RECESSIVE DYSTROPHIC EPIDERMOLYSIS BULLOSA: A CASE REPORT

EPIDERMÓLISE BOLHOSA DISTRÓFICA RECESSIVA: RELATO DE UM CASO CLÍNICO

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RESUMO

Apresentamos um relato de caso de um menino de 6 anos que apresentava epidermólise bolhosa distrófica recessiva desde o nascimento, mas que nessa idade veio apresentando cárie e dificuldade em realizar sua higiene bucal. A epidermólise bolhosa é uma doença hereditária rara na infância, causada por mutações no gene do colágeno tipo VII, caracterizada por ser uma doença bolhosa mucocutânea incurável e, muitas vezes, fatal, apresentando manifestações bucais e sistêmicas. Foi destacado as características clínicas da doença e os cuidados odontológicos gerais realizados.

Palavras-chave: Epidermólise Bolhosa. Cuidados odontológicos. Criança.

ABSTRACT

We present a 6-year-old boy with recessive dystrophic epidermolysis bullosa, who has suffered from his birth, but at this age, he has been presenting caries and difficulty in performing his oral hygiene. Epidermolysis Bullosa is a rare hereditary disease in childhood, caused by mutations in the type VII collagen gene, incurable and often fatal mucocutaneous blistering disease with oral and systemic manifestations. We highlight the clinical features of the disease and the general dental care performed.

Key words: Epidermolysis Bullosa. Dental Care. Child.

INTRODUCTION

Epidermolysis bullosa (EB) is a rare group of genetically heterogeneous diseases, characterized by deficiencies in the adhesion of the connective tissue to the epithelium[1-3]. Junctional EB and dystrophic EB, are the most severe types. Recessive dystrophic epidermolysis bullosa (RDEB) is a hereditary disease caused by mutations in the type VII collagen gene[1-5].

Common clinical manifestations are mechanical fragility of the skin and mucosae, such as oral, pharyngeal and esophageal mucosas, with severe non-healing and painful oral wounds and with tissue shrinkage. The tongue is affected by ankyloglossia, losing the lingual papillae, and microstomia. Patients with RDEB has also predisposition to oral carcinoma, agenesis, anomalies hypoplastic tooth and alveolar bone resorption. The difficulty to hold the toothbrush due to the joined fingers and the fragility of oral tissues upon minor trauma contribute to a poor oral hygiene and the increased risk of carie and periodontal disease[2-4, 6-10].

This paper documents a rare case of a child diagnosed with RDEB, describing the clinical manifestations and the dental treatment performed as well as suggestions of dental care managements that dentists should adopt in order to provide best quality of life to these patients.

REPORT CASE

An 6-years-old boy came to the Clinic of Pediatric Dentistry for general dental care, with presence of caries and difficulty in performing his oral hygiene. He had been diagnosed with RDEB. At the time of admission, the informed consent has been obtained and the patient had anemia and he was undergoing medical treatment using cephalexin, iron supplement and mineral oil. Physical examination revealed regional ulcerations throughout the body, located at the back (Figure 1A), right knee (Figure 1B), left hand with fingers with ischemia and no nails (Figure 1C). It was also observed dystrophic right hands and upper limb with xeroderma, deformity, fibrosis between fingers (Figure 1D), and left foot with fingers without nails (Figure 1E).

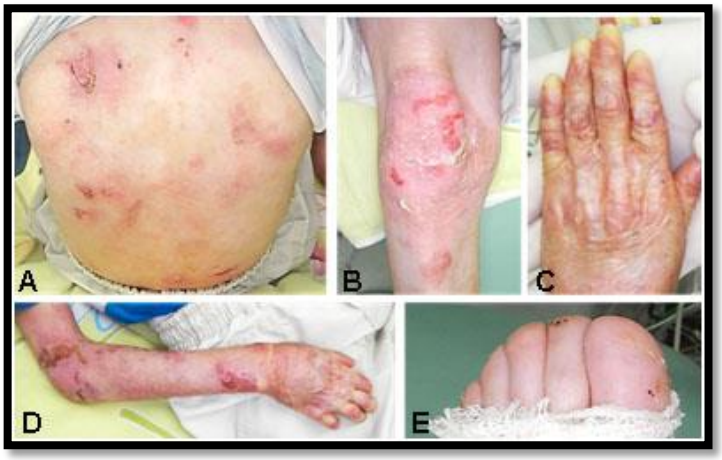

Figure 1- Physical examination (Fig. 1A. regional ulcerations located at the back, Fig. 1B. right knee, Fig. 1C. left hand with fingers with ischemia and no nails, Fig. 1D. dystrophic right hands and upper limb with xeroderma, deformity, fibrosis between fingers, and Fig. 1E. left foot with fingers without nails).

