

Orthodontic care for patients with epidermolysis bullosa

Epidermolysis bullosa (EB) is a diverse group of disorders with blister formation in response to mechanical trauma. Patients with EB requiring orthodontic treatment require special consideration

Epidermolysis bullosa (EB) is a diverse group of disorders characterised by increased skin fragility and blistering of the skin following minor or insignificant trauma or traction of the skin (Fine, 2010). The condition came to public attention following the broadcast of documentary 'The Boy Whose Skin Fell Off' which told the story of Jonny Kennedy who had EB (Channel 4, 2004). In addition to skin fragility and blistering, depending on the type of EB that is present, internal blistering can occur in the oral cavity, the larynx and the pharynx. This internal blistering may lead to reduced opening of the temporomandibular joint, tender soft tissues in the oral cavity, reduced saliva production and decreased mobility of the tongue due to the fusion of the tongue to the floor of the mouth.

Because of these symptoms, designing orthodontic appliances for these patients who have been diagnosed with epidermolysis bullosa can present

challenges that need to be overcome for satisfactory treatment. The conventional wire and acrylic components of orthodontic appliances will usually be intolerable for patients with EB.

This article looks at orthodontic appliances that have been used for patients with EB at London's Great Ormond Street Hospital. The appliances are designed to be free of contact with the mucosa and be completely tooth borne.

Etymology

The skin is composed of different layers. The outer is called the epidermis and the inner layers are the dermis. 'Lysis' means breakdown and 'bullosa' means blister filled with fluid; therefore, epidermolysis bullosa means breakdown and blistering of the skin.

Aetiology

EB is caused by a mutation in the keratin or collagen gene (Prockop and Ala-Kokko, 2005).

Symptoms

The main indication of EB is the eruption of fluid-filled blisters on the skin, most commonly on the hands and feet in response to friction. Depending on the type of EB present, blisters will usually develop in various areas. In mild cases, blisters heal without scarring. Symptoms and other signs of EB may include blistering of the skin; deformity or loss of fingernails and toenails; internal blistering in the oral cavity, the larynx, pharynx, oesophagus,

upper airway, stomach, intestines and urinary tract; hyperkeratosis (thickening of the skin on the palms and soles of the feet); scarring alopecia (scalp blistering, scarring and hair loss); atrophic scarring (thin-appearing skin); milia (small white pimples on the skin); excessive sweating, dysphagia (difficulty with swallowing) and dental anomalies, such as tooth decay from poorly formed tooth enamel.

Classification

Traditionally, EB types have been classified according to skin morphology (Fine, 2010). The classification system for inherited EB is recommended by an international committee of experts, based on the ultrastructural level of skin separation, clinical phenotype, and genotype gives three major EB types with distinctive clinical subtypes. This ultrastructural level of separation is determined using transmission electron microscopy and/or immunofluorescence antigenic mapping (Fine et al, 2000). See *Figure 1* for an interpretation of how EB types and subtypes are classified.

Epidermolysis bullosa simplex

Epidermolysis bullosa simplex (EBS) describes the type of EB that manifests at the dermoepidermal junction (Freedberg et al, 2003). The dermoepidermal junction is where the epidermal and the dermal layers of the skin join (*Figure 2*). *Figure 3* shows the blistering on the hands and feet of a patient with EBS.

James Green is a maxillofacial and dental laboratory manager at Great Ormond Street Hospital for Children NHS Foundation Trust in London. He is also a volunteer speaker for DebRA, the national charity working for people in the UK with EB.

