Dystrophic epidermolysis bullosa in a child

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Abstract

Epidermolysis Bullosa (EB) is a form of severe skin adhesion defect due to the disruption of the dermal- epidermal junction. It is classified into simplex and dystrophic forms depending on the level at which the junction is compromised. Repeated ulcerations and bullae formation in the mouth lead to scarring that brings about various changes in the oral cavity. These include loss of sulcular depth, ankyloglossia, limited mouth opening and other dentoalveolar changes. At present while there is no cure for EB, the therapeutic approaches are essentially aimed at controlling the infections and maintaining an acceptable quality of life. Dental management should aim at maintaining a functional dentition that would help in mastication and favour nutrition. Oral manifestations and dental management in a child diagnosed with dystrophic EB since birth are presented here.

Keywords: Blistering, dystrophic epidermolysis bullosa, epidermolysis bullosa

Introduction

Epidermolysis bullosa (EB) comprises a group of genetically determined skin fragility disorders characterized by blistering of the skin and mucosa following mild mechanical trauma. Dystrophic Epidermolysis Bullosa (DEB) is a subtype of EB with a well understood pathogenesis. The main presenting feature of DEB is trauma induced blisters and healing with scarring. The dystrophic forms of EB are characterized by deformities of the skin including coalescence of the fingers, nail changes and milia formation, and have either autosomal recessive or dominant inheritance.[1] Prevalence of DEB is not known precisely though it is found to occur in all races worldwide with equal predilection in both the genders.[2] In the recent past, investigators have discovered the molecular basis for all of the major inherited blistering disorders.[3]

Case Report

A seven year old male patient was referred to the Pedodontic clinic for oral examination by the Paediatric Dermatologist. Parents reported that the child had been diagnosed with EB since birth when he developed a blister on the tongue during nursing. No significant family history or consanguinity was reported. The child's milestones were normal. Parents reported that due the blisters in the mouth they avoided brushing his teeth and had started brushing his teeth at six and half years of age. When presented to our department, the child had been brushing only in the morning with a soft brush and herbal toothpaste. Diet of the child was mainly vegetarian, non spicy and broken into smaller bite sizes for ease of eating.

General clinical examination revealed a small and thinly built child with normal speech. Both hands had pseudosyndactyly, that is, mitten deformity of the hands and the finger nails were missing [Figure 1]. While the skin was dry and wrinkled on arms, neck, ear, the face had scabs and the scalp showed alopecia [Figure 2]. Microstomia was observed [Figure 3]. Buccal mucosa and palate appeared greyish white and ankyloglossia with erythematous patches was present [Figure 3]. Lingual sulcus was reduced in depth, while labial sulci and buccal vestibule were absent [Figure 4]. Both upper and lower labial frenum were absent. All deciduous teeth were present and all first permanent molars had erupted. The dentition was non-spaced with retroclined lower anteriors, and anterior deep bite. Dental caries was present in relation to 51, 52, 61 and 54. Skin biopsy was done and sent for histopathological and immunopathological examination. Histopathology confirmed the diagnosis of EB while immunopathology report confirmed the diagnosis of DEB stating that collagen band VII was absent at basement membrane zone (BMZ) ; and, laminin band V was present and keratin 14 showed a normal pattern. Barium meal radiograph revealed the presence of an esophageal stricture. During the first visit, oral prophylaxis was completed followed by fluoride varnish application. Parents were counselled on maintenance of the oral hygiene and avoiding cariogenic food. Carious teeth were restored in the subsequent visit.

