

Amelogenesis Imperfekta: Review and Case Report

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Abstract

Amelogenesis imperfecta is a hereditary anomaly that affects enamel formation and is observed in primary and permanent dentition. Amelogenesis imperfecta (AI) is an inherited disease characterized by a marked defect in the enamel structure of primary and permanent teeth without any systemic disease. In many cases, it shows an autosomal dominant, autosomal recessive or X-linked genetic transmission. Developmental enamel defects; they are defects that can be hereditary or acquired as a result of the cessation of development during the development of enamel tissue. Defects can cause problems such as sensitivity, aesthetic problems and caries susceptibility. Therefore, it has been stated that defects can negatively affect the quality of life. Developmental enamel defects, early diagnosis and preventive treatments are primarily conditions that require a multidisciplinary approach. The main concerns of patients with AI are caries tendency, poor aesthetics and widespread sensitivity. Treatment of these defects is extremely important in terms of aesthetics, function and phonation, as well as the correction of psychosocial problems associated with them. Many factors such as age, socioeconomic status, type and severity of the disease, and oral condition at the time of treatment are effective in the treatment planning of AI patients. In this study, information about AI was given by reviewing the literature, and a patient with amelogenesis imperfecta who applied to our clinic in adulthood was followed up by performing treatment procedures aimed at eliminating aesthetic, function, phonation and psychosocial problems, as well as increasing the psychosocial quality of life of the case.

Case Report (HRU IJDOR 2021; 1(1):14-18)

Keywords: Amelogenesis imperfecta, Enamel defects, Loss of vertical dimension

Introduction

Amelogenesis Imperfecta (AI); it is a rare hereditary developmental anomaly that shows genetic inheritance, affects the enamel structure, quantity and composition of primary and permanent teeth, with or without systemic diseases (1). Anomaly is seen in the ectodermal structures forming the tooth, whereas the mesodermal structure is completely normal (2). It was first described as hereditary brown tooth in 1890. It was later named brown hypoplasia of enamel by Finn. from 1890 to 1938, it was not considered clinically independent of dentinogenesis imperfecta (3-5).

In the epidemiological study, its incidence was reported to vary between 1/718 and 1/14000 (6). Its prevalence was reported as 1/700 in Northern Sweden, 1/14000 in America, 43/10.000 in Turkey, 10/10.000 in Argentina and 1.25/10000 in Israel, depending on the populations studied (5). Depending of these data, its prevalence in the world is 0.5% on average. However, a racial tendency of AI has not yet been reported (7). In AI, in addition to the irregular structure of the enamel, the primary clinical problem is sensitivity to thermal stimulus (8). In addition, congenital tooth deficiencies, loss of vertical dimension, anterior region open bite, pulpal calcifications, dentin dysplasia, root and crown resorption, hypercementosis, root malformations, anomalies in eruption of teeth, taurodontism can be seen (9). In addition, these teeth are

extremely resistant to caries (7). It can be seen as autosomal dominant, autosomal recessive or rarely linked to X chromosome (10).

In the current classification, AI is divided into four types according to the anatomical and histological features of enamel: hypoplastic, hypomatured, hypocalcified, and hypomatured-hypoplastic observed with taurodontism; although these four groups are divided into many subgroups according to their clinical, histological, radiological and genetic characteristics, similar symptoms such as tooth sensitivity, aesthetic and functional insufficiency are observed in patients (11).

Among the AI types, the most common hypoplastic type is thought to occur with insufficient deposition of the enamel matrix (12). Pits or grooves are observed on the shiny, yellow, hard enamel surface (11). Conical or cylindrical dental crowns have diastema (13).

The formation of hypomature organic matrix is normal, but the maturation of the enamel crystal structure is defective. Normal-thickness enamel is stained opaque white, yellow-brown or red-brown in color and slightly softer than normal. It is difficult to distinguish between enamel and dentin on the radiogram (14-16).

In the hypocalcified type, enamel is relatively normally formed, but mineralization is weak. Shortly after the teeth erupt, the enamel is separated from the dentin. Hypocalcified enamel and exposed dentin are easily stained by environmental factors; teeth appear yellow-brown. Patients have complaints of tooth sensitivity. In the last group, it is seen as a combination of hypomaturity-hypoplasia types. Numerous white-yellow mottled spots or brown small teeth can be seen on the vestibule surfaces of the teeth. Proximal contacts may not be present (11).

The diagnosis of AI is made by clinical anamnesis, radiographic evaluation and genetic studies. Although clinical and radiographic diagnostic methods are frequently used, the definitive diagnosis should be made by genetic studies, since these diagnostic criteria are not objective (17).

The negative effects of AI on aesthetic appearance, function, occlusion, phonation, gingival health and psychological health also negatively affect the quality of life of patients (18). The aim of the treatment of AI patients is to eliminate the aesthetic function and phonation problems that the patient has lost, to solve the problems associated with morphological structure disorder and tooth sensitivity and to take them under control. A multidisciplinary treatment approach with a combination of periodontal, prosthetic and restorative treatments is

required for the evaluation, diagnosis, treatment and long-term maintenance of oral health (11). Treatment planning should be shaped according to the patient's age, socioeconomic status, type and severity of AI, and the intraoral situation at the time of treatment planning. In line with this information, although different approaches than routine dental treatments were considered, a standard procedure for successful treatment could not be established due to the lack of long follow-up periods (17).