Email: jamesgreen@nhs.net

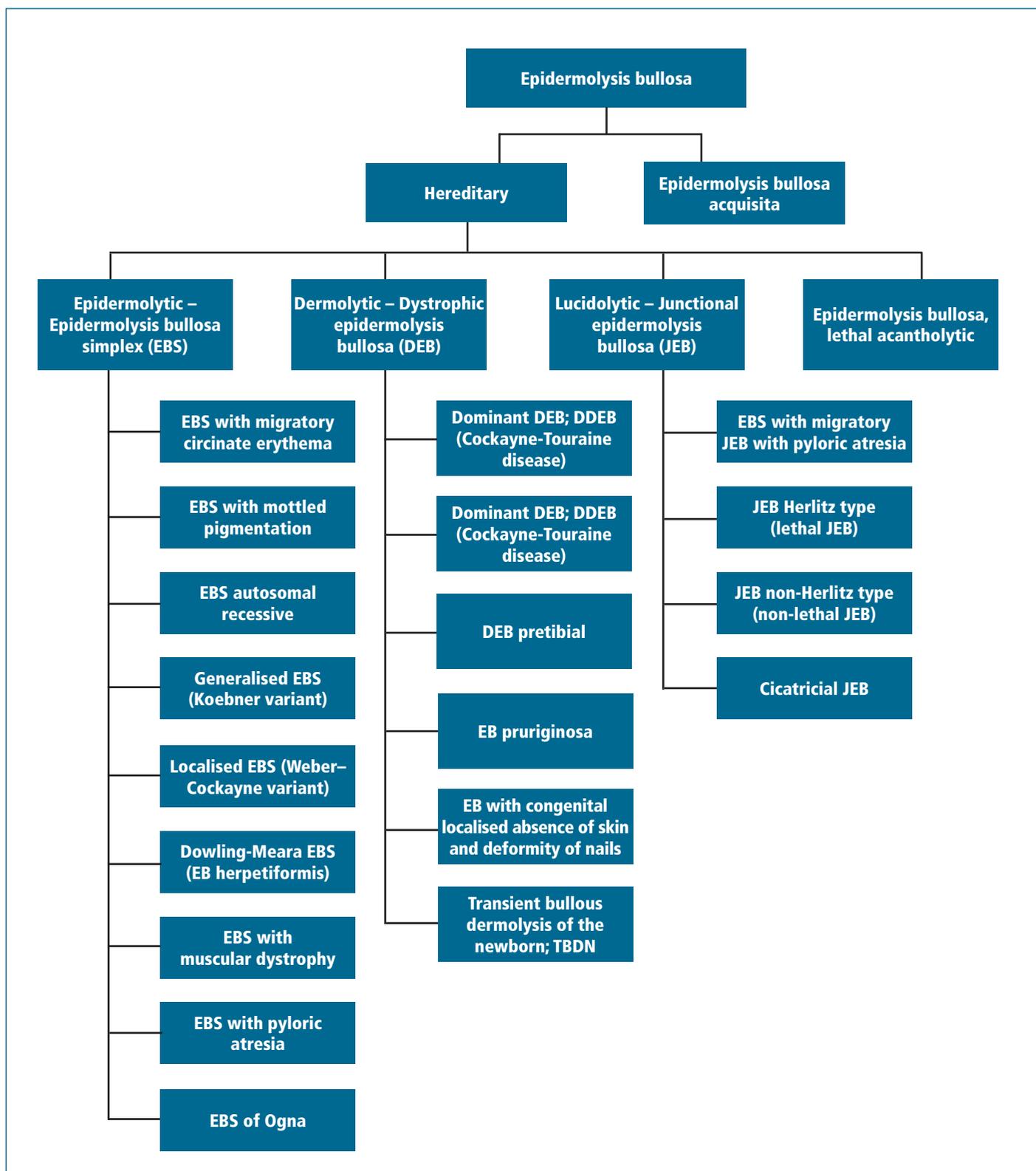


Figure 1. Classification of EB types

Junctional epidermolysis bullosa

Junctional epidermolysis bullosa (JEB)

describes the type of EB where blisters occur within the lamina lucida of the basement membrane zone (Freedberg et al, 2003). Figure 4 shows an infant with Herlitz JEB.

Dystrophic epidermolysis bullosa

Dystrophic epidermolysis bullosa (DEB) describes the type of EB that occurs as



Figure 2. The dermoepidermal junction



Figure 3. The hands and feet of a patient with EBS (courtesy of DebRA)



Figure 4. An infant with Herlitz JEB (courtesy of DebRA)

sublamina densa basement membrane zone separation (Freedberg et al, 2003). Figure 5 shows an example of a young boy with the recessive type of DEB.

Diagnosis

Skin biopsy

A skin biopsy requires the removal of a small piece of the affected skin and examining it under a microscope to reveal where the skin is separating and the type of EB that the patient has. Specialised tests, such as electron microscopy or immunoelectron microscopy, can also be required to substantiate the diagnosis.

