Amelogenesis Imperfecta: Report of a Successful Transitional Treatment in the Mixed Dentition

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ABSTRACT

The term amelogenesis imperfecta is applied to a clinically heterogeneous group of hereditary disorders that interfere with the normal development of dental enamel. These disorders cause a deficiency in the enamel's quantity and/or the quality that may result in poor dental esthetics. The purpose of this paper was to describe a case of hypoplastic amelogenesis imperfecta in an 8-year-old girl whose dissatisfaction with the appearance of her teeth led to impaired social functioning. Since the patient was in the mixed dentition stage, a temporary treatment aiming to improve dental esthetics, preserve oral function, and allow for the recovery of the patient's self-confidence was performed by a multidisciplinary team. (**J Dent Child 2008;75:201-6**)

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Amelogenesis imperfecta (**AI**) consists of a diverse group of genetically determined enamel defects not associated with any other systemic disorder. AI results from single-gene mutations that follow autosomal dominant, autosomal recessive or X-linked patterns of inheritance.[1-3] All or some of the teeth in both primary and permanent dentition are affected.[1-3]

AI can occur as 3 main types, depending on the stage of enamel formation primarily affected: (1) hypoplastic; (2) hypomaturation; and (3) hypocalcified. (Table 1) The differential diagnosis among these 3 types is usually based on clinical and radiographic evaluation and on the mode of inheritance of the trait, as determined from family pedigrees.[1,2] An affected individual may be classified as having one type of AI and, at the same time, have areas in one or more teeth that show enamel defects characteristic of other forms of AI, due to environmental and/or other genetic challenges.[1] Table 1 describes the main phenotype characteristics of each type of AI .

Recently, molecular studies and mutational analyses have established a correlation between AI and certain genes encoding specific enamel proteins.[5] Alterations in the amelogenin gene (AMELX) are responsible for X-linked AI, whereas mutations in the enamelin gene (ENAM), enamelysin gene (MMP-20), and kallekrein-4 gene (KLK-4) cause hypoplastic or hypomaturation AI with the autosomal pattern of inheritance.[5-8] Genes responsible for the autosomal-dominant hypocalcified AI, however, have not been identified yet.[5]

In addition to the compromised appearance of their teeth, AI patients may face chewing function problems due to dental sensitivity and the reduced clinical crowns caused by attrition or incomplete eruption.[9-12] Anterior open bite is also a common finding associated with AI. Ravassipour et al[13] found that, among 54 AI subjects, 42% had either dental and/or skeletal open bite malocclusions and observed

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*Table 1. Phenotype Characteristics of the 3 Main Types of Amelogenesis Imperfecta **

Type	Clinical features	Radiographic features
I. Hypoplastic (deficiency in the quantity of enamel)	• Thin, hard, and glossy enamel • Surface may show pits and grooves • Coloration: Yellow to brownish • Incomplete eruption, delayed eruption, coronal resorption before eruption	• Pulpal calcifications • Normal contrast between enamel and dentine; the reduced thickness of enamel makes its visualization difficult
II. Hypomaturation (abnormally matured enamel)	• Snow-capped teeth (mottled areas with flecks of opaque white enamel) The defects are limited to the incisal and occlusal portions of the teeth • It can resemble dental fluorosis	• Lack of contrast between enamel and dentine
III. Hypocalcified (insufficiently mineralized enamel)	• Opaque soft enamel, easily removed • Coloration: Honey to light brown • Dentinal hypersensitivity • Incomplete eruption, delayed eruption, coronal resorption before eruption	• Lack of contrast between enamel and dentine • Pitted crowns

** Classification of amelogenesis imperfecta as reported by Witkop,[1] Seow,[2] and Hunter et al.[3]*

that enamel phenotype severity was not associated with the presence or severity of this type of malocclusion. Aren et al[14] studied the dental and craniofacial characteristics of a sample of 33 female and 10 male AI patients. They showed that anterior open bite was present in 50% of the hypoplastic AI patients, 31% of the hypomaturation AI patients, and 60% of the hypocalcified AI patients. This association may be due to a genetically determined anomaly of craniofacial development[15,16] or to environmental factors such as abnormal tongue positioning caused by dental sensitivity.[2,17] The true reasons for the higher prevalence of anterior open bite among AI patients, however, remain unclear.[2,13,15,17]

Another aspect that cannot be underestimated is the psychological impact on patients since this pathology generally gives the teeth an unsatisfactory appearance. Therefore, patients may show low self-esteem and excessively introspective behavior, which affects their socialization.[11,12,18] A study conducted by Coffield et al[19] aimed to associate AI with a negative psychosocial outcome. The results showed that more AI patients reported being teased about their teeth and being unhappy with the color, shape, and size of their teeth than did the unaffected patients. They also presented higher levels of social avoidance and distress, and fewer subjects with AI were married or in a committed relationship. Also found among AI patients were higher levels of dysfunction, discomfort, and disability attributable to oral conditions and higher levels of fear of negative evaluation, especially at younger ages.