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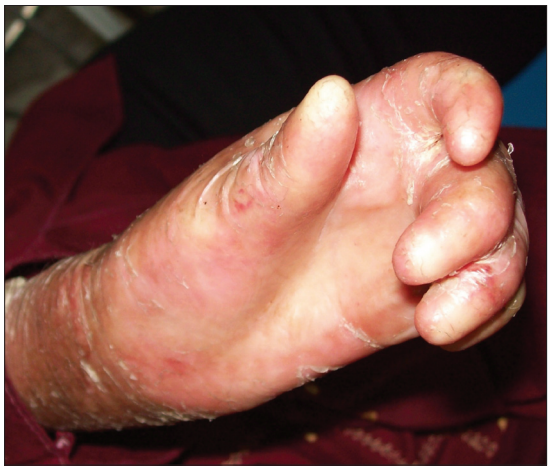

Figure 1: Pseudosyndactyly and absent finger nails seen in the patient in the case report

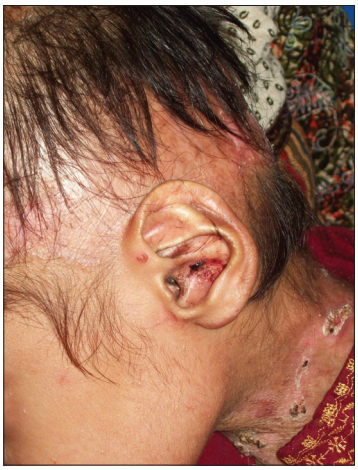

Figure 2: Alopecia and scabs on neck seen in the patient in the case report

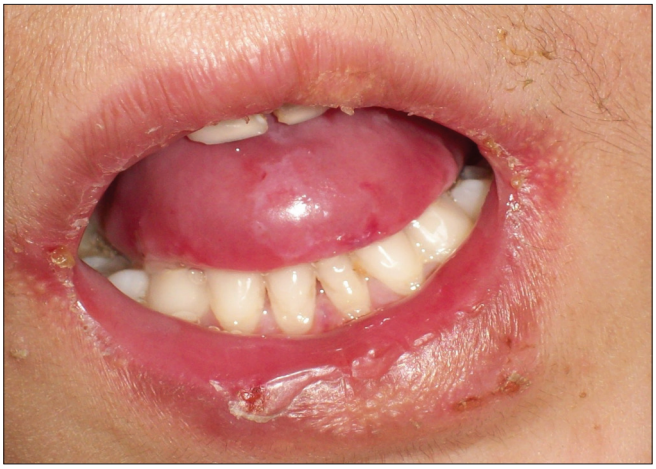

Figure 3: Limited mouth opening, ankyloglossia, retroclined lower anterior teeth seen in the patient in the case report

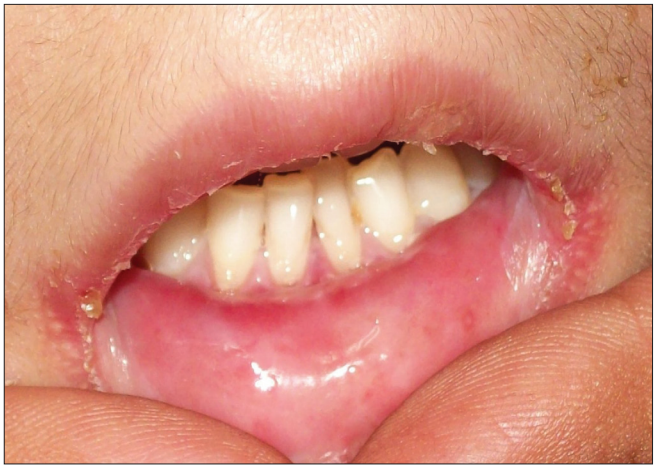

Figure 4: Obliterated labial sulcus seen in the patient in the case report

Patient was advised using a 0.05% sodium fluoride mouthwash and was placed on a recall every six months.