Genetic testing

The majority of types of EB are inherited and the diagnosis can be confirmed with genetic testing. This would usually involve a blood sample being taken and being analysed in a laboratory to confirm the diagnosis.

Epidemiology

Age

Except in very mild cases of EBS, which can remain undetected until adulthood or occasionally remain undiagnosed altogether, EB will be diagnosed shortly after birth.

Prevalence

According to Fine et al (1999), 50 cases of epidermolysis bullosa occur per one million live births. Of these cases, approximately 92% are EBS, 5% are DEB, 1% are JEB, with the remainder unclassified (see Figure 6).

Mortality and morbidity

Patients with dominantly inherited epidermolysis bullosa simplex and dystrophic epidermolysis bullosa and milder JEB subtypes may not affect a patient's life expectancy. However, in most cases infancy is a difficult time for patients with EB. Blistering caused by any type of the disorder may be complicated by infection and /or sepsis (blood poisoning). Severe forms of EB increase the risk of death in infancy; patients with the Herlitz



Figure 5. A young boy with recessive DEB (courtesy of DebRA)



Figure 7. A conventional appliance to procline the maxillary incisors



Figure 8. An epidermolysis bullosa lower removable appliance with inclined plane to procline the maxillary incisors

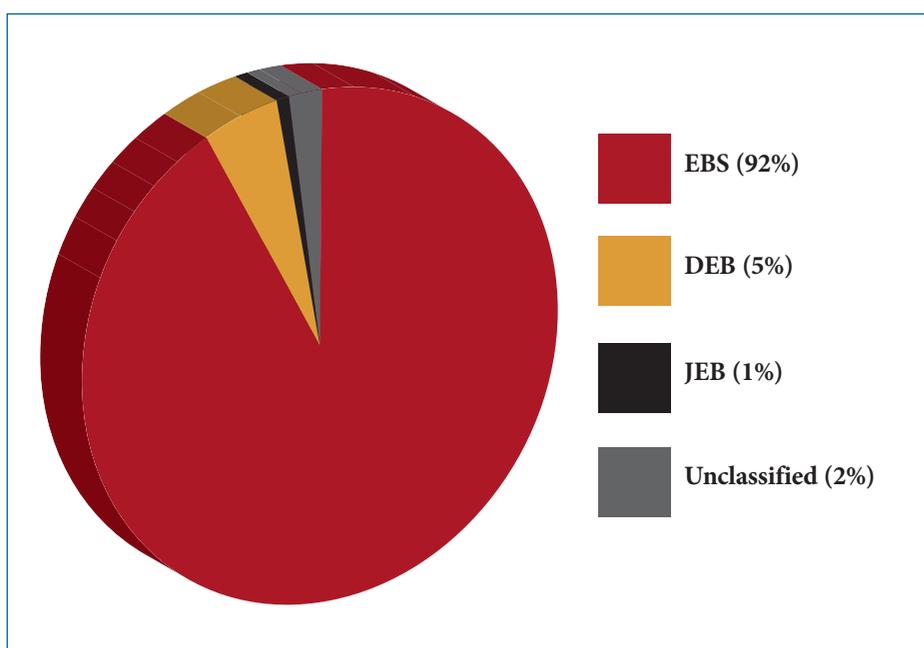


Figure 6. Prevalence of EB types

subtype of JEB (Figure 4) are at the highest risk during infancy and have an estimated mortality rate of 87% in the first year of life. For those who survive childhood, the most common cause of mortality is metastatic squamous cell carcinoma (SCC; a form of skin cancer). SCC manifests specifically in patients with recessively inherited EB and are usually 15–35 years of age. It was reported by Risser et al (2009) that from 1979–2002, the overall age-adjusted annual mortality rate from bullous skin diseases was 0.103 deaths per 100 000 population.

