Dowling-Degos Disease in the Anogenital Region

Dowling-Degos disease (DDD) is a benign, rare genodermatosis (reticulate pigmented anomaly) of flexure sites with autosomal dominant inheritance (1,2).The disease is caused by a loss-of-function mutation of keratin 5 (KRT5) present on the chromosome 12q gene (3). It usually affects the younger population, most commonly 20-30 years of age, with some patients being older and with a predominance in the female population (4). The disease is characterized by formation of dark, hyperpigmented macules which are confined to the flexure sites, most commonly over the axillae, groin area, and neck, along with scattered, comedo-like lesions and pitted acneiform scars (3,5). The diagnosis is established based on clinical and histopathological correlation.

We report the case of a 39-year-old patient who presented with a dark brown discoloration of the skin in the area of vulva, perineum, and perianal region (**Figure 1**) with occasional itching sensation that had suddenly appeared a year before presentation at our Department. Additionally, sparce brown macules were found in the left axillary region that had appeared a few months earlier. Histopathology of the skin showed fine and irregular elongation of the interpapillary cones with hyperpigmentation. Based on her clinical presentation and histopathology, the diagnosis of DDD was established. The patient was unsuccessfully treated with adapalene gel and refused the recommended oral retinoid therapy, as well as laser therapy.

Dowling-Degos disease can present as an isolated disease or can be linked to other clinical entities. Usually, it presents with flat macules which are 3-5 mm in diameter and can vary in color from light brown to black (6). Furthermore, the disease is almost always asymptomatic, but pruritus has been reported in some cases (6), as observed in our patient. Even though DDD is primary a disease of the flexures, there have been reports of patients that have presented with hyperpigmented macules on the dorsum of the hands and feet (7). The affected areas in our patient were the anogenital region and left axillary region, and even though this combination of areas is rather uncommon, to our knowledge two similar cases have been reported in the literature (6,8). The most notable histopathological findings of DDD are elongation of rete ridges of the epidermis as well as hyperpigmentation, usually found in the lower third of the elongated rete ridges (6); both of those features were present in the skin biopsy specimen of our patient. Both the clinical picture and pathohistological findings are crucial for the diagnosis of DDD, and we can conclude that the findings of our patient were consistent with DDD.

There are a number of closely related entities to Dowling-Degos disease: Galli-Galli disease (GGD), reticulate acropigmentation of Kitamura (RAPK), Haber disease, and symmetrical acropigmentation of Dohi. Galli-Galli disease has an almost identical clinical presentation, the only difference between those two entities being the presence of acantholysis on biopsy in GGD (9). RAPK presents with hyperpigmentation on the dorsum on the hands and feet, and that pattern has been observed in some patients with DDD as well as GGD (6,7,10-12). However, it differs from DDD in the presence of palmar and plantar pits and slight depression of pigmented lesions (6). Haber disease also has a very similar clinical presentation to DDD, with the presence of dark papules on flexure sites; however, central facial telangiectatic erythema was

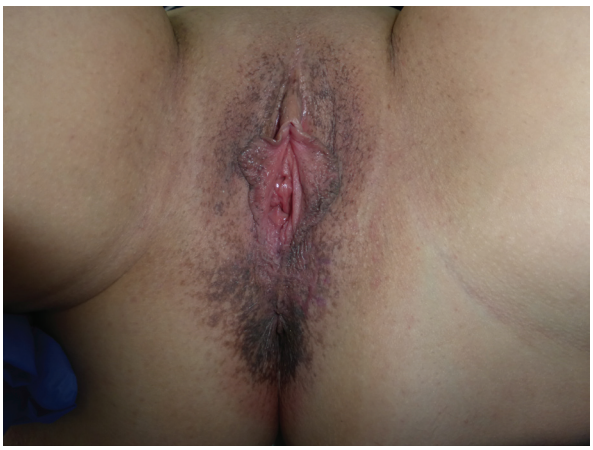

Figure 1. A dark brown discoloration of the skin in the area of vulva, perineum, and perianal region, histologically diagnosed as Dowling-Degos disease.

observed only in Haber disease (13). The clinical features of symmetrical acropigmentation of Dohi are the presence of hyperpigmented macules on the dorsum of the hands and feet, but intermingled areas of hypopigmented macules can also be observed, and the onset of the disease is earlier (infancy and early childhood) when compared with DDD (6,14).

There are no successful treatments for DDD. Topical steroids may reduce the itching. Hydroxyquinone, a topical retinoid (adapalene gel), can be used for fading the pigmentation, but there rapid recurrence was reported when treatment was ceased (15). Systemic retinoids have also been unsuccessful. Er:YAG laser treatment has been reported to be effective, but only in a few cases (6,16,17).

The goal of this paper was to present the case of a patient with DDD on the vulva, perineum, and perianal region as well as to describe the relationship of DDD with other members of the hyperpigmentative disease family.

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Received: February 13, 2021

Accepted: December 1, 2022