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Dystrophic Epidermolysis Bullosa: Rare Case Reports with Review of Literature.

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Abstract:
Epidermolysis Bullosa (EB) constitutes a group of rare genetically transmitted diseases with several methods of inheritance and various degrees of severity and presentation. It is an autoimmune disease characterized by the presence of IgM and IgG antibodies at the level of basement membrane zone. It is an inherited autosomal dominant or recessive disease with incidence of 8-10 per million births. It is equally prevalent in both sexes and occurs in all racial & ethnic groups. It usually presents at birth with various subtypes (simplex, junctional, dystrophic, Kindler syndrome). Over 34 different forms of EB have been identified. The prominent clinical characteristic is blistering & erosions of the skin and oral mucous membrane. This paper documents a case of 2siblings diagnosed with EB. The patients presented with both new & old skin and oral lesions following minor trauma, owing to the fragile skin & mucosa. The most common oral manifestations are painful blisters affecting all the mucosal surfaces. These lesions tend to heal with scar formation in the mouth, mucosa & conjunctiva. Microstomia, ankyloglossia, atrophy of tongue, obliteration of buccal & vestibular sulci, were also noticed. Multiple white spots called milia were seen on both ears. The nails had become severely dystrophic and were lost. The purpose of this case report & review of literature is to emphasize on the importance of proper diagnosis based on clinical observation & histopathological investigations, which will aid in formulating treatment outline, following various precautionary measures inorder to improve health.

Key Words: *Blisters, Erosions, Fragile skin, Milia, Dystrophic nails.*

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I. Introduction:

EB is a rare genetic disorder. It is characterized by blisters & erosions on the skin & oral mucosa. The cause may be due to altered enzyme activity & collagen destruction. [1] The pathogenesis is related to contributing defect in the epithelium & connective tissue. Due to extreme fragility of skin & mucous membrane there appears blister formation following minor trauma.[2] The 4 major forms of EB are simplex, junctional, dystrophic- autosomal dominant & autosomal recessive, kindler syndrome etc.[1,6] [Fig. 1] EB usually presents at birth/ during 1st year of life & lesions heal with scar formation. Oral manifestations vary in involvement & severity according to the different forms.[1] Intraoral manifestations include both new & old lesions on labial and buccal mucosa with fibrosis &lesions on anterior palate, multiple carious lesions, restricted mouth opening, ankyloglossia , gingival inflammation & gingival recession.[1,3,4] Extra orally lesions may appear on skin, ear, conjunctiva, nails. Milia formation is also evident. Healing is with scarring & hypopigmentation of areas with such lesions. Diagnosis was based upon clinical history & immunofluorescence mapping which showed lgG & lgM deposition at the level of basement membrane zone. Other diseases with similar presentation & which occurs since birth include epidermolytic ichthyosis, incontinentia pigmenti, neonatal pemphigus & staphylococcal scalded skin syndrome. It is important to distinguish EB from these diseases. [1] The following case report describes the clinical presentation of 2patients with EB. The aim was to diagnose the patient based on clinical & oral manifestations & immunofluorescence mapping which helps in framing an optimal treatment plan. The major dental complications are increased risk of dental caries and formation of new lesions.[5] Dental treatment is always aimed at avoiding the formation of new bullae.[5] It involves the use of soft toothbrushes with modifications in brushing technique and fluoride therapy.[4,5] Soft diet, iron, vitamins and dietary

supplements play a role in maintaining proper oral health. This article also discusses the management of patients with EB.

