Dystrophic epidermolysis bullosa associated with non-syndromic hypodontia

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ABSTRACT

Epidermolysis bullosa (EB) is a genetic disease associated with fragility and bullous lesions of the skin and mucous membranes. There are various patterns of inheritance and histopathology. The disease is associated with systemic and oral manifestations, among which may be dental decay necessitating oral rehabilitation. The aim of this article is to present the course of the condition in a child with dystrophic EB and also to report an association between EB, hypodontia, and supernumerary teeth which has not been reported earlier in literature.

Key words: Vesiculobullous lesions, dystrophic epidermolysis bullosa, oligodontia, microstomia, ankyloglossia

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INTRODUCTION

Epidermolysis bullosa (EB) is a wide spectrum of rare genetic disorders characterized by marked fragility of the skin and mucous membranes in which vesiculobullous lesions occur in response to trauma, heat, or no apparent cause. EB was first reported as a separate entity by von Hebra in 1870 and it affects 1 of 50,000 births. Lesions usually start to appear at birth or within the first 6 months of life.[1,2]

CASE REPORT

A12-year-old boy, a product of consanguineous marriage, was referred from the Department of Dermatology, Civil Hospital for favor of dental check-up. Patient was apparently alright at the time of birth. Patient developed vesicles and bullae 6 days after birth, over the face, scalp, trunk, and extremities, with loss of finger and toenails and inability to open the mouth wide. Oral hygiene was not practiced due to the formation of bullae following toothbrush trauma. The patient complained of toothache with food impaction in lower left posterior teeth region. History of pain, getting aggravated upon mastication and relieved upon medication was elicited from the patient. Pain was dull, intermittent, and non-radiating in nature. His diet was limited to soft or pureed foods. He had normal cognitive function and attended school. No allergies were reported.

Dermatological Examination revealed patchy alopecia over the scalp. Hyperkeratosis of palms and soles was seen. Sclerosis of fingers was present with fixed flexion deformity at distal interphalangeal joints and "claw-like" hands [Figure 1]. Fingernails are all intact except left thumbnail. Complete loss of toenails was seen [Figure 2]. Skin examination revealed generalized scarring with hypopigmented and hyperpigmented areas over the chest, back, and trunk. Nikolsky's sign was found to be positive in ankle region in the patient. Intraoral examination revealed healed lesions over the lower labial and lingual mucosa in canine-premolar region. White patch was seen over the right retromolar region [Figure 3] with the overlying skin giving a collapsed/shrunken/shriveled appearance, suggestive of a freshly ruptured bulla. Oral hygiene was very poor and generalized gingival inflammation was seen [Figure 4]. He had a mixed dentition with severe occlusal carious lesions in first premolars and upper second molars. Both maxillary and mandibular lateral incisors and premolars were clinically missing. Detailed intraoral examination was not possible due to limited mouth opening caused by scarring of the lesions.

INVESTIGATIONS

Orthopantomogram [Figure 5] reveals missing maxillary and mandibular lateral incisors and second premolars. It also revealed caries in lower

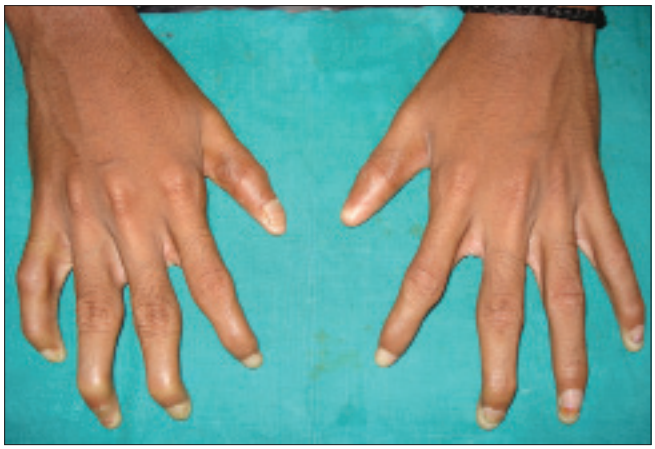

Figure 1: Sclerosis with fixed flexion deformity of distal Interphalangeal joints giving a claw-like appearance