Discussion

Abnormalities in collagen VII form the molecular basis for DEB. In DEB skin, the collagen VII is either reduced or absent, and electron microscopy reveals paucity, rudimentary structure or complete lack of the anchoring fibrils. Several hundred mutations in the gene for collagen VII, COL*7A1*, have been noted in both the recessive and dominant forms of DEB. Since all DEB subtypes are caused by mutations in the COL*7A1* gene, DEB is classified as Severe generalised recessive DEB, Recessive DEB, and dominant DEB.[4] Severe generalised recessive DEB is the most severe form, previously known as Hallopeau-Siemens subtype, and starts at birth with extreme fragility of skin. Mitten deformities are seen in the hands and feet due to excessive scarring. Dental abnormalities are common including dystrophic teeth and severe caries. There is a high risk of developing squamous cell carcinoma. In recessive DEB, blistering is less severe and dental problems are less extensive. Mutilating deformities are not seen. Dominant DEB is milder, starts at birth though it decreases in intensity with advancing age. Teeth are normal in this form.[2]

In the present case, repeated blistering and ulceration which healed with scarring, lead to microstomia. The long standing nature of the disease in this case had lead to the elimination of the buccal and vestibular sulci, marked tongue atrophy and ankyloglossia. In children suffering from DEB, scarring of the fingers and hands poses a problem in maintaining the oral hygiene. Dental caries is common and may be mild or rampant. It has been found in studies that in terms of enamel chemistry, teeth from patients with recessive dystrophic EB are essentially the same as those from healthy individuals containing > 90% mineral per volume, thereby ruling out enamel hypoplasia in these teeth.[5] Therefore, the disease does not predispose the tooth in any way to carious attack. Poor oral hygiene, and diet rich in sugar and carbohydrates, which is necessary to provide sufficient caloric intake to maintain the growth, are the two

most important etiological factors in high caries experience. It has also been reported that patients with recessive EBD have elevated levels of albumin in the saliva predisposing them to high rate of dental caries.[6] Parents' indulgence, a common characteristic in cases like this in which an actual life-threatening condition is present, has been found to contribute significantly to poor oral status.[7]

Oesophageal strictures in DEB contribute to nutritional problems like limited intake of food, protein loss and impaired growth.[2] The combined effects of malnutrition and scarring cause a marked inhibition of facial growth. This contributes significantly to the marked dento-alveolar disproportion and consequently to dental crowding.[8] Oral care aims at maintaining a functional dentition that would help in efficient mastication, and thereby favour nutrition of the patient, and involves continuous follow-up of dietary advice, oral hygiene maintenance and fluoride therapy. The treatment for patients with EB is multidisciplinary. Secondary infections must be treated with topical and/or systemic antibiotics and a protein-rich diet with iron and zinc supplements must be provided. Systemic treatment remains primarily palliative and dental treatment is aimed at avoiding the formation of new bullae during perioperative management.[7]

Treatment of these patients is planned out in phases. Oral hygiene, dietary instructions and elimination of gingival inflammation form the first phase; restorative procedures form the second phase; and, extractions of the carious destroyed teeth, and recall system, form the third and fourth phase of the treatment, respectively.[9] Caries prevention in individuals involves modification of toothbrushing techniques, diet modification and adjuvant therapies. Soft and small head toothbrush with short bristles should be recommended in case of severe microstomia. Parental help in toothbrushing is required. Adjuvant therapies like the use of fluoride varnish and chlorhexidine have been recommended. Since these individuals can be sensitive to flavouring agents, neutral, non-flavored and alcohol free formulations should be advised. Since diet is a key factor in causing caries a thoughtful dietary plan should be made with the help of a nutritionist. Purée diets are recommended because of the lesions involving the oral mucosa and oesophagus. Increased fluid intake while eating and rinsing with warm water would help in oral clearance.[10]

Limited mouth opening poses a problem in dental treatment. However, there are no contraindications for restorations, endodontics, and placement of stainless steel crowns. Care must be taken when planning removable dentures where the tolerance depends on the mucosal fragility of each patient.[10] General precautions like lubricating the lips, buccal mucosa and instruments with Vaseline; and, placing the suction tip on tooth surface, can help prevent bullae formation during the course of the dental treatment.

Preventive programs have shown to be effective in preventing dental caries in individuals with DEB. Restoration of decayed teeth is essential for functional dentition. Dental management of patients with DEB is an important part of the multidisciplinary approach in the care of these patients.

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