# A case of dyschromatosis universalis hereditaria with adermatoglyphia: A rare association

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## ABSTRACT

Dyschromatosis universalis hereditaria (DUH) is a rare, autosomal dominant genodermatosis with a peculiar reticulate pigmentary change, consisting of hyperpigmented macules mingled with hypopigmented lesions to give an overall impression of mottling. We herein report a case of DUH with adermatoglyphia in a young male with family history of the disorder.

**Key words:** Adermatoglyphia, dyschromatosis universalis hereditaria, reticulate pigmentation

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## INTRODUCTION

Dyschromatoses are a group of reticulate pigmentary disorders characterized by the presence of both hyper- and hypo-pigmented macules that are small in size and irregular in shape. It is a spectrum of diseases that includes dyschromatosis universalis hereditaria (DUH), dyschromatosis symmetrica hereditaria (DSH) or acropigmentation of Dohi and a segmental form called unilateral dermatomal pigmentary dermatosis. DSH was the first reported as a clinical entity by Toyama in 1929.[1] In recent years, with the development of genetic analysis, ADAR1 or DSRAD gene mutations have been found in DSH but not in DUH, and therefore these two diseases are now regarded as two different entities. DUH was first described by Ichikawa and Hiraga in 1933. It presents as a generalized reticulate pigmentation.[2] Adermatoglyphia or immigration delay disease is characterized by congenital absence of epidermal ridges and eventually finger prints. We are presenting a case of DUH associated with adermatoglyphia in a young Indian male with family history of the disorder.

## CASE REPORT

A 31-year-old unmarried male presented with progressively increasing, generalized mottled pigmentation since the age of 3 years. The lesions had started over neck and trunk that spread centrifugally to the extremities over a period of 10 years. There was no history of photosensitivity. There was no history of handling any chemical directly or any significant history of drug intake. There was history of similar lesions in the younger brother. His younger sister was normal. There was no history of consanguinity among the parents who were also normal. On dermatological examination, multiple hyper- and hypo-pigmented macules of varying size ranging from 2 to 30 mm were present in a reticulate pattern without atrophy over the forehead [Figure 1], trunk [Figures 2 and 3], upper and lower extremities [Figure 4]. Reticulate pigmentation was present also on palms with absent dermatoglyphics distal to the distal creases [Figure 5]. Dystrophic changes in the fingernails including longitudinal striations, ragged cuticles and pterygium in one fingernail were seen [Figure 6]. Mucous membranes and soles were normal. Systemic examination did not reveal any abnormality. Routine investigations, including complete blood counts, urinalysis, liver function tests, renal function tests, and serum electrolytes were within normal limits. Venereal Disease Research Laboratory test and enzyme-linked immunosorbent assay for human immunodeficiency virus were both nonreactive. Histopathological examination from hyperpigmented lesions showed mild hyperkeratosis. The basal layer showed

