KERATODERMA HEREDITARIA MUTILANS WITH ACANTHOSIS NIGRICANS (VOHWINKEL DISEASE)
(A case report with a brief review)

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Summary

A case of keratoderma hereditaria mutilans in a 24 years old lady with associated marked ichthyosis and acanthosis nigricans is reported. A short literature review is given.

Keratoderma hereditaria mutilans is a rare hereditary disease which is transmitted by an autosomal dominant gene. It was first described by Vohwinkel in 1929[1]. The disease is characterised by diffuse palmoplantar keratoderma, keratosis on limbs and fibrous constriction bands around the digits occurring in early childhood. To the best of our knowledge its association with acanthosis nigricans has not been described in the literature so far. In this report a case of keratoderma hereditaria mutilans with severe ichthyosis vulgaris and acanthosis nigricans is presented.

Case Report

A 24 years old hindu female, a resident of Calcutta, being frustrated of her malady reported to the hospital on 7-12-78 with the complaints of thickening of the skin of palms and soles present since birth and constrictions of most of the toes and fingers which developed gradually over 16 years. The pain and disfiguration of the left little finger was so much that the finger had to be amputated in 1969 (Fig. 1). She complained of pain in the right little finger for 6 months. She had 3 brothers and 4 sisters. Her immediate elder brother who was the only affected sibling was reported to have thickened skin of palms and soles as well as scaliness on the body.

On clinical examination marked diffuse honeycombed hyperkeratosis of palms (Fig. 1) and severe keratosis leading to cracks of soles (Fig. 2) were evident. Keratosis also involved entire palmar surface of the fingers and proximal parts of the plantar surface of the toes. Keratosis was gross enough to appear verrucous. It was linear in distribution on the dorsa of feet (Fig 3). Creases on the dorsa of hands were prominent on account of the marked thickening (Fig. 4). Well circumscribed hyperpigmented plaques simulating lesions of acanthosis nigricans were noted symmetrically over axillae, antecubital (Fig. 5) and popliteal fossae (Fig 6). Ainhum like constrictions of toes were noted particularly on the 3rd, 4th and 5th toes bilaterally (Fig. 3). The 3rd and 4th toes on both sides

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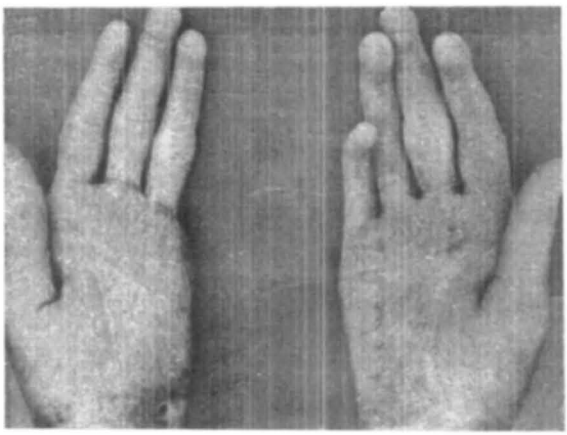

Fig. 1 Honeycombed hyperkeratosis of palms, amputated left little finger and bulbous swelling of the terminal end of the fingers.

were dorsally displaced and the 5th toes were grossly atrophic (Fig. 3). Constrictions were seen over all the fingers near the proximal part of distal phalanges making the tips of the fingers bulbous in appearance (Fig. 1). Nails were found to be moderately dystrophic. Ichthyotic changes were present over the entire surface of the body but were especially prominent over legs (Fig 2) where scales were coarser and over the back where the skin simulated lizard skin (Fig. 7). Scalp showed diffuse hair loss and no other abnormality.

General systemic examination revealed no significant finding. No abnormality was detected in CNS, ear, nose or throat.

Routine investigations were within normal limits. Serum β-carotene was 75 μg% and vit-A was 81 μg%. Wassermann and Kahn tests were negative. ECG was normal. X-ray of hands and feet showed mild decalcification involving the phalanges. Biopsies were done from left leg, left cubital fossa and left palm. These were reported as Ichthyosis vulgaris (Fig 8), acanthosis nigricans (Fig 9) and severe hyperkeratosis (Fig 10) respectively.

Comments

Though Hyde and Montgomery[2] described this type of cases as early as 1905, the detailed case report of Vohwinkel[1] of a 24 years old white woman having diffuse palmoplantar hyperkeratosis and annular constriction of digits created wide spread interest. The most severe form of the disease was reported by Presseby and Bonte[3] who referred to this syndrome as "progressive destructions of hands and feet". Gibbs and Frank[4] reviewed this disease in great detail. In India, Haldar[5] and Kapoor et al[6] reported similar cases.

