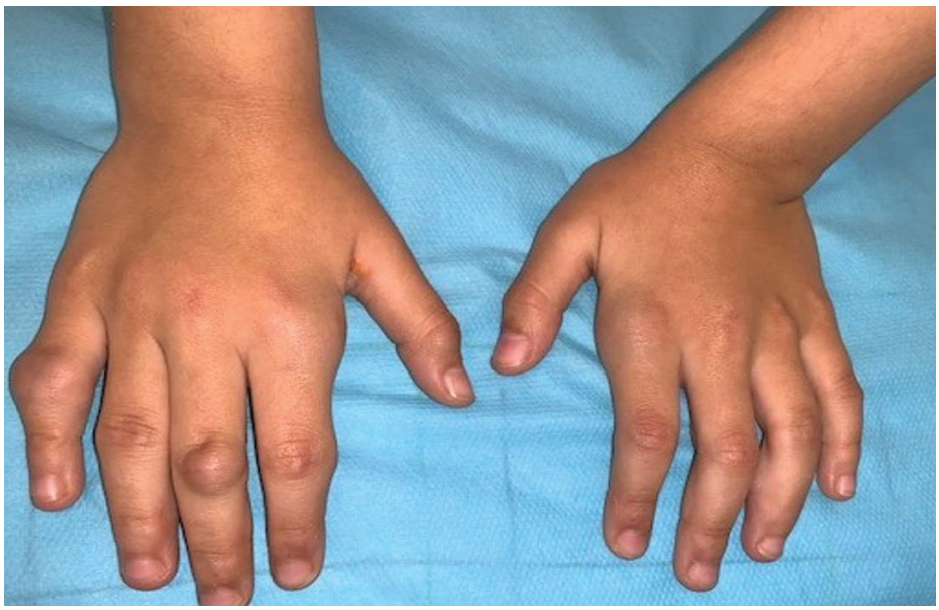

Figure 3: Xanthoma on the middle finger of the right hand.