Case report

A rare case report of Dowling- Degos disease: Reticulate acropigmentation of kitamura overlap

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Abstract

Dowling Degos disease (DDD) and Reticulate Acropigmentation of Kitamura (RAPK) belong to a rare group of autosomal dominant inherited reticulated pigmentary disorders. We are reporting a case of a 38 year old female patient who presented to us with multiple asymptomatic dark coloured lesions over the dorsa of the hands and feet, axilla, groin, periorbital region, front and sides of neck. There were also multiple dark comedo-like lesions, perioral pitted acneiform scars and multiple palmar pits. Based on the clinical and histopathological findings, a diagnosis of an overlap of DDD and RAPK was made. Such overlaps of features of reticulate pigmentation have been rarely mentioned in the literature.

Keywords: Dowling Degos disease, Reticulate Acropigmentation of Kitamura, reticulate pigmentation, overlap

Introduction

Dowling Degos disease (DDD) and Reticulate Acropigmentation of Kitamura (RAPK) are a group of rare autosomal dominant genodermatosis, clinically characterized by symmetric, asymptomatic reticulated pigmented macules affecting the flexures in the former and dorsa of hands and feet in the latter.[1] Additionally, DDD also manifests with scattered comedo-like lesions(dark dot follicles) and pitted acneiform scars.[2] We herein present a case with overlap features of DDD and RAPK which is rare.

Case report

A 38 year old female patient, presented to us with asymptomatic dark coloured lesions over the hands since fifteen years. The lesions progressively increased in number to involve the neck, trunk and other parts of the body like axilla and groin. She also complained of multiple asymptomatic pits around the mouth since twelve years. There was history of similar lesions in her son, but he was not available for examination. There was no history of consanguinity.

General physical examination and systemic examination did not reveal any abnormality. Cutaneous examination revealed multiple pits of varying sizes from 1-3mm around the perioral region (Figure-1) with dark comedo-like lesions distributed over the chin, cheek, neck, axilla, cubital fossae, back and abdomen (Figure-2). There were multiple hyperpigmented macules in a reticulate pattern over the dorsa of hands (Figure-3), forearms, front and sides of neck, periorbital region and axillae (Figure-4). Few tiny palmar pits were also noted.

The skin biopsy, taken from a hyperpigmented macule over the dorsum of hand revealed elongation of rete ridges with branching and increased melanin pigmentation in the lower part of the rete ridges. Few pseudo horn cysts were also seen (Figure-5).

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Discussion

DDD and RAPK are classified under reticulated pigmentary disorders, which follow an autosomal dominant pattern of inheritance. There have been a few reports in literature of patients exhibiting features of both DDD and RAPK, indicating that they may be the same disease with variable phenotypic expression.[3]

DDD was first described by Dowling in 1938 and Degos in 1954. It is mainly characterized by reticulated hyperpigmented macular lesions predominantly distributed over the flexures (neck, axilla, cubital fossa, groin).[4] The degree of pigmentation varies and in some patients, it may be confluent. It is also characterized by open comedo-like lesions of the face and neck and pitted perioral acneiform scars. Hair and nails are normal. The genetic defect in DDD is attributed to the loss of function mutations in the keratin 5 gene (KRT5) situated in the keratin gene cluster on chromosome 12q13, resulting in haploinsufficiency.[5] Interestingly, similar pigmentary changes are also observed in epidermolysisbullosa simplex with mottled pigmentation, which is as a result of a specific KRT5 gene mutation. Recently, another genetic defect of DDD has been reported in the gene locus mapping to chromosome 17p13.3 and chromosome 1q21 with mutations located in the DSRAD gene.[6] Galli- Galli disease is a rare acantholytic variant of DDD.[7]

RAPK is mostly reported in Asian ethnic groups. It usually develops during the first and second decades of life and is characterized by reticulate hyperpigmented macules over the dorsa of hands and feet with few palmoplantar pits.The reticulate pigmentation may sometimes gradually involve other parts of the body as well. Sunlight may play a role in aggravating the condition and the lesions gradually darken over time.[8]

Histopathology reveals elongation of rete ridges with increased melanin pigmentation at their tips, which is similar to DDD. Occasionally few horn cysts may be seen.[9]

Our patient had diffuse reticulate pigmentation of face, flexures with dark comedo-like lesions and perioral pitted acneiform scars resembling DDD and reticulate acral pigmentation over the dorsa of both hands and feet with few palmar pits resembling RAPK.