Even in the face could observe regional ulcerations located in the external pinna and neck (Figure 2A), forehead, eyelids and nose (Figure 2B). Extra oral examination showed ulcerated lesions and fibrous scars bands in some places of the lips (Figure 2C).

Intra oral examination revealed ulcerated lesions in tongue with reduction of lingual papillae, microstomia, ankyloglossia and crowding (Figure 2D). The patient also had a severe scarring of the mouth and hemorrhagic lesions extending throughout the mucosa. Poor oral hygiene leaves to six carious lesions on the primary

teeth, all asymptomatic. Oral hygiene measures were not practiced due to the ulcerated oral lesions following tooth brushing trauma and the difficulty in holding the toothbrush since her hands and fingers had become claw-like through repeated scarring.

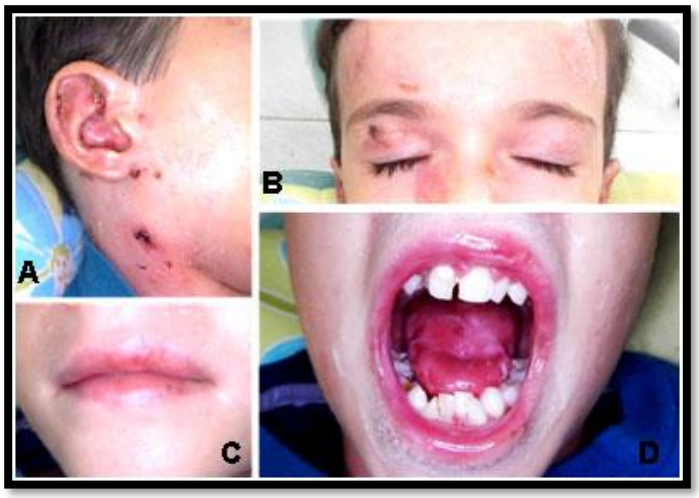

Figure 2- Extra oral (Fig. 2A. regional ulcerations located in the external pinna and neck, Fig. 2B. forehead, eyelids and nose, and Fig. 2C. ulcerated lesions and fibrous scars bands in some places of the lips) and intra oral examinations (Fig. 2D. ulcerated lesions in tongue with reduction of lingual papillae, microstomia, ankyloglossia and crowding).

The preoperative care included a panoramic radiograph, that confirmed the presence of caries, but no bone pathology was observed. Three days before the next appointment, an extra-soft toothbrush tuft single was indicated, artificial saliva three times a day was prescribed; and one day before the appointment, chlorhexidine mouthwash (0.12%) twice a day and Bepantol (Bayer HealthCare, Berlin, Germany) for lips three times a day were also prescribed.

The transoperative care included the use of mattress to reduce friction of the body with dental chair and minimize local temperature, it was also applied of Bepantol (Bayer HealthCare, Berlin, Germany) in lips and perioral area before the restorative procedures, that were performed with atraumatic restorative treatment without anesthesia (Figure 3A) with ionomer cement and noninvasive sealants with fluoride (Figure 3B). During the treatment was given him a break, and a popsicle was given to the patient in order to reduce the sensitivity of the mucosa (Figure 3C) to finish the dental treatment (Figure 3D). The goals of this phase of the therapy were mainly to provide the patient with an effective psychological conditioning, motivate and stimulate him commitment with the treatment.

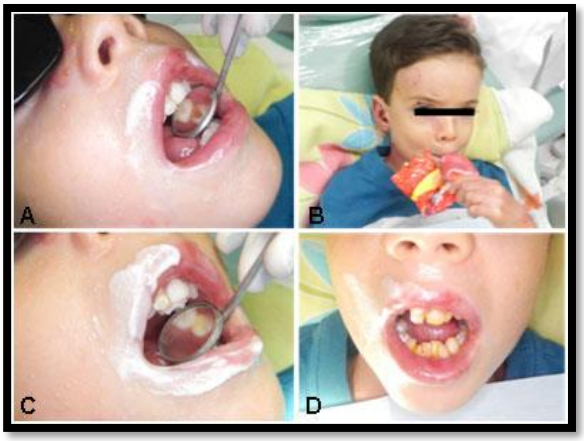

Figure 3. Intra oral examination (Fig. 3A. before the restorative procedures, Fig. 3B. atraumatic restorative treatment without anesthesia with ionomer cement, and noninvasive sealants with fluoride, Fig. 3C. break with popsicle in order to reduce the sensitivity of the mucosa, Fig. 3D. the end of the dental treatment.