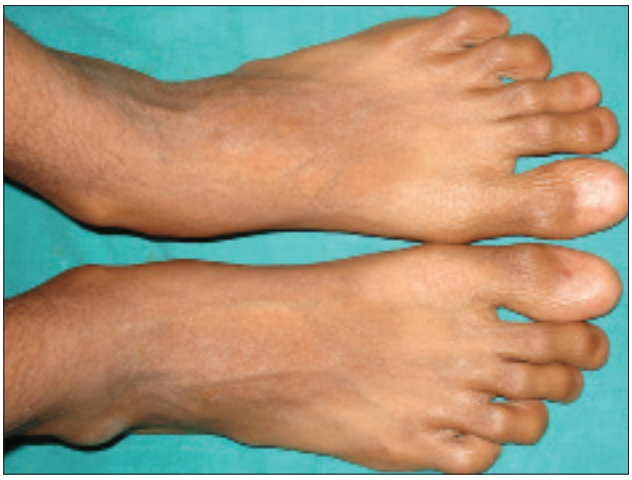

Figure 2: Loss of toenails since childhood

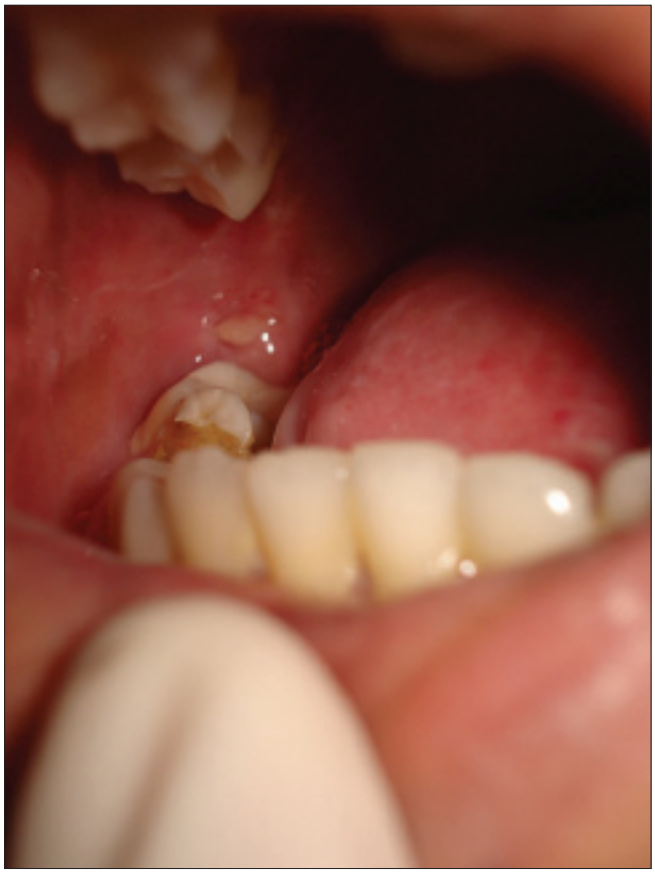

Figure 3: White patch seen over the right retromolar region suggestive of a freshly ruptured bulla

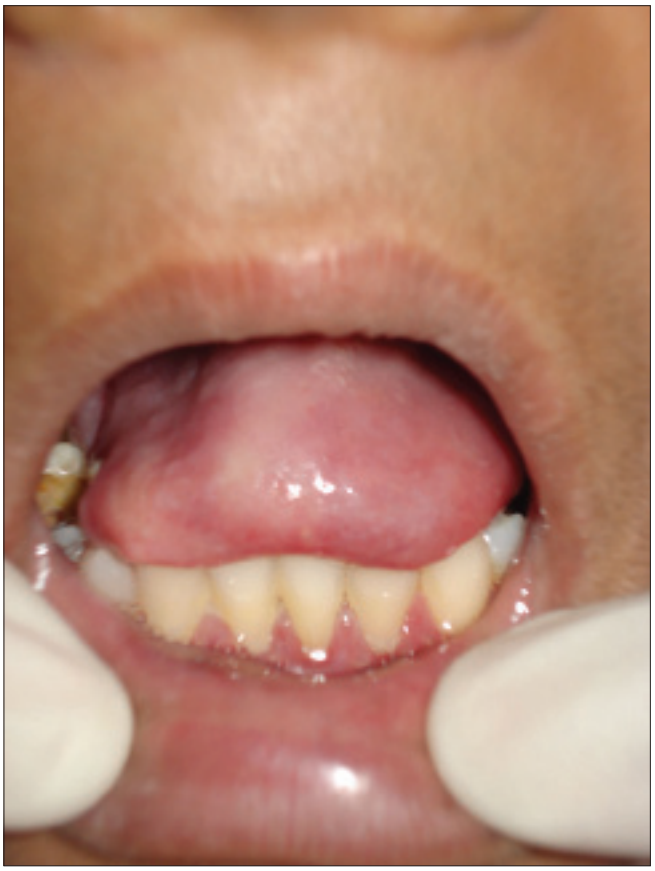

Figure 4: Gingival inflammation seen in free gingiva in lower anterior region with ankyloglossia and microstomia

right and left first molars and in upper left central incisor. Grossly decayed upper left deciduous second molar and a retained carious deciduous lower left second molar were present.

Routine blood examination and abdominal ultrasound revealed no abnormality. Based upon the history and general systemic examination, a diagnosis of EB of recessive dystrophic type was seen.

