Dowling Degos Disease, A rare genetic disorder

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Key Clinical Message

Dowling-degos disease (DDD) is an uncommon autosomal dominant disorder characterized by reticulate pigmentation. It is important to diagnose the disease as many other cutaneous conditions are associated with it. Here, we have a case report of DDD with clincial features of downs syndrome.

Results

On the basis of clinical and histological findings, diagnosis of DDD was made.Patient was counseled about the prognosis of disease, treatment options and is under regular follow-up.

Discussion

DDD is a rare autosomal dominant genodermatosis but can also occur sporadically. It is characterized by asymptomatic, progressive, reticulate hyperpigmented macules with a flexural distribution. Other cutaneous findings include comedo-like hyperkeratotic follicular papules on the neck, perioral pitted acneiform scars.[5]Loss-of-functional mutation in the keratin 5 (KRT5) gene on chromosome 12q13 is reported. Mutation of the gene results in disruption in melanosome transfer and trafficking. Gene locus mapping to chromosome 17p13.3 has also been reported. Other genes involved in pathogenesis of DDD are POGLUT1(protein O-glucosyltransferase 1) ,POFUT1 (protein O fucosyltransferase 1), and PSENEN (presenilin enhancer protein 2 gene).[4]

Epidermal cysts, multiple keratoacanthomas, squamous cell carcinoma, abscess, hidradenitis suppurativa, seborrheic keratosis, and pilonidal cysts are reported to be associated with DDD.[2] These disorders are not present in our case and other family members. Histopathology of skin biopsy in classical DDD reveals thinning of the epidermis. Elongated, thinned, and branched (antler like) rete ridges with increased melanin pigmentation at their tips, but no increase in the melanocyte number.[6] Histopathology of skin biopsy of our patient also revealed these classical findings. DDD with features of downs syndrome has not been reported till date in literature. Karyotyping in daughter would have revealed the association between DDD and downs syndrome.As the patient denied the intervention, association could not be established.

It is a rare genetic disorder and only few cases have been reported in our country. There is no definitive cure for Dowling-Degos disease. Different treatment options are depigmenting agents such as hydroquinone, retinoids and laser therapy such as fractional Erbium YAG laser.[7]

There is paucity of reported cases of DDD in literature. This condition should be kept in mind in the differential diagnosis of pigmented lesions of flexural sites.

Authors Contribution Statement

Prof. Dr. Mahesh Mathur: Conceptualization; Formal analysis; Resources; Supervision; Validation; Visualization; Writing-original draft

Dr. Neha Thakur: Conceptualization; Formal analysis; Resources; Supervision; Validation; Visualization; Writing-original draft.

