Recessive dystrophic Epidermolysis bullosa. Two case reports with 20-year follow-up

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Abstract
These two case reports highlight the enormous clinical difficulties faced by dentists in providing satisfactory long-term dental care to patients who suffer from Epidermolysis bullosa. Problems of bullae formation in oral soft tissues and subsequent scarring are outlined in relation to the difficulty of maintaining satisfactory oral hygiene and a diet leading to minimal dental caries experience. The behavioural problems of maintaining patient compliance for preventive and restorative dentistry in this painful and debilitating disease are illustrated in these case reports. Difficulties in providing restorative care, either under local anaesthesia or general anaesthesia are discussed, and a novel replacement of non-viable carious anterior teeth using a nine-unit porcelain fused to metal Rochette type bridge is presented. Dental management of patients with Epidermolysis bullosa should commence at birth, and non-compliance in dental attendances should be followed up by social workers to prevent the disastrous oral morbidity that frequently occurs in such patients.

Key words: Recessive dystrophic Epidermolysis bullosa, dental caries, Rochette bridge.

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Introduction
Satisfactory dental management of children and adults who suffer from Epidermolysis bullosa presents one of the most difficult clinical challenges to both paediatric and general dentists alike. Management problems may be divided into three areas: soft tissue lesions including periodontal disease, malformation and diseases of dental hard tissues, and preventive and behavioural management.

Epidermolysis bullosa represents a group of mainly hereditary skin disorders manifested by an exceptional tendency of the skin and mucosa to form bullae and vesicles following minor friction and trauma. Up to 23 types of Epidermolysis bullosa have been described in recent literature, often with close similarities in clinical manifestations. It is recommended that analysis of all cases be undertaken using electron microscopy, immunofluorescence and immunochemical studies.

Differential diagnoses include bullous impetigo (pemphigus neonatorum), Ritter’s disease, porphyria congenita, congenital syphilis, and juvenile bullous dermatitis herpetiformis. Dentally, amelogenesis imperfecta should be considered.

Clinically, two main types were described originally: the simplex type in which scarring does not occur and mucous membranes are rarely affected; and the dystrophic type in which scarring occurs, and where three forms have been reported based on the pattern of inheritance. These are an autosomal recessive lethal type where bullae do not heal and death occurs in infancy; an autosomal dominant dystrophic form associated with severe scarring; and an autosomal recessive form termed Recessive dystrophic Epidermolysis bullosa in literature before 1980, in which scarring occurs, physical development is impaired and ectodermal defects of hair, nails and sweat glands are present.

A recent classification (Table 1) is based on the anatomical level of tissue cleavage following mechanical trauma to the skin or mucous membrane. Distinct subgroups are then delineated on the basis of phenotypic characteristics and mode of inheritance.

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Cementum has been reported in early literature; however, the occurrence of enamel hypoplasia in Type III recessive dystrophic forms is controversial in recent literature.2,7

Case reports

Patient A

Patient A, a Caucasian male, was noted at birth to have blisters on the hands and feet. He was transferred to a major paediatric teaching hospital, where medical management was then undertaken by a specialist dermatologist. A clinical diagnosis was made of recessive dystrophic Epidermolysis bullosa.

The patient had a male sibling (Patient B) four years older, who had also been diagnosed previously with the condition, although with less severity. There was no family history of the condition.

By the age of four years Patient A had lost all his finger and toe nails. The knees, ankles, elbows and mouth showed many blisters. At the age of 10 years the fingers were webbed together and fused in flexion. Medical care was essentially supportive, with the use of vitamin E supplements and creams. A trial of the drug phenytoin was undertaken with no apparent clinical improvement. The patient was noted to ‘require dental attention’. Subsequently, all four first permanent molars and all deciduous molars were removed under general anaesthesia by a private general dental practitioner.

At age 11 years the dermatologist noted ‘shocking dental caries’ and referred the patient to the dental department of a major paediatric teaching hospital.

