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**Case Report**

# A rare case report of progressive symmetric erythrokeratoderma in five generations of an Indian family

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## ABSTRACT

Erythrokeratoderma is a rare group of disorders of autosomal dominant inheritance characterised by localised erythema and hyperkeratosis. Within a broad spectrum of phenotypes atleast two are delineated: a) Erythrokeratoderma variabilis and b) Progressive symmetric erythrokeratoderma. Here we are reporting a case of progressive symmetric erythrokeratoderma in an Indian family where five successive generations were involved.

**Keywords:** Erythrokeratoderma, Five generations, Autosomal dominant, Progressive symmetric erythrokeratoderma

## INTRODUCTION

Erythrokeratoderma is a rare group of disorders inherited mainly in autosomal dominant manner, characterised by localised erythema and hyperkeratosis of body parts that is either stationary or migratory.

Within a broad spectrum of phenotypes, at least two disorders can be delineated[1]:

1. Erythrokeratoderma variabilis or *Mendes de Costa syndrome*
2. Progressive symmetric erythrokeratoderma or *Gottron syndrome*

Progressive symmetric erythrokeratoderma (PSEK), first described by Darier in 1911, later renamed by Gottron is characterised by well demarcated, erythematous, hyperkeratotic plaques that are symmetrically distributed over the extremities, buttocks and, often face.[2] The trunk is spared but palms and soles may be involved. Large geographical but symmetrical fine scaly plaques with an orange-red erythema appear in infancy or at times childhood. The plaques usually remain stable in location and appearance but may undergo partial regression at puberty. Keratoderma is also seen in patients with occasional complains of pruritus. Although it is considered to be autosomal dominantly inherited, 40% of cases occur sporadically.[3]

## CASE REPORT

An 18 year old girl came to Dermatology OPD with complains of dusky red erythematous hyperkeratotic plaques present bilaterally over her hands, feet, buttocks and knees (shown in Figure 3 (A-C). According to her the lesions exaggerated during winter and resolved during summer. She was suffering from these symptoms since her childhood. She also had thickened soles and palms with hyperpigmentation and thickening over her dorsal hand and feet. According to her the plaques developed over the same sites during winter and resolved in summer. On examination mainly the extensors were involved; some lesions were present on her buttocks, knees and elbow also. The lesions were polycyclic and bizzarely shaped with hyperkeratosis and underlying

erythema at the margins; she had mild pruritus in the lesions occasionally.

On taking history from her we found that five generations of her family were involved with similar disease. History of consanguinity was present in her maternal side of family members; her father did not have any symptom of the disease but her mother had similar disease features. According to her, in her mother's family two of her sisters and one brother had similar lesions on body since childhood; her maternal grandfather had similar disease who inherited it from his mother. According to the patient, her elder sister had the disease whose daughter was also affected with similar features. We found that the severity of the presentation of the disease in various family members was variable. Some had very mild erythema with hyperkeratosis while a few had more severe hyperkeratosis with erythema. The inheritance pattern has been described using the given flowchart (Figure 1).

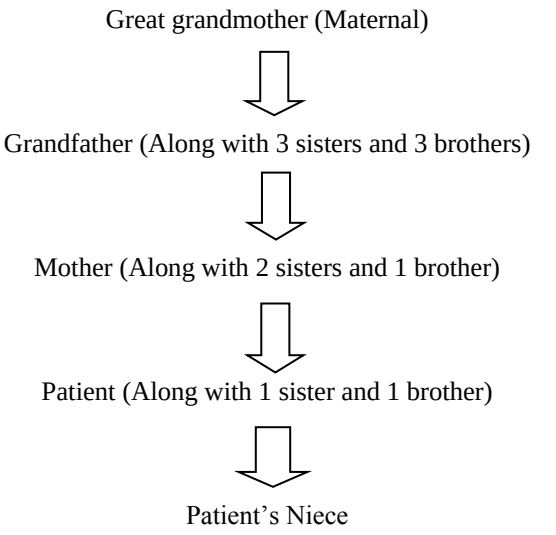


**Figure 1: The given flowchart shows the inheritance pattern in the patient's family.**

A differential diagnosis was considered for erythrokeratoderma variabilis progressiva and progressive symmetric erythrokeratoderma. We took a biopsy of the lesion from the girl's foot.

On histopathological examination sparse superficial perivascular lymphocytic infiltrate with slight epidermal hyperplasia was present. The epidermis also showed mild focal spongiosis with a normal granular layer and a moderately thickened stratum corneum. The stratum corneum had lamellated parakeratosis and orthokeratosis. The images of histopathology are given below (Figure 2). These findings with clinical correlation were suggestive of progressive symmetric erythrokeratoderma.

