

Peeling skin syndrome

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DESCRIPTION

A 30-year-old man born of a non-consanguineous marriage presented with asymptomatic peeling of skin throughout his body, sparing palms and soles, since birth. There was no preceding history of redness or fluid-filled lesions over the affected area. The duration of skin shedding varied between winters and summers, occurring, on average, for 7 days in winter and 5 days in summer. Rubbing of the skin during bathing would initiate the process. No similar symptoms were present in his other family members. During examination, minor rubbing on the volar aspect of his forearm produced peeling of skin (figure 1). Histopathological examination showed sparse superficial perivascular lymphocytic infiltrates with hypergranulosis and lamellated orthohyperkeratosis in the epidermis. An incomplete cleft was seen above the granular layer (figure 2). Generalised peeling skin syndrome is caused by autosomal recessive ichthyosiform genodermatoses, which is characterised by periodic or continuous peeling of superficial layers of skin.¹ The underlying defect is a mutation in gene encoding for corneodesmosin, which plays an important role in intercellular adhesion between corneocytes in the cornified cell layer. There are two types of peeling skin syndrome: generalised and acral. The generalised form is further divided into two variants: type A (non-inflammatory and asymptomatic, characterised by generalised peeling) and type B (presenting with congenital ichthyotic erythroderma).² The acral variant is confined to the peeling of the palms and soles, and is associated with homozygous mutation in transglutaminase 5 (TGM5).² TGM5 is expressed in the epidermal granular layer, where it cross-links a variety of

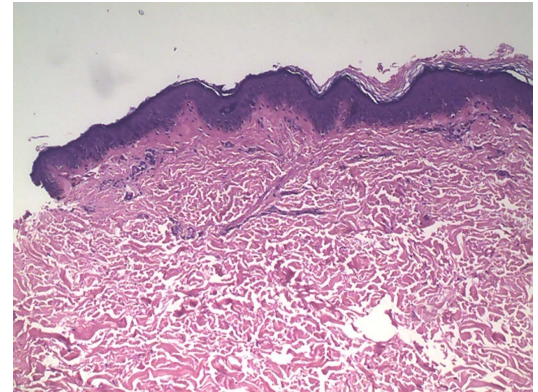


Figure 2 Photomicrograph showing hypergranulosis lamellated orthohyperkeratosis in the epidermis. An incomplete cleft is seen above the granular layer (H&E, ×40).

structural proteins and plays an important role in the formation of the cornified cell envelope. Disease associations have been reported and include fragile hair, koilonychia, onycholysis, chapping, keratoderma, sexual dysfunction, anosmia, short stature, primary amenorrhoea, cheilitis, keratosis pilaris, melanonychia, clubbing and hyperhidrosis. Abnormal biochemical parameters such as altered tryptophan levels, aminoaciduria, high copper level, higher IgE and ceruloplasmin, iron and iron-binding capacity and abnormal epidermal retinoid metabolism have also been reported.³

Learning points

- ▶ Generalised peeling skin syndrome is an autosomal recessive ichthyosiform disorder characterised by asymptomatic continuous or periodic generalised peeling of the skin, with or without trauma.
- ▶ The defect is a mutation in gene encoding for the corneodesomin, which plays an important role in adhesion of the corneocytes.
- ▶ An acral mild variant is confined to the palms and soles, and is associated with mutation in transglutaminase 5, resulting in defective cornification.

Competing interests None declared.

Patient consent Obtained.

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Figure 1 Clinical photograph showing peeling of superficial skin over the volar aspect of the forearm after induced mild trauma.



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