GENETICS AND GENODERMATOSES

A CASE REPORT OF FAMILIAL HYPERCHOLESTEROLEMIA TYPE 2A

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Introduction: Familial hypercholesterolemia (FH) is an autosomal dominant condition characterized by high total and LDL cholesterol, slightly low HDL cholesterol and normal triglyceride concentrations and clinically by frequent tendon & tuberous xanthomata with premature onset of cardiovascular disease. The incidence of homozygous FH (HoFH) is very low (1 in million people). However, heterozygous FH occurs in 1of 500 people, and is frequently detected by routine medical health check-up.

Case: A 12 year old male child presented with the chief complaints of asymtomatic multiple yellowish lesions over various parts of body for about three years. Fresh lesions started appearing previously on knees and then elbows and buttocks. There was no family history of consanguinity, dyslipidemia and acute myocardial infarction. Dermatological examination of the child showed: multiple tuberous xanthomas of the knees, elbows and the gluteal region.Laboratory investigations; Plasma lipid levels of the child were: total cholesterol (TC); 712mg/dl, low density lipoproteins (LDL); 576mg/dl, triglycerides (TG); 127mg/dl and high density lipoprotein (HDL); 28 mg/dl. Urea, creatinine, liver function tests and thyroid function tests were within normal limits. Histopathology of the biopsy specimens confirmed the diagnosis of xanthoma. Lipid profile of the other family members are normal limits. With the available laboratory data and clinical findings, it was established that the patient had Homozygous FH. He was advised ophthalmology and cardiology consultation, low cholesterol diet, treated with Tab Atorvastatin 20 mg.

Discussion: Xanthomas are benign tumours characterized by skin coloured or yellowish plaques and nodules. Tuberous xanthomas are commonly found over the extensor aspect of elbows and knees. In our patient, the lipid profile was suggestive of Type IIa hyperlipidaemia, with markedly elevated total and LDL cholesterol levels and normal triglyceride levels.The diagnosis of familial hypercholesterolaemia is essentially based on clinical assessment and biochemical parameters.