CASE 11

Patient: A 27-year-old Thai male

Chief Complaint: Dyspigmentation of hands and feet since childhood

Present Illness: Progressive asymptomatic dyspigmentation on dorsum of hands and feet since he was young. Neither history of chemical exposure nor history of erythema was noted preceding the skin lesions.

Past History: No history of UV sensitivity. Previously healthy and not taking any medication.

Family History: No consanguineous marriage in the family. No history of pigmentary skin disorder in family.

Dermatological Examination: Localized hypopigmented and hyperpigmented macules at dorsal of both hands and feet.

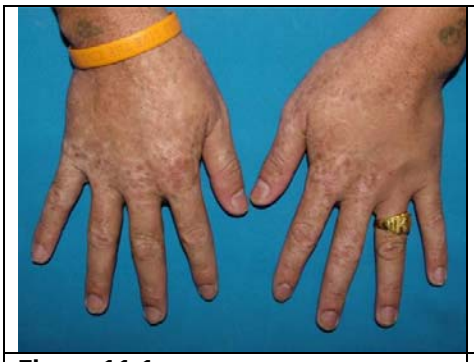

Figure 11.1

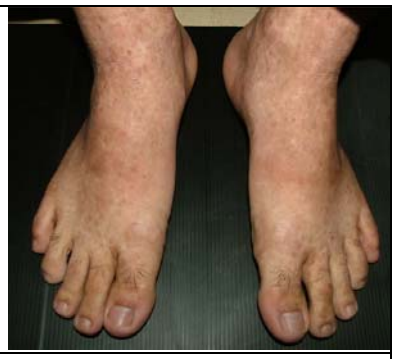

Figure 11.2

Diagnosis: Reticulate acropigmentation of Dohi (Dyschromatosis symmetrica hereditaria)

Treatment: Advice and genetic counseling

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Discussion:

Reticulate acropigmentation of Dohi (RAD) or Dyschromatosis symmetrica hereditaria (DSH) is considered a localized form of dyschromatosis universalis hereditaria, firstly reported by Toyama in 1929. It is characterized by both hyperpigmented and hypopigmented macules of varying size on face and dorsal surfaces of hands and feet, occasionally on the arms and legs.[1]

It is a rare autosomal dominant inheritance pattern with high penetrance. 20% of patients have no family history of the disease and presumably represent spontaneous mutations. However, autosomal recessive inheritance has been reported.[2] The vast majority of reports have come from East Asia (e.g. Japan, Korea, China); nevertheless, this disorder has also been observed in European, Afro-Caribbean and Indian patients.[3, 4]

Mutation of the adenosine deaminase acting on RNA 1 (ADAR1 or DSRAD) gene which causes RAD, were detected in Japanese and Chinese families with this disorder.[5-7]

Patients develop the hyperpigmented and hypopigmented macules in infancy or early childhood.[2] The macules often increase in size and number until adolescence; then, they stabilize and persist indefinitely. It favors the distal extremities and is usually most marked on the dorsal aspect of the hands and feet. It spares the palms, soles and mucous membranes. Spontaneous clearing of lesions has not been observed. The hyperpigmented macules may darken following sun exposure.

There have been non-cutaneous associations reported with RAD. Mostly are neurological disorders including dystonia and idiopathic brain calcification.[8-10] A patient with neurofibromatosis and RAD had been reported.[11]

Histology of hyperpigmented lesions shows increased melanin pigments at the basal layer. Decreased or absent melanin pigment is observed in hypopigmented lesions.

The condition belongs to a group of autosomal dominant reticulate pigmentary disorders that also include reticulate acropigmentation of Kitamura (RAPK), and Dowling-Degos disease (DDD).

RAPK typically presents during the first or second decade of life as a slowly progressive network of hyperpigmented macules on the extensor surfaces of the hands and feet. Punctiform pits and breaks in the epidermal rete ridge pattern are often present. In contrast to the findings in RAD, hypopigmented macules are not observed. The abnormality is characterized histologically by epidermal atrophy and an increased number of basal melanocytes.[2, 12]

DDD [2, 12] is a slowly progressive genodermatosis that manifests as reticulate hyperpigmented macules in flexural areas including the axillae, groin, inframammary areas, neck, and rarely the wrists, face, arms, and scalp. The onset is usually in the fourth decade of life. Dark comedo-like lesions on the neck and pitted perioral scars are other features of this disease. Pigmented filiform epidermal projections are seen histopathologically. Basal melanocytes are unaffected in number.

Other differential diagnoses of the dyspigmentation are dyschromatosis universalis hereditaria,

mild form of xeroderma pigmentosum and pigment disorders due to exposure to chemicals or radiation

Current treatments of RAD are unsatisfactory. Neither PUVA nor topical corticosteroids can improve the dyschromatosis. Split-thickness autografts and laser therapy may improve cosmetically for some patients.[13]

References

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