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DOWLING DEGOS DISEASE: A RARE GENODERMATOSIS

Dermatology

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ABSTRACT

Dowling-Degos disease (DDD) is a rare autosomal dominant disorder, classically characterized by acquired reticular hyperpigmentation in flexural sites. Onset is typically after puberty and commonly occurs in the third to fourth decade of life. Mutations in genes involved in melanosome trafficking and melanocyte and keratinocyte proliferation, differentiation, and cellular communications have all been implicated as etiologies. Hyperpigmentation is often recalcitrant to treatment, as evidenced by the varying success of topical and laser therapies.We report a 42-year-old female presented with dark lesions which initially developed over the hands and face and gradually progressed to involve the axillae, inframammary folds, neck, groin, forearms, hands, and feet since 11 years of age.

KEYWORDS

autosomal dominant, flexural sites, reticulate pigmentary disorder

INTRODUCTION

Dowling–Degos disease (DDD) is a rare, autosomal dominant pigmentation disorder marked by reticulate hyperpigmentation, typically emerging after puberty and worsening over time. Patients may also experience itching, burning, or inflammation. Histologically, DDD features thinned suprapapillary epidermis, elongated rete ridges, and basal melanin deposition.Though once thought to result from a single gene, DDD is now known to be genetically heterogeneous, with five identified causal genes: KRT5, POFUT1, POGLUT1, PSENEN, and GLMN. Each gene correlates with specific clinical patterns—for example, KRT5 affects flexural areas, POGLUT1 the extremities, and PSENEN is linked to hidradenitis suppurativa. GLMN mutations may lead to glomuvenous malformations.There is no curative treatment, though ablative laser therapy can reduce pigmentation, with risks like postinflammatory hyperpigmentation.[1]

Here we report a 42-year-old female with typical flexural involvement and acral sites were also affected, showing its clinical variability.

CASE REPORT

A 42-year-old female presented with asymptomatic dark-colored lesions since 11 years of age. Lesions initially appeared over the hands and face and gradually progressed to involve the axillae, inframammary folds, neck, groin, forearms, hands, and feet.

Family history revealed similar lesions in her mother and siblings, suggestive of autosomal dominant inheritance

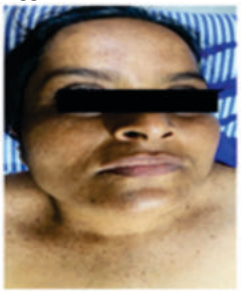
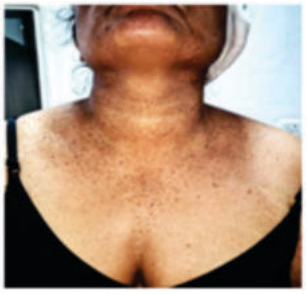



Fig 1 & 2 shows multiple, discrete to confluent brown to dark brown macules arranged in a reticulate pattern over the face, V of neck

On Examination:

Multiple discrete to confluent brown macules arranged in a reticulate pattern over face, V of neck, anterior and lateral aspect of neck, both axilla ,flexor and extensor aspect of both forearms, dorsal aspect of both hands, inframammary area, anterior and medial aspect of thighs,region around both popliteal fossae, dorsum of both feet.

Dermoscopy demonstrated an irregular reticular network with follicular plugging.

On Histopathology – Epidermis is thinned out with features of elongation of rete ridges. Also noted thinning of suprapapillary dermis. Dermis shows sparse inflammatory cell infiltration form of lymphocytes. Increase amount of melanin is noted in stratum basal layer.

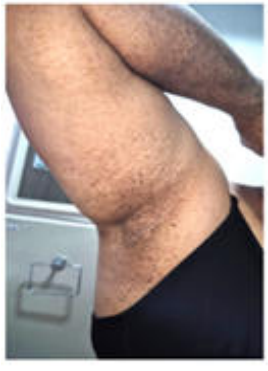
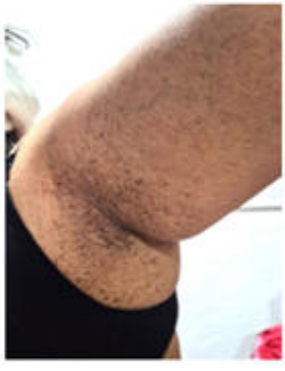



Fig 3& 4 shows multiple, , discrete to confluent brown to dark brown macules arranged in a reticulate pattern over both axill

