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CASE REPORT

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Stomatological management and implant-supported rehabilitation in a patient with recessive dystrophic epidermolysis bullosa

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Abstract

Inherited epidermolysis bullosa (EB) is a disease that causes epithelium fragility due to a protein anomaly caused by a genetic mutation. Epidermolysis bullosa clinical manifestations are bullae and cutaneous-mucosal erosions. Epidermolysis bullosa is a rare disease, with different clinical presentations depending on the type and subtype. The stomatological treatment depends on the oral manifestations and EB types. There is no high level of recommendations due to the limited cases described in the literature. We describe an implant-supported dento-maxillary rehabilitation of a 49-year-old patient suffering from a newly diagnosed hereditary recessive EB with disabling oral manifestations. In the current case, the diagnosis of recessive dystrophic epidermolysis bullosa has been confirmed, and adequate dental care was carried out taking into account the disease constraints.

KEYWORDS

epidermolysis bullosa, oral manifestation, prosthetic rehabilitation

1 | INTRODUCTION

Inherited epidermolysis bullosa (EB) is a genetic disease. It results from an abnormality in specific proteins' constitution involved in the epidermal cohesion. The pathology is clinically expressed by the formation of bullae and cutaneous-mucosal erosions. The EB prevalence reaches 20 cases per million inhabitants.[1] Fine et al. [dataset][2] describe four major classes of inherited EB: epidermolysis bullosa simplex (EBS), expressing bullae in the epidermis; junctional epidermolysis bullosa (JEB), expressing bullae in the lamina lucida; dystrophic epidermolysis bullosa (DEB), expressing bullae in the upper dermis; and Kindler syndrome, a specific entity expressing bullae on multiple epidermis and dermis layers.

The proteins responsible for cutaneous cleavage differ according to the type of epidermolysis. Penetrance is variable depending on individuals and tissues. Till date, 17 genes have been identified in EB. The transmission mode is either autosomal dominant or recessive. However, the mutation can also occur *de novo* in the early embryonic life.[2]

We report a functional and aesthetic implant-supported rehabilitation of a patient with a newly diagnosed inherited recessive EB with disabling oral manifestations.

2 | CASE REPORT

A 49-year-old patient was referred to the stomatology department for generalized dental pain, causing eating difficulties and masticatory discomfort due to the presence of internal jugal flanges. The patient requested a total dental avulsion and prosthetic rehabilitation. The patient described a medical history of disease-causing the formation of mucosal and cutaneous bullae. Based on the clinical presentation at birth, resulting in oral and dental lesions; esophageal stenosis, operated on twice; multiple bullae located mainly on the feet, hands, and scalp; and genital lesions, an "epidermolysis bullosa" was suspected.

The work-up included a clinical examination and medical imaging (Figure 1). The clinical examination revealed an exacerbated gingival sensitivity secondary to ulcerations, erosions related to bullae rupture, multiple caries caused by brushing difficulties, a generalized periodontosis, and numerous osteitis foci in the maxilla and mandible. Oral vestibule obliteration, secondary to retractile fibrous scarring following the bullae rupture was highlighted. It resulted in a 20 mm inter-dentally mouth opening limitation, and bilateral loss of the maxillary and mandibular vestibules. Moreover, the patient presented a macroglossia and a microstomia, DEB clinical characteristic signs.[3]

The patient was an only child, with no family history of a genetic disease. The patient was not taking any medication and his medical follow-up was focused on esophageal stenosis.

Before the treatment, a biopsy of a perineal bullous area was performed. A genetic advice was requested. A "trio mendelioma" testing the parents and the patient was prescribed, revealing a *COL7A1* gene mutation, and confirmed the DEB diagnosis. Both parents were heterozygous carriers, explaining why none of them developed the disease. Notwithstanding, the patient was a homozygous carrier and thus expressed the disease.

A total dental avulsion with alveolar preservation was performed. One month after the procedure, the patient showed good mucosal healing (Figure 2). Nevertheless, bullae on the palate and the jugal mucosa, and bilateral scarring were observed. The mouth opening reached 40 mm between the two edentulous ridges.

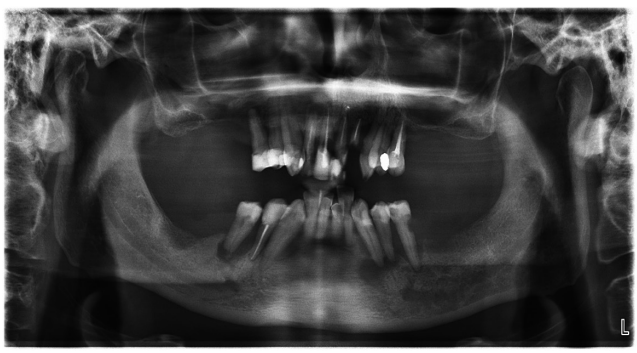

FIGURE 1 Pre-extraction surgery orthopantomogram showing periodontal disease, dental infections, and caries

During the care, the type of rehabilitation was questioned. A removable prosthesis was not recommended.[1,4] The prosthesis friction on the mucosa could foster bullae formation and the vestibule loss would not allow satisfactory prosthetic stability. In addition, the limitations of oral access for impression trays due to the microstomia and the limited mouth opening would have complicated the prosthesis management.

During the bone healing, a Therabite® (manufacturer: Atos Medical AB) device (Figure 3) was prescribed to promote mouth opening.[4] However, the device's mouthpieces were unsuitable due to the patient's particular anatomical conditions. Therefore, we custom-made polylactic acid (PLA) resin mouthpieces using computer-aided design and manufacturing techniques. The modeling was done using Blender® software (open source: https://blender3d.fr/), and the mouthpieces were printed using a Makerbot 3D plus® (Manufacturer: Makerbot USA) printer (Figure 3).

Six months after the dental extractions, four maxillary and four mandibular Branemark® (manufacturer: Nobel Biocare, Sweden) implants with external hexagonal connections were placed under general anesthesia. Healing screws were placed during the procedure to avoid a second surgery. Besides, a partial debridement of the jugal flanges and synechiae was performed to help mouth opening.

The patient was thoroughly followed. Good mucosal healing with no bullae formation or erosion was observed. A postoperative orthopantomogram confirmed the implants' good position (Figure 4).

Fixed prostheses, placed on the multi-unit prosthetic abutment, with very limited contact with the gingival ridge, were manufactured and positioned to prevent the formation of bullae in the oral mucosa. An orthopantomogram was taken to confirm the prosthetic abutments fit (Figure 4).

Standard impression trays were impossible to use due to the patient's anatomy. Therefore, we realized a double-mixed open impression without an impression tray. Normally, the double-mix technique corresponds to "taking" an impression with two materials of different viscosity. These two materials have different consistency. The low viscosity material is injected first on the preparations and then covered by an impression tray loaded with the high viscosity material. The first material records the details. The second placed in the impression tray exerts compression on the first injected material and records the adjacent elements' anatomy. In the current case, once the impression transfers were in place, we placed a high-viscosity silicone bead (Optosil® manufacturer: Kulzer)

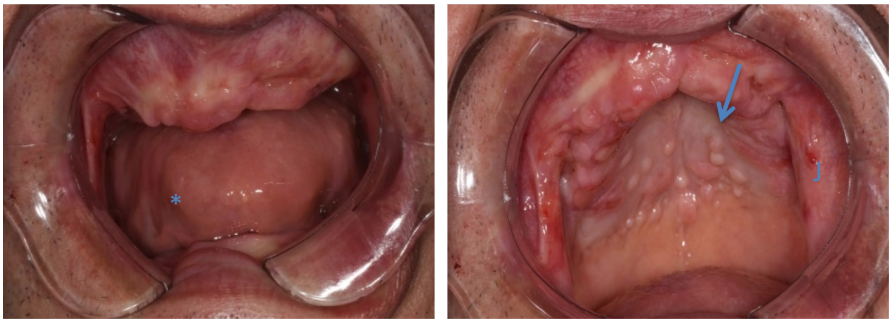

FIGURE 2 Intraoral view 1 month after the procedure. The mucosal healing is good. The presence of bullae on the palate (→), macroglossia (*), and voluminous bilateral jugal flaps (J) can be observed

