Images in Clinical Practice

Pediatric tuberous xanthomas

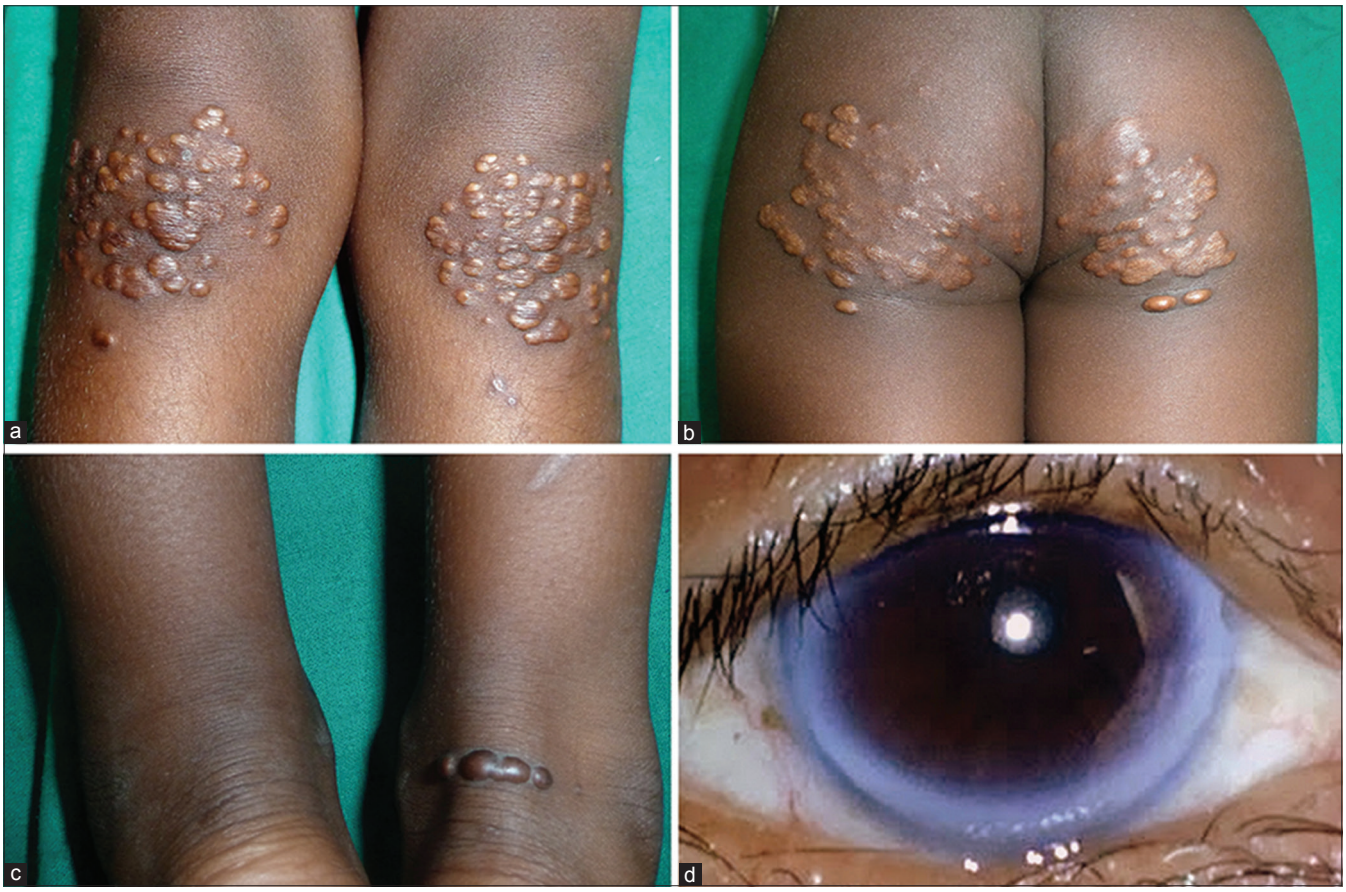

Figure 1: Yellowish-brown, clustered, smooth surfaced, nodules over (a) knees, (b) buttocks, and (c) right ankle (d) arcus juvenilis

A 7-year-old boy, developed, asymptomatic, gradually progressive smooth, mobile, yellowish, soft, and elevated nodules over knees, buttocks, and right ankle since the last three years [Figure 1a-c]. Slit lamp examination revealed arcus juvenilis [Figure 1d]; lipid profile revealed markedly elevated low density lipoproteins indicating severe primary type II homozygous familial hypercholestrolemia. These findings exclude other differential diagnoses of pediatric xanthomas such as phytosterolemia, cerebrotendinous xanthomatosis, and Alagille syndrome.

Due to high risk of development of early coronary artery disease despite statin therapy, strict adherence to lifestyle modifications is warranted. Family members should be screened and appropriately treated. Specialist management includes ezetimibe-induced diminished cholesterol absorption and its "emergency" removal by plasmapheresis.

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