When treating AI child patients, professionals should adopt a comprehensive approach and plan a treatment capable of satisfying both patients and parents. The treatment implies motivation and involvement on the part of the young patient and the family. It is, thus, extremely important that all the steps and treatment options be carefully explained to the patients' families, along with the difficulties related to the rehabilitation process.

The bonding between the restorative material and the defective enamel is one of the main obstacles during the rehabilitation of an AI patient. For all types of AI, according to Seow and Amaratunge,[20] it is possible to obtain an etched surface after the use of 37% phosphoric acid. Some types, however, lend themselves to better etching and bonding than others. It has been shown that an enamel treatment with 5% sodium hypochlorite for 1 minute, after acid conditioning, enhanced enamel bond strength in cases of hypocalcified AI.[21,22]

According to the type of AI and to the patient's age, many treatments have been proposed and various strategies may be used to overcome the compromised esthetics and function. There is general agreement that, when the patient is in primary or mixed dentition, the main goal is to provide a treatment that can reestablish esthetics and chewing function and reduce dental hypersensitivity and attrition until the patient approaches adulthood—when a permanent treatment can be planned.[23-25]

Transitional treatment can be obtained by placing stainless steel crowns on permanent and primary molars and strip crowns or direct and indirect composite veneers on anterior teeth.[1,2,25,26] Some authors have also suggested the use of anterior stainless steel crowns with veneered facings.[1,2,23] A new protocol of laboratory-fabricated composite crowns and veneers luted adhesively on primary teeth without previous tooth preparation by rotary instruments has also been proposed.[10] The use of porcelain-fused-to-metal crowns in posterior teeth and porcelain crowns or veneers in anterior teeth are recommended for permanent treatment.[1,2,9,11,12] A successful treatment requires a multidisciplinary approach that may include, besides the pediatric dentist, an orthodontist, a periodontist, an oral surgeon, and a speech therapist.

CASE DESCRIPTION

An 8-year-old girl sought treatment at the Faculty of Dentistry of the Rio de Janeiro State University, Rio de Janeiro, Brazil. Her chief complaint was the yellowish coloration of her teeth, which, according to her mother, made her ashamed of smiling and had a negative influence on her relationship with other children. Her medical history was unremarkable. The clinical examination showed that the primary maxillary canines and maxillary and mandibular molars were present, together with the permanent mandibular canines, maxillary and mandibular incisors and maxillary and mandibular first molars. The gingival tissues around most of the permanent teeth showed mild inflammation and a noticeable enlargement. The enamel surface of the primary teeth was smooth, but the enamel surface of the permanent teeth was rough. The permanent teeth presented a corroded-like appearance and were not completely erupted. Tongue thrusting and anterior open bite were also observed (Figure 1). The patient had no complaints regarding sensitivity to cold or heat. Only the permanent mandibular right first molar presented a caries lesion.

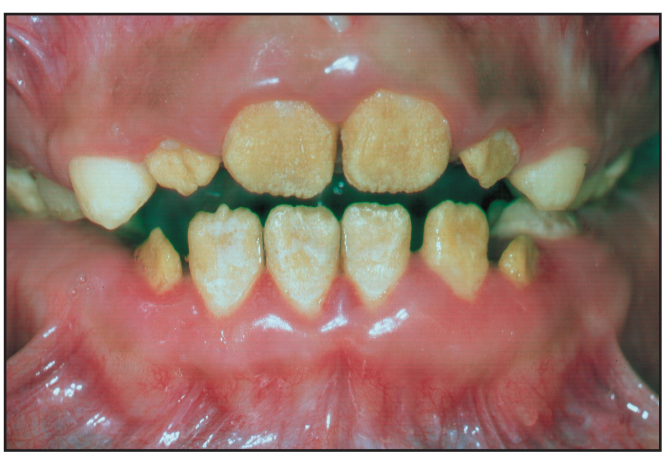

Figure 1. Primary and permanent anterior teeth showing enamel anomaly. Presence of anterior open bite.