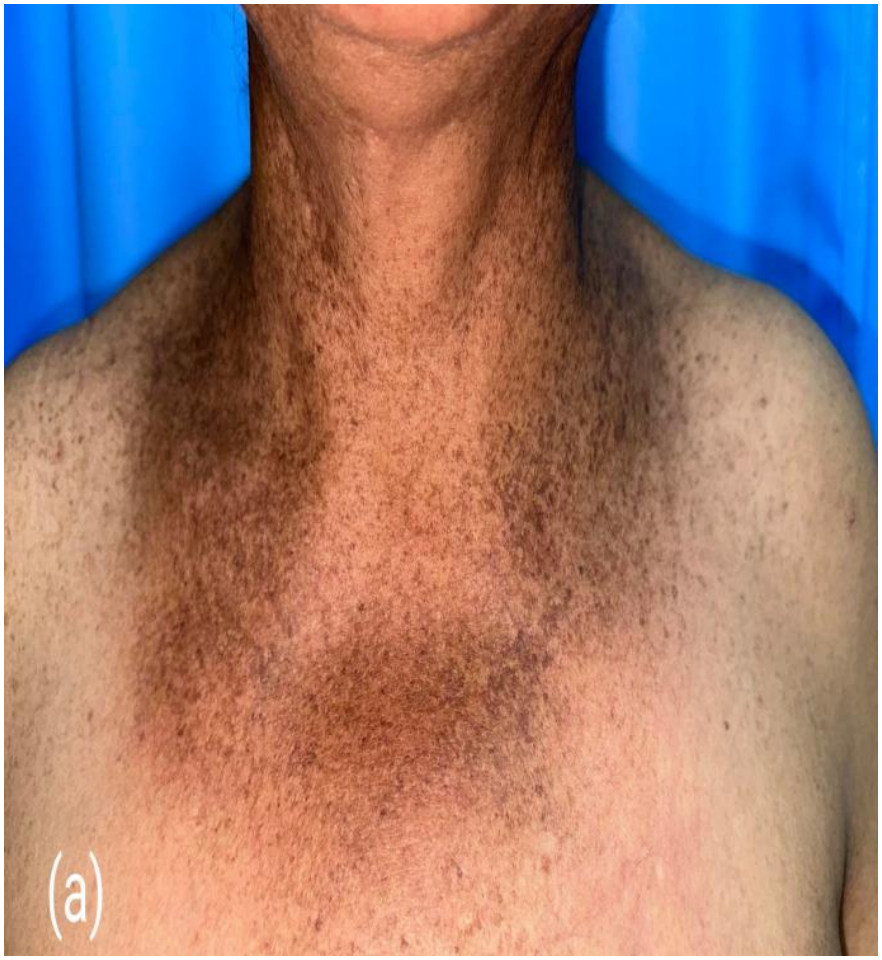
Dr. Nabita Bhattarai: Conceptualization; Formal analysis; Resources; Supervision; Validation; Visualization; Writing-original draft.

Dr. Supriya Paudel: Formal analysis; Resources; Supervision; Visualization; Writing-original draft; Writing- review and editing

Dr. Sandhya Regmi: Data curation; Investigation; Resources; Visualization; Writing- review and editing.

Dr. Sambidha Karki: Data curation; Investigation; Resources; Visualization; Writing - review and editing.

