Content available at: https://www.ipinnovative.com/open-access-journals

Journal of Oral Medicine, Oral Surgery, Oral Pathology and Oral Radiology

Journal homepage: http://www.joooo.org

Case Report

When skin and smiles shatter: An extraordinary case of epidermolysis bullosa dowling-meara with devastating oro-dental involvement

Jyoti C[1*], Praveenkumar Ramdurg[1], Naveen Srinivas[1], Lingaraj S Harihar[1], Mohammed Fazil[1], Harshita K Arabbi[1]

[1]Dept. of Oral Medicine and Radiology, P.M.N.M. Dental College and Hospital, Bagalkot, Karnataka, India

Abstract

Epidermolysis Bullosa (EB) is a spectrum of rare inherited disorders characterized by skin and mucosal fragility. Oral manifestations—particularly enamel hypoplasia, blistering, and mucosal atrophy—can be severe and functionally limiting in the more aggressive subtypes such as EB simplex, Dowling-Meara (EBS-DM).Case Summary: We report the case of a 21-year-old female diagnosed with EBS-DM, the most severe variant of EB simplex caused by mutations in KRT5 or KRT14. She presented with facial and limb scarring, dystrophic nails, generalized enamel hypoplasia, over-retained deciduous teeth, multiple impacted teeth, and grossly decayed molars. Panoramic radiography confirmed periapical pathology, enamel thinning, and root shortening. Her medical history included lifelong vesiculobullous lesions and a background of consanguineous parentage. Dental management involved atraumatic extractions, fluoride therapy, and preventive protocols under a multidisciplinary team approach.Conclusion: This case emphasizes the importance of early recognition and trauma-free dental care in EB. Individualized treatment plans, psychosocial support, and genetic counseling are critical for improving function and long-term quality of life.

Keywords: Epidermolysis bullosa dowling-meara, Enamel hypoplasia, KRT5, KRT14, Blistering disorder, Oral manifestations, Multidisciplinary care.

Received: 05-07-2025; **Accepted:** 29-07-2025; **Available Online**: 29-09-2025

1. Introduction

Epidermolysis bullosa (EB) comprises a group of genodermatoses resulting from mutations that affect proteins responsible for epithelial integrity. EB is classified into four major types based on the level of tissue cleavage: EB simplex (EBS), junctional EB, dystrophic EB, and Kindler syndrome.[1-3] Among these, Dowling-Meara is the most severe and generalized form of EBS. It arises from mutations in the KRT5 or KRT14 genes, which code for keratin 5 and 14—key cytoskeletal proteins in basal keratinocytes. Defective assembly of keratin filaments leads to cytolysis and intraepidermal blistering.

EB has an incidence of approximately 1 in 50,000 live births.[1] Though cutaneous features are the most evident, oral manifestations can be devastating and often under-recognized.[4,5] Enamel hypoplasia, delayed eruption, oral blistering, mucosal scarring, ankyloglossia, and early tooth loss are among the most disabling features affecting function, nutrition, and psychosocial well-being. This case report documents a young female with EBS-DM presenting with extensive cutaneous and dental findings. It highlights the need for coordinated dermatologic-dental management, supported by examples from similar reports.

2. Case Report

A 21-year-old female presented to the department of oral medicine and radiology with complaints of persistent pain and food impaction in the lower posterior region of the oral cavity. She also reported marked thermal sensitivity and

*Corresponding author: Jyoti C
Email: jcp7760@gmail.com

https://doi.org/10.18231/j.jooo.67659.1759123213

significant difficulty in maintaining oral hygiene, attributed to longstanding mucosal fragility and orofacial discomfort. The patient's medical history revealed the presence of recurrent vesiculobullous eruptions since birth, predominantly affecting the face, limbs, and trunk. These lesions typically ruptured spontaneously, resulting in serous discharge and healing with atrophic scarring. Although no familial history of similar dermatological manifestations was elicited, the patient was born of a consanguineous union. Extraoral examination revealed characteristic perioral scarring, hyperpigmented macules over the extremities, dystrophic nails, and digital contractures with webbing, which contributed to compromised manual dexterity and impaired oral hygiene practices (**Figure 1**).

