

Case Report

Occurrence of epidermolysis bullosa along with Amelogenesis imperfecta in female patient of India

A. P. Javed¹, Prashanth Shenai², Laxmikanth Chatra², K. M. Veena², Prasanna Kumar Rao², Rachana Prabhu²

¹Department of Oral Medicine and Radiology, Annoor Dental College, Muvattupuzha, Kerala, ²Department of Oral Medicine and Radiology, Yenepoya Dental College, Yenepoya University, Deralakatte, Mangalore, Karnataka, India

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Address for correspondence:
Prof. Prashanth Shenai,
Professor, Department of
Oral Medicine and Radiology,
Yenepoya Dental College,
Yenepoya University,
Deralakatte,
Mangalore - 575018,
Karnataka, India
E-mail: ydcomr@gmail.com

ABSTRACT

Epidermolysis bullosa (EB) is an inherited disorder, which is characteristically presented as skin blisters developing in response to minor injury. Junctional variety of EB is also associated with enamel hypoplasia. Amelogenesis imperfecta presents with abnormal formation of the enamel both in deciduous and permanent dentition. This article describes a previously unreported case of Amelogenesis imperfecta with complete loss of enamel in a young female patient with EB.

Key Words: Amelogenesis imperfect, epidermolysis bullosa, vesicles and bullae

INTRODUCTION

Epidermolysis Bullosa (EB) is a group of genetically determined rare disorder, where mutations coding for targeted proteins involved with keratin filament assembly promote architectural alterations in the epithelial basement membrane complex. EB occurs at the time of birth or in early infancy. This is commonly observed in children and can be minor or much more severe, which is very different case by case. The children who have this disorder are often called by several different names such as butterfly children (because their skin is as fragile as a butterfly's wings) and cotton wool babies. It has an incidence rate of approximately, 400,000-500,000 people who are affected world-wide and no definitive treatment have yet been developed.^[1] It can affect both sexes equally and in any racial and ethnic group. The development of blisters following minor or insignificant

trauma to skin or mucosal surfaces leading to non-healing large ulcer formation is characteristic of this disorder. Disruptions to cellular adhesion will facilitate increased fragility of the skin and mucosal surfaces. Lesions may arise spontaneously, often with compromised wound healing with scarring in various EB subpopulations. Oral features seen with EB include mucosal vesicles and bulla that are frequently painful, exuberant granulation, tissue proliferation, and abnormal teeth usually affecting enamel that is complete or partial. The dentition may be affected severely by enamel hypoplasia and/or dental caries depending on the EB type. Dahl indicated that all patients with junctional EB (JEB) suffered from enamel hypoplasia. It has since been confirmed in a large prospective study that generalized enamel hypoplasia is limited to JEB types.^[2] Amelogenesis Imperfecta may present as hypoplastic, hypomineralized or both and the teeth affected may be discolored, sensitive or prone to disintegration. Amelogenesis Imperfecta (AI) is due to the malfunction of the proteins in the enamel, ameloblastin, enamelin, tuftelin and amelogenin.^[3] The exact incidence of AI is still uncertain, but the prevalence can vary from 1:700 to 1:14,000.^[4]

A rare case of a female patient suffering with JEB with Amelogenesis imperfecta is being reported.

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CASE REPORT

An 18-year-old female patient reported to department of Oral Medicine and Radiology, Yenepoya Dental College, Yenepoya University, Mangalore with a complaint of discolored teeth since childhood. Her past dental history revealed similar type with early loss of tooth structure in deciduous dentition. Her medical history revealed presence of multiple dermal lesions, which started appearing immediately after birth, which was later diagnosed as EB. She is second child from a consanguineous marriage and her sister is also affected with similar dermal lesions since childhood. Her gait was abnormal owing to non-healing ulcer in feet. Blisters were seen on the lower part of feet with irregular borders and yellowish slough present at the base of the lesions [Figure 1]. Scarring of healed lesions on knee was also noticed [Figure 2].

On examination, diffuse reddish pseudo membranous area is seen extending from external occipital protuberance and spreading bilaterally until ear and extends until scapula. Loss of hair is seen with respect to that area. There was presence of itching and burning sensation in the affected area. Surface was erythematous with yellowish slough [Figure 3]. On intra oral soft tissue examination, a vesicle was seen on the right rugae area on the hard palate [Figure 4]. However, there was no abnormal eruption pattern noticed. From a functional point of view, she had been avoiding hard food substances and carious lesion was noticed affecting enamel and dentin of teeth.

On detailed hard tissue examination, it was found that she had a normal complement of teeth. Height of teeth was reduced because of complete chipping of enamel and exposing dentin. Underlying dentin appears to be normal. The surfaces of the teeth were rough. The teeth, in general, exhibited a yellowish



Figure 1: Ulcerative lesion on feet



Figure 2: Healed lesion on knee



Figure 3: Dermal lesion in occipital region



Figure 4: Vesicle in the anterior palate

brown discoloration, with diffuse pitting present on the exposed tooth surfaces, more prominent on the labial and buccal aspects. The emergence pattern and timing of teeth seemed to be within the normal range. No occlusal disharmony is present [Figure 5].

