Hereditary Dystrophic Epidermolysis Bullosa A Rare Case Report

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Abstract:

Epidermolysis bullosa (EB) is a diverse group of disorders characterized by blister formation with tissue separation occurring at variable depths in the skin and oral mucosa depending on the specific EB type. Oral tissue fragility and blistering is common to all EB types. However oral debilitation as a result of soft tissue scarring is primarily limited to the recessive dystrophic EB subtypes. Due to extreme mucosal fragility such patients are managed by modified dental care practices. This case report describes a rare case of recessive dystrophic EB.

Keywords: Dystrophic, Epidermolysis bullosa, Recessive

Introduction

Epidermolysis bullosa (EB) represents a group of hereditary skin disorders manifested by an exceptional tendency of the skin and mucosa to form bullae and vesicles as a result of trauma or friction. Although the specific pathogenesis of these disorders still remains unknown, bullae formation has been associated with various basic defects including structural and/or biochemical abnormalities of keratin, hemidesmosomes, anchoring fibrils, anchoring filaments, and physicochemically altered skin collagenase.[1] Recent genetics studies have linked certain EB types to a keratin type VII collagen gene defect.[2]

Around 23 distinct types of EB have been reported, each displaying its unique clinical presentation, extracutaneous involvement, mode of inheritance, and level of tissue cleavage. However, these subtypes are classified into three main groups based on the level of tissue separation that develops following mechanical trauma to the skin.[3] Blistering occurs within the epidermis, within the basement membrane, or beneath the basement membrane in simplex, junctional, and dystrophic forms of inherited EB, respectively. The ultrastructural level of separation in blistered tissue is determined using transmission electron microscopy and/or immunofluorescence anti-genic mapping.[4]

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Characterizing morphologic features- including the hemidesmosomes, anchoring fibrils, and subbasal dense plates and the relative expression of numerous basement membrane-specific antigens, such as type VII collagen, GB3,19DEJ-1, and chondroifin 6-sulfate proteoglycan- also are useful aids in further determining EB types and subtypes.[4]

Mode of inheritance and clinical features, such as the severity and distribution of cutaneous and extracutaneous findings, has also been considered in the final classification of each EB subtype.[5]

Cutaneous findings show great variation and may include blisters, crusted erosions, milia, scarring, granulation tissue, pigmentation changes, cicatricial alopecia, and absence or dystrophy of nails. Extracutaneous involvement may involve the eyes, teeth, oral mucosa, esophagus, intestinal tract, anus, genito-urinary tract, and musculoskeletal system.[6] In general, specific EB subtypes tend to display characteristic combinations of cutaneous and extracutaneous features, although there may still be considerable clinical variation in the type and severity of these manifestations.

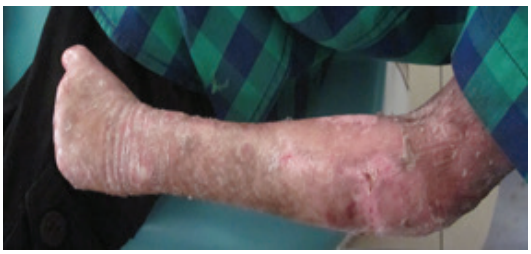
Here we present a rare case of hereditary dystrophic epidermolysis bullosa.

Case Report:

A 15 year old male patient reported to the Department of Pedodontics and Preventive Dentistry with a chief complaint of pain in upper front tooth region since one week. On eliciting the medical history, it was found that he was a known case of hereditary dystrophic epidemolysis bullosa. His natal, neonatal and family

Upon physical examination, bullae were present all over the body, showing various stages of eruption and healing. Hands and feet showed digital webbing and severe mitten deformities resulting from the continual process of blistering and scarring around the digits (Fig 1).

Patient showed limited mouth opening due to perioral scarring (Fig 2 and 3). Intraorally there was presence of oral mucosal bullae, bleeding and plaque accumulation (Fig 4). Root stumps were present involving 16, 12, 21, 22, 26, 36 and 46. Deep carious lesions were present in 31, 41, 37 & 47. OPG confirmed the clinical findings showing numerous root stumps and carious teeth. Accordingly the treatment was planned and the patient is scheduled for treatment.