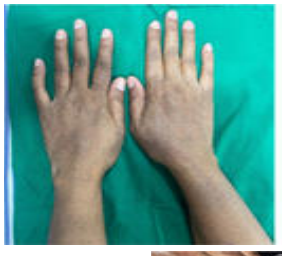
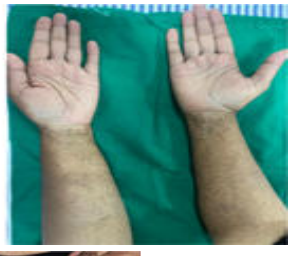
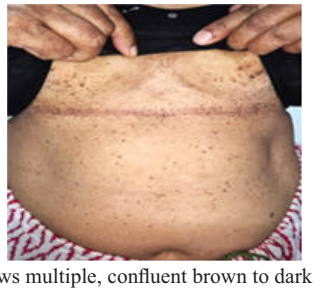
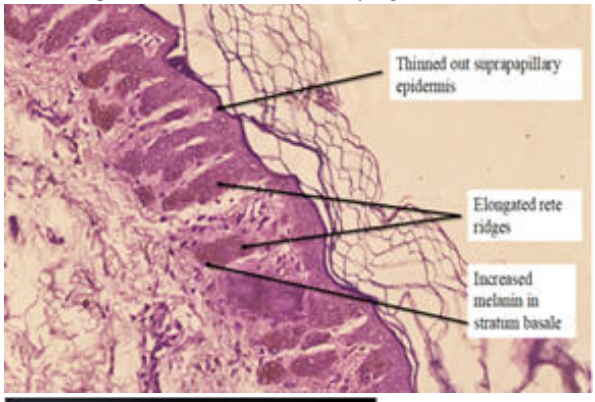




Fig 5,6 & 7 shows multiple, confluent brown to dark brown macules

arranged in a reticulate pattern over dorsum of both hands, flexor and extensor aspect of forearms, inframammary region.



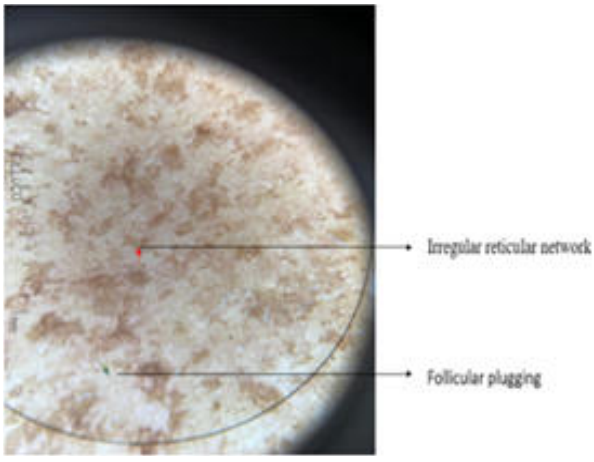


Fig 8&9 shows histopathological and dermoscopic findings.

DISCUSSION

Dowling–Degos disease (DDD) is a rare autosomal dominant genodermatosis characterized by reticulated hyperpigmentation predominantly affecting flexural areas such as the axillae, groin, and inframammary regions. The condition usually manifests after puberty, most commonly in the third or fourth decade of life, and follows a slowly progressive course. Patients may report progressive darkening of skin folds, occasionally accompanied by pruritus or irritation, with lesions sometimes extending to the neck, trunk, and inner thighs.

DDD is genetically heterogeneous and associated with mutations in genes involved in epidermal structure and pigmentation, particularly KRT5, which plays a critical role in keratinocyte integrity and melanosome transport. Additional genes implicated in DDD include POFUT1, POGLUT1, PSENEN, and GLMN, which are associated with distinct subtypes and overlapping clinical syndromes, such as hidradenitis suppurativa and glomuvenous malformations. Many of these mutations affect the Notch signaling pathway, which is essential for keratinocyte and melanocyte proliferation, differentiation, and intercellular communication.

The pathogenesis of DDD involves defective melanosome trafficking and abnormal keratinocyte–melanocyte interactions, resulting in characteristic reticulated pigmentation. Histopathologically, DDD is characterized by elongated, branching (antler-like) rete ridges, thinning of the suprapapillary epidermis, and increased basal layer pigmentation. Additional findings may include dermal melanophages and a mild perivascular lymphohistiocytic infiltrate. The Galli–Galli variant is distinguished by suprabasal acantholysis without dyskeratosis.

Clinically, DDD presents with lentigo-like macules, brown papules, and reticulated hyperpigmented patches in intertriginous areas. Comedone-like lesions on the back and neck, perioral pitted scars, and palmar pits may also be observed. The disease is progressive but shows no gender or racial predilection.

Diagnosis is based on characteristic clinical features and confirmed by histopathological examination of a skin biopsy. Genetic testing may aid in subtype classification and screening for associated conditions, although routine laboratory or imaging investigations are not required. Treatment remains challenging, as no definitive cure is available. Topical depigmenting agents, retinoids, and corticosteroids have shown variable and limited efficacy. Laser therapies, such as Er:YAG laser, may provide cosmetic improvement; however, recurrence and post-inflammatory hyperpigmentation are common. Management is therefore focused on symptomatic relief, cosmetic concerns, and genetic counseling.

CONCLUSION

This case highlights a rare pigmentary disorder with typical flexural involvement- Dowling Degos Disease. In this case, acral sites were also affected, showing its clinical variability. Reporting such cases is important to avoid misdiagnosis with other reticulate pigmentary disorders and to highlight the role of clinicopathological correlation and family history in reaching the correct diagnosis.

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