An intensive preventive programme including diet counselling and oral hygiene instruction was instituted, and restoration of carious teeth was undertaken using local anaesthesia. The patient became uncooperative for restorative treatment over successive visits, even with the use of nitrous oxide relative

Table 1. Classification of Epidermolysis bullosa

<table>
<thead>
<tr>
<th>Type</th>
<th>Description</th>
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<tr>
<td>Type I</td>
<td>Intra-epidermal forms: non-scarring; autosomal dominant and X linked.</td>
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<tr>
<td>Type II</td>
<td>Junctional forms: associated with hemidesmosome defect and skin atrophy; autosomal recessive.</td>
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<tr>
<td>Type III</td>
<td>Dermal forms: characterized by scarring and atrophy, autosomal dominant and recessive.</td>
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<tr>
<td>Type IV</td>
<td>Epidermolysis bullosa acquisita: acquired non-hereditary form.</td>
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Recessive dystrophic Epidermolysis bullosa as traditionally reported in the literature has been more recently termed scarring Epidermolysis bullosa with dermolytic blisters (Hallopeau-Siemens type). This type of Epidermolysis bullosa may be divided into four autosomal recessive subtypes which are difficult to differentiate.2,6

Systemic features include bullae formation on hands, feet, elbows and knees; which are initially noted at or soon after birth. Bullae leave painful ulcers on rupturing; and healing is often followed by scarring and tissue contraction.3,6 Contractures and pseudosyndactyly of digits may result in the formation of claw-like hands. Frequently the upper oesophagus may become stenotic, leading to dysphagia.

Oral features include repeated blistering and scar formation leading to limited oral opening, ankyloglossia, elimination of buccal and vestibular sulci, perioral stricture, severe periodontal disease and alveolar bone resorption, atrophy of the maxilla with mandibular prognathism, increased mandibular angle, and predisposition to oral carcinoma. Routine dental care or even normal toothbrushing may cause bullae formation on the lips and oral mucosa.3

Histologically, bullae occur below the periodic acid Schiff-positive basement membrane, with hemidesmosomes and tonofibrils being absent or reduced in numbers.3 Hypoplasia of enamel with absence of prismatic structure and defective cementum has been reported in early literature, however the occurrence of enamel hypoplasia in Type III recessive dystrophic forms is controversial in recent literature.2,7

Fig. 1. – Patient A at 16 years: panoramic radiograph.
analgesia. At age 13 years his dental condition had deteriorated despite a preventive programme which included the use of chlorhexidine and fluoride gels for use at home. Over the next three years, four cancelled appointments and 13 broken appointments were recorded.

At age 16 years the patient and his mother requested that all dental treatment be completed under general anaesthesia, due to the painful and unsatisfactory treatment attempted previously under local anaesthesia.

At this time the patient was referred to author CBO. An examination was conducted, and a panoramic radiograph (Fig. 1) and periapical radio-graphs (Fig. 2a, b) of the extensively carious incisor teeth were taken.

A diagnosis was made of rampant dental caries associated with coronal destruction of multiple teeth (Fig. 3, 4). Multiple Class II, III and V lesions were charted. The anterior teeth were non-vital. An unerupted, impacted tooth 45 was seen on the panoramic radiograph. Oral hygiene was poor, mucosal ulceration was present, and access to the mouth was limited. The patient expressed intense dislike and anxiety regarding further dental treatment under local anaesthesia.

Fig. 2a, b. – Patient A at 16 years: periapical radiographs, teeth 12 11 21 22.

Fig. 3, 4. – Patient A at 16 years: intra oral views of dentition; note extensive coronal destruction. (Note mirror view in Fig. 4.)
Dermatology, anaesthesia and prosthodontic consultations were obtained. The alternatives of a maxillary partial denture or a Rochette bridge to replace the maxillary anterior teeth were discussed with the patient.\textsuperscript{8,9} (Maryland type etched casting bridges had not yet been described in the literature.)

