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Case Report

Dyschromatosis universalis hereditaria in clinical practice– A rare case report

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Abstract

Dyschromatosis Universalis Hereditaria (DUH) is an AD (autosomal dominant) genetic skin disorder, rarely sporadic and is characterized by a combination of different types of pigmented macules in a reticulate or mottled arrangement all over the body. DUH usually develops during childhood. It differs from other pigmentary disorders because the pigmentation changes are widespread, symmetric, and often affect the face, trunk, and limbs. It was previously believed that DUH resulted from an abnormal number of melanocytes, excessive melanosome production, or improper distribution of melanin within the epidermal melanin units. DUH treatment options are limited, as there are no proven therapies to reverse or prevent skin colour changes. Current management focuses on minimizing sun exposure, using sunscreen, and addressing cosmetic concerns. While DUH is primarily a cosmetic concern, the condition can cause psychological distress due to the visible nature of the lesions.

Keywords: Dyschromatosis universalis hereditaria, Dyschromatosis symmetrica hereditaria or acropigmentation of dohi, Dyskeratosis congenita / Zinsser engman cole syndrome, Xeroderma pigmentosum, Generalized Dowling degos disease Or Reticulate pigmented anomaly of flexures, Incontinentia pigmenti (bloch Sulzberger), Naegeli franseshetti syndrome, Chronic arsenic toxicity

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1. Introduction

Ichikawa and Hiraga first described DUH in Japan in 1933.[1] DUH is a rare genodermatosis characterized by a distinctive pigmentary change, which consists of hyperpigmented macules of varying sizes interspersed with hypopigmented lesions, creating an overall mottled appearance.[2] The ABCB6 gene, situated on chromosome 2q36, codes a protein that belongs to the ABC (ATP Binding Cassette) transporter family. This protein is responsible for transporting molecules across cell membranes. It plays a key role in copper homeostasis, and mutations in ABCB6 can disrupt copper regulation, affecting tyrosinase function and leading to abnormal synthesis of melanin.[1] ABCB6 is also present on exosomes, helping transport melanosomes (containing melanin) to keratinocytes for melanin distribution across the skin.[1,3] Early childhood marks the onset of pigmentation, which progressively spreads and goes to a deeper extent. Once fully formed, these dyschromatic lesions do not vary in location or color and remain the same throughout life.

According[4] to Nuber et al. DUH is not a disease affecting several melanocytes, but rather a disorder of the activity of melanocytes or the melanosome synthesis rate.[5] Over 80% of patients develop dyschromatosis by 7 to 8 years of age, and few patients have dyspigmentation at birth. The dyschromia involves hands, feet, and even soles and palms, but it spares the mucous membranes. There is no variation with season. There is no spontaneous regression as age increases. Most common typical nail alterations include pterygium formation, hyperpigmentation, and dystrophy. Additional associated features may include photosensitivity, coxa valga, and neurosensory hearing defects.[6] Abnormalities of nerve tissues and connective tissue can be associated with dyschromatosis universalis hereditaria.[7-10]

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2. Case Report

A 26-year-old unmarried female presented with complaints of multiple, asymptomatic generalized hyperpigmented macules varying in size from 0.5 to 1 cm, interspersed with spotty hypopigmented macules noted over the upper chest, lower abdomen, face, neck, and dorsum of b/l feet. Lesions started developing at the age of 7 years and progressed as age increased. Hyperpigmented macules mixed with hypopigmented macules started over the upper chest and then gradually progressed to the face, upper limbs, foot, and axilla.(**Figure 1-5**) A diffuse, ill-defined pigmented patch without scaling was noted over the bilateral axilla. No atrophy, telangiectasias, or scaling were noted over the affected areas. Palms, soles, and mucosa are not affected. There is no history of photosensitivity, handling any detergents or chemicals, or drug intake prior to the development of lesions, seizures, or ocular disturbances. She was born to consanguineous parents, and three generations of family members are affected, including the grandfather, father, and brother, all of whom have similar complaints.

