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"A RARE CASE REPORT OF DYSCHROMATOSIS UNIVERSALIS HEREDITARIA"

Dermatology

Dr. Akshata Yadav. S Post Graduate

Dr. Rajashekar T.S* Prof And HOD *Corresponding Author

Dr. Sureshkumar. K Associate Professor

ABSTRACT

Dyschromatosis universalis hereditaria (DUH) is a rare genetic skin condition that involves a mixture of hyperpigmented and hypopigmented macules with mottled pigmentation. Lesions usually start on the limbs and extend to the trunk over time. Cases in the Indian population are extremely uncommon, with the majority of recorded cases emanating from Japan. This case report describes a 65-year-old Indian male who had atypical involvement of scalp along with other common site like hands, legs trunk and back, with DUH as diagnosis.

KEYWORDS

Dyschromatosis universalis hereditaria, Autosomal dominant

INTRODUCTION

The rare autosomal dominant genodermatosis known as dyschromatosis universalis hereditarian (DUH) is typified by asymptomatic hyperpigmented and hypopigmented macules that are widely distributed on the trunk, limbs, and occasionally the face. The asymptomatic hyperpigmented and hypopigmented macules that are extensively dispersed on the trunk, limbs, and occasionally the face are the hallmark of dyschromatosis universalis hereditarian (DUH), a rare autosomal dominant genodermatosis.

A mutation in the ADAR1 gene causes this disease. The incidence is 0.3 per 100,000.

Case Report: A 65 year old male presented with multiple hypopigmented lesions over the trunk, arms, forearms, legs, scalp since he was 1 year old. The lesions initially started over both the legs and gradually progressed to involve thighs, new lesions were noticed over the period of time over the back, both hands progressing to involve the elbows and scalp. Patient gave history similar lesions were present in the father and his sister. There was no history of seizures or reduced sweating. No history photosensitivity or any other systemic compliants.

On cutaneous examination multiple asymptomatic, generalized, 0.5-1cm hyperpigmented macules interspersed with hypopigmented macules distributed dorsum of both the hands (Image 1) involving foreams and elbows, dorsum of both the feet(Image 2), back(Image 3), few over neck, chest, scalp (Image 4) with sparing of palms and soles. Mucosa spared.

Dermascopic Findings: Brownish pseudoreticular pigment network, uniform distribution of brown gray dots (red arrow).

Biopsy from forearm hyper and hypopigmented macules showed Melanin pigmentation in the epidermis basal layer slightly high, and there may be some indication of pigmentary incontinence. Differentials of xeroderma pigmentosum, A diagnosis of DUH was suggested by the presence of both hyperpigmented macules interspersed between the depigmented macules and reduced and elevated basement membrane pigmentations observed in the depigmented and hyperpigmented regions, respectively.

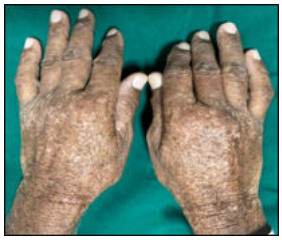

Figure-1

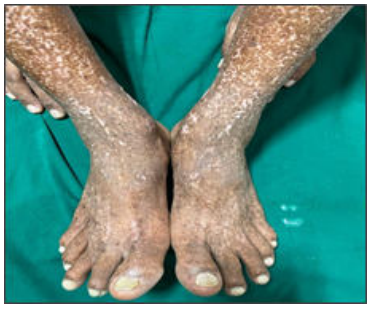

Figure-2:

(Fig 1&2) Multiple asymptomatic, generalized, 0.5-1cm hyperpigmented macules interspersed with hypopigmented macules present over the dorsum of both hands and feet, legs.

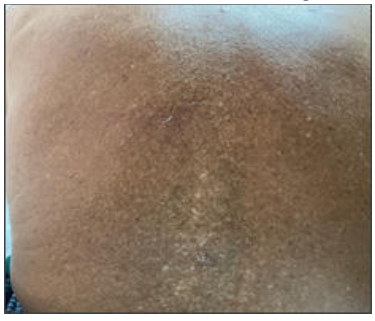

Figure-3 : Multiple hypopigmented and hyperpigmented macules present over the back

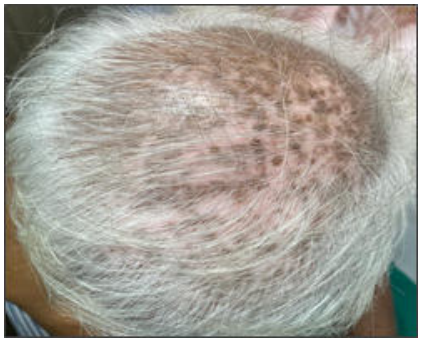

Figure-4 : Multiple depigmented macules interspersed beteem the normal skin present over the scalp.

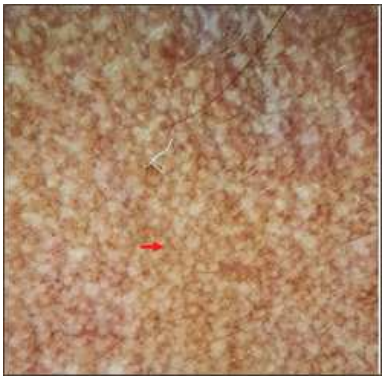

Figure-5: Brownish pseudoreticular pigment network, uniform distribution of brown gray dots (red arrow).

DISCUSSION:

Ichikawa and Hiraga discovered DUH for the first time in 1933, it has been documented worldwide, including in India, and primarily affects people of East Asian heritage.[1] Dyschromatosis universalis hereditaria (DUH) is a rare genetic pigmentary disorder due to mutation in the ABCB6 gene. Other genes include SASH1, PER 3, and KITLG (DUH type 2).[2]

There are two loci known to cause DUH: one on chromosome 12q21–6q25.2 in two Chinese families and one on chromosome 6q24.2–6q25.[3,4]

Mutation of these genes leads to disruption in the melanin synthesis and melanosome transport. Inheritance pattern can be autosomal dominant, autosomal recessive and few sporadic cases have been documented.[5]

This condition is characterized by the presence of multiple asymptomatic, diffuse, and mottled hyper and hypopigmented macules distributed all over the body and sometimes on face.[6] The illness usually manifests in early childhood and progresses slowly or non-progressively.

It is a Melanosome synthesis disorder in epidermal melanin units. Age-related progression or reversal of DUH lesions is nonexistent. Numerous diseases have been linked to DUH, including learning disabilities, neurosensory hearing impairments, ocular abnormalities, and deafness,insulin-dependent diabetes mellitus, x-linked ocular albinism, mental retardation, epilepsy, small stature.[7] The differential diagnosis for DUH includes residual leukoderma, vitiligo, xeroderma pigmentasum, and dyskeratosis congenita.

Different modalities are available for the treatment which includes Q switched alexandrite laser for hyperpigmented lesions,NBUVB therapy and camouflage. These have show some improvement in pigmentation in few studies.

CONCLUSION:

In this condition lesions primarily appear on various areas of the body, the scalp is less commonly involved when compared to other regions like the limbs, and torso. Scalp involvement in Dyschromatosis Universalis Hereditaria (DUH) is generally rare. Here we report a case of DUH with lesions distributed over atypical sites like scalp and with an autosomal dominant inheritance pattern making it a unique case.

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