II. Case Report:

A 19 year old boy and 20 year old girl reported to the department of oral medicine and radiology for treatment with chief complaint of reduced mouth opening and ulcers on palate and buccal mucosa. As reported by the parents, ulcers, blisters and vesicles on skin and oral mucosa started in the female patient 2 hours after birth whereas the same features were noticed in the male patient 2 days after birth. No similar history was previously depicted in the family. On extraoral examination, multiple blisters and vesicles were seen all over the body with involvement of hands, feet, elbow, knees, trunk, scalp and eyes in both the patients.[Fig. 2.b, 4.a] Healed scars with hypopigmentation were also seen in previously involved areas of skin.[Fig. 2.a,b; 4,b] The nails were dystrophic and were indistinguishable. Face was not severely involved and white milky spots called milia were seen on ears and hands. [Fig. 2.c] Intraoral examination revealed ulcers on palate, buccal mucosa and gingiva. [Fig. 5.a] Previous oral lesions healed with scar formation and it resulted in the formation of Microstomia. [Fig. 3.a] Tongue movement was also limited because of ankyloglossia. Both the patients had poor oral hygiene with thick plaque deposition. Gingiva was red and edematous. [Fig. 3.b] Both the patients complained of dysphagia and burning sensation in mouth on intake of food. This could be related to recurrent esophageal lesions that healed with scarring. No abnormalities were detected on systemic examination. OPG revealed no major abnormality except root stumps with 26 and severely decayed tooth with 16. Informed consent were received from the patients for histopathological investigations. Histopathological examination of biopsy specimen showed hyperkeratosis, mild acanthosis, intact basal cells and sub-basal cleft. [Fig. 6] Skin and oral biopsy was taken which on immunofluorescence mapping (IFM) confirmed the diagnosis of DEB. Type VII collagen was not detected in the skin of patients on IFM. Deposition of IgG and IgM antibodies were seen at the level of basement membrane zone. The goal of treatment in this case was to prevent formation of new lesions and their resulting complications. Supportive measures in the form of steroids and antibiotics were used to prevent secondary infection and promote healing of lesions. These patients also had increased risk of caries development because of poor oral hygiene. [Fig. 5.b.] Both the patients were given oral hygiene instructions. Dental plaque was controlled with supragingival scaling. With minimal intervention such as frequent topical fluoride application and use of soft bristle tooth brushes, risk of caries development was prevented to a larger extent. Decayed teeth were restored without analgesia and with all precautionary measures. Lubricants were used to protect commissures and intraoral soft tissues. Removal of root stumps was important as its sequel would be difficult to manage. Extraction of root stumps with maxillary 1st molar was carried out under local anesthesia without any complications. To avoid trauma to the fragile mucosa, necessary precautions were followed including use of gentle pressure during gingival reflection. Patient is on follow-up every 3 months to avoid complex treatments. Patient was also referred to dermatologist for treatment of skin lesions.

III. Discussion:

EB is a rare genetic disorder. It is characterized by blisters and erosions on the skin & oral mucosa. The four major forms of EB are: Simplex, Junctional, Dystrophic- autosomal dominant & recessive and kindler syndrome. [1,6] The incidence is approximately 8-10 per million births.[6] Oral manifestations vary according to the subtype in severity & frequency.[1] Although it is reported that it is equally prevalent in both the sexes[2,4,6] lesions started appearing earlier in female patient in our case. Also the lesions were found to be more severe in female patient. Dental management was not very simple. [5] This findings were in relation with the findings reported by Thais M. oliveira et al in his study. [5] However no relation between severity and genetic makeup (association with X chromosome) has been reported till date. In dystrophic type of EB, malformed teeth with early development of dental caries and gingival inflammation due to plaque accumulation is usually present. [1, 2] These features were also seen in our case. The patients presented with extensive dental caries & poor oral hygiene. It was also reported by Wright et al in his study that there is an increase in susceptibility of dental caries in patients with EB.[5,2] The different reasons can be cariogenic diet rich in sucrose or difficulty in plaque removal with tooth brush. A delayed dental maturity based on demirjian's system was present in our patients. The same was also reported by katayoun Esfahanizadeh et al in his study. Intra oral examination revealed ulcers on palate, buccal mucosa & gingiva, obliteration of buccal vestibule & ankyloglossia.[1,3,4] Oral lesions healed with scar formation and Microstomia was also revealed. These findings were consistent with the findings reported by Parushetti et al in his study.[1] Based upon the clinical findings we ruled out the possibility of pemphigus and mucous membrane pemphigoid as fragile skin, nail dystrophy, milia and scarring are characteristics of EB.[1] The treatment goals were to minimize the risk of trauma in oral cavity, prevent blister formation & future complications.[1] As oral ulceration during dental treatment is unavoidable it was prevented by lubricating oral mucosa with hydrocortisone cream, petroleum jelly and triamcinolone before starting any dental procedure.[1,2]To reduce discomfort due to blisters, Magic mouthwash was recommended to be applied on

