International Journal of Paediatric Dentistry 2005; **15:** 146–150

Junctional epidermolysis bullosa: a case report

A. MOMENI & K. PIEPER
Department of Paediatric and Community Dentistry, Philipps-University Marburg, Marburg, Germany

Summary. Epidermolysis bullosa (EB) is a group of genetically determined disorders characterized by blistering of the skin and mucosae. There are three major forms – simple, junctional and dystrophic – and each has several varieties. The present case report describes a male child with junctional EB. The aim of the report is to present the dietary situation and the dental status of the child, examples of potential dental and nutritional consequences, and the therapeutic interventions possible for children with this disease.

Introduction

Epidermolysis bullosa (EB) describes a group of hereditary mechanobullous disorders which are all characterized by blistering of tissues containing stratified epithelia [1,2]. The disorder occurs in every racial and ethnic group throughout the world and affects both sexes equally. Köbner coined the term 'epidermolysis bullosa' in 1886, but even before this time, Legg and Brocq had already provided a clinical description of the disease [3]. The disease often becomes manifest at birth or during the first year of life. Epidermolysis bullosa has been classified into three major types depending upon the histological level of tissue separation [4]: (1) epidermolysis bullosa simplex is characterized by discontinuities in the epithelial keratinocyte layer; (2) junctional epidermolysis bullosa involves separation within the basement membrane; and (3) dystrophic epidermolysis bullosa is characterized by discontinuities in the underlying connective tissue. Each type of EB has various subtypes and these may vary in severity [5]. Skin biopsies are needed for appropriate diagnosis and classification for affected subjects.

The incidence of recessive EB is approximately one in 300 000 births, and for dominant EB, this figure is one in 50 000 [6]. Epidermolysis bullosa may also cause bullae to form on inner organs, such as the oesophagus. Depending on the type, its efflorescences may or may not leave a scar on healing. As well as the formation of bullae, there have been descriptions of nail and hair dystrophy, and alopecia in affected individuals. There have also been reports of enamel hypoplasia [7,8].

Correspondence: A. Momeni, Philipps-University Marburg, Department of Paediatric and Community Dentistry, Georg-Voigt-Str. 3, 35033 Marburg, Germany. E-mail: momeni@mailer.uni-marburg.de

Case report

Clinical examination

The patient attended the present authors' centre for the first time at 5 years of age in February, 2003. Prior to this, his private dentist had recommended treatment involving the use of general anaesthesia and hospital admission since outpatient treatment was considered inadvisable. The patient was referred to the hospital because of the risk entailed by treatment involving general anaesthesia. At the time of presentation, the patient was cooperative, and thus, easy to examine. He had suffered from EB junctionalis atrophicans mitis since birth (i.e. generalized atrophic benign epidermolysis bullosa, GABEB, a mutation in the COL 17 A gene). The patient had two sisters, aged 4 and 1·5 years, who were unaffected by the disease. His parents were also unaffected and were not consanguineous. Both sets of grandparents came from the same small village in Sri Lanka. This type of illness had not previously appeared in the family.

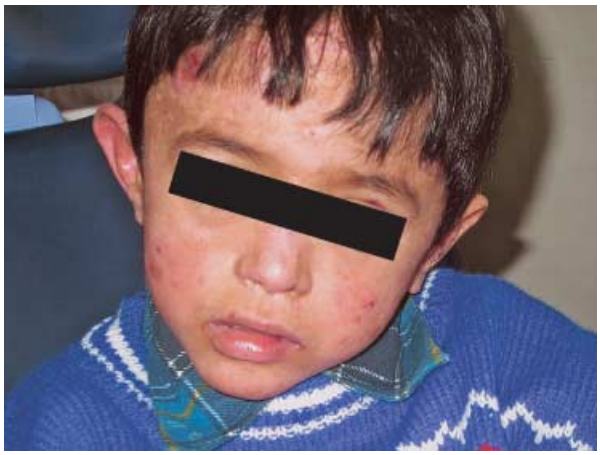

Fig. 1. Facial photograph of the patient at 5 years of age.

