Progressive Symmetrical Erythrokeratoderma (PSEK) – A Mimicker of Psoriasis, a Rare Case Report

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Abstract:
Progressive symmetrical erythrokeratodermia (PSEK), also known as Gottron's disease, is a rare autosomal dominant genodermatosis with variable penetrance presenting as sharply defined, erythematous and hyperkeratotic plaques symmetrically over the limbs and buttocks. This is a report of two siblings presenting with these lesoins which started at the age of 5 years in the male and 3 years in the female born out of third degree consanguineous marriage. Both were treated with 0.5mg/kg oral isotretinoin and moisturisers containing urea which showed drastic improvement in two months. Histopathology showed marked ortho hyperkeratosis and focal irregular areas of acanthosis and focal papillomatosis. It's important to rule out psoriasis which is it's closest differential diagnosis. Lesions progress during childhood and stabilise after puberty

Key-words – Progressive symmetrical erythrokeratodermia (PSEK), Gottron's disease, Genodermatosis

Introduction- Progressive symmetric erythrokeratoderma (PSEK) is a rare autosomal dominant disorder belonging to erythrokeratoderma characterized by non-migratory, erythematous or hyperpigmented, symmetric plaques that are usually distributed on the extremities, buttocks, and sometimes the face. The erythematous aspect of erythrokeratoderma appears to be less pronounced or clinically significant in individuals with Type IV to VI skin. There can be associated neurological abnormalities. [(1)]

Case report

Two siblings, 15-year-old female and 11-year-old male presented with itching over the body since 3 years of age in the female child and since the age of 5 years of age in the male child

History of the presenting illness- The patients were apparently alright till their mentioned ages when they developed itchy small red coloured raised lesions over the dorsa of bilateral feet which gradually progressed in size and involved the knees, elbows and dorsa of both the hands in that order in a span of 5 years in the male and 7 years in the female child. There was history of third-degree consanguineous marriage with no such complaints in the family.
On examination, there were well defined, sharply demarcated, symmetrical skin-coloured to erythematous hyperkeratotic plaques covering the dorsa of both hands and feet (Fig-1a and 2a).

Histopathological examination showed ortho hyperkeratosis, focal irregular areas of acanthosis and focal papillomatosis in epidermis. The upper dermis was loose with focal spongiosis with aggregates of mixed inflammatory infiltrates of lymphocytes, macrophages and occasional neutrophils (Fig 3).

Both were prescribed oral isotretinoin 0.5 mg/kg/day, moisturiser containing 20% urea which showed dramatic improvement in two months (Fig- 1b and 2b). They continued the same treatment for one more month and later maintained on moisturiser containing 10% urea. There was no recurrence of lesions on follow-up for the next 6 months.

Discussion-"Erythrokeratodermas" are a group of rare genodermatosis which compromises two important non-syndromic conditions –progressive symmetric erythrokeratoderma (PSEK) and erythrokeratoderma variabilis (EKV) – and some syndromes like as KID (keratitis, ichthyosis, deafness) syndrome and HID (hystrix-like ichthyosis withdeafness) syndrome[3]. PSEK is eponymically known as Darier- Gottron syndrome though it was first described by Darier. [4]
Most cases of PSEK exhibit an autosomal dominant inheritance pattern characterized by incomplete penetrance and variable expressivity, though sporadic occurrences are also frequently observed. Additionally, there have been a few reports of autosomal recessive inheritance. The molecular mechanisms underlying PSEK remain largely undefined, with no definitive candidate genes identified to date. Notably, a mutation in the GJB4 gene, which encodes the connexin 30.3 protein commonly associated with EKV, was identified in a singular case of PSEK.[5]. However, this mutation has not been detected in any other cases examined.[6-8]

The typical clinical manifestation of PSEK involves symmetrically distributed, nonmigratory, large, well-defined, erythematous, hyperkeratotic plaques characterized by pronounced peripheral erythema. Predominantly, these lesions appear on the extensors of the extremities and occasionally on the face, while generally sparing the trunk. PSEK predominantly presents during infancy or childhood and displays no

preference for any gender. [1]The progression of lesions is gradual throughout childhood and usually stabilizes during the teenage years.[2]

EKV, psoriasis, and pityriasis rubra pilaris are considered primary differential diagnoses for PSEK. [3]. However, the latter two conditions can be effectively distinguished from PSEK through clinical examination and histopathological analysis.

