Epidermolysis Bullosa: what the dental practitioner needs to know- a case report

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Abstract:

The term Epidermolysis Bullosa (EB) encompasses a rare group of disorders characterized by structural fragility within the skin and selected other tissues, resulting in recurrent blister formation. The blisters break and inevitably lead to atrophic scarring. The clinical presentation may range from blistering of the hands and feet only to a more generalized blistering of the skin and oral cavity, and injury to many internal organs. While the cause is largely hereditary, a rare acquired form is also present. Diagnosis and subclassification is usually made through detailed family history, along with the results of immunofluorescence; antigenic mapping, transmission electron microscopy, and in some cases, by DNA analysis.

The oral and dental findings are characteristic, and often the reason why the patient seeks medical care. Hence the dentist plays a key role in the diagnosis and early management of the disease. In this article we present an interesting case of EB with characteristic oral and radiographic features, and special emphasis is paid to highlighting the diagnostic and management protocol to help the general dental practitioner.

Key words: Epidermolysis, Bullosa (EB), Enamel defects, Bulla.

Introduction:

Epidermolysis bullosa (EB), a term used to describe a group of rare, mainly hereditary, chronic, non-inflammatory diseases of mucous membranes and skin, is characterized by the development of bullae as a result of mild to moderate trauma. The bullae easily burst, causing ulcerative lesions which usually heal with scarring. Though the inheritance is either autosomal dominant or recessive, there is also an acquired form. The hereditary types of EB present at or within a few weeks of birth, can affect all populations and racial groups and there is no apparent predilection for males or females.

With 3 main types of inherited forms, and more than 23 subtypes along with an acquired form, most classifications are either based upon mode of inheritance, anatomic location or distribution of lesions and associated morbidity. The most generally acknowledged and simplest classification is the one based on the anatomical level of tissue cleavage [1, 2].

Type I. Intraepidermal forms (Epidermolysis bullosa simplex).

Type II. Junctional forms (Junctional epidermolysis bullosa).

Type III. Dermal forms (Epidermolysis bullosa dystrophica dominant and recessive).

Type IV. Epidermolysis bullosa acquisita.

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Ten distinct genes encoding key molecular components of hemidesmosome anchoring complex and associated keratin filament network are recognized as underlying the three major forms of EB, and several sub-types of Epidermolysis Bullosa Simplex\(^{[3,6]}\).

All three main types of EB produce oral defects which are very pathognomonic. Bullae can be initiated on skin or mucous membranes at sites of trauma or pressure (Nikolsky’s sign) and on rupturing they leave painful erosion which heals with scar formation. As the oral mucosa is a site of constant minor trauma while eating food or tooth-brushing, this oral mucosal scarring and contracture can lead to tongue-tie, obliteration of the sulci, limited opening, lingual depapillation and atrophy of the palatal folds. The cicatrization following blistering can lead to entrapment of epithelial cells particularly in the hard palate, and give rise to milium cysts. Defects in enamel are a common feature and may present with thinned enamel and localized or generalized hypoplasia. Fine or coarse enamel pitting defects or thin or uneven enamel lacking prismatic structure is also common. The amelodentinal junction has been found to be smooth but the mineral and chemical composition of dental enamel in EB is no different from normal and does not predispose the teeth to caries, although the prevalence of dental caries is significantly increased in individuals with junctional EB and recessive EB. The fingers of patients with EB are often destroyed with resorption of phalanges, and in advanced cases, hands can appear club-shaped. Trauma to limbs leads to bulla formation and subsequent healing with scarring, which is prominent on the knuckles and knees. Management of patients with EB presents its own difficulties, and since the oral manifestations can be painful and warrant immediate attention, the dentist plays an important role in the multi-specialty care. We present a previously undiagnosed case of EB with characteristic oral findings.

**Case Report:**

A 16-year old male patient presented for dental care with a chief complaint of stains on his teeth. The patient gave a history of getting wounds quite easily on his hands and legs, particularly at the joints, which healed with scarring. General physical examination showed multiple abrasions on his knees and shins, and scarring on his knuckles (Fig 1, 2). The patient also said that he occasionally developed blisters in his mouth which would burst to leave painful ulcers, only to heal in about 1 week, but at the time of presentation he did not have any of these complaints.

Intra-oral examination revealed severe generalized pitting defects of enamel with superficial staining of teeth (Fig 3). A panoramic radiograph was taken which revealed the generalized nature of the enamel pitting defects (Fig 4). The oral mucosa appeared normal. An incisional biopsy from the skin lesions confirmed the diagnosis of Epidermolysis Bullosa Simplex.
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The patient was counseled on his condition and advised to undergo genetic testing. However, he only wanted to get his teeth cleaned and hence underwent thorough oral prophylaxis and was lost to follow-up.

Discussion:

With the major types and several subtypes of EB, the clinical presentations vary depending on the disease. Symptoms may be severe with patients presenting with extensive scarring and fibrosis of the hands (Mitten-deformity), to mild blisters and scarring at the joints. As our patient had EB Simplex form, his skin lesions were not very extensive. The dental appearance however was of concern to him. What makes this case interesting is that he was a previously undiagnosed case of EB, and the dental and clinical findings led us to follow through with a biopsy that confirmed the disease process.

The goal of treatment of all patients with EB is to prevent blisters from forming and complications. How much treatment is needed depends on how severe the disease is. Recommendations often include avoiding skin damage (trauma) and hot environments. If any blistered areas become crusted or exposed, application of an antibiotic ointment to wound-like areas would be necessary.

The Dental considerations in management of patients with EB also depend on the degree of severity of the disease. For swallowing difficulties, oral steroids for short periods of time are recommended. Long-term steroids however are generally not advised. Candida infection that may develop in the mouth or oesophagus following steroid use would advocate the use of topical antifungal medications. The patient must be advised to incorporate regular dental visits into his routine as the incidence of caries is slightly high in patients with EB. However, the cause of caries is probably due to lack of oral cleansing. There is no direct relationship between the extent of oral blistering and caries experience. The salivary flow rate has been investigated and no difference was found between EB individuals and controls. Dental extractions or minor oral surgical procedures can successfully be done under general anesthesia[7] or intra-ligamentary technique to minimize surface trauma.

Along with the above methods, the dentist must be aware that proper nutrition is also important as protein helps injured skin tissue to recover. Patients must be advised to avoid eating hard or brittle foods such as nuts, crusty pizza, hard toast and the like.

Referral to a physiotherapist to help maintain joint motion and limit contractures or to a plastic surgeon for grafting of skin from denuded areas may be warranted.

It is important to know that the acquisita form of EB may be treated with oral steroids and other medications that suppress the immune system. However, as with all immunosuppressants, these increase the risk of infection or damage the liver or kidneys.

Studies using interferon, gene therapy, protein replacement therapy and cell-based therapies are underway and show promising results[8].

Areas of leukoplakia and oral squamous cell carcinoma (OSCC) have also been reported to develop in patients with EB, particularly affecting the lingual mucosa[9,10]. Hence the role of the dentist should not be underestimated in helping reach a timely diagnosis, thereby preventing more serious complications from occurring.
References:


