Case report

Amelogenesis Imperfecta- A case report

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Abstract

Amelogenesis Imperfecta (AI) is a diverse collection of inherited diseases that exhibit quantitative or qualitative tooth enamel defects in the absence of systemic manifestations. Enamel findings in AI are highly variable, ranging from deficient enamel formation to defects in the mineral and protein content. The affected teeth are disturbed in coloration, thickness and resistance. The rehabilitation of AI in a child must take into account the development of child’s teeth, the health of the periodontal tissues and the maxillary and mandibular growth. The purpose of this case report is to present the esthetic and functional rehabilitation of the teeth in a 13yr old patient with AI.

Key words: Amelogenesis Imperfecta; Enamel dysplasia; Heterogenous developmental disorder; Full mouth rehabilitation.

Introduction

Amelogenesis Imperfecta (AI) is a group of inherited defects of dental enamel formation that show both clinical and genetic heterogeneity (1). AI is also known by varied names such as hereditary enamel dysplasia, hereditary brown enamel, and hereditary brown opalescent teeth (2). Amelogenesis Imperfecta may affect all or only some of the teeth in the primary and/or permanent dentition (3). It occurs in the general population in the approximate range of 1 in 14,000 to 1 in 16,000(4). This disorder has been categorized into three main groups, hypoplastic, hypomaturation and hypocalcified and subdivided fourteen subtypes based on phenotype and patterns of inheritance (5).

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These enamel defects are a result of gene mutations associated with amelogenin protein and enamelin protein, which are secreted during the amelogenesis process. However, most forms of AI do not have a defined molecular basis at this time (6). AI may occur in isolation or in association with other abnormalities like cone rod dystrophy. It may show autosomal dominant, autosomal recessive, sex-linked and sporadic inheritance patterns with autosomal-dominant form being the most common (7). The disorder may create unaesthetic appearance, dental sensitivity and attrition. In these patients, the pulp and dentin are usually normal and the teeth are caries resistant (4). Dental features associated with AI include: quantitative and qualitative enamel deficiencies; pulpal calcification, taurodontism and root malformations; failed tooth eruption and impaction of permanent teeth; progressive root and crown resorption; congenitally missing teeth; and anterior and posterior open bite occlusions (8).

Dental radiographs of AI teeth provide important information to the clinician with respect to the degree of enamel mineralization to design an appropriate treatment plan. Evaluation of enamel density a change in AI teeth are generally made by contrasting the enamel with the dentin and if the enamel has a radiopacity similar to or less than that of dentin is considered mineral deficient.

Treatment planning for patients with AI is related to many factors: The age and socioeconomic status of the patient, the type and severity of the disorder, and the intraoral situation. An interdisciplinary approach is necessary to evaluate, diagnose and resolve esthetic problems using a combination of prosthodontic, orthodontic and restorative treatment (2).

The present paper reports the diagnosis and treatment planning of AI in a young male patient.

Case report

A 13yr old male patient reported with the complaint of extreme dissatisfaction with the esthetic appearance of his teeth and his mother confirmed that the patient had been socially affected by this problem. No significant medical and dental history was seen.

On intraoral examination, gingiva was observed to be swollen and the following teeth were present: 11,12,53,54,55,16,21,22,63,64,65,26,31,32,73,75,36,41,83,84,46.(figures 1-3). The enamel of all the teeth was yellow in color. The surfaces of the teeth were rough and irregular in shape and much smaller than normal with considerable tooth sensitivity. The enamel over the crowns was very thin and there was

Figure 1 : Pre operative view of lower posteriors

Figure 2 : Pre operative view of upper posteriors
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chipping on probing of enamel in relation to all the teeth. Stains and debris was present on the teeth. The occlusion of the patient revealed anterior open bite. Attrition of the molars had resulted in the decrease of the vertical dimension of occlusion. The diagnosis of Amelogenesis Imperfecta was made and the patient’s parent was informed of the diagnosis and the possible treatment options. A full mouth rehabilitation of the patient was planned.