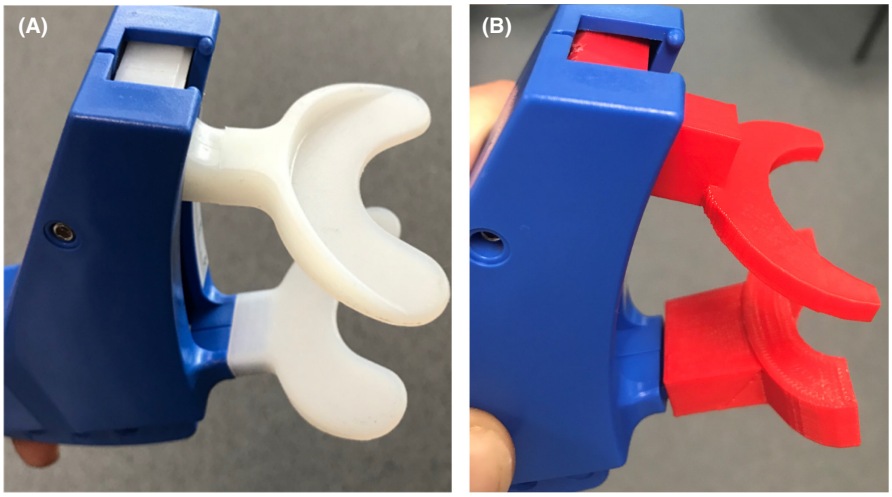

FIGURE 3 Therabite® mouth-opening exerciser: (A) original mouthpieces, (B) custom-made mouthpieces made in the Stomatology Department by computer-aided design and 3D printing

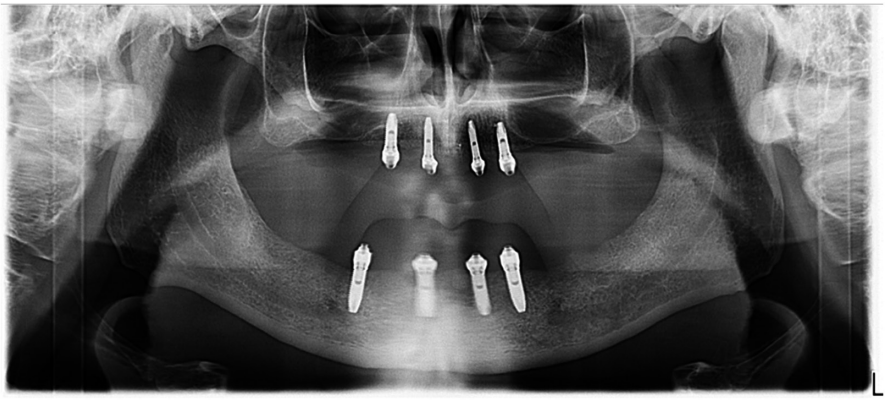

FIGURE 4 Orthopantomogram after prosthetic abutments placement

with its opening at the level of the implant transfers, leaving a space between the bead and the mucosa. We then applied a low-viscosity silicone (Aquasil light® manufacturer: Dentsply) in this space with a slight compression. In the normal double-mix technique, the two materials polymerize in a single step, whereas in our technique, the impression is made in two steps. Thereafter, we performed the different prosthetic steps following a standard procedure: occlusal rim, mock-up trial, and prosthetic trial. The prostheses were screwed onto the four maxillary and four mandibular implants (Figures 5 and 6), and an occlusion check was performed. At the recall appointment 6 months later, the patient demonstrated that he was able to keep the dentures clean and keep the oral tissues clean and healthy by gentle brushing and frequent rinsing.