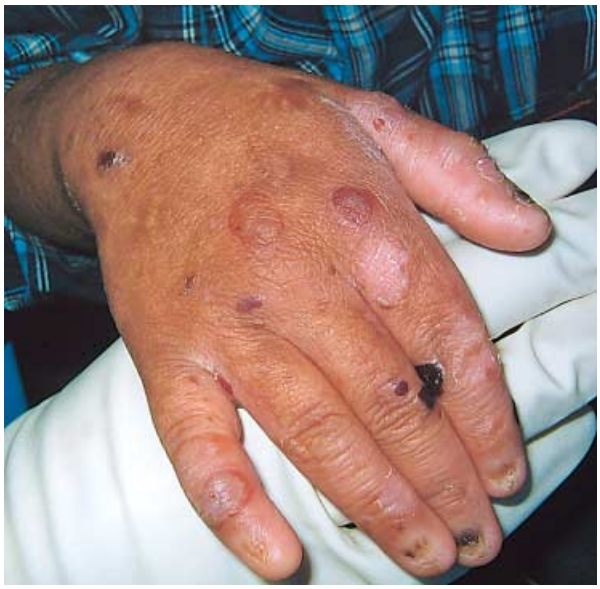

Fig. 2. View of the right hand showing absence of nails, scarring and bullae.

On examination, the patient's skin was covered with multiple pigmented scars. In addition, there were numerous blisters involving his head (Fig. 1) and hands, most of them filled with blood. He had no nails on his fingers (Fig. 2) and there were bald spots on his head (Fig. 3).

The results of the medical examination were unremarkable. His parents reported that blisters appeared on the patient's skin at the slightest touch or defensive movement. These could be large or small, sometimes filled with blood, and depending on their size and location, they could leave a scar on healing. Bullae also affected mucous membranes in his

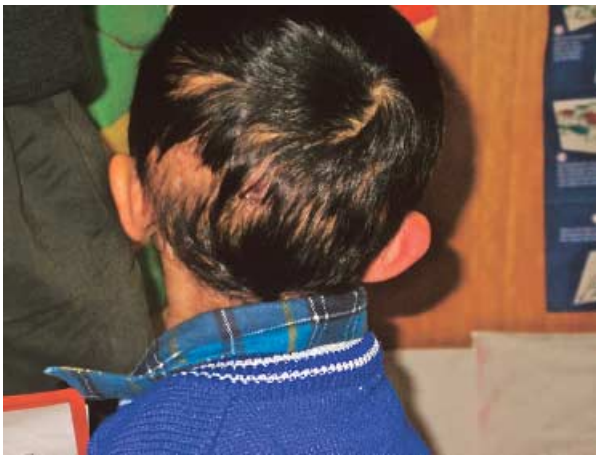

Fig. 3. Back of the head showing areas of alopecia.

mouth and his anal region, which caused considerable difficulty when eating or defecating. In addition, the patient had trouble carrying out oral hygiene measures, which may have contributed to the development of caries.

There were no bullae on the oral mucosa at the time of the first examination. All primary teeth were present on investigation. All the primary molars (Fig. 4), as well as teeth 53, 52, 62 and 63, exhibited atypical lesions on the occlusal surfaces and incisal edges, combining characteristics of caries and erosion. Active caries lesions were present in primary upper central incisors (51 and 61) (Fig. 5). Caries were attributed to special dietary habits and lack of oral hygiene measures.

Dietary anamnesis/oral hygiene

The patient had been wholly breast-fed for a short time, and was then bottle-fed until he was 12 months of age. Because drinking out of a bottle caused him to develop blisters on his tongue, he began to drink out of a cup at a very early age. Overall, the patient's food intake was severely inhibited because of the blisters forming in his mouth during his first years of life. Between his first and fourth year of life, his general condition was so poor at times that he had had to be tube-fed by means of a straw. By the end of his second year, the patient was being given milk gruel or baby food. Since he frequently had extreme difficulty ingesting food, he was often only able to eat ice cream. In addition, the patient was fed yoghurt, either with or without fruit flavourings. At present, he enjoys eating soups, and drinks water, milk and