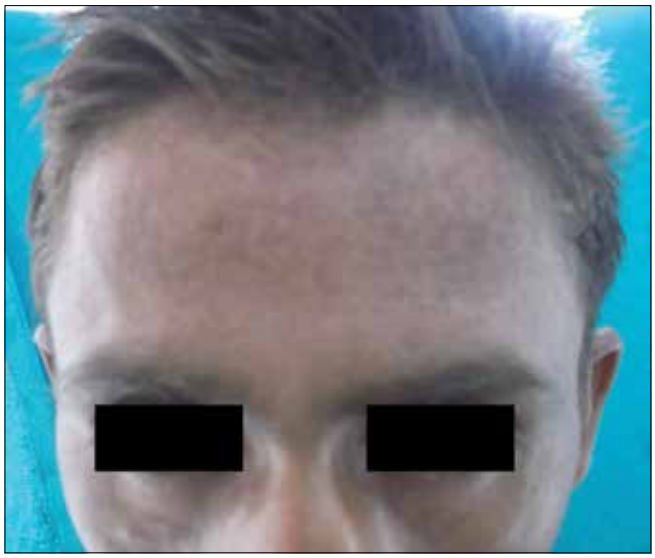

**Figure 1:** Symmetrically distributed hyper- and hypo-pigmented lesions on forehead

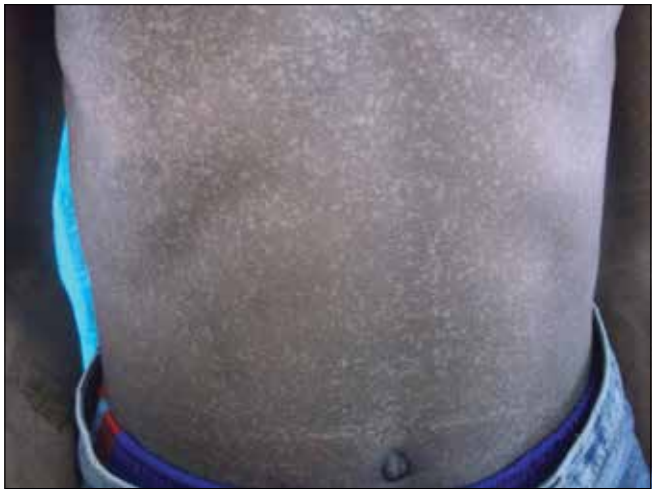

**Figure 3:** Generalized symmetrically distributed hyper- and hypo-pigmented lesions on abdomen

pigmentation with some coarse melanocyte granules, predominantly in the lower 2–3 layers of the epidermis. There was prominent melanin incontinence with melanophages in the dermis [Figure 7]. Sparse lymphohistiocytic infiltrates were seen in the perivascular areas. This confirmed the diagnosis of DUH.

## DISCUSSION

Dyschromatosis universalis hereditaria is a rare genodermatosis that is commonly encountered in Japan; however, rare familial cases have been reported from Europe, China, and India.[3-6] DUH is an autosomal dominant (AD) disorder with variable penetrance, but a few individuals show an autosomal recessive pattern.[6,7] Although the precise etiology of this disorder is not yet known and the culprit gene still unidentified, DUH locus has recently been mapped to chromosome 6q24.2-q25.2, but

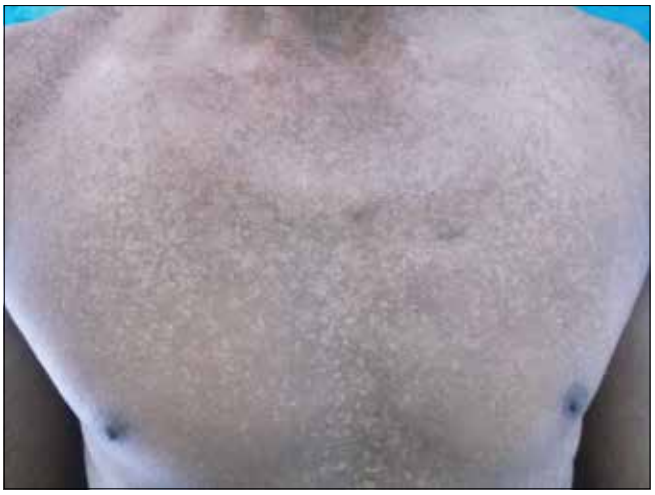

**Figure 2:** Generalized symmetrically distributed hyper- and hypo-pigmented lesions on chest

**Figure 4:** Reticulate pigmentation over legs

has not been confirmed by many.[8] In our patient, there was history of similar lesions in the younger brother.

In DUH, skin lesions are usually present before 6 years of age with equal sex preponderance. The trunk and extremities are the dominant sites. Facial lesions were seen in almost 50% of affected individuals, but involvement of palms and soles is rare.[5] In our case, trunk and extremities were the predominant sites with involvement of face. Palms were involved, with sparing of the soles.

