A Rare Case: Epidermolysis Bullosa in a Child Patient with Amelogenesis Imperfecta

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ABSTRACT

Epidermolysis bullosa (EB) is an inherited disorder affecting the skin and mucous membranes, characterized by blister formation following minor trauma. It is a chronic mechanobullous disease related to the specific abnormal or absent proteins. The disease is associated with conspicuous clinical and oral manifestations. The oral involvement of EB includes generalized enamel hypoplasia, dental caries, limited mouth opening, ankyloglossia, microstomia and obliteration of the vestibule.

Amelogenesis imperfecta (AI) is a hereditary disorder with dental enamel defects and enamel hypoplasia both in deciduous and permanent dentition.

There is very limited information in the literature, which indicate the presence of EB together with AI.

The aim of this report is to present the clinical and radiographic manifestations and dental management of EB simplex in a child patient with hypoplastic form of AI.

Keywords: Amelogenesis imperfect, epidermolysis bullosa, hereditary disorder

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INTRODUCTION

Epidermolysis bullosa (EB) is an inherited disorder affecting the skin and mucous membranes, characterized by blister formation following minor trauma (1). It is a chronic mechanobullous disease related to the specific abnormal or absent proteins (2,3). EB was first described by Von Hebra in 1870 and the estimated frequency is 1 of 50,000 births (4,5). Approximately 400,000-500,000 people are affected worldwide and there is no specific treatment available (6).

Diagnosis is based on clinical symptoms and skin biopsy. Individuals with EB are classified based on the ultrastructural level within which blisters occur (7). In the report of the "Third International Consensus Meeting on Diagnosis and Classification of EB (2008)", it was reclassified into four major types; simplex, dystrophic, junctional and the mixed type (or called Kindler Syndrome) (8). The character and extend of oral involvement varies depending on EB types (9). EB simplex has the best prognosis and oral soft tissue lesions typically heal without scarring (2, 10). Gingival blisters or erosion, enamel hypoplasia, dental caries, malocclusion and premature loss of teeth are the most common dental manifestations in patients with Dystrophic form of EB and it is divided into two main groups as recessive and dominant (8,9,11). Especially, generalized form of recessive dystrophic EB demonstrates the most severe oral involvement characterized by obliteration of the vestibule and ankyloglossia (9). Individuals with junctional EB have an increased fragility of the oral mucosa that is accompanied by blister formation and ulceration mostly heals without scarring (2).

Both dentitions could be affected by enamel hypoplasia and/or dental caries depending on the EB type (9). Furthermore, in junctional type of EB, individuals are at increased risk for developing dental caries and enamel lesions that differ from generalized pitting to generalized hypoplasia (2, 12). However, the patients with EB simplex have the same

currency of dental caries as the unaffected population due to limited intraoral soft tissue morbidity and the standard shaping of tooth (2).

Amelogenesis imperfecta (AI) is a hereditary disorder with dental enamel defects and enamel hypoplasia both in deciduous and permanent dentition. Classification of specific AI types primarily is based on genetic inheritance and clinical appearance. AI can be inherited as autosomal dominant, autosomal recessive, or X-linked (13). According to Witkop classification system, which is based on clinical appearance (phenotype), AI is categorized into four major types: hypoplastic, hypomatured, hypocalcified, and hypomatured-hypoplastic enamel with taurodontism (14).

Occurance of EB along with AI, although rare, has been reported in the literature, however there is not enough information about clinical findings and management of this combine situation.

The aim of this report is to present the clinical and radiographic manifestations and dental management of EB in a child patient with hypoplastic form of AI.

CASE REPORT

A 12-year old male patient diagnosed with EB simplex referred to the Akdeniz University, Faculty of Dentistry, Department of Pediatric Dentistry with the complaint of pain caused by dental caries and discolored anterior teeth. In accordance with EB, his medical history revealed the presence of multiple dermal lesions, which started to appear immediately after birth. No significant family history or consanguinity was reported. His growth and development were compatible with the ideal measurements of a 12 year-old child. He had normal cognitive function and regular school attendance. No allergies were reported.

As shown in Figure 1, during the general body inspection, nail dystrophy involving only toenails and blister formation healed without scarring were seen. According to information received from parents, warm and humid weather increased the frequency of occurrence of lesions.

