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Case Report

Keratoderma Hereditariamutilans: A Rare Disorder of Keratinization

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ABSTRACT

Keratoderma hereditariamutilans is a rare, autosomal dominant disorder of keratinization characterized by palmoplantar keratoderma and ainhum-like constrictions. We report herein a 16-year-old deaf –mute girl, since birth with palmoplantar hyperkeratosis with a honeycomb-like appearance, constricting band in her bilateral little finger, starfish keratosis on extensor surface of joints and nail changes.

Key words: Palmoplantar keratoderma, ainhum like.

INTRODUCTION

Palmoplantar keratoderma (PPKs) are a heterogeneous group of disorders characterized by hyperkeratosis of the palms and soles. The majority of the disorders of this group are hereditary and a few are acquired. Inherited PPKs can further be classified into diffuse, focal PPK and punctuate. Keratoderma hereditariamutilans (KHM) is also known as Vohwinkel syndrome. KHM is an autosomal dominant type of diffuse transgradient palmoplantar keratoderma characterized by honeycomb appearance, pseudoainhum leading to auto amputation, stellate keratosis on knuckles, and associated with sensorineural deafness. The most common mutation responsible is in the gene encoding connexin26, [1] which is found in cochlea as well as in palmoplantar epidermis.

CASE REPORT

A 16-year-old female deaf-mute since birth born of nonconsanguinous marriage by full term normal vaginal delivery after an uneventful pregnancy. Presented with persistent, palmoplantar keratoderma (Fig 1a,1b) since the age of 6 months. Palmoplanter keratoderma progressed steadily, and there was the development of tight band around little finger of both hands since the age of around 2 years. These bands and PPK subsequently lead to deformities of both the hands. (Fig1a)There were starfish-like keratosis over the knuckles and hyperkeratotic papules over the dorsum of both feet. A constriction band around little finger. There was no history of recurrent infections, teeth loss, photosensitivity, photophobia, sweating abnormalities, muscle weakness or walking disability. No h/o any other systemic complaints. She had 3 sisters and one brother, none of them had similar complaints or any other disorder of keratinization.

Examination of the patient revealed diffuse yellowish thickening of the palms

(Fig 1a), and soles (Fig 1b), honeycombed (Fig. 2), by keratotic lesions encroaching on dorsum of hands and feet (Fig. 3), but there was no perilesional erythema. The keratosis was marked leading to clawing of both the hands. (Figure1a) A constricting fibrous band was seen around bilateral little finger (Fig4). Nails showed gross thickening with horizontal ridges, and loss of cuticle (Fig5) there were starfish-shaped keratosis on the dorsa of the fingers and knees (Fig 6a) elbows (6b) and linear keratosis on the flexor aspect of forearms. Hair, mucous membranes, and teeth were normal. Similarly, ocular, and musculoskeletal examination was normal. Patient on bedside tests done for hearing was found to be deaf.

