Dental Rehabilitation in a 3-Year-Old With Junctional Epidermolysis Buallosa

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Introduction

Junctional epidermolysis bullosa (JEB) is a rare genetic condition marked by extreme fragility of the skin and mucosal membranes. Among the four EB subtypes, JEB is the least common, comprising about 1% of cases. It typically presents at birth and, in severe forms, is associated with high mortality—up to 40% within the first year and 62% by adolescence.

Oral manifestations include enamel hypoplasia, mucosal blistering, gingival bleeding, and feeding difficulties, often leading to poor growth. Despite the known dermatologic burden of JEB, dental management remains underreported and must be approached with caution.

Case Description

A 3-year-old girl, known to have JEB since birth, presented to the PSMMC pediatric dental clinic with oral discomfort, gingival bleeding, and sensitivity to food and beverages. Her mother reported feeding difficulties and decreased food intake due to pain. She was born to consanguineous parents and has a history of skin blistering and erosions, especially over pressure-prone and trauma-exposed areas. Oral symptoms significantly interfered with her nutritional status and oral hygiene practices.

Figure 1.

Genetic Findings

Whole-exome sequencing revealed a homozygous likely pathogenic variant in the COL17A1 gene: NM_000494.3:c.2068G>T, resulting in p.(Glu690*), a nonsense mutation associated with JEB.

Clinical Findings

Extraoral

- Generalized scarring, erosions, hypopigmentation
- Areas of healed and active blistering.

Intraoral

- Fragile gingiva with spontaneous bleeding
- Generalized enamel hypoplasia (mild to severe)
- Gingival inflammation
- Carious lesions on primary molars
- Pitting and irregularities on anterior teeth
- Reduced mouth opening
- Bullae noted on the floor of the mouth and tongue

Treatments

Planning & Precautions

Due to the extent of caries, tissue fragility, and limited tolerance for intraoral procedures, full mouth rehabilitation was planned under general anesthesia.

Transport & Positioning:

- Patient was gently lifted using a blanket to avoid skin contact.
- All surfaces (e.g., stretchers, tables) were padded to reduce pressure and friction injuries.

Preoperative Protection:

- Non-adherent dressings were applied to all pressure points and vulnerable skin areas.
- ECG leads, pulse oximeter probes, and BP cuffs were secured using non-adhesive techniques.
- A foam interface was placed beneath IV hub

Anesthesia

Induction:

- Oral midazolam (0.5 mg/kg) was administered preoperatively.
- Due to chronic nasal stenosis and mucosal fragility, nasal intubation was avoided

Intubation:

- A smaller-diameter endotracheal tube was carefully inserted via oral route using a fiber optic bronchoscope.
- The tube was lubricated with water-based gel.

Intraoperative Protection:

- A pre-moistened throat pack was placed.
- Suction tips were only applied to hard surfaces (teeth).

Restorative Treatment

- **Primary molars:** Full-coverage stainless-steel crowns (SSCs) were placed to restore function, protect compromised enamel
- **Anterior teeth:** Composite restorations were used to seal pitting and enamel defects.
- **Atraumatic technique:** High polishing of restorations and careful instrumentation minimized mechanical irritation.

Preventive Measures

- Fluoride application every 3 months.
- Caregiver instruction included:
- Brushing with a small-headed, soft-bristled toothbrush soaked in warm water.
- Use of non-irritating oral care products.
- Dietary counseling to avoid acidic and abrasive foods.

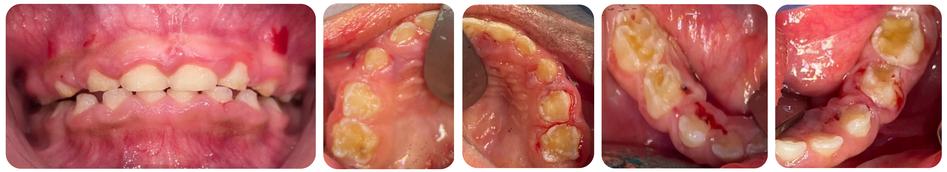

Figure 2. Pre-treatment Intraoral Photograph

Figure 3. 6 Months Follow-up Photograph

Postoperative Care & Outcome

Postoperative Care:

- **Pain Management:** Oral analgesics and topical fusidic acid for wound care.
- **Magic Mouthwash Application:** Tapped gently on affected gingiva using pre-soaked and squeezed gauze to avoid rubbing trauma.
- **Lip and Skin Care:** Ongoing use of emollients and protective dressings for any facial blistering or crusting.
- **Nutritional Support:** Soft, non-acidic diet to minimize oral trauma.

6-Month Follow-Up Findings:

- Marked reduction in gingival inflammation
- Improved oral hygiene
- No new carious lesions
- Restoration stability maintained
- The patient resumed normal eating and demonstrated healthy weight gain

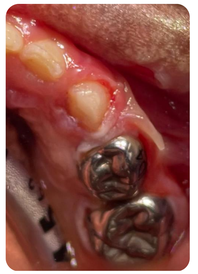

Figure 4. Immediately After Procedure

Discussion

JEB poses distinct challenges in pediatric dentistry due to its significant oral and systemic implications.

Oral Manifestations patients often present with enamel hypoplasia, dental caries, gingival fragility, and oral blistering—contributing to pain, feeding difficulties, and malnutrition.

Genetic Basis mutations in genes like COL17A1 impair hemidesmosomal proteins essential for enamel development, increasing susceptibility to dental defect.

Multidisciplinary Coordination is essential in JEB management, especially involving pediatric dentists, dermatologists, anesthesiologists, and nutritionists.

Dental treatment restorative techniques should prioritize long-term durability with minimal manipulation of tissues.

Anesthesia Planning must be personalized to avoid mucocutaneous trauma, with careful airway management and atraumatic monitoring.

Prevention and Education are critical—caregiver training, dietary counseling, and fluoride protocols help maintain oral health and reduce recurrence.

Nutrition malnutrition is a common concern in JEB, often resulting from painful oral lesions that limit intake. A soft, non-irritating diet and tailored nutritional support are essential components of care.

Early Referral to dental care is vital to initiate preventive measures before complications develop.

Conclusion

Despite the complexity of JEB, positive outcomes are achievable with early intervention and multidisciplinary care, children with JEB can maintain improved oral function, reduced discomfort, and better overall quality of life.

References: