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.
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.

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**Figure 2:** Xanthomas on both knees.

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