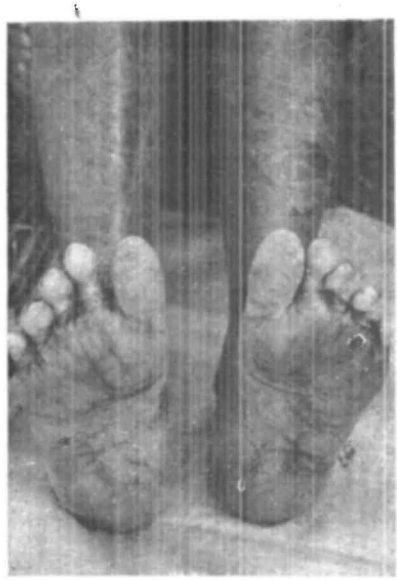

Fig. 2 Hyperkeratosis leading to cracks on the soles and severe ichthyotic changes over the legs.

In the case reported here there was classical honeycombed keratosis of the palms (Fig. 1), keratosis leading to fissures on soles (Fig. 2) and warty keratosis with linear configuration on dorsum of the hands and feet (Fig. 3). Pain in the right little finger was probably of

Fig. 3 Ainhum like lesions on toes, 3rd and 4th toes dorsally displaced and 5th toe grossly atrophied. Hyperkeratosis with linear configuration.

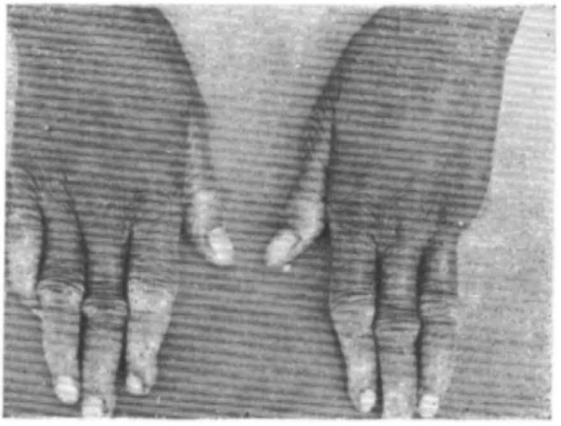

Fig. 4 Hyperkeratosis of the dorsum of the hands.

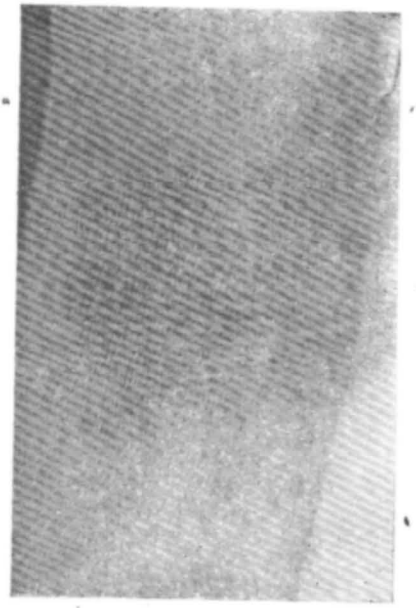

Fig. 5 Acanthosis nigricans type of lesion over the anticubital fossa.

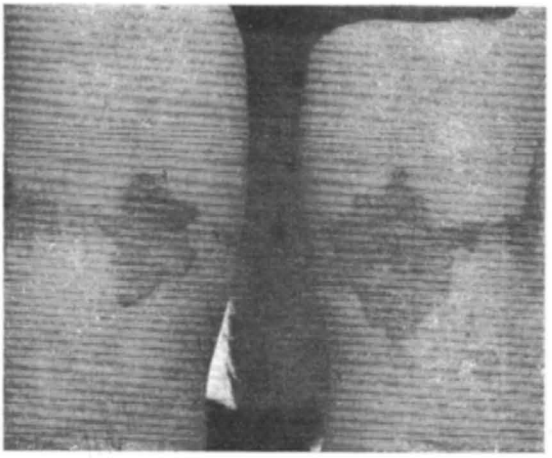

Fig. 6 Acanthosis nigricans type of lesion over the popliteal fossae and ichthyosis over the back of the legs.

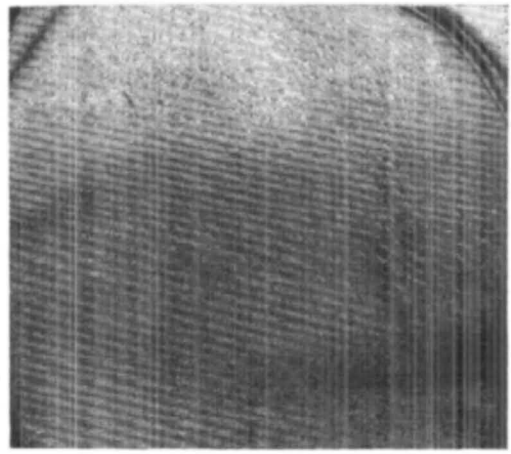

Fig. 7 Ichthyotic changes simulating lizard skin over the back of the trunk.