![Figure 3: Pre-operative image depicting upper and lower anteriors](image)

![Figure 4: Post-operative view of lower posteriors](image)

The patient was placed on an intensive oral hygiene program because of his inadequate oral hygiene. Treatment included was oral prophylaxis, composite build up for anteriors and stainless steel crown for the molars (figures 4-6). Periodic topical fluoride application was advised. The patient is presently under regular follow up with emphasis on the diet control and oral hygiene. Also the patient is advised for regular radiographic analysis and the patient is prescribed on remineralizing pastes for the subsequent teeth which are erupting.

![Figure 5: Post-operative image depicting upper posteriors](image)

![Figure 3: Post-operative view of anteriors](image)

Discussion

Inherited enamel defects that occur in the absence of a generalized syndrome are collectively designated as Amelogenesis Imperfecta (AI) (9).

Genetically, currently, there are five proven candidate genes for AI: amelogenin (AMELX), enamelin (ENAM), enamelysin (MMP20), kallikrein 4 (KLK4), and distal-less homeobox 3 (DLX3). There are also two unproven candidate genes: ameloblastin (AMBN) and tuftelin (TUFT1) (9). AI may be inherited by X-linked autosomal dominant or recessive, or sporadic inheritance.
However, we did not attempt to trace the gene responsible in our case.

AI is currently classified into 14 distinct subtypes based on various phenotypic criteria; however, the gene responsible for each phenotype has not been defined. Many classifications of AI have evolved since the original division into hypoplastic and hypocalcified types in 1945. Some have been exclusively based on the phenotype (appearance); others have used the phenotype as the primary discriminant and the mode of inheritance as a secondary factor in diagnosis. Similarly, it was the phenotype in the present case that aided in diagnosing the condition. The most recent classification was given by Aldred et al., in 2003 based on the mode of inheritance as phenotype (clinical and radiographic), molecular defect (when known), biochemical result (when known) (7,9).

Amelogenesis imperfecta has marked psychosocial effects, which suggests that it’s very important to have teeth and oral esthetics for a normal psychosocial development (6). A primary goal for the treatment is to address each concern as it presents with an overall comprehensive plan that outlines anticipated future treatment needs. In accordance, we advised for regular follow up even after treating the condition. Clinician treating children and adolescents with Amelogenesis Imperfecta must address the clinical and emotional demands of these disorders with sensitivity. It is important to establish good rapport with the child and the family early. Timely intervention is critical to spare the patient from psychosocial consequences of these potentially disfiguring conditions. A comprehensive and timely approach is reassuring to the patient and family and may help decrease their anxiety. However, the restoration of esthetics and function of teeth in childhood patients suffering from Amelogenesis Imperfecta often represents to the dentist a major challenge. Attention should first be given to the patient’s level of oral hygiene and dietary habits, which can compromise the rehabilitation procedures. Poor oral hygiene is a recognized problem in patients with AI, because of the rough enamel surface which causes plaque retention and to the sensitivity experienced when brushing (6). In agreement the present patient had tooth sensitivity and poor oral hygiene. Therefore, meticulous oral hygiene, calculus removal, and oral rinses can improve periodontal health, fluoride applications and desensitizing agent may diminish tooth sensitivity (10). It is common for AI patients to have spacing between the teeth may reduce the susceptibility to interproximal caries.

Amelogenesis Imperfecta presents with problems of socialization, function and discomfort which may be managed by early vigorous intervention, both preventively and restoratively. A multidisciplinary approach consisting of an orthodontist, prosthodontist and endodontist should be planned (2). Similar approach was undertaken in the present case in accordance to a previous report on similar condition (2).

Conclusions

Amelogenesis Imperfecta is a heterogenous developmental disorder which presents with severe dental anomalies (2). The complexity of the management of patients with Amelogenesis Imperfecta supports the suggestion that the dental profession should have appropriate methods for the rehabilitation of rare dental disorders. The psychological effects of Amelogenesis Imperfecta on affected individuals, even in children are significant. Although more simple, the treatment on the children must be performed with attention, in order to re-establish both the function and the patient/ parents expectations to prevent serious problems in the future occlusion and social adaptation of the patient.
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References