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Case Report

Amelogenesis Imperfecta from Diagnosis to Rehabilitation-A Case Report

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Abstract

During odontogenesis, enamel is normally synthesized as an extracellular matrix, a phenomenon known as amelogenesis. The failure of this process can lead to malformationof dental enamel, called Amelogenesis Imperfecta (AI). AI, by causing a qualitative and/or quantitative deficiency of dental enamel, presents characteristics such as hypersensitivity, unsatisfactory aesthetics, reduced vertical dimension, anterior open bite,plaque accumulation, and greater susceptibility to caries and gingivitis. Thus, our objective is to report a case of AI, describing the main characteristics of the disease, the diagnosis and the rehabilitative treatment plan, aiming to improve the patient's aesthetics and reestablishing the function of the stomatognathic system. Patient M.A.S.B.,female, 5 years old, leucoderma, was admitted at the University of Patos de Minas Dental Clinic with a complaint of pain in the teeth. During the clinical interview, the person in charge reportedthat the child's teeth had the same defects as those of her mother and brother. During clinical examination it was observed: generalized loss of tooth structure, color change, rough surfaces and hypersensitivity. Treatment involved restorations with glass ionomer cement and composite resin, extraction of residual roots from the maxillary incisors, and confection of a functional and aesthetic space maintainer. Early diagnosis associated with the correct treatment plan is essential for a more conservative approach focused on preventing the effects of AI. In advanced cases, restoring function and aesthetics is paramount to improve the patient's quality of life.

Keywords: amelogenesis imperfecta; clinical features; oral rehabilitation.

Introduction

Amelogenesis imperfecta (AI) is a group of hereditary alterations that exhibit anomalies in the primary enamel and permanent dentition [1]. It is considered a rare genetic alteration, since its frequency is depicted between 1:14000 and 1:700 [2]. However, in some populations where consanguineous marriages are customary, this frequency may be even higher [3]. Four main types are noted for AI, as: (i) hypoplastic, (ii) hypomineralized, (iii) hypomaturity, and (iv) hypoplastic-hypomaturity AI with taurodontism [1].

AI can be expressed in isolation, without any other non-oral symptoms, or as a phenotype of syndromic conditions, such as renal enamel syndrome(OMIM # 204690; *FAM20A*) and Jalili syndrome (OMIM # 217080; *CNNM4*). To date, more than 10 genes have been identified as being involved in the pathogenesis of AI [4].

The AI clinical characteristics depend on the type of the presented defect and on the stage of enamel formation, as: exaggerated dental sensitivity, compromised esthetics, loss of vertical dimension, plaque accumulation and, consequently, greater susceptibility to caries and gingivitis [5, 6, 7, 8]. Besides that, early teeth eruption, progressive resorption of roots and crowns, short roots and gingival hyperplasia, can also be associated with AI [5, 9]. In addition, craniofacial features can be observed, such as the inverted curve of Spee, restricted maxillary arch, and anterior/posterior open bite. These are considered severe features and can be directly linked to issues in the patient's social life.

Rehabilitation in AI cases brings numerous challenges for dental surgeons because, besides involving an interdisciplinary approach, there is no guarantee of a relapse-free treatment. It is necessary to maintain case follow-up, respecting the stages in which the alteration is found and the age of the patient [5].

Based on the above, the purpose of this paper is to report a case of AI, describing the main characteristics of this disease, its diagnosis, and the rehabilitative treatment plan, aiming to improve the patient's aesthetics,

in addition to restore the her stomatognathic system function.

Case Report

M.A.S.B., 5 years old, female, leucoderma, was admitted at the University Center of Patos de Minas Dental Clinic, accompanied by her guardians, with pain in several dental elements. During the clinical interview, her mother reported dissatisfaction with the color and the quality of her daughter's teeth, claiming that they were yellowish and weak. In addition, she complained that the child had the same defects as her own and her brother's teeth. It was also reported that the patient had already received dental treatment, but due to the child's difficulty in understanding and accepting, it was unsatisfactory. The patient was submitted to clinical and radiographic exams, in addition to conditioning sections, oral hygiene instructions and treatment to restore aesthetics and function.

During the extraoral clinical examination no alteration was identified that would justify any dysfunction, whether muscular and/or articular. The intraoral examination revealed generalized loss of tooth structure, color change, rough surfaces, hypersensitivity and aesthetic impairment (Figures 1 and 2). Furthermore, the patient presented malocclusion and loss of vertical dimension. On panoramic radiography exam, it was possible to observe normal characteristics for both dentin and pulp, however, the absence of the enamel lamina was noticed (Figure 3).