Dyschromatosis universalis hereditaria may be associated with abnormalities of dermal connective tissue, nerve tissue, or be associated with other systemic complications, but no such features existed in our patient.[7,9] Abnormalities of hair and nails have also been reported, but only nail changes were present in our patient.[10]

It has been suggested in the past that DUH is a disorder of melanocyte number. On the basis of recent electron microscopic study, it has been suggested that DUH may be a disorder of

**Table 1: Differential diagnoses of DUH**

| Disorders | Inheritance | Genetics | Onset | Distribution of lesions | Skin morphology | Pathology | Additional features |
|---|---|---|---|---|---|---|---|
| Dyschromatosis Universalis Hereditaria | AD occasionally AR | 6q24.2eq25.2 or 12q21eq23 | Early childhood | Trunk, limbs, face, almost all over the body | Mottled hyperpigmented and hypopigmented macules | Pigmentary incontinence with increase or decrease in melanin content of the keratinocytes in basal layer, normal melanocyte number | Associated abnormalities were rarely seen though cases with tuberous sclerosis, photosensitivity and neurosensory hearing defect, small stature and high-tone deafness, X-linked ocular albinism have been reported |
| Dyschromatosis Symmetrica Hereditaria | AD occasionally AR | DSRAD gene, ADAR1 gene | Early childhood | Back of hands and feet, face | Mottled hyperpigmented and hypopigmented macules | Pigmentary incontinence with increase or decrease in melanin content of the keratinocytes in basal layer, normal melanocyte number | None |
| Dyskeratosis congenita | X-linked occasionally AD | X-linked: DKC1 AD: TERC | Early childhood | Neck, upper chest, upper arms | Reticulate hyperpigmented and hypopigmented macules | Melanophages in upper dermis | Mucosal leukoplakia; nail dystrophy; dental changes; hair abnormalities; ocular changes; hyposthenic body builds; dysphagia; bone marrow dysfunction; tumor predisposition |
| Xeroderma pigmentosum | AR | Various genes | Early childhood | Sun-exposed areas | Lentigines and hyperpigmented macules | Hyperkeratosis, chronic inflammatory infiltrate in upper dermis, irregular accumulation of melanin in basal layer, melanocyte number can be increased | Xerosis; atrophy; telangiectasia; skin tumors |
| Generalized Dowling Degos disease | AD | Not identified | Young adulthood | Generalized, flexural areas, trunk, limbs | Mottled hyperpigmented and hypopigmented macules, papules | Mild hyperkeratosis, thinned epidermis, downward proliferation of rete ridges, basal hyperpigmentation | Facial pits, comedo-like papules, palmar pits, broken epidermal ridges, café-au-lait spots |
| Incontinentia pigmenti (Bloch-Sulzberger) | X-linked dominant | X-linked: NEMO | At birth | Along Blaschko lines | Hyper-pigmentation in stage 3, hypopigmentation in stage 4 | Hyper-pigmentation due to pigmentary incontinence in dermal melanocytes | Alopecia; nail dystrophy; hypo/adontia; eye abnormalities; CNS involvement |
| Naegeli Franceschetti Jadassohn syndrome | AD | 17q21 | Early childhood | Neck, chest, and abdomen | Brown or gray-brown reticular pigmentation | Variable pigmentation of basal keratinocytes; pronounced pigmentary incontinence | Dental anomalies; palmar/plantar hyperkeratosis; hypohidrosis; nail dystrophy |
| X-linked reticulate amyloidosis | Not inherited | | Depends on the accumulation of arsenic in target tissues and its metabolism and elimination | Trunk, areola, flexural creases, any part of the body may be affected | Guttate hypopigmentation superimposed on hyperpigmentation "raindrops on a dusty road" | Increased melanin in epidermis, no melanocytic proliferation | Increased risk of internal malignancies |
| RAPK | AD | Not identified | First and second decade | Dorsa of hands and feet | Mottled pigmentation with atrophy | Epidermal atrophy with club like elongation of rete ridges and excess of melanin in basal layer | Pits and breaks in dermatoglyphics on palms and soles |

AD: Autosomal dominant; AR: Autosomal recessive, CNS: Central nervous system, DUH: Dyschromatosis universalis hereditaria

