

Familial Disseminated Comedones without Dyskeratosis, A Rare Genodermatosis: A Case Report

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ABSTRACT

Background: Familial disseminated comedones without dyskeratosis (FDCWD) is a rare inherited genodermatosis characterized by widespread comedones, papulonodular lesions, and variable scarring. Only a few families with this condition have been reported worldwide, and its etiopathogenesis and underlying molecular mechanisms remain poorly understood.

Case Report: A 24-year-old man presented with multiple papulonodular lesions and comedones involving the face and trunk. The lesions initially appeared at the age of 20 years and progressively increased in number and extent. Cutaneous examination revealed multiple scattered comedones with blackish keratotic plugs, punctate depressions, skin coloured to hyperpigmented nodules with prominent follicular openings containing keratotic plugs, hyperkeratotic papules, hypertrophic and atrophic scars, and post-inflammatory hyperpigmented macules. The back showed extensive involvement. Systemic examination was unremarkable. Similar clinical features were present in seven affected individuals of both sexes across three successive generations, suggesting an autosomal dominant pattern of inheritance. Histopathological examination showed orthokeratotic hyperkeratosis, markedly dilated follicles containing keratin plugs, and an arborizing proliferation of basaloid cells surrounding the dilated follicles, without evidence of dyskeratosis. Genetic testing was not performed due to limited resources and financial constraints. Based on the clinical, histopathological, and familial findings, a diagnosis of familial disseminated comedones without dyskeratosis was made. The patient was started on oral isotretinoin at a dose of 0.5 mg/kg/day and followed up in the Dermatology Outpatient Department.

Conclusion: FDCWD is a rare genodermatosis with characteristic clinical and histopathological features. The presence of similar manifestations across three successive generations in this case supports an autosomal dominant pattern of inheritance. The limited number of reported families highlights the importance of documenting additional cases to improve understanding of the clinical and histopathological spectrum, inheritance pattern, and long-term progression of this rare disorder.

Keywords: *Familial disseminated comedones; Genodermatosis; Comedones; Autosomal dominant inheritance; Without Dyskeratosis*

INTRODUCTION

Familial disseminated comedones without dyskeratosis (FDCWD) is a rare genodermatosis characterized by the development of widespread comedones with subsequent scarring. Only a few families with this condition have been reported worldwide. Due to its rarity, its etiopathogenesis and underlying molecular mechanisms remain poorly understood.¹⁻³ We report a

case of a 24-year-old man with familial disseminated comedones without dyskeratosis, with similar clinical features observed in multiple generations of his family.

CASE REPORT

A 24-year-old man presented to the Dermatology Outpatient Department with multiple papulonodular lesions and comedones involving the face and trunk.

Cutaneous examination revealed multiple scattered comedones measuring approximately 1–4 mm, containing blackish keratotic plugs (Fig. 2).

Figure 1: PEDIGREE CHART

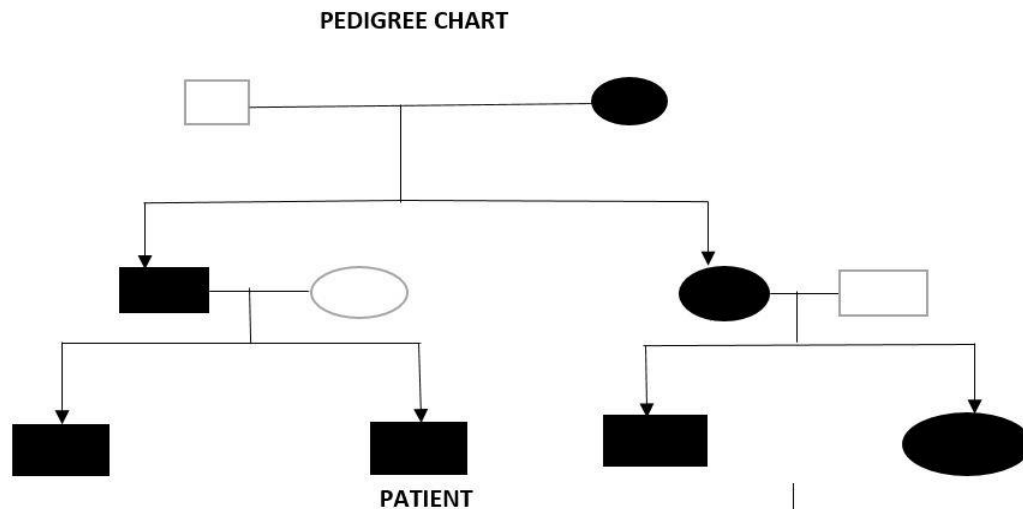


Figure 2: On cutaneous examination there were multiple scattered comedones(1-4mm) filled with blackish keratotic plug and skin- coloured to hyper-pigmented nodules (1- 3cm) with follicular openings and keratotic plugs distributed over neck and trunk. A few hypertrophic and atrophic scars were also present along with multiple post-inflammatory brown to black macules.



