DOWLING- DEGOS DISEASE- A CASE SERIES
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Abstract: Dowling Degos Disease or Reticular pigmented anomaly of flexures, is a rare autosomal dominant pigmentary disorder characterized by acquired reticulate pigmentation of flexures, groin, neck. We describe a series of 6 cases of Dowling Degos disease belonging to 5 separate families presenting over a period of two years. These cases have been proven by histopathology. We present this case series as this disease might not be as rare as it has been reported so far. This is probably because Dowling Degos Disease is easily missed.

Keyword: Reticular pigmented anomaly of flexures, Autosomal dominant.

INTRODUCTION-Dowling – Degos Disease (DDD) is a rare genodermatosis, characterized by acquired reticular hyperpigmentation that begins in the axilla & groin and later involves other body folds. We present a series of 6 cases, 5 females and one male patients belonging to five separate families.

CASES- We came across 6 cases with flexural pigmentation, pitted perioral scars and dark-dot follicles over a period of 13months (July 2010-August 2011) [Table 1]. Pruritus was a feature in half of our patients, while the rest were asymptomatic. Two of the patients (cases 1 and 2) were related [Daughter- father]. Two patients (cases 1 and 4) had oral mucosal involvement and one patient had hypopigmented macules over the trunk and extremities (case 2). In one patient (case 4), the pigmented macules demonstrated the Koebner’s phenomenon. One female patient (case 5) had associated lesions of hidradenitis suppurativa over both axillae.

FIGURE 1- Numerous pigmented macules and papules distributed symmetrically over flexor aspects of both forearm

FIGURE 2- Reticular pigmentation over neck

FIGURE 3- Hypopigmented macules over trunk and extremities

FIGURE 4- Reticular pattern of maculo-papular lesions over neck
Histopathological examination in all patients showed increased pigmentation of basal layer, with finger-like rete ridges with thinning of suprapapillary epidermis with the antler-like pattern.

Figure 10. Increased pigmentation of basal layer with finger-like rete ridges and thinning of suprapapillary epidermis. Melanophages seen in upper dermis. (H & E, 10x)

Figure 11. One patient had horn cysts in the epidermis.

