Touraine–Solente–Gole syndrome: The elephant skin disease

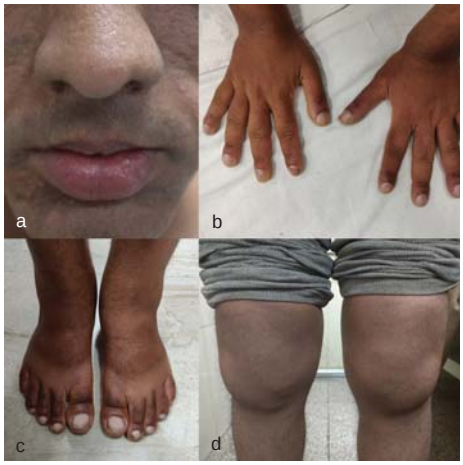

FIG 1. Changes in the skin and joints

FIG 2. Changes in the eyes

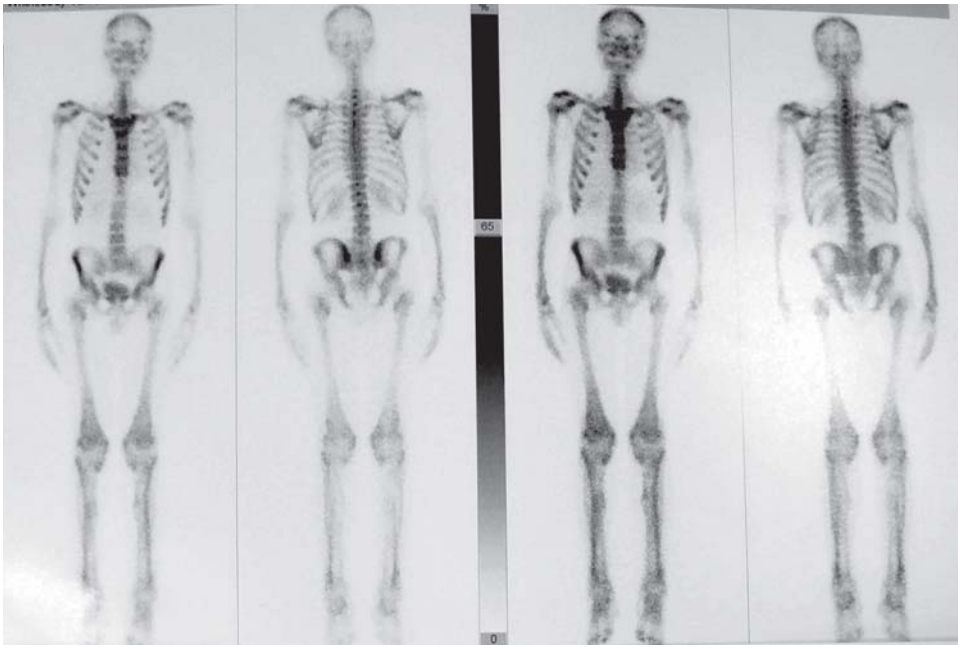

FIG 3. Technetium-99m bone scan showing symmetrical diffuse uptake in the long bones

Pachydermoperiostosis (Touraine–Solente–Gole syndrome) is a rare genetic disease, with multiple systemic manifestations involving hypertrophic skin changes, digital clubbing and periostosis.[1] A 23-year-old man presented with a history of thickening and drooping of his eyelids for the past 3 years. He complained of repeated swelling of his hip, knee and wrist joints. On examination, he had thickened, wrinkled facial skin with furrowing. There were deepened nasolabial folds and seborrheic hyperplasia (Fig. 1a). There was evidence of clubbing in the fingers (Fig. 1b) and toes (Fig. 1c) along with hyperhidrosis of the palm and sole. The knee joints also appeared swollen (Fig. 1d). There was diffuse thickening of the tarsus and lid margin with two small chalazions near the right lower lid margin with moderate ptosis in both eyes (Fig. 2a). Congestion and palpebral conjunctival hyperplasia were noted bilaterally (Fig. 2b). Technetium-99m bone scan showed symmetrical diffusely increased uptake in long bones of the appendicular skeleton and the periarticular region (Fig. 3). A diagnosis of pachydermoperiostosis was made. He was managed with intralesional injection of triamcinolone acetonide for chalazion, and topical antihistamine and lubricants for ocular irritation.

The disease has an estimated prevalence of 0.16% with onset in adolescence.[2,3] The man:woman ratio is 7:1, with men being severely affected.[4] It has autosomal dominant inheritance.[5] Three clinical forms are described: complete (periostosis and pachydermia), incomplete (periostosis without pachydermia), and forme fruste (pachydermia with minimal periosteal

change). While the typical course of the disease is self-limiting, non-steroidal anti-inflammatory drugs may be given to manage pain.[6] To control the cutaneous manifestations, steroids, isotretinoin[7] and botulinum toxin[8] have been tried.

Conflicts of interest. None declared

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