In patients with AI, loss of vertical dimensional is seen, which increases with age due to the wear of the teeth. Treatments such as overdenture prostheses, crown veneers, stainless steel crowns and onlay restorations can be applied to regulate the occlusion of the patients (19-23). In full crown applications, difficulties are encountered such as the age of the patient, the short crown and root length of the teeth, and the excess number of teeth to be restored (24). On the other hand, overdenture prosthesis treatments supported by soft tissues, tooth roots or modified teeth prevent loss of vertical dimension, especially in AI cases in children and young patients, and lay a suitable basis for treatments that can be applied in older ages. In addition to these, it contributes to the aesthetics of the individual, providing the chewing function and bringing them to a psychologically healthier state. The fact that overdenture prostheses can be applied in a short time and is a non-invasive method can also be counted among its advantages. The most preferred restorative material due to aesthetic concerns is composite resins. However, this method fails in some patients due to structural defects in enamel and dentin (17,24).

Case Report

Thirty-two-year-old female patient applied to the Faculty of Dentistry of Harran University with widespread tooth sensitivity and aesthetic complaints due to the appearance of her teeth, and she hesitated to talk (Figure 1-2). In the clinical examination, it was determined that the enamel layer in all teeth was very thin, rough and eroded, and the teeth were yellow-brown in color and opaque. The patient's posterior group teeth were worn and the teeth were yellow. Although the teeth were in preparation tooth form, they were in contact in the occlusion. It was determined that the loss of vertical dimension of the occlusion decreased and the lower face height decreased due to the crown of the patient's teeth. Gingivitis, the presence of bleeding gingiva on probing and the presence of supragingival calculus on the lingual surfaces of the lower incisors were detected due to the patient's lack of tooth brushing habit and the presence of hypocalcified teeth. It was observed that the parts of the enamel defects under the

gingiva facilitate plaque accumulation and make cleaning difficult.

When the patient smiles, in addition to these images, an appearance in which the gingiva is visible between the cervicals of the teeth and the upper lip border was determined (Gummy smile). After the Informed Consent Form was obtained from the patient, periodontal treatment was performed in the first appointment and oral hygiene training was given. Treatment was given an appointment for two weeks later. Restorative treatments were completed in the second appointment. Prosthetic treatment was planned for the loss of vertical dimension.

Figure 1A,B. Clinical intraoral view of the patient.



1A.



1B.

Figure 2. Radiological panoramic view.



AI is a rare disease that affects the quality and quantity of tooth enamel, negatively affecting aesthetic appearance, function, occlusion, phonation and gingival health. AI causes psychological and physiological problems that seriously decrease the quality of life in many patients. Therefore, these patients should be treated with a comprehensive and multidisciplinary approach (11).

In patients with AI, gingival inflammation and related soft tissue deformities are seen as a result of tooth sensitivity, difficulty in brushing, inability to achieve physiological lubricity, deterioration of food distribution and increased plaque retention, which are seen due to the defected morphological structure (25).

There are many treatment options available to rehabilitate the dentition with amelogenesis imperfecta (22). However, the factors to be considered while making a decision on this issue are; patient's age, socioeconomic status, condition of teeth, periodontal health, condition of impacted teeth, occlusion and psychosocial status of the patient (24). While planning the treatment for our patient, treatment was planned with a multidisciplinary approach in line with the patient's complaints.

In our case, the amount of bacterial plaque was reduced by giving oral hygiene training at the beginning. In a study on AI, prosthetic rehabilitation of patients; metal-ceramic crowns, composite restorations, stainless steel crowns, laminate applications, onlay applications, overdenture applications (5).

Today, a treatment plan with full porcelain restorations is preferred for patients with AI. In the field of aesthetic dentistry of full porcelain restorations; it has advantages such as bonding to dentin, restoring function and acceptable aesthetic appearance. However, marginal compatibility and bonding problems have been reported as disadvantages for laminate veneer restorations (7). Sarı and Uşmez applied two treatment plans together in a study they conducted. Anterior teeth were restored with porcelain laminate veneers and maxillary and mandibular

posterior teeth with full porcelain crowns (26). Çakmak et al. reported that by using overlay prostheses, which are a kind of overdenture, in pediatric patients with AI, chewing habits were restored, aesthetic expectations were met, and the patient's speech improved (27). They also reported that the construction of this type of prosthesis was completed in a short time and the cost was low (5). In a study related to AI, they stated that they observed that patients adapt better to their friend circles in the post-treatment follow-up and that their school success and self-confidence level increased due to the increase in school attendance at school age (28). Similarly, the fact that our patient started the treatment increased her self-confidence and the desire to come to her appointments on time and complete the treatment as soon as possible.

Conclusion

AI is the change in quality and quantity of tooth enamel. When deciding on the treatment of the patient with AI, factors such as the type of anomaly, the patient's age, socioeconomic status, the extent of eruption, and periodontal health should be taken into account. It should be known that clinical changes can be seen depending on the classification. In AI cases, early treatment can prevent loss of aesthetics, function, loss of vertical dimension and sensitivity, and the patient's rate of being psychologically affected by this situation can be reduced. The long-term maintenance of the treatment results is acceptable for the physician and the patient if the applied prosthetic restorations are paid attention to the marginal harmony, occlusal and interface relations and regular periodontal prophylaxis is performed. Since primary teeth and permanent teeth are affected together in AI patients, preventive measurements before eruption of permanent teeth. In such patients, it is important to start treatment as early as possible in order to increase tissue destruction over time and to prevent early tooth loss.

Data Availability

The data that support the findings of this study are available upon reasonable request.

Consent

Informed consent was obtained from the patient. Written informed consent was obtained from the patient before the study.

Conflicts of Interest

No conflicts of interest related to this paper.

Contributions of the authors:

Study conception and design: YY, ZA

Analysis and interpretation of the data: YY, ZA

Draft manuscript preparation: YY, ZA

Critical revision of the work: YY, ZA

All authors reviewed the results and approved the final

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