Diffuse Palmoplantar Keratoderma, Onychodystrophy, universal Hypotrichosis and Cysts

Dear Editor,

Clouston syndrome, also called hidrotic ectodermal dysplasia (HED), is an autosomal dominant ectodermal dysplasia characterized by a clinical triad of onychodystrophy, generalized hypotrichosis, and palmoplantar keratoderma (1). Herein we report the case of a 24-year-old male with the distinctive clinical triad associated with multiple epidermoid cysts, which probably reflects the phenotype of Clouston syndrome.

A 24-year-old male presented to our Department with diffuse thickening of the skin of his palms and soles since infancy. He also complained of sparsity to near absence of body hair and also reported thickening of the nails and multiple swellings involving the genitals and head since childhood. There was no history of consanguinity or of recurrent painful paronychia or abnormality in sweating. The patient denied any history of deafness, diminution of vision, redness, or watering of the eyes.

On examination, diffuse hyperkeratosis of the palms and soles was observed (Figure 1 a, b) However, there was no extension of this hyperkeratosis to the dorsal aspects of the hands and feet or any proximal extension to the forearms or legs. Extensor aspects of the elbows and knees did not reveal any hyperkeratotic skin lesions. The nails were yellowish-brown, thickened, and hyperconvex, which was more pronounced in the finger nails than the toe nails (Figure 1 c, d). There was no associated paronychia.

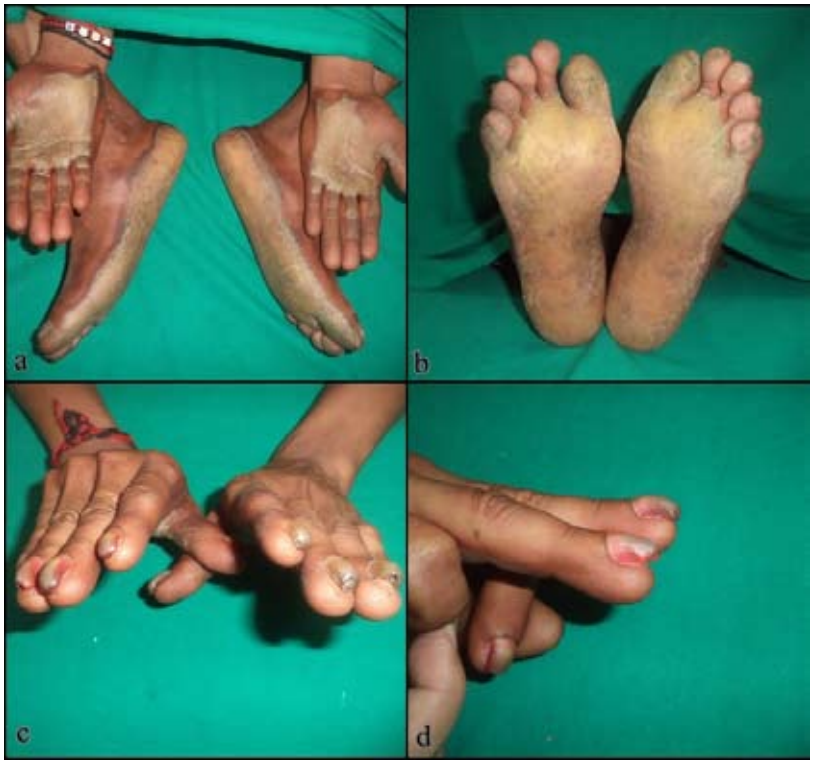

Figure 1. Diffuse palmoplantar keratoderma (a, b). The finger nails are thickened, yellowish-brown and hyperconvex (c, d).