Intraoral findings included generalized enamel hypoplasia, evidenced by enamel pitting, discoloration, and surface roughness. The mandibular left first molar (tooth 36) exhibited complete loss of coronal structure due to severe attrition, while the mandibular right first molar (tooth 46) was grossly carious, associated with tenderness suggestive of periapical involvement. Over-retained primary teeth were observed in regions 84, 64, 54, and 51. Erupting third molars were noted in areas 18, 38, and 48, and impacted permanent teeth were identified in the regions of 13, 21, 23, and 45. Additional findings included atrophic gingiva, although no active ulcerations were noted intraorally (**Figure 2**). A panoramic radiograph (**Figure 3**) corroborated the clinical findings, revealing multiple impacted permanent teeth (13, 21, 23, 45, 18, 38, 48), over-retained deciduous teeth (84, 64, 54, 51), extensive caries in tooth 46 with a periapical radiolucency, generalized thinning of enamel, root shortening, and evidence of alveolar bone loss.

Based on the constellation of mucocutaneous, dental, and radiographic findings, a provisional diagnosis of Epidermolysis Bullosa was made by the department of oral medicine and radiology, and the patient was referred for dermatological and genetic evaluation.

To confirm the diagnosis, a peripheral blood sample was collected in an EDTA vial and submitted to a certified molecular diagnostics laboratory. Genomic DNA was extracted using a spin-column–based method, and next-generation sequencing (NGS) was conducted. Genomic DNA was extracted using a spin-column–based method, and a custom epidermolysis bullosa gene panel was employed for next-generation sequencing (NGS), targeting both hotspot and non-hotspot regions of EB-associated genes, including KRT5, KRT14, COL7A1, and others. Library preparation was carried out using hybrid-capture enrichment, and sequencing was performed on the Illumina platform. The analysis identified a heterozygous missense mutation in the KRT14 gene (c.373C>T; p.Arg125Cys), which is characteristically associated with the Dowling-Meara subtype of Epidermolysis Bullosa Simplex. The variant was classified as pathogenic according to the American College of Medical Genetics (ACMG) guidelines. Although parental genetic testing could not be performed due to logistical constraints, the patient's consanguineous background and clinical features further supported the molecular diagnosis. The results were clearly explained to the patient, who was then referred for genetic counseling to understand the hereditary implications and to aid in future family planning.

In view of the pronounced mucosal fragility and heightened risk of trauma-induced blistering, invasive dental procedures were deferred. The patient was advised to maintain meticulous but gentle oral hygiene using ultra-soft toothbrushes and alcohol-free mouth rinses. Topical fluoride therapy was initiated, and future dental interventions were planned to be performed under controlled conditions using minimally invasive techniques. A structured three-month preventive recall protocol was established. Additionally, the patient was referred for psychological support and continued genetic counseling to address the broader emotional, systemic, and hereditary concerns related to her condition.

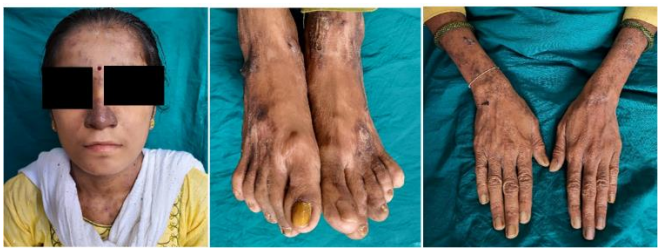

Figure 1: Extraoral image showing perioral scarring and hyperpigmentation

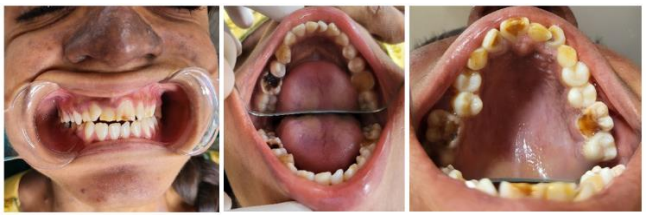

Figure 2: Intraoral photo revealing generalized enamel hypoplasia and grossly decayed molars

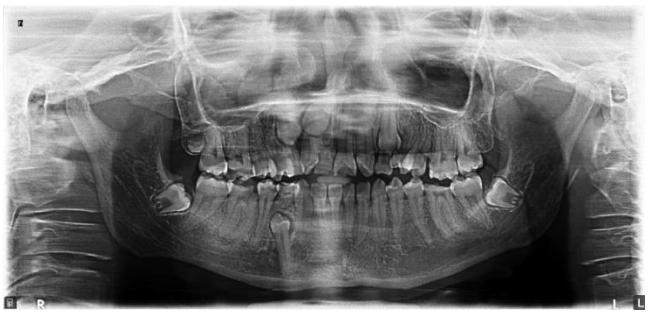

Figure 3: Panoramic radiograph showing impacted teeth, retained deciduous teeth, periapical radiolucency, and enamel thinning