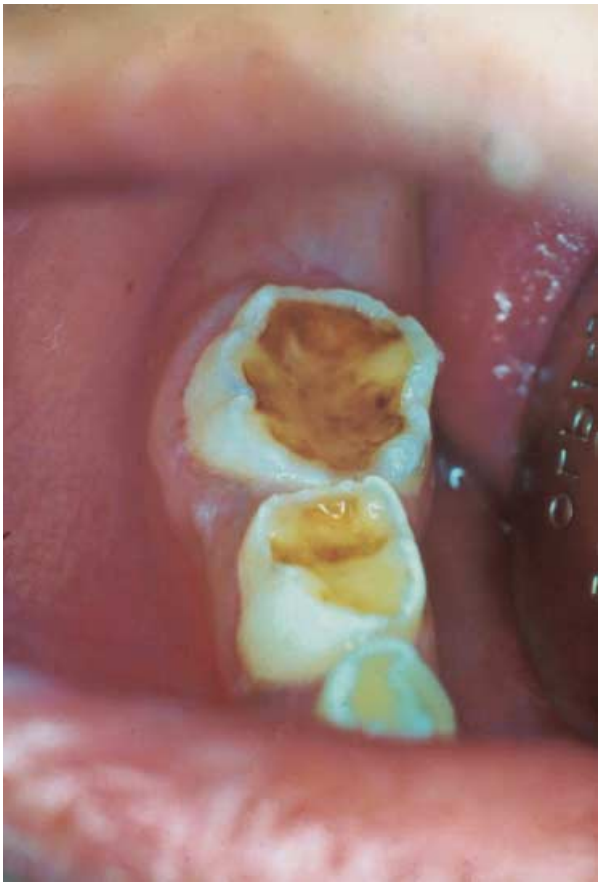

Fig. 4. Lower right primary molars (84 and 85) showing defects of occlusal surfaces.

fruit juices. In addition, he is occasionally given iron preparations.

The patient's parents first attempted to clean his teeth when he was between 1·5 and 2 years of age, which proved to be extremely difficult at first. Only when he was approximately 4 years old did it become considerably easier to brush his teeth. At present, the patient cleans his own teeth twice a day, in the morning and evening. He was given fluoride tablets for 3 years.

Management

After exploration of the patient's medical history and feeding practices, the present authors decided to focus on restorative and preventive treatment. This treatment included the restoration of teeth 51 and 61, instruction in proper tooth cleaning and oral hygiene, professional prophylaxis, and topical application of a fluoride varnish at recall every 3 months.

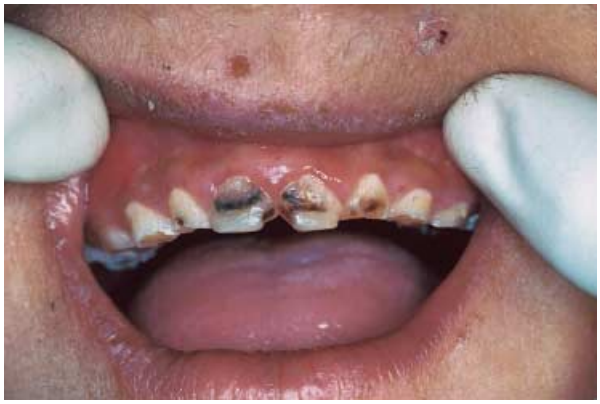

Fig. 5. Anterior view of upper arch showing defects of labial, mesial and occlusal surfaces.

Check-ups performed so far have shown an unmistakable improvement in oral hygiene. Since the patient's parents are making every effort, and thanks to regular reinforcement at the frequent recall visits, his chances of retaining his primary teeth are considered to be generally good.

Discussion

Dental management of individuals with EB has been reported previously by several authors [9–20].

Examinations have shown that the caries prevalence among individuals with dystrophic EB and junctional EB is significantly higher than among healthy people [21]. Similar observations have been made with respect to the prevalence of plaque and gingivitis. However, the patients examined in these studies show no significantly higher prevalence of *Candida albicans*, *Lactobacillus casei* or *Streptococcus mutans* [22].

Wright *et al*. reported that none of their patients with EB showed any evidence of a decreased salivary flow rate [23]. Despite the severe cutaneous and extracutaneous involvement associated with inherited EB, the above authors found no evidence to support the hypothesis of abnormal salivary function or mucosal immunity in this disease. Taken together, these findings suggest that the rampant dental caries seen in the various forms of EB are more likely to be attributable to nonsalivary factors, such as enamel involvement, soft-tissue alterations and/or diet.

