Case Report

Dyschromatosis symmetrica hereditaria: a pedigree study

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Abstract: Dyschromatosis symmetrica hereditaria (DSH), also known as reticulate acropigmentation of Dohi, is an autosomal dominant disease with high penetrance, characterized by hypo- and hyper-pigmented macules in varying sizes on the dorsal extremities with reticulated pattern. This paper presents an unmarried Chinese male patient with typical dermatological lesions. Four generations of the family with 26 people, 9 cases suffered from the same disease and symptoms.

Keywords: Dyschromatosis symmetrica hereditaria, dyschromatosis, reticulated acropigmentation of Dohi

Introduction

Dyschromatosis symmetric hereditaria (DSH) characterized by the presence of hyper- and hypo-pigmented macules arranged in a reticular pattern. DSH is a rare genodermatosis, autosomal dominant with high penetrance, but some sporadic cases were reported. It is most commonly found in Japanese patients, but there have been cases in some parts of Asia and Europe [1, 2]. In this study, we presented a case of DSH with typical dermatological lesions in an unmarried Chinese male with a family history of the disorder.

Case presentation

A 19-year-old unmarried male patient presented with multiple small hyperchromic and hypo-chromatic macules distributed symmetrically on the dorsum of the hands and feet (Figure 1A, 1B). The lesions appeared in infancy and progressed with age. The face was spared. Mucous membrane, hair, and nails were uninvolved. Systemic examination did not reveal any abnormality. There was no history of photosensitivity or photophobia. There was no history of preceding dermatosis or associated systemic involvement.

Family survey revealed that 9 cases were found in a Han Chinese family of 26 people in four generations, suffered from the same course and lesion (Figure 2). The similar distributions of skin lesions could be found on feet and even on trunks. Based on the clinical features, a diagnosis of DSH was made. Skin biopsies were not performed because clinical characteristics were apparently sufficient for the diagnosis of DSH.

Laboratory examination including complete blood test, renal and liver function tests, serum HIV, and HbsAg were negative or within normal limits. No treatment was recommended. The patient received orientation and remains in ambulatorial follow-up.

Discussion

DSH is a genodermatosis characterized by multiple small hypo- and hyper-pigmented macules, with irregular size and shape, symmetrically distributed on the backs of the hands and feet. Some patients also show freckle-like macules on the face. Palms, soles, and mucous membranes are free from the disorder [1, 2]. The skin lesions commonly spreading with age until adolescence and last a lifetime. These abnormalities are otherwise asymptomatic and do not affect general health. However, there are isolated reports of association with neurofibromatosis type I, thalassemia, polydactyly, and torsion dystonia [3].
Since 2003, a genetic mutation has been identified on chromosome 1q11-1q21 as being responsible for the production and distribution changes of melanin [4]. Miyamura et al. first identified heterozygous mutations of the gene DSRAD or ADAR1, responsible for coding the double-stranded adenosine deaminase specific RNA as the cause of DSH [5]. Despite more than 90 different mutations in the DSRAD gene have been described in the literature, it is still uncertain how these changes can cause the same phenotype [6].

This disease must be differentiated from other pigmented disorders such as reticulate acropigmentation of Kitamura, marked by presence of atrophy and absence of hypopigmented lesions; Dowling-Degos disease with reticulate hyperpigmentation in the body's folds, accompanied by comedogenic lesions on the back and the neck and depressed or pitted scars; initial cases of xeroderma pigmentosum, distinguished by the development of more serious symptoms of xerosis, atrophy, telangiectasia, and tumors in photo-exposed areas; dyschromatosis universalis hereditaria with predominating lesions on the trunk that start in childhood [7, 8].

No treatment is effective for this genodermatosis. The exact frequency of the DSH is unknown since the main changes are confined to the skin with no systemic involvement, and many cases remain unreported [1, 2]. We report this case due to its rare occurrence in this part of the world and in view of the need of proper diagnosis and guidance for such patients.

Disclosure of conflict of interest

None.

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