Our Dermatology Online

Case Letter

A case of delayed diagnosis of Dowling–Degos disease

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Sir,

Dowling–Degos disease (DDD) is a rare genodermatosis characterized by acquired reticular hyperpigmentation of flexural sites, comedo-like lesions, and pitted facial scars. The classic disease is inherited by the autosomal dominant pattern. Herein, we present here the case of a fifty-year-old female diagnosed with Dowling–Degos disease.

A fifty-year-old female was admitted to our outpatient clinic with a fifteen-year history of brown spots on the neck, face, axilla, wrist, and vulva. Dermatological examination revealed pitted, periorbital scars and pigmented brown macules on the face, axilla, dorsum of the hands, inner face of the wrists, inguinal folds, external genitalia, and multiple, small, brown papules with variable hyperkeratosis on the chest (Figs. 1 and 2). The mucosal membranes, hair, and nails were normal. There were similar pitted lesions in the patient's two children as well. Routine laboratory parameters, including the blood glucose level, lipids, and insulin, were in the normal range. Two punch biopsy specimens were taken from the trunk and axilla. Histopathology of the skin biopsy revealed orthokeratosis on the superficial layer, keratin horns in the epidermis, an elongated and increased melanin pigment on the rete ridges, and mild dermal infiltration (Fig. 3). Based on the clinical and histopathological features, a diagnosis of Dowling–Degos disease was established and referred to the genetics department.

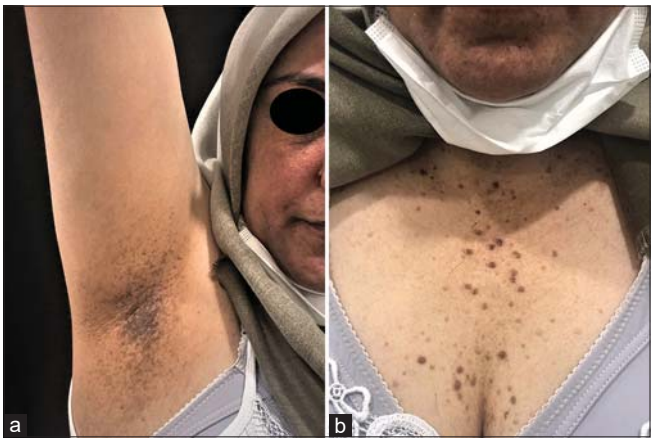

Figure 1: (a) Reticulate hyperpigmentation on the axilla. (b) Multiple, small, brown papules with variable hyperkeratosis on the chest.

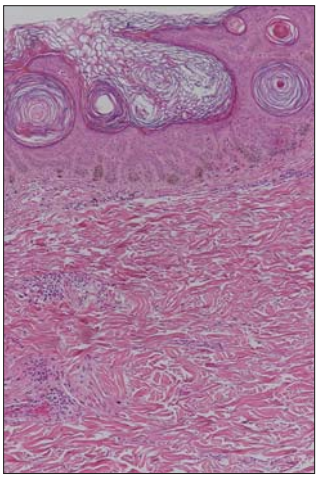

Figure 2: Histopathology revealing keratin horns in the epidermis and an elongated and increased melanin pigment on rete ridges (H&E; 10×).

Dowling–Degos disease, also known as the reticular hyperpigmented anomaly of the flexures, is a rare genodermatosis first described by Dowling and Freudenthal in 1938 distinguished by acanthosis nigricans [1]. DDD is characterized by acquired reticulate pigmentation of the flexures, neck, groin, wrist, face, vulva, and scrotum. Other associated skin manifestations are pitted, perioral, acneiform scars, comedo-like, hyperkeratotic papules, epidermal, trichilemmal cysts, and hidradenitis suppurativa [2,3]. The onset of the disease is typically after puberty and

How to cite this article: Topal S, Topal IO, Bilgi H. A case of delayed diagnosis of Dowling–Degos disease. Our Dermatol Online. 2023;14(1):114-115.

Submission: 18.09.2022; **Acceptance:** 02.12.2022
DOI: 10.7241/ourd.20231.31

the third to fourth decade of life. Mutations in the keratin-5 gene affecting the transfer of melanosomes to melanocytes and keratinocyte differentiation have been found in the pathogenesis of the disease. Atypical clinical presentations are fingernail dystrophy, dyschromatosis universalis hereditaria-like lesions; localized areas such as vulva have been reported in the literature so far [4]. There are several defined dermoscopic features in Dowling–Degos disease, which are irregular, brown pigmentations surrounding a hypopigmented center in a reticular pattern [5]. Pigmented lesions of genital involvement, especially on the vulva, may be rarely seen. Our patient has also genital involvement along with other sites (Fig. 2). Ho Song Kang et al. presented a case of Dowling–Degos disease with vulva involvement [6]. Histopathologically, the disease is characterized by increased pigmentation of the basal layer, thinning of the underlying suprapapillary epithelium, and downward elongation of the rete ridges [7]. Involvement of the infundibulum of the hair follicle is the unique and distinctive feature of the reticular pigmented anomaly [6]. A differential diagnosis should be made from Galli–Galli disease, Haber syndrome, reticulate acropigmentation of Kitamura, dyschromatosis symmetrica hereditaria (acropigmentation of Dohi), and acanthosis nigricans [7]. The most closely related disease is Galli–Galli disease (GGD), which is also an autosomal dominant genodermatosis with loss of function in KRT-5. GGD is clinically indistinguishable from DDD and histologically differentiated with the presence of acantholysis. GGD may also be considered an acantholytic variant of DDD [4]. Reticulate acropigmentation of Kitamura was excluded because of acral involvement and childhood onset. Furthermore, epidermal atrophy was not present in our case, as in Kitamura [7].

DDD is diagnosed based on clinical and histological findings. Numerous treatment options, such as topical hydroquinone, tretinoin, adapalene, and corticosteroids, have been tried for DDD, yet no treatment options have been completely successful in eliminating the lesions. Er: YAG lasers have been proven to be effective by some case reports [7]. Our patient is being followed up with topical tretinoin and methylprednisolone. The results of the genetic samples taken from the patient and her two children have not been concluded yet.

Since Dowling–Degos disease is a rare dermatosis, an average of a hundred familial cases have been reported in the literature so far. We present this case in order to review its diagnosis, emphasize genital involvement, and review its distinction from diseases with reticular pigmentation, particularly in familial cases.

Consent

The examination of the patient was conducted according to the principles of the Declaration of Helsinki.

The authors certify that they have obtained all appropriate patient consent forms, in which the patients gave their consent for images and other clinical information to be included in the journal. The patients understand that their names and initials will not be published and due effort will be made to conceal their identity, but that anonymity cannot be guaranteed.

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Source of Support: This article has no funding source.

Conflict of Interest: The authors have no conflict of interest to declare.