

# Dyschromatosis Universalis Hereditaria

Bista M,<sup>1</sup> Agrawal S,<sup>1</sup> Agrawal Y<sup>2</sup>

<sup>1</sup>Department of Dermatology and Venerology,

<sup>2</sup>Department of Pathology,

B.P. Koirala Institute of Health Sciences,  
Dharan, Nepal.

## Corresponding Author

Muna Bista

Department of Dermatology and Venerology,

B.P. Koirala Institute of Health Sciences,

Dharan, Nepal.

E-mail: [anumbista@gmail.com](mailto:anumbista@gmail.com)

## Citation

Bista M, Agrawal S, Agrawal Y. Dyschromatosis Universalis Hereditaria. *Kathmandu Univ Med J.* 2021;73(1):146-7.

## INTRODUCTION

Dyschromatoses are a group of rare pigmentary genodermatoses characterized by the presence of multiple small irregular hyperpigmented and hypopigmented macules over the body. Dyschromatosis universalis hereditaria (DUH), Dyschromatosis symmetrica hereditaria (DSH) and Unilateral dermatomal pigmentary dermatosis (UDPD) are the entities included in it. Pigmentary change appears in a generalized pattern in DUH as opposed to DSH where the lesions are distributed symmetrically over the extremities only. In UDPD, there is segmental pattern of the pigmentary disorder. DUH clinically presents as multiple pinpoint to pea-sized hypo- and hyper-pigmented macules distributed in a reticulated pattern within the first few years of life. The mode of inheritance of DUH is usually both autosomal dominant and autosomal recessive patterns. However, very few sporadic cases have also been found. Here we report a sporadic case of DUH.

## ABSTRACT

Dyschromatosis universalis hereditaria (DUH) is a rare pigmentary genodermatosis usually inherited in autosomal dominant fashion characterized by multiple pinpoint to pea-sized hypo- and hyper-pigmented macules arranged in reticulate pattern that develops within the first few years of life. An 11 years old boy presented with multiple gradually progressive asymptomatic hypopigmented macules on hyperpigmented background on trunk, extremities and face since 2 years of age. Family history was absent. Punch biopsies revealed increased number of melanocytes in the epidermis with basal cell vacuolar alteration and pigmentary incontinence and perivascular infiltration by lymphocytes and melanophages in the dermis. We herein present a sporadic case of dyschromatosis universalis hereditaria.

## KEY WORDS

*Dyschromatosis universalis hereditaria, Reticulate pigmentation*

## CASE REPORT

An 11 years old boy presented to the skin outpatient department of B.P. Koirala Institute of Health Sciences with the appearance of multiple hypopigmented macules on hyperpigmented background on trunk, extremities and face since 2 years of age. The lesions were asymptomatic, however were gradually progressive. There was no history of photosensitivity or any other systemic complaints, chemical exposure or drug intake. There was no history of similar complaints or consanguinity in the family.

On examination, multiple hypopigmented macules of size ranging from few millimeters to a centimeter were distributed over the face, trunk and extremities in reticulate pattern with relative sparing of the palms and soles. There was no atrophy or telangiectasia of the affected skin. Hair, nails, teeth and oral mucosae were normal. Systemic examination was unremarkable. Routine investigations

including complete blood counts, liver function tests, renal function tests and urinalysis were sent and were within normal limits.

Punch biopsies were taken which showed increased number of melanocytes in the epidermis with basal cell vacuolar alteration. The dermis showed increase pigmentary incontinence and perivascular infiltration by lymphocytes and melanophages. Based on this clinical presentation and histopathological examination, diagnosis of DUH was made although the family history was negative in our patient.

## DISCUSSION

Dyschromatoses is a group of genetic pigmentary disorders characterized by the presence of macules which can be both hyperpigmented and hypopigmented. It chiefly comprises of three conditions i.e. Dyschromatosis universalis hereditaria (DUH), Dyschromatosis symmetrica hereditaria (DSH) or acropigmentation of Dohi and a segmental form called unilateral dermatomal pigmentary dermatosis (UDPD). The first case to be reported was that of DSH described by Toyama in 1929.<sup>1</sup> Following this DUH was first described by Ichikawa and Hiraga in 1933.<sup>2</sup> The cases of dyschromatoses are predominantly seen in Japan but there have been various case reports from different parts of the world. The prevalence of DUH is said to be 0.3 per 100,000.<sup>3</sup>

DUH is mostly inherited as autosomal dominant (AD) pattern with variable penetrance, but various cases of autosomal recessive pattern and sporadic cases have also been described.<sup>4</sup> In our case there was no history of similar condition in any of the family members. Regarding

the pathogenesis of the disease, it is not very clear but it has been linked to mutation of the ABCB6 gene which is thought to be involved in the transfer of melanosome to keratinocytes.<sup>5</sup>

Clinically, the lesions of DUH appear in infancy or early childhood usually before the age of 5 years with trunk and extremities as the dominant sites but face may also be involved in almost 50% of the cases. Palms, soles and mucosa are usually spared but some cases of the involvement have also been reported. In our case, the lesions were predominant over the trunk and extremities and few over the face with sparing of the palms and soles. Rarely, it has been seen in association with X-linked ocular albinism, tuberous sclerosis, photosensitivity and neurosensory hearing defect, small stature and high-tone deafness.<sup>6-9</sup> No such association could be elicited in our case.

The histopathological examination of the skin lesion shows evidence of increase (if hyperpigmented) or decrease (if hypopigmented) in melanin in the basal cell layer. Occasionally, pigmentary incontinence may be present.<sup>10</sup> In our case, findings were consistent with DUH and hence clinical diagnosis of DUH was supported by the histopathology.

This case of DUH is reported because of its rarity. The disease was earlier considered to be limited to Japanese population but now it is being reported from various parts of the world. There have been very few case reports from the Indian subcontinent especially that of sporadic cases. Hence we highlight the importance of diagnosis of the condition even in absence of significant family history.

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