VOLUME - 12, ISSUE - 02, FEBRUARY - 2023 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Dermatology

A CASE SERIES OF PROGRESSIVE SYMMETRIC ERYTHROKERATODERMIA IN DIFFERENT AGE GROUPS

Dr. Soundarya S	Assistant Professor, Department Dermatology venereology and leprosy, Chettinad hospital and research institute.
Dr. T U Pooja	Junior Resident, Department Dermatology venereology and leprosy, Chettinad hospital and research institute.
Prof. Jayakar Thomas	Emeritus Professor, Department Dermatology venereology and leprosy, Chettinad hospital and research institute.
Dr. Brindha	Junior Resident, Department Dermatology venereology and leprosy, Chettinad hospital and research institute.

ABSTRACT Progressive symmetric erythrokeratodermia (PSEK) is an autosomal dominant inherited disorder with genetic mutation. It is one among the broad spectrum of erythrokeratoderma characterised by erythematous to hyperkeratotic scaly plaques with non-migratory pattern.In this article , we present 3 cases of Progressive symmetric erythrokeratoderma (PSEK) along with the review of literature.

KEYWORDS : Erythrokeratoderma , Progressive symmetric erythrokeratodermia , autosomal dominant, genodermatosis , extensor aspect, non-migratory, symmetrically distributed

INTRODUCTION

Erythrokeratoderma is a rare group of disorder characterized by widespread erythematous plaques, which are either migratory (Erythrokeratodermia Variabilis or Mendes Da Costa syndrome) or stationary (Progressive symmetric erythrokeratodermia)[1].

Progressive symmetric erythrokeratodermia (PSEK) also known as Gottron's syndrome, first described by Darier in 1911[1][2], is a heterogenous genodermatosis[3] of autosomal dominant inheritance characterised by well demarcated, erythematous to hyperkeratotic plaques with minimal scaling that distributed bilaterally symmetrical over the knees, elbow, dorsal aspect of hands and feet, buttocks and occasionally thighs, upper arm, face.

The palms and soles are usually spared. PSEK may not be present at the time of birth, it usually manifests during infancy and rarely during adult life. It progresses in early childhood[4]. PSEK becomes stable and some may begin to regress at puberty. The molecular basis of PSEK is unclear, but some studies suggest the mutation in loricrin gene[1][2].

Case Report

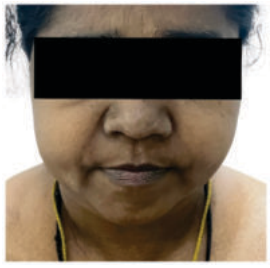
Case 1

A 45-year-old female presented to dermatology OPD with complaints of hyperpigmented plaques with symmetrical distribution over trunk, upper arm, forearm, dorsal aspect of hand and feet, buttocks, thigh, knee, shin and face since childhood. It was progressive in nature. There was no migration of lesions present.

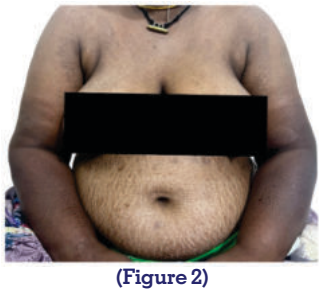
Patient was asymptomatic throughout the course. She is deaf and mute since birth. There was no significant past medical history. On eliciting history from her companion, we could find that there are similar lesions for her brother.

On physical examination, multiple, well defined, slightly erythematous to hyperpigmented keratotic scaly ichthyotic plaques distributed symmetrically over the trunk, upper arm, forearm, dorsal aspect of hand and feet, buttocks, thigh, knee, shin and face.

Diagnosis of Progressive symmetric erythrokeratodermia were considered. To conclude with a diagnosis skin biopsy was taken from the left shoulder region.



(Figure 1)



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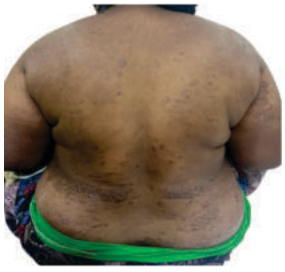

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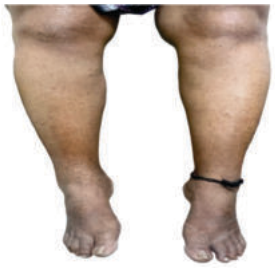
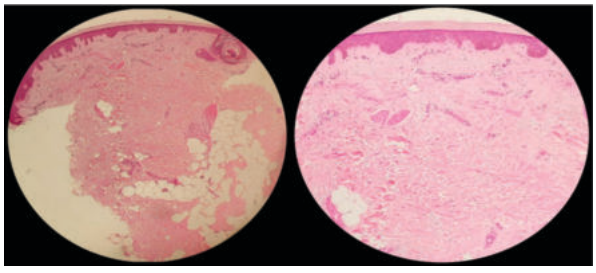


(Figure 4)

Multiple, well defined, slightly erythematous to hyperpigmented keratotic scaly ichthyotic plaques distributed symmetrically over her face (figure 1), trunk and upper limbs (figure 2), back (figure 3), both lower limbs (figure 4)

Histopathological report revealed mild hyperkeratosis with irregular acanthosis, papillomatosis, focal basal cell vacuolation and spongiosis. Papillary dermis showed mild perivascular mononuclear inflammatory infiltrates and sclerosis.

