European Journal of Research in Dentistry 2018; 2-1: 23-27
DOI: 10.12990/MDJ.2018.14

CASE REPORT / OLGU SUNUMU

Clinical Findings of Children With Epidermolysis Bullosa: Case Reports of Three Siblings

Büllozalı Çocukların Klinik Bulguları: Üç Kardeş Vaka Çalışması

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Abstract

Epidermolysis bullosa (EB) is a group of inherited bullous disorders characterized by blistering of the skin and mucosa in response to trauma. EB patients require special precautions during dental treatment because of the high risk of lesioning the soft tissue when handling cutting instruments close to the skin and oral mucosa In this case report, clinical and oral findings of three siblings with epidermolysis bullosa, whose parents were relatives, are presented. The first case was a 5-year-old girl with skin lesions, microstomia, anklyoglossia and poor oral health. The second case was a 4-year-old girl having only deformity on her foot fingernail without any oral tissue involvement. The third case was a 2-year-old boy with numerous dermal blisters, ankyloglossia and poor dental status. Oral health of these children was maintained by improving oral hygiene, preventive measures and regular visits.

Keywords: Epidermolysis bullosa, oral health, oral manifestations

Öz

Epidermolizis bülloza, (EB) deri ve mukozada travmaya karşı cevap olarak meydana gelen bül oluşumuyla karakterize ailesel geçişli bir vezikülobüllöz hastalıktır. EB hastalarının dental tedavilerinde kullanılan aletlerin yumuşak doku lezyonu oluşturması riski yüksektir ve özel önlem gerektirmektedir. Bu vaka raporunda, ebeveynleri akraba olan, EB'lı üç kardeşin klinik ve oral bulguları sunuldu. Birinci vaka deri lezyonları, mikrostomi, ankiloglossi ve kötü ağız hijyenine sahip 5 yaşındaki bir kız çocuğu idi. İkinci vaka olan 4 yaşındaki kız çocuğunda sadece ayak tırnağında deformite mevcuttu. Üçüncü vaka, deride çok sayıda kabarcığı bulunan ankiloglossi ve kötü ağız sağlığına sahip 2 yaşındaki bir erkek çocuk idi. Bu çocukların ağız sağlığı, oral hijyenin sağlanması, koruyucu uygulamalar ve düzenli kontroller ile idame edildi.

Anahtar Kelimeler: Epidermolizis büllosa, Ağız sağlığı, Oral bulgular

Introduction

The term epidermolysis bullosa (EB) describes a heterogeneous group of inherited blistering mucocutaneous disorders which have a specific defect in the attachment mechanisms of the epithelial cells, either to each other or to the underlying connective tissue (1, 2). This dermatological condition is a severe autoimmune disease (3, 4).

The incidence of EB was reported varying between 1:50 000 to 1:500 000 live births (1). The most reliable figures on prevalence of EB are derived from the National Epidermolysis Bullosa Registry, which collected cross-sectional and longitudinal data on about 3300 patients in the United States from 1986 to 2002 (5, 6, 7,8). Over a 5-year period (1986 to 1990), the prevalence of EB was estimated to be approximately 8 per million live births (5). Data from the Australasian Epidermolysis Bullosa Registry provided a prevalence estimate of 1 case per 100 000 live

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Submitted / Gönderilme: 21.03.2018 **Accepted/Kabul:** 05.05.2018

births. Prevalence rates ranging from 15 to 32 cases per 100 000 have been estimated in the United Kingdom (6, 7) EB can be seen in all racial and ethnic groups and genders are affected equally (1, 9, 10).

EB can be inherited in either autosomal dominant or recessive fashion. Four major forms of EB have been described, depending on the level of skin involvement: EB simplex (EBS) involving the intra-epidermal layer; junctional EB (JEB) involving the lamina lucida; dystrophic EB (DEB) involving the sublamina densa; and Kindler syndrome involving multiple layers of the epidermis (11, 12, 13). However, more than 20 subtypes of EB have been recognized in the literature (1).

The etiopathogenesis of all forms of EB resides in mutations in more than 10 different genes coding for mutated proteins located at different levels in the epidermis/dermis (14). Indeed, mutations in transglutaminase 5, plakoglobin, plakiphilin 1 and demosplakin are responsible for the suprabasal form of EBS; mutations in keratins 5/14, plectin, BP230, kindlin-1, exophilin 5 for the basal form of EBS; mutations in integrins α6β4, integrin α3, collagen type XVII, and laminin-322 for the JEB and, mutations in collagen type VII for both dominant and recessive forms of DEB (12, 15).

