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## Case report

### A LATE ONSET CASE OF SPORADIC DYSCHROMATOSIS UNIVERSALIS HEREDITARIA

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## ABSTRACT

Dyschromatosis universalis hereditaria (DUH) is an autosomal dominant inherited rare genodermatosis wherein patient presents with hypopigmented and hyperpigmented macules of varying sizes in a reticulate pattern. We report a rare case of Dyschromatosis universalis hereditaria in a 23 year old male patient with no affected family members suggesting the possibility of sporadic mutation. Patient born of non consanguineous marriage presented with both hypopigmented macules and hyperpigmented keratotic papules with progressive diffuse hyperpigmentation over the trunk and both the extremities. Other system examination was normal. Histopathological examination showed pigment incontinence with collagenisation of the dermis. A diagnosis of Dyschromatosis universalis hereditaria was made based on history, clinical morphology and histopathology.

**Keywords:** Dyschromatosisuniversalis, Reticulate, Genodermatosis, Pigmentation

## INTRODUCTION

Dyschromatosis are pigmentary disorders which presents with both hyperpigmented and hypopigmented macules clinically. Various conditions present with dyschromatosis like genodermatosis, inflammatory skin diseases, infections, drugs, chemical use and nutritional disorders. Two types of dyschromatosis are described namely dyschromatosis universalis hereditaria and dyschromatosis symmetrica hereditaria or acropigmentation of Dohi, wherein there is predominantly acral distribution as opposed to DUH with generalized distribution. Autosomal dominant DUH is a genodermatosis described first by Toyama<sup>1</sup> in Japan in 1929 and again in 1933 in Germany by Ichikawa and Hiraga. It's been suggested that DSH could be a subtype of DUH. Only if, cloning of the unidentified causative genes is done, we can arrive at a conclusion whether DSH is a subtype of DUH or not.

## CASEREPORT

A 23 year old unmarried male born of non consanguineous marriage presented to us with complaints of diffuse darkening of skin and multiple black warty skin lesions along with whitish discolouration over arms, trunk and legs of 5 years duration. Initially, it started over both the legs and gradually progressed to involve the thighs and trunk over last two years. No history of burning or itching sensation on sun exposure. There was no history suggestive of any drug intake or chemical exposure. No other family members suffer from similar skin lesions.

Dermatological examination revealed diffuse hyperpigmentation all over the body with multiple hypopigmented macules and hyperpigmented keratotic papules varying in size from a few mm to 3 mm in diameter (Figure 1,2). His palms, soles and mucous membrane were normal. Ophthalmological

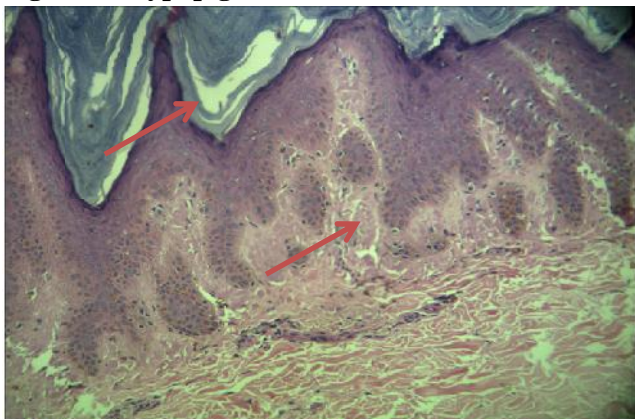
and ENT examinations were normal. Systemic examination was normal. All other routine investigations were within normal limits. VDRL and HIV were negative. A biopsy was taken from two sites: hyperpigmented keratotic papule (A) and the hypopigmented macule (B).



**Fig 1: Hyperpigmented keratotic papules admixed with hypopigmented macules over both the legs**



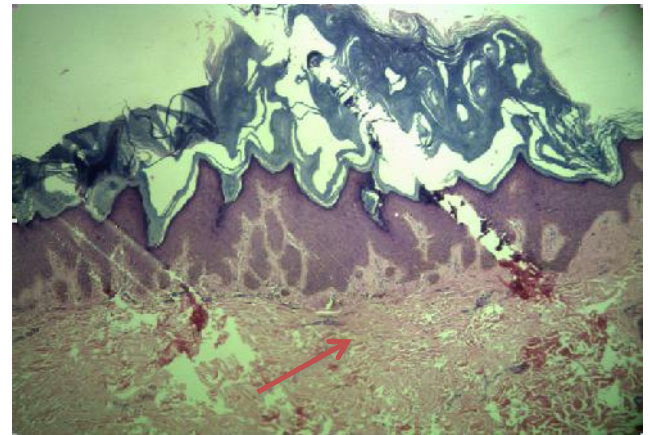
**Figure 2: Hypopigmented macules over the trunk**



