ANNALI ITALIANI DI CHIRURGIA

Digital Edition
e-publish on-line
ISSN 2239-253X

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A case report

Ann Ital Chir, Digital Edition 2020, 9
pii: S2239253X20032454 - Epub, May 11
free reading: www.annitalchir.com

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Huriez syndrome associated with basal cell carcinoma: A case report

Huriez syndrome is a rare cancer-prone genodermatosis confined to the hands and feet connects with an increase in squamous cell carcinoma on affected skin. Its diagnosis is complex due to not well defined symptoms and since only few cases are described in literature. The differential diagnoses are many and the treatment is focused only on symptoms control and tumours eradication.
Our case report is highly interesting because add new knowledge about this disease describing a new important feature of the syndrome. For the first time in literature we describe the arising of basal cell carcinoma from affected skin.

KEY WORDS: Tumors, Basal cell carcinoma, Huriez syndrome, Palmoplantar keratoderma

Introduction

Huriez syndrome, an autosomal dominant genodermatosis also referred as "sclerotylosis", was firstly described in two large families from Northern France in 1969 by Huriez et al. [1]. The syndrome is a rare disease, classified either as a disorder of the connective tissue or as a keratoderma. It is characterized by congenital scleroatrophy of the extremities, palmoplantar keratoderma and hypoplastic nail changes [2].
A peculiar feature of this syndrome is the development of squamous cell carcinomas (SCC) in the atrophic skin of palms and soles [3].
We describe the first reported case of a patient affected by Huriez syndrome developing basal cell carcinoma on affected skin of the hand.

Pervenuto in Redazione Gennaio 2020. Accettato per la pubblicazione Marzo 2020
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Case Report

In 2010, a 79-year-old woman observed an abnormal and dystrophic toenail growth associated with severe impairment of movements. At that time, she presented at our Plastic Surgery Department where she underwent topic treatment and was referred to dermatology department.
In 2014, after the worsening of her clinical symptoms a diagnosis of Huriez syndrome was made by the Department of Dermatology.
Then at the physical examination, she showed a diffuse thickening of palms and soles, associated with thin dorsum skin both of hands and feet. Moreover she showed hypohidrosis, hyperpigmentation, sclerotylosis and scleroatrophy of palms and soles, associated with recurrent fissuring and superficial chronic painful erosions.
Furthermore the hands were characterized by sclerodactyly with a sclerodermiform-like onset leading to cicatricial retraction of the phalangeal joints, with a diffuse contraction of the hand and a diffuse fingers ankylosis. The dystrophic nails showed longitudinal ridging and aplasia, enclosed in the dermis. The keratoderma was

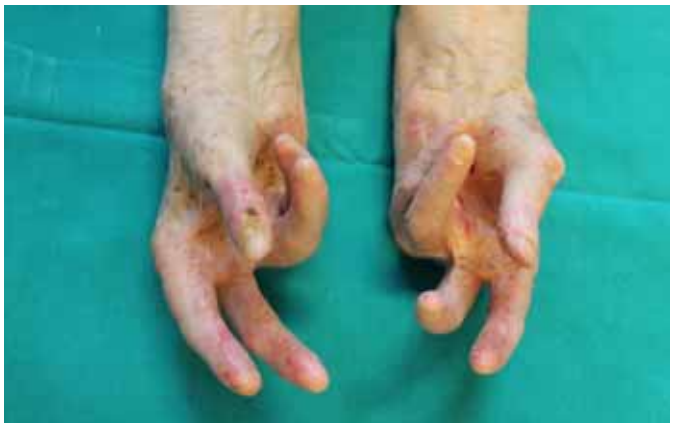

Fig. 1.

Fig. 2.

more uniform and radiated from the palm along the fingers in the hands in comparison with the soles where it was not uniform accentuated over pressure sites (Figs. 1, 2).
X-rays were negative for pathological joint changes.
During standard dermatological follow-up, a persistent ulcerative lesion of the first finger of the right hand was noticed. It was treated conservatively with aureomycin ointment once a day. Topic corticosteroids were prescribed due to diffuse dermatitis of the upper and lower limbs.
Because of the increased risk of developing SCC in Huriez Syndrome, in 2015 for concomitant rheumatoid arthritis she started methotrexate treatment in place of systemic steroids.
Due to painful non-healing ulcers of palms and soles, in 2017 the patient underwent hyperbaric therapy and antalgic therapy.

