Case 11

A 23-year-old Thai man from Ang Thong

Chief complaint: Skin dyspigmentation for 10 years

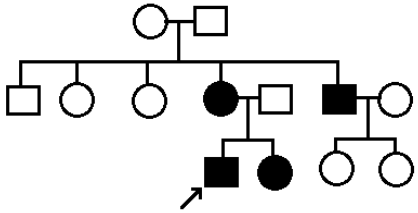
Present illness: The patient developed asymptomatic and slowly progressive abnormality of his skin color since school-aged year. He denied any sun-sensitivity, previous exposure to arsenic or herbal medication. He is otherwise healthy

Personal history:

No current medication.

Family history:

His younger sister, mother and aunt had the same dyspigmented skin



Skin examination:

Generalized spotty hypopigmented and hyperpigmented macules at face, lips, trunk and extremities including acral skin.

Eye, oral mucosa, nail: normal

Histopathology:

S11-00715A:

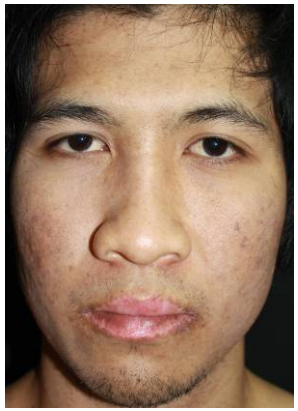
(Masson-Fontana satin from hyperpigmented lesion)

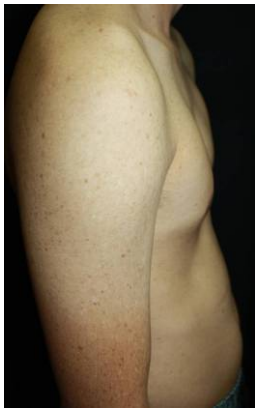
-abundant basilar melanin and melanocytes with some melanophages

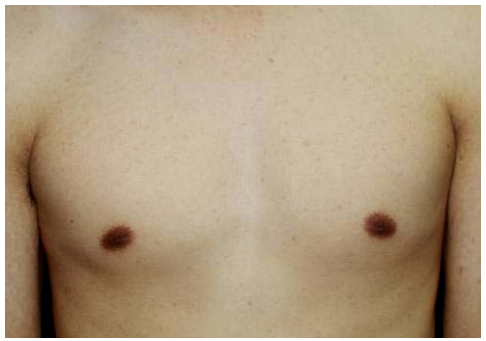
S11-00715B:

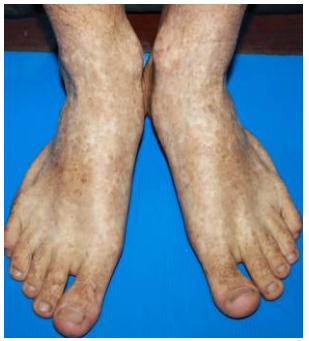
(Masson-Fontana satin from hypopigmented lesion)

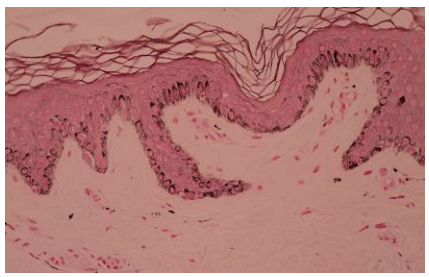
-scant basilar melanin and melanocytes with occasional necrotic melanocytes and few melanophages

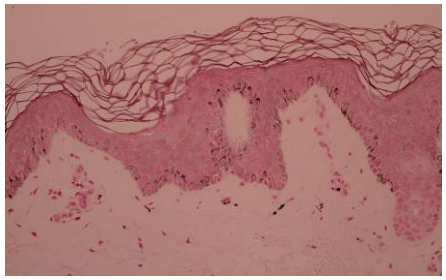












Diagnosis: Dyschromatosis universalis hereditaria (DUH)
Treatment: Patient education, Sun avoidance, Sunscreen
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Consultant: Natta Rajatanavin

Discussion:

Dyschromatosis universalis hereditaria (DUH) is a rare cutaneous disorder with autosomal dominant and sometimes recessive inheritance[1]. It characterizes by development of asymptomatic, hyperpigmented macules, intersperse with hypo- and depigmented macules of varying sizes and shapes. DUH is typically generalized. Facial lesions are seen in 50% of affected individuals[2] but palms, soles and mucosa are rarely involved. However, those uncommon areas have been reported[3, 4]

Table 1 shows differential diagnosis of cutaneous disease with dyschromia. The main differentiation to be made in this case is Dyschromatosis symmetrica hereditaria (DSH or reticulate acropigmentation of Dohi). DSH characterizes by asymptomatic small hyperpigmented macules scatter on the face and hypopigmented macules on the dorsal aspects of the extremities that appear in infancy or early childhood and commonly stop spreading before adolescence [5]. DSH and DUH are thought be two distinct diseases because DSH linked to mutation of the double-stranded RNA-specific adenosine deaminase gene (ADAR1 or DSRAD) on chromosome 1q21.3, while no such mutation found in DUH patients[6]. Histopathology, DUH shows a focal increase or decrease in melanin content of the basal layer depending on the type of lesion biopsied, pigmentary incontinence, and some melanophages in the upper dermis but normal in the number of melanocytes[7]. Hypo-and hyperpigmented lesions of DSH show decrease and increase melanin, respectively[8, 9] with normal number of melanocyte and no melanophage was found. Although our patient's abnormalities predominate at face and extremities, the presence of few truncal lesion and melanophages in upper dermis suggest the diagnosis of DUH rather than DSH.

The pathogenesis of DUH is not known but was proposed by Nuber et al.[10] that DUH is a disorder of abnormal melanosome synthesis rate, represented by higher in fully melanized melanosome numbers in melanocytes and keratinocytes in hyperpigmented skin comparing with hypopigmented skin. Melanocyte numbers are normal in both areas. Autosomal dominant DUH was mapped to chromosome 6q24.2-q25.2, whereas autosomal recessive form was mapped to chromosome 12q21-q23[1].

There is no effective standard treatment for DUH at present. Patient education, reassurance and also suggestion of sun avoidance were done.

Table I Differential diagnosis of dyschromatoses[11].

Location of lesions	Diagnosis	Inheri-tance	Morphology	Pathology
Almost all the body	Dyschromatosis universalis hereditaria	AD*	Motted hyperpigmentation and hypopigmented macules	Increase or decrease in basal melanin, normal melanocyte number, melanophages in the upper dermis[4]
Almost all the body	Amyloidosis cutis dyschromica	AR	Motted hyperpigmented and hypopigmented macules	Amorphous eosinophilic masses (amyloid) in the papillary dermis
Almost all the body	Generalized Dowling–Degos disease	AD	Motted hyperpigmentation and hypopigmented macules, papules	Mild hyperkeratosis, thinned epidermis, downward proliferation of rete ridges, basal hyperpigmentation
Back of hands and feet, face	Dyschromatosis symmetrica hereditaria	AD*	Motted hyperpigmentation and hypopigmented macules	Increase or decrease in basal melanin, normal melanocyte number, no melanophage
Any part of the body	Chronic arsenic toxicity	Not inherited	Guttate hypopigmentation superimposed on hyperpigmentation 'raindrops on a dusty road'	Increased epidermal melanin, no melanocytic proliferation
Sun-exposed areas	Xeroderma pigmentosum	AR	Lentigines and hypoerpigamented macules	Flat epidermis, melanin pigment irregular proliferation of rete ridges, basophilic degeneration of dermal collagen

*Reported case of AR[11] and sporadic [9]

References

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