

Images/Instrument in Dermatology/Dermatosurgery

Familial hypercholesterolemia

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A 4-year-old female presented with multiple yellowish papules and plaques over knees, buttocks, legs, and gluteal cleft for 1 year, clinically diagnosed as xanthomas. Her mother had yellowish plaques in the bilateral periorbital region for 3 years clinically diagnosed as xanthelasma palpebrarum [Figure 1a].

Lipid profile of daughter revealed raised cholesterol (804 mg/dL) and low-density lipoproteins (859.4 mg/dL). Lipid profile of mother showed raised cholesterol (329 mg/dL) and low-density lipoproteins (235.6 mg/dL). Histopathology from child's knee showed multiple histiocytes, and lymphoplasmacytic infiltrate involving dermis and subcutis, consistent with xanthoma [Figure 1b and c]. Based on lesions, lipid profile, and histopathology, diagnosis of Type 2a dyslipidemia (familial hypercholesterolemia) was confirmed.^[1,2]

Cutaneous manifestations reported with familial hypercholesterolemia include tendinous xanthoma (40–50%), xanthelasma (23%), and tuberous xanthomas (10–15%). Less commonly, subperiosteal xanthoma (below knee and over olecranon) may be seen. Rarely, intertriginous xanthomas in the finger webs, axillae, buttocks, antecubital, and popliteal fossa may also be seen, which are pathognomonic of familial hypercholesterolemia.^[3]

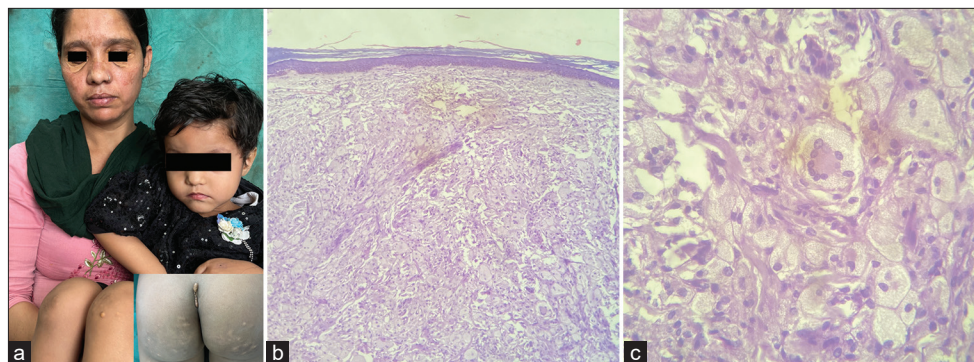


Figure 1: Clinical image. (a) Daughter shows multiple yellow papules over the bilateral knees, legs, and gluteal cleft (Inset); and mother shows periorbital yellowish flat plaques. Histopathology showing (b) thin epidermis, flattened rete ridges, and lymphoplasmacytic infiltrate in dermis and subcutis (H and E × 100). (c) Lymphoplasmacytic infiltrate and Touton giant cells (H and E × 400).

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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.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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