Intraoral examination revealed poor oral hygiene, generalized gingival inflammation with calculus deposition due to abundant plaque accumulation, variable size and shape of crowns, lack of contact between adjacent teeth, enamel hypoplasia and dental caries. The oral mucosa appeared normal and no oral blisters were observed. He had a mixed dentition with dental carious lesions in 26, 46 and 36 with a combined periodontal-endodontic lesion. As shown in Figure 2, all teeth showed a yellow discoloration with the attrition of the incisal and occlusal surfaces. There was evidence of thin enamel layer with normal structure interrupted with rough areas and irregularly shaped empty spaces.

Panoramic radiograph revealed a normal sized and shaped pulp chamber and root canal spaces. The enamel was completely lost and radiopaque dentin is acknowledged (Figure 3). As a result of clinical and radiologic evaluations, we arrived at diagnosis of hypoplastic type of AI.

The treatment started with scaling, root planning and endodontic therapy of the left mandibular first molar, which was diagnosed as combined periodontal-endodontic lesion. Direct composite restorations of the mandibular and maxillary anterior teeth were performed. Furthermore, stainless steel crown was placed on the right mandibular first molar and it was planned to place stainless steel crown on the other molars in the future appointments (Figure 4a, 4b, 4c). The patient was followed up every 3 months for one year. In this period, new dental caries was occurred and new restorations were performed on each appointment (Figure 5). Besides restorations, methods that prevent dental caries such as fluoride varnish (Duraphat, Colgate Palmolive, New York, NY, USA) were applied. The patient has been

informed for the improvement of the oral hygiene and he was advised using a %0.05 sodium fluoride mouthwash. The patient is under regular control and his dental treatments are in progress in our clinic.

DISCUSSION

EB is a group of rare genetic disorders that involve the blister formation following mild trauma (4). The disease is associated with conspicuous clinical and oral manifestations, which include oral tissue fragility and blistering of the skin and mucosa (4, 9, 15). Blisters that occur in the oral mucosa heal with scar formation usually start to appear at birth or during the course of the first year of life. However in the present case, there was no intraoral blister or scar formation.

A recent study, including 252 patients with EB, reported that the prevalence of caries, scored as DMFS, was significantly higher in junctional and recessive dystrophic EB than among healthy people and simplex type. Additionally in this study, developmental enamel defects were seen more in junctional EB than the other EB types which have a prevalence of enamel defects similar to unaffected individuals. According to this study, it appears to be the oral blistering and enamel hypoplasia are the primary determinants leading these patients to dental involvement and high risk of caries formation (12). However, in our case, there is no clear link between the intensity of oral blistering and caries formation and the cause of dental caries is possibly related to poor oral hygiene and irregular tooth surfaces.

Kirkham, et al. (2000) indicated that patients with junctional EB had developmentally enamel defects such as enamel hypoplasia due to reduced mineral per volume content (16). Even though generalized enamel hypoplasia is accepted as a feature of junctional EB, Çağırankaya, Hatipoğlu and Katipoğlu (2006) reported a 17-year-old boy with EB simplex

associated with enamel hypoplasia (17). Similarly we presented a patient with EB simplex associated with enamel hypoplasia in the form of AI.

Based on the phenotype, AI is divided into four major types: hypoplastic, hypomatured, hypocalcified, and hypomatured-hypoplastic enamel with taurodontism (14). Hypoplastic type of AI is characterized by a defect in the formation of the enamel matrix causing thin enamel with or without pitting and also square-shaped crown and lack of contact between adjacent teeth. In the hypocalcified type, the enamel is poorly mineralized, soft, and can be easily abraded (18). Hypomaturation type is related to enamel maturation and the enamel appears mottled, opaque white to red-brown coloration, and is softer than normal (19). On the basis of clinical and radiological examination the patient was diagnosed with hypoplastic type of AI.

There are many determinants, which affect the planning of treatment in AI patients such as the patient's age and socioeconomic condition, disorder's type and intensity and intraoral status during the treatment is scheduled (20). Total coverage of all teeth with fixed partial dentures was diagnosed as the best approved treatment, but the patient's unaccomplished skeletal development during the diagnosis ruled out this as a possible instant choice of treatment.

An increased risk of caries is the main dental problem for patients with EB. Prevention of dental caries and periodontal disease must be emphasized at an early age, because of severe limitations and complications of providing dental treatment to patients with EB (6). As a recommendation, continuous oral hygiene must be provided and dental appointments must be frequently made. Oral hygiene procedures must be modified for patients with EB, in order to prevent the formation of new oral blisters. The constant use of an alcohol free fluoride mouthwash and a manual toothbrush with small head size and extra soft filaments should be recommended (5). Topical fluoride application should be performed during the

deciduous and permanent dentition.