A study published in 2001 investigated how effective a medicine called Sucralfate was for patients with EB [24]. This is a complex salt of sucralfate and aluminium hydroxide. Sucralfate has been used

in other contexts, and for example, may reduce pain caused by oral mucositis during radiation therapy and recurrent aphtous stomatitis. The patients in the above study, who all suffered from EB, showed a great improvement both in spontaneous pain and in the reduction of the number of blisters. A 90% reduction of plaque and a 100% reduction of bleeding and gingival inflammation was also noted, which was probably a result of a lower level of mucosal pain, allowing the patients to perform good oral hygiene and to eat proper food.

Studies have repeatedly documented the occurrence of junctional EB involving enamel hypoplasia [see 7,8]. In these patients, enamel hypoplasia may also promote the development of caries.

Because caries was extensive at presentation, the present authors were unable to confirm clinically that their patient had enamel hypoplasia. When recording the previous history of the patient's eating habits, they found that this correlated very closely with the damage to his teeth. The pattern of disease was unusual, but the authors suspected that plaque had formed on the chewing surfaces as a result of a past history of insufficient oral hygiene or from eating soft foods containing sugar. Demineralization was found to have occurred beneath the plaque. Caries only stopped progressing when oral hygiene measures had improved sufficiently.

Conclusions

Early dental management and preventive care to minimize caries development and to improve oral health is very important for patients with EB. Furthermore, they are often advised to reduce their consumption of cariogenic foods. Topical fluoride applications at regular intervals are also indicated. Ongoing patient monitoring must be stressed to minimize the need for restorative treatment in patients with EB.

Acknowledgements

The authors would like to thank the German Society of Dental, Oral and Craniomandibular Sciences for their support.

Résumé. L'épidermolyse bulleuse (EB) est un groupe de pathologies d'origine génétique caractérisées par la formation de bulles au niveau cutané et muqueux. Il existe 3 formes majeures: simple, jonctionnelle et dystrophique, chacune ayant plusieurs variétés. Cet article présente le cas d'un garçon présentant une EB jonctionnelle, avec pour objectif d'exposer la situation alimentaire et l'état dentaire de l'enfant, des exemples de conséquences potentielles dentaires et nutritionnelles, ainsi que les interventions thérapeutiques possibles pour les enfants porteurs de cette maladie.

Zusammenfassung. Epidermolysis bullosa (EB) ist eine Gruppe von genetisch bedingten Krankheiten mit Blasenbildung von Haut und Mukosa. Es gibt drei Klassen: Einfache, junktionale und dystrophische, jede hat mehrere Unterformen. Dieser Fallbericht beschreibt ein Kind mit junktionaler EB. Ziel dieses Beitrags ist es, die Ernährungsbedingungen und den Zahnstatus zu zeigen ebenso wie Beispiele potentieller dentaler und ernährungsbezogener Konsequenzen und mögliche therapeutische Interventionen für Kinder mit diesem Krankheitsbild.

Resumen. La Epidermolisis ampollosa (EA) es un grupo de alteraciones determinadas genéticamente caracterizadas por ampollas de la piel y mucosas. Hay tres grandes formas: simple, de la unión y distrófica, cada una tiene sus variaciones. El informe de este caso describe un niño varón con EA de la unión: El objetivo del informe es presentar la situación de la dieta y el estado dental del niño, ejemplos de las consecuencias potenciales, tanto dentales como nutricionales y las intervenciones terapéuticas posibles para los niños con esta enfermedad.

References

1 Gedde-Dahl T. *Epidermolysis Bullosa: A Clinical, Genetic and Epidermological Study*. Baltimore, MD: Johns Hopkins Press, 1971.
2 Gedde-Dahl T, Anton-Lamprecht I. Epidermolysis bullosa. In: Emery AEH, Rimoin D (eds). *Principles and Practice in Medical Genetics*. Edinburgh: Churchill Livingstone, 1990: 855–876.
3 Leiber B, Olbrich G. Epidermolysis bullosa syndrome. In: Band I (ed.). *Die Klinischen Syndrome*, 6th edn. München: Urban & Schwarzenberg, 1981: 298–299.
4 Fine JD, Bauer EA, Briggaman RA, *et al.* Revised clinical and laboratory criteria for subtypes of inherited epidermolysis bullosa. A consensus report by the Subcommittee on Diagnosis and Classification of the National Epidermolysis Bullosa Registry. *Journal of the American Academy of Dermatology* 1991; **24**: 119–135.
5 Fine JD, McGrath J, Eady R. Inherited epidermolysis bullosa comes into the new millennium: a revised classification system based on current knowledge of pathogenic mechanisms and the clinical, laboratory, and epidemiologic findings of