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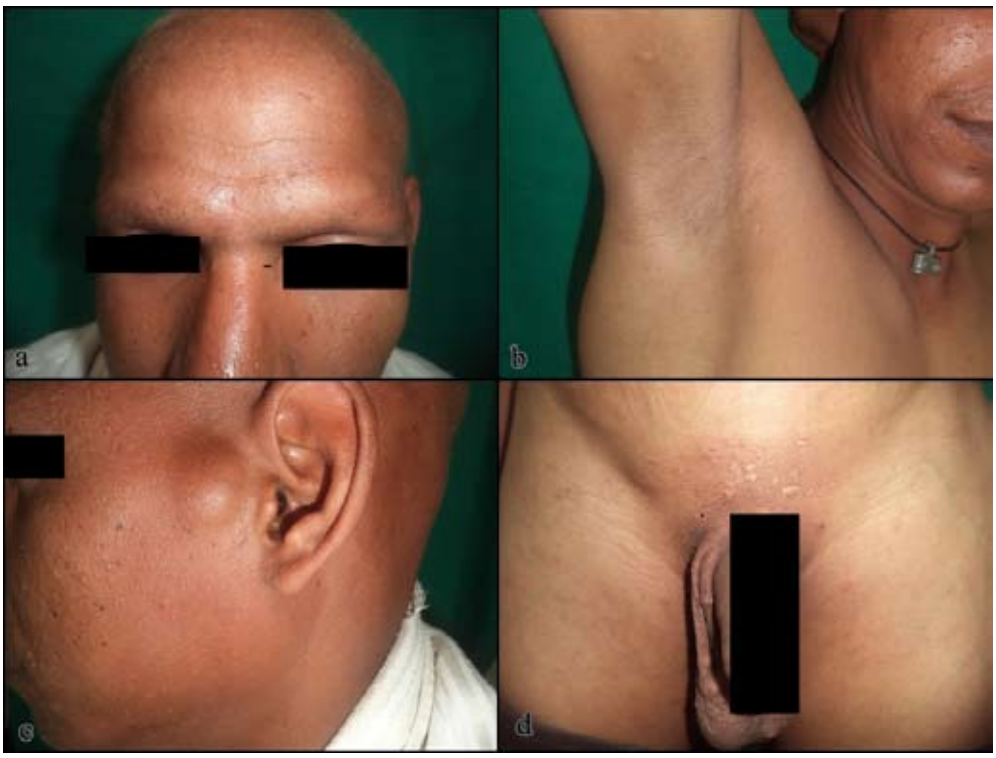

Figure 2. There is sparse to nearly absent hair on the eyebrows, scalp, axilla, moustaches, and beard (a, b, c). Swellings over the face and scrotum (c, d).

The scalp hair was very sparse, fine, and pale in color, reaching just a length of 3-4 mm in some places while totally absent in other places. The hair from the beard, eyebrows, eyelashes, moustaches, and pubic and axillary regions was very sparse to nearly absent (Figure 2 a, b, c). General body hair was also absent. In the left pre-auricular area there was a 3×2.5 cm swelling, soft to firm in consistency, non-tender, and non-pulsatile with no sinus or scar over it (Figure 2c). Multiple similar swellings of variable size measuring 0.6 to 1.3 cm were present over the scrotum (Figure 2 d). Systemic examination including oro-dental and ophthalmological examination was unremarkable. Physical tests for hearing were normal. Nail clippings for KOH examination did not reveal any fungal components. Fine needle aspiration from the pre-auricular swelling was consistent with epidermoid cyst. The classical triad of onychodystrophy, universal hypotrichosis, and palmoplantar hyperkeratosis with normal sweating and teeth indicated a diagnosis of Clouston syndrome.

Hidrotic ectodermal dysplasia was first described in a French-Canadian kindred (2). However, it has subsequently been described in other ethnic and geographical areas. There is a mutation in the β gap junction protein gene which codes for the protein connexin 30 (Cx30) (3). This condition primarily affects the hair, nails, and skin, while sparing the teeth and sweat glands. The hair is sparse and pale, and the alopecia can be patchy or total. Hair loss may lead to total alopecia by puberty. The eyelashes are short and sparse, and the eyebrows as well as axillary and pubic hair are also sparse or absent (1), as in our case. During infancy, the nails are typically milky white, gradually thickening throughout childhood. The nail plate is short, thick, slow-growing, and discolored, which was consistent with our patient's nail changes. Diffuse palmoplantar hyperkeratosis is a characteristic sign which may extend to the dorsum of the hands and feet (4). However, our case had no transgradient component.

There are other less common abnormalities reported in Clouston syndrome, which include conjunctivitis, strabismus, congenital cataract, oral leucoplakia, diffuse eccrine poromatosis, sensorineural hearing loss, thickened skull bones, and tufting of the terminal phalanges (2,5-8). However, to the best of our knowledge, the presence of epidermoid cysts in Clouston syndrome has not been previously reported, making our case a unique clinical presentation. Pachyonychia congenita is a very close differential diagnosis for this entity. However, universal hypotrichosis and the lack of oral leukokeratosis were the differentiating features in our case. Additionally,

palmoplantar keratoderma in pachyonychia congenita is mainly focal rather than diffuse, as in our case. However, genetic studies are needed to establish such a diagnosis.

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