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Case Report

Early diagnosis of Voh Winkel syndrome-inherited honeycombed hands and hearing loss

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ABSTRACT

Voh-Winkel Syndrome with approximately 50 cases reported in literature is a rare inherited Palmoplantar keratoderma characterised by honeycomb like hyperkeratosis, hearing loss or ichthyosis. It also features constricting fibrous bands that develop at the base of fingers/toes and later get strangulated to cause autoamputation called pseudoainhum. It is an autosomal dominant disorder with mutations in loricrin and connexin genes leading to hearing loss and ichthyosis variants respectively and manifests in infancy and becomes apparent later in childhood or adult life. Early diagnosis of this condition and management by multiple specialities given the associated features therefore becomes an absolute necessity. An emphasis on dermatological examination to manage hyperkeratosis using topical and systemic therapy, otorhinolaryngology to manage hearing loss as well as plastic surgery to alleviate symptoms arising due to the constriction bands is mandatory for effective management of this condition. Here we report a 22-year-old female patient who came with Palmoplantar keratoderma, bilateral hearing loss and pseudo-ainhum.

Keywords: Palmo-plantar keratoderma, Hearing loss, Pseudo-ainhum

INTRODUCTION

Voh-Winkel syndrome or Keratoderma hereditarian mutilans is an autosomal dominant palmoplantar keratoderma. Recent molecular biological studies indicate mutations of connexin-26 and loricrin wherein missense mutation of connexin 26 and insertional mutation of loricrin are responsible for deafness variant and ichthyosis variant respectively.[1] Nail dystrophy, alopecia, spastic paraplegia, myopathy, onychogryphosis and other neurological abnormalities may also occur in this disorder. There is some overlap of the clinical features of Vohwinkel syndrome with that of other disorders and the treatment of it can vary from simple measures such as saltwater soaks and paring, to topical keratolytics, systemic retinoids or even reconstructive surgery with total excision of the hyperkeratotic skin followed by skin grafting. Here, we present a 22-year-old patient whose father, paternal aunt and grandfather had similar complaints of thickening of skin of hands and feet, hearing impairment and fibrotic band around left fifth toe that can aid in comprehensive understanding of this condition by a dermatological clinician.

CASE REPORT

A 22-year-old female patient born out of a non-consanguineous marriage presented with asymptomatic thickening of skin of hands and feet since the past 20 years and progressed steadily with development of fibrotic band around left fifth toe. She also had decreased hearing bilaterally that progressed to loss of hearing due to which the history was provided by a friend that accompanied her. She had sought medical advice for the same but no clear

diagnosis was made and the management was not satisfactory. Cutaneous examination revealed multiple hyperpigmented, hyperkeratotic plaques present over bilateral palmar aspect of hands and plantar aspect of feet with few hyperpigmented papules coalescing to form plaques over dorsum of hands, feet and knuckles. Pseudoainhum was also present over base of left fifth toe (Figure 1-3).

Figure 1: Multiple hyperpigmented, hyperkeratotic plaques over the dorsum of foot along with pseudo-ainhum over the left fifth toe.

Figure 2: Multiple hyperpigmented, hyperkeratotic papules coalescing to form plaques over the dorsum of hands.

All basic blood investigations were done and found to be normal. Histopathological examination revealed features suggestive of Palmoplantar keratoderma hyperkeratosis of stratum corneum, compact column of parakeratosis, thickening of stratum granulosum. Some acanthosis and papillomatosis. Patient was also referred to the Department of Otorhinolaryngology in view of hearing loss where Rinne's test showed air conduction greater than bone conduction. Further, pure tone audiometry showed moderate to severe bilateral sensory neural hearing loss (Figure 4). Based on the patient's history, clinical presentation, biopsy findings and Pure tone audiometry reports; a diagnosis of Voh Winkel syndrome was established. The patient received treatment with topical retinoids and keratolytics, systemic retinoids were deferred since the patient was to be married in 6 months. Hearing aid was advised for hearing loss.

