Universal Small White Macules with Autosomal Dominance in a 19-year-old Female

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CASE REPORT

A 19-year-old girl with skin phototype IV visited our ward complaining of spotted hypopigmentation on her neck, trunk, and extremities (Fig. 1). The history could be traced back to her infancy when a few pin-point whitish spots were noted on her abdomen. Those spots increased in size and number during her childhood with dissemination to her whole trunk, legs and arms. A significant exaggeration of this condition with almost tripled density of these lesions occurred in recent 6 months when a larger amount of sun exposure was noted. However, her face, hands and feet were normal in appearance, and the complexion was the same all over the body.

A closer view showed some densely distributed, sharply-demarcated, off-white macules measuring from 0.1x0.1 cm to 0.3x0.3 cm in size. There was no skin atrophy, erythema, or telangiectasia (Fig. 2). A skin biopsy was performed from her left forearm.

Under light microscope, the epidermis from the hypopigmented skin was devoid of melanin, while the adjacent non-hypopigmented skin contained more melanin in the basal layer than healthy individuals (Fig. 3). This result was further confirmed by Fontana-Masson stain (Fig. 4). There was no atrophy in epidermis, and the dermis was unremarkable.

Throughout the course there was no contributory history noted, and the hair, nails, teeth and mucosae of this girl were normal. Her family history of the same condition was compatible with the autosomal dominant (AD) pattern (Fig. 5).
DIAGNOSIS: Dyschromatosis Universalis Hereditaria

DISCUSSION

Dyschromatosis universalis hereditaria (DUH) is a rare, autosomal dominant genodermatosis included in the spectrum of reticulate pigmentary dermatoses. The typical presentation is generalized and random distribution of small hypo- and hyperpigmented macules with appearance in infancy or early childhood. The skin appendages and teeth are usually normal, and the hands and feet are usually spared. The disease progression usually stagnates before adolescence. The differential diagnosis of DUH includes dyschromatosis symmetrica hereditaria (acropigmentation of Dohi), unilateral dermatoal pigmented dermatosis, dermato-orthia pigmentosa reticularis, Naegeli-Franceschetti-Jadassohn syndrome, dyskeratosis congenita, xeroderma pigmentosum, chronic radiodermatitis, dyschromic amyloidosis, pellagra, and topical application of chemicals.

Our patient was devoid of hyperpigmentation and showed marked disease progression in early adulthood. These presentations were incompatible with the reported typical findings of DUH. However, the diagnosis of DUH was made by the sparing pattern of dorsal hands and feet, the absence of systemic defects, skin atrophy and telangiectasis, the generalized distribution of lesions with no exaggeration on sun-exposed areas, and the negative history of nutritional deficiency and topical chemical exposure. This diagnosis was further supported by pathological findings. The enhanced expression of melanin in normal-pigmented skin may be related to the disease progression rather than solar exposure because the complexion was the same all over the body.

Dyschromatosis universalis hereditaria was firstly reported in Japan. Most of the cases are east Asian, but familial cases recognized in India, Saudi Arabia and Europe have shown DUH is a disease of at least 3 different races. However, there are distinct spite of the same skin phototype, and family members seem to have identical skin manifestations and clinical course. This fact is suggestive of the importance of the genetic role in the pathomechanism.

Recent studies have shown DUH is a disorder of melanosome synthesis rather than melanocyte number. The proposed mechanisms include defects in melanosomal packaging and transfer, and remnants of an evolutionary process that has been mediated by embryonic neural reflexes and is expressed in genetically predisposed individuals and races. Perhaps identification of the causative gene will make this peculiar disease more clear or even give us a practical method for cosmetic use.

REFERENCES