RESEARCH PAPER | Medical Science | Volume : 5 | Issue : 2 | Feb 2015 | ISSN - 2249-555X

Clinico-Pathological Correlation of Localised Dowling-Degos Disease: a Case Report

KEYWORDS: Dowling-Degos disease (DDD), hyperpigmentation.

Bhushan Malhari Warpe

Hindalco Colony, MIDC Taloja, Navi-Mumbai-410208 Maharashtra

ABSTRACT *A 26-years-old young man came with complaints of abnormal dark skin colour (hyperpigmentation), particularly in the back of the hands and intertriginous folds between digits, progressively spreading since last five years without any family history of the same. He was diagnosed with clinical diagnosis of hyper-pigmented disorder under evaluation most probably medication induced. He had no personal and family history of such skin disease or diabetes mellitus. We received a skin biopsy of 0.3 x 0.3 cm which was totally embedded for histopathology reporting. Section studied through skin reveals epidermis, dermis and subcutaneous tissue. Epidermis shows hyperkeratosis, irregular acanthosis with focal filliform down growth of epidermis. Multiple keratin horn cysts were seen in the down growth. The tips of rete ridges showed pronounced hyperpigmentation. Histopathological impression was given as Dowling-Degos disease (DDD). DDD is a rare genetic skin condition without definite cure, although its prevalence is unknown. So we report a DDD case in a young man with clinico-pathological correlation.*

Introduction:

Dowling-Degos disease (DDD) appears to be a rare genetic skin condition, although its prevalence is unknown. Dark dot disease, DDD, Dowling-Degos-Kitamura disease, reticular pigment anomaly of flexures and reticulate acropigmentation of Kitamura are the various names used by people worldwide. DDD and its variants can either be inherited from one parent (autosomal dominant) or appear without a family history (sporadic). It is not curative as it is a genetic disorder.1-4

The onset of classic DDD is in adult life, most commonly in the 20s or 30s but sometimes later. It slowly becomes more extensive with time but not life threatening. DDD only affects the skin and there are no internal effects. Skin affected by DDD can be itchy, especially in summer when the effects of heat, perspiration and friction aggravate the pigmentation and itch. Blistering is not a feature of this disease.1-2

DDD is a skin condition characterized by a lacy or net-like (reticulate) pattern of abnormal hyperpigmentation, particularly in the body's folds and creases. These skin changes typically first appear in the armpits and groin area and can later spread to other skin folds such as the crook of the elbow and back of the knee. Less commonly, pigmentation changes can also occur on the wrist, back of the hand, face, scalp, scrotum (in males), and vulva (in females). 3-4

In our case, the back of hands and intertriginous areas between digits was the site of localized skin involvement with hyperpigmentation (Fig. 1 inset). No internal effects were seen with no family history of such skin lesions (sporadic case) in our case.

Case report:

A 26-years-old young man came with complaints of abnormally dark skin coloring (hyperpigmentation), particularly in the back of the hands and intertriginous folds between digits, progressively spreading since last five years without any family history of the same. These areas of hyperpigmentation do not darken with exposure to sunlight and caused no health problems. It was not associated with skin irritation and itchiness. Since the patient used to live in a red light area of Kamathipura area of Mumbai, he thought it was some sexually transmitted skin infection. He had no alleged sexual contact with prostitutes in his area. He was diagnosed with clinical diagnosis of hyper-pigmented disorder under evaluation most probably medication induced. He had no personal and family history of such skin disease or diabetes mellitus.

We received a skin biopsy of 0.3 x 0.3 cm which was totally embedded for histopathology reporting. Section studied through skin reveals epidermis, dermis and subcutaneous tissue. Epidermis shows hyperkeratosis, irregular acanthosis with focal filliform down growth of epidermis (Fig. 1). Multiple keratin horn cysts (Fig. 2) were seen in the down growth. The tips of rete ridges showed pronounced hyperpigmentation. Histopathological impression was given as Dowling-Degos disease.

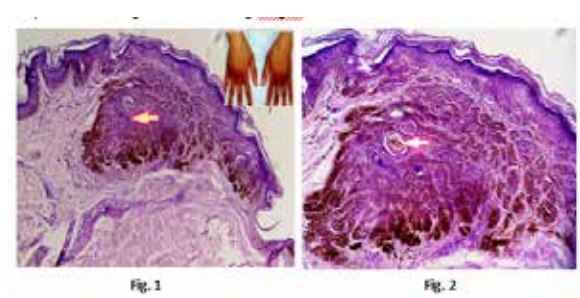

Figure 1: Microphotograph - Epidermis shows hyperkeratosis, irregular acanthosis with focal filliform down growth of epidermis (arrow). Fig.1 Inset : Clinical photograph shows hyperpigmentation in the back of the hands and intertriginous folds between digits.

Figure 2: Microphotograph - Multiple keratin horn cysts (arrow) were seen in the down growth of epidermis. The tips of rete ridges showed pronounced hyperpigmentation.