Figure 3: Plantar aspect of bilateral feet showing keratoderma.

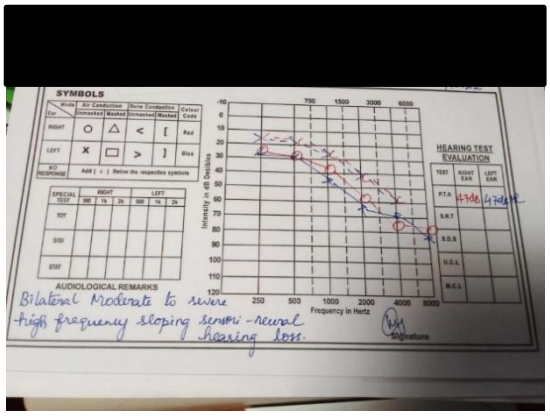

Figure 4: Pure tone audiometry of the patient showing Bilateral Sensory neural hearing loss.

Plastic surgery opinion was also sought for the pseudo ainhum. Genetic study was also advised outside since our institution did not have facilities for the same. Three months after presentation her skin lesions improved. She has since been lost to follow-up.

DISCUSSION

Vohwinkel syndrome is a rare palmoplantar keratoderma which manifests in childhood and becomes more evident in adulthood.[2] It was first described in 1929 by Vohwinkel in a 24-year-old woman who, since age 2 years, had a diffuse honeycombed palmar and plantar keratosis, in addition to distal interphalangeal creases. The constrictions led to autoamputation.[3] The patient's daughter also had similar complaints. Our case had similar features but had not progressed to a level of autoamputation. Vohwinkel syndrome is characterized by a Palmoplantar Keratosis, digital constriction bands leading to pseudoainhum formation and hearing impairment or deafness. It begins within the first few years

of life and appears as a callus-like thickening on the palms and soles and is frequently referred to as having a "honeycomb" pattern.[4] Apart from sensory neural deafness which was seen in our patient; other features like myopathy, spastic paraplegia, mental retardation, acanthosis nigricans, ichthyosiform dermatitis, alopecia, and nail abnormalities are also seen.[5] It has been classified into two variants: an ichthyosis-associated variant and a deafness-associated variant. The variant with ichthyosis, also called Camisa's variant, is associated with the loricrin mutation whereas the one with deafness is associated with Connexin mutation; mainly that of connexin-26. A missense mutation is seen of the *GJB2* gene coding connexin-26, a gap junction protein.[6] Connexins are building blocks of gap junctions that are plasma membrane complexes that facilitate and regulate the passage of small molecules between cells. Several other rare mutations have also been described. Connexins 26, 30, 30.3, 31, and 43 are related to cutaneous diseases associated with multiple organ involvement. Mutations in connexin 26 are linked to Vohwinkel syndrome, keratitis-ichthyosis deafness and hystrix like ichthyosis deafness syndromes, palmoplantar keratoderma with deafness, deafness with Clouston-like phenotype, and Bart-Pumphrey syndrome thereby making them close differentials.[7] Another mutation found is an insertional mutation of the loricrin gene on epidermal differentiation complex on 1q21. This protein plays an integral role in the formation of the cornified cell envelope. An ichthyotic variant has also been found with a 730insG mutation.[8] Halting the progression of pseudo-ainhum before it leads to auto amputation of the affected toes/fingers and early detection of hearing impairment so as to facilitate hearing aids for the patients; decreasing the social stigma and isolation faced by them due to the disabilities arising from the disorder is very important. Early diagnosis and treatment thus become imperative.

CONCLUSION

We have herein presented a case of Voh Winkel syndrome deafness associated variant who presented with palmoplantar hyperkeratosis as well as hearing impairment and pseudo-ainhum of toe. There was family history of similar lesions which followed an autosomal dominant pattern. We have emphasized the rarity of this entity and the need for proper diagnosis and synergistic management of the condition by the dermatologist along with the otorhinolaryngology and surgical departments for proper monitoring and care of hearing loss and pseudo-ainhum.

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