3. Discussion

EBS-DM is the most severe variant of EBS and is linked to autosomal dominant mutations in the KRT5 and KRT14 genes.[6] These keratins are essential for maintaining the structural integrity of basal keratinocytes; mutations result in cytoskeletal instability, leading to intraepidermal cleavage and blistering with minimal trauma. Clinically, EBS-DM presents with grouped blisters in a herpetiform pattern, nail dystrophy, palmoplantar keratoderma, and progressive scarring.[7]

Molecular diagnostics, including gene sequencing of KRT5 and KRT14, can confirm diagnosis and guide prognosis in EB patients.[8]

Oral manifestations of EB are frequently under-recognized but have significant functional and psychological impacts.[4] In EBS-DM, enamel hypoplasia is common and can resemble amelogenesis imperfecta. However, the key distinction lies in the coexistence of mucosal fragility and systemic vesiculobullous features. Wright et al. demonstrated that enamel in EB patients is structurally compromised due to defective ameloblast function and is further exacerbated by systemic inflammation and nutritional deficiencies during odontogenesis.[5]

Intraoral features such as enamel hypoplasia, rampant caries, retained primary teeth, ankyloglossia, and depapillated tongue are consistent with literature reports.[5] Additionally, delayed tooth eruption has been reported in EB patients, likely due to chronic inflammation, altered development, and nutritional deficits. Other similar dental anomalies in patients with dystrophic EB, including retained teeth, root shortening, and early tooth loss.[9] Panoramic radiography is often preferred over intraoral imaging due to the risk of inducing mucosal trauma from film placement.

Management of dental care in EB patients requires a minimally invasive approach tailored to reduce trauma. Atraumatic extractions and fluoride therapy are mainstays of care. Oral rehabilitation approaches such as prosthetic interventions and preventive sealants may also be beneficial in selected cases, particularly in young patients with advanced dental damage. Additional preventive strategies include use of CPP-ACP, silver diamine fluoride (SDF), and desensitizing agents.[10] In the current case, atraumatic extractions were carried out under topical anesthesia, and fluoride varnish was applied to promote remineralization and reduce dentinal hypersensitivity. Behavioral guidance with use of ultra-soft toothbrushes and non-alcoholic rinses was emphasized.

Multidisciplinary care is vital in EB to address systemic, dental, psychological, and genetic concerns. The chronic nature of EB, coupled with visible disfigurements and pain, significantly affects patients' emotional and social well-being, underscoring the need for psychosocial intervention. Amerio et al. stressed the importance of coordinated oral-dermatologic management in improving quality of life and reducing complications.[11] Our patient benefited from joint care involving oral medicine, dermatology, psychological counseling, and genetic consultation—highlighting a model of holistic EB care.

A similar case reported by Kar et al. described generalized enamel hypoplasia and multiple impacted teeth in a patient with EBS-DM, with clinical manifestations closely paralleling our findings.[12] In contrast, a case presented by Agrawal et al. involved a patient with the same subtype who exhibited complete anodontia and required extensive oral rehabilitation with full dentures.[13] Both cases, along with ours, underscore the phenotypic variability in oral manifestations and the need for patient-specific treatment protocols. In another study by Yoon et al., a pediatric patient with EBS-DM presented with severe early-onset gingival erosions, requiring tailored periodontal interventions and protective dental splints.[14] Such comparative observations further reinforce the importance of early diagnosis and multidisciplinary preventive strategies.

Future directions in EB management are increasingly focusing on curative strategies, including gene therapy, protein replacement, and stem cell transplantation. Sun et al. demonstrated long-term skin regeneration using ex vivo-corrected autologous keratinocyte stem cells. Similarly, phase I clinical trials using gene-modified skin grafts show promising results in reversing disease phenotype.[15] Although these therapies are in early phases, they underscore the potential for transformative treatments in severe EB subtypes.

EBS-DM presents a complex oral and systemic disease burden. This case reaffirms the necessity for early diagnosis, trauma-free dental protocols, and comprehensive interdisciplinary management to enhance functional outcomes and improve the patient's quality of life.

4. Conclusion

This case underscores the critical importance of recognizing oral signs of EB in early stages. Prompt, trauma-free dental care, preventive measures, and coordination with dermatologic and genetic teams are vital to manage disease burden. Individualized care improves not only oral function but also overall well-being and self-esteem in affected individuals.

5. Patient Consent

Obtained in writing.

6. Ethical Approval

Not required for case reports

7. Source of Funding

None.

8. Conflict of Interest

None.