Management of patients with EB requires a multidisciplinary approach, encompassing neonatologist, genetic counselor, pediatrician, dermatologist, physiotherapist, plastic surgeon, gastroenterologist, dietitian, psychologist, special nurse and dentist. Pediatric dentists play an important role to improve the quality of life of patients with EB in functional and esthetical aspects. In these patients, preventing and controlling tooth decay are the main factors to provide a healthy dentition. Preserving the dentition not only decreases the possibility of soft tissue trauma but also improves the nutrition, which is necessary for healthy growth and development. A rare case of EB simplex associated with AI that required extensive dental treatment and long-term follow-up is presented.

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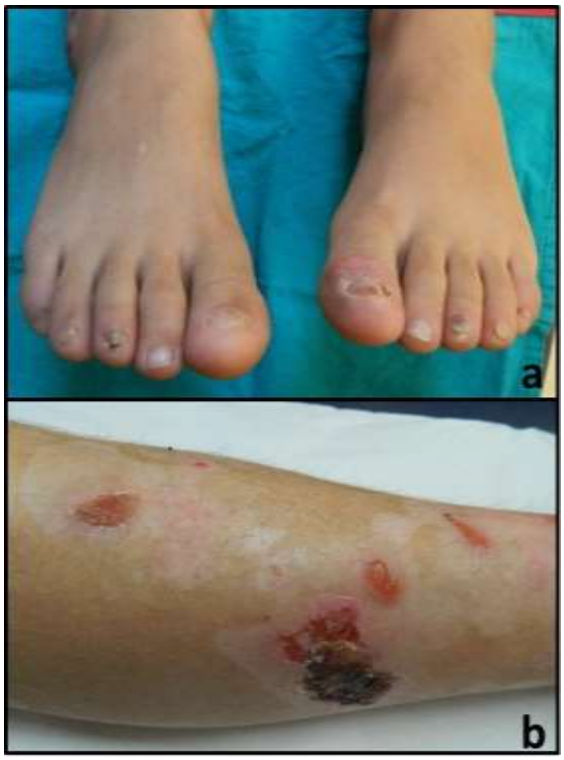

Fig 1: During the general body inspection, (a) nail dystrophy involving only toenails and (b) blister formation healed without scarring were seen

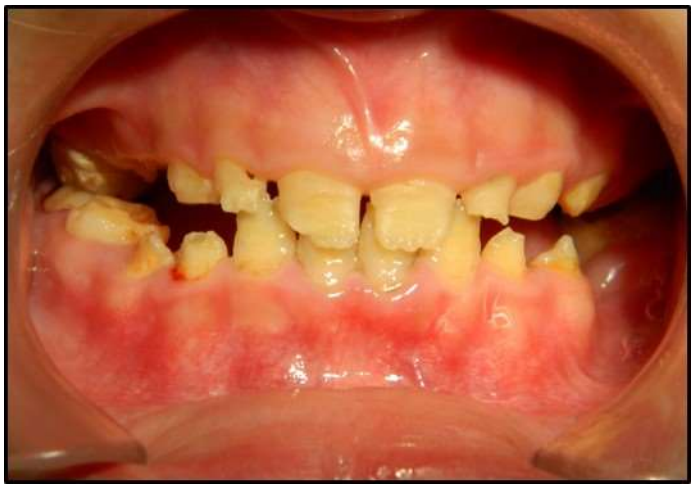

Fig 2: All teeth showed a yellow discoloration with the attrition of the incisal and occlusal surfaces