3 | DISCUSSION

Epidermolysis bullosa is a rare disease, with a variety of clinical presentations depending on the type and subtype, making recommendations with high levels of

FIGURE 5 View of the patient without implant-supported dentures (left) and with implant-supported dentures (right)

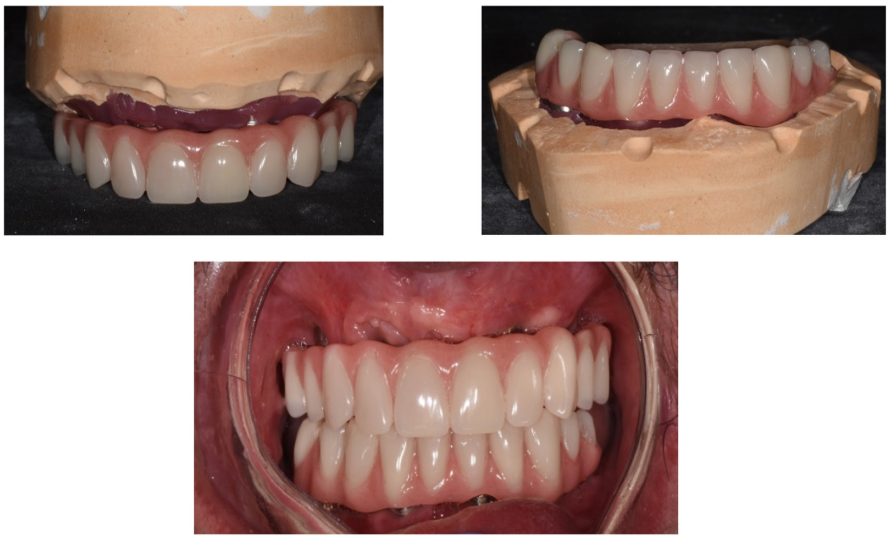

FIGURE 6 Intraoral and extraoral implant prosthesis

evidence difficult. Because of its rarity, the majority of published articles on EB are case reports. Over time, the literature has been enriched and helped guide the management of our case. A diagnosis of certainty is crucial for those patients. The search for bullous episodes in the patient's history and the physical examination of pathognomonic signs are fundamental, allowing a diagnostic hypothesis to be established. The differential diagnosis should consider infectious causes: impetigo, acute staphylococcal epidermolysis, and primary herpetic gingivostomatitis; autoimmune diseases: pemphigus, cicatricial pemphigoid, gravidic pemphigoid, linear Iga A dermatosis, acquired epidermal bullous disease; as well as erythema multiforme and Stevens–Johnson syndrome. The definitive diagnosis is obtained by genetic analysis, completed by anatomical-pathological examination of a skin biopsy.[5]

In the current case, the observed clinical signs of DEB had previously been described in the literature: mucocutaneous erosions and ulcerations, microstomia, periodontosis, and vestibular obliteration. Nevertheless, no depapillated tongue or dento-maxillary disharmony had been noticed in our patient.[6]

Epidermolysis bullosa is a multi-system disease, with a high mortality rate in some forms and numerous comorbidities. It is essential that EB patients are managed holistically, with a multidisciplinary team working in close collaboration. DEB is a severe form of EB, with many oral manifestations. An early bucco-dental follow-up facilitates the prevention and treatment of often inevitable oral decay.

Prosthetic rehabilitation using implants seems to be an interesting solution. Indeed, studies show that DEB does not contraindicate implants or prosthetic rehabilitation. In a study of 28 patients, 161 implants were placed, only two implants failed.[7] Another study of 38 dental implants showed a success rate of 97.9%.[8] This mucocutaneous disease does not affect osseointegration or bone healing, allowing a bone graft to be performed if necessary before placing an implant.[9] Although, the prosthetic space must be evaluated generally being limited by the presence of jugal synechiae. Oral opening exercises or Therabite

therapy is recommended. A vestibuloplasty, allowing a partial debridement of the synechiae, can be considered to improve this space.[10] Rubbing of the prosthesis should be avoided to prevent the formation of bubbles and erosions, which is why a fixed prosthesis on an implant is preferred. Surgical procedures must be as atraumatic as possible to preserve the mucosa.[9]