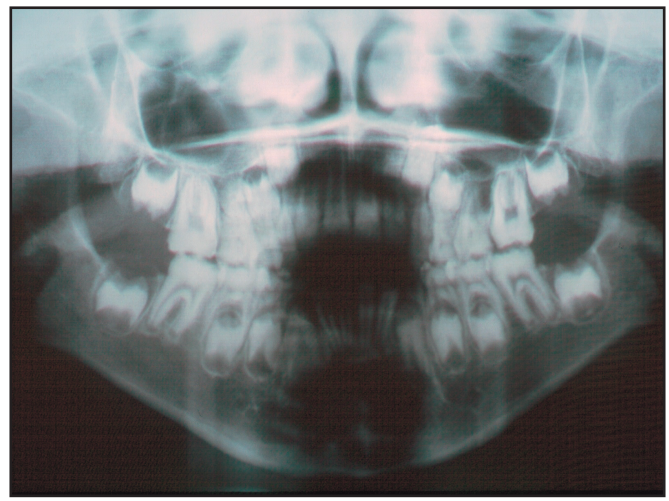

Figure 2. Panoramic radiograph showing agenesis of the maxillary left second premolar.

Radiographic examination comprised periapical and panoramic X rays. The panoramic radiography showed that both primary and permanent teeth had thin enamel, whose radiodensity was higher than that of the dentine. It also detected agenesis of the maxillary left second premolar (Figure 2). In addition, periapical X rays revealed the presence of pulpal calcifications in primary molars. The diagnosis of hypoplastic AI was established based on clinical and radiographic data as well as on the exclusion of other possible etiologic factors, such as tricho-dento-osseous syndrome, dental fluorosis, or acquired enamel defects. Initial treatment consisted of oral hygiene instruction, dietary counseling, impression for study casts, and filling of the carious molar with glass ionomer cement (Vitremer/3M ESPE, USA). Gingivoplasty was undertaken to create a normal and functional gingival shape in the permanent maxillary and mandibular first molars and permanent mandibular incisors.

After a healing period of 30 days, the permanent maxillary and mandibular first molars were restored with stainless steel crowns (Unitek/3M ESPE, USA; Figures 3 and 4), and the permanent maxillary and mandibular incisors received direct composite veneers (Fill Magic/Vigodent, Brazil and Durafill/Kulzer, Germany; Figure 5). A removable orthodontic appliance was used to discourage the habit of placing the tongue between the teeth before and during the act of swallowing. The patient was referred to a speech therapist, but did not undergo treatment. After the first phase of treatment, the patient (Figure 6) and her family were satisfied with the results. The patient's motivation to change oral hygiene behavior was confirmed by the improvement of her gingival health. She was put on a 3-month recall regimen for routine preventive treatment, as the absence of the remaining permanent teeth precluded the performance of any other treatment.

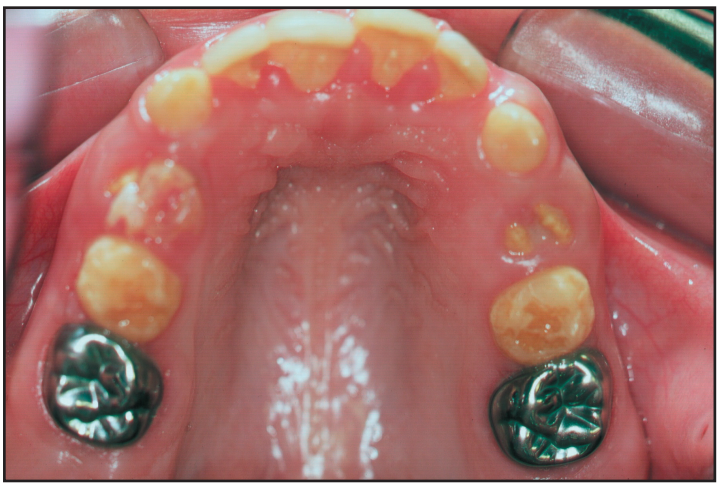

Figure 3. Upper occlusal view after treatment.

