

Case Report

Huriez syndrome: a rare palmoplantar keratoderma

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ABSTRACT

The Huriez syndrome is a rare autosomal dominant transgradient palmoplantar keratoderma which is characterized by scleroatrophy of the fingers, nail changes and squamous cell carcinomas in affected skin. Herein, we present a non-familial case of very rare plamoplantar keratoderma with scleroatrophy - the Huriez syndrome in a 45 year old female patient.

Keywords: Palmoplantar keratoderma, Transgradient, Autosomal dominant

INTRODUCTION

The palmoplantar keratodermas (PPK) are a heterogeneous group of acquired or hereditary disorders defined by excessive thickening of skin of palms and soles.¹ The genetically determined PPKs may form a part of ectodermal syndromes or associated with other systemic anomalies like cardiomyopathy, impaired hearing, neuropathy, neuro-developmental defects and various carcinomas.² The gene defects in the palmoplantar keratodermas lies in the genes encoding intra cellular structural proteins, desmosomal proteins, gap junction components and enzymes.

Huriez syndrome is a very rare autosomal dominant syndrome characterized by diffuse keratoderma, red atrophic skin on the dorsum of the hands and feet, sclerodactyly, nail abnormalities and high incidence of squamous cell carcinomas, first reported by Huriez et al in two families from Northern France.³⁻⁵ The squamous cell carcinoma of affected skin appears in the third to fourth decade and which is unusually prone to metastasis.

The chromosome 4q23 has been implicated but the underlying gene defect is still unknown.⁶

CASE REPORT

A 45 year old female, born out of a non-consanguineous marriage presented with complaints of diffuse non erythematous thickening of skin of the palms and soles with sparing of central area of palm and instep area of feet since the age of five years. The lesion started from first inter digital space of thumb and index finger of both hands and progressed to involve palms, knuckles and soles by the age of ten years associated with episodes of peeling and fissuring of skin. Thickening leads to fissuring and pain with progressive binding down, ultimately leading to auto amputation of the right fifth toe at thirteen years of age. There was progressive binding down of the affected skin of the fingers with hyper curvature of nails. No history of blistering and oozing from the lesions or sweating abnormality. The knees, ankles or elbow and mucosal areas were not involved. There was no history of Raynaud's phenomenon, photosensitivity, mental retardation and exposure to

chemicals or drugs that could cause binding down of skin. No history suggestive of cardiovascular, respiratory, gastrointestinal, musculoskeletal and auditory system abnormality. All the family members, parents and siblings were normal.

The General physical and systemic examination was normal.

On local examination there was diffuse hyperkeratosis with fissuring involving bilateral hands and feet with sparing of central palm and in step area of feet and the lesions were not well defined at some sites. The fifth toe of right foot was auto amputated at the fifth metatarsal joint (Figure 1.a, 1b, 1c, 1d). The dermatoglyphics were absent over the diffuse thickening of both hands (Figure 2). The nails had hyper curvature, longitudinal ridging and binding down of overlying skin.



Figure 1a: Keratoderma over bilateral hands.



Figure 1b: Palmoplantar keratoderma with sparing of central palm.

No abnormality on the routine blood investigations and radiological examination. Excision biopsy was done and histopathological examination revealed hyperkeratosis with irregular parakeratosis and a moderate perivascular infiltrate. Based on the history and clinical findings, we made a diagnosis of Huriez syndrome with differential diagnosis of Papillon-Lefevre syndrome. The patient showed marked improvement to topical emollients and oral retinoids.



Figure 1c: Keratoderma over lateral aspect of dorsum of left foot.



Figure 1d: Transgradient keratoderma with amputated right fifth toe.



Figure 2: Ink imprints of bilateral hands showing loss of dermatoglyphics.

DISCUSSION

The clinical patterns of keratodermas are grouped as diffuse, focal, striate and transgradient but no absolute distinction between them. The clinical presentation and manifestations of a palmoplantar keratoderma may vary with site, age, gender or occupation.¹ Transgradient keratoderma extends beyond palmoplantar skin in the form of corn or callosity on the pressure points on the fingers, knuckles or other body sites. The Papillon-Lefevre syndrome is an autosomal recessive keratoderma with extracutaneous features characterized by redness and thickening of palms and soles associated with periodontitis and frequent pyogenic skin infections. This

syndrome is caused by homozygous mutations in the gene encoding the lysosomal protease cathepsin C.⁷

Huriez syndrome and Papillon-Lefevre syndrome are transgradient PPKs closely resembling to our patient, but predominant feature of PPKs with scleroatrophy, nail changes, non-erythematous lesions and lack of mucosal and systemic involvement tilt the clinical diagnosis towards the very rare type of autosomal dominant inherited Huriez syndrome. The Huriez syndrome should be differentiated from other genodermatosis like Rothmund-Thomson syndrome, dyskeratosis congenita, xeroderma pigmentosum as it can present in varied clinical manifestations.

CONCLUSION

The palmoplantar hyperkeratosis can lead to minor inconvenience to major functional and social disability. The psychosocial impact of such rare disorder will definitely cause low self-esteem and decrease the quality of life with advancing disease, so crux lies in early recognition and counseling for proper care with liberal use of emollients and oral retinoids for severe disease

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