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Research Article

DYSCHROMATOSIS UNIVERSALIS HEREDITARIA: A RARE GENODERMATOSES IN INDIA

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A B S T R A C T

Dyschromatosis universalis hereditaria (DUH) is a rare genodermatoses characterized by hyperpigmented and hypopigmented macules which is inherited most commonly in an autosomal dominant manner. We hereby report a case of DUH in a female with family history in an autosomal recessive manner.

INTRODUCTION

The dyschromatoses are a group of disorders characterized by the presence of both hyperpigmented and hypopigmented macules, many of which are small in size and irregular in shape[1]. It includes dyschromatosis symmetrica hereditaria (DSH) and dyschromatosis universalis hereditaria (DUH), both of which are seen most commonly in Japan[2] and acropigmentation of Dohi and a segmental form called unilateral dermatomal pigmentary dermatosis (UDPD).

In 1933, Ichikawa and Hiraga were the first to describe DUH as well-demarcated brown macules admixed with various-sized hypopigmented macules in a generalized as opposed to acral distribution seen in DSH.[3]

Case Report

A 35-year-old female hailing from south east India came with the chief complaints of multiple light and dark coloured lesions all over the body since birth. Initially few lesions were present over the trunk later gradually progressed to involve rest of the body. There was no history of itching, photosensitivity or chemical use or drug intake. There was no history of consanguinity among the parents but history of similar lesions were present in siblings.

Cutaneous examination revealed multiple, well-defined to ill-defined hypopigmented and hyperpigmented macules of various sizes, distributed bilaterally symmetrically on the trunk, upper and lower limbs, face, palms and soles [Fig 1-4].

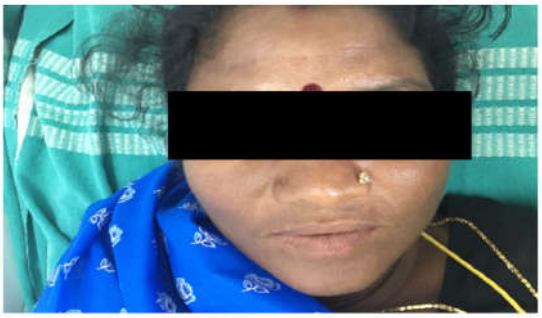

Fig 1 A 35-year-old female patient whose facial involvement was spared

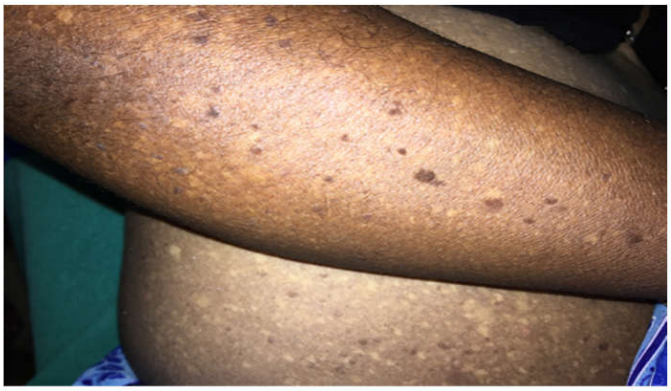

Fig 2 multiple hyper pigmented and hypopigmented macules present over the right upper limb and trunk.

There was no erythema, atrophy or telangiectasia of the affected skin. Mucosae were spared. Hair, nails, and teeth were normal. Systemic examination was normal. Ophthalmic and otorhinolaryngological examination revealed no abnormalities.

