

## INTRODUCTION

Epidermolysis Bullosa (EB) is a rare inherited disorder that is characterized by fragile epithelium that results in blisters following minimal trauma.<sup>1</sup> Dystrophic ED results from a type VII collagen mutation, leading to separation below the basement membrane, and progressive scarring.<sup>2</sup> In the oral cavity, EB significantly impacts oral hygiene, leading to increased caries risk. Scarring can cause limited opening which reduces access for restorability, and ultimately a reduction in quality of life.

## ETIOLOGY AND EPIDEMIOLOGY

Dystrophic EB is caused by a mutation in the COL7A1 gene, impacting type VII collagen, leading to significant oral and perioral involvement.<sup>1</sup> It can be both autosomal dominant, and recessive with the latter being more severe. Recessive dystrophic EB affects approximately 1 in 500,000 people. It is a lifelong condition with progressive scarring in skin, mucous membranes, esophagus, and other areas as well.

## DIAGNOSIS AND MANAGEMENT

Diagnosis for EB includes evaluating the clinical presentation, conducting a family history, and a skin biopsy. However, the gold standard for diagnosis is genetic testing.

Management of patients with EB include minimizing trauma to tissues through lubrication of the lips and tissues, being gentle with the tissues, avoiding any frictional forces, and using smaller instruments for intraoral use.<sup>3</sup>

Focusing on prevention is essential for lifelong oral health through frequent recall visits, minimally invasive techniques and education for families.

## CASE REPORT

A 21-year-old female presented to Healthy Smiles for Kids by AltaMed for a recall exam with chief complaint of broken teeth. Clinical exam noted severe mucosal fragility, limited opening, extensive scarring, multiple broken teeth, failing restorations, and high caries presence.

Patient reported difficulty finding providers comfortable treating her teeth due to her medical conditions.

Patient was unable to take intraoral radiographs on the day of recall, but panoramic radiographs were obtained that day as well as from previous dental visits.



Figure 1 Panoramic radiograph at previous visit in 2019



Figure 2 Panoramic radiograph at recall exam in 2024

## DENTAL IMPLICATIONS or CLINICAL FEATURES

EB has many features that are associated with oral complications.

Soft tissue findings include fragile mucosa, recurrent blistering leading to scarring, and ulcerations. The scarring can lead to vestibular obliteration, and microstomia, which can make restorative management and dental access difficult.<sup>2</sup>

Patients have an increased caries risk due to plaque accumulation secondary to limited ability to maintain oral hygiene. Patients can have increased enamel defects (more common in junctional EB) which increases the risk for caries.

Due to these challenges, comprehensive dental care is often indicated in a hospital setting. Due to the extent of decay and the complications of the disease more definitive treatment like extractions will be needed. The challenge becomes that as these children age out of pediatric hospitals finding an adult facility to treat them is often difficult if not impossible.

For our patient, we are working with a pediatric dentist in San Diego for multidisciplinary care including an oral surgeon for full mouth extractions and possible immediate implant placements. A multidisciplinary approach including prosthodontic consultation will be necessary.

## CONCLUSION

Dystrophic EB presents significant challenges for dental providers due to severe fragility of mucosa, limited opening, and increased caries risk.

Preventative focused care should be emphasized early in life to improve dental outcomes.<sup>4</sup> Management with interdisciplinary care is ideal to navigate the challenge of managing patients with this condition.<sup>5</sup> This case highlights the challenges we face as pediatric dentist to transition children with complex medical conditions to adult providers. A conversation must begin to help identify solutions for treating complex medical conditions like EB as the patient transitions out of pediatric dental specialty care.

## REFERENCES

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