



## DOWLING-DEGOS DISEASE: A CLINICO-HISTOPATHOLOGICAL CASE SERIES

## Dermatology

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## ABSTRACT

Dowling Degos disease is a rare genodermatosis with autosomal dominant inheritance. In this case series, we report 3 cases of Dowling Degos disease which were evaluated based on their clinical and histopathological findings. The patients presented with similar findings such as reticulate hyperpigmentation of the flexures, hyperpigmented papules and comedo-like lesions. Family history of similar lesions was present. On histopathology, elongation of rete ridges and hyperpigmentation of the basal layer was seen in all the 3 cases which are the characteristic features of DDD. Hence, in our case series clinical examination and histopathological examination played a vital role in establishing the diagnosis of Dowling-Degos disease.

## KEYWORDS

## INTRODUCTION

Dowling Degos disease is a rare genodermatosis with autosomal dominant inheritance. It was first described by Dowling and Freudenthal.<sup>1</sup> Onset commonly occurs in the third to fourth decade of life.<sup>2</sup> The pathophysiology of this condition has been linked to mutations in genes that impact melanosome transfer, melanocyte and keratinocyte development. KRT5, POFUT1, POGUT1, and, most recently, PSENEN are some of these genes.<sup>3</sup> Reticulate hyperpigmentation in a flexural pattern, comedo-like lesions, and pitted perioral acneiform scars are its distinguishing feature. Histopathology reveals an "antler horn" or "stag horn" pattern due to the finger-like elongation of rete ridges.<sup>4</sup>

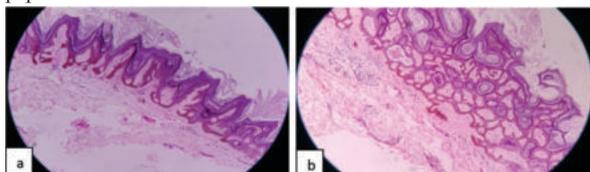
## Report:

## Case 1:

A 62-year-old male patient presented with reticulate hyperpigmentation over neck and upper back; multiple hyperpigmented papules over face, neck, flexures of limbs since 30 years of age. All these lesions were asymptomatic. Similar complaints were present in mother, siblings and his daughter. Histopathological examination revealed hyperkeratosis, thin filiform downgrowth of basal layer which was extensively pigmented. Horn cysts were noted. Dowling-Degos disease was confirmed. In view of cosmetic concern, patient was treated with topical adapalene gel 0.1% following which mild improvement was noted.



**Figure 1:** 62 years old male patient showing  
**Figure 1a:** showing reticulate hyperpigmentation over the neck and multiple hyperpigmented papules on the trunk  
**Figure 1b:** reticulate pigmentation and multiple hyperpigmented papules over axilla



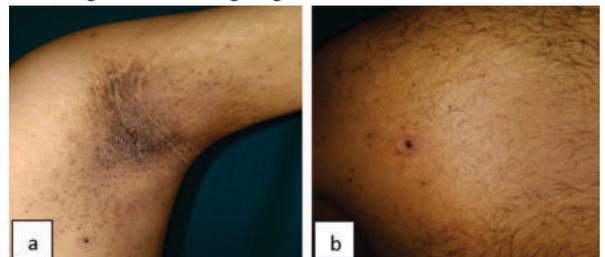
**Figure 2:** Histopathological examination of skin

**Figure 2a:** hyperkeratosis, thin filiform down growth of basal layer which was extensively pigmented

**Figure 2b:** Horn cysts seen

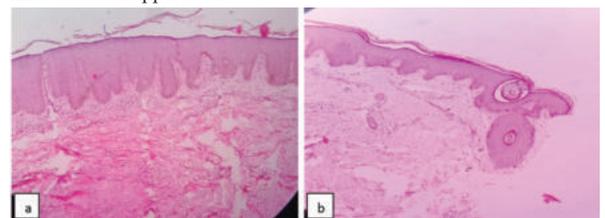
## Case 2:

A 23-year-old male presented with complaints of multiple papules and comedo-like lesions over bilateral axillae, neck, back and bilateral arms since 10 years. History of similar lesions was present in family members- father and grandmother. Histopathological examination revealed acanthosis with elongation of rete ridges, hyperpigmentation. Thus, diagnosis of Dowling-Degos disease was established.



