Journal of Pakistan Association of Dermatologists. 2016;**26 (2)**:150-153.

Case Report

Rare case report of dyschromatosis universalis hereditaria

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Abstract Dyschromatosis universalis hereditaria (DUH) is a rare genodermatosis reported initially and mostly in Japan. We report a case of DUH in a child with no family history but cosmetic disfigurement and psychological impairment were the presenting symptoms.

Key words
Dyschromatosis universalis hereditaria.

Introduction

Dyschromatoses are a group of disorders characterized by presence of hyperpigmented and hypopigmented macules which are of varying sizes and irregular in shape. It comprises of a spectrum of diseases which include dyschromatosis universalis hereditaria (DUH) or acropigmentation of Dohi and segmental form called unilateral dermatomal pigmentary dermatosis (UDPD).[1,2,3] Dyschromatosis symmetrica hereditaria (DSH) was first reported as a clinical entity by Toyama in 1929.[4] It is characterized by symmetrical distribution of hyperpigmented and hypopigmented macules on the extremities more so over the dorsum of hands and feet.

In 1933, Ichikawa and Hiraga described dyschromatosis universalis hereditaria (DUH) which was basically the same disorder but with distribution pattern all over the body and not merely acral preponderance. It is expressed that DSH may be subtype of DUH but can be affirmed only after gene cloning.

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Case Report

A 14-year-old child, born of non-consanguineous marriage, presented with asymptomatic pigmented macules, which first appeared about 4 years back, primarily over the trunk and progressively involved the whole body, extremities, hands, feet and oral cavity although palms and soles were spared. In due course, hypopigmented macules appeared all over with interspersed hyperpigmented skin. The skin markings over the hypopigmented macules were decreased and dermatoglyphics over tips of all fingers were not very prominent.

There was no history of any previous dermatoses, drug intake, systemic illness, or exposure to chemicals. There was no history of photosensitivity. Developmental milestones were normal and family history was not significant.

Dermatological examination revealed symmetrically distributed reticulated, hyper- and hypopigmented macules over the whole body. (**Figure 1-5**). Hypopigmented macules showed minimal atrophy with total absence of skin markings. There was no erythema or telangiectasia. Buccal mucosa, palatal mucosa and tongue were involved. Scalp hair, teeth and mucosa elsewhere were normal. No abnormality was found in other systems.

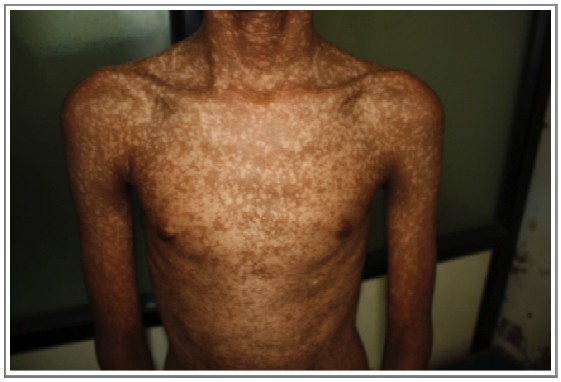

Figure 1 Anterior view of the neck, chest, abdomen and forearm showing hyperpigmented and hypopigmented macules.

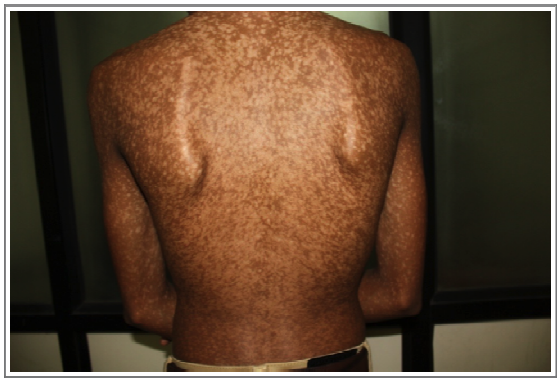

Figure 2 Hyperpigmented and hypopigmented macules on the posterior aspect of trunk.

