DOI: http://dx.doi.org/10.3315/jdcr.2012.1112

Journal of Dermatological Case Reports

PHOTOLETTER TO THE EDITOR

Dyschromatosis universalis hereditaria: an infrequently occurring entity in Europe

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Abstract

Dyschromatosis universalis hereditaria (DUH) is a rare genodermatosis mainly described in asian subjects. Here, we report a case of a caucasian 11-year-old boy with DUH and an unaffected twin brother. Parents were not consanguineous. A review of the main phenotical, clinical and hystological aspects of this rare entity is exhibited. Differential diagnose might be stablished with several pigmentary disorders, so Dermatologist might have this entity in mind to make a correct diagnose, specially in cases with no response to typical treatments. (*J Dermatol Case Rep*. 2012; 6(3): 96-97)

Key words:

dyschromatosis, dyschromatosis universalis hereditaria, pigmentary disorder

Dyschromatosis universalis hereditaria (DUH) is a rare genodermatosis. Generalized hyperpigmented and hypopigmented macules characterize its clinical appearence. It belongs to the group of dyschromatosis. Dyschromatosis symmetrica hereditaria (DHS) presents an acral distribution and unilateral dermatomal pigmentary disorder (UDPD) segmental lesions.[1,2] DUH has a difficult differential diagnosis. Early diagnose is very important in order to detect DUH associated comorbidities.[1,3]

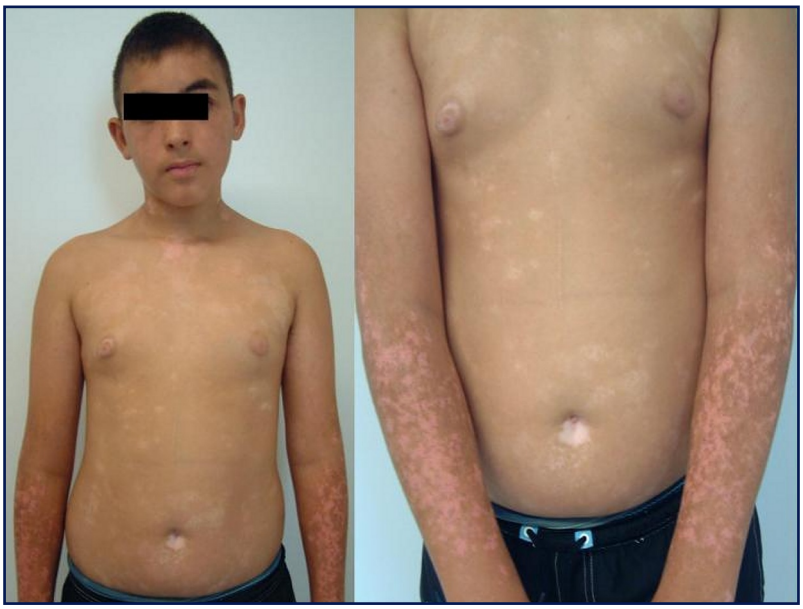

Figure 1

Clinical appearance. Hypopigmentated and hyperpigmentated lesions in a reticular pattern located on trunk and limbs.

A 11-year-old boy was referred to our Department with the diagnose of vitiligo. He was born in Tenerife, Spain. On physical examination hypopigmented and hyperpigmented macules distributed in a reticular pattern located on trunk and limbs were observed, specially on distal areas. Hair, nails and mucoses had a normal appearence. His medical history showed a mild mental retardation. His parents were not consanguineous and he had an unaffected twin brother. Familial history did not revealed other affected members or other remarkable conditions. Parents described that lesions started when he was 2 years old. He had been treated as vitiligo for three years with topical steroids, immunomodulators, moisturizers and fotoprotector without any improvement. Blood test with immunoglobulin and thyroid profile showed normal ranges. Imaging studies, including abdominal ultrasonography and torax radiography showed normal features. Cutaneous biopsy from hyperpigmentated and hypopigmentated areas were performed. Fontana, HE and HMB45 stains showed hypopigmentated lesions with normal melanocyte number but a lack of melanin. Hyperpigmentated areas showed normal melanocytes with and increase of pigmentation and mild incontinence. With this clinical, blood test and hystological findings the diagnose of DUH was stablished.