Multiple punctate depressions were also noted. In addition, skin-coloured to hyperpigmented nodules measuring approximately 1–3 cm were present, with prominent follicular openings containing keratotic plugs. A few hyperkeratotic papules were also observed. Both hypertrophic and atrophic scars were present, along with multiple post-inflammatory hyperpigmented macules distributed over the face, neck, and trunk. The back showed extensive involvement (Fig. 3).

Figure 3: : On cutaneous examination there were multiple scattered comedones(1-4mm) filled with blackish keratotic plug and skin- coloured to hyper-pigmented nodules (1- 3cm) with follicular openings and keratotic plugs distributed over face and upper back.



Systemic examination was unremarkable, with no skeletal, auditory, ocular, or intellectual abnormalities. Similar clinical features were reported in affected family members spanning three successive generations, suggesting an autosomal dominant pattern of inheritance (Fig. 1). A total of seven individuals of both sexes across three generations were affected.

Histopathological examination of a representative lesion showed orthokeratotic hyperkeratosis of the epidermis and markedly dilated follicles containing keratin plugs (Fig. 4). An arborizing proliferation of basaloid cells was observed surrounding the dilated follicles (Fig. 5). No evidence of dyskeratosis was identified.

Figure 4: Histopathological section of low power field image shows orthokeratotic hyperkeratosis with dilated follicle filled with keratin plug.

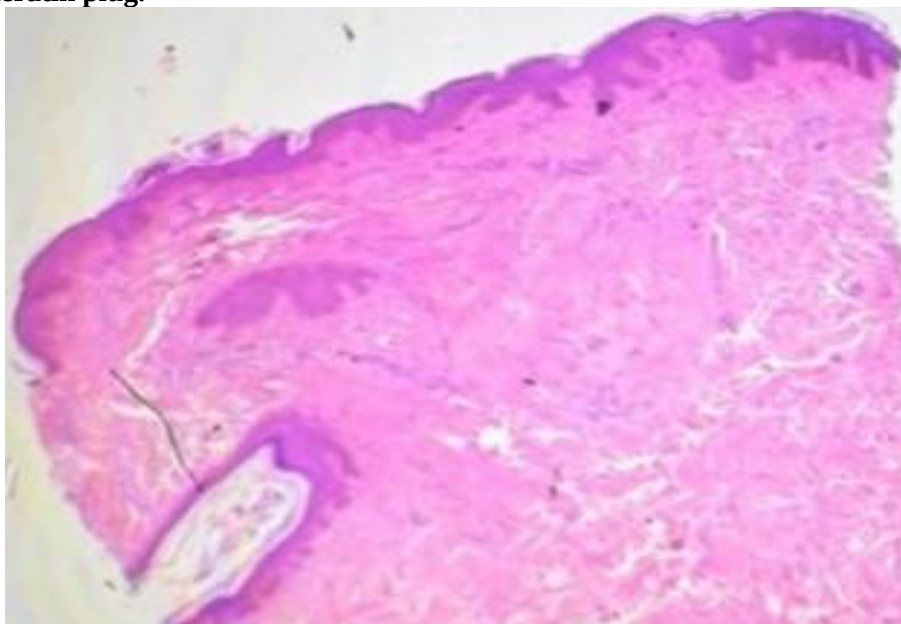
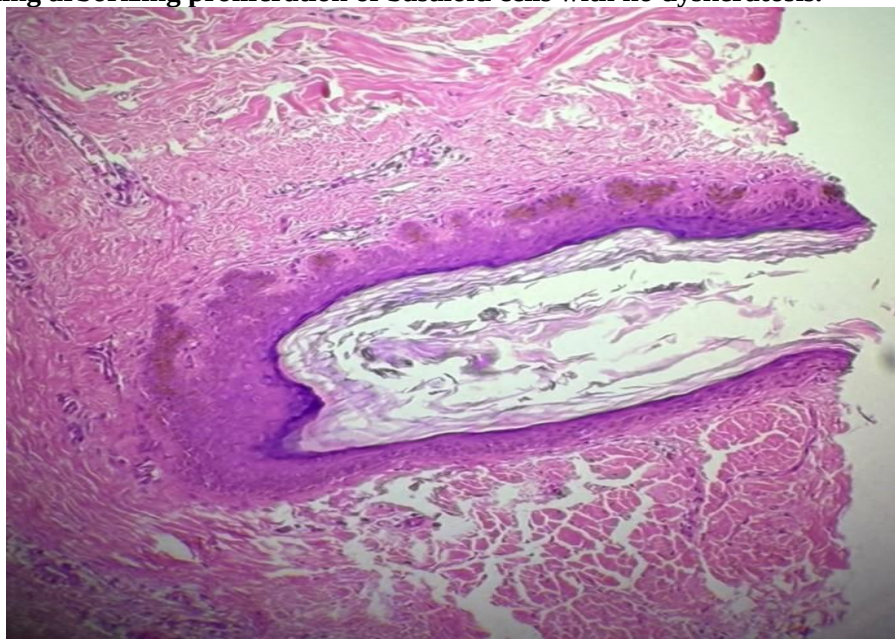


Figure 5: Histopathological sections from high power field image revealed dilated follicles filled with keratin plug with surrounding arborizing proliferation of basaloid cells with no dyskeratosis.