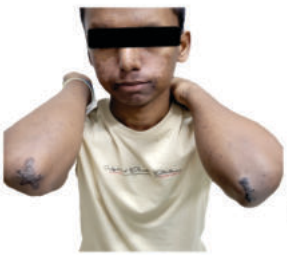


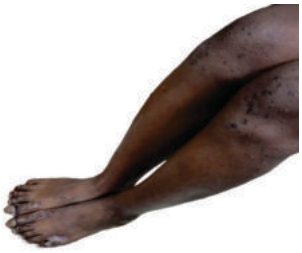
These findings were consistent with Progressive symmetric

erythrokeratodermia. The patient was managed with oral isotretinoin, topical keratolytics and emollients

Case 2

A 13-year-old boy presented to dermatology OPD with complaints of erythematous, scaly plaques present symmetrically over both the feet, elbows and knees, face. It started to appear at his age of 2 and initially it was small and progressed to the current size. Patient was asymptomatic. There is no significant past or family history present. On examination, well defined hyperkeratotic plaque with scaling was present symmetrically over both the elbows and knees. Hypopigmented scaly plaques distributed symmetrically over both feet and face.





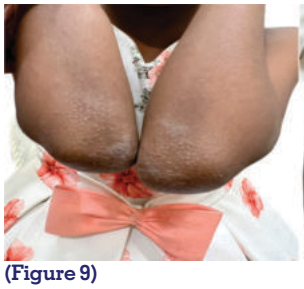
(Figure 7) **(Figure 8)**

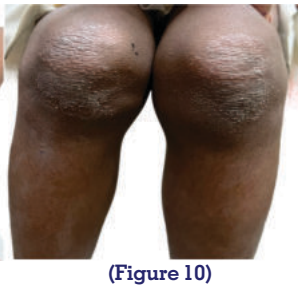
well defined hyperkeratotic plaque with scaling over both the elbows (figure 7) and knees (figure 8). Hypopigmented scaly plaques distributed symmetrically over both feet (figure 8) and face (figure 7)

To confirm the diagnosis skin biopsy was taken from right elbow region for histopathology and it was consistent with Progressive symmetric erythrokeratodermia. Then the patient was started on emollients, topical keratolytics, and oral acitretin. Patient improved symptomatically.

Case 3

A 10 old girl came with complaints of erythematous scaly plaques present over both elbows and knees for the past 6 years. It had a gradual onset. Patient was asymptomatic. There is no significant past or family history. On physical examination, well defined erythematous to pigmented scaly plaque presented symmetrically over both elbow and knees. Differential diagnosis of Progressive symmetric erythrokeratodermia & Juvenile Pityriasis rubra pilaris were considered.





(Figure 9) **(Figure 10)**

well defined erythematous to pigmented scaly plaque presented symmetrically over both elbow (figure 9) and knees (figure 10)

Biopsy was taken to narrow down the diagnosis and histopathology was suggestive of Progressive symmetric erythrokeratodermia. Patient treated with emollients and topical keratolytics.

DISCUSSION

Incidence of PSEK is not yet known, as it is a rare disorder. Few autosomal recessive cases have also been reported. It is also caused due to genetic mutation in GJB4gene encoding connexin 30.3 protein[1][5]. In our study with history elicited there was no familial association found in the last 2 case series. The classification on types of erythrokeratoderma is not yet confined as it is a rare disease, there are some well-defined and atypical types

1. Progressive symmetric erythrokeratodermia (Gottron's syndrome)
2. Erythrokeratodermia Variabilis (Mendes Da Costa syndrome)
3. Erythrokeratoderma en cocardes (Degos syndrome)[6]
4. Localised erythrokeratoderma
5. Erythrokeratoderma like lesions in KID syndrome (Keratitis, ichthyosis, deafness)[2]
6. Progressive partially symmetrical erythrokeratodermia with peripheral neuropathy and deafness
7. Erythrokeratoderma with ataxia[7]
8. Annular migrating erythrokeratoderma
9. Erythrokeratoderma with periorificial lesions[8]

Differential diagnosis of PSEK includes Pityriasis rubra pilaris, Psoriasis, Erythrokeratodermia Variabilis and Eczema. Therefore, clinical and histological findings should be correlated with each other to form a conclusion of PSEK. In our case 1 as the patient was deaf and mute differentials of Erythrokeratoderma like lesions in KID syndrome (Keratitis, ichthyosis, deafness) and Progressive partially symmetrical erythrokeratoderma with peripheral neuropathy and deafness could also be considered but in this case the patient was asymptomatic and doesn't show any associated features of these 2 types of erythrokeratoderma as well as biopsy was more in favour of PSEK.

Clinical features of PSEK include well defined, large, symmetrically distributed erythematous to hyperkeratotic plaque with absence of migratory pattern, which develop gradually mainly over the extensor aspect of extremities and rarely face. It mainly develops during infancy or childhood. Treatment options in PSEK include oral retinoids like Isotretinoin and acitretin[1] which has shown good response by downregulating the plaques and scaling. Topical calcipotriol has also shown the same effect[1][9]. Other options that can be considered for symptomatic improvement which include emollients, keratolytics, topical steroids and topical retinoids. The usage of retinoids was successful, but on cessation of the drug recurrence was report in some cases[9].

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