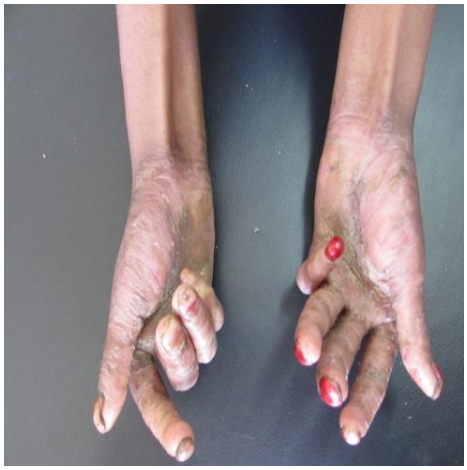


Figure1a honey combing and clawing of hands

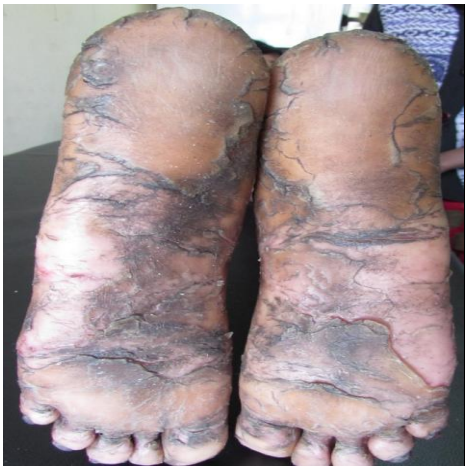


Figure 1b planter keratoderma

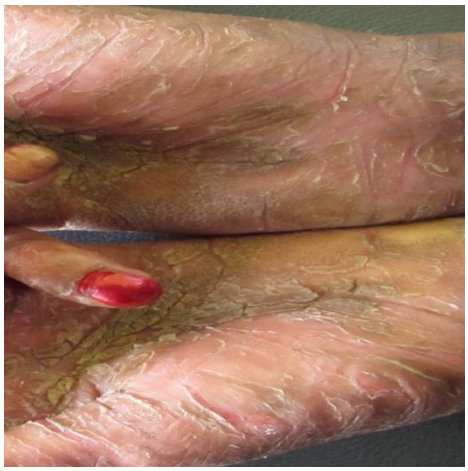


Figure 2: showing honey combing

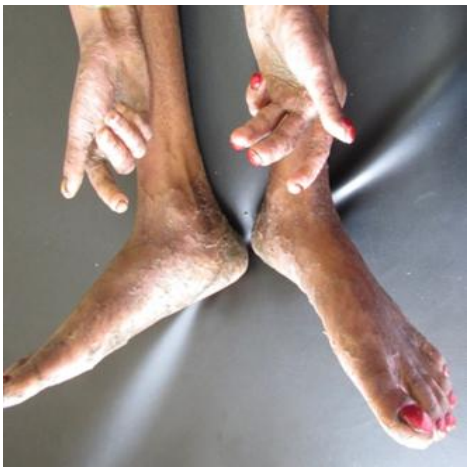
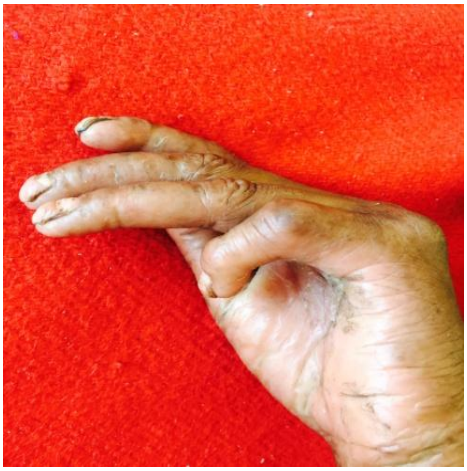


