CASE REPORT

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Dowling-degos Disease: A Case Report

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Abstract

Dowling-degos disease (DDD) is a rare genodermatose inherited as autosomal dominant trait characterized by brown to black macules located symmetrically in flexural sites. Lesions are not congenital, and the age of onset is highly variable. It is more common in women. Herein, we present the case of a male patient whose clinical and histopathological findings are consistent with DDD and then review existing literature. A 35-year-old male patient presented to the dermatology clinic with black lesions in his flexural sites since childhood. He had no subjective symptoms such as itching or pain in his symmetrically located lesions. His mother and cousins had similar lesions. On the histopathological examination of the lesion sample taken from the inguinal region, fine filiform branchings were found in the epidermis, the rete ridges showed tendency to merge, and there was budding in the rete ridges, which showed hyperpigmentation. The adjacent epidermis showed keratin cysts and mild perivascular mononuclear inflammatory cell infiltration in the superficial dermis. Based on these histopathological findings, the diagnosis was DDD. Fractional erbium YAG laser yielded good clinical outcome.

DDD should be kept in mind in the differential diagnosis of hyperpigmented lesions in flexural sites.

Keywords: Dowling-degos disease, hyperpigmentation, flexural sites

INTRODUCTION

Dowling-degos disease (DDD) is a rare pigmentation disorder and inherited as an autosomal dominant trait (1,2). The real prevalence of DDD is unknown (3). It is characterized by small round symmetrical or asymmetrical dark-brown to black pigmented macules located symmetrically, especially in flexural sites of the axillae, inguinal regions, head, neck, arms, or trunk (1,4). It may be seen in any age group, ranging from early adolescence to young adulthood. Lesions progress slowly over the years (5).

CASE PRESENTATION

A 35-year-old male patient presented to the dermatology clinic with symmetrically located black lesions on eyelids, axillae, and inguinal regions. His history revealed that the lesions were present since he was a child and did not cause subjective symptoms such as itching or pain nor regressed spontaneously. He had no complaints other than recurrent froncules in the inguinal region, and he had no history of chronic illnesses, smoking, alcohol consumption, or any medication. As regards family history, his mother and cousins had experienced similar lesions. Systemic examination and routine laboratory results were normal. Dermatological examination revealed smooth, contoured dark-brown to black macules of 2-4 mm in diameter located in eyelids, perineum, and inguinal region (Figure 1). Histopathological examination of the lesion sample taken from the inguinal region showed basket-wave orthokeratosis, fine filiform branching of the epidermis, tendency to merge in the rete ridges, and budding of the rete ridges, which showed hyperpigmentation. The adjacent epidermis showed keratin cysts and mild perivascular mononuclear inflammatory cell infiltration in the superficial dermis (Figure 2, 3).

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Despite different clinical presentation findings, reticular pigmentation disorders were taken into consideration in the differential diagnosis. Among these disorders, Galli-Galli disease was excluded because there was no suprabasal acantholysis and parakeratosis in the epidermis, Kitamura's reticular acropigmentation was rejected by the lack of atrophy in the epidermis and its specific clinical site in acral regions, and Haber syndrome was ruled out by the absence of keratotic follicles, erythema, and telangiectasias on the face. Based on his histopathological and clinical findings, he was diagnosed with DDD.

