VOHWINKEL'S DISEASE

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A case of mutilating keratoderma of Vohwinkel associated with ichthyosis, high frequency hearing loss, mental retardation and bony changes seen in a 14-year-old boy in a non-familial pattern is reported.

Key words : Vohwinkel's disease, High frequency hearing loss, Ichthyosis, Mental retardation, Bone changes.

Palmo-plantar keratodermas are a group of hereditary and acquired disorders characterised by diffuse or discrete as well as focal thickening of the palms and soles. Genetically inherited palmo-plantar keratodermas are of several types in which the dominantly inherited, keratoderma hereditarium mutilans of Vohwinkel is a rarity. It is characterised by diffuse, honeycombed, keratotic thickening of the palms and soles, starfish like keratosis of the dorsal aspects of the hands and feet, linear keratosis of the elbows and knees and annular fibrous constricting bands leading to progressive autoamputation of the digits[1]. A few variants of this disease have been described as isolated case reports with possible recessive mode of inheritance[2]. According to Gibbs and Frank[1], very few cases of this disorder have been described in the literature to define the exact mode of inheritance and the pattern of the disease. As far as ascertained, only 4 cases have been reported from India[3-6].

Case Report

A 14-year-old boy had thickening of the palms and soles and flexural parts of the body of 8 years duration. He was the second child of consanguineous parents and was apparently normal at birth. The mile stones had been delayed. When the child was 6-year-old, the parents noted thickening of the skin first over the palms and soles and later over the dorsal aspects of the hands, feet, knees, elbows and body-folds namely gluteal, perianal, inguinal, popliteal and axillary regions. There was no history of any systemic illness. None of the family members were affected by a similar process.

Thickening of the palms and soles was diffuse and honeycombed with fissuring (Fig. 1).

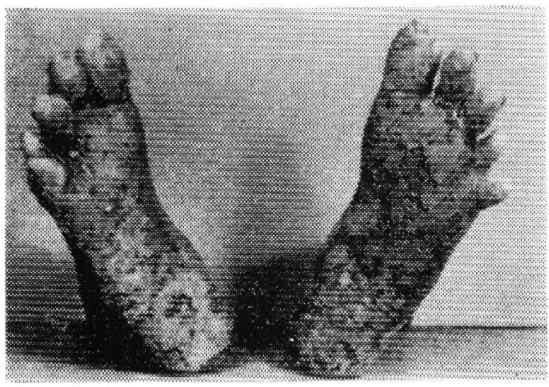

Fig. 1. Gross thickening and fissuring of the skin over soles. Also note the malalignment of the toes.

Dorsa of the hands, elbows, knees, feet, perianal, gluteal region and groins also showed keratotic thickening of the skin with accentuated markings.

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Starfish-like keratosis were seen in the popliteal fossae (Fig. 2). Constricting bands were present

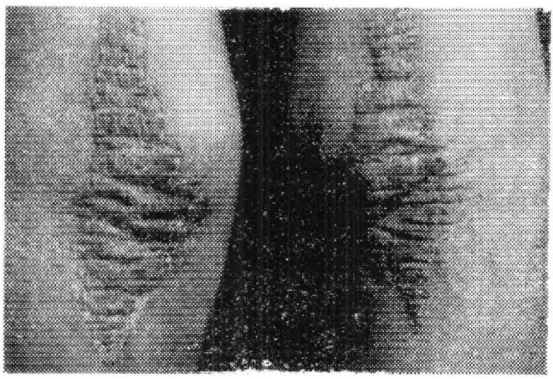

Fig. 2. Starfish-like keratosis in the popliteal fossae.

near the interphalangeal joints of the medial three fingers of the right hand and also to a minimal extent over the middle and ring fingers of the left hand (Fig. 3). Distal to the constric-

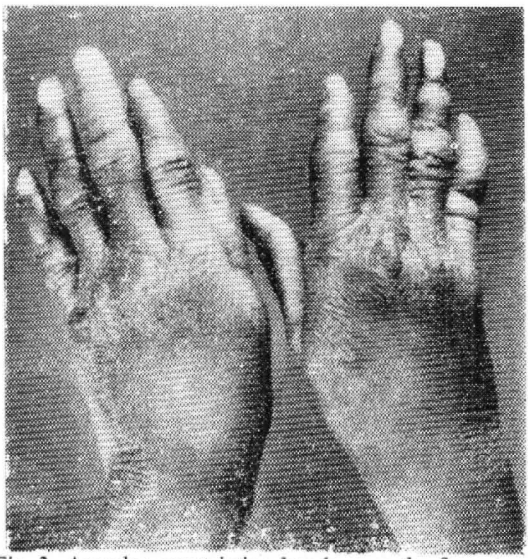

Fig. 3. Annular constricting bands over the fingers and keratotic thickening of the skin over the dorsa of hands.

tion, the fingers were bulbous with tapering of tips and increased curving of the nails. Ichthyosis was present over both the legs. Toes were not aligned properly, the 4th toe of the left foot being pushed dorsally (Fig. 1). Subungual hyperkeratosis was observed in the finger nails. Toe nails showed gross dystrophic changes. Hair, teeth, oral mucosa and eyes were normal. In addition, he had moderate degree of mental retardation and defective audition in both the ears.

