

An Interesting Case of Dyschromatosis Universalis Hereditaria

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Abstract

Dyschromatosis universalis hereditaria is an autosomal dominant disorder but can be inherited recessively or sporadically with peculiar pigmentary changes of different sizes, intermingling between hyperpigmented and hypopigmented macules. Here, we report this interesting and a rare case of dyschromatosis universalis hereditaria in a young male involving the entire body including the palms and soles sparing the face.

Keywords: Interesting, case, universe

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INTRODUCTION

Dyschromatosis universalis hereditaria is a rare genodermatosis that presents with unusual pigmentary changes of varying sizes, consisting of hyperpigmented hypopigmented macules. It was first described and reported in Ichikawa and Hiraga in 1933. It is an autosomal dominant disorder but it may be transmitted sporadically or by autosomal recessive trait.[1]

Dyschromatosis includes a set of disorders which are characterized by pigmentary changes, presence of small irregular hypopigmented and hyperpigmented macules. It is a spectrum which includes

Dyschromatosis universalis hereditaria (DUH)

Dyschromatosis symmetrica hereditaria (DSH)

Acropigmentation of Doh

Segmental form called unilateral dermatomal pigmentary dermatosis (UDPD)

DUH has been found to be associated (but rarely associated) with Epilepsy, short stature, deafness, albinism, photosensitivity, mental retardation, ocular abnormalities [2]

It is rarely reported in India but is mainly seen in the east asian population mostly in Japan.

CASE REPORT

A 21-year-old unmarried male came to our dermatology outpatient department with asymptomatic, blackish, and whitish discoloration of the skin over the body for the past 16 years. The lesions appeared when the patient was 5 years of age over the palms and soles and gradually progressed to involve the abdomen, upper limbs and lower limbs. Sparing of face was noted. The lesions did not get worsened with exposure to sunlight.

He was born to non-consanguineous parents. The patient has no siblings. There was no history of similar lesions in family members.

On examination of the skin, there were multiple, discrete, hyperpigmented, and hypopigmented macules of size 2mm symmetrically distributed on the upper and lower limbs, palms, and soles with sparing of the face

The mucous membrane showed hyperpigmented macules on the tongue and buccal mucosa of both sides. Examination of nails showed longitudinal ridging of both the fingernails

Examination of scalp and hair was normal. Systemic examination did not reveal any abnormality. Laboratory tests including complete haemogram, renal function tests, and liver function tests, were normal. HIV, and HBsAg was non-reactive. The USG of the abdomen was normal.

Hence a provisional diagnosis of dyschromia Universalis hereditaria was made.

A biopsy was taken from the hypopigmented and hyperpigmented skin lesion on the trunk to confirm the diagnosis.

Section showed skin with basket weave hyperkeratosis, increased melanin pigment in the basal layer, and few areas showed no melanin pigment. The papillary dermis showed melanophages, and perivascular inflammatory cells infiltrate. Hence the diagnosis was confirmed.



Fig 1. Both the palms and soles show hypo pigmented and hyperpigmented macules



Fig 2. Clinical picture shows hyper pigmented and hypo pigmented macules on the forearm



Fig 3. Clinical picture shows multiple, discrete, hyperpigmented, and hypopigmented macules on the trunk



Fig 4a and 4b: showing longitudinal ridging of the fingernails

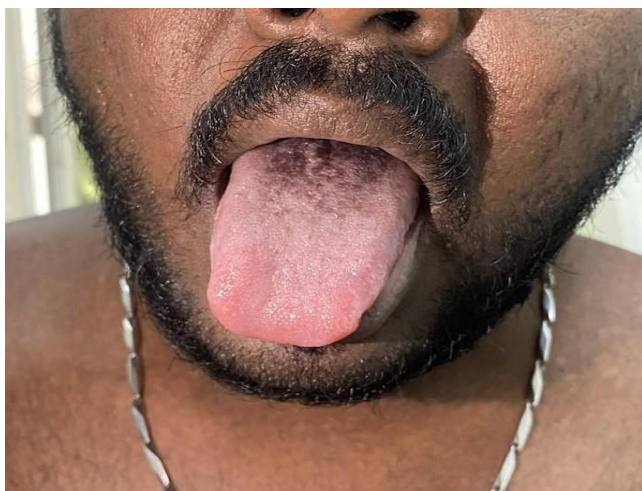


Fig 3a and Fig 3b. Clinical photograph shows pigmented macules over the tongue and buccal mucosa



Fig 5. shows sparing of face

affected sites while facial lesions are seen in almost 50% cases.

