Clinical and Experimental Health Sciences

Case Report / *Olgu Sunumu*

Extra- and Intraoral Clinical Findings of Recessive Dystrophic Epidermolysis Bullosa: A Case Report

Resesif Distrofik Tip Epidermolizis Büllozada Ağız Dışı ve İçi Klinik Bulgular: Vaka Raporu

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Abstract

Epidermolysis bullosa (EB) is a heterogeneous group of inherited blistering mucocutaneous disorders that have a specific defect in the attachment mechanisms of the epithelial cells, either to each other or to the underlying connective tissue. Recessive dystrophic type EB (Hallopeau–Siemens syndrome) represents repeated episodes of cutaneous breakdown, which heal with scarring, resulting in the fusion of the fingers into a mitten-like deformity. The vesicle/bulla can be intraorally observed and the depth of the buccal vestibule may decrease because of scarring. In this case report, the intra-/extraoral findings of a 25-year-old male patient, who was diagnosed with recessive dystrophic type EB immediately after birth, are presented. Extraoral examination revealed mitten-like hands and feet because of scar tissue formations on the hands and feet. Similarly, during intraoral examination, scar tissue formations were observed that had caused denuded mouth opening and limited tongue functions. In addition, periodontal problems and dental caries were observed because of limited plaque control. This case report emphasizes the importance of knowledge regarding patients with EB and presents some intraoral findings.
Keywords: Oral manifestations, Hallopeau–Siemens disease, epidermolysis bullosa dystrophica, case report

Öz

Epidermolizis bülloza (EB), epitel hücrelerinin kendi arasında veya altındaki bağ dokusuyla tutunmasını sağlayan mekanizmalardaki bozukluk sonucu meydana gelen, klinik olarak vezikül/bül oluşumuyla karakterize, kalıtımsal mukokutanöz hastalık grubudur. Resesif distrofik tip EB (*Hallopeau-Siemens* sendromu), kutanöz dokuda tekrar eden doku yıkım süreçleri ve skar dokusuyla iyileşmeye bağlı olarak parmaklarda birleşme, eldiven tipi parmaklar, tokmak el/ayak bulgularının gözlenebildiği bir EB alt grubudur. EB hastalarında ağız içinde de görülebilen vezikül/bül oluşumları, bukkal vestibül alan darlığı, ağız açıklığında kısıtlanma, ağız içinde skar dokularının oluşmasına sebep olabilir. Bu vaka raporunda, doğumundan hemen sonra resesif tip EB tanısı konan 25 yaşındaki hastanın ağız içi ve ağız dışı klinik bulguları sunulmuştur. Yapılan ağız dışı muayene sonucunda el ve ayaklarda skar dokusu oluşumlarına bağlı tokmak el ve ayak görüntüleri izlendi. Benzer şekilde, ağız içinde meydana gelen skar dokularının ağız açıklığında ve dil fonksiyonunda sınırlama meydana getirdiği gözlendi. Sınırlı plak kontrolüne bağlı olarak periodontal problemler ve diş çürükleri saptandı. Bu vaka raporu, EB hastalarının ağız içi bulguları hakkında bilgi sahibi olunmasının önemini vurgulamaktadır.
Anahtar kelimeler: Ağız içi bulgular, Hallopeau-Siemens hastalığı, distrofik tip epidermolizis bülloza, vaka raporu

INTRODUCTION

Epidermolysis bullosa (EB) is a rarely observed genetic disorder that causes noticeable vesiculobullous lesions on the skin and mucous membranes and that occurs because of mechanical trauma, heat, or no clearly distinct cause (1). The incidence of EB is between 1/50.000 and 1/500.000 live births worldwide (2). EB can affect any person regardless of racial or ethnic group or gender and can occur at birth or during the first year of life (2, 3).

EB can be categorized into three main conditions depending on the place of blistering in the dermal–epidermal junction, such as simplex, junctional, and dystrophic (4, 5); more than 20 subtypes have been reported considering the severity and mode of inheritance (1). Alterations may occur in the epidermal basal layer (EB simplex), lamina lucida (junctional type EB), or lamina densa (dystrophic type EB).

