Oral manifestations and dental care of epidermolysis bullosa patient: a case report

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ABSTRACT

Aim: The aim of this report is to present clinical features of patients with Epidermolysis Bullosa (EB) and describe effective methods of dental management for these individuals. Case Report: Epidermolysis Bullosa (EB) is a rare inherited disorder with skin and oral manifestations. The patient in this report was 25-year-old female, has been diagnosed with Junctional EB at birth, presented to
University at Buffalo clinics for a routine dental appointment. Clinical examinations revealed classical mucocutaneous features of EB such as; multiple hemorrhagic bullae with scar tissues, limited mouth opening, dental caries, poor oral hygiene, enamel pitting, and occlusal wear. A proper dental management care was provided. Conclusion: This case report presented clinical features of individuals with Epidermolysis bullosa (EB), focusing on proper methods of dental care for affected patients by this inherited disorder.

Keywords: Epidermolysis Bullosa, Bullae, Blister, Case Report

1. INTRODUCTION

Epidermolysis bullosa (EB) is a rare genetic disorder with mucocutaneous features of developing blisters and erosions due to severe epithelial damage, which may happen spontaneously or from any minor trauma. Because of severity of this autoimmune condition, it is usually detected at birth or early childhood but it can affect people from different age groups (Sawamura et al., 2010; Silva et al., 2004). Several types and subtypes of EB have been reported in the literature showing varying degree of severity in clinical presentations and complications. EB have four major types, simplex, junctional, dystrophic, and Kindler’s syndrome, and more than thirty different subtypes. These types and subtypes are classified based on the level of cleavage within the epithelial – connective tissue zone. One of the important factors that plays a role in the severity of clinical presentation is inheritance patterns (autosomal dominant / autosomal recessive) (Silva et al., 2004; Fine et al., 2008; Fine et al., 2014).

EB simplex is the most common and the mildest form of EB which leaves less scar tissue after skin blistering. Also, it may exhibit oral manifestations especially after trauma of oral mucosa. However, teeth in individuals with the simplex variant are similar to unaffected people in terms of dental caries and tooth wear (Liu et al., 1998). Unlike to the EB simplex type, the junctional form of EB considers the most severe variant and usually shows aggressive features affecting both skin and oral mucosa with increase tissue fragility and blisters ulceration. Mutated genes in junctional EB forms, such as; LAMA3, LAMB3, LAMC3, COL17A1, ITG6A, and ITGB4, affect normal formation of epithelial cells and teeth. Microstomia, less soft tissue flexibility with scar formation, blister ulcerations, pitting enamel hypoplasia, and dental caries are the most common oral presentations in the junctional types and subtypes of EB (Momeni & Pieper, 2005; Wright et al., 1994). Another common type of EB is the dystrophic form, which has multiple inheritance patterns that determine the severity of clinical phenotype. For example, the dominant dystrophic types are not fatal and usually show mild ulceration and limited oral involvement, however, the individuals with recessive inheritance pattern represent a severe form of EB that influence body functions causing limitations in movements especially in hands and feet (Das & Sahoo, 2004). Frequent secondary infections are common in generalized recessive cases because of healing challenges after the big ulcerations. In addition, severe dental caries, limited mouth opening and ankyloglossia are not uncommon oral features in recessive dystrophic types (Wojnarowska et al., 1983). Kindler syndrome is a rare condition, which shares features of blistering and ulcercations with the other forms of EB. This syndrome usually manifests in early infancy accompanied with photosensitivity, hyperkeratosis of palms and soles, and small clusters of blood vessels (Burch et al., 2006). Both the Kindler syndrome and the severe generalized recessive dystrophic form are at increased risk of developing squamous cell carcinoma (Weber et al., 2001).

The diagnosis of EB is usually confirmed by a skin biopsy, genetic analysis, and a prenatal testing if there is a family history (Hsu et al., 2014; Fasshi & McGrath, 2010). For treatment of EB, wounds care would be the proper treatment for mild cases and a multispecialty intensive care for the severe cases. For instance, the intensive care may include; systemic antibiotics to treat infections, surgical interventions for ocular complications, and dental restorations for decayed teeth with periodic screening for oral squamous cell carcinoma. All these medical services should be provided along with preventive methods to avoid serious and possible fatal complications (Krämer, 2010; Siqueira et al., 2008, Krämer et al., 2012). The aim of this article is to report a patient was diagnosed with EB, describing all the clinical and oral features and discussing the difficulties faced by the patient and clinicians to perform a proper dental management.

