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*Case Report*

# Keratosis Linearis with Ichthyosis Congenita and Sclerosing Keratoderma Syndrome – An Unusual Presentation

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## ABSTRACT

Keratosis linearis with ichthyosis congenita and sclerosing keratoderma syndrome is a rare autosomal recessive disorder which is characterised by palmoplantar keratoderma, linear hyperkeratotic plaques, ichthyosiform scaling, pseudoainhum and plaques distributed linearly in the flexures. A 7-year-boy presented with ichthyosiform scaling over body since birth, hyperkeratotic plaques over palms and soles. On cutaneous examination, diffuse ichthyosiform scales were present all over body with non-transgradient type of palmoplantar keratoderma with bilaterally symmetrical linear hyperkeratotic plaques over 5$^{th}$ metatarsophalangeal joint. In our case, hyperkeratotic plaque was seen in extensor region over the feet. This case is being reported on account of its rarity and rare presentation.

**Keywords:** Keratosis linearis with ichthyosis congenita and sclerosing keratoderma syndrome, Pseudoainhum, Palmoplantar keratoderma, Ichthyosis

## INTRODUCTION

Keratosis linearis with ichthyosis congenita and sclerosing keratoderma (KLICK) syndrome (OMIM 601952) is a rare autosomal recessive disorder. It is characterised by congenital non-bullous ichthyosiform scaling, sclerosing palmoplantar keratoderma (PPK), constrictive bands on fingers and toes (pseudoainhum) and linear hyperkeratotic plaques in the skin folds.[1] Approximately 14 documented cases identified through literature review.[2] We describe a case of KLICK syndrome with linear keratotic plaques on extensors, not previously reported.

## CASE REPORT

A 7-year-old boy, born to non-consanguineous parents, presented with ichthyosiform scaling over body since birth, hyperkeratotic plaques over palms and soles. Similar lesions were present in two elder siblings. All three children were born in a collodion membrane. There was no history of erythroderma, autoamputation of digits and sensorineural hearing loss. On cutaneous examination, diffuse ichthyosiform scales were present over body [Figure 1], non-transgradient type of palmar keratoderma and sclerosis was limited to distal interphalangeal joint of fingers [Figure 2a]. Psedoainhum was present in all the digits of fingers and toes [Figure 2b and c]. Sclerosis was present till metatarsophlangeal

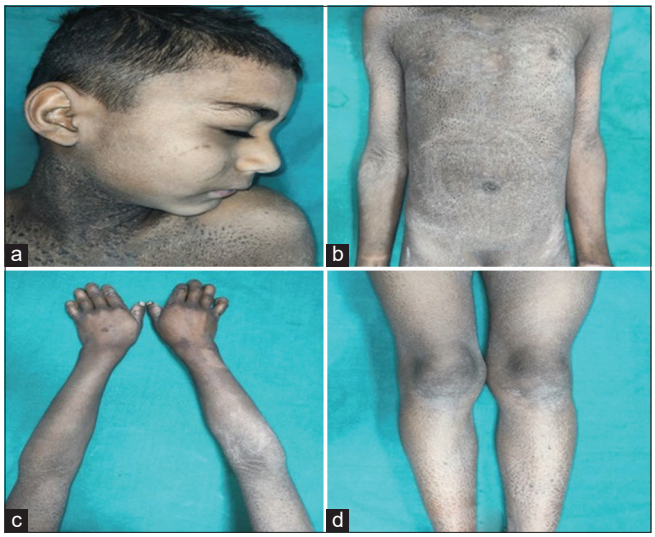


**Figure 1:** (a-d) Ichthyosiform scaling present all over body.

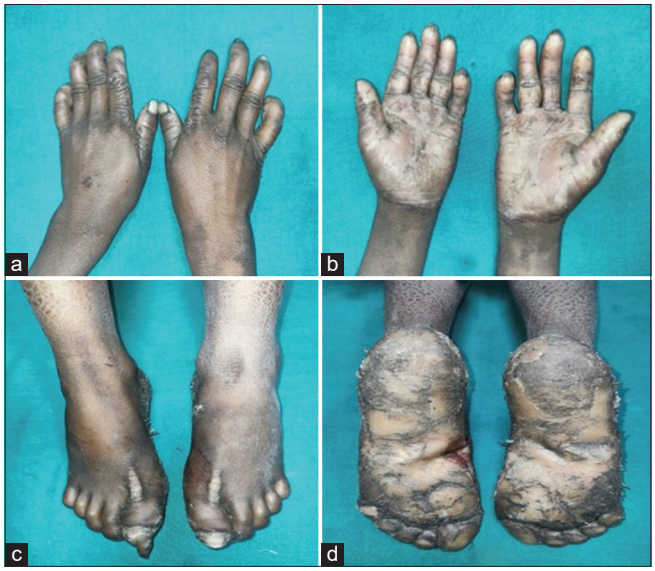


**Figure 2:** (a) Non-transgradient type of palmar keratoderma and sclerosis was limited to distal interphalangeal joint of fingers. (b and c) Psedoainhum was present in all the digits of fingers and toes. (c) Sclerosis was present till metatarsophlangeal joint in all the digits of feet and bilaterally symmetrical linear hyperkeratotic plaques were seen dorsal first metatarsophalangeal joint. (d) Non transgradient type of plantar keratoderma.

joint in all the digits of feet and bilaterally symmetrical linear hyperkeratotic plaques were seen dorsal first metatarsophalangeal joint [Figure 2 c]. Non transgradient type of plantar keratoderma [Figure 2d]. Hair, nails and teeth were normal with no systemic abnormality. Histopathology from linear hyperkeratotic plaque on feet revealed hyperkeratotic stratified squamous epithelium with acanthosis and elongated rete ridges along with areas of hypergranulosis [Figure 3]. Genetic workup was not done due to financial constraints and non-availability at our hospital. The patient is currently being treated with oral retinoids (Acitretin 25 mg/day) with emollients and humectants such as 30% urea creams.