References

1. Fine JD, Bruckner-Tuderman L, Eady RA, Bauer EA, Bauer JW, Has C, et al. Inherited epidermolysis bullosa: updated recommendations on diagnosis and classification. *J Am Acad Dermatol*. 2014;70(6):1103–26. https://doi.org/10.1016/j.jaad.2014.01.903.
2. Horn HM, Tidman MJ. The clinical spectrum of dystrophic epidermolysis bullosa. *Br J Dermatol*. 2002;146(2):267–74. https://doi.org/10.1046/j.1365-2133.2002.04607.x.
3. Has C, Bauer JW, Bodemer C, Bolling MC, Bruckner-Tuderman L, Diem A, et al. Consensus reclassification of inherited epidermolysis bullosa and other disorders with skin fragility. *Br J Dermatol*. 2020;183(4):614–27. https://doi.org/10.1111/bjd.18921.
4. Wright JT. Oral manifestations in the epidermolysis bullosa spectrum. *Dermatol Clin*. 2010;28(1):159–64. https://doi.org/10.1016/j.det.2009.10.022.
5. Wright JT, Fine JD, Johnson L. Dental caries risk in hereditary epidermolysis bullosa. *Pediatr Dent*. 1994;16(6):427–32.
6. Khani P, Farokh Forghani S, Ataei Kachoei Z, Zekri A, Ghazi F. Analysis of KRT5 and KRT14 gene mutations and mode of inheritance in Iranian patients with clinical suspicion of Epidermolysis bullosa simplex. *Med J Islam Repub Iran*. 2020;34:43. https://doi.org/10.34171/mjiri.34.43.
7. Mariath LM, Santin JT, Schuler-Faccini L, Kiszewski AE. Inherited epidermolysis bullosa: update on the clinical and genetic aspects. *An Bras Dermatol*. 2020;95(5):551–69. https://doi.org/10.1016/j.abd.2020.05.001.
8. Zhang J, Yan M, Liang J, Li M, Yao Z. A novel *KRT5* mutation associated with generalized severe epidermolysis bullosa simplex in a 2-year-old Chinese boy. *Exp Ther Med*. 2016;12(5):2823–6. https://doi.org/10.3892/etm.2016.3717.
9. Yavuz Y, An I, Yazmaci B, Akkus Z, Ortac H. Evaluation of clinical and oral findings in patients with epidermolysis bullosa. *Medicina (Kaunas)*. 2023;59(7):1185. https://doi.org/10.3390/medicina59071185.
10. Al-Nerabieah Z, AlKhouli M, Dashash M. Preventive efficacy of 38% silver diamine fluoride and CPP-ACP fluoride varnish on molars affected by molar incisor hypomineralization in children: a randomized controlled trial. *F1000Res* [Internet]. 2024 [cited 2025 Sep 22];12:1052. Available from: https://doi.org/10.12688/f1000research.136653.3.
11. Amerio P, Ferrucci SM, Galluzzo M, Napolitano M, Narcisi A, Levi A, et al. A multidisciplinary approach is beneficial in atopic dermatitis. *Dermatol Ther (Heidelb)*. 2024;14(6):1443–55. https://doi.org/10.1007/s13555-024-01185-1.
12. Kar AK, Parkash H, Jain V. Full-mouth rehabilitation of a case of generalized enamel hypoplasia using a twin-stage procedure. *Contemp Clin Dent*. 2010;1(2):98–102. https://doi.org/10.4103/0976-237X.68601.
13. Agrawal PV, Lahoti KR, Rathi N, Jagtap A, Tasgaonkar A, Kotnis R. 3D printed denture for a pediatric patient with complete anodontia: a case report. *Int J Clin Pediatr Dent*. 2025;18(1):114–8. https://doi.org/10.5005/jp-journals-10005-3027.
14. Yoon RK, Ohkawa S. Management of a pediatric patient with epidermolysis bullosa receiving comprehensive dental treatment under general anesthesia. *Pediatr Dent*. 2012;34(3):251–3.
15. Sun BK, Siprashvili Z, Khavari PA. Advances in skin grafting and treatment of cutaneous wounds. *Science*. 2014;346(6212):941–5. https://doi.org/10.1126/science.1253836.

Cite this article: Jyoti C, Ramdurg P, Srinivas N, Harihar LS, Fazil M, Arabbi HK. When skin and smiles shatter: An extraordinary case of epidermolysis bullosa dowling-meara with devastating oro-dental involvement. *J Oral Med Oral Surg Oral Pathol Oral Radiol*. 2025;11(3):112–115.