**Figure 3:** 23 years old male patient showing  
**Figure 3a:** showing reticulate pigmentation and multiple hyperpigmented papules on axilla

**Figure 3b:** showing multiple hyperpigmented papules and comedo-like lesions on upper back



**Figure 4:** Histopathological examination of skin of a 23-year-old male patient revealing

**Figure 4a:** acanthosis with elongation of rete ridges and hyperpigmentation

**Figure 4b:** epithelial strands extending into the superficial dermis resulting in an 'antler-like' pattern

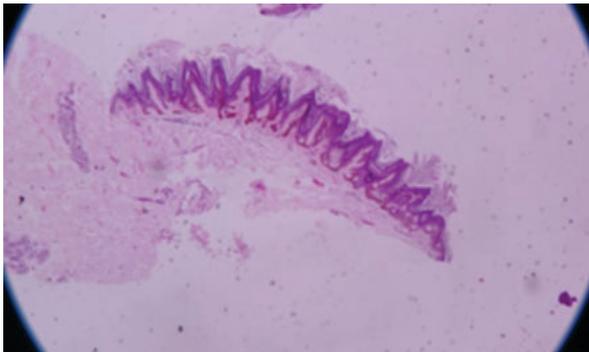
## Case 3:

An 80-year-old female patient presented with comedo-like lesions and hyperpigmented papules over face, trunk and bilateral upper limbs. Similar lesions were present in son and grand-daughter. Histopathology revealed elongation of rete ridges with hyperpigmentation of basal layer.

In the above cases, diagnosis of Dowling-Degos disease was established based on the clinico-histopathology.



**Figure 5:** 80 year old female patient showing reticulate hyperpigmentation and hyperpigmented papules involving the antecubital fossa



**Figure 6:** Histopathological examination of skin of a 80 year old female patient revealing elongation of rete ridges with hyperpigmentation of basal layer.

## DISCUSSION

Dowling-Degos disease (DDD) is a rare autosomal dominant disorder, classically characterized by acquired reticular hyperpigmentation in flexural sites.<sup>1</sup> Less than 50 cases of DDD have been documented in the literature since the condition was first diagnosed in 1938. DDD is caused by loss of function of keratin protein due to mutations in the gene that encodes keratin 5 which disrupts the arrangement of the keratin intermediate filaments. Additionally, decreased keratin 5 prevents transfer of melanosomes to keratinocytes, which results in the pigmentary symptoms of Dowling-Degos illness.<sup>5</sup> In our study, all the three cases complained of hyperpigmented papules and comedo-like lesions were reported in 2 cases. Positive family history of similar lesions was common for all the three cases. Different diagnostic modalities for DD is based on patient's history, clinical and physical examination and can be confirmed by histopathological examination.<sup>4</sup> In our case series, elongation of rete ridges and hyperpigmentation of the basal layer were the consistent histopathological findings. These elongated rete ridges are referred to as an "antler-like" pattern which are the characteristic features of DDD<sup>4</sup>. Different treatment option for DDD include systemic and topical retinoids, Adapalene, hydroquinone and Laser therapy: combination of Q-switched Nd: YAG and fractional carbon dioxide laser and intense pulsed light. In our study, first case was treated with topical adapalene gel 0.1% following which mild improvement was noted. Acanthosis nigricans is the commonest differential diagnosis for DDD and may be distinguished clinically based on the presence of velvety plaques; histologically by less pronounced elongation of the rete ridges. In addition, there is no follicular involvement. Other differential diagnosis include Neurofibromatosis type 1 which shows freckling in the axillae and groin, confluent and reticulate pigmentation and reticulate acropigmentation of Kitamura.

## CONCLUSION

Dowling-Degos disease is a rare genodermatosis. Though it affects a very less population of the worldwide without any significant mortality, affected patients may experience significant distress and anxiety due to the skin manifestations. This leads to decrease in the quality of life. Our case series highlighted the importance of clinical

examination and the vital role of histopathological examination in establishing the diagnosis of Dowling-Degos disease which helped in treatment.

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