DUH was first described in Japan in 1933 by Ichikawa and Hiraga.[2] Clinical reports have been described in Japan, China, Taiwan, Tunicia, India, Iraq, Saudi Arabia and Niger, South America and South Africa. In Europe is a rare disease and it has been described as isolated clinical cases with an indian background.[1,2,3,4,5]

Autosomical dominant (AD), recessive (ar) and sporadic inheritance have been described. However AD inheritance is most common.[4] Specific DUH ethiopathogeny is still unknown, but several theories have been proposed: a) an abnormal melanocyte synthesis rate or an abnormal melanocyte activity, with a normal appearence and number of melanocytes. The keratinocytes and melanocytes of the hyperpigmentated lesions contained numerous, fully melanized melanosomes and, in contrast, the melanosomes were absent from both keratinocytes and melanocytes of hypopigmentated lesions.[2,4] b) Interference with the neural-melanocytic interaction in early embriogenic life could explain deffects in melanocytes maduration.[1]

DHU has a heterogeneous phenotype. The face is affected only in 50% of cases and soles-palms and mucoses are usually unaffected.[1,2] Several comorbidities have been associated with DHU as short stature, photosensitivity, theeth abnormalities, deafness and cataracts.[1] Differential

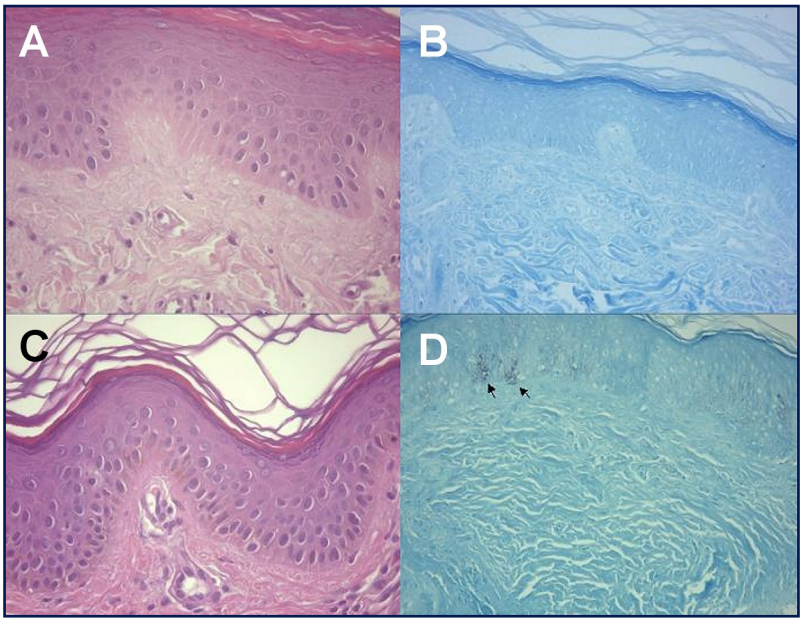

Figure 2

(A, B) Hypopigmentated areas hystological appearence. (C, D) Hyperpigmentated lesions hystological features.

diagnosis with other entities as other dyschromatiosis, generalized reticulate pigmentary dermatosis (Naegeli-Franceschetti-Jadassohn, dermatophatia pigmentosa reticularis or dyskeratosis congenita), xeroderma pigmentosum, residual leukoderma, amyloidosis cutis discromia and vitiligo should be stablished. The most typical missdiagnoses are xeroderma pigmentosum and vitiligo.[3,5] Our patient was treated as vitiligo for three year without any improvement.

The genetic study is not necessary because the most of cases present a non-specific mutation and several cases are sporadic. Some authors have described mutations in chromosome 6 (6q24.2-q25.2) and others in chromosome 12 (12q21-q23).[1,5] However, DSH have been associated with different mutations in the gene ADAR1, this mutation is not present in patients with DUH and it is other reason to consider them two different entities.[3] The diagnose of DUH is mainly clinical. Our case theorically could be the result of a post-cygotical mutation (sporadic inheritance), because the patient had a cutaneous non affected twin brother.[1,3]

We report a rare clinical case of DHU in a Caucasian Spanish boy with an unaffected twin brother.

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