Conclusion – Transmission in PSEK is autosomal dominant with sporadic cases occurring in 50% cases, affecting both the genders equally. The lesions slowly progress in size and number during childhood and stabilise after puberty. The inherited cases have lifelong symptoms but there are reports of spontaneous improvement after many years in sporadic cases. Documenting such highlights the role of genetic counselling and expands the clinical understanding of PSEK, reducing the likelihood of misdiagnosis and diminishing the necessity to correlate it with a syndrome.

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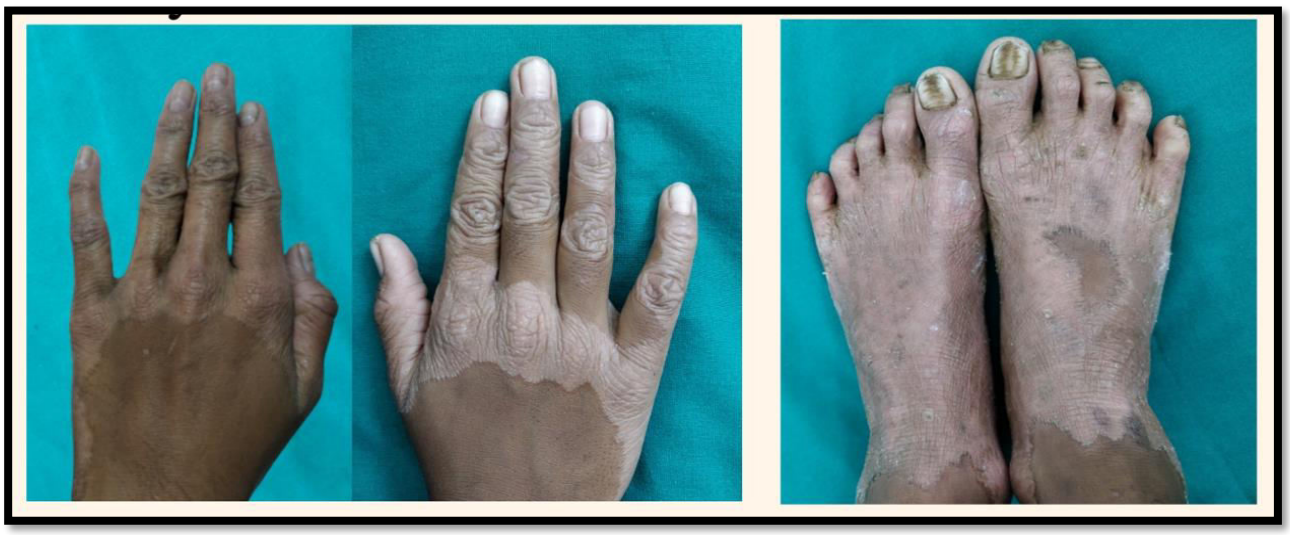
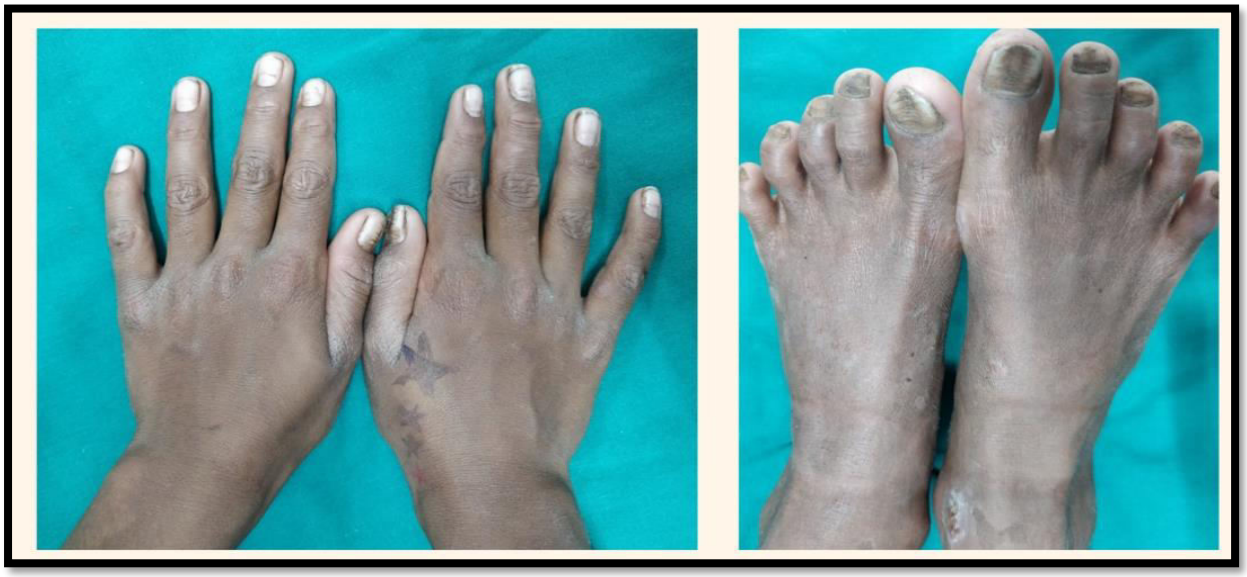
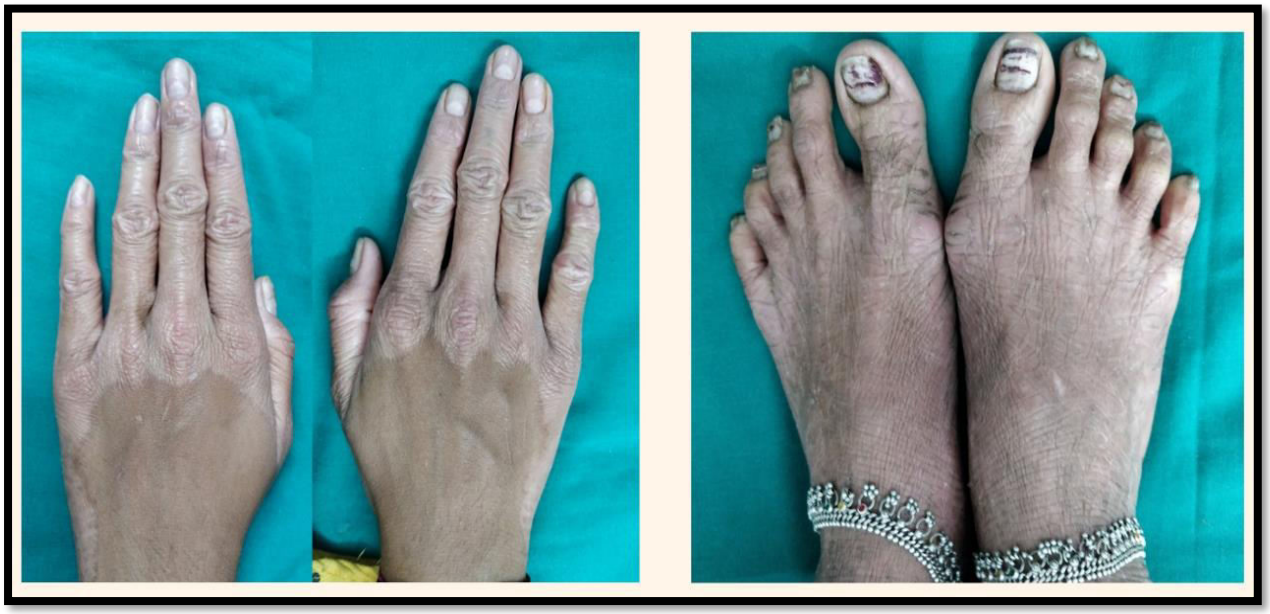