The patient was started on low dose oral isotretinoin (0.5 mg/kg/day) and topical keratolytics (salicylic acid 3% and urea 10%). After two months of treatment her lesions improved significantly. She was followed up regularly at one month interval for 6 months.

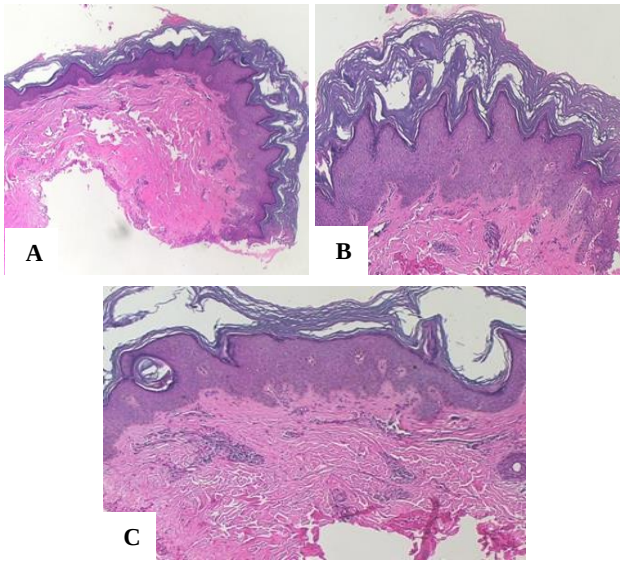


**Figure 2 (A-C): Biopsy shows sparse superficial perivascular lymphocytic infiltrate with slight epidermal hyperplasia. The epidermis also shows mild focal spongiosis with a normal granular layer and a moderately thickened stratum corneum. The stratum corneum shows lamellated parakeratosis and orthokeratosis.**

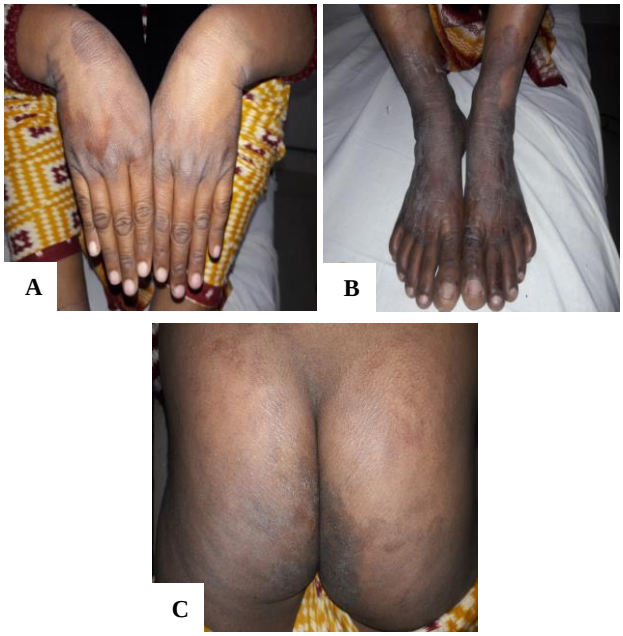


**Figure 3: (A) Hyperkeratotic erythematous lesion on hands); (B) Hyperkeratotic erythematous lesion on feet; (C) Similar erythematous lesions on buttocks.**

## DISCUSSION

The prevalence of PSEK in the general population is unknown. The disorder was first described by Darier in

1911. Since then, fewer than 100 cases have been described in the medical literature.[4]

PSEK is typically caused by a genetic mutation in an as of yet unidentified gene(s), which may occur at random (i.e., spontaneous new mutation) or be inherited as an autosomal dominant trait. Recently, a mutation in GJB4, which encodes connexion 30.3, was found in patients from Netherlands with either PSEK or EKV. To discuss the potential causes of PSEK, the risk of having children with this disorder and the possibility of genetic testing, genetic counselling may be of benefit for affected individuals and their families.[5]

The genetic analysis in PSEK patients has shown mutation in the loricrin protein with the gene coding region located on chromosome 1q21.3.[6] Recently, a novel locus on chromosome 21q11.2-21q21.2 was identified for PSEK in a Chinese family. These studies only suggest that further research is required to identify the mutation causing this disorder.

The treatment options for PSEK include topical keratolytics, emollients, steroids, retinoids and oral retinoids (Acitretin and Isotretinoin). Use of topical calcipotriol with the good response has been reported in a patient.[7]

These case reports are significant as they spread awareness about rare diseases worldwide and encourage research workers to find the definite pathology and management of such cases.

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