Poikiloderma a varied presentation - Huriez syndrome

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ABSTRACT

Huriez syndrome is a rare autosomal dominant genodermatosis characterized by the triad of congenital scleroatrophy of the distal extremities, palmoplantar keratoderma (PPK) and hypoplastic nails. We report the case of a 25 year old male, with nonfamilial Huriez syndrome.

Key words: Nonfamilial, poikiloderma, squamous cell carcinoma

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INTRODUCTION

Huriez syndrome is a rare autosomal dominant genodermatosis first reported in two families from Northern France by Huriez *et al*. in 1963.[1,2] It is characterized by the triad of congenital scleroatrophy of the distal extremities, palmoplantar keratoderma and hypoplastic nails.[1-3] The importance of early diagnosis lies in the fact that squamous cell carcinoma (SCC) arises in the scleroatrophic area in around 15% of the affected individuals, which is characterized by its early onset mostly in the third to fourth decades of life, more aggressive behavior and metastasis.[1,3-5]

CASE REPORT

A 25-year-old male, born out of a nonconsanguineous marriage presented to the outpatient department of our hospital with complaints of pigmentation of skin, painful fissures of the hands and feet, with hypohidrosis since birth. There was tightening of skin of the distal extremities since 15 years. His family members were unaffected. There was no history of Raynaud's phenomenon, photosensitivity, growth or mental retardation, exposure to chemicals or any drugs that would be responsible for his sclerotic skin. On examination, both palms and soles had diffuse scleroatrophy with fissuring and hyperpigmentation [Figure 1]. The skin had reduced pinchability on the distal extremities with generalized atrophy, dyschromia, telangiectasia, and xerosis [Figure 2]. There was ulnar deviation at the left wrist due to contracture along the ulnar border of the palm. A raised tender nodular lesion measuring around 1 cm × 1.5 cm was present on the left thenar eminence; there was no ulceration or bleeding from the lesion [Figure 3].

Nail changes were characteristic with minimal growth of the third and fifth toenail bilaterally and absence of nail of the second right toe [Figure 4]. All the finger and toenails had longitudinal ridging and discoloration. Routine blood investigations were within normal limits. Biopsy was performed from three different sites viz., the left thenar nodule, right palm and dorsal aspect of proximal third of the left forearm.

Excisional biopsy of the thenar nodule revealed mild hyperkeratosis with dysplasia of the basal keratinocytes and formation of small buds extending into the papillary dermis, features suggestive of actinic keratosis, which is a precursor of SCC. Biopsy of palms and soles revealed massive hyperkeratosis and acanthosis and that of the sclerosed skin on the left forearm showed acanthosis with an accentuated granular layer, orthohyperkeratosis with sparse mononuclear cell infiltrate [Figure 5]. Immunohistochemistry analysis showed a marked reduction in CD1a, Lag+ and S-100 epidermal Langerhans cells [Figures 6 and 7], which is a characteristic finding of Huriez syndrome.[6] Based on the history, clinical findings and histopathological features, we made a diagnosis of nonfamilial Huriez syndrome.

The margins of the excised nodule were free of any precursor neoplastic changes; thus, it was