Dental considerations

Depending on the type of EB that a

patient presents with, blistering can occur internally in the mouth, the larynx and the pharynx. Decreased opening of the temporomandibular joint, fragile oral mucosa, reduced saliva production and decreased mobility of the tongue due to the fusion of the tongue to the floor of the mouth can be caused by internal blistering. Dental treatment is also dependent on the type of EB that the patient presents with. Those with EB simplex may be able to be treated as normal patients while those with recessive dystrophic EB present the most severe oral manifestations which complicate dental treatment, although successful dental treatment under general anaesthetic for

patients with the most severe forms of EB have been described (Block and Gross, 1982; Wright 1984, 1990). Good dental hygiene is essential for patients with EB and a dentist familiar with the disorder should be consulted if possible. Because of enamel defects patients with JEB or DEB will develop caries regardless of the standard of oral hygiene. Patients should avoid mouthwashes containing alcohol and use saline rinses to clean the mucosa (American Academy of Pediatric Dentistry Council on Clinical Affairs, 200–2009).

Orthodontic appliances

Because of the symptoms described, orthodontic treatment with conventional appliances would be impossible for many patients with epidermolysis bullosa. A conventional removable orthodontic appliance consists of an acrylic palatal plate with stainless steel wire components which provide active and retentive elements. Both acrylic resting on the fragile oral tissue and stainless steel wire elements could prove painful and impossible for

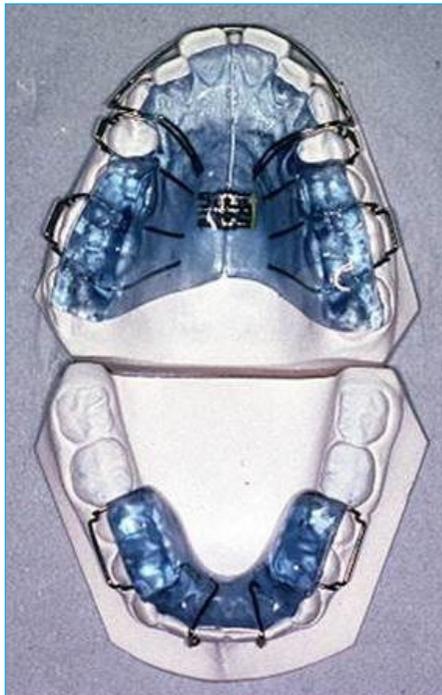


Figure 9. A conventional twin block appliance

patients with EB to tolerate. Figure 7 shows a maxillary removable appliance for a patient without EB. The T springs will be used to procline the maxillary incisors. Depending on the type of EB that is present, an appliance such as this would irritate the delicate oral tissues and could aggravate or create blistering. Appliances for patients with EB need to be designed without the conventional elements of a removable orthodontic appliance, i.e. no stainless steel wires or acrylic resting on the palate.

Appliances need to be completely tooth borne (i.e. resting on the teeth without any pressure on the mucosa) and prepared using heat polymerised acrylic rather than auto polymerised acrylic to reduce the probability of residual monomer in the appliance irritating the oral environment. Auto-polymerising acrylic resins also have higher cytotoxicity than heat-polymerising acrylic resins (Kim et al, 2002).

Extra care needs to be taken to ensure that the appliance is totally smooth and free of any corners or edges that could cause trauma to the patient's delicate tissues. As with the appliance in Figure 7,



Figure 10. An epidermolysis bullosa twin block

Box 1. The four golden rules of designing orthodontic appliances for patients with epidermolysis bullosa

Appliances should be designed to be:

- Free of wire components
- Prepared using heat-polymerised acrylic
- Rounded, smooth and free of any corners or edges
- Tooth borne and free of any pressure on the gingival tissue

the appliance in Figure 8 is for a patient with instanding maxillary incisors. The appliance is worn on the lower teeth and uses an inclined plane to guide the teeth into their correct position.

Correction of Class II Division II Malocclusion

The use of a twin block appliance (Clark, 1982) has proved to be a popular and successful way to treat Class II Division II malocclusions. The Twin Block Appliance is a two part functional appliance with acrylic base plates retained with stainless steel clasps and bite blocks to provide a functional mandibular displacement. One such appliance is shown in Figure 9.

As discussed previously, a patient with EB and oral involvement would not be able to tolerate this appliance and an alternative design is shown in Figure 8.

The epidermolysis bullosa twin block (EBTB) uses bite blocks that are similar to those described by Clark. The EBTB is prepared in heat polymerised acrylic and fits over the teeth, hence is tooth borne and does not rest on the mucosa

and does not need stainless steel clasps to retain it in position (Figure 10). Also note the difference between the blocks on the appliances. The blocks on the EBTB (Figure 10) are very rounded and smooth and resemble ice cubes while those on the conventional appliance (Figure 9) are more clearly defined blocks. The author's 'four golden rules' for designing appliances for patients with EB are summarised in Box 1.