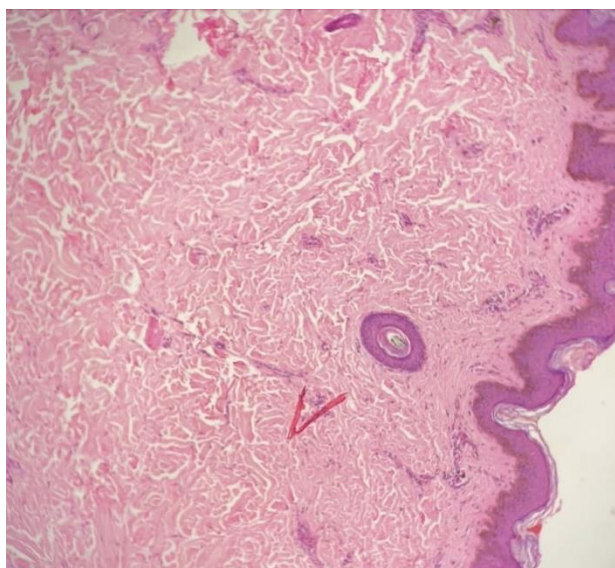
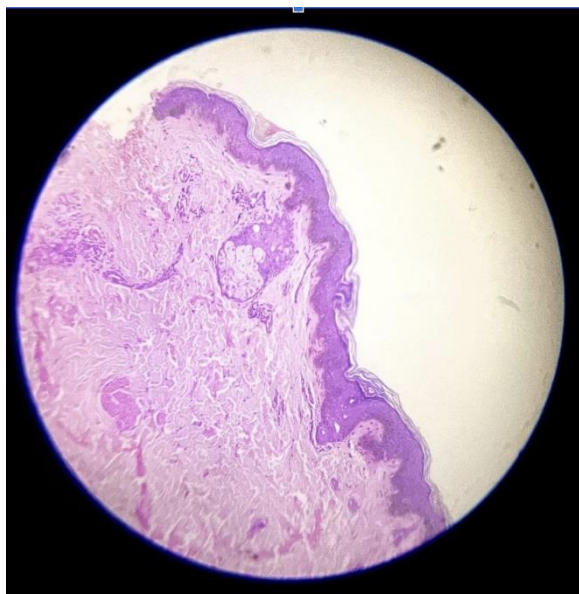


Fig 6a - 10x, Fig 6b - 40x

Histopathology of skin lesions shows Basket weave hyperkeratosis.

Increased melanin pigment in the basal cell layer.

Papillary dermis show: melanin incontinence and perivascular inflammatory cell infiltrate

DISCUSSION

DUH is a rare genodermatosis which was first reported in 1933. It manifests with a peculiar mixture of hypo pigmented and hyperpigmented macules all over the body. DUH is most often reported in Japan however few cases of DUH were also described in Europe, South america, India and Saudi Arabia.

In DUH, the trunk and the extremities are the commonly

Palms and soles are involved very unusually, however our patient had involvement of palms and soles. Majority of the cases show an autosomal dominant pattern. In this case, we could not elicit any significant family history. [3]

The histopathology shows a focal decrease or increase in melanin in the basal layers and occasional pigmentary incontinence.[4] An ultrastructural skin investigation stated that DUH is a disorder of Melanosome synthesis rate and melanocyte activity and not a disorder of melanocyte number.

Eighty two percent of patients that have been reported so far showed clinical symptoms before the age of 6 years. [5] DUH can also be associated with dermal connective tissue, nerve tissue abnormalities or can also be associated with other systemic complications, no such features were seen in our patient. In DUH the lesions occur at the sites where they are unexposed to sun. Lesions are benign and do not worsen with age. [6]

DUH has been reported with increasing frequency in other races besides being confined in the Japanese population. Even though only a few cases have been reported in india. It has to be differentiated from xeroderma pigmentosum, in which the lesions are confined to sun exposed areas.

The other differential diagnosis for this condition are dermatopathia pigmento reticularis, dyskeratosis congenita (DKC) These dyschromias lead to cosmetic disfigurement that can bare a psychological troll on the patient.[7]Role of NBUVB as a treatment modality can be evaluated and tried for better cosmetic appearance, but so far there has been no effective treatment modality.

In conclusion, we present here an interesting and rare case of Dyschromatosis universalis hereditaria involving the entire body including the palms and soles with sparing of face.

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