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IMAGES IN PAEDIATRICS.....

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Tumorous xanthomas in a young male with familial hypercholesterolaemia

A 16 year old boy presented with massive bilateral tumorous swellings over both buttocks (fig 1). He had a strong family history of premature coronary artery disease, with one of his first degree relative dying at the age of 18 years. On examination, his weight was 52 kg, height 168 cm, body mass index (BMI) 18.43 kg/m², and blood pressure 110/76 mmHg. He had evidence of bilateral xanthelasmas, premature arcus cornea, tuberous xanthomas over both buttocks and elbow joints, and tendon xanthomas over the Achilles tendon and extensor tendons of the hands. The systemic examination was unremarkable. Lipid profile revealed total cholesterol 460 mg/dl, triglyceride 192 mg/dl, high density lipoprotein cholesterol (HDL-C) 30 mg/dl, and low density lipoprotein cholesterol (LDL-C) 368 mg/dl. Fasting plasma glucose and serum insulin levels were 87 mg/dl and 2.4 µU/ml respectively with calculated HOMA-IR of 0.51. The lipid profile of his father revealed total cholesterol of 267 mg/dl, triglyceride 229 mg/dl, HDL-C 38 mg/dl, and LDL-C 183 mg/dl. The lipid profile of his mother showed total cholesterol of 254 mg/dl, triglyceride 148 mg/dl, high density lipoprotein 35 mg/dl, and low density lipoprotein 191 mg/dl. Fine needle aspiration from the gluteal swelling was consistent with a xanthomatous lesion. He received atorvastatin 80 mg and ezetimibe 10 mg; six months later he showed modest decrease in size of the xanthomas.

Xanthomas are important clinical manifestations of disordered lipid metabolism. Tuberous xanthomas are nodular, firm, painless subcutaneous swellings that develop in those areas which are subjected to repeated trauma such as elbows, knees, and buttocks. They may arise as single or multiple and vary in size from pea sized to lemon/mango sized. Sometimes they may coalesce to form large tumorous swelling as shown in this case. Tuberous xanthomas are usually associated with raised low density lipoprotein levels. If untreated, hypercholesterolaemia is associated with premature atherosclerotic disease and cardiovascular mortality.

Figure 1 Huge xanthomas over both buttocks (consent was obtained for publication of this figure).

However, there is a poor co-relationship between circulating LDL-C levels and corresponding size of the xanthomas. Even with successful treatment and normalisation of LDL-C levels, these tumorous swellings may not regress completely as was seen in our patient. Early and sustained aggressive treatment is required for prevention of future cardiovascular morbidity and mortality.

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