Dowling–Degos disease with dyschromatosis universalis hereditaria-like pigmentation in a family

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ABSTRACT
Dowling–Degos disease is a rare autosomal dominant inherited pigmentary disorder characterized by reticulate pigmentation of the flexures, prominent comedone like lesions and pitted scars. Dyschromatosis universalis hereditaria is characterized by the presence of hypopigmented as well as hyperpigmented macules. We report a family showing features of both these diseases.

Key words: autosomal dominant, dyschromatosis universalis hereditaria, familial, reticulate pigmented anomaly of flexures

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Dowling–Degos disease (DDD) or reticular-pigmented anomaly of the flexures is a rare autosomal dominant inherited genodermatosis. It is characterized by multiple freckle-like hyperpigmented macules mainly affecting the flexures. Additional features include comedones and pitted scars near the angle of the mouth.1,2 Dyschromatosis universalis hereditaria (DUH) is a hereditary skin disease characterized by the presence of widespread hypopigmented as well as hyperpigmented macules.3,4 We report a family who had DUH-like pigmentary alterations in addition to the classical presentation of DDD.

Case reports

Case 1
A 51-year-old female presented with gradually progressive pigmentation in the axillae, groin and neck since early childhood. For the last 3–4 years she had noticed unusual mottled pigmentation on her body. The flat-pigmented lesions first appeared in the axillae, and the pigmentation increased in intensity and similar lesions gradually spread to involve other sites. Examination revealed symmetrical deep brown reticular pigmentation of the neck, axillae and the groin folds (fig. 1). The patient had multiple comedones over the face, neck and trunk along with pitted scars on her chin, angle of mouth and forehead (fig. 2). On the trunk she had multiple hypopigmented macules interspersed with a few hyperpigmented irregular macules (fig. 3). There was no atrophy in any of the lesions. Palms, soles, nails and mucosae were normal. Punch biopsies were taken from both the reticular pigmented lesions on the neck and mottled pigmented macules on the abdomen. The biopsy from the neck revealed pigmented filiform epithelial downgrowths arising from the epidermis. In some areas follicular plugging was evident, while biopsy from the abdomen showed atrophic epidermis with increased pigmentation of the basal layer along with melanin incontinence.
Case 2

The 30-year-old daughter of case 1, had slowly progressive reticular brownish pigmentation involving the neck and axillae since early childhood and had been developing an asymptomatic mixture of hypopigmented and hyperpigmented lesions over her trunk and abdomen over the past year (fig. 3). During the past year she had developed pigmentation on the flexures as well. Her younger sister also had similar lesions in an almost identical distribution, suggesting an autosomal dominant inheritance.

Discussion

DDD was first characterized by Wilson-Jones and Grice in 1978 in their description of 10 patients with the disorder. It is an autosomal dominant inherited pigmentary disorder usually of adult onset, but may occur in childhood. It is characterized by reticular pigmentation of the flexures, which may be associated with dark comedone-like lesions and pitted scars. The pigmentation most commonly affects the axillae, groins, submammary folds and neck but sometimes can spread to involve the face, chest, perineum, natal clefts and wrists. The pigmentation consists of numerous small, discrete, round to oval pigmented macules, which resemble freckles. The confluence of lesions toward the vault of the axillae and the centre of the genitocrural folds was observed by Smith et al. The pigmentation is progressive, symmetrical, often extensive and completely asymptomatic. It is exaggerated by sun exposure.

Comedones and pitted scars most characteristically occur around the lateral margins of the mouth, but can involve other areas of the face, neck, axilla, thighs, etc. DDD has been associated with hidradenitis suppurativa, mental retardation and trichilemmal cysts. DDD has been reported in association with reticulate acropigmentation of Kitamura and is considered to be part of the spectrum of the same genodermatosis.

The affected skin shows pigmented filiform epidermal down-growths arising from the epidermis. An increased number of melanophages has been observed, with no quantitative increase in the number of melanocytes.

Dyschromatosis on the other hand is a rare pigmentary genodermatosis characterized by asymptomatic, irregular, hyperpigmented and hypopigmented macules forming a reticulate pattern. Based on the distribution of these lesions, two major types have been described: dyschromatosis symmetrica hereditaria, localized to the acral areas and dyschromatosis universalis hereditaria, with widespread involvement.

DDD has a wider clinical spectrum than the original descriptions. The biopsy from the abdominal lesions revealed epidermal atrophy and melanin incontinence, as observed in Kitamuras disease, which is already known to overlap with DDD. As the two morphological lesions did not occur alone in other family members, it further reinforces that the trunk lesions were part of the spectrum of DDD rather than a separate entity.

To the best of our knowledge this is the first report of DDD with DUH-like pigmentation in a family apparently following an autosomal dominant inheritance pattern. It remains to be seen if DUH-like pigmentation is also a part of the spectrum of reticulate pigmentary disorders. If further patients like the two index cases are reported, it may bolster this possibility. Alternatively, elucidation of the genetic basis of these conditions may prove or disprove this assertion.

References