Case Report

Siblings with Dyschromatosis Universalis Hereditaria A Rare Case Report

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Abstract

Dyschromatoses are a group of genodermatosis characterized by the presence of both hyperpigmented and hypopigmented macules of variable shapes and sizes. Here, we report siblings presented with asymptomatic progressive mottled pigmentation of reticulate pattern over the trunk and limbs since 6 years of their age without any systemic or other cutaneous illness. They were born to nonconsanguineous parents following an uneventful pregnancy. Their paternal grandfather had a similar appearance. Histological examination was consistent with dyschromatosis universalis hereditaria (DUH). Based on the clinical and histological findings, a diagnosis of DUH was made. We report this case of rare genodermatosis in siblings affected by the same disease.

Keywords: Dyschromatosis universalis hereditaria, genodermatosis, siblings

Introduction

Dyschromatoses are a group of genodermatosis characterized by the presence of both hyperpigmented and hypopigmented macules of variable shapes and sizes. The spectrum of dyschromatoses includes dyschromatosis universalis hereditaria (DUH), dyschromatosis symmetrica hereditaria (DSH), acropigmentation of Dohi, Dowling–Degos disease, and a segmental form called unilateral dermatomal pigmentary dermatosis.[1]

Case Report

Two siblings, a 21-year-old female and a 17-year-old male, respectively, presented to the dermatology outpatient department (OPD) with multiple hypopigmented and hyperpigmented macules arranged in a reticulate pattern over the trunk and bilateral upper and lower limbs. The face, palms, and soles were spared. The lesions were noticed 6 years after birth in both of them and increased progressively with age. There was no history of photosensitivity, seizures, or any other systemic illness. They were born of nonconsanguineous marriage by normal vaginal delivery and had normal developmental milestones. There was a history of similar lesions in their paternal grandfather. On cutaneous examination, multiple, well-defined, bilaterally symmetrical both hyperpigmented and hypopigmented macules of variable sizes and shapes were present in a reticulate pattern over the trunk and bilateral upper and lower limbs with sparing of the face, palms, and soles in both the patients [Figures 1 and 2]. Atrophy or telangiectasias were absent in the affected areas. The hair, teeth, nail, and mucosa were normal. There was no eye; ear, nose, and throat, or any other systemic abnormalities. Microscopically, H- and E-stained sections of the skin biopsy from both siblings showed the presence of an increase in melanin pigment extending to the stratum spinosum. The dermis was unremarkable except for the presence of mild periadnexal lymphocytic infiltrate [Figure 3]. Their paternal grandfather was not able to visit OPD as he was old and debilitated. Based on the findings, a clinical diagnosis of DUH was made, which was confirmed after histopathological results.

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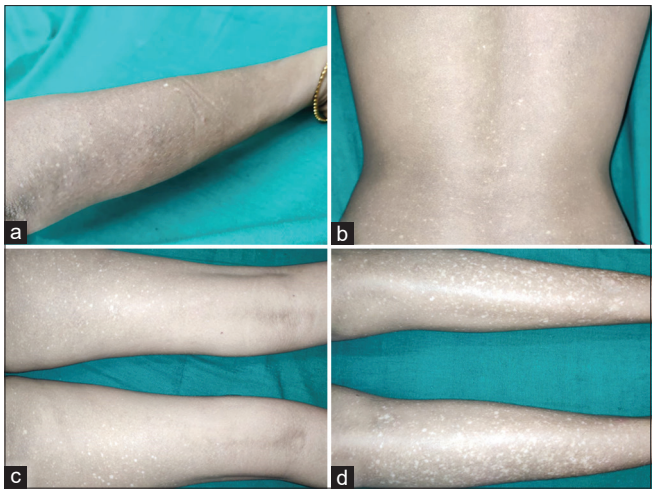

Figure 1: A female with multiple hypopigmented and hyperpigmented macules over the (a) right forearm, (b) back, (c) posterior aspect of bilateral thighs, and (d) bilateral legs

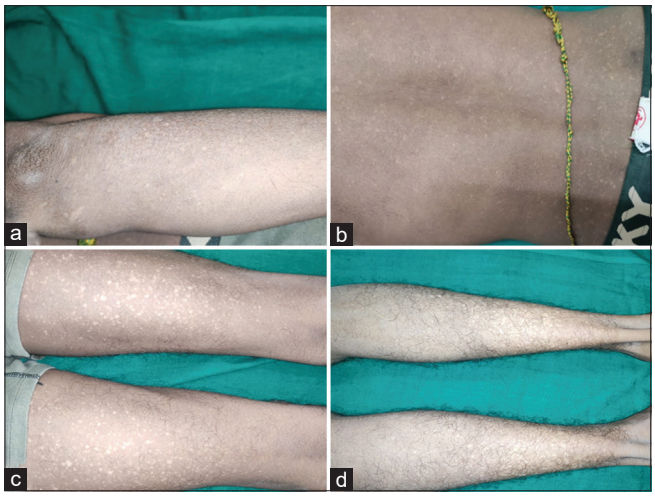

Figure 2: A male with multiple hypopigmented and hyperpigmented macules over the (a) right forearm, (b) back, (c) anterior aspect of bilateral thighs, and (d) bilateral legs