Routine investigations on blood, stools and urine were normal. VDRL was non-reactive. Serum proteins were within normal limits. Skin biopsies from the right palm and the popliteal fossa showed massive hyperkeratosis, hypergranulosis, acanthosis and a mild inflammatory infiltrate in the upper dermis consistent with the diagnosis of palmo-plantar keratoderma. X-ray of the hands showed constricting bands near the interphalangeal joints of the medial three fingers of the right hand and also to a minimal extent over the middle and the ring fingers of the left hand. Soft tissue swelling distal to constricting bands and demineralisation of all the metacarpals and phalanges were also evident. X-rays of both the feet showed malalignment of the metatarsals and phalanges with osteolytic changes of the distal phalanges and minimal demineralisation of metatarsals and phalanges. Audiogram revealed bilateral high frequency hearing loss of neural type.

Comments

Hyde and Montgomery[7] first reported patients with palmar and plantar hyperkeratosis associated with constriction of the digits. However, Vohwinkel[8] in 1925 gave a detailed account of this association in a 24-year-old female under the name keratoderma hereditarium mutilans. Later, sporadic case reports of this interesting disorder have appeared in the literature from time to time with several atypical features from different parts of the world[3-13].

No doubt, the patient reported here is a case of Vohwinkel's disease because he had all the characteristic features of the disease namely honeycombed hyperkeratosis of the palms and soles, constricting bands, starfish like keratosis and linear keratotic lesions. However, we became more interested when we observed certain unusual findings like non-familial occurrence, ichthyosis, bilateral high frequency hearing loss of neural type, mental retardation and bone changes. Further, the distribution of lesions was relatively extensive and only a few cases have been reported with keratotic lesions over the perianal, gluteal regions and groins.

Vohwinkel's disease is inherited through an autosomal dominant gene although in its variants an autosomal recessive mode of inheritance has been suggested[2]. Referring to the reported cases of keratoderma hereditarium mutilans of Vohwinkel in negroes, Gibbs and Frank[1] noticed absence of familial incidence similar to the case reported here. Nevertheless, history of consanguinity, non-familial pattern and association of several unusual findings observed in our patient suggest either an autosomal recessive mode of inheritance, or a mutation in the gene.

Association of ichthyosis in Vohwinkel's disease has been noted by some workers [3, 5, 6, 10, 11] particularly the cases reported from India. Haldar et al[3] described a case of Vohwinkel's disease associated with severe ichthyosis and acanthosis nigricans. Reddy and Gupta[5] have observed non-bullous type of ichthyosiform erythroderma in Vohwinkel's disease. However, in our patient the ichthyosis was minimal and confined only to the extensor aspect of the legs.

The occurrence of high frequency hearing loss in Vohwinkel's disease was also reported by Gibbs and Frank[1]. Similarly, Drummond[12] reported a deaf-mute having Vohwinkel's disease.

Mental retardation in Vohwinkel's disease as noted in the present case has not been recorded so far in the literature. This association may reflect a common developmental anomaly of the structures derived from neuroectoderm. In fact such a feature has been observed in other forms of keratodermas[2] such as Griether's syndrome, Richner-Hanhart syndrome, Darier's disease, etc.

In the common varieties of palmo-plantar keratoderma, roentgenographic findings do not contribute to the diagnosis, but in the mutilating form, characteristic features of this entity are seen as described by Presly and Bonta[13]. In the initial stage of the disease they observed demineralisation of bones of the hands and feet with resorption of tufts of all the distal phalanges in addition to constriction and soft tissue swellings. Similar findings have been noted in our case. In the later part of the disease there may be total destruction of bones of the hands and feet with fusion of the carpal bones, thinning of the distal aspects of the radius and ulna, periosteal reaction and soft tissue hypertrophy with nodular, warty, infected, macerated, keratotic masses. These late features are considered characteristic of this disease and may help to differentiate from other disorders like scleroderma, rheumatoid arthritis, TAP, syringomyelia, leprosy, yaws, syphilis, frostbite, ergot poisoning, etc., where ainhum like constrictions with resorption of terminal phalanges may be seen[13].

Features like acanthosis nigricans, pseudopelade type of alopecia and plantar bulla as observed by other workers[3, 4] were not seen in our patient.

References

1. Gibbs RC and Frank SB: Keratoma hereditaria mutilans, Arch Derm, 1966;94:619-625.
2. Ebling FJG and Rook A: Disorders of keratinisation, In Text Book of Dermatology, 3rd Ed, edited by

Rook A et al, Blackwell Scientific Publications, Oxford and Edinburgh, 1979; p 1303.

3. Haldar B: Mutilating keratoderma, Ind J Dermatol, 1972;17:95.
4. Kapoor TR and Singh OP: Vohwinkel syndrome, Ind J Dermatol, 1977;43:214.
5. Reddy BSN and Gupta SK: Mutilating keratoderma of Vohwinkel, Inter J Dermatol 1983, 22:530:533.
6. Chaudhuri AK and Haldar B: Keratoderma hereditaria mutilans with acanthosis nigricans, Ind J Dermtol Venereol Leprol, 1980; 46:299-304.
7. Hyde JN and Montgomery FH: A Practical Treatise of Disease of the Skin, Harry R Kimpton, Medical Publisher and Bookseller, London, 1905; p 609.
8. Vohwinkel KH: Keratoderma hereditarium mutilans, Arch Dermatol Syphilol, 1929;158:354.
9. Ruiz-Maldonado R and Lozano Ferral N: Quoted by Ebling FJG and Rook A (2).
10. Wirz F: Keratoma hereditarium mutilans, Arch Dermatol Syphilol, 1930;159:311.
11. Grschebin S: Quoted by Gibbs and Frank (1).
12. Drummond M: A case of unusual skin lesions, Irish J Med Sci; Feb 1939, p 85.
13. Presly NL and Bonta FJ: The roentgen appearance of mutilating palmoplantar keratosis, Amer J Roentgen, 1961;86:944.