Figure 1: *Dental elements of the lower arch with loss of tooth structure, rough surfaces and color change, causing aesthetic compromise*.

Figure 2: *Dental elements of the upper arch with loss of tooth structure and color change, causing, aesthetic compromise*

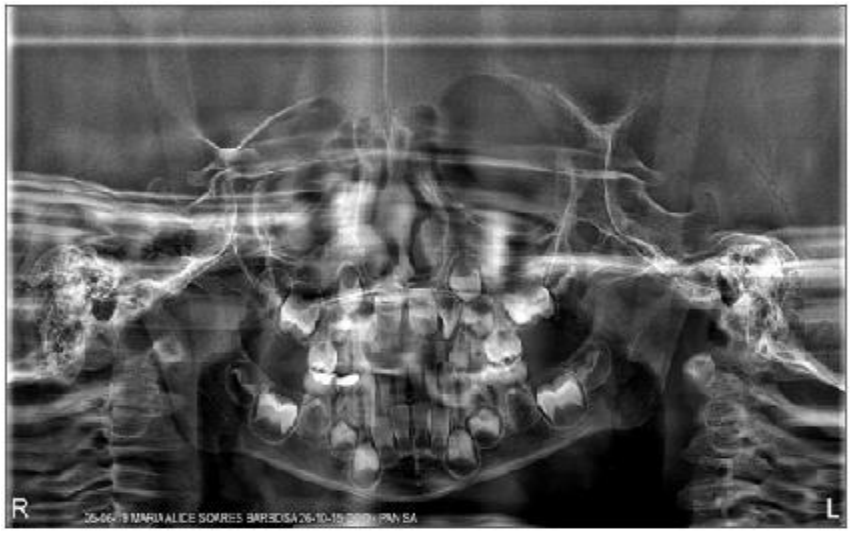

Figure 3: *Panoramic radiographt showing the absent of the enamel lamina, while the dentin and the pulp show normal aspects*

Taking into consideration the clinical interview, the extra and intraoral clinical examination, the radiographic findings and the absence of systemic alterations that would justifydental enamel malformation, the most likely diagnostic hypothesis was AI. Based on this, the patient's plan, which is described in detail below, was elaborated aiming at the return of function and aesthetics, starting with the urgency and conditioning procedures of the patient, followed by environment adaptation and restorative phase.

The first treatment phase was dedicated to the urgencies and patient conditioning, since she was extremely afraid of the dentist, the instruments, and the noise of the high and low rotation motors:

- Extraction of the residual roots of elements 61 and 62 (1st week) (figure 4);
- Extraction of the residual roots of elements 51 and 52 (2nd week);
- Conditioning of the patient through drawings and videos related to dentistry(3rd week);
- Conditioning of the patient by means of play and prophylaxis with the lowrotation, Robinson's brush, rubber cup and prophylaxis paste (4th week).

The second treatment phase focused on eliminating infective foci anddecreasing hypersensitivity:

- Removal of decayed tissue, pulp protection with calcium hydroxide andrestoration with glass ionomer cement on element 85 (5th week);
- Removal of decayed tissue and restoration with composite resin on elements74 and 75 and application of resin sealant on elements 55 and 65 (6th week);
- Topical application of fluoride in the whole mouth (5th and 6th week).

The third treatment phase aimed at restoring the patient's function and æsthetics:

- Restoration with composite resin on elements 53, 63 and 73 (7th week);
- Restoration with composite resin using celluloid matrix on elements 72 and82 (8th week) (figure 5);
- Restoration with composite resin on elements 71 and 81 (9th week);
- Fabrication of a functional and esthetic space maintainer (10th week).

