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DYSCHROMATOSIS UNIVERSALIS HEREDITARIA WITH DYSCHROMATOSIS SYMMETRICA HEREDITARIA- AN UNUSUAL PRESENTATION IN THE SAME FAMILY

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ABSTRACT
Dyschromatosis universalis hereditaria (DUH) is a rare autosomal dominant genodermatosis frequently reported in japanese literature. Both autosomal recessive and sporadic cases though infrequent have been reported. We hereby report a case of DUH with dyschromatosis symmetrica hereditaria (DSH), the two disorders of the same spectrum present in different individuals of the same family.

KEYWORDS: Dyschromatosis Symmetrica Hereditaria, Dyschromatosis Universalis Hereditaria, Genodermatosis.

INTRODUCTION
Dyschromatosis is defined as a disorder with 'freckle like' hypo and hyperpigmented macules of small size and irregular shape.[1] It is a spectrum of disorder which includes DUH, DSH also known as acropigmentation of dohi and a segmental form known as unilateral dermatomal pigmentary dermatosis (UDPD). DUH is characterized by hyper- and hypopigmented macules distributed over the entire surface of the skin while DSH has a symmetric reticulate pigmentation over the extremities mainly over acral aspects.[2] In this case report, we describe a case of DUH and DSH in the same family with the mother presenting with DUH while her son with DSH.

CASE REPORT
A 32 year old married women presented to us with the chief complaints of multiple light to dark brown colored flat skin lesions of variable sizes since the age of 4 years. The condition gradually progressed to involve the entire body by the age of 10 years.(Figure 1) There was no history of photosensitivity or exposure to any chemical substance triggering the onset of such lesions. There was no history of any drug intake. There was no history of consanguinity among her parents or in her own marriage. Her family history was negative but her 11 year old son has started developing a few hyper- and hypopigmented lesions on ventral aspect of bilateral forearm in symmetric fashion since the the age of 5.(Figure 2) The condition is non-progressive in her son.

Cutaneous examination of the lady revealed multiple hyper-and hypopigmented macules coalescing to form patches at variable places in a symmetric fashion on her neck, bilateral axilla, forearms arms, trunk, back, inner aspect of her thighs, legs. Her face, palms and soles were relatively spared. Mucosa was not involved. On cutaneous examination of her son hyper- and hypopigmented macules were present symmetrically on the ventral aspect of bilateral forearm. No other site seem to be involved. There was no apparent erythema, atrophy or telangiectasia in both the patients. Systemic examination of both mother and her son did not reveal any abnormality. Routine laboratory investigations including complete blood count, hepatic and renal function tests, serum electrolytes were within normal range. ECG and chest X-ray results did not show any signs of abnormality. Patient did not give her consent for histopathological examination, therefore skin biopsy could not be performed. We counseled the patient regarding the usually benign and non-progressive nature of the condition, the genetic transmission of the disorder and possibility of having its occurance in her future progeny and thereafter. We also told her about its rare neurological, ophthalmological and hematological complication.