4 | CONCLUSIONS

We describe an implant-supported dento-maxillary rehabilitation of a 49-year-old patient suffering from a newly diagnosed hereditary recessive EB with disabling oral manifestations. In the current case, the diagnosis of recessive dystrophic epidermolysis bullosa has been confirmed and adequate dental care was carried out taking into account the disease constraints after implementation of the recommendations from the scientific literature. The diagnosis of DEB was fundamental not only for therapeutic stomatological management but also for identifying other manifestations of this multi-system disease in our patient. The psychological benefit to this patient was significant: the stomatological treatment relieved his pain, allowing him to regain the pleasure of eating and to find a smile; moreover, the patient received a definitive diagnosis of his disease.

AUTHOR CONTRIBUTIONS

Pierre Mestrallet MD DDS[a], Adnane Wardani DDS[b], Laurence Evrard MD DDS PhD[a]. [a]Department of Stomatology, Oral and Maxillofacial Surgery, Hôpital Erasme, Université libre de Bruxelles, Brussels, Belgium. [b]Department of Dentistry, Hôpital Erasme, Université libre de Bruxelles, Brussels, Belgium. Pierre Mestrallet contributions: contributed to the different surgical and prosthetic steps, writing, editing. Adnane Wardani contributions: contributed to the different prosthetic steps, writing, editing. Laurence Evrard contributions: supervised all the steps, editing.

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None.

CONFLICT OF INTEREST

Not declared.

DATA AVAILABILITY STATEMENT

The datasets generated during and/or analyzed during the current study are available from the corresponding author on reasonable request.

ETHICAL APPROVAL

Ethics committee Erasme hospital Approval: reference: p2021/261/NA.

CONSENT

Written informed consent was obtained from the patient to publish this report in accordance with the journal's patient consent policy.

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REFERENCES

1. Mirgaine A. Les es "uncited": bulleuses hbulleuses hL le point en; 2016;118.
2. Fine J-D, Bruckner-Tuderman L, Eady RAJ, et al. Inherited epidermolysis bullosa: updated recommendations on diagnosis and classification. *J Am Acad Dermatol*. 2014;70(6):1103-1126.
3. Wright JT, Fine J-D, Johnson LB. Oral soft tissues in hereditary epidermolysis bullosa. *Oral Surg Oral Med Oral Pathol*. 1991;71(4):440-446.
4. KräKrr S, Lucas J, Gamboa F, Peson LB. Oral soft tissues in hereditary epidermolysis bullosa. Orainical practice guidelines: Oral health care for children and adults living with epidermolysis bullosa. *Spec Care Dent Off Publ Am Assoc Hosp Dent Acad Dent Handicap. Am Soc Geriatr Dent*. 2020;40 Suppl 1:3-81.
5. Chiaverini C, Bourrat E, Mazereeuw-Hautier J, Hadj-Rabia S, Bodemer C, Lacour J-P. Hereditary epidermolysis bullosa: French national guidelines (PNDS) for diagnosis and treatment. *Ann Dermatol Venereol*. 2017;144(1):6erJ.
6. Ergun GA, Lin AN, Dannenberg AJ, Carter DM. Gastrointestinal manifestations of epidermolysis bullosa a study of 101 patients. *Medicine*. 1992;71(3):121-127.
7. Chrcanovic BR, Gomez RS. Dental implants in patients with epidermolysis bullosa: a systematic review. *Oral Maxillofac Surg*. 2019;23:389-394.
8. Oral rehabilitation with bone graft and simultaneous dental implants in a patient with epidermolysis bullosa: a clinical case report.
9. Nowak AJ. Oropharyngeal lesions and their management in epidermolysis bullosa. *Arch Dermatol*. 1988;124(5):742742i.
10. Brain JH, Paul BF, Assad DA. Periodontal plastic surgery in a dystrophic epidermolysis bullosa patient: review and case report. *J Periodontol*. 1999;70(11):1392-1396.

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