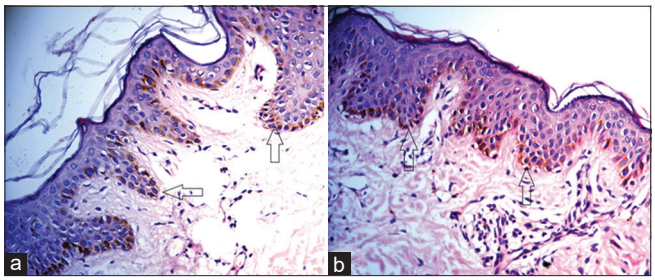

Figure 3: Presence of increased melanin pigment deposition (white arrow) in the epidermis in the (a) male Sibling and (b) female sibling (H and E, ×400)

Discussion

Various types of dyschromatoses based on the distribution of lesions include DUH characterized by both hyper- and hypopigmented macules all over the body which was first reported in 1933 by Ichikawa and Higara.[2] DSH, also known as acropigmentation of Dohi, described in 1929 by Toyama,[3] involves only the acral areas. Both are common in Japan. Few cases of DUH among Europeans, South Americans, and Indians have also been described.[4]

The etiology of this disorder is not known completely. A variable autosomal mode of inheritance of DUH has been described.[2] Their paternal grandfather also had a similar appearance in our case. Some clinicians have suggested that DSH might be a subtype of DUH. The RNA-specific adenosine deaminase gene (ADAR1, DSRAD) responsible for DSH was described first in Japanese families.[5] Mutations in the ADAR1 have been reported in Chinese families that confirmed that this gene is responsible for DSH in different ethnic groups.[6] Sixteen novel mutations in the ADAR1 have been reported by Suzuki *et al*.,[7] with no such mutations in patients with DUH. Furthermore, they did not find the two diseases in the same family, denoting that these two conditions are completely different.

Skin lesions usually present in the 1st years of life in DUH. The trunk and extremities are the dominant sites. Facial lesions were seen in almost 50% of the affected individuals, but the involvement of the palms and soles is rare.[8] Hair and nails abnormalities may also occur.[4] In our case, the face, palms, and soles were spared. The hair, nails, teeth, and mucosae appeared normal.

Nuber *et al*.[9] indicated that DUH is a disorder of melanosome synthesis rate or melanocyte activity and not a disorder of melanocyte number. Association of DUH has also been found with abnormalities of dermal connective tissue, nerve tissue, and other systemic complications.[4] No such features characterized our patients.

Other diseases characterized by cutaneous dyschromia must be differentiated from DUH. The most important is xeroderma pigmentosum in our country.[8] Both siblings had no signs of XP (no poikiloderma, atrophy, or telangiectasia). Usually, there is no worsening or progression of disease with age. In all reports, there was no spontaneous regression.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patients have given their consent for their images and other clinical information to be reported in the journal. The patients understand that their name and initials will not be published and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

References

1. Kumar S, Bhoyar P, Mahajan BB. A case of dyschromatosis universalis hereditaria with adermatoglyphia: A rare association. Indian Dermatol Online J 2015;6:105-9.
2. Kenani N, Ghariani N, Denguezli M, Sriha B, Belajouza C, Nouira R. Dyschromatosis universalis hereditaria: Two cases. Dermatol Online J 2008;14:16.
3. Toyama I. Dyschromatosis symmetrica herediteria. Jpn J Dermatol 1929;29:95-6.
4. Sethuraman G, Srinivas CR, D'Souza M, Thappa DM, Smiles L. Dyschromatosis universalis hereditaria. Clin Exp Dermatol 2002;27:477-9.
5. Miyamura Y, Suzuki T, Kono M, Inagaki K, Ito S, Suzuki N, *et al.* Mutations of the RNA-specific adenosine deaminase gene (DSRAD) are involved in dyschromatosis symmetrica hereditaria. Am J Hum Genet 2003;73:693-9.
6. Liu Q, Liu W, Jiang L, Sun M, Ao Y, Zhao X, *et al.* Novel mutations of the RNA-specific adenosine deaminase gene (DSRAD) in Chinese families with dyschromatosis symmetrica hereditaria. J Invest Dermatol 2004;122:896-9.
7. Suzuki N, Suzuki T, Inagaki K, Ito S, Kono M, Fukai K, *et al.* Mutation analysis of the ADAR1 gene in dyschromatosis symmetrica hereditaria and genetic differentiation from both dyschromatosis universalis hereditaria and acropigmentatio reticularis. J Invest Dermatol 2005;124:1186-92.
8. Al Hawsawi K, Al Aboud K, Ramesh V, Al Aboud D. Dyschromatosis universalis hereditaria: Report of a case and review of the literature. Pediatr Dermatol 2002;19:523-6.
9. Nuber UA, Tinschert S, Mundlos S, Hauber I. Dyschromatosis universalis hereditaria: Familial case and ultrastructural skin investigation. Am J Med Genet A 2004;125A: 261-6.