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A Rare Case Of - Keratitis Ichthyosis Deafness Syndrome

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Keywords:

Keratitis-Ichthyosis-Deafness syndrome, congenital ichthyosis, palmoplantar keratoderma, sensorineural hearing loss.

Abstract:

Background: Keratitis-Ichthyosis-Deafness (KID) syndrome is a rare congenital ectodermal disorder characterized by ichthyosiform skin lesions, sensorineural hearing loss, and ocular involvement, with considerable phenotypic variability and occasional familial occurrence.
Case Presentation: A 20-year-old male presented with generalized scaling and hyperpigmented patches since birth, associated with palmoplantar hyperkeratosis and hearing impairment. A similar clinical presentation was observed in his 11-year-old cousin, who had more extensive cutaneous involvement and earlier onset of hearing loss. Cutaneous examination in both patients revealed generalized ichthyosiform scaling with palmoplantar keratoderma, while systemic findings were unremarkable. Skin biopsy demonstrated marked hyperkeratosis and acanthosis consistent with congenital ichthyosis. Based on clinical features, family history, and histopathology, a diagnosis suggestive of KID syndrome was made.
Conclusion: These cases emphasize the phenotypic variability and possible familial nature of KID syndrome. Early recognition is essential for multidisciplinary management, genetic counseling, and long-term follow-up to prevent complications.

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Introduction

keratitis-ichthyosis-deafness (KID) syndrome is a rare congenital ectodermal dysplasia characterized by the classical triad of progressive keratitis, ichthyosiform skin lesions, and congenital sensorineural hearing loss. The condition was first described by Skinner and colleagues in 1981 and is now recognized as a genetic disorder primarily caused by mutations in the GJB2 gene located on chromosome 13, which encodes the gap junction protein connexin-26. Dysfunction of this protein leads to impaired intercellular communication in epithelial tissues, explaining the multisystem involvement of the disease affecting the skin, eyes, and inner ear [1,2].

KID syndrome is extremely uncommon, with approximately 100 cases reported worldwide, and most cases occur sporadically, although autosomal dominant inheritance has been documented. Clinical manifestations usually begin in infancy or early childhood with generalized hyperkeratotic plaques, palmoplantar keratoderma, alopecia, nail dystrophy, and varying degrees of hearing impairment. Ocular involvement, often developing later, includes vascularizing keratitis, corneal

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neovascularization, photophobia, and progressive visual impairment, which may lead to blindness if untreated [3,4].

Patients with KID syndrome are also predisposed to recurrent bacterial and fungal infections due to impaired epithelial barrier function and immune dysregulation. Additionally, there is an increased risk of cutaneous malignancies, particularly squamous cell carcinoma of the skin and mucosa, making long-term surveillance essential. The rarity of the condition and its diverse clinical presentation often lead to delayed diagnosis and management challenges, emphasizing the importance of reporting individual cases to improve understanding of its natural history and therapeutic approaches [5,6].

Case Report

A 20-year-old male presented to the dermatology outpatient department with complaints of multiple asymptomatic hyperpigmented patches associated with generalized scaling of the skin since birth. The lesions were characterized by fish-like scales with lichenification involving the bilateral upper limbs, lower limbs, and trunk. Examination also revealed few skin-colored hyperkeratotic plaques with fissuring over both palms and soles. There was no history of pain, itching, or discharge from the lesions.

A positive family history was noted in his first cousin brother, an 11-year-old boy, who presented with similar dermatological complaints since birth. The younger patient exhibited multiple hyperpigmented patches and plaques with fish-like quadrangular scales and lichenification involving the bilateral upper and lower limbs, trunk, face, and scalp, with diffuse scaling over the scalp. Both patients had associated hearing impairment, which was more severe and had an earlier onset in the younger child.

Cutaneous examination in both cases showed generalized ichthyosiform scaling with palmoplantar hyperkeratosis. No significant nail or mucosal abnormalities were observed. Systemic examination was within normal limits.

Skin biopsy obtained from representative lesions of both patients revealed features consistent with lamellar ichthyosis, including marked hyperkeratosis, acanthosis, and mild inflammatory infiltrate in the dermis, thereby confirming the diagnosis.

Based on the clinical presentation, family history, and histopathological findings, a diagnosis of congenital ichthyosis with associated hearing impairment was made in both patients.

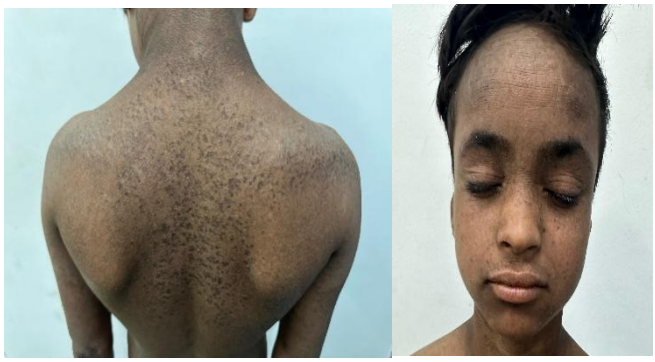

Figure-1: Multiple hyperpigmented and hyperkeratotic papulo-plaque lesions over trunk and face

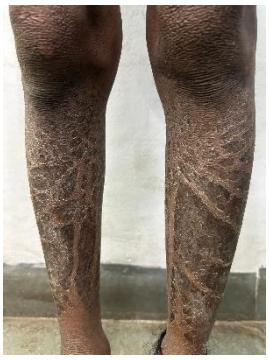

Figure-2: Hyperpigmented and hyperkeratotic plaque over lower 1/3rd of extensor aspect of the leg.