Panoramic radiograph revealed a normal pulp chamber and root canal spaces. The enamel was completely lost, radiopaque dentin is clearly appreciated [Figure 6]. Based on history, clinical and radiographic features we arrived at diagnosis of hypoplastic, rough, autosomal recessive AI. Patient was advised to undergo biopsy and immunofluorescence test, but owing to unwillingness of patient's parents regarding non-healing of wound, tests could not be carried out. Aesthetic restoration was carried out with the composite veneering in relation to maxillary central incisors and left lateral incisor and patient is undergoing restoration of other teeth [Figure 7]. The patient was also referred for dermatological treatment.

DISCUSSION

EB is a diverse, heterogeneous group of conditions characterized by fragility of the skin that results in blisters caused by little or no trauma. EB is of 3 major categories, which includes- EB simplex (intraepidermal skin separation); JEB (skin separation in lamina lucida); and dystrophic EB (sublamina densa separation).^[5] Ten genes are known to harbor mutations in the major types of EB and the level of expression of these genes within the cutaneous basement membrane zone and in extracutaneous tissues as well as the types and combinations of the mutations, explain in general terms the phenotypic variability.^[6]

Molecular genetic studies revealed that Junctional form of EB is caused by mutations in the genes encoding COL17 or laminin 332. Enamel hypoplasia also has been reported in junctional form of EB. This is because deficiency of epithelial-mesenchymal junction molecules, such as COL17 can lead to the pathological mechanisms that can result in enamel hypoplasia.^[6] Disruption of the COL17 gene leads to abnormal interaction between enamel epithelium and the underlying mesenchyme via the epithelial-mesenchyme junction, resulting in defective ameloblast differentiation. Epithelial-mesenchymal interactions via the epithelial-mesenchymal junction are important for tooth morphogenesis, and

hemidesmosome components are thought to regulate the proliferation and differentiation of tooth forming cells including ameloblast.^[7] Since, there is the presence of enamel hypoplasia, which occurs only in association with the JEB and this case goes in favor of junctional enamel hypoplasia.



Figure 5: Clinical appearance of teeth



Figure 6: Panoramic radiograph-complete loss of enamel and loss of proximal contacts



Figure 7: Composite veneer on maxillary anterior teeth

Enamel hypoplasia in the form of Amelogenesis imperfecta is seen in this present case. Amelogenesis imperfecta can have different inheritance patterns depending on the gene that is altered. Based on the phenotype, Amelogenesis imperfecta is divided into four major categories-hypoplastic, hypomaturational, hypocalcified, and hypoplastic with taurodontism. Hypoplastic with taurodontism, which are again subdivided into 15 subtypes by phenotype and secondarily by mode of inheritance.^[4] Clinically, a skeletal anterior open bite is seen in approximately 50% of patients with Amelogenesis Imperfecta of either X-linked or autosomal inheritance however, in the present case, it was not evident. Such an association might be regarded as a syndrome, but this does not appear as such in any classification. The significance of this common association has yet to be elucidated. Diagnosis involves exclusion of extrinsic environmental or other factors, establishment of a likely inheritance pattern, and recognition of phenotype and correlation with the dates of tooth formation to exclude a chronological developmental disturbance.^[7] Radiographically, the enamel may appear totally absent. When present may appear as a thin layer, chiefly over the tips of the cusps and on the interproximal surfaces. In some cases, calcification is so much affected that enamel and dentin seem to have the same radio density, making differentiation between the two difficult.^[8] In the present case, complete absence of enamel was seen in radiograph.

The management of EB is primarily preventive and supportive, consisting of prevention of trauma, careful wound care, nutritional support, and infection control. Surgical procedures are indicated when deformities are caused by the blistering and scarring. Steroid therapy is controversial for EB. Since, EB are genetic disorders, no drug is capable of correcting the molecular defect. Gene therapy is potentially, a future therapy. Recently, researchers have reported sustainable genetic correction of JEB, patient skin tissue with laminin gene delivery. Clinical physicians should provide genetic counseling for families at risk for EB. The prognosis of EB depends on the severity of the illness.^[7]

There is also a need for diet supplements, such as vitamins, proteins, and iron in order to avoid anemia.

The use of vitamin E and immunosuppressive drugs have also been suggested for the treatment of EB.^[9] Dental treatment is aimed at avoiding the formation of new bulla during perioperative management and the choice of anesthetic method is one of the main issues for dentists and anesthesiologists. Special dental concerns involve the use of soft toothbrushes and irrigation techniques.

CONCLUSION

To the best of our knowledge, present case is first case report of EB along with Amelogenesis imperfecta. Dermal lesion and its association with dental anomalies have made management difficult. Palliative care was given for dermal lesions and aesthetic rehabilitation as part of dental management.

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