There are no effective treatments for these conditions. Topical adapelene, 20% azelaic acid, systemic retinoids and Erbium doped yttrium aluminum garnet(Er:YAG) laser have been tried as therapeutic options.[10]So, our patient had an overlap of features of DDD and RAPK, which has been rarely mentioned in the literature.

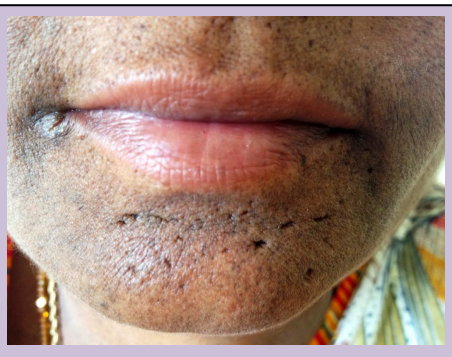

Figure 1: Perioral pitted scars

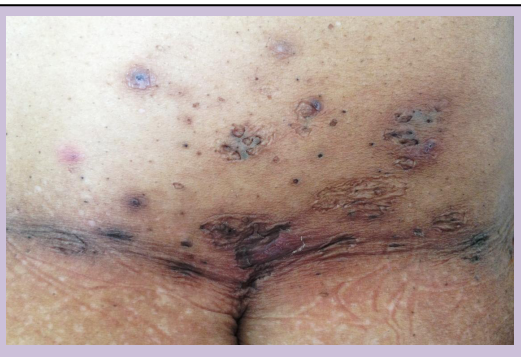

Figure 2: Comedo-like lesions in

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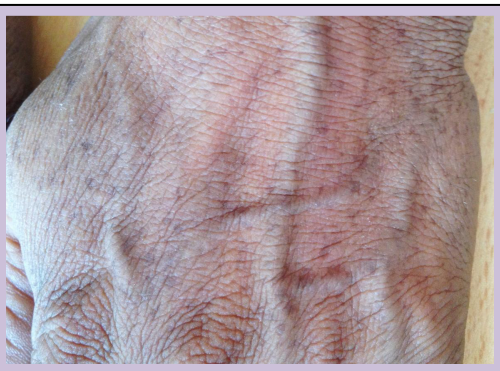

Figure 3: Reticulate hyperpigmented macules

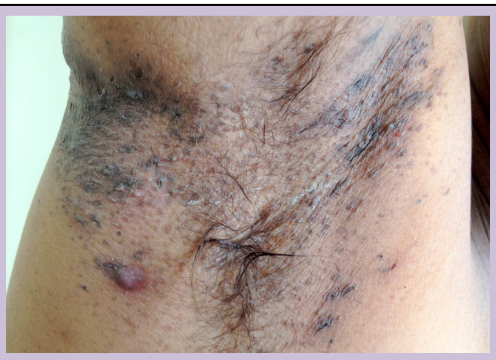

Figure 4: Reticulate hyperpigmentation in

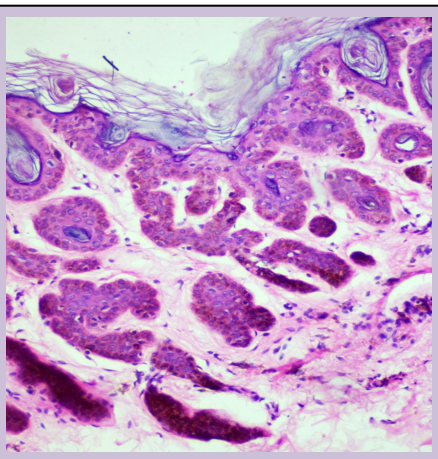

Figure 5: H&E (40x) section showing pseudo horn cysts and branched rete ridges with increased pigmentation at their tips.

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