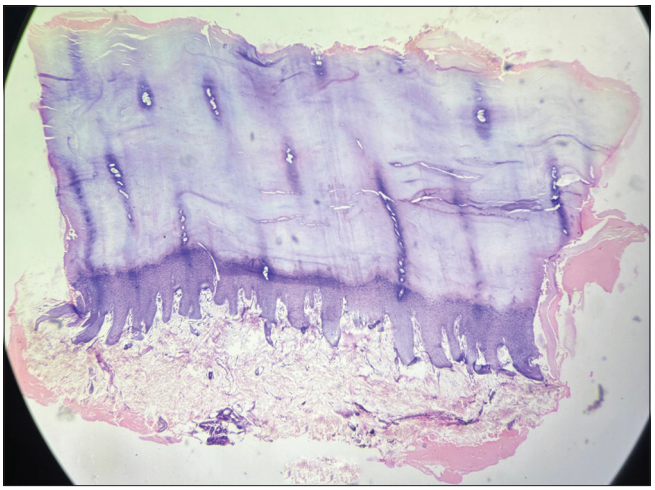

**Figure 3:** Histopathology showing hyperkeratotic stratified squamous epithelium with acanthosis and elongated rete ridges along with areas of hypergranulosis (Haematoxylin and eosin ×10).

## DISCUSSION

KLICK syndrome is a rare genodermatosis characterised by PPK, sclerosis of digits, ichthyosis congenita and linear hyperkeratotic plaques in the flexor regions and axillae.[1]

In 1989, Pujol *et al.* reported four individuals from a consanguineous family exhibiting symptoms resembling KLICK syndrome.[3] In 2006, Chaves *et al.* reported a patient showing typical symptoms of KLICK syndrome with erythematous lesions.[1] KLICK syndrome occurs due to recurrent homozygous 1-bp deletion in the 5'UTR of the POMP gene. This encodes a proteasome maturation protein which is crucial for stabilising the proteasome complex. POMP is a widely expressed protein serving as a chaperone for proteasome maturation. Consequently, the syndrome results from diminished POMP levels, leading to inadequate proteasome activity in differentiating keratinocytes.[2] Disruption in proteosome assembly and decreased activity may cause skin inflammation and hyperkeratosis. Inflammation from hyperactivated innate immunity can trigger autoinflammatory keratinisation disorders (AiKDs). AiKD includes diseases such as pustular psoriasis, hidradenitis suppurativa, KLICK syndrome and porokeratosis.[4]

The list of differential diagnoses includes the keratitis, ichthyosis, deafness syndrome, Vohwinkel syndrome and Olmsted syndrome, as mentioned in Table 1. The most important differential is Vohwinkel syndrome.[5]

**Table 1:** Differential diagnosis of KLICK syndrome.

| Features | KLICK syndrome | Vohwinkel syndrome | KID syndrome | Olmstead syndrome |
|---|---|---|---|---|
| Inheritance | AR | AD | Sporadic (mostly) AR, AD (rarely) | Sporadic (mostly) AR (rare) |
| Mutation | POMP encoding proteosome maturation 40 | Connexin 26 | Connexin 26 | K1, K10, TRPV3 |
| PPK | Diffuse, transgradient | Diffuse, transgradient, honey-comb like | Reticulate PPK with stippled appearance | Mutilating transgradient PPK |
| Ichthyosis | Present | Present | Diffuse scaling, transient congenital erythroderma | Absent |
| Pseudoainhum | Present, fissuring and flexion deformities of digits | Present | Absent | Present, fissuring and flexion deformities of digits |
| Specific cutaneous features | Parallel linear arrays of keratotic papules in the flexural areas of the extremities | Stellate keratoses on knuckles ('starfish appearance') | Linear and spiny hyperkeratosis on face, flexures and extensors of joints and extremities. Perioral rugae | Hyperkeratotic plaques over periorificial and perineal regions |
| Auditory | Normal | Sensorineural hearing loss | Severe sensorineural hearing loss | Normal |
| Ophthalmological | Normal | Normal | Vascularising keratitis leading to blindness | Normal |
| Other features | SCC (complication) | Alopecia, nail abnormalities, spastic paraplegia, myopathy. Epithelioma cuniculatum (complication) | Scarring alopecia of scalp, eyebrows, eyelashes, body hair. Dystrophic nails. May have one or more component of follicular occlusion triad. | SCC and malignant melanoma (complications) |

KLICK: Keratosis linearis with ichthyosis congenita and sclerosing keratoderma, PPK: Palmoplantar keratoderma, AR: Autosomal recessive, AD: Autosomal dominant, KID: Keratitis, ichthyosis and deafness, POMP: Proteosome maturation protein, TRPV: Transient receptor potential vanilloid.

No definite treatment guidelines are for KLICK syndrome. It consists of oral retinoids like acitretin. Others are topicals such as keratolytics and emollients which result in significant improvement in patient's condition. However, recurrence occurrence when the treatment is stopped.[6]

In our case, hyperkeratotic plaque was seen over extensor region over the feet. This case is being reported on account of its rarity and rare presentation.

## CONCLUSION

We are reporting a case of 7-year-old boy with features of KLICK syndrome with linear keratotic plaques on extensors. We are presenting this case to contribute to medical literature by providing comprehensive account on KLICK syndrome and shedding light on its atypical presentation.

### Ethical approval

Institutional Review Board approval is not required.

### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

### Financial support and sponsorship

Nil.

### Conflicts of interest

There are no conflicts of interest.

### Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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