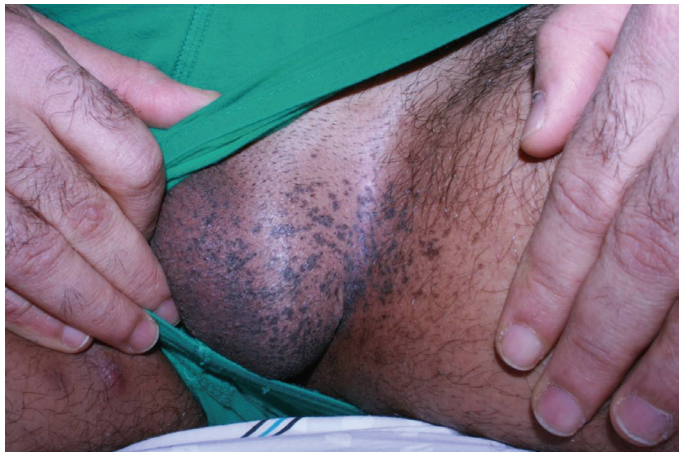
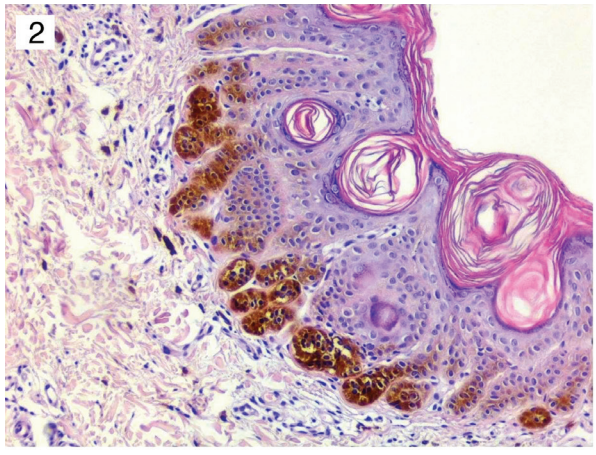


Figure 1. Dark-brown to black macules located in the inguinal region



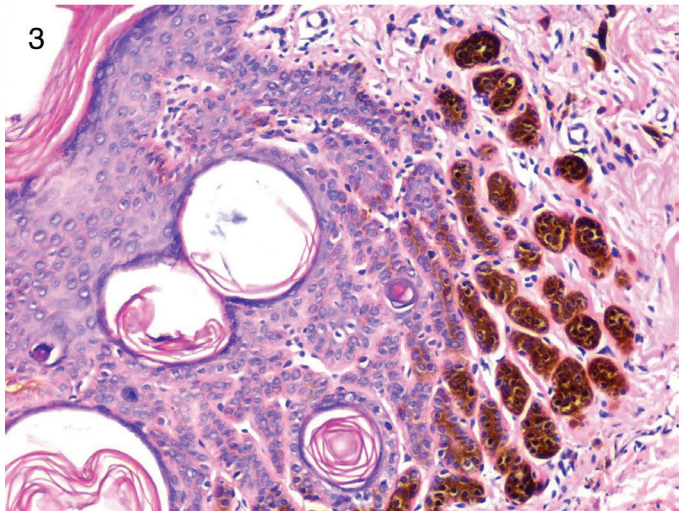


Figure 2, 3. Budding of the rete ridges, which showed hyperpigmentation. The adjacent epidermis showed keratin cysts and mild perivascular mononuclear inflammatory cell infiltration in the superficial dermis (hematoxylin and eosin staining, x200)

Two sessions of fractional erbium YAG [(Er): YAG] laser was applied to axillary lesions, which had provided good clinical outcome (Figure 4).

Informed consent was taken from the patient for the publication of this case and any accompanying images.

DISCUSSION

Recent studies have focused on the mutations of the *keratin 5* gene *(KRT 5)* in the pathogenesis of DDD (6,7). The *KRT 5* gene plays an important role in the cell-to-cell adhesion of the keratinocytes, transfer of melanosomes into the keratinocytes, transportation of organelles, and support of the nuclear structure. Because of the mutations in this gene, hyperpigmented reticular macules are seen in the body, especially in the flexural sites (6).

Although sporadic cases are reported, the patients mostly have a family history, as seen in our case (8). DDD is thought to be inherited as an autosomal dominant trait, but in published series and cases, it is more commonly seen in women than in men. As a rare disorder, only a few cases are reported in our country, and the number of the affected women appeared higher than that of men (4,9). In our case, the patient is also a male.