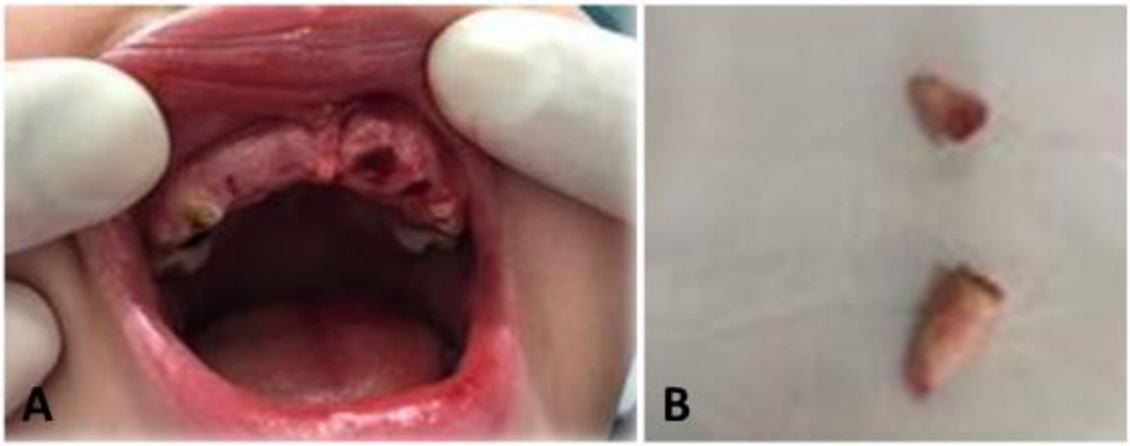

Figure 4: A: *Immediate post extraction of residual roots. B: Residual roots*

Figure 5: *Composite resin restoration on the lower lateral incisors, making a comparison with the lower central incisors*

Discussion

AI is a group of genomic origin disorders that affect both the structure and chemical composition of dental enamel [10]. In addition, it can be associated with morphological and/or biochemical changes in other parts of the human body [11].

During tooth development, the enamel is normally synthesized as an extracellularmatrix and this phenomenon is defined as amelogenesis. It happens in two stages, wherethe first (secretory stage) is responsible for the production of the mineralized protein matrix, which will result in the adult enamel, and the second (maturation stage), responsible for the degradation of the matrix and the completion of mineralization [11, 12].

Some of these variations affected genes that encode structural enamel proteins (amelogenin, ameloblastin, c4orf26, enamelin), enzymatic proteins (kallikrein 4, *MMP20)*, cellular proteins *(WDR72, FAM83H, COL17A1)*, transcription factors (*MSX2*, DLX3) and cell receptor *(ITGB6)* [13].

There are several researchers who have set out to describe the forms of AI. Molecular defects, phenotypes, mode of inheritance and biochemical analysis are used as arguments to defend the most varied types of classification, with at least 13 models [5, 12, 14]. The most accepted classification in the literature, however, is divided into three main types (hypoplastic AI, hypomaturated AI, and hypocalcified AI) and is based on the stages of the enamel formation process [15]. In hypoplastic AI, a failure occurs during the secretory phase of amelogenesis and, as a result, the tooth enamel is thinner than normal. On radiography, it is possible to observe a contrast with the dentin [5, 13].

In hypocalcified AI, the failure happens during the beginning of mineralization and, therefore, the enamel has an adequate thickness, but it is softer, presents a yellow-orange coloration and, because the matrix is badly calcified, it is quickly lost. Radiographically, the enamel has less radiopacity than the dentin [5, 15]. Finally, inhypomaturation AI, the defect occurs in the maturation phase, with the enamel showing normal thickness, but much softer than normal. Its appearance varies from white to darkbrown and, on radiography, it presents the same radiopacity as dentin [15, 16]. In the present case, it was observed that the patient, besides having cases in her family of AI, presented a friable enamel with yellowish coloration and, radiographically, a more radiopaque dentin than the enamel. Thus, based on her past history and these clinicaland radiographic findings, we reached the diagnosis of hypocalcified amelogenesis imperfecta.

The primary discriminator for the AI classification is usually the phenotype, but some argue that the primary structure should be the mode of inheritance, since the molecular basis of all AI has already been established. Thus, in this classification model, the clinical and radiographic features would be the secondary discriminators of AI [11].

Because AI causes both aqualitative and quantitative deficiency of dental enamel, some clinical features and consequences become evident. Besides those already mentioned, it is possible to observe rough enamel texture, stained appearance, ogival palate, delayed eruption, dental impaction, presence of cavities in the teeth and diastemas[9, 17]. As a result of such characteristics, these patients have a significantly higher level of distress and social isolation than individuals without the condition, thus suffering a social impact on education, work, and interpersonal relationships [18, 19, 20].