Conclusion

Due to the severity of the condition, it could be argued that orthodontics for these children with EB should not be attempted. However, it is the author's opinion that the aim should be to make life as normal as possible for them. Today orthodontic treatment is a normal part of growing up for many children and if a child is unhappy with the way their teeth look, most children have the option of orthodontics—why not those with EB? DN

Further information

DebRA is the national charity working on behalf of people in the UK with EB. Visit the DebRA website at www.debra.org.uk. The photographs in figures 3, 4 and 5 are provided courtesy of DebRA.

American Academy of Pediatric Dentistry Council on Clinical Affairs (2008-2009) Guideline on management of dental patients with special health care needs. *Pediatric Dentistry* 30(Suppl 7): 107–11

Block MS, Gross BD (1982) Epidermolysis bullosa dystrophica recessive: Oral surgery and anesthetic considerations. *J Oral Maxillofac Surg* 40: 753–8

Channel 4 (2004) The Boy Whose Skin Fell Off. 25 March 2004. Available at: <http://www.channel4.com/programmes/the-boy-whose-skin-fell-off/4od#3131855> (accessed 24 March 2012)

Clark WJ (1982) The Twin Block traction technique. *Eur J Orthod* 4: 129–38

Fine JD, Bauer EA, McGuire J, Moshell A, eds (1999) *Epidermolysis Bullosa: Clinical, Epidemiologic, and Laboratory Advances and the Findings of the National Epidermolysis Bullosa Registry*. Johns Hopkins University Press, Baltimore

Fine JD, Eady RA, Bauer EA et al (2000) Revised classification system for inherited epidermolysis bullosa: report of the second International Consensus Meeting on diagnosing and classification of epidermolysis bullosa. *J Am Acad Dermatol* 42(6): 1051–66

Fine JD, Eady RA, Bauer EA et al (2008) The classification of inherited epidermolysis bullosa (EB): Report of the Third International Consensus Meeting on Diagnosis and Classification of EB. *J Am Acad Dermatol* 58(6): 931–50

Fine JD (2010) Inherited epidermolysis bullosa: past, present, and future. *Ann N Y Acad Sci* 1194: 213–22

KEY POINTS

- **Epidermolysis bullosa is a rare condition that is usually diagnosed shortly after birth with only 50 cases being reported per one million live births.**
- **The main symptom of the condition is the eruption of blisters on the skin that occur most commonly on the hands and feet but also arise in other areas.**
- **Blistering can occur internally in the mouth, in the larynx and in the pharynx.**
- **Patients requiring orthodontic treatment need special consideration because of these symptoms.**

Freedberg IM, Eisen AZ, Wolff K et al (2003) *Fitzpatrick's Dermatology in General Medicine*. 6th edn. McGraw-Hill Medical Publishing Division, New York

Gedde-Dahl T Jr (1971) *Epidermolysis Bullosa. A clinical, genetic and epidemiologic study*. The John Hopkins Press, Baltimore

Kim SK, Chang IT, Heo SJ, Keak JY (2002) Cytotoxicity of denture base resins. *Journal of Korean Academy of Prosthodontics* 40(4): 309–22

Prockop DJ, Kuivaniemi H, Tromp G (1993) Heritable disorders of connective tissue. In: Kasper DL, Braunwald E, Fauci AS et al (eds).

Harrison's Principles of Internal Medicine. 16th edn. McGraw-Hill Medical Publishing Division, New York

Risser J, Lewis K, Weinstock MA (2009) Mortality of bullous skin disorders from 1979 through 2002 in the United States. *Arch Dermatol* 145(9): 1005–8

Wright JT (1984) Epidermolysis bullosa: Dental and anaesthetic management of two cases. *Oral Surg Oral Med Oral Pathol* 57: 155–7

Wright JT (1990) Comprehensive dental care and general anaesthetic management of hereditary epidermolysis bullosa: A review of fourteen cases. *Oral Surg Oral Med Oral Pathol* 70: 573–8