EB is diagnosed by examining the basal membrane with transmission electron microscopy, immunohistochemical analysis, and other supplementary examinations such as immunofluorescence, optical microscopy, and enzymatic analysis. In some cases, fetoscopy for prenatal diagnosis of EB may be suggested (1, 2).

All major types of EB patients show skin and mucosa fragility in different levels clinically. Repeated episodes of cutaneous breakdown heal with scarring on hands and feet resulting in the fusion of the fingers into a mittenlike deformity in DEB patients (7).In addition, patients with EB have some oral alterations such as dental anomalies of shape, position, and structure (hypoplasia and hypomineralization); tongue denudation and limited mobility, ankyloglossia, and microstomia (11, 16,17, 18, 19, 20, 21).

Dental treatment is aimed at avoiding the formation of new bullae during treatment procedures under local anesthesia. Therefore, dental management of DEB patients requires lubrication of mucosal tissues and dental instruments in order to avoid adherence (22). The suction tip is advised to lean on hard dental tissues to avoid further epithelial sloughing (22). Special dental management involve the use of soft toothbrush and non-irritant irrigation techniques. Soft diets are also recommended because of the lesions involving the oral mucosa and gastrointestinal tract (23).

The aim of this case report was to present clinical and oral findings of three siblings with EB, whose parents had consangionus marriage.

Case Reports

Our first case was a 5-year-old girl with DEB, who was born with the skin lesions on the legs and hospitalized immediately (Figure 1). She had skin lesions on the ears, scalp, knees, elbows and fingers, and lesions on oral mucosa with poor oral hygiene. Intraoral examination was conducted with difficulty due to microstomia and limited opening. The patient had anklyoglossia and a smooth tongue with obliteration of the oral and lingual vestibules. Her dental status was poor with multiple advanced dental caries in most of her teeth.

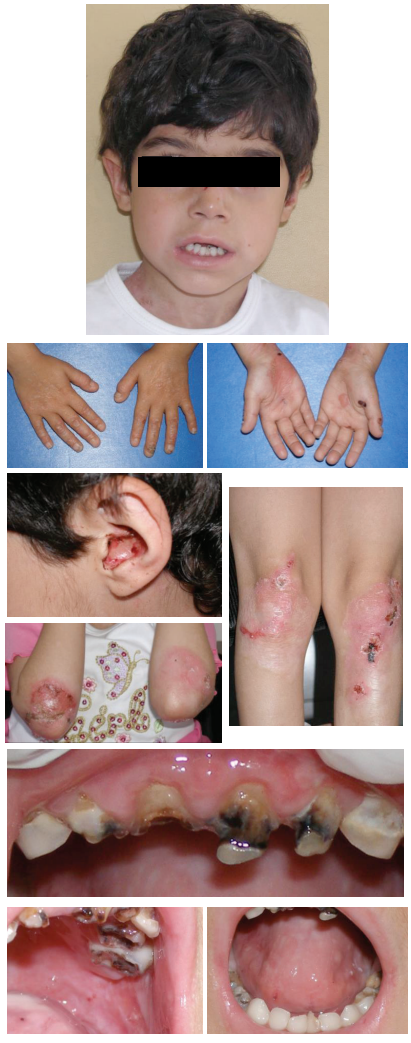

Figure 1. Case 1 showing skin and oral manifestations.

The second case was a 4-year-old girl with EBS (Figure 2). When she was born, she had blisters on her feet that healed within 20 days and thereafter she had not presented any lesions related with EB except deformity on her right foot fingernail. No involvement of oral mucosa was seen along with normal structured teeth.