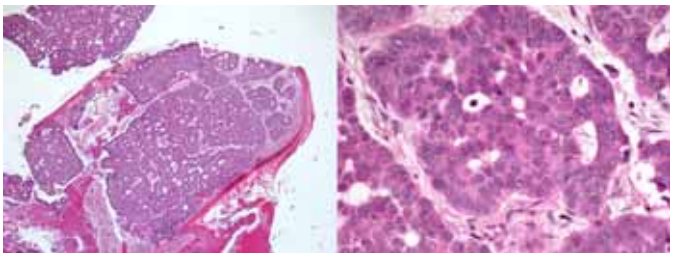

Fig. 3.

At the end of 2017 she presented at the dermatology outpatient clinic complaining of an indolent, nodular lesion of the right thumb. Clinically there was high suspicious of basal cell carcinoma. After discussion with the patient a conservative approach with imiquimod was started and complete remission was obtained. Nevertheless, at the end of 2018 a recurrent ulceration at the radial surface of the same finger, evaluated as BCC relapse, was found.
For this reason, the patient was referred to our Plastic Surgery Department to undergo a diagnostic incisional biopsy.
The lesion was nodular, indurated and ulcerated. The pathological report was positive for ulcerated nodular type basal cell carcinoma, with numerous mycotic colonies (Fig. 3).
According to the latest guidelines, a radical surgical treatment of the BCC was proposed.
In detail, the surgical planning comprended wide excision directly followed by reconstruction with skin graft or dorsal metacarpal flap.
Such operation is usually accomplished as day surgery under local or regional anesthesia with light sedation.
Two-step reconstruction procedure, to get free margins assessment before accomplishing wound repair, appeared to be a second choice, due to lesion small dimensions and in order to avoid prolonged fuctional impairment of the hand.
Instead, the use of dermal substitute for reconstruction after tumor resection routinely is not considered in our institute, relating to the subsequent inflammatory response that may play a role in tumor local relapse.
For severe skin retractions concomitant commissuroplasties were proposed to restore fingers mobility, but patient refused.
At that time plantar surfaces were still affected by a multitude of ulcerated lesions compatible with Huriez syndrome.
Currently the patient is attending regular 6 months' follow up both in plastic and dermatology outpatient clinic.

Discussion

The diagnosis of this rare cancer-prone genodermatosis confined to the hands and feet is complex since only few cases are described, besides some cases may remain

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Table I - *Huriez syndrome differential diagnosis*

Disease	Genetics	Onset	Clinical features
Huriez syndome[1]	Autosomal dominant	neonatal/puberty Progression in adulthood	- not severe palmoplantar keratoderma - nail changes - extremities scleroatrophy - skin cancer related
Unna-Thost syndrome[1]	Autosomal dominant	Early childhood	- Diffuse severe nonepidermolytic palmoplantar keratoderma - hyperhidrosis
Scleroderma[1,2]	Non-hereditary autoimmune disease	Adulthood	- systemic symptoms (i.e pulmonary and pericardial fibrosis, bowel obstruction) - Raynaud's phenomenon - sclerodactyly
Dyskeratosis congenita[2,3]	Autosomal dominant X linked recessive	Childhood	- nail dystrophy - poikiloderma - mucosal leukoplakia - bone marrow failure
Rothmund Thomson syndrome[3]	Autosomal recessive	Neonatal	- photosensitivity - poikiloderma and hyperkeratosis - cataract - growth retardation - skin cancer related - radial ray malformations
Pigmented xerodermoid[3]	Autosomal recessive	Puberty/Adulthood	- skin dryness and keratoses - photophobia - hyper-pigmented freckle-like macules - skin cancer related

1. Patrizi A, Di Lernia V, Patrone P: *Palmoplantar keratoderma with sclerodactyly (Huriez syndrome).* Journal of the American Academy of Dermatology, 1992; 26(5), 855-57.
2. Çelik NS, Yaşar Ş, Aytekin S, Güneş P: *A Rare Syndrome Resembling Scleroderma: Huriez Syndrome*. Skin Appendage Disorders, 2017; 4(2), 82-85.
3. Jairath V, Mohan M, Chandra S, Kharge P, Fernendes C: *Poikiloderma a varied presentation. Huriez syndrome.* Indian Dermatology Online Journal, 2015; 6(1), 27.

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undiagnosed because not well defined clinical presentation and symptoms.

The sclerodactyly not associated with Raynaud's phenomenon is the most prominent feature of the syndrome [5]; over the years our patient faced a severe progression of this feature causing important functional impairment due to skin sclerosis and painful ulcerated lesions on the feet.

The differential diagnosis of Huriez syndrome are among disorders characterized by hands sclerosis and palmoplantar keratoderma (Table I). It is distinguished from scleroderma by early onset and progression during adulthood and by absence of systemic symptoms and Raynaud's phenomenon [2].