Postoperative care included: topical buccal dexamethasone (0.1 mg/ml - 2 times a day for 15 days); nystatin (100,000 IU – 3 times a day, during 15 days); gel for dry mouth moisturizing (3-5 times per day, constant use); artificial saliva solution (3 times a day, constant use); solution of sodium fluoride 0.05% (03 times daily,

constant use). The patient was following after 15 days showed a significant plaque reduction and intraoral lesions. Anemia and micronutrient measure was investigated and the patient is following with good dental hygiene and weight gain. The patient is currently being followed up, with visits every 6 months.

DISCUSSION

Collagen VII is the main constituent of the anchoring fibrils, important adhesive structures that attach the epidermis to the dermal extracellular matrix. Two disorders are caused by dysfunction of collagen VII, both characterized by skin and mucosa fragility, epidermolysis bullosa acquired and dystrophic epidermolysis bullosa[1-5]. Epidermolysis bullosa acquired and dystrophic epidermolysis bullosa share high clinical similarities with significant difference in patients' age of onset and pathogenesis[1,3,6-11]. Our patient presented with severe RDEB, microstomia leading to limited oral opening, presence of caries, malnutrition and multiple ulcerated and healing lesions. Because of that, the routine dental care was necessary and important and we described the clinical manifestations and the dental treatment performed as well as suggestions of dental care management that dentists could be adopt in order to provide the effective and the best dental treatment to these patients.

Individuals with the RDEB typically have extreme fragility of their oral and perioral mucosa, and ulcerations can affect all areas of the oral mucosa, including the tongue[2,4]. The continuous process of blister formation and healing with scarring results in marked changes in the oral architecture increasing discomfort during feeding, reducing the food intake, resulting often in anemia[11-13], as this case. The soft tissues defining the oral opening fail to grown normally due to scarring, resulting in a typically markedly restricted oral aperture[2,4,11-13]. Thus, the use of topical corticosteroids or systemic to the symptomatic treatment of soft tissue lesions, plus oral lubricate substances can prevent to appearance new bullous due stimulation of salivary flow and reduction of intraoral friction[12-13]. However, it is extremely important that contact between these patients and the dentist earliest as possible to adequate prevention of oral lesions and caries, and to submit the patient to less invasive dental procedures and with lower cost. So, the dentist is important link in the quality of life of these patients[2,4,12-13].

The clinical and radiographic findings provided evidence that this child was at caries activity. The soft consistency-high-sucrose diet, the difficulty to hold the toothbrush due to the joined fingers and the fragility of oral tissues upon minor trauma contribute to a poor oral hygiene and the increased risk of carie[8,12-13]. Instructions on toothbrushing and dietary habits were emphasized at each treatment appointment in this case. The treatment strategies included the adoption of preventive measures for adequacy of the oral conditions, which comprised elementary instruction on oral health care, diet counseling and improvement of oral care skills by means of supervised toothbrushing and training of the parents on toothbrushing and flossing. The gentle use of a soft baby-size toothbrush was encouraged as an effort to minimize the mucosa's trauma[2,4,8,12-14]. Restorations should carefully adapted and highly polish; secondary infections should prevent with the use of oral antiseptic; and the

administration of local anesthesia should avoid whenever possible, because may cause blister formation[8,12,14-15].

To minimize discomfort for the patient during the dentistry care, inpatient care teams should understand these considerations and modify the care plan accordingly as given him a break and a popsicle in order to reduce the sensitivity of the mucosa. Prior preparation by the inpatient care team will facilitate the dentistry treatment of safe and effective care and greatly improve the overall patient experience[2,4,8,11-15].

CONCLUSÃO

In despite of patients with RDEB needs to be assist by a multidisciplinary team and requires extreme care professional, the dentist should be prepared to recognize the oral needs of these patients to assist in early diagnosis, providing prevention and rehabilitation procedures of these patients of adequate form, avoiding trauma and ulceration[8,12,14-15]. Because of that, our case could help dentists outlining the best possible dental treatment to minimize the possible complications resulting from dental treatment and to provide the effective dental treatment to these patients.

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