Initial management consisted of deep sedation with premedication with omnopon and scopolamine, and induction with intra-venous diazepam and ketamine. Ketamine is a dissociative anaesthetic agent which maintains the oropharyngeal reflex. Xylocaine local anaesthesia was used also. Amalgam restorations were placed, and non-viable teeth were extracted. In view of the bloody field created, it was decided to allow healing before undertaking a second procedure to complete the remaining restorative needs. A second deep sedation session using the same technique was undertaken two months later, when all remaining treatment was completed. The asymptomatic impacted tooth 45 was left in as removal would have caused extensive soft tissue trauma.
Following completion of restorative and exodontic treatment, an intensive preventive regimen using chlorhexidine gluconate gel and sodium fluoride gel for home use was commenced. Three months after surgery, alginate impressions were obtained with difficulty using cut down impression trays. A nine unit porcelain fused to metal Rochette bridge replacing teeth 12,11,21,22 was constructed and inserted using enamel bonded, chemically cured resin composite (Concise). Such a prosthesis was deemed viable as there were no opposing occlusal scarring of circumoral soft tissues.

Remaining mandibular buccal teeth had retroclined over the intervening nine months from the restorative treatments, presumably due to the progressive scarring of circumoral soft tissues.

A preventive recall programme was instituted; however, patient attendance was irregular. At age 20 years lateral oblique radiographs were taken, and two amalgam restorations and two fissure sealants placed. At 21 years the bridge loosened and was reinserted with light-cured resin composite (Silux).

At patient age 23 years the bridge loosened. It was removed, and after sand-blasting the metal framework was re-inserted using Panavia, a resin luting agent recently introduced for use with Maryland bridges. Extensive decalcification and caries was present on palatal surfaces of the remaining teeth. All lesions were restored with glass ionomer cement Ketacfil prior to reinsertion of the bridge.

The patient failed to attend for any dental care for the following four years. At age 27 years he presented complaining of pain to cold foods from the maxillary left quadrant. Deep caries with a near exposure was found in tooth 23. A panoramic radiograph (Fig. 6) and periapical radiograph of tooth 23 were taken prior to restoration with composite resin Silux. It was suspected that endodontic treatment might be required for this tooth in the future.

Three other carious lesions were subsequently restored with amalgam and glass ionomer cement Ketacfil. A preventive programme using chlorhexidine gel and sodium fluoride mouth rinse was reintroduced. Records show that 17 prescriptions were given for chlorhexidine and fluoride preparations over an eight-year period. At age 28 years plastic surgery on both right and left hands was undertaken to improve function (Fig. 7).

At age 29 the patient moved to an interstate capital city which had a non-fluoridated water supply. He had resided previously in a city with an optimally fluoridated water supply from age 13 years to 29 years. The bridge dislodged at age 30 years at which stage management was undertaken by author LFBo. Extensive decalcification and dental caries were present, as seen in the panoramic radiograph taken at this time (Fig. 8). Restorations were placed and a small maxillary acrylic partial denture constructed. It was felt that his natural teeth had a poor prognosis. Full maxillary and mandibular dentures, if well constructed and highly polished, may perhaps be tolerated by the patient if they do not cause excessive trauma and blistering.

The patient has been unemployed for lengthy periods since leaving school at the age of 16 years. Temporary employment has been obtained doing light manual work, and in keyboard data entry. He has seemed overwhelmed by his medical condition, and disinterested in dental care except for symptomatic care for pain or aesthetics of maintaining anterior teeth. This has been reflected in limited compliance with recall programmes and preventive strategies.

**Patient B**

Patient B, the male sibling of Patient A and 4 years older, also was diagnosed clinically with Recessive dystrophic Epidermolysis bullosa at birth. He has suffered from soft tissue lesions of hands and feet and also has similar, although less severe, lesions of oral soft tissues, limited oral opening and reduction of buccal and vestibular sulci with associated periodontal disease.