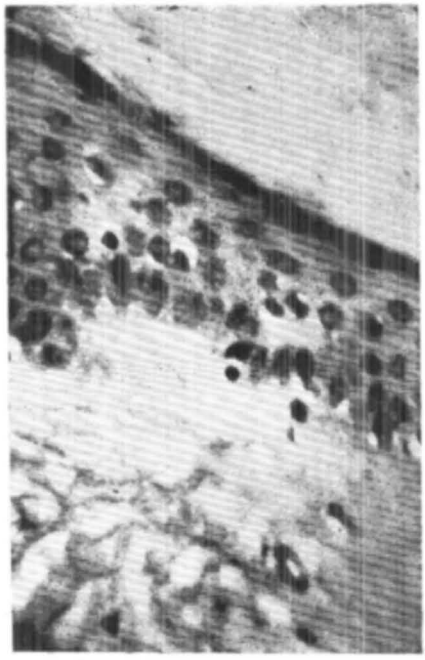

Fig. 8 Showing hyperkeratosis with less prominent granular layer (HE stain×400).

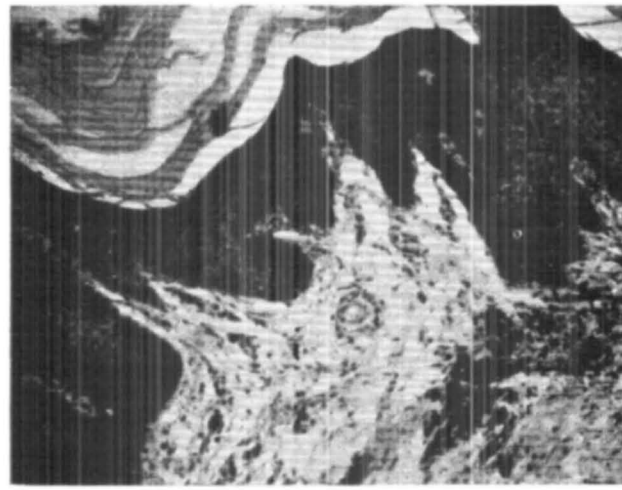

Fig. 9 Showing features of acanthosis nigricans (HE stain × 40)

ischaemic origin caused by constriction by the bands of the digital vessels. Marked ichthyosis (Fig 2 and Fig 7) was a prominent feature in our case as was described by Wirz[7] and Grschebin[8]. There was diffuse hair loss of the scalp similar to that reported by Gibbs and Frank[4] but there was no deafness. In our case both the 3rd and the 4th toes were dorsally displaced (Fig. 3). Gibbs and Frank[4] had reported such involvement on the 4th toe only. A significant feature in our case was the presence of acanthosis nigricans type of lesions in axillae, antecubital fossae (Fig. 5) and popliteal fossae (Fig. 6). The association of acanthosis nigricans with keratoderma hereditaria mutilans can be explained by the fact that true benign acanthosis nigricans is determined by an irregularly dominant gene[9] and very often there is co-existence of other genetic abnormalities implicating disorder of keratinisation. The elder brother of the patient who was found to have moderate degree of palmoplantar hyperkeratosis and ichthyosis vulgaris indicates the genetic etiology in this case. Vit-A deficiency was thought in the past to be a causative factor in many keratinising disorders[10] and it had also been used as a therapeutic agent mostly on an empirical basis[11]. Serum β-Carotene and Vit-A levels in our case were found to be within normal limits (50-100 μg%). Mutilating acropathy as has been reported by Phillips and Murray[12] was ruled out in this case as there was no sensory loss or history of painless trauma. Mild decalcification of phalanges was due probaly to disuse.

Local applications of salicylic acid 20%, urea 30%, retinoic acid 0.3%[13] and administration of Vit-A orally (150,000 units daily) and even by injection (600,000 units biweekly) for adequate period given empirically failed to improve the condition permanently and the therapeutic problem remains still unsolved.

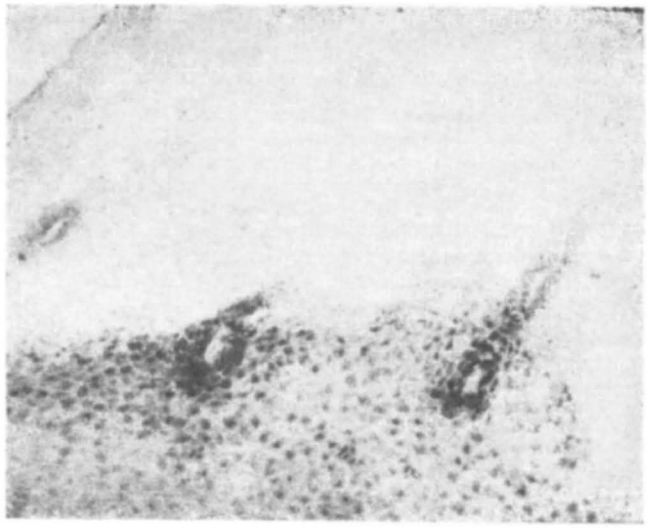

Fig. 10 Showing severe degree of hyperkeratosis (HE stain ×40)

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