COURSE OF CONDITION

Due to the extent of his restorative needs, limited mouth opening, and poor level of cooperation, it was decided to render treatment in the operating room under general anesthesia, but the parents were not willing for the same and the patient was placed on antibiotic regimen and instructed to rinse the mouth with water after eating, and apply chlorhexidine with a cotton

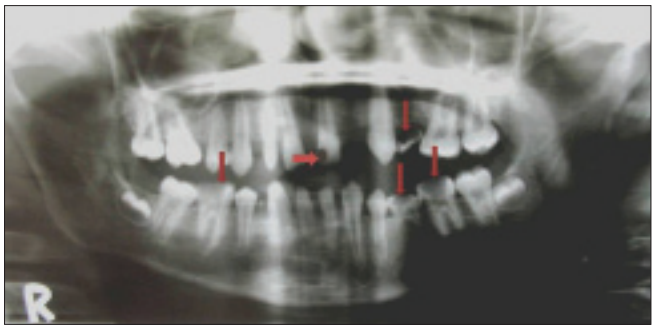

Figure 5: Orthopantomogram depicting congenitally missing lateral incisors and second premolars and multiple carious teeth

swab twice daily. Dental care through plaque control and careful prophylaxis was advised.

DISCUSSION

The case presented here illustrates many features that are characteristic of generalized recessive dystrophic EB (DEB). Having unaffected, consanguineous parents suggest an autosomal-recessive inheritance form of DEB.[1] No syndromes have been identified within the family. The usual mode of inheritance in non-syndromic hypodontia is autosomal-dominant, but autosomal-recessive inheritance, X-linked, and polygenic or multifactorial models of inheritance have been reported.[3] The clinical presentation and family history appear to be consistent with autosomal–dominant Hypodontia, which is quite similar to previously reported conditions associated with mutations within the *PAX 9* gene but further genetic testing is required for confirmation. Matalova *et al*. concluded in their study that the molecular basis of the defect is not completely understood, despite identification of several mutations in *MSX1* and *PAX 9* genes that seem to be crucial for tooth agenesis and mutations in the *AXIN2* gene that cause oligodontia together with a predisposition to colorectal cancer.[3]

More than 20 distinct variants of EB have been identified and classified according to the phenotypic characteristics, mode of inheritance, and the ultrastructural level at which the blisters occur. These have further been classified into three major subgroups based on specific level of tissue cleavage.[4]

1. EB simplex is usually dominantly inherited with cytolysis of the basal layer of the epidermis and is characterized by intraepidermal blistering with relatively mild blistering of the skin and mucous membranes. Lesions typically heal without scarring
2. Junctional EB is recessively inherited and lesions occur in the lamina lucida or epidermal–dermal interface with variable hemidesmosomal abnormalities
3. DEB is inherited in a dominant as well as recessive form and is usually associated with cleavage in the sublamina lucida plane. There is excessive collagenolysis resulting in reduced or absent collagen VII.

Non-syndromic hypodontia is a common term, including different phenotypes ranging from hypodontia of one tooth (excluding third molars) to oligodontia and anodontia. The usual mode of inheritance is autosomal-dominant, but autosomal-recessive inheritance, X-linked, and polygenic or multifactorial models of inheritance have been reported.[5]

ORAL MANIFESTATIONS

The oral involvement of EB, in its mildest form, includes intraoral blistering that heals rapidly, while some patients may present with severe intraoral blistering and subsequent scar formation, causing microstomia and even perioral carcinomas. Additional oral manifestations include milia, lingual papilla atrophy, caries, enamel hypoplasia, and rapid attrition of teeth. The recessive form of DEB has the most significant oral lesions such as microstomia, ankyloglossia, and rampant caries.[1,2] Defective enamel, poor oral clearance of foods, and inability to achieve adequate oral hygiene may lead to rampant decay.[1,4]

DERMATOLOGICAL MANIFESTATIONS

Cutaneous blistering is the most common dermatological finding which may include corneal erosions, scarring alopecia, erosions in tracheal epithelium, esophageal strictures, deformities of the hands and feet, loss of nail beds, congenital pyloric atresia, and late onset muscular dystrophy. Energy demands are increased along with the need for increased caloric intake; failure to achieve this may lead to premature death during infancy.[4,6]

TREATMENT

There is no specific regimen for managing EB; however, much emphasis has been placed on supportive measures. The comprehensive oral care of children with DEB can be difficult because of the severe limitations such as limited mouth opening, formation of intraoral bullae, and mucosal sloughing with trauma from hard foods and tooth brushing and difficulty in deglutition, imposed by the condition. This can impact not only dental management but all other specialties as well. These children are often on hypercaloric formulas, which, combined with poor oral hygiene and swallowing issues, results in high caries rates. The development of scarring and hand contractures further complicates oral hygiene issues. Thus, prevention is crucial. Dentists play a central role in early intervention. Patients may require frequent follow-up for cleaning and topical fluoride application. Home-care regimen

should include brushing with a soft-bristled toothbrush and regular use of non-alcohol-based fluoride rinses.

Further evaluations using enzymatic analysis, electron microscopy, immunofluorescence, and immunohistochemistry are necessary to confirm the diagnosis of EB and characterize the histological type. Since genetic tests are now available, these are often subsequently used to determine the exact mutation.

CONCLUSION

- A rare case of DEB associated with hypodontia and supernumerary teeth that required extensive dental treatment is presented
- Early intervention with a focus on prevention, strict home-care regimen, dietary counseling, and frequent follow-up with oral health provider, is critical for optimal oral health
- A review of oral care considerations in management of DEB is presented.

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