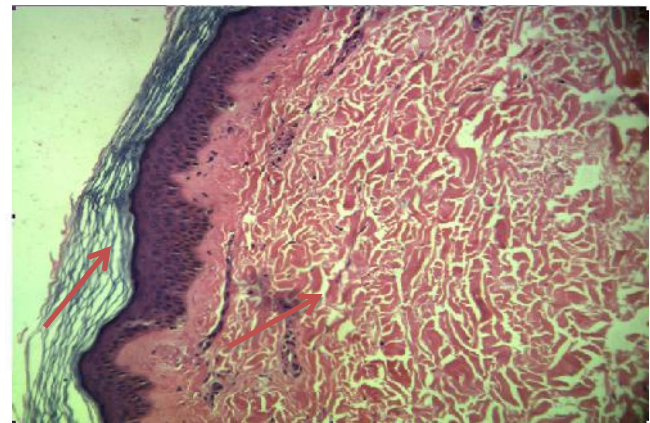
**Fig3: Histopathological examination A – Low power (10X): Hyperkeratotic squamous epithelium with increased basal pigmentation & pigment incontinence in the dermis**

Histopathological examination of the hyperpigmented keratotic papule (A) – (Figure 3,4) showed hyperkeratotic squamous epithelium with increased

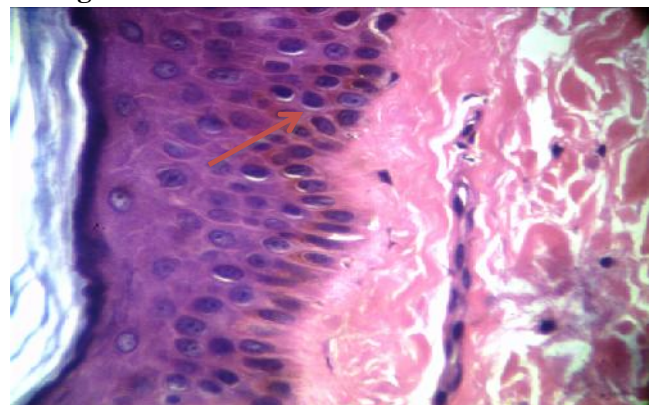
basal pigmentation and pigment incontinence in the dermis with collagenisation of dermis, whereas hypopigmented macule (B) – (Figure 5,6) showed thinning of epidermis with blunting of rete pegs. Collagenisation of dermis is noted.



**Fig 4: Histopathological examination A – Low power(10X): Collagenisation of dermis**



**Fig 5: Histopathological examination B: Low power(10X): Epidermal thinning and dermal collagenisation**



**Fig6: Histopathological examination B: High power(40X): Blunting of rete pegs**

A diagnosis of Dyschromatosis universalis hereditaria (DUH) was made based on history, clinical

morphology and histopathology and the patient was counseled on the benign course of the disease.

## DISCUSSION

Pigmentary dermatosis of reticulate type includes a group of disorder clinically presenting with hypopigmented and hyperpigmented macules. The two major forms are Dyschromatosis symmetrica hereditaria (DSH) and Dyschromatosis universalis hereditaria. Usually DUH has an autosomal dominant inheritance but sometimes it can be inherited recessively also. This disease is most commonly seen in Japan; though there are few case reports from Europe, India and China, fewer cases show familial involvement. The absence of family history in our case suggests a sporadic mutation. Cutaneous clinical morphology shows scattered hypo and hyperpigmented mostly, guttate macules of varying sizes and shapes<sup>2</sup> with irregular border spanning the entire body. Disease presentation is usually during the first few years of life and few cases show late onset of this disease. Most commonly trunk and extremities are affected, the face is rarely involved, though our case presented with diffuse hyperpigmentation. Palms, soles and mucous membrane tend to be spared as in our case, although few isolated cases of palms, soles, oral mucosa and nail involvement have been reported. Various conditions associated with DUH are tuberous sclerosis<sup>3</sup>, Dowling degos disease<sup>3</sup>, X linked ocular albinism<sup>3</sup>, photosensitivity, learning difficulties, insulin- dependent diabetes mellitus, mental retardation and erythrocyte, platelet and tryptophan metabolism abnormalities. Other associations include grand mal epilepsy, high tone deafness and small stature. Most cases of DUH run a benign course.

The pathogenesis of DUH remains unclear. In a genetically prone individual, during the early stages of embryo formation there is a hindrance in the neural-melanocytic interaction. Other explanations put forth for the pigment anomaly in DUH are defective melanosome production and distribution in epidermal melanin units<sup>3</sup> or because of the small number of melanocytes. The ultra structure investigations have shown different levels of melanocyte activity without abnormal pigment production or transfer. Causative DUH gene was mapped to 6q24.2-q25. 2 (OMIM 127500). One of the main differential diagnosis to be considered in

cases of DUH is xerodermapigmentosum as both the condition shows the involvement of sun exposed areas. But in DUH, lesions occur in unexposed sites as well. There is no atrophy or telangiectasia. Most cases don't progress or worsen with age<sup>4</sup>.

Once the disease was thought to be confined to Japanese, now DUH is being increasingly reported in other races as well. This disorder has been reported in two Bantu females by Findlay and Whiting<sup>5</sup> and in an Iraqi girl by Rycroft et al<sup>6</sup>. In view of ruling out another important genodermatosis namely xerodermapigmentosum, DUH gains its importance. Only few isolated case reports have been notified in India so far<sup>7,8</sup>.

## CONCLUSION

As there are only a few case reports from India with no familial involvement and with late onset of disease presentation, our case report of DUH assumes paramount significance

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**Conflict of interest: Nil**

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