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Dry skin and blistering in childhood

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Clinical findings

A 5-year-old boy presented with a history of dry scaly skin. He had been born at term, with no collodion membrane or erythroderma noted at delivery. Skin changes were noted soon after birth, with widespread dryness and occasional blistering, mainly affecting the toes. Previous treatment with emollients and topical corticosteroids had not resulted in improvement. There was no family history of dermatological disease, and his parents were not related. Physical examination revealed mild, light-grey hyperkeratosis, particularly on the extensor aspects of the skin overlying the joints and the dorsal surfaces of the feet. In addition, superficially denuded areas with collarette-like borders (known as the Mauserung phenomenon) were seen, most notably on the knees (Fig. 1a–c). The palms, soles, head and neck were spared, and the hair and all nails were normal.

Histopathological findings

Histological examination of a skin biopsy taken from the ankle region showed marked hyperkeratosis of the stratum corneum and a prominent granular layer with vacuolization (Fig. 2a).

What is your diagnosis?



Figure 1 (a) Light-grey hyperkeratosis overlying the ankle and the dorsa of the feet; (b) superficially denuded area with collarette border on the skin overlying the knee (known as the Mauserung phenomenon, or moulting), which develops due to superficial blistering and shedding of the stratum corneum; (c) hyperkeratosis of the ankle, with well-defined peeling skin, consistent with the Mauserung phenomenon, i.e. small patches of apparently normal skin in the middle of areas of hyperkeratosis.

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Learning points

- SEI is a rare autosomal dominant genodermatosis that can be inherited or occur on a sporadic basis.
- Key features include hyperkeratosis, blistering and localized superficial skin peeling. Blistering improves with age.
- Histologically, epidermolysis is seen in the granular layer.
- Genetic testing for a mutation in *KRT2* can be useful in confirming the diagnosis.

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