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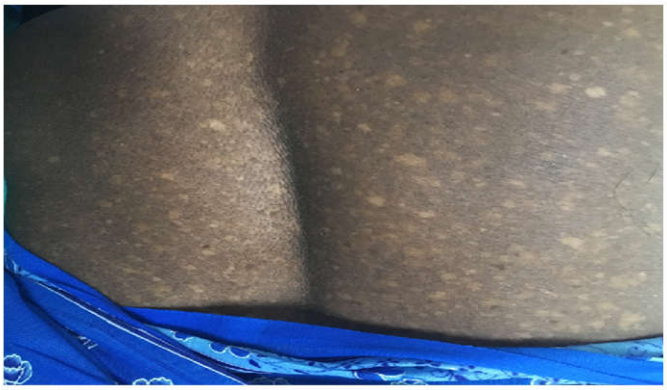

Fig 3 multiple hyper pigmented and hypopigmented macules present over the back

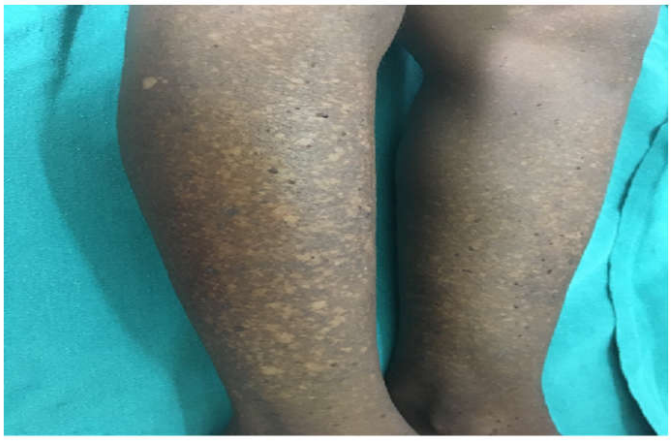

Fig 4 multiple hyper pigmented and hypopigmented macules present over the lower limbs.

The biopsy was taken from both the hyperpigmented and hypopigmented lesions. Epidermis in both the biopsies were thinned out and the basal layer in the hyperpigmented lesions showed increase in pigment whereas in the hypopigmented lesions it showed decrease in pigment. Dermis in both the biopsies showed mild perivascular lymphomononuclear infitrate.

DISCUSSION

DUH is a rare genodermatosis which has been reported most often from Japan. Over 80% of DUH patients develop dyschromia by 6 years of age, and ~20% have dyspigmentation at birth[4]. A subset of DUH is caused by heterozygous mutations in *ABCB6*, which encodes an ATP-binding cassette transporter protein that is expressed in keratinocytes and melanocytes. Additional autosomal dominant and recessive forms of DUH have been mapped to 6q24.2-q25.2 and 12q21-q23, respectively[5]. A few have inherited it in an autosomal recessive fashion.

Previously it was believed to be a disorder of melanocyte number. In a recent ultrastructural skin investigation, Nuber *et al*. indicated that DUH is a disorder of melanosome synthesis rate or in melanocyte activity and not a disorder of melanocyte number.[8]

In DUH, skin lesions are usually present in the 1st year of life. The trunk and extremities are the dominant sites. Hyperpigmented macules on the face can resemble ephelides or lentigines.

The dyschromia may involve the palms and soles as well as the dorsal aspects of the hands and feet, but it spares the mucous membranes. There is no seasonal change or spontaneous regression with age. Extracutaneous abnormalities reported in isolated cases of DUH include short stature and high-frequency deafness; abnormalities in erythrocytes, platelets and tryptophan metabolism; bilateral glaucoma and unilateral cataract; and seizures[6-7].

Differential Diagnosis includes Xeroderma pigmentosum which may have similar dyspigmentation, but it is in a photodistribution and associated with photosensitivity, premature actinic damage. However, lesions in DUH has a benign course and does not show telangiectasia or atrophy. Other differential diagnosis includes DSH, dyskeratosis congenita, chronic radiodermatitis, incontinentia pigmenti (Bloch-Sulzberger), generalized Dowling-Degos disease, Naegelie-Franceschetti-Jadassohn syndrome, chronic arsenic toxicity.[9]

From India there have been very few isolated case reports in the past.[10,11]. Despite its rarity it has its significance as it forms an important differential diagnosis of xeroderma pigmentosum. The dyschromias leads to cosmetic disfigurement which causes significant psychosocial consequences. And an autosomal recessive inheritance pattern was seen in our patient.

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