DISCUSSION-DDD was first described by Dowling in 1938 and later by Degos and Ossipowski in 1954. [2], [3] It is a rare autosomal dominant disorder which shows female predominance. Most cases have been reported from Asian and Mediterranean countries. Mutation in keratin 5 has been implicated. [4] A genome-wide linkage analysis of 2 German families mapped this disease to 12q. In this case series, all but one patient had a positive family history and female: male ratio was 5:1. DDD usually presents post-pubertally. [5] In this series the median age of onset was 20 years. Clinical presentation includes small round pigmented macules resembling freckles which become more numerous and reticulate with time. Usual sites of involvement are axillae and groin. Other sites include intergluteal and inframammary folds, neck, scalp, trunk and arms. Involvement of the genitalia, particularly pigmented lesions of the vulva can occur. [6] Pigmentation is symmetrical and progressive. The lesions are usually asymptomatic but may be associated with pruritus. Dark brown papules may also occur at the affected sites due to lichenification. Other features include comedo-like lesions or dark-dot follicles on the back and neck, pitted perioral or facial scars, and epidermoid cysts. [7] Hypopigmented macules and papules may occur rarely. [8] Additionally, speckled macules involving the dorsum of the hands, the proximal
<table>
<thead>
<tr>
<th>No</th>
<th>Age (Y)</th>
<th>Sex</th>
<th>Age of onset</th>
<th>Sites of Macular-Papular pigmented Lesions</th>
<th>Pitted comedo-like lesions</th>
<th>Others</th>
<th>Symptoms</th>
<th>Family history</th>
<th>HPE</th>
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<td>2</td>
<td>5</td>
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<td>30</td>
<td>Face, neck, axillae, groin, trunk, forearms. (Figure 2)</td>
<td>Present</td>
<td>Hypopigmented macules over trunk &amp; extremities (Figure 3)</td>
<td>Asymptomatic</td>
<td>Present</td>
<td>Confirmatory</td>
</tr>
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<td></td>
<td>3</td>
<td>F</td>
<td>12</td>
<td>Axillae, Cubital fossae, forearms, Face, neck, back (Figure 4)</td>
<td>Absent</td>
<td>Absent over zygoma, cheeks, forehead</td>
<td>Pruritus</td>
<td>Present</td>
<td>Confirmatory</td>
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<td>4</td>
<td>2</td>
<td>F</td>
<td>20</td>
<td>Axillae, trunk, extremities (Figure 5)</td>
<td>Present</td>
<td>Pigmented macules over buccal mucosa</td>
<td>Pruritus</td>
<td>Present</td>
<td>Confirmatory</td>
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<td>2</td>
<td>F</td>
<td>20</td>
<td>Axillae</td>
<td>Present (Figure 6)</td>
<td>Absent Hidradenitis suppurativa lesions over axillae (Figure 7)</td>
<td>Asymptomatic</td>
<td>Present</td>
<td>Confirmatory</td>
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<tr>
<td></td>
<td>4</td>
<td>F</td>
<td>45</td>
<td>Axillae, groin, waist, inframammary, inner aspect of forearms (Figure 8 &amp; 9)</td>
<td>Absent</td>
<td>Present</td>
<td>Pruritus</td>
<td>Absent</td>
<td>Confirmatory</td>
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nail folds, or the scrotum may be seen. Fingernail dystrophy may be present. The finding of speckled macules on the scrotum is isolated and limited to the scrotal and penile skin. This pigmented eruption on the male external genitalia is possibly a cutaneous marker of underlying testicular carcinoma. Association with hidradenitis suppurativa has been described. Our patients presented with the classical findings of reticulate pigmented macules over the body folds as well as the forearms, thighs and face. Pitted perioral scars were present in 4 of 5 patients and comedone-like lesions over the face were present in three patients. Pruritus was a complaint in three patients while the other three were asymptomatic. Our male patient also had hypopigmented macules over the trunk and extremities. One patient had lesions of hidradenitis suppurativa over the axillae while two patients had pigmentation of buccal mucosa. In one patient, koebnerization of the pigmented macules was observed. Histopathology is diagnostic with thin branching, heavily pigmented digitate elongations of rete ridges with tendency to spare the suprapapillary epithelium. In some cases horn cysts may be seen. Wilson-Jones summarized these findings as ‘demonstrating dusky dappled disfigurements and dark-dot depressions and disclosing digitate downgrowths delving dermally’. The histopathological picture from the pigmented macular-papular lesions in all our cases showed thin, branching, finger-like elongations of rete ridges. In one patient, horn cysts were also appreciated.

Differential Diagnosis includes acanthosis nigricans, axillary freckles of neurofibromatosis, reticulate acropigmentation of Kitamura, Haber’s Syndrome and Galli-Galli disease. In our patients, the closest differential diagnosis was reticulate acropigmentation of Kitamura. However, Kitamura’s reticulate acropigmentation is characterized by a network of freckle-like areas of pigmentation which develop on the dorsa of the hands in the first two decades. The reticulate pigmentation may subsequently involve most parts of the body. Palmar pits and breakages of epidermal ridge pattern are also found. Another unique feature seen is the atrophic nature of these lesions. In our patients, palmar pits were absent and dermatoglyphics was normal. Also, the pigmentation began over the flexures and not over the extremities and atrophia lesions were absent. Haber’s Syndrome is an autosomal dominant disease with rosacea-like facial erythema and keratotic plaques and pitted scars that begin in childhood. Galli – Galli disease is similar in clinical features to DDD but on histopathology acantholysis was not seen and hence it is considered an acantholytic variant of DDD. In this case series, acantholysis was not seen in any patient. Treatment with topical hydroquinone, tretinoin, adapalene and steroids have shown varying success. Topical steroids may decrease the pruritus. Er:YAG laser pulse energy between 1000 and 1200 mJ three consecutive passes has lead to good results in some patients. Daily application of topical tretinoin 0.05% was tried for 3 months in our patients without any success and was thus discontinued.

CONCLUSION:
This case series indicates that DDD is probably not as rare as thought to be. This is could be because DDD is easily missed due to its subtle presentation which may be overlooked especially in dark-skinned individuals and also because of the asymptomatic nature of the disease in many patients. In addition to the classical features present in all our patients, rare findings included presence of hypopigmented macules in one patient. Also one patient lacked similar family history and had onset of lesions as late as 45 years of age. Other interesting findings were of oral mucosal pigmentation in two patients and presence of koebner’s phenomenon in one patient. These features have not been reported in DDD in the literature so far.

BIBLIOGRAPHY

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