oral mucosa. Magic mouthwash contains sucralfate, tetracycline, lidocaine that promotes formation of protective layer against any trauma and favors re-epithelialization of the damaged mucosa. For multiple carious lesions & increased risk of dental caries- oral hygiene instructions, increased fluid intake, water jet systems, soft toothbrushes and fluoride mouth rinses were recommended.[4,5] Topical application of neutral sodium fluoride & fluoride varnishes were effective. It is important to preserve the natural dentition in patients with EB as prosthetic rehabilitation is complicated and retention is difficult because of scarring of oral tissue due to trauma or distortion.[1,2] Tooth extraction was recommended in our case because of extensive caries and defects of enamel on teeth. Blister formation may also occur during administration of local anesthesia (LA) during tooth extraction. So, care was taken to deposit LA slowly & deep into the tissues to avoid such complications. Also use of small sized instruments, short shaft dental burs & handpieces with small size head were indicated.[2] Topical antibiotics & oral antiseptics were prescribed to promote healing & prevent secondary infections.[1,2,5] Advanced treatment modalities include gene & cell therapy, intradermal injection of allogenic fibroblasts, bone marrow transplantation, stem cell transplantation and recombinant proteins infusions (Type 7 collagen in wounded skin).[2] We hope that this article would be helpful with relevant clinical aspects & typical oral findings in patients with EB so as to provide excellent dental treatment. This information could also be used in diagnosis of patient in clinic, their management& in research work.

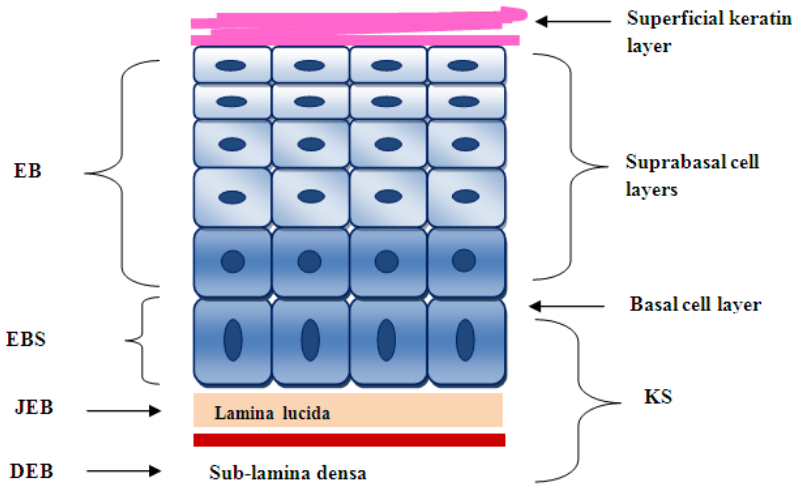


Fig. 1. Diagram showing different types of EB depending on the cell layer involved.