2. CASE REPORT

In 2017, a 25-year-old Caucasian female patient came to University at Buffalo, School of Dental Medicine, Post Graduate clinics for a routine dental appointment. During the clinical interview, the patient reported that she has been diagnosed with Junctional EB at birth. Extra oral examinations revealed multiple hemorrhagic bullae with scar tissues, and erosions throughout the face and body (Figure 1, A). Nail dystrophy was also present (Figure 1, B, C).
Figure 1 A. Facial scarring, and small bullae with erosions.

Figure 1 B. Hemorrhagic bullae with scar tissues, and erosions cover hands. Also, nail dystrophy was present in all fingers.
In addition, intra oral examination showed limited mouth opening, multiple ulcerations and erythematous oral mucosal tissues with vesicles (Figure 2, A, B, C, D). Dental caries with poor oral hygiene, and occlusal wear were noted (Figure 3). The patient was aware of the findings and she was well motivated to receive periodontal and restorative care. The etiological risk factors of the dental problems including soft diet, home dental care, and other systemic and local cofactors were discussed with the patient.

**Figure 1 C.** Similar characteristic features of Junctional EB, shown at Figure 1B which include hemorrhagic bullae with healing scar tissues, and erosions but at the palms of both hands.

**Figure 2 A.** Microstomia of the patient with junctional EB, causing by scar formation.
Figure 2 B. the buccal mucosa of the patient showing ulcerated blisters in the healing process.

Figure 2 C. Other fragile mucosal tissues with erythematous changes at the hard palate. Note, the occlusal wear of all teeth.

Figure 2 D. Similar ulcerated bullae on the tip and lateral border of the tongue.
The medical history of the patient was significant for neck pain and a recent cervical disc surgery. In addition, she was allergic to Trimethoprim/sulfamethoxazole, clindamycin, and doxycycline. For the dental history, the patient has received orthodontic treatment, root canal treatment, restorations, and extractions. She was also very sensitive to hot, cold and sweet drinks/food at most teeth. Bruxism (parafunctional grinding of teeth) has been reported as well. The patient continued to receive the required dental treatment, mainly scaling and dental fillings. Moreover, she was seeing her primary physician and dermatologist on regular basis.

3. DISCUSSION
Epidermolysis bullosa (EB) is a rare inherited disorder featured by blisters formation and erosions due to tissue fragility to any mild trauma, and it usually affects skin and mucosa (Sawamura et al., 2010; Silva et al., 2004; Fine et al., 2008). The case presented in this article showed the most common clinical features of EB, especially the junctional type. Skin features included; hemorrhagic blisters, atrophic nails, erosions, and scar tissue formation. Oral features were also presented including; extensive dental caries, mucosal ulcers, poor oral hygiene, and tooth wear (Winter & Brook, 1975; Kummer et al., 2013). These oral features are related to the disorder itself and also to inability to perform a good oral hygiene practice by the patient. It has been reported that EB individuals have a higher susceptibility to dental caries compared to healthy people (Wright et al., 1994). All the oral and dental findings in this case report were consistent with the literature that has reported similar presentations and outcomes. Therefore, preventive dental programs are essential to maintain teeth by providing oral hygiene instructions, using a special handle for toothbrush, rinsing with water frequently, receiving regular dental services and implementing dietary modifications.

Because there is no cure for EB, most of the management protocols are supportive to relieve pain and help affected individuals to live a better life physically and emotionally. Maintaining an active patient – medical provider relationship is critical to keep the patient safe from secondary infections or worsening the complications. On the other hand, dental clinicians should adjust their instruments and procedures to overcome some challenges such as limited mouth opening, dry mouth, or mucosal ulcers. In such cases, pediatric instruments and lubricants for oral mucosa are basic requirements in order to provide a proper dental management. Moreover, local anesthesia may cause some blistering, so, it should be avoided when it is possible. In general, precautions need to be performed to protect skin and oral mucosa of the patient every single visit.

4. CONCLUSION
Patients with bullous conditions that have dermal and mucosal manifestations, such as Epidermolysis bullosa (EB), require a special and periodic dental care. All dentists and dental hygienists should understand the needs of these patients to provide appropriate preventive and dental services. This report presented difficulties faced by the dental provider and discussed methods of management making the patient fully involved and have some responsibilities to ensure better home care and outcomes.

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Informed consent: Written & Oral informed consent was obtained from the patient included in this case report. Additional informed consent was obtained from all individual participants for whom identifying information is included in this manuscript.

REFERENCE