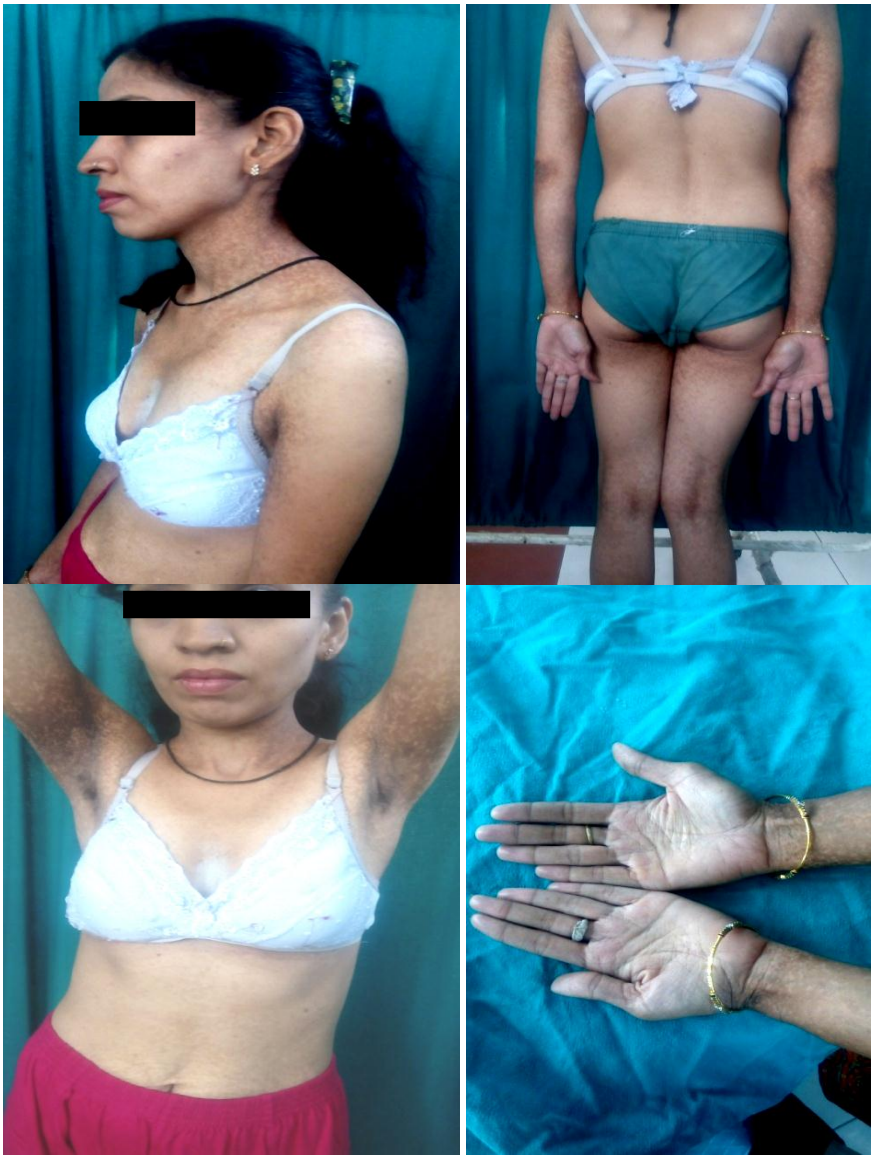

Figure. 1: Generalised hyper- and hypopigmented macules diffusely present all over the body in the mother.

Figure. 2: A few hyper- and hypopigmented macules over ventral aspect of forearm in the son.

DISCUSSION

DUH, a rare genodermatosis was first described by Ichikawa and Hiraga in 1933.[3] The gene responsible for DUH maps to chromosome 6q24.2-q25.2.[2] It is characterised by hyper-and hypopigmented macules distributed over entire surface of the skin. The another variant of dyschromatosis known as DSH was first described by Toyama in 1929.[4] It is caused by a heterozygous mutation in ADAR1 gene located on chromosome 1q21 which mediates post-transcriptional modification of m-RNA.[5] This impaired RNA editing during melanoblast migration leads to formation of either hypo or hyperactive melanocytes and that the most affected melanocytes migrate farthest to the extremities, i.e. the hands and feet.[5] Both the conditions have been commonly reported in japenese population but these are being reported with increasing frequencies in other races as well. Various associations reported with DUH include coxa valga, nerve compression, small stature, high tone deafness, photosensitivity, neurosensory hearing defect, epilepsy and mental retardation.[6,7]

Various differential diagnosis to be considered in a case of DUH are dyskeratosis congenita, generalized dowling degos, incontinentia pigmenti, Naegelie-Franceschetti-Jadasshon, chronic arsenic toxicity and an important one for DSH is xeroderma pigmentosum as both involve photoexposed parts.[8]

In our case, mother is suffering from DUH while her son has got DSH, an unusual presentation as members of the same family are suffering from different disorders of the same group, a rarely reported entity in the literature so far.

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