large, well-defined patient cohorts. *Journal of the American Academy of Dermatology* 2000; **43**: 135–137.
6 Davidson BCC. Epidermolysis bullosa. *Journal of Medical Genetics* 1965; **2**: 233–242.
7 Wright JT, Johnson LB, Fine JD. Development defects of enamel in humans with hereditary epidermolysis bullosa. *Archives of Oral Biology* 1993; **38**: 945–955.
8 Kirkham J, Robinson C, Strafford SM, *et al*. The chemical composition of tooth enamel in junctional epidermolysis bullosa. *Archives of Oral Biology* 2000; **45**: 377–386.
9 Howden EF, Oldenburg TR. Epidermolysis bullosa dystrophica: report of two cases. *Journal of the American Dental Association* 1972; **85**: 1113–1118.
10 Endruschat AJ, Keenen DA. Anesthetic and dental management of a child with epidermolysis bullosa dystrophica. *Oral Surgery, Oral Medicine and Oral Pathology* 1973; **36**: 667–671.
11 Crawford EG, Jr, Burkes EJ, Briggaman RA. Hereditary epidermolysis bullosa: oral manifestations and dental therapy. *Oral Surgery, Oral Medicine and Oral Pathology* 1976; **42**: 490–500.
12 Kahn SB, Trieger N. Epidermolysis bullosa hereditaria letalis – a case report with special emphasis on oral manifestations. *Journal of Oral Medicine* 1976; **31**: 32–35.
13 Caroll DL, Stephan MJ, Hays GL. Epidermolysis bullosa – review and report of case. *Journal of the American Dental Association* 1983; **107**: 749–751.
14 Wright JT. Epidermolyiss bullosa: dental and anesthetic management of two cases. *Oral Surgery, Oral Medicine and Oral Pathology* 1984; **57**: 155–157.
15 Lanier PA, Posnick WR, Donly KJ. Epidermolysis bullosa – dental management and anesthetic considerations: case report. *Pediatric Dentistry* 1990; **12**: 246–249.
16 Moghadam BK, Gier RE. Epidermolysis bullosa: oral management and case reports. *ASDC Journal of Dentistry for Children* 1992; **59**: 66–69.
17 Sipahier M. Epidermolysis bullosa: a case report. *Quintessence International* 1994; **25**: 839–843.
18 Olsen CB, Bourke LF. Recessive dystrophic epidermolysis bullosa. Two case reports with 20-year follow-up. *Australian Dental Journal* 1997; **42**: 1–7.
19 Bohaty B, Spencer P, Dunlap C, Wandera A. Epidermolysis bullosa: case report of appropriate classification of subtype because of an early dental exam. *Journal of Clinical Pediatric Dentistry* 1998; **22**: 243–245.
20 Liu HH, Chen CJ, Miles DA. Epidermolysis bullosa simplex: review and report of case. *ASDC Journal of Dentistry for Children* 1998; **65**: 349–353.
21 Wright JT, Fine JD, Johnson L. Dental caries risk in hereditary epidermolysis bullosa. *Pediatric Dentistry* 1994; **16**: 427–432.
22 Harris JC, Bryan RA, Lucas VS, Roberts GJ. Dental disease and caries related microflora in children with dystrophic epidermolysis bullosa. *Pediatric Dentistry* 2001; **23**: 438–443.
23 Wright JT, Childers NK, Evans KL, Johnson LB, Fine JD. Salivary function of persons with hereditary epidermolysis bullosa. *Oral Surgery, Oral Medicine and Oral Pathology* 1991; **71**: 553–559.
24 Marini I, Vecchiet F. Sucralfate: a help during oral management in patients with epidermolysis bullosa. *Journal of Periodontology* 2001; **72**: 691–695.