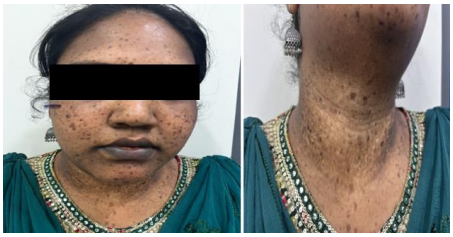

Figure 1: Multiple discrete hyperpigmented macules interspersed with hypopigmented macules noted over face and neck

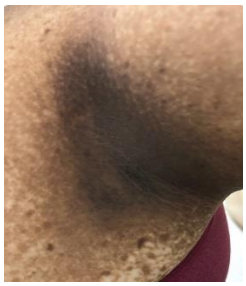

Figure 2: Ill-defined diffuse hyperpigmented patch with few hyperpigmented and hypopigmented macules in the periphery noted over both axillae

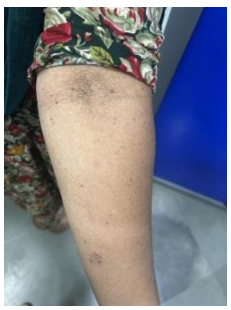

Figure 3: A few hypopigmented and hyperpigmented macules noted over the left forearm

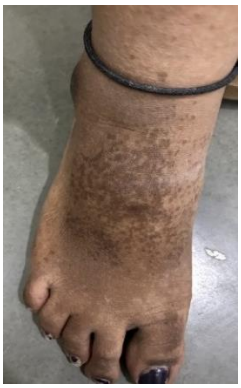

Figure 4: Multiple pigmented macules coalescing to form an ill-defined pigmented patch over the dorsum of the right foot

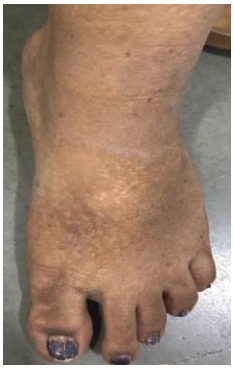

Figure 5: Multiple hypopigmented macules noted over the dorsum of the left foot.

The patient was advised to undergo a biopsy, but patient denied the same.

3. Discussion

Dyschromatosis is a rare genetic condition characterized by the presence of both hyperpigmented (dark) and hypopigmented (light) macules, often interspersed in a mottled or reticulated pattern. These lesions typically appear in an early age and can affect various parts of the body, including the hands, feet, face, trunk, and limbs. Rarely, hair, nails, and oral mucosa may also be involved. DUH is more common in Japanese people and is rarely seen in Indian population

Two classic dyschromatoses are present: Dyschromatosis universalis hereditaria, Dyschromatosis symmetrica hereditaria, and new form acquired brachial cutaneous dyschromatosis. Extracutaneous Manifestations seen in DUH are short stature, high-frequency deafness, abnormalities in erythrocytes and platelets, abnormal tryptophan metabolism, unilateral cataract, bilateral glaucoma, and seizures. Lesions of DUH have to be differentiated from DSH, Xeroderma pigmentosum, Dyskeratosis congenita, Incontinentia pigmenti, Generalized Dowling Degos disease, Chronic arsenic toxicity, Chronic radiodermatitis, Naegeli Franseshetti syndrome.(**Table 1,2**)

Table 1: Comparison of inherited disorders of dyschromatosis and related conditions

Disease	**Dyschromatosis universalis hereditaria [DUH]**	**Dyschromatosis symmetrica hereditaria [DSH] or Acropigmentation of Dohi**	**Dyskeratosis congenita / Zinsser engman cole syndrome**	**Xeroderma pigmentosum**
Inheritance	AD, Occasionally AR	AD, Occasionally AR	X-linked, occasionally AD Increased risk in successive generation due to progressive telomere shortening due to defective telomere function	AR
Genetics	ABCB6 gene on keratinocytes and melanocytes of 6q.	Double-stranded RNA-specific adenosine deaminase is encoded by the ADAR1 gene /DSRA gene.[8]	X-linked: DKC1 AD: TERC leads to the formation of short telomeres and causes cell arrest and apoptosis.	The responsible genes are involved in the functioning of nucleotide excision repair (NER pathway) responsible for the removal and replacement of damaged DNA. Diminished DNA repair in cells exposed to UVB rays. Photosensitivity Carcinogenesis.
Onset	Early age of onset	Early onset in childhood	Early age of onset	Early childhood Heterozygous disorder with seven subtypes (complementation groups A-G) and a variant XP-V
Distribution of lesions	Face, Trunk, limbs. Can occur all over the body (even over palms and soles), but mucous membranes are spared.	Face, Dorsum of hands and feet Palms, soles, and mucous membranes are Normal.	Neck, arms, forearms, and upper chest.	Sun-exposed sites.
Skin morphology	Mottled, interspersed hyperpigmented and hypopigmented macules.[6]	Symmetrical hyperpigmented and hypopigmented macules that increase in size and number as adolescence is reached and later stabilize. Darkening after sun exposure can be observed.	TRIAD Lacy Reticulated hyperpigmented macules (sometimes admixed with hypopigmented macules) Nail dystrophy (pterygium) Mucosal Leukoplakia In some patients, Poikiloderma can be present.	hyperpigmented macules and Lentigines are seen over affected areas.
Pathology	Pigmentary incontinence with normal number of melanocytes.	Basal layer keratinocytes have increased melanin with a normal number of melanocytes. There is pigment incontinence.	Presence of melanophages in the upper dermis.	Hyperkeratosis is noted in the epidermis. There is melanin accumulation in the basal cell layer of epidermis. There can be an increase in the number of melanocytes. The upper dermis shows a chronic inflammatory infiltrate.
Additional cutaneous clinical features	Tuberous sclerosis photosensitivity.	No other additional features noted.	Hair abnormalities squamous cell carcinoma (mouth, anus).	Xerosis Atrophy Photosensitivity pigmentation Telangiectasia Skin tumors