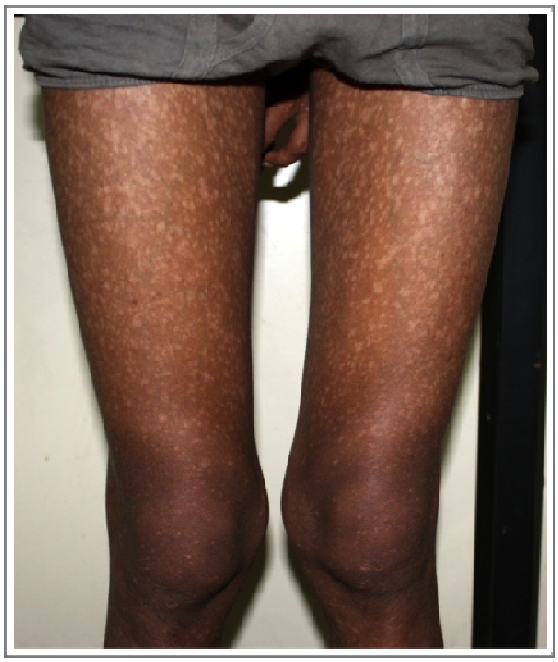

Figure 3 Hyperpigmented and hypo pigmented macule over thighs and knees.

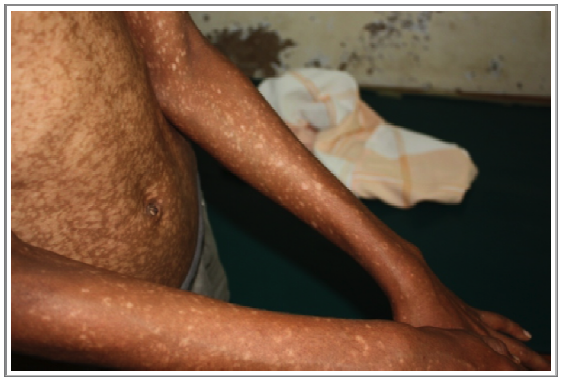

Figure 4 Hyperpigmented and hypopigmented macule over left arm, forearm, hands and abdomen.

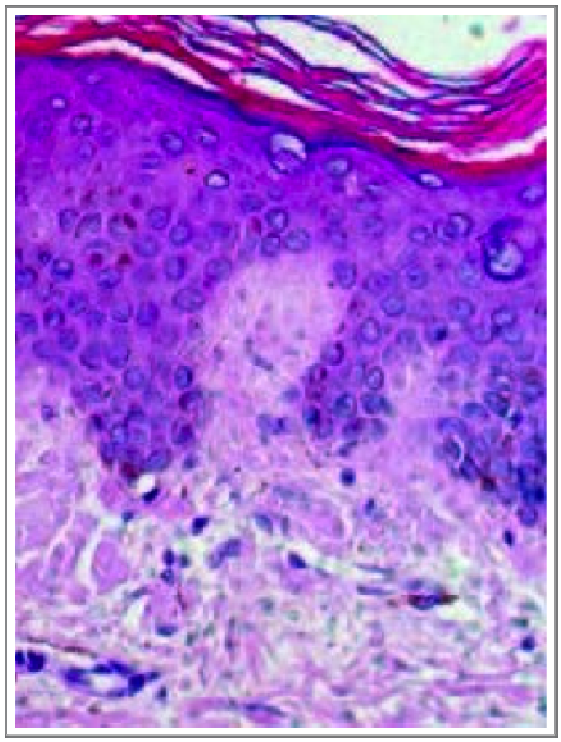

Figure 5 Histopathology of skin showing basal layer pigmentation and pigmentary incontinence.

Histopathology of the skin showed varying degree of basal layer pigmentation and pigmentary incontinence **(Fig. 6)**.