In the case in question, we noticed that the patient, in the first sessions, presented great difficulty in relating to others, was introspective, resisted dental treatment, avoided eye contact, and did not smile. However, as the treatment progressed and her teeth were restored, we could notice a clear improvement in her behavior and relationships. The patient played, hugged and smiled a lot to show her new teeth. Thus, we ratify what is described in the literature and highlight the importance of rehabilitative and aesthetic treatment to improve the patient's self-esteem and interpersonal relationships.

For an adequate treatment plan elaboration, it is of fundamental importancenot only to confirm the diagnosis of AI, but also to know how to identify both the type and severity of the disease. This requires differential diagnosis with dental fluorosis, molar-incisor hypomineralization, early carie lesions, dentinogenesis imperfecta type III and tetracycline staining [12, 21]. By diagnosing the presence of AI in family members of the presented case, the diagnosis became less challenging.

Close the diagnosis of AI is a very complex task, once it has an enormous clinical variability, presenting up to 14 subtypes [10, 15]. Therefore, a detailed information of the family history, as well as clinical and radiographic examinations to analyze the density and the thickness

of the enamel layer, are extremely important to diagnose AI [21]. In addition, when possible, it is important to take deoxyribonucleic acid (DNA) samples,molecular genetics and a morphological analysis using electron microscopy scanning sections [22, 23, 24]. Based on the family history and both clinical and imaging examinations, it was already possible to identify the diagnostic hypothesis, however, to obtain confirmation, a genetic mapping was requested from the patient's family, which was not possible to be performed due to her scarce financial conditions.

In the past, the standard treatment for AI consisted, basically, in multiple extractions followed by the making of total prosthesis, generating psychological problems for patients, especially young ones [25]. Nowadays, thanks to advances in dental materialsand greater technical and scientific knowledge on the part of professionals, the treatment modalities are varied and, to determine the best conduct to be employed, it is necessary to take into account the degree of AI severity, age and socioeconomic status of thepatient [21, 26, 27]. However, regardless of the treatment plan chosen, the final objective is always the same: control the vertical dimension, improve aesthetics and eliminate dentin sensitivity, always preserving the dental structures as much as possible [14, 21, 24, 28].

Because the treatments are mostly lengthy and complex, the multidisciplinary follow-up, with periodontal, odontopediatric, orthodontic, endodontic and even psychological therapy, becomes essential for the success of the case [21, 26, 27]. Ideally, the diagnosis of AI should always be early so that it is possible to perform all steps of thetreatment, which begins during childhood. In this initial phase, the treatment consists basically of oral hygiene orientation, topical fluoride application to reduce sensitivity, prevention of atrophy lesions and provisional restorations with glass ionomer cement (GIC) [7].

In the present case, when the family sought dental care for the patient, the oral problems caused by the AI were already advanced. Therefore, for the residual roots, it was necessary to resort to surgery; for the cavitated posterior teeth with active or inactive carie lesions, we performed the restoration withGIC; for the anterior teeth, which had already lost tooth enamel, a composite resin restoration was performed using celluloid matrix. In addition, following what is recommended in the literature, we performed oral hygiene orientations for both the child and the family and topical fluoride application.

For complications caused by AI ranging from mild to moderate, microabrasion, the application of fluoride varnish or amorphous casein-calcium phosphopeptide (CPP- ACP), are effective treatment options. The application of 37% phosphoric acid with pumice stone or 6% hydrochloric acid with silica can also be used for the treatment of mild to moderate AI [29]. In contrast, these treatments require frequent applications and do not produce totally satisfactory aesthetic results, becoming an inconvenient factor [30, 31].

An effective alternative to treat definitively the consequences of mild and moderate AI is the application of infiltrating resin. It presents low viscosity and, thus, is able to flow into the enamel porosity, hardening the tissue and improving aesthetics. Moreover, the application of the infiltrating resin is considered minimally invasive and less aggressive than conventional procedures [32, 33]. Because they have properties that confer greater aesthetic stability and functional conditions, such as the maintenance of disocclusion guides, ceramics are an excellent treatment option for more severe AI [7, 31, 34]. However, in order to have a good adaptation, a greater dental wear is necessary,a conduct that goes against what is recommended for the AI treatment [35].

Conclusion

The diagnosis of AI is challenging, so a very detailed interview and a thorough clinical examination, as well as complementary imaging exams associated with agood knowledge of the condition is necessary. Moreover, it is of fundamental importanceto make an early diagnosis and a correct treatment plan, aiming at a more conservativeapproach and focusing on the prevention of the AI effects.

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