Int J Clin Exp Med 2017;10(2):3987-3989
www.ijcem.com /ISSN:1940-5901/IJCEM0023212

Case Report

Dyschromatosis symmetrica hereditaria: a pedigree study

Wei Chen[1], Jianjun Liu[1], Mengmeng Yin[1], Zekun Wang[1], Jianzhong Zhang[2], Junlian Liu[1]

[1]*Department of Dermatology,* [2]*Department of Pathology, 306 Hospital of PLA, Beijing, PR China*

Received January 4, 2016; Accepted November 24, 2016; Epub February 15, 2017; Published February 28, 2017

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

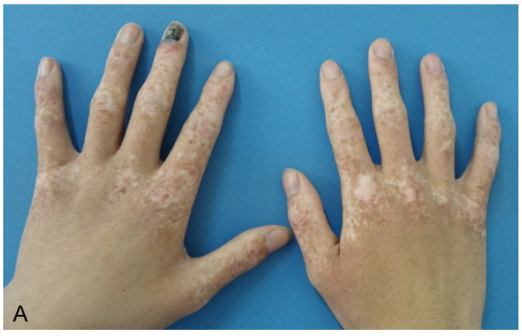
A 19-year-old unmarried male patient presented with multiple small hyperchromic and hypochromic macules distributed symmetrically on the dorsum of the hands and feet (**Figure 1A**, **1B**). The lesions appeared in infancy and progressed with age. The face was spared. Mucous membrane, hair, and nails 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].



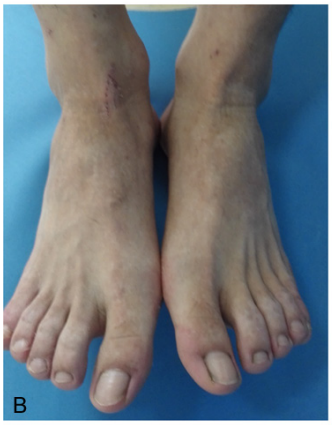


Figure 1. A, B. Scaly, hyper- and hypo-pigmented macules distributed symmetrically on the dorsum of the hands and feet.

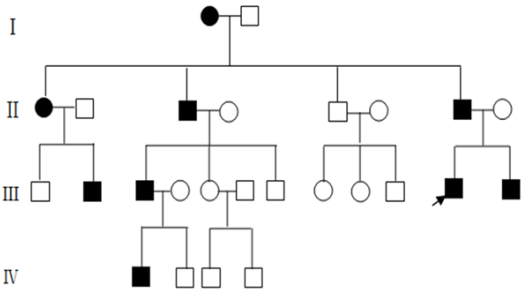


Figure 2. Family pedigree. Arrow indicates the patient presented with DSH, open symbols indicate unaffected individuals, blackened symbols indicate affected individuals, squares indicate men, and circles indicate women. Blackened bars indicate the chromosome region shared by affected members of the pedigree.

Since 2003, a genetic mutation has been identified on chromosome Iqll-Iq2l 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 pigmentary 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.

Address correspondence to: Dr. Junlian Liu, Department of Dermatology, 306 Hospital of PLA, 9 North Anxiang Road, Chaoyang District, Beijing 100101, PR China. Tel: +86-10-66356729; Fax: +86-10-64871261; E-mail: liujunlian306@sina.com

References

[1] Hayashi M and Suzuki T. Dyschromatosis symmetrica hereditaria. J Dermatol 2013; 40: 336-343.

[2] Muller CS, Tremezaygues L, Pfohler C and Vogt T. The spectrum of reticulate pigment disorders of the skin revisited. Eur J Dermatol 2012; 22: 596-604.

[3] Consigli J, Zanni MS, Ragazzini L and Danielo C. Dyschromatosis symmetrica hereditaria: report of a sporadic case. Int J Dermatol 2010; 49: 918-920.

[4] Zhang XJ, Gao M, Li M, Li M, Li CR, Cui Y, He PP, Xu SJ, Xiong XY, Wang ZX, Yuan WT, Yang S and Huang W. Identification of a locus for dyschromatosis symmetrica hereditaria at chromosome 1q11-1q21. J Invest Dermatol 2003; 120: 776-780.

[5] Miyamura Y, Suzuki T, Kono M, Inagaki K, Ito S, Suzuki N and Tomita Y. Mutations of the RNA-specific adenosine deaminase gene (DSRAD) are involved in dyschromatosis symmetrica he-

reditaria. Am J Hum Genet 2003; 73: 693-699.

[6] Liu Q, Wang Z, Wu Y, Cao L, Tang Q, Xing X, Ma H, Zhang S and Luo Y. Five novel mutations in the ADAR1 gene associated with dyschromatosis symmetrica hereditaria. BMC Med Genet 2014; 15: 69.

[7] An JM, Ko BJ, Cho MK and Whang KU. A case of sporadic dyschromatosis universalis hereditaria. Ann Dermatol 2015; 27: 467-468.

[8] Vachiramon V, Thadanipon K and Chanprapaph K. Infancy- and childhood-onset dyschromatoses. Clin Exp Dermatol 2011; 36: 833-838, quiz 839.