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Figure-3: Prominent dermatoglyphics

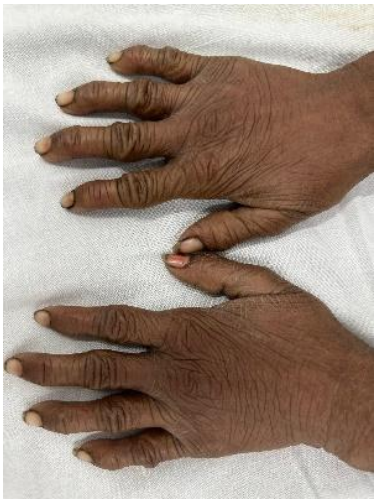

Figure-4: Beaked nails

Discussion

Keratitis-Ichthyosis-Deafness (KID) syndrome is a rare congenital ectodermal disorder characterized by the triad of vascularizing keratitis, ichthyosiform erythrokeratoderma, and sensorineural hearing loss. The condition is most commonly associated with heterozygous missense mutations in the *GJB2* gene encoding connexin-26, which plays a critical role in epidermal differentiation and cochlear ion transport. Disruption of connexin channels results in abnormal keratinization and auditory dysfunction.

Most cases reported in the literature are sporadic; however, familial inheritance with autosomal dominant transmission has also been described. The presence of similar clinical manifestations in two related individuals in our report supports a hereditary etiology. **Kalezić et al. (2022)** reported a familial case with a heterozygous p.Asp50Asn mutation demonstrating comparable cutaneous and auditory features, reinforcing the role of genetic predisposition in familial clustering.

palmoplantar hyperkeratosis with fissuring, generalized ichthyosiform scaling, hyperpigmented patches, and Cutaneous findings in our patients, including generalized ichthyosiform scaling, are consistent with previously described dermatological manifestations. According to **Kalezić et al. [7] (2022)**, palmoplantar keratoderma and hyperkeratotic plaques are commonly observed and may worsen with age due to progressive keratinization abnormalities.

However, unlike several published cases where erythematous plaques, alopecia, and nail dystrophy were prominent, our patients demonstrated mainly scaling and lichenification without significant appendageal involvement. This variation reflects the phenotypic heterogeneity noted in KID syndrome. **Asgari et al. [9] (2020)** emphasized that patients with identical genetic mutations may exhibit markedly different clinical severity, supporting variable expressivity as a hallmark of the disorder.

Both individuals in our report had congenital hearing impairment, which is considered the most consistent clinical feature of KID syndrome and typically presents as severe bilateral sensorineural hearing loss. Interestingly, the younger child exhibited earlier onset and greater severity compared with the adult patient, further demonstrating intrafamilial phenotypic variability.

Ocular manifestations were not prominent in our patients at presentation, whereas keratitis with corneal neovascularization is reported to develop in the majority of cases over time. **Pérez-Rueda et al. [4] (2021)** noted that ocular findings may appear later in childhood or adolescence, suggesting the need for long-term ophthalmologic follow-up even in asymptomatic individuals.

Histopathological examination in both patients revealed marked hyperkeratosis and acanthosis consistent with lamellar ichthyosis. Histology in KID syndrome is often nonspecific and overlaps with other congenital ichthyoses, making clinicopathological correlation essential. **Kalezić et al. [7] (2022)** similarly reported nonspecific histological findings, emphasizing that diagnosis is

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primarily clinical with genetic confirmation when available.

Differential diagnoses include lamellar ichthyosis, Vohwinkel syndrome, Clouston syndrome, and other connexin-associated ectodermal dysplasias. Increasing recognition of overlap among connexin disorders has highlighted the importance of molecular testing for definitive diagnosis, as discussed by **Asgari et al. [8] (2020)**.

Patients with KID syndrome are at increased risk of complications such as recurrent infections, chronic mucocutaneous candidiasis, and cutaneous squamous cell carcinoma due to impaired epithelial barrier function. **Kalezić et al. [7] (2022)** reported malignant transformation in long-standing lesions, underscoring the importance of regular surveillance and multidisciplinary management.

Hearing rehabilitation with cochlear implantation may significantly improve quality of life; however, abnormal skin healing and infection risk may complicate surgical outcomes.

The present case is noteworthy due to the occurrence in two related individuals suggesting familial transmission, absence of significant ocular involvement at presentation, and variable severity of hearing impairment within the same family. These findings further support the phenotypic diversity and diagnostic complexity associated with KID syndrome.

Conclusion

Keratitis-Ichthyosis-Deafness syndrome is a rare genodermatosis with significant clinical heterogeneity and potential familial occurrence. The presence of congenital ichthyosiform scaling associated with hearing impairment should prompt consideration of this diagnosis, even in the absence of prominent ocular findings at presentation. The occurrence of similar manifestations in related individuals in our report suggests a possible hereditary pattern with variable expressivity. Histopathological findings may overlap with other forms of congenital ichthyosis, making clinicopathological correlation essential. Early diagnosis is important to facilitate multidisciplinary care, hearing rehabilitation, genetic counseling, and long-term surveillance for complications such as infections and cutaneous malignancy. Increased awareness among clinicians can aid in timely recognition and improved patient outcomes.

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