Genetic testing was not performed because of limited resources and financial constraints. Based on the clinical presentation, characteristic histopathological findings, and involvement of multiple generations of the family, a diagnosis of familial disseminated comedones without dyskeratosis was made.

DISCUSSION

Familial disseminated comedones without dyskeratosis (FDCWD) is a rare autosomal dominant genodermatosis characterized by widespread and extensive comedonal involvement of the skin. It is distinguished histopathologically by dilated follicular structures filled with lamellar keratin and filiform or arborizing extensions of basaloid cells arising from the outer root sheath, without evidence of acantholysis or dyskeratosis.¹⁻³

FDCWD is an extremely rare dermatosis, with only a few affected families reported in the literature to date.¹⁻³ Previously reported cases have demonstrated predominant involvement of the trunk, variable clinical severity, a chronic progressive course, and limited response to treatment. Histopathologically, crateriform follicular dilatation containing lamellar keratin is a characteristic finding.^{1, 3}

The presence of affected individuals in successive generations in our case supports an autosomal dominant pattern of inheritance, consistent with previously reported familial cases.^{1, 3} Molecular studies have also provided evidence for a genetic basis of the disorder. Whole-genome linkage analysis followed by whole-exome sequencing in an affected family identified a heterozygous one-base-pair insertion, c.84_85insT (p. Leu28Phefs*93), in the *PEN2* gene, located within the linked region on chromosome 19.⁴

In our case, the characteristic clinical presentation, corresponding histopathological findings, and clear familial pattern involving three successive generations strongly support the diagnosis of FDCWD. The

clinical and histopathological findings are in concordance with those described in previously reported cases. However, only a limited number of families with this condition have been documented in the literature. The paucity of reported cases highlights the importance of documenting additional cases to expand the understanding of its clinical and histopathological spectrum, inheritance pattern, and long-term progression.

Strengths

- The present case represents a rare familial genodermatosis with a clearly documented pattern of involvement across three successive generations.
- The identification of seven affected individuals of both sexes within the family provides supportive clinical evidence for an autosomal dominant inheritance pattern.
- The diagnosis was supported by characteristic clinical and histopathological findings, including widespread comedones, follicular dilatation with keratin plugs, and arborizing basaloid cell proliferation without dyskeratosis.
- Detailed clinical documentation and histopathological examination contribute to the limited literature on familial disseminated comedones without dyskeratosis.

Limitations

- Genetic testing could not be performed because of limited resources and financial

constraints; therefore, the underlying genetic mutation could not be molecularly confirmed in the present patient or affected family members.

- The diagnosis was based on the characteristic clinical presentation, histopathological findings, and familial pattern rather than molecular confirmation.
- Long-term treatment response and follow-up data are limited.
- Although multiple affected family members were identified, detailed clinical and histopathological evaluation was not available for every affected individual.
- As this is a single-family case report, the findings cannot be generalized to all patients with familial disseminated comedones without dyskeratosis.

CONCLUSION

Familial disseminated comedones without dyskeratosis is a rare inherited genodermatosis characterized by widespread comedones, papulonodular lesions, and variable degrees of scarring. In the present case, the presence of similar clinical manifestations in three successive generations supports an autosomal dominant pattern of inheritance. Recognition of the characteristic clinical and histopathological features is important for establishing the diagnosis and differentiating this condition from other disorders presenting with multiple comedones. Genetic counselling may be considered for affected individuals and their families. Further genetic and molecular studies are required to elucidate the underlying pathogenesis and may contribute to the development of more effective therapeutic strategies for this rare disorder.

Declarations

Ethical Approval

Ethical approval was obtained from the Institutional Ethics Committee prior to the preparation and submission of this case report. The study was conducted in accordance with the ethical principles of the Declaration of Helsinki and applicable institutional guidelines.

Informed Consent

Written informed consent was obtained from the patient for publication of the case report and accompanying clinical and histopathological images.

Consent for Family Information

Appropriate consent was obtained for reporting relevant familial clinical information and the pedigree presented in this case report.

Conflict of Interest

The authors declare that they have no conflict of interest.

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Author Contributions

All authors contributed to the clinical evaluation, interpretation of findings, preparation and critical revision of the manuscript, and approved the final version for publication.

Data Availability

All relevant information supporting the findings of this case report is included in the manuscript. Additional identifiable patient information is not publicly available to protect patient confidentiality.

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AI Use Disclosure

The authors declare that no generative artificial intelligence tools were used to generate or substantially modify the scientific content of this manuscript.

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