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Cutaneous Xanthomas in Familial Hypercholesterolemia

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A 9-year-old girl presented with asymptomatic skin lesions for the past 5 years. Similar lesions were reportedly present in her father, who died at the age of 40 years from coronary artery disease. Examination revealed tuberous xanthomas of varying sizes over the knees [Figure 1a] and buttocks [Figure 1b]. Tendinous xanthomas over the Achilles tendon and xanthelasma palpebrarum [Figure 1c and d] were also noted. On ophthalmic examination, corneal arcus was not found. Patient's mother denied consent for biopsy. Her lipid profile was as follows: Total serum cholesterol: 616 mg/dL, low-density lipoprotein: 552 mg/dL, triglycerides: 88 mg/dL and high-density lipoprotein: 46 mg/dL. Diagnosis of familial hypercholesterolemia (FH) type II was made as the above presentation fulfilled the Dutch Lipid Clinic Network criteria. Molecular genetic testing was not available. Her electrocardiogram and echocardiogram were normal. The patient was advised lifestyle modification and was started on tablet atorvastatin 5 mg daily. Patient's mother was advised to bring her younger son to our OPD for screening for familial hypercholesterolemia.



Figure 1: Tuberous xanthomas over the knees (a) and buttocks (b). Tendinous xanthomas were found over the Achilles tendon (c shown by black arrowheads). Xanthelasma palpebrarum (d).

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Varied types of xanthomas can be seen in FH, among which tendinous xanthomas are highly specific for FH. Prompt identification of these lesions can result in early diagnosis and treatment initiation, thereby preventing future complications like cardiovascular diseases.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

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