Figures



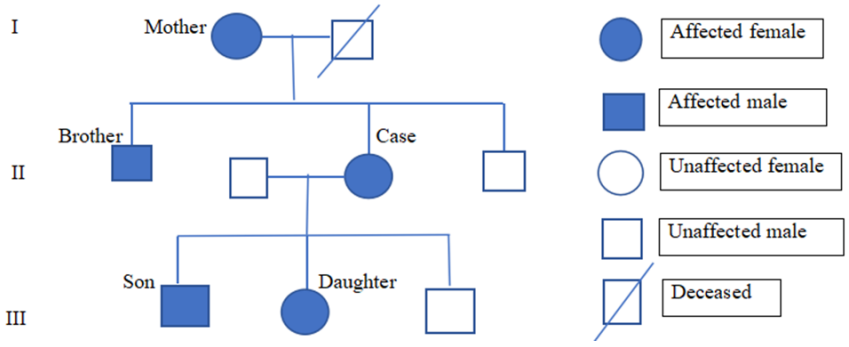
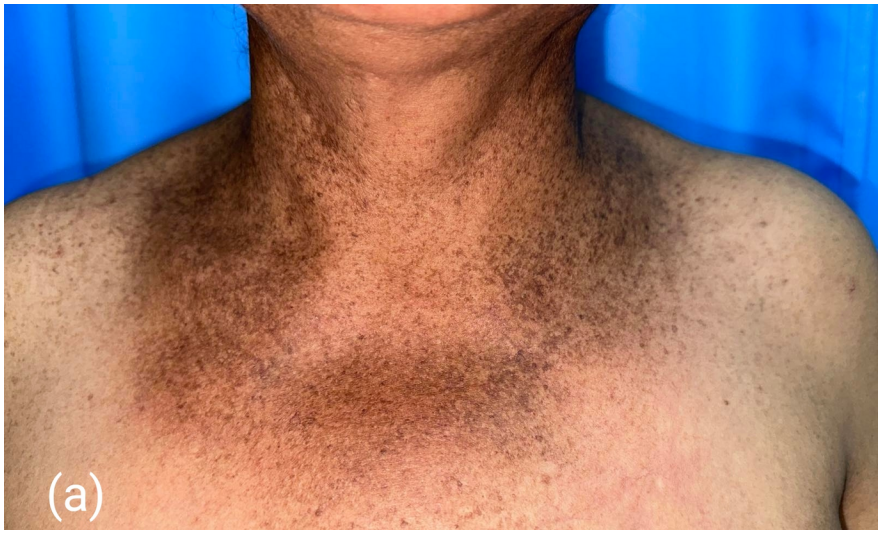
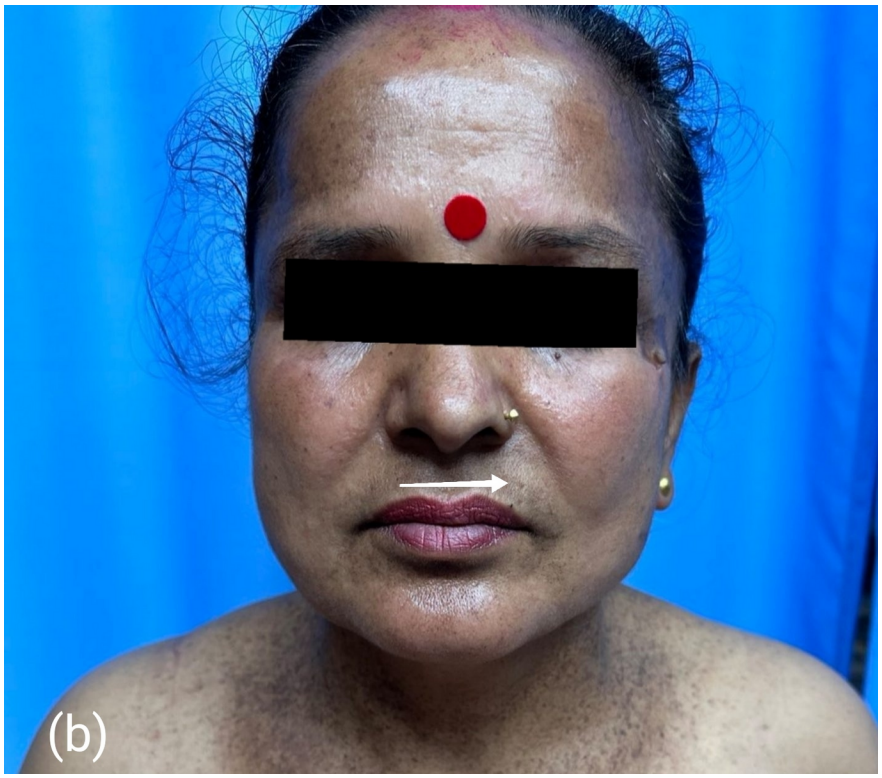
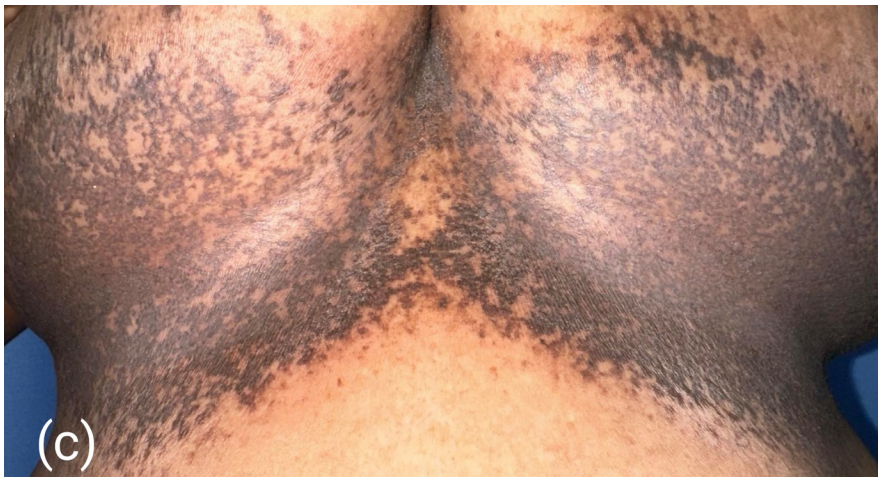