Table 2: Differential diagnosis of reticulate and patterned pigmentary disorders

Disease	**Generalized Dowling degos disease Or Reticulate pigmented anomaly of flexures**	**Incontinentia Pigmenti (bloch Sulzberger)**	**Naegeli Franseshetti syndrome**	**Chronic arsenic toxicity**
Inheritance	AD	X-linked dominant Lethal in utero	AD	Not inherited
Genetics	Loss of function mutations in gene that encode non helical head domain present on keratin 5 that leads to haploinsufficiency. Mutations in POFUT1 and POGLUT1, causes defect in the Notch signaling pathway crucial for melanocyte proliferation and differentiation. Loss of function of these enzymes affects the melanocyte function, such as pigmentation.	X linked: NEMO (NF-κB essential modulator.	17q21	
Onset	Adult age of onset	Occurs at birth	Neck, chest, and abdomen	Depends on the metabolism in body accumulation and removal of arsenic in the target tissues.
Distribution of lesions	flexural areas, trunk, lower and upper limbs.	It is distributed along the lines of Blaschko. It also affects Ocular (mainly 1/3rd of patients), dental, and central nervous system disorders.	Early age	Trunk, flexural creases, any part of the body may be affected.
Skin Morphology	Reticulate hypopigmented and hyperpigmented macules are noted over intertriginous sites and flexors.	4 Stages are present Stage 1 or Vesiculobullous stage (tense bulla develops linearly). Stage 2 – It becomes verrucous as it grows, between 1 to 6 months of age. Stage 3 or Hyperpigmentation stage. Stage 4 leads to Hypopigmentation or Atrophic stage.	Reticular pigmentation noted over body which be brown in colour.	Rains over a dusty road" is the metaphor for guttate hypopigmentation superimposed over hyperpigmentation.
Pathology	Mild hyperkeratosis with thinned-out epidermis, basal hyperpigmentation with downward proliferation of rete ridges.	Hyperpigmentation due to pigmentary incontinence in dermal melanocytes.	Variable pigmentation of basal keratinocytes; pronounced pigmentary incontinence noted.	Increased melanin in epidermis, no melanocytic proliferation
Additional cutaneous clinical features	Pits over palms comedo-like papules, pitted face scars can be seen. Can be associated with Hidradenitis Suppurativa. Broken epidermal ridges café-au-lait macules.	Alopecia nail dystrophy	Hyperkeratosis in palmar and plantar areas. Decreased sweating over affected areas. nail dystrophy can be seen in some cases.	Increased chances of developing internal malignancies.

4. Conclusion

We are presenting this case for its rarity. It is very essential to differentiate between closely related conditions. Though DUH is primarily a cosmetic condition and usually does not affect life expectancy or cause systemic involvement, its psychological impact on patients can be significant due to the visible skin changes. Currently, there is no definitive treatment; however, supportive management and counseling are essential, and cosmetic options may be considered in selected cases.

5. Authors Contributor Roles

1. Dr. Ojasvi Virendra Saoji: Funding acquisition, Writing – original draft.
2. Dr. Gopalakrishnan Kunjaram: Supervision, Writing – review editing.
3. Dr. Jayakar Thomas: Supervision, Writing – review editing.

6. Source of Funding

None.

7. Conflict of Interest

None.

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