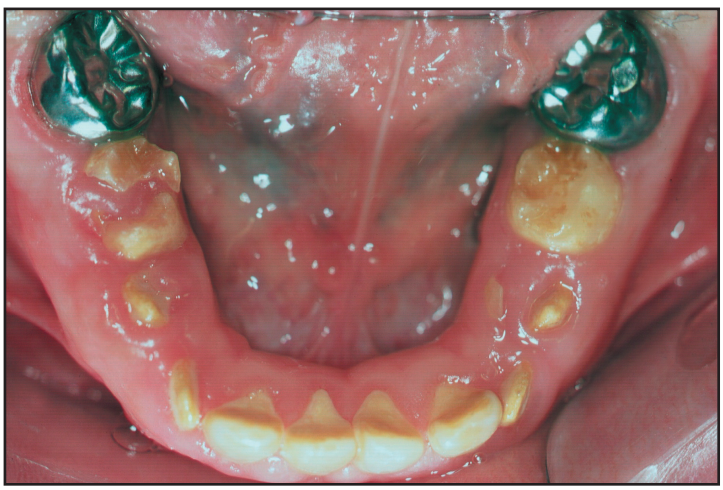

Figure 4. Lower occlusal view after treatment.

DISCUSSION

The most widely accepted classification system for AI is the one described by Witkop in 1989,[1] which divides AI into 3 basic types: hypoplastic; hypomaturation; and hypocalcified. This classification considers the clinical aspects and inheritance pattern and comprises 3 major groups, each with a large number of subtypes. There may be cases in which clinical features of different types of AI coexist. Thus, classifying a patient as having one specific type of AI is a major challenge to the clinician. Some authors have reported that genetic mappings can be a useful tool in identifying to which AI group a patient belongs.[5-8] Although these advances in the diagnosis of AI by molecular and biochemical methods are extremely important, this kind of sophisticated technique is not yet routinely available.

Our patient was diagnosed as having AI because she presented abnormal enamel thickness in the primary teeth and generalized enamel hypoplasia of the permanent teeth and did not have any systemic diseases that could be associated with these defects. The following features suggested that the child belonged to the AI hypoplastic group[1]: thin enamel; different patterns of radiodensity between enamel and dentine; hard enamel surface; the absence of dentinal hypersensitivity; and the presence of pulpal calcifications. In the primary dentition, the enamel defects were mild and could not be easily seen. This may explain why the family sought dental care for the child only after the permanent anterior teeth erupted.

Despite the difficulties related to the classification of the different types of AI, all efforts were made to establish the most accurate diagnosis, since it has a direct impact on the treatment. For example, if the patient had the hypocalcified type, deproteinization with 5% sodium hypochlorite could have been performed.[21,22] As in the hypoplastic type, the concentration of proteins is normal, this procedure was considered unnecessary.

Since the patient's permanent teeth were the most affected but had been recently erupted—thus presenting large pulp chambers—a provisional treatment was adopted to maintain pulp health and conserve the available tooth structure. Moreover, the aims of this first phase of the treatment were to allow for the normal development of the dentition and re-establish esthetics, facial harmony, and chewing function until adulthood, when a permanent treatment could be planned.[23-25] Efforts were also made to provide the patient with a relatively affordable treatment that could decrease her self-consciousness and increase her sociability, since that was her mother's chief complaint. The dental literature shows that AI can have an overwhelming effect on a person's social interaction, self-image, and self-perceived quality of life.[19]

After receiving oral hygiene instruction and dietary counseling, the patient was sent to a periodontist. He confirmed that the presence of gingival hyperplasia had led to a reduction in the cervical-occlusal height of the crowns, and he recommended periodontal surgery. After gingivoplasty, it was possible to make restorations within biologically acceptable cervical limits.[9,11] In the anterior teeth, direct composite veneers were placed to recover esthetics, as suggested by some authors.[20,26] Stainless steel crowns were placed on the permanent first molars to halt attrition of the occlusal surfaces and decrease the chance of loss of vertical dimension in the future, as well as to protect the dentinal-pulp complex from chemical and thermal attacks.[2,25] At that point, the recovery of vertical dimension was not a concern due to the risk of aggravating the anterior open bite.

During the follow-up, a displacement of the stainless steel crown of the permanent mandibular left first molar was observed after 19 months, and a displacement of the composite veneer of the permanent maxillary left lateral incisor was observed after 10 months. These restorative procedures were redone. In the case of the anterior tooth, the unsuccessful restoration may be explained by the thin enamel layer available for bonding.