Routine hematological investigations, serum electrolytes, liver function tests, kidney function tests, and urine and stool examination showed no abnormality. Ultrasonography of the abdomen was normal.

Discussion

Reticulate pigmentary dermatoses (RPD) comprise of a heterogeneous group of rare dis-

orders characterized by hyperpigmented macules which coalesce in a reticular pattern, interspersed with hypopigmented macules.[1]

Based on the distribution, morphology and arrangement of the lesions, RPD are divided into two broad categories: acral RPD and generalized RPD. The various acral RPD include reticulate acropigmentation of Kitamura, acropigmentation of Dohi, acral melanosis and heterochromia extremitarium. The differential diagnoses of generalized RPD are dyschromatosis universalis hereditaria (DUH), dermatopathia pigmentosa reticularis (DPR), Naegeli–Franceschetti–Jadassohn syndrome and dyskeratosis congenita (DKC).[1,2,3]

DUH was first described in 1929 by Toyamo,[4]a rare clinically heterogeneous genodermatosis characterized by both hyper- and hypopigmented macules forming a reticulate pattern.[1]

The pathogenesis of DUH is unclear although variable autosomal inheritance has been postulated with occurrence of few sporadic cases[2] and also a probable inherent abnormality of melanosomes or melanin processing.[5]Electron microscopic picture shows that the hyperchromic macules contain numerous fully melanized melanosomes forming melanosome complexes, but the melanosomes are absent from both keratinocytes and melanocytes in the achromic macules.[6]

Dyskeratosis congenita (DKC)is caused predominantly by missense mutations in the DKC1 gene linked to Xq28,8 although autosomal forms may harbour abnormalities in the RNA component of telomerase.[7] Clinically DKC is characterized by a triad of reticulate pigmentation of the skin resembling poikiloderma atrophicans vasculare, nail dystrophy with failure of the nails to form a nail plate and leucokeratosis of the oral mucosa.[5] The lesions usually appear early in the first few months of life; however, sporadic cases can manifest over a period of time as in the present case. DUH may be associated with abnormalities of hair, teeth, nails, and various other systems,[8] high tone deafness,[6] small stature,[6] and ocular albinism. The important essential features of DKC are caused predominantly by missense mutations in the DKC1 gene linked to Xq28,[8] although autosomal forms may harbour abnormalities in the RNA component of telomerase.[9] Clinically it manifests as atrophy, telangiectasia, and pigmentation of skin (poikiloderma), nail dystrophy, and oral leukoplakia.[7] Bone marrow failure and malignancy[7] commonly manifest in the second and third decades. Our patient had no poikiloderma, bone marrow involvement, or signs of malignancy.

Dermatopathia pigmentosa reticularis comprises of the clinical triad of reticulate hyperpigmentation, non-scarring alopecia and onychodystrophy.[7] Other concurrent associations include adermatoglyphia,[10] hypohidrosis[11] or hyperhidrosis,[10]palmoplantar hyperkeratosis and non-scarring blisters on the dorsum of the hands and feet.[11] Our patient had no alopecia, palmoplantar hyperkeratosis, or blistering and had absence of dermatoglyphics only over fingertips.

Naegeli–Franceschetti–Jadassohn syndrome is a type of ectodermal dysplasia which mainly affects sweat glands, nails, teeth, and skin and is characterized by complete absence of dermatoglyphics, reticulate hyperpigmentation that eventually diminishes over the period with advancing age,[12] palmoplantar keratoderma, decreased sweating, enamel defects, dental anomalies, skin blistering, and nail dystrophy.[13] Our case had progressive hypo- and hyperpigmentation, partial loss of dermatoglyphics and no keratoderma or dental enamel anomalies.

Our patient showed features typical of DUH (**Figure 1-5**)with hyper- and hypopigmented macules over the sole with loss of dermatoglyphics over hypopigmented macules.

Despite its rarity, DUH assumes significance as several rare associations are reported.[14]

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