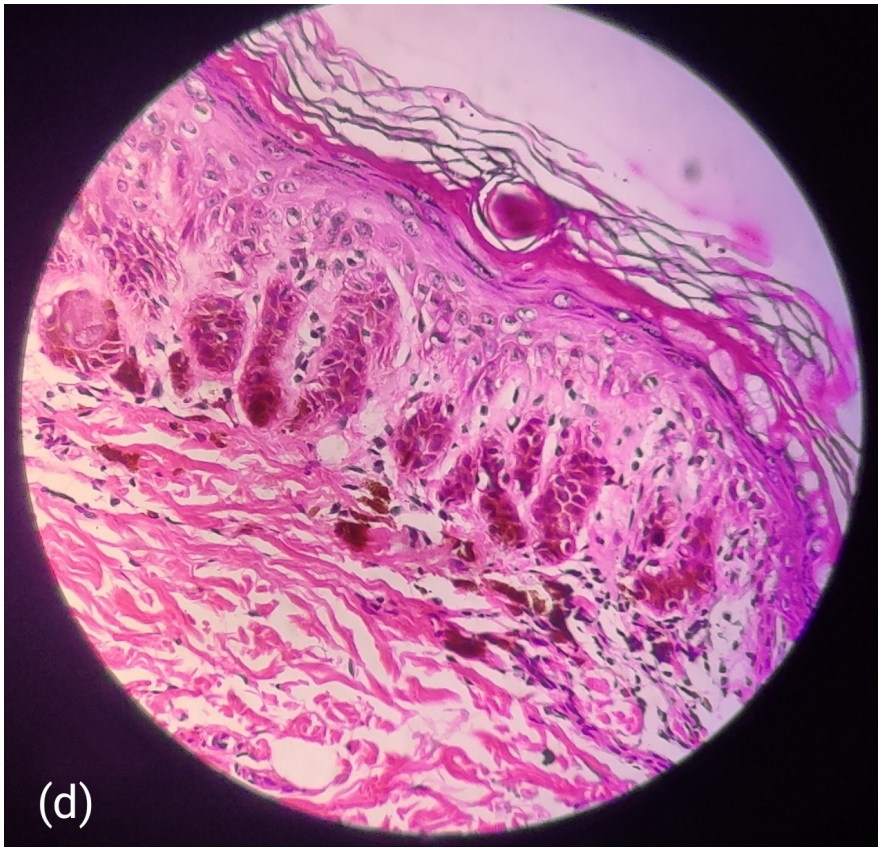


Figure 2: Pedigree of the family with Dowling Degos Disease.









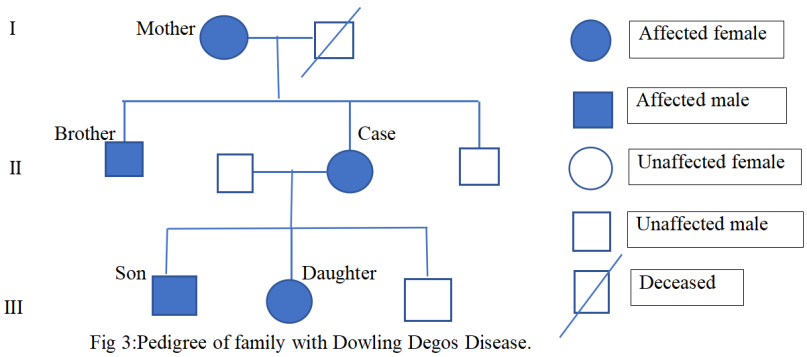


Fig 3:Pedigree of family with Dowling Degos Disease.