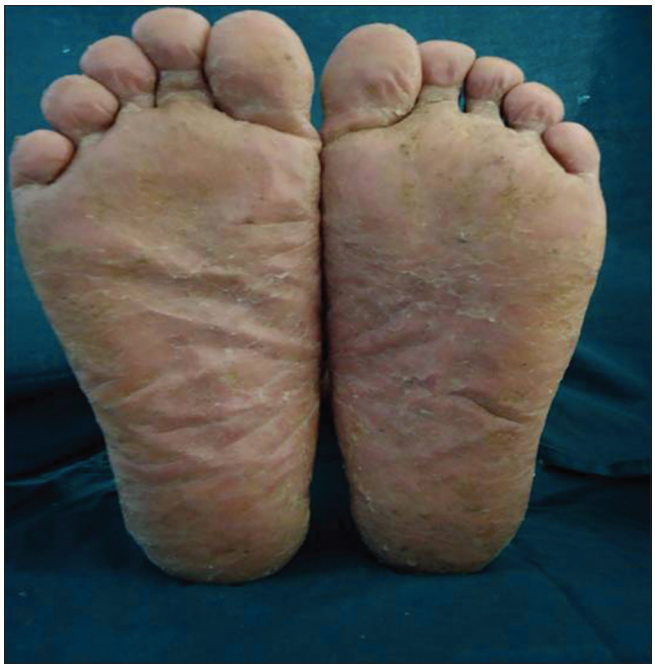

Figure 1: Plantar keratoderma

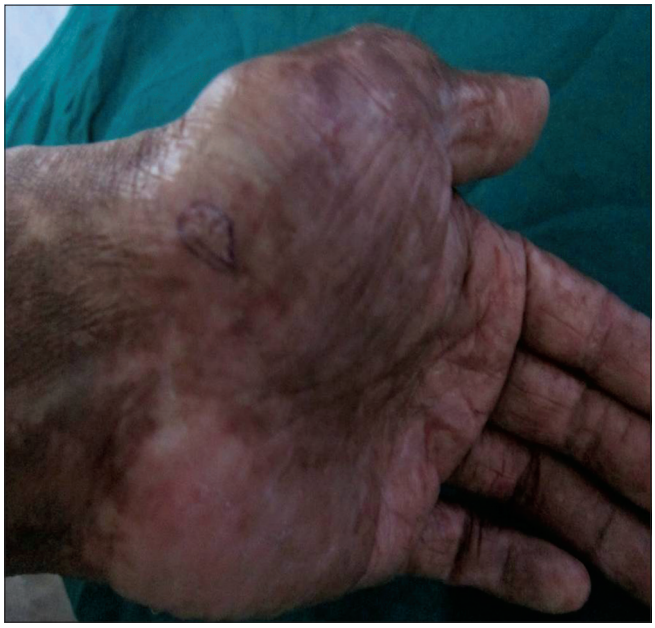

Figure 3: Nodule over the palmar aspect of left hand (which later showed actinic keratosis)

deemed adequate. The patient was given topical tazarotene and emollients and advised a six monthly follow-up. He has been on regular follow-up for more than three years now, and there is no other nodular eruption or growth suggestive of SCC till date.

DISCUSSION

Huriez syndrome, also referred to as sclerotylosis was first described by Huriez *et al*. as an autosomal dominant genodermatosis.[1-5] In addition to its occurrence in French

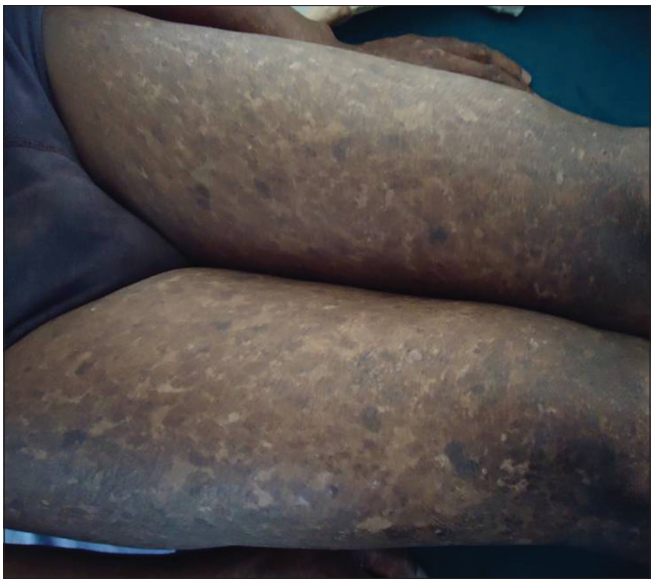

Figure 2: Genralised atrophy, dyschromia and xerosis; the hallmark features of poikiloderma