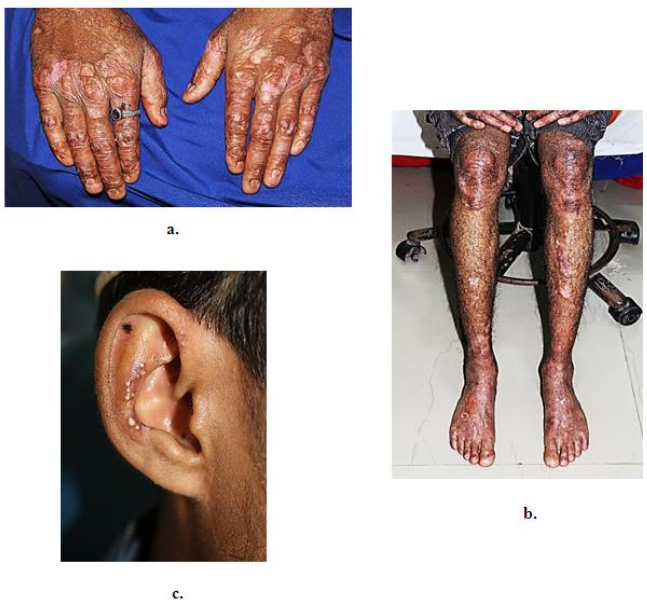
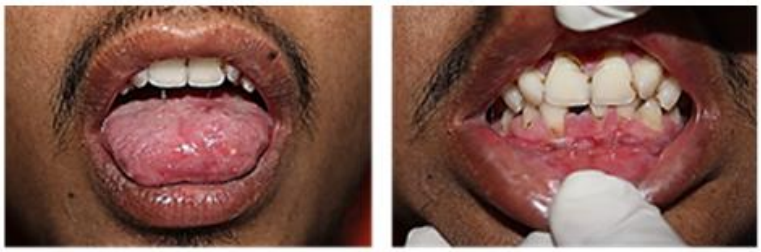


Fig. 2. Extra oral lesions in male patient.

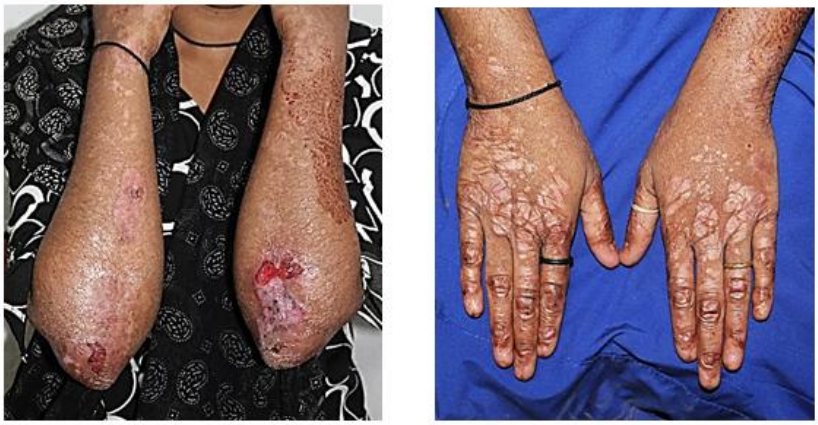
a. Multiple bullae and vesicles on hands with erosions and scarring.
b. New bullae and previous healed scars on lower extremity.
c. Multiple skin milia on ear.



a b.

Fig. 3. Intra oral lesions in male patient.

a. **Healed lesions and ulcers on tongue with reduced mouth opening.**
b. **Gingival recession & inflammation with lesions on labial mucosa & gingiva.**



a b.

Fig. 4. Extra oral lesions in female patient.

a. **New lesions in the form of bullae and denuded skin on elbows.**
b. **Healed lesions with scarring on back of hands.**

a. b.

Fig. 5. Intra oral lesions in female patient.

a. **Multiple vesicles and bullae on palate.**
b. **Multiple carious teeth**

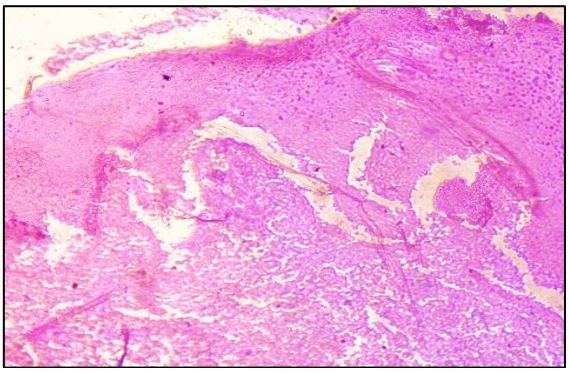


Fig. 6. Histopathological examination of biopsy specimen from an oral lesion showed hyperkeratosis, mild acanthosis, intact basal cells and sub-basal cleft.

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