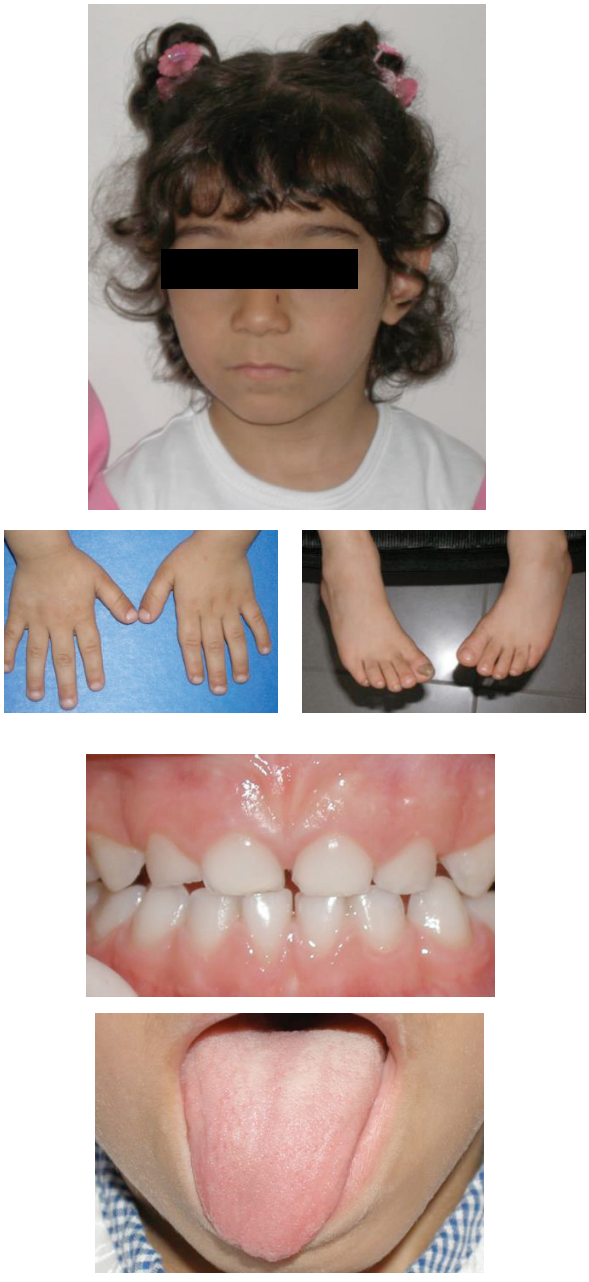

Figure 2. Case 2 with normal intraoral structures.presenting only a minor deformity on her right foot fingernail.

Our last case was a 2-year-old boy with DEB, who had oral mucosa lesions when he was born (Figure 3). His skin lesions appeared after he refused breastfeeding when he was 2 months old. He had numerous blistering on his face, ears, trunk, hand and feet with mittenlike deformity. Similar to the first case, the patient was ankyloglossia. His dental status was poor with caries in most of his teeth.

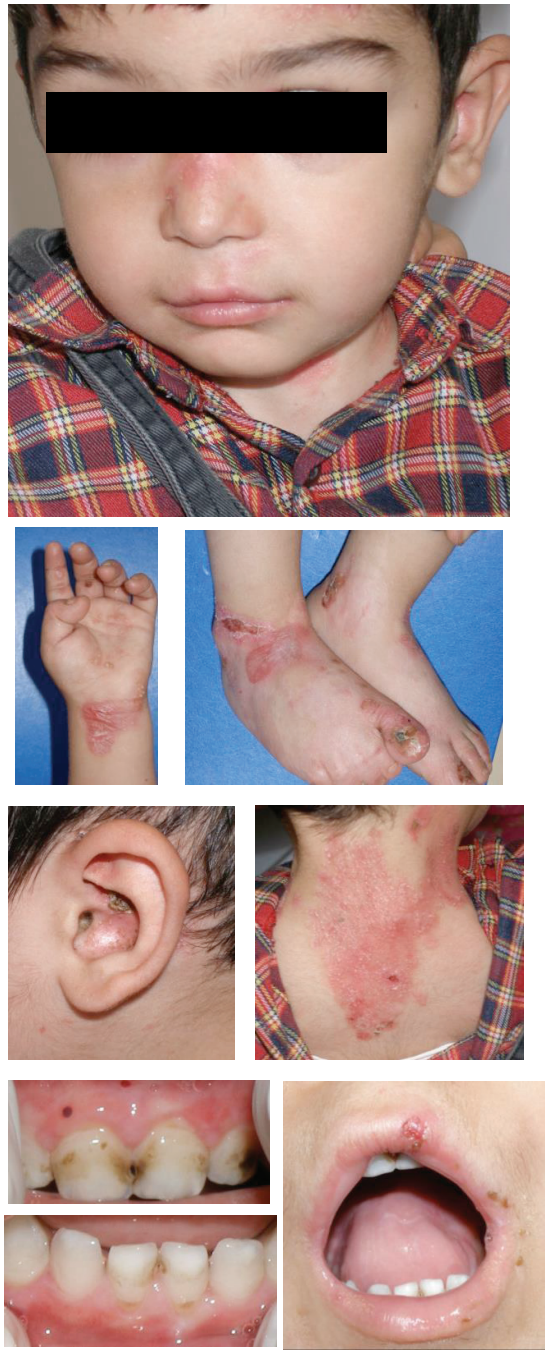

Figure 3. Case 3 showing lesions on skin and oral mucosa.