Figure 3: Transgradient PPK



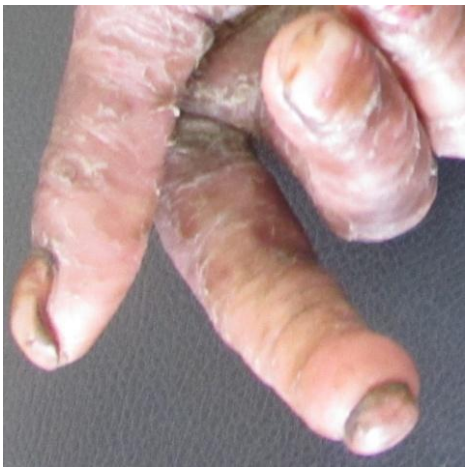


Figure 4: pseudo ainhum in the little finger

Figure 5: nail changes

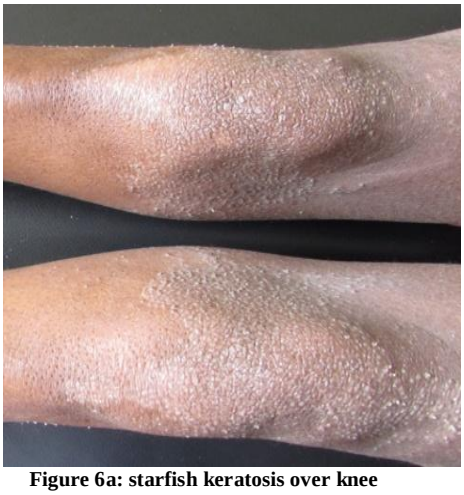


Figure 6a: starfish keratosis over knee

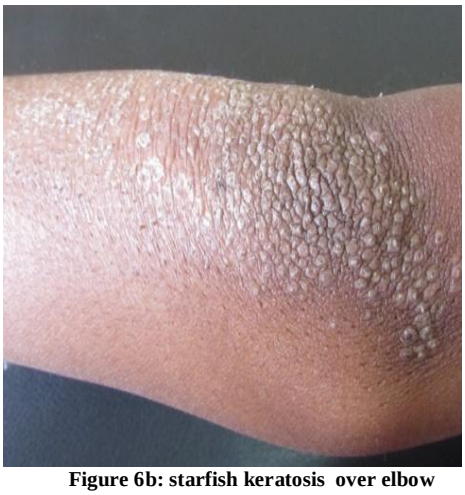


Figure 6b: starfish keratosis over elbow

Routine investigations were normal. Histopathological examination showed - hyperkeratosis, spotty parakeratosis, regular acanthosis, and preserved granular layer. Audiometry revealed bilateral sensorineural hearing loss. We started the patient on oral isotretinoin (20 mg/day) which led to significant improvement after 1 month of therapy with decreased keratoderma and arrest of the progression of fibrotic bands.

DISCUSSION

In 1929, Vohwinkel first described this syndrome in a 24-year-old woman who had a diffuse honeycombed palmar and plantar keratosis, in addition to distal interphalangeal constrictions since 2 years of age. The constrictions ultimately led to auto-amputation. The patient also had mild-to-moderate sensorineural deafness. Vohwinkel syndrome is a rare autosomal dominant palmoplantar keratoderma, with approximately 50 cases reported in the literature, which manifests in childhood and becomes more evident in adulthood. [2] Keratoderma hereditariamutilans also known as "Vohwinkel syndrome" and "mutilating palmoplantar keratoderma" is a rare autosomal dominant disorder although few sporadic cases have also been described. No racial and sex predominance is reported. The lesions start appearing in infancy or early childhood and gradually progress with age. It begins as shiny or translucent papular hyperkeratosis, gradually becoming confluent on hands and feet, warty papules on the knuckles and other extensor sites, coalesce into pathognomic 'starfish keratosis.' The edge of the keratoderma at the wrists and Achilles tendon consists of spiky digitate hyperkeratotic projections onto normal skin, sometimes showing koebnerization. Multiple keratosis on digits produce circumferential hyperkeratosis, which predisposes to the formation of cicatricial bands and auto amputation. The little finger and fifth toe are most commonly affected as was seen in our case.

Two types of mutations of epidermal differentiation have been identified in Vohwinkel syndrome. One is a novel missense mutation of the gap junction protein beta 2 (GJB2 gene) coding connexin-26, a gap junction protein. [3,4] This mutation is associated with the classic, sensorineural hearing loss-associated Vohwinkel syndrome. In Camisa syndrome there is an insertional mutation of the loricrin gene on the epidermal differentiation complex on 1q21. A phenotype associated with ichthyosis. [5] This protein is synthesized in the granular layer; it migrates to the cell periphery, where it is deposited beneath the plasma membrane. It links to several other proteins, forming cornified cell envelope. Thus, it promotes an effective barrier between the

body and the environment. [5] This mutation presents a palmoplantar keratoderma, generalized thin desquamation on the extensor surfaces of the finger joints, and constricting bands around the digits(pseudo-ainhum).Other reported findings of classic Vohwinkel syndrome are ,congenital alopecia universalis, [6] pseudopelade type alopecia, acanthosis nigricans, spastic paraplegia, [7] myopathy, nail changes, mental retardation, bullous lesions on the soles, and seizures. [8] The differential diagnosis of Vohwinkel's syndrome includes other types of keratoderma that can be associated with auto-amputation of the digits: Olmsted syndrome, acral keratoderma, congenital pachyonychia, Sybert's palmoplantar keratoderma, Meleda disease and Gamborg-Nielsen palmar and plantar keratoderma. [9] As genetic and molecular defects have not been elaborated in all these conditions, they are differentiated on clinical grounds mainly. The treatment is very difficult and tends to be symptomatic: keratolytic agents and systemic retinoid [10] are used for the treatment of hyperkeratosis, but without consistent results. [2,11] Excision of the constricting bands, the use of grafts and flaps have been an alternative to hyperkeratosis and auto amputation of the digits. The case is being presented because the disease was sporadic and extensive involvement was there. She had complete sensorineural deafness with extensive palmoplantar keratoderma leading to disability and deformity of hands.

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