In Huriez syndrome keratoderma is slight and not homogenous, this is why patients with prominent keratoderma, associated with hyperhidrosis and with occasional sclerodactyly or nail changes, are more likely to be affected by Unna-Thost syndrome [1,6].

In some severe plantar and palmar keratodermas sclerosis may be present, but in Huriez syndrome this is a prominent feature.

Huriez syndrome normally appears at early age, persists unmodified in adulthood and may be genetically trasmissed [1]. Our patient was 79 years old presenting worsening of clinical symptoms since puberty. In her genealogical tree her mother and brother had the same disturb, instead her children result unaffected.

The life expectation in Huriez syndrome is not reduced, except for tumor's complications [3].

In literature, the association between Huriez syndrome and aggressive SCC of the affected skin is largely described [1,3,4,6-10], occurring in around 15% of affected individuals. Affected patient carry a >100-fold higher risk for the development of aggressive SCC of the skin [3,4].

SCC is the most feared complication of the syndrome, leading to early metastasis. Even if the pathogenetic mechanism of tumorigenesis is unknown, sclerosis, atrophy, and scarring are well recognized risk factors for SCC.

Despite it's not atypical that different skin tumors arise simultaneously in the same patient, we believe that our case report is highly interesting because there is not current description of Huriez syndrome associated with basal cell carcinoma arising from affected skin.

Clinically, the development of SCC and other non-mela-

noma skin cancers are similar to Marjolin's ulcer, which relates to aggressive SCC arising in chronic ulcer, scar tissue and burn scar [11]. In our patient, the palmar skin was fragile with scleroatrophia, process similar to scarring, thus predisposing to skin cancer [3,11].

Conclusion

At present the treatment of Huriez syndrome is focused only on symptoms control and tumors eradication, maybe future genetic research may provide new informations regarding the disease and may promote development of specific treatment.

References

1. Huriez C, Deminatti M, Agache P, Mennecier M: *Une génodysplasie non encore individualisée: La génodermatose scléro-atrophiante et kératodermique des extremités fréquemment dégénerative.* Sem Hop 1968; 44:481-8.

2. Burton JL, Ebling FJG: *Disorders of connective tissue.* In: Rook A, Wilkinson DS, Ebling FJG, et al., eds. Textbook of Dermatology. 4th ed.London: BlackwellScientific, 1986: p.1813.

3. Sekar SC, Srinivas CR: *Huriez syndrome.* Indian J Dermatol Venereol Leprol 2008; 74:409-10.

4. Hamm H1, Traupe H, Bröcker EB, Schubert H, Kolde G: *The scleroatrophic syndrome of Huriez: A cancer-prone genodermatosis.* Br J Dermatol, 1996; 134(3):512-18.

5. Patrizi A, Di Lernia V, Patrone P: *Palmoplantar keratoderma with sclerodactyly (Huriez syndrome).* Journal of the American Academy of Dermatology, 1992; 26(5), 855-57.

6. Delaporte E, N'guyen-Mailfer C, Janin A, Savary JB, Vasseur F Feingold, et al.: *Keratoderma with scleroatrophy of the extremities or sclerotylosis (Huriez syndrome): A reappraisal.* British Journal of Dermatology, 1995; 133(3), 409-16.

7. Watanabe E, Takai T, Ichihashi M, Ueda M: *A Non familial Japanese case of Huriez syndrome:p 53 expression in squamous cell carcinoma.* Dermatology, 2003; 207:82-4.

8. Guerriero Cl, Albanesi C, Girolomoni G, De Simone C, Capizzi R, Amerio P, et al.: *Huriez syndrome: Case report with a detailed analysis of skin dendritic cells.* Br J Dermatol, 2000; 143(5):1091-96.

9. Lee YA, Stevens HP, Delaporte E, Wahn U, Reis A: *A gene for an autosomal dominant scleroatrophic syndrome predisposing to skin cancer (huriez syndrome) maps to chromosome 4q23.* The American Journal of Human Genetics, 2000; 66(1), 326-30.

10. Çelik NS, Yaşar Ş, Aytekin S, Güneş P: *A rare syndrome resembling scleroderma: Huriez syndrome.* Skin Appendage Disorders, 2017; 4(2), 82-85.

11. Oruç M, Kankaya Y, Sungur N, Özer K, Işık VM, Ulusoy MG, et al.: *Clinicopathological evaluation of Marjolin ulcers over two decades.* Kaohsiung J Med Sci; 2017; 33(7):327-33.

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