Cases were managed by bathing daily and drying with gentle movements to prevent trauma. An antibacterial ointment was used on the lesions and gauze wrapped around the affected areas. Oral hygiene instructions were given,

soft toothbrush was recommended, and dental therapy was performed under local anesthesia. The pedigree of the family is presented in Figure 4.

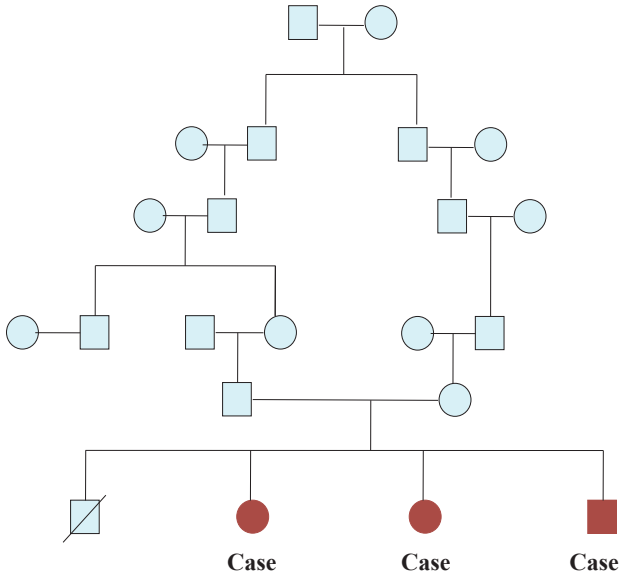

Figure 4. Pedigree of the family

Discussion

EB is a heterogeneous group of rare genetic disorders characterized by marked fragility of the skin and mucous membranes in which vesiculobullous lesions occur in response to trauma and other physical, termal and chemical causes (23). Due to the consanguineous marriage of parents, fetoscopy for prenatal diagnosis of EB could had been suggested to prevent the birth of other children with this disorder.

EB is a challenge to health professionals because there is no definitive cure (13, 17, 24) . Although all major types of EB has skin and mucosal lesions, DEB is the most aggresive form of EB (11, 25). The first and third cases were diagnosed DEB and they had oral manifestations such as mucosal erosions, reduced vestibular sulcus depth, ankyloglossia, microstomia and caries. But the second case, which was diagnosed as EBS, had only skin lesions on her feet and no involvement of oral mucosa. These dermal and oral findings of our cases are consisted with reported literatures (12,13,15,16).

EB patients require special precautions during dental treatment because of the high risk of lesioning the soft tissue when handling cutting instruments close to the skin and oral mucosa (16, 18). Cariogenic food, limited mouth opening as a result of persistent wounds and poor oral hygiene caused by pain are the predisposing factors to dental caries (19, 20). In these cases, minimal intervention has so far preserved the oral cavity and monthly topic fluoride application helped to control dental caries (26).

The patient maintains continuous contact with the health team to avoid complex treatments. Numerous alternative therapies have being used for the treatment of blisters. The application of aloe vera gel diminishes the subdermal temperature by providing a refreshed sensation, reducing the healing period and promoting the antimicrobial activity (27, 28). This product possesses buffering capacity, immunological effect, and a self-cleaning effect (28). EB treatment is generally focused on support rather than perforating the blisters since this procedure contributes to acceleration of the healing process and lateral spreading of the blisters. Currently, researchers are focusing their attention on gene and cell therapy, recombinant protein infusions, intradermal injections of allogenic fibroblasts and stem cell transplantation (29). Other developing therapies are directed toward the enhancement of wound healing and better quality of life for EB patients (2, 9, 22, 26, 29).

A multidisciplinary approach involving the nutritionist, pediatrician, dermatologist, plastic surgeon, hematologist, gastroenterologist, ophthalmologist, cardiologist, pediatric dentist, nurse and occupational therapist is essential (9, 10, 12, 29).

Conclusion

This case report emphasizes that patients with EBS need special precautions during dental treatment because of probable blister formation. Moreover, these patients require an early multidisciplinary approach to improve their quality of life, with the dentist playing an important role in oral health management.

Acknowledgement

This study was supported by the grant SAG-D-250608-0165.

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