Mutations in the type VII collagen gene cause dystrophic type EB, which can be inherited in both dominant and recessive forms (3). As a main component, collagen VII is a fibrous protein of the anchoring fibrils (1, 2, 6). Anchoring fibrils are considered to form a structural link between the basement membrane of epidermis and the fibrillar collagens in the dermis. The insufficiency of anchoring fibrils results in structural inadherence between the epidermis and underlying dermis layer.

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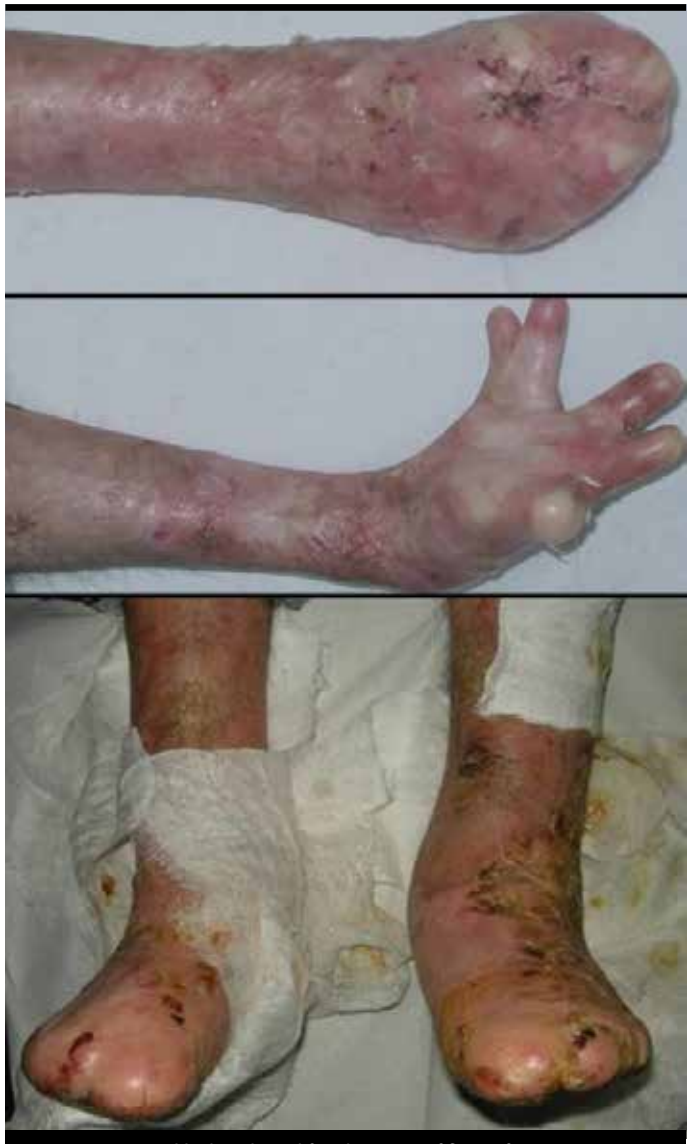

Figure 1. Mitten-like hands and feet because of finger fusions

Figure 2. Intraoral clinical and radiographical findings of patient

Figure 3. Limitation of mouth opening because of scar formations on labial commissura

Recessive dystrophic type EB (Hallopeau–Siemens syndrome) is a subtype of EB with an autosomal recessive inheritance pattern (7). Because of recessive dystrophic type EB, the skin becomes fragile, and blistering and severe skin ulceration forms, even with minimal mechanical trauma. Repeated episodes of cutaneous breakdown, which then heal with scarring, can result in fusion of the fingers and in a mitten-like deformity (2). Flexural contractures occur in the knee, elbow, wrist, and ankle joints (1). The development of vesicle or bulla occurs on the entire gastrointestinal tract and oral cavity, making eating and defecating very painful (7, 8).

The recessive dystrophic form of EB presents intraoral alterations with the formation of a vesicle that rapidly tears and that afterwards leaves erosive surfaces on the mucosa of cheeks, tongue, uvula, lips, and rest of the oral cavity. Reduced or no function of the tongue, ankyloglossia, microstomia, and reduced vestibule sulcus depth can be observed as a result of the continuous healing processes of this disease (1, 9). Dental anomalies of the form, position, and structure-like hypoplasia and hypomineralization may present, too (10). Because of these oral manifestations and improper oral hygiene, these patients are more prone to high caries incidence and gingival disease severity.

This case report aimed to show the intra-/extraoral clinical and radiographical findings of a patient with recessive dystrophic type EB.

CASE REPORT

In this case report, the intra-/extraoral findings of a 25-year-old male patient, who was diagnosed as recessive dystrophic type EB immediately after birth, are presented. Written informed consent, that allows the sharing of clinical findings and images of patient for scientific purposes, was signed by the patient Parents of the patient had a consanguineous marriage. Fusions of the hand fingers had started at 4 years old and he had undergone four operations. Repeated scar tissue formations on the hands and feet had caused mitten-like hands and feet (Figure 1).

Intraoral clinical examination revealed improper oral hygiene level, supra- and subgingival calculus, and plaque existence. Clinical periodontal parameters were measured as follows: mean plaque index, 2.08±0.45; gingival index, 2.53±0.23; and probing depth, 3.46±1.02. Moreover, severe dental hard tissue loss because of caries, intraoral scar tissue formations, hopeless teeth, and remnants roots were observed on the intraoral and radiographical examination (Figure 2).

Because of intraoral scar tissue formations, the tongue functions were restricted and the buccal vestibule depth was shallowed (Figure 2). Similarly, the scar tissues on the corner of the lips limited the mouth opening, as can be seen in Figure 3. The patient was under the control of a dermatologist and there was no internal organ involvement. The patient was using only topical medications containing corticosteroid (0.05% Klobetazole 17-propiyonate, 2*1, Dermovate®; GlaxoSmithKline, London, UK) and dexpanthenol (50 mg/g D-panthenol, 3*1, Pantenol Pomad®; Saba, İstanbul, Turkey) to improve epidermal wound healing and relief pain.

Dental treatment was planned as a minimal invasive multidisciplinary approach to gain periodontal and dental health with functional dentition. In the treatment plan, involving the extraction of hopeless teeth, initial periodontal treatment consisted of oral hygiene instructions, scaling, and root planning; restorative prosthodontic treatments were planned but the patient discontinued the treatment (11). Unfortunately, follow-up of the patient could not be performed.

DISCUSSION

EB is a group of inherited bullous disorders outlined by blistering of the skin and mucosa in response to trauma (7). Dystrophic EB has either an autosomal- dominant or recessive pattern of inheritance and is associated with the loss of fibrils of anchorage and increased collagen disintegration on the superficial dermis due to excessive synthesis of collagenase (1, 2). In our case, the patient, whose parents had a consanguineous marriage, was diagnosed with a recessive pattern of dystrophic type EB immediately after birth.

Scar tissue healing on the fingers had caused finger fusions of the hands and feet and mitten-like hands. Due to intraoral/perioral scar tissue formations because of the torn bulla, the patient had sequels like tongue denudation, limitation of the mouth opening, a reduction of buccal vestibule depth, and phonation problems. Restricted functions of the hand and mouth opening made plaque control difficult. Due to this, periodontal problems and dental caries were observed. Moreover, the patient was psychologically compromised because of the aesthetic and functional problems. Internal organ involvement, like with the gastrointestinal tract, may be observed in recessive dystrophic type EB (8), but in our case there was no internal organ involvement.

CONCLUSION

Dentists must be aware of intra- and extraoral manifestations of patients with EB and work co-operatively with dermatologists to manage patients with EB.

Informed Consent: Written informed consent was obtained from patient who participated in this study.

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