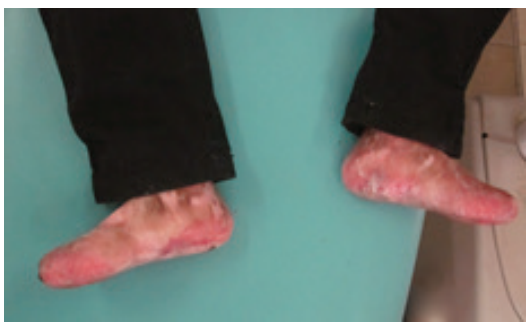


Fig 1. Hand and feet showing digital webbing and severe mitten deformities

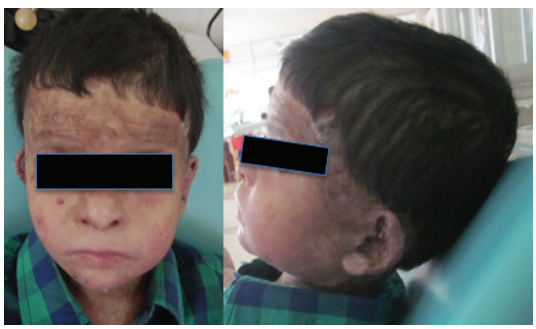


Fig 2: Frontal and lateral view of the face

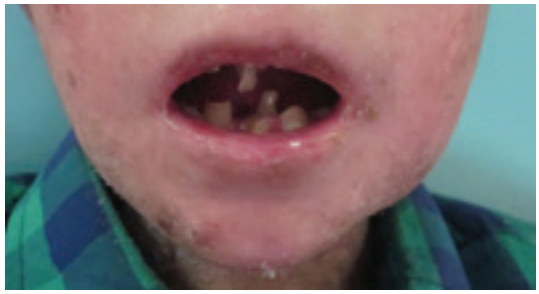


Fig 3: Peri-oral scarring and limited mouth opening

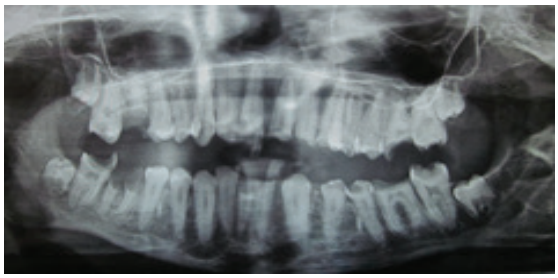


Fig 4: OPG showing multiple carious teeth

Discussion

Due to its rare occurance and scarcity of literature on dental treatment of patients suffering from epidermolysis bullosa, the management of such patients presents as a challenge for the dentists. Apart from limited mouth opening and perioral and intraoral bullae associated with scarring, the uncooperative behaviour of such patients further complicates the management.

General management consists primarily of palliative topical care. Eroded skin surfaces can be covered with nonadherent dressings after applying a topical antibiotic such as bacitracin, silver sulfadiazine, or mupirocin. Oral nutritional supplements including iron and zinc may be beneficial in individuals suffering from anemia, and liquid preparations high in protein and calories may help patients with growth retardation. Dental management depends on the severity of the condition. If the patient shows milder form, then routine dental treantment with few modifications can be carried out. However in severe cases showing extreme fragility of oral mucosa, altered approach to oral rehabilitaion and anaesthetic management. When administering local anesthesia, the anesthetic solution should be injected deeply into the tissues slowly enough to prevent tissue distortion, which may cause mechanical tissue separation and blistering.

Nerve blocks are far less likely to form blisters since they do not place the mucosal surface under pressure by depositing a bolus of fluid near the tissue surface. When manipulating tissues of individuals with those EB types most prone to mucosal blistering (severe generalized recessive dystrophic EB), only compressive forces should be applied because these are less likely than lateral traction or other shear forces to induce tissue separation. Lubricating the patient's lips and any tissue to be contacted also will reduce the likelihood of shear forces and tissue damage.[7] The generalized enamel hypoplasia characteristic of junctional EB often is best managed in the child with stainless steel crowns as to protect all of the teeth. Patients with severe generalized dystrophic EB who develop extensive dental caries frequently require stainless steel crowns on all of the primary teeth.[8]

Preventing dental caries is most challenging in individuals with severe mucosal involvement since they often are faced with an extremely cariogenic diet and are least able to able to perform routine preventive procedures. In patients prone to oral blistering oral hygiene may best be accomplished with a soft-bristled, small-headed tooth-brush. In addition to systemic administration of fluorides, fluoride rinses also may help control caries.[9]

Hereditary dystrophic EB presents as a challenge for dental practitioner as described in our case report. However by employing modified treatment approach such cases can be handled efficiently. Hence dentists should be aware of the condition, its various systemic and oral manifestations and methods for management of such cases.

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