Chapter 18
Hyperkeratotic Plaques on the Extensor Extremities and Punctate Palmoplantar Keratoderma in a Boy

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An 8-year-old boy presented to dermatology with reddish lesions on his extremities that were predominantly observed on the elbows and knees, and thickening of the palms and soles since early childhood (Figs. 18.1, 18.2, and 18.3). The lesions were mainly distributed over the extensor surfaces. They started as erythematous, scaly papules and progressed into hyperkeratotic plaques covering large areas; however, they were not migratory in nature. The patient's developmental milestones were typical. The boy was born out of a non-consanguineous marriage, and none of his family members had similar lesions. Skin examination revealed well-defined, erythematous, hyperkeratotic plaques distributed symmetrically over the dorsal aspect of the fingers and feet, knuckles, elbows, and knees. Hyperkerototic, punctate lesions were present over pressure-prone areas on the soles and palms (Fig. 18.4). Toenails showed pits, and a median canalicular ridge was present on the big toes. The review of systems was unremarkable, and routine serologic parameters were within normal limits.

Based on the Case Description and Photographs, What Is Your Diagnosis?

- Erythrokeratoderma variabilis
- Epidermolytic ichthyosis
- Progressive symmetric erythrokeratoderma
- Olmsted syndrome
- Psoriasis

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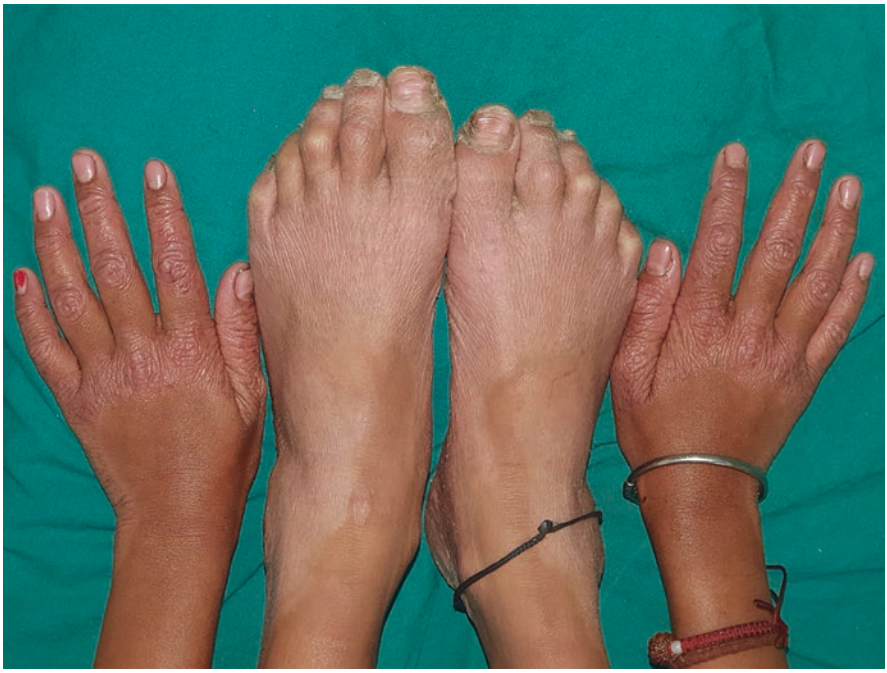

Fig. 18.1 Large hyperkeratotic plaques, almost perfectly symmetrically distributed over the dorsal feet and hands

Diagnosis

Progressive symmetric erythrokeratoderma (PSEK). The diagnosis is supported by the history and clinical findings, including the symmetric involvement of the extensor surfaces of the extremities and non-migratory nature of hyperkeratotic lesions.

Discussion

PSEK is a rare genodermatosis. It is an autosomal disorder of cornification with variable penetrance associated with frameshift mutations in loricrin gene [1]. Positive family history is observed in half of the cases - the rest are due to spontaneous insertional mutations in loricrin gene. PSEK usually manifests in the first decade of life, but cases with adult onset have have been reported [1]. Unlike erythrokeratoderma variabilis (EKV), the closest differential diagnosis, the neonate with PSEK has no skin lesions at birth.

The condition is characterized by well-defined, erythematous, hyperkeratotic plaques affecting the extensor surfaces of the extremities, buttocks, and

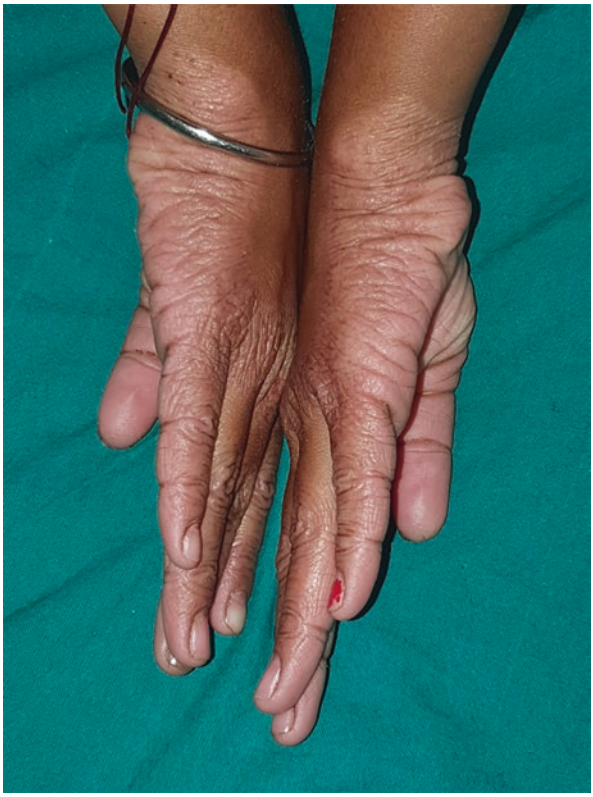

Fig. 18.2 Symmetric hyperkeratotic plaques on the lateral aspect of the hands

occasionally the face; the trunk is typically spared, although lesions on the chest and abdomen have been reported [1–3]. The lesions are almost perfectly symmetric. These hyperkeratotic plaques are slowly progressive, increasing in number and size throughout early childhood before either stabilizing, regressing or disappearing sometime later during life [4, 5]. Waxing and waning may exceptionally occur. Calloused skin on the palms and soles (PPK; palmoplantar keratoderma) is not uncommon, and can be disabling [5].

The histopathological features of PSEK include orthokeratosis, focal parakeratosis, mild or moderate acanthosis, normal or prominent granular layer, and a perivascular lymphocytic infiltrate in the papillary dermis [6]. PSEK is diagnosed based on the history and clinical findings, especially as loricrin gene analysis is difficult in hospital settings [1].

Here, we report a rare presentation of PSEK associated with punctuate palmoplantar keratoderma. We started the patient on topical keratolytics, emollients, and oral acitretin. Topical calcipqotriol has been helpful, but topical retinoids and steroids have shown variable efficacy [3].

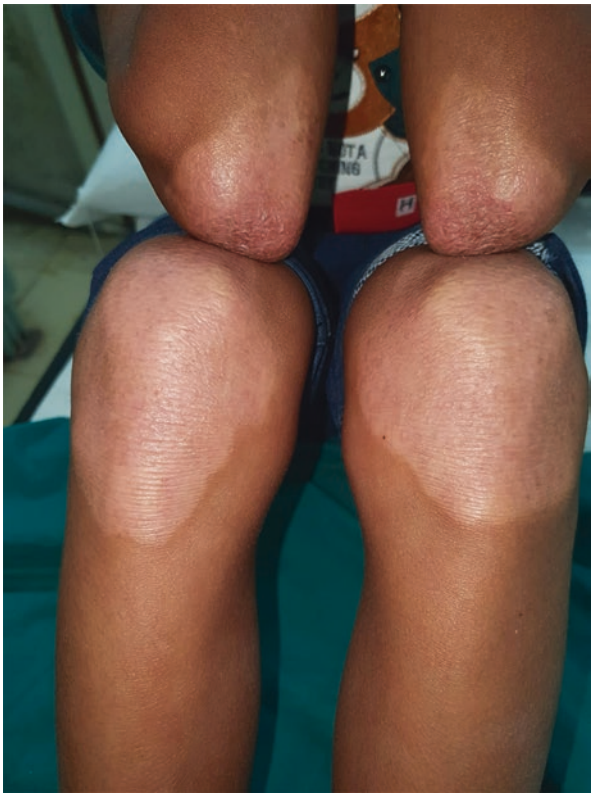

Fig. 18.3 Hypopigmented thickened skin with accentuated skin markings on the elbows and knees

Key Points

- Erythrokeratoderma is a rare group of genodermatoses - the majority of individuals with features of erythrokeratoderma belong to the clinical spectrum of EKV or PSEK.
- PSEK typically presents with symmetrically distributed, non-migratory, large, well-defined, erythematous, hyperkeratotic plaques. The lesions are strikingly symmetrical and affect predominantly the extensor surfaces of the extremities and sometimes the face.
- PSEK usually presents in early childhood and, unlike EKV, not at birth.
- Punctate type of PPK was noted in our patient - this is an exceptional association of PSEK.

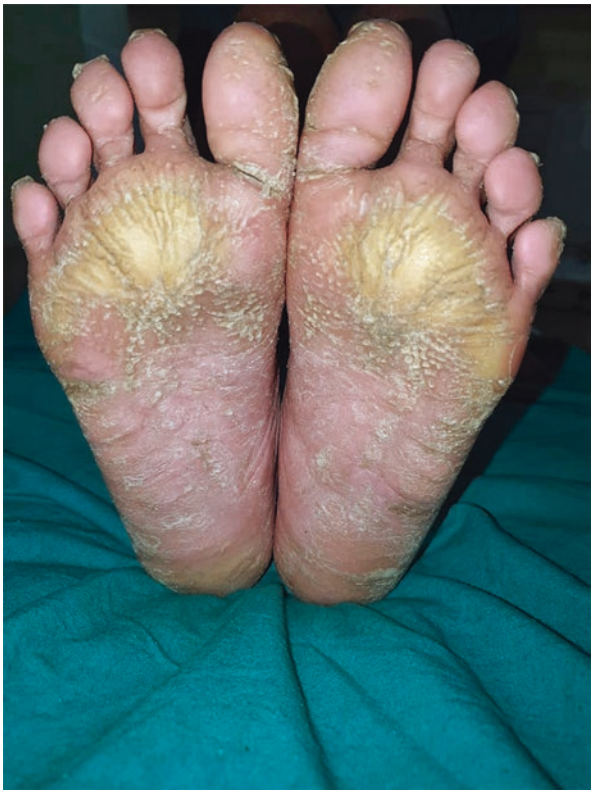

Fig. 18.4 Punctate keratoderma is noted on the soles

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