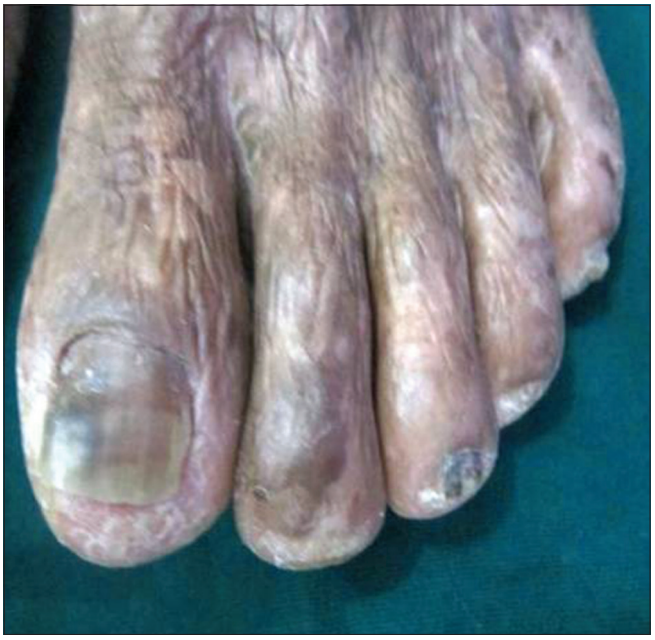

Figure 4: Hypoplastic nails with longitudinal ridging

patients, it has also been reported from India, Japan, Tunisia, Germany, and Italy.[3,5] There have been only few reports about its sporadic occurrence.[3,5,7,8] Ours is also a nonfamilial case of Huriez syndrome. There was no history of consanguinity, and none of the patient's parents or siblings were affected.

There is a great individual variation in the clinical manifestations of Huriez syndrome, which should be differentiated from other genodermatosis.

There is a clinical similarity with dyskeratosis congenita, which is a rare multisystem disorder with both autosomal

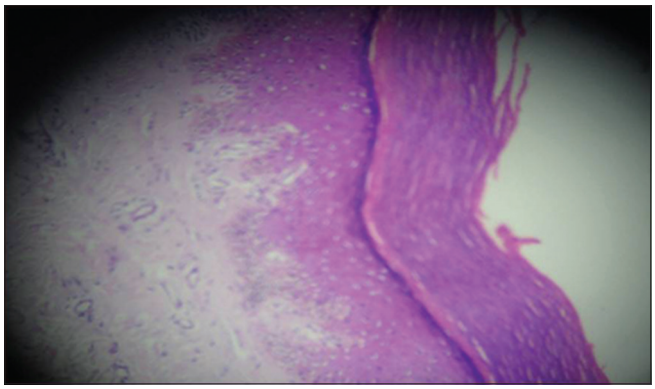

Figure 5: H and E, ×40 showing epidermal hyperkeratosis and acanthosis and dermis showing a sparse mononuclear cell infiltrate

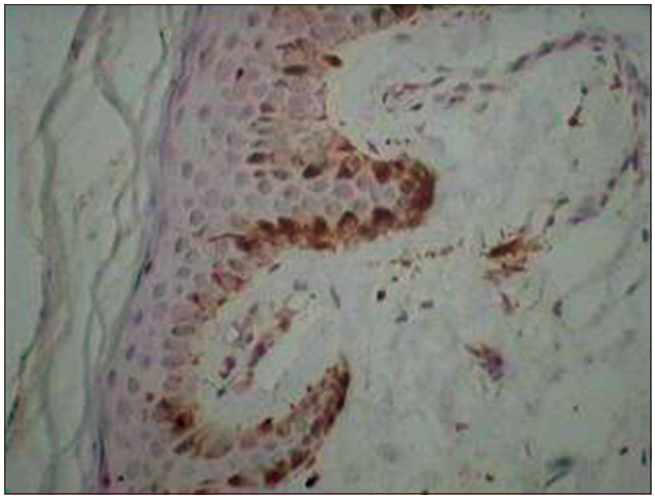

Figure 6: Immunohistochemical analysis showed a marked reduction in the number of S100+ epidermal Langerhans cells