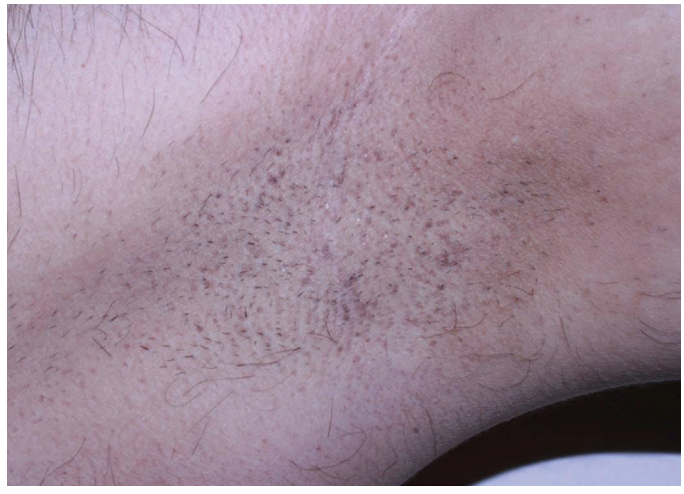


Figure 4. Axillary region after treatment

The age of disease onset is variable. It may be seen in any age group, ranging from early adolescence to young adulthood. Most of the cases are seen within the fourth decade (1). In our case, the lesions were present since childhood.

In DDD, lesions arise from the flexural sites and slowly spread to other sites. The axillae, inguinal regions, face, neck, arms, and trunk are mostly commonly affected. Other clinical findings that are less commonly seen in patients with DDD are atrophic scars around the mouth, comedone-like lesions in the face, neck and trunk, and epidermal and trichilemmal cysts (1,2). None of these findings were observed in our patient.

Histopathological findings specific to DDD are elongation of the rete ridges, filiform formation, interconnecting branching, and basal hyperpigmentation. Follicular infundibulum may be involved. Moderate orthokeratosis or hyperkeratosis, melanophages in the papillary dermis, and mild perivascular mononuclear inflammatory cell infiltration may be seen (1). Most of the defined findings were also seen in our case.

Other reticular pigmentation disorders such as Galli-Galli disease, Kitamura's reticular acropigmentation, and Haber syndrome were taken into consideration in the differential diagnosis (5,10). In Galli-Galli disease, which is a variant of DDD, besides the characteristic histological findings, suprabasal dyskeratotic acantholysis is also present. In Kitamura's reticular acropigmentation, atrophic hyperpigmentated papules begin on the outer surfaces of acral areas such as in hands and feet. In Haber syndrome, the first lesions are facial eruptions. Some studies have shown that all these diseases are inherited as autosomal dominant trait and may show similar clinical and histopathological findings and a patient may present with more than one of these diseases; therefore, these diseases should be evaluated within the same spectrum (11). The differential diagnosis also includes neurofibromatosis type 1 and acanthosis nigricans (3). Although the lesions in the axillary and inguinal areas in neurofibromatosis type 1 may show similarity to DDD, the presence of multiple neurofibromas is one of the important findings in the distinction. In acanthosis nigricans, there is no reticular pigmentation, as seen in DDD.

Till date, DDD has no known definitive treatment. Some studies have reported that topical steroids, azalaic acid, topical retinoids, depigmentation agents, and laser therapies especially CO_2 or Er: YAG laser are effective treatment of DDD (8,12,13). In the present case, two sessions of fractional Er: YAG laser was applied to the axillary as treatment and had provided good clinical outcome.

CONCLUSION

Since DDD is a rare disease that predominantly affects women and reported cases are limited, this condition should be kept in mind in the differential diagnosis of hyperpigmented lesions of flexural sites, not only in women but also in men. As there is no known definitive treatment, development of new treatment modalities is warranted.

Ethics

Informed Consent: Informed consent was taken from the patient for the publication of this case and any accompanying images.

Peer-review: Externally and internally peer-reviewed.

Authorship Contributions

Surgical and Medical Practices: S.Ş.E., T.Y., P.E.Z., Concept: S.Ş.E., T.Y., P.E.Z., Design: S.Ş.E., T.Y., P.E.Z., Data Collection or Processing: S.Ş.E., T.Y., P.E.Z., Analysis or Interpretation: S.Ş.E., T.Y., P.E.Z., Literature Search: S.Ş.E., T.Y., P.E.Z., Writing: S.Ş.E., T.Y., P.E.Z.

Conflict of Interest: No conflict of interest was declared by the authors.

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