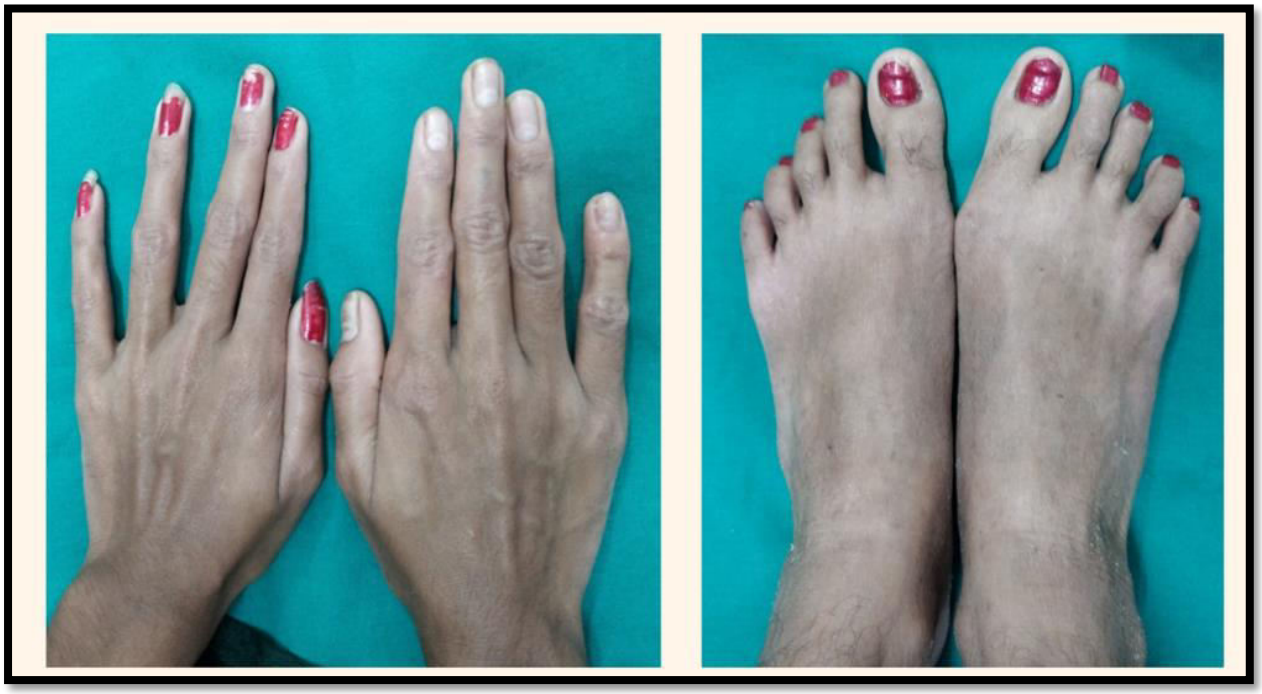
1a



1b



2a



2b

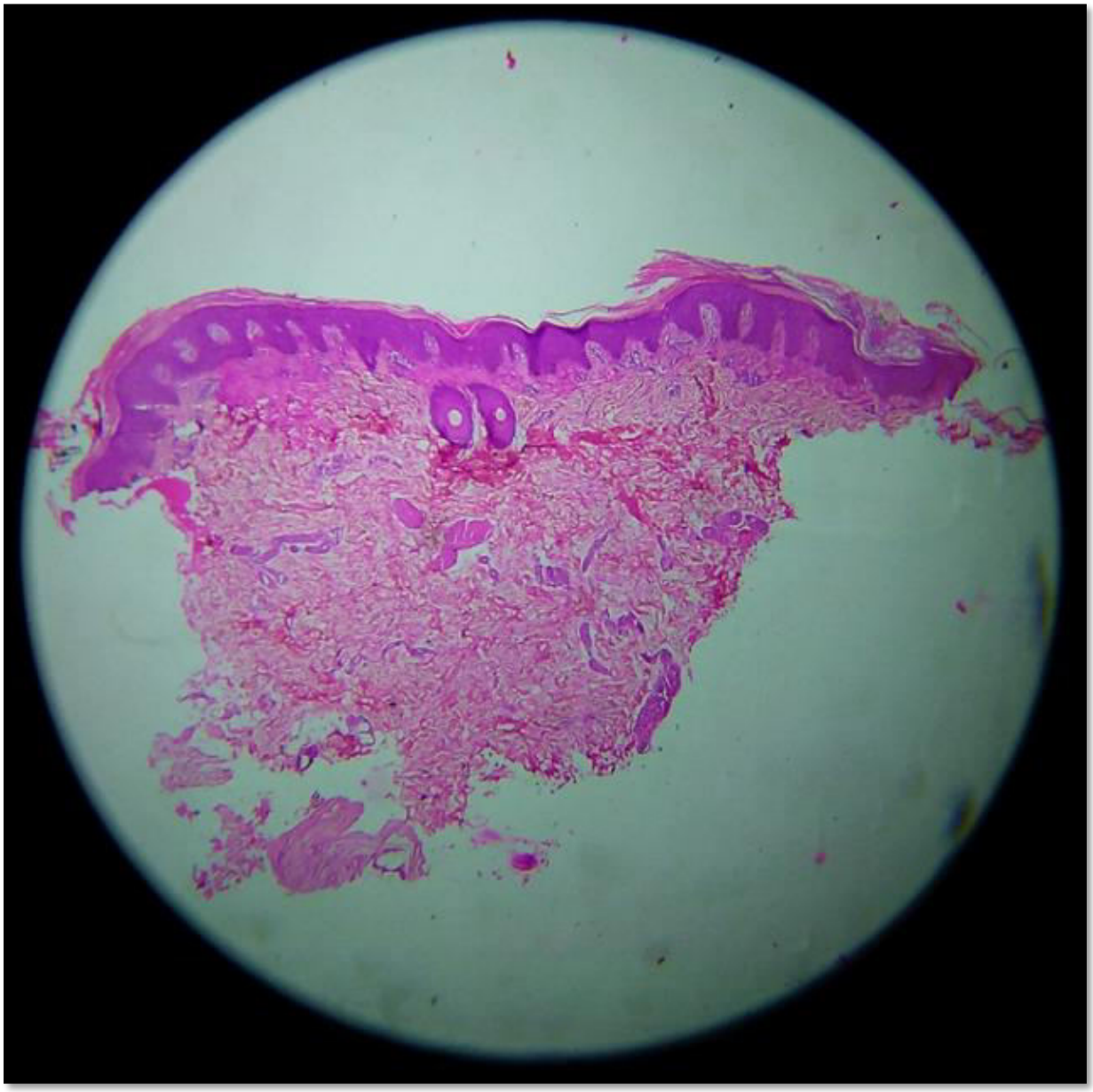


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Figure legends-

1a- Dorsa of both hands and feet showing well defined, sharply circumscribed erythematous hyperkeratotic lesions in the male child before treatment

1b- Dorsa of both hands and feet showing well defined, sharply circumscribed erythematous hyperkeratotic lesions in the male child after two months of treatment

2a- Dorsa of both hands and feet showing well defined, sharply circumscribed erythematous hyperkeratotic lesions in the female child before treatment

2b- Dorsa of both hands and feet showing well defined, sharply circumscribed erythematous hyperkeratotic lesions in the female child after two months of treatment

3 - The epidermis showed ortho hyperkeratosis, focal irregular areas of acanthosis and focal papillomatosis. The upper dermis was loose with focal spongiosis with aggregates of mixed inflammatory infiltrates of lymphocytes, macrophages and occasional neutrophils.

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The manuscript has been read and approved by all the authors, that the requirements for authorship have been met, and that each author believes that the manuscript represents honest work