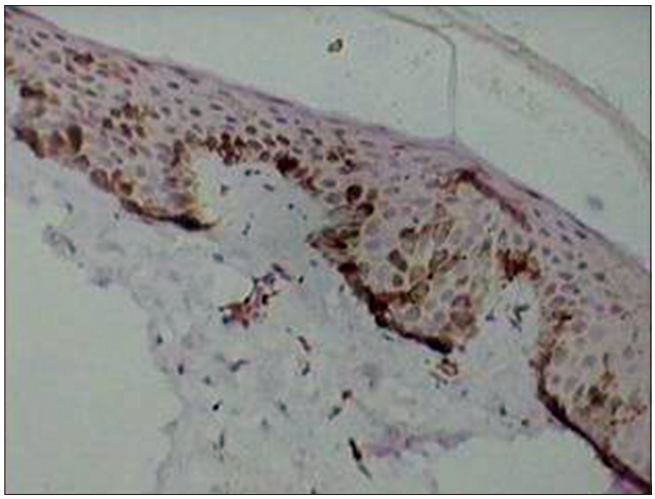

Figure 7: Immunohistochemical analysis showing marked reduction in CD1a+ epidermal Langerhans cells

dominant and recessive modes of inheritance, characterized by the triad of nail dystrophy, poikiloderma, and mucosal leukoplakia. Histopathology of the skin is nonspecific and includes epidermal atrophy, increased vascularity of the dermis with a sparse lymphocytic infiltrate, and pigment-laden macrophages.[9]

Rothmund–Thomson syndrome is another rare disorder characterized by photosensitivity, poikiloderma, cataract, and growth retardation. A common ocular feature seen in 73% of patients is the presence of bilateral, rapid onset (2-3 months), anterior subcapsular cataracts. SCC may arise from the involved skin over the areas of hyperkeratosis mostly in adult patients.[9]

Pigmented xerodermoid, a rare genodermatosis resembling xeroderma pigmentosum (XP), presents mostly in the first and second decades of life and is outlined by features of dryness, pigmentation and parchment-like skin, with freckle-like hyperpigmented macules on the sun exposed areas. Premalignant actinic keratoses may develop at an early age. Patients with XP under 20 years of age have a greater risk of cutaneous basal cell or SCC or malignant melanoma.[9]

The exact identification and characterization of the gene causing Huriez syndrome still remains an enigma. Deminatti *et al*. proposed probable linkage of the gene involved in Huriez syndrome with that of the MNSs blood group, which was later mapped to chromosome 4 (4q28-31).[10] Lee *et al*. localized the position of the gene to be 4q23.[4] Watanabe *et al*., in their study found positive staining of p53 in atypical keratinocytes in their patients and suggested that p53 mutations may be responsible for development of actinic keratoses and SCC in cases of Huriez syndrome.[5] We did not perform any genetic studies in our patient; this is one limitation of our study, but the diagnosis in Huriez syndrome was based on clinical findings, immunohistochemistry characterized by a marked reduction in CD1a, Lag+ and S-100 epidermal Langerhans cells.[6]

A multidisciplinary approach with dermatologists, geneticists and plastic surgeons is vital in addition to close medical supervision because of high cancer risks.[4] However, a close surveillance for malignancy and aggressive treatment may be necessitated. In a few cases, topical retinoids have been given.[3]

Affected individuals carry a >100-fold higher risk for the development of aggressive SCC of the skin.[11] In our patient the excision biopsy of the raised tender nodule over the left thenar eminence revealed actinic keratosis, which is a precursor of SCC. We want to emphasize the need for a high index of suspicion, early diagnosis, timely intervention and regular follow-up in such cases so as to avoid the dreaded complication of SCC.

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