Volume : 5 | Issue : 2 | Feb 2015 | ISSN - 2249-555X

Discussion:

Dowling 1 first delineated this genodermatosis as a distinct entity in 1938. In 1954, Degos and Ossipowski 2 described a patient with a similar case. Few patients with reticulate pigmented anomaly, also known as Dowling-Degos disease (DDD), have been reported.

Dowling-Degos disease (reticulate pigmented anomaly) is a rare condition affecting both sexes but more common in females. DDD tends to develop early in adult life, with the onset of pigmentation occurring in individuals before they are aged 24 years. The flexural pigmentation has its onset from childhood to adult life. It may be intense, with a brownish black color and sometimes steel blue or navy overtones. However, if the condition is less severe, it is stippled in shades of brown. No verrucous or velvety papillomatosis is present, as might be seen in acanthosis nigricans.1-2

Dowling-Degos disease (reticulate pigmented anomaly) is characterized by flexural pigmented reticulate macules and sometimes comedo-like papules on the back and/or the neck (dark dot follicles). Some patients have pitted perioral scars. Pruritus of affected flexural areas may be the only symptom. In both male and female patients, pigmented reticulate macules may also be evident on the genitalia, where they may be seen alone. The pigmented eruption on the male external genitalia may be a cutaneous marker of underlying testicular carcinoma,3 although the association is probably fortuitous. Bilateral nephroblastoma in familial Hay-Wells syndrome has been associated with familial reticulate pigmentation of the skin, 4 another possibly fortuitous association.

Typical clinical Dowling-Degos disease (reticulate pigmented anomaly) may histopathologically be Galli-Galli disease. Galli-Galli disease is a rare genodermatosis in the spectrum of reticulate hyperpigmentation, probably best regarded as an acantholytic variant of Dowling-Degos disease (reticulate pigmented anomaly). 5

Dowling-Degos disease (reticulate pigmented anomaly) is often familial and appears to be inherited in an autosomal dominant manner. 6 A gene locus believed responsible in one Chinese patient was mapped to 17p13.3. 7 A genome-wide linkage analysis of two German families mapped this disease to 12q. 8 This region includes the keratin gene cluster, which was screened for mutations.

Loss-of-function mutations were identified in the keratin 5 gene (KRT5) in all affected family members and in 6 unrelated patients with Dowling-Degos disease (reticulate pigmented anomaly). Another study found the same KRT5 mutation in patients with reticulate pigmented anomaly and its acantholytic variant, Galli-Galli disease. 9 This variant has a genotype/phenotype correlation with mutations in the keratin 5 (KRT5) gene.

No treatment is effective for Dowling-Degos disease (reticulate pigmented anomaly). Topical retinoic acids, topical steroids, hydroquinone, tretinoin, and systemic retinoids have been used without success. Dowling-Degos disease has been successfully treated with the fractional Er:YAG laser. The patient and his or her family should be educated about the common autosomal dominant nature of Dowling-Degos disease (reticulate pigmented anomaly). 10

REFERENCE

1. Dowling GD, Freudenthal W. Acanthosis nigricans. Proc Roy Soc Med. 1938; 31:1147-50. | 2. Degos R, Ossipowski B. Reticulated pigmentary dermatosis of the folds: relation to acanthosis nigricans. Ann Dermatol Syphiligr (Paris) 1954; 81(2):147-51. | 3. Schwartz RA, Birnkrant AP, Burgess GH, Stoll HL Jr, Yaqub M, Fox MD. Reticulate pigmented anomaly. Cutis. Oct 1980; 26(4):380-1. | 4. Drut R, Pollono D, Drut RM. Bilateral nephroblastoma in familial HayWells syndrome associated with familial reticulate pigmentation of the skin. Am J Med Genet. Jun 15 2002; 110(2):164-9. | 5. Müller CS, Pföhler C, Tilgen W. Changing a concept controversy on the confusing spectrum of the reticulate pigmented disorders of the skin. J Cutan Pathol. Jan 2009; 36(1):44-8. | 6. Brown WG. Reticulate pigmented anomaly of the flexures. Case reports and genetic investigation. Arch Dermatol. Jul 1982; 118(7):490-3. | 7. Li CR, Xing QH, Li M, et al. A gene locus responsible for reticulate pigmented anomaly of the flexures maps to chromosome 17p13.3. J Invest Dermatol. Jun 2006; 126(6):1297-301. | 8. Betz RC, Planko L, Eigelshoven S, et al. Loss of function mutations in the keratin 5 gene lead to Dowling Degos disease. Am J Hum Genet. Mar 2006; 78(3):510-9. | 9. Hanneken S, Rütten A, Pasternack SM, Eigelshoven S, El ShabrawiCaelen L, Wenzel J, et al. Systematic mutation screening of KRT5 supports the hypothesis that Galli Galli disease is a variant of Dowling Degos disease. Br J Dermatol. Mar 5 2010; [Medline]. | 10. Yun JH, Kim JH, Choi JS, Roh JY, Lee JR. Treatment of Dowling Degos disease with fractional Er:YAGlaser. J Cosmet Laser Ther. Mar 6 2013;[Medline].