Dental management was undertaken by author CBO for the patient from age 22 years for a period of 13 years. The patient has been almost caries free; however, he has experienced continuing calculus build up around the mandibular anterior teeth. Dental management has consisted of regular recall examinations, scaling of teeth using a careful and gentle technique, and a preventive programme using chlorhexidine and sodium fluoride gels. Patient compliance has been excellent.

The patient has had professional employment since obtaining a tertiary educational qualification. He has acquired and has maintained keyboard skills, despite severe scarring of both hands and contractures of fingers.

**Discussion**

Recessive dystrophic Epidermolysis bullosa presents the dentist a most difficult problem, and is probably more taxing of modern preventive and restorative techniques than almost any other condition. The painful, traumatic and unsatisfactory nature of restorative procedures undertaken using local anaesthesia is reflected in the 17 broken or
cancelled appointments of Patient A in his early teenage years. However, as prosthetic treatment is at best of doubtful value in these cases, it is imperative that the natural dentition be maintained to help patients socially, psychologically and nutritionally.

Preventive strategies must be aimed at plaque-related diseases of dental caries and periodontal disease. Diminished oral opening and progressive obliteration of the labial sulcus make oral hygiene difficult. Minor trauma from toothbrushing may cause eruption of oral bullae, causing discomfort and scarring. Patient dexterity in performing oral hygiene measures is often limited by hands and fingers that have become claw-like through repeated scarring. Adequate oral hygiene should be encouraged with the gentle use of small headed soft toothbrushes and small sponge applicators. The physical removal of bacterial plaque may be supplemented with chemical inhibition by the use of 0.2 per cent chlorhexidine gluconate solution twice daily. This may be swabbed around the mouth in young children, and in the older child it may be used as a mouthwash. Chlorhexidine is adsorbed to oral mucosal surfaces where it exerts an antibacterial effect for several hours. This property may also assist in reducing secondary infection of ulcerated mucosal surfaces, thereby reducing discomfort and promoting more rapid healing. The astringent taste may be unacceptable to some patients, and the brown discoloration of the acquired pellicle may require professional removal every few months.

The use of fluoride to prevent dental caries is another vital component in prevention. Neutral pH sodium fluoride mouthwashes are useful, while acidic fluoride preparations cause discomfort when oral ulceration occurs. Neither patient received fluoride supplements in early life, and the water supply was not fluoridated until Patients A and B were 13 and 17 years of age respectively. Poor compliance with prescribed fluoride mouth rinses and gels was the norm for Patient A.

Diet constitutes a major difficulty in caries control. Nutritional advice may be indicated as coarse foods are not well tolerated, and a high caries rate is often the norm. Restorative or exodontic treatment performed under local anaesthesia inevitably causes blistering and progressive scarring. The resulting discomfort to Patient A presumably resulted in non-compliance for subsequent dental treatment and non attendance for review examinations.

Modifications to operative technique in patients with Epidermolysis bullosa include the use of extreme care of fragile tissues, using little pressure and no sliding movements of tissues, and entering the mouth on as few occasions as possible. The air syringe and suction must be used carefully, and the use of adhesive tape to locate an intravenous cannula or to protect the eyes under general anaesthesia must be avoided.

Extensive dental treatments, where all restorative and exodontic needs may be completed at one session, may be better conducted under endotracheal intubation or deep sedation. It has been shown that concerns of laryngeal obstruction are largely unfounded with a skilled intubation technique. It could be argued that Patient A would have been better served if dental treatment had been carried out under general anaesthesia on his initial referral at age 10 years for specialist paediatric dental care.

The low dental caries experience of Patient B may be attributed to dietary and salivary factors as indicated by his propensity for calculus formation, as well as compliance with preventive dental care strategies.

The case history of Patient A, where there was no regular dental care until the age of 10, shows the disastrous consequences of dental caries in such patients. Patients with Epidermolysis bullosa and their families should receive regular preventive dental care and counselling from birth. Ideally non-compliant patients should be followed up by social workers to prevent the dental morbidity which frequently occurs in patients who suffer from Epidermolysis bullosa.

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