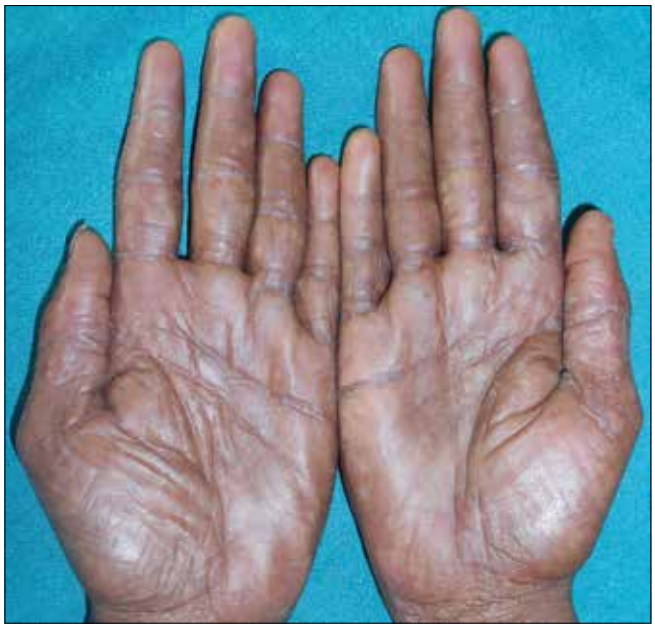

**Figure 5:** Absent dermatoglyphics on fingertips

**Figure 6:** Dystrophic changes in fingernails

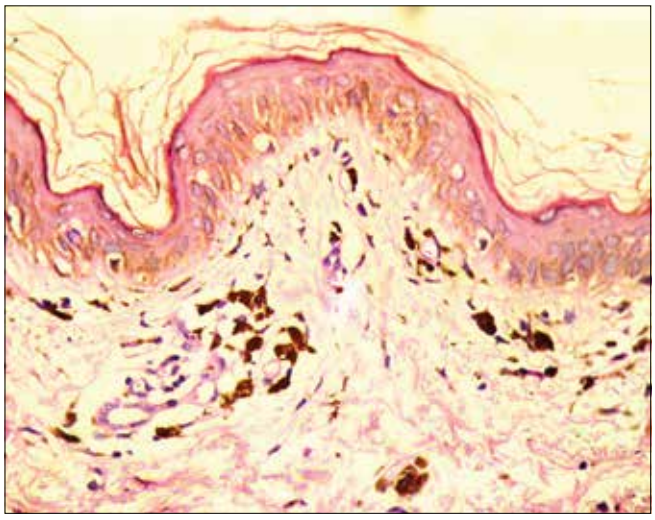

**Figure 7:** Histopathology from hyperpigmented lesion showed increased melanin deposition in the basal layer of epidermis with prominent pigmentary incontinence (H and E, ×400)

melanosome production and distribution in epidermal melanin units rather than a disorder of melanocyte number.[11]

Adermatoglyphia is caused by a point mutation in the splice-site of a 3' exon of the gene for SMARCAD1-helicase, which is mapped to chromosome 4q22.[12] It is an autosomal dominant disorder and has been reported in association with other reticulate pigmentary disorders such as dermatopathia pigmentosa reticularis (AD), Naegeli–Franceschetti–Jadassohn syndrome (AD with keratin 14 gene defect) and Benson's syndrome (AD ectodermal dysplasia). Our patient showed adermatoglyphia distal to the distal creases in both the hands, but there were no other cutaneous features suggesting overlapping of adermatoglyphia with these reticulate pigmentary disorders.

Lesions of DUH have to be differentiated from other inherited reticulate pigmentary disorders such as dermatopathia pigmentosa reticularis (DPR), Naegeli–Franceschetti–Jadassohn syndrome, and dyskeratosis congenita [Table 1]. Naegeli–Franceschetti–Jadassohn syndrome is characterized by complete absence of dermatoglyphics, reticulate hyperpigmentation, palmoplantar keratoderma, abnormal sweating, subtle developmental anomalies of the teeth and hair.[13] DPR is characterized by triad of reticulate hyperpigmentation, noncicatricial alopecia, and onychodystrophy.[13-15] DUH can also present with mild palmoplantar hyperkeratosis and onychodystrophy.[10]

To the best of our knowledge, our report is the first case of DUH with ademathoglyphia from India. Although it is possible that our patient had two distinct genetic disorders by sheer chance, it is important to record any such clinical overlapping, since it may indicate genetic linkage. Due to unavailability and cost factor, we could not perform gene mapping, which might have explained their precise association.

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