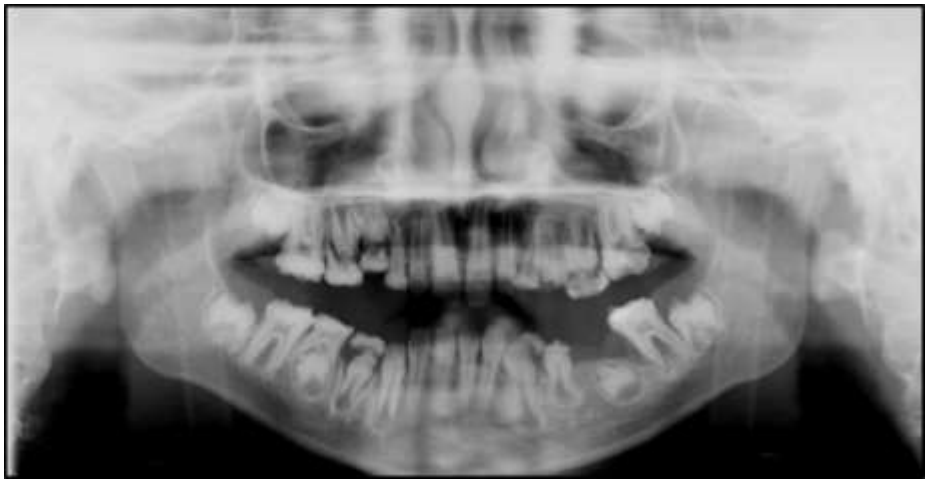

Fig 3: Panoramic radiograph revealed a normal sized and shaped pulp chamber and root canal spaces. The enamel was completely lost and radiopaque dentin is acknowledged

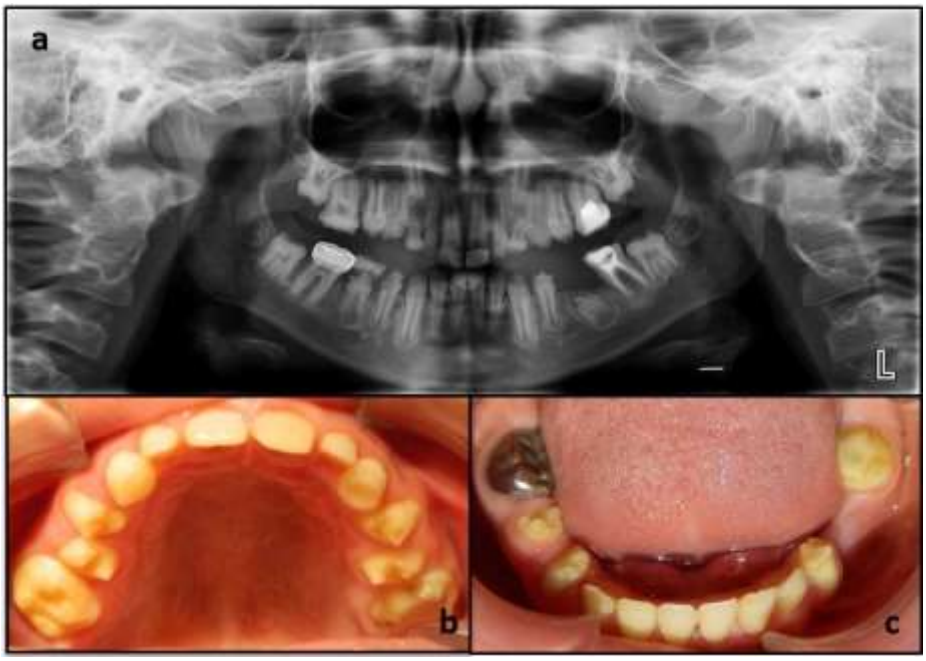

Fig 4: (a) Endodontic therapy of the left mandibular first molar and (b) direct composite restorations of the mandibular and maxillary anterior teeth were performed. (c) Stainless steel crown was placed on the right mandibular first molar

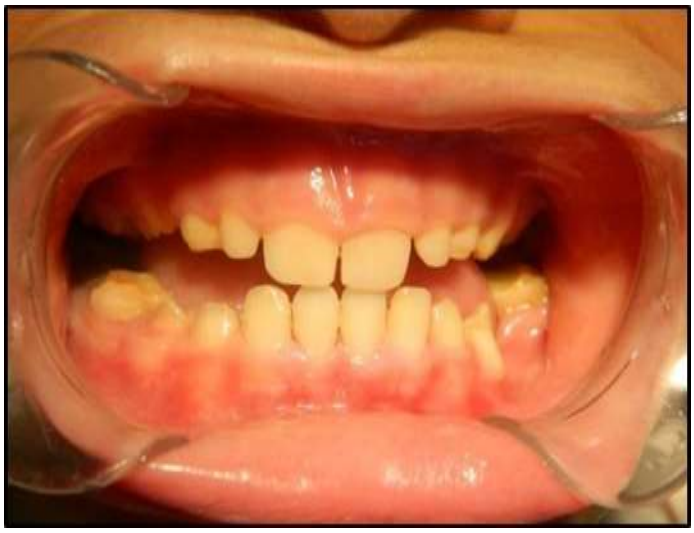

Fig 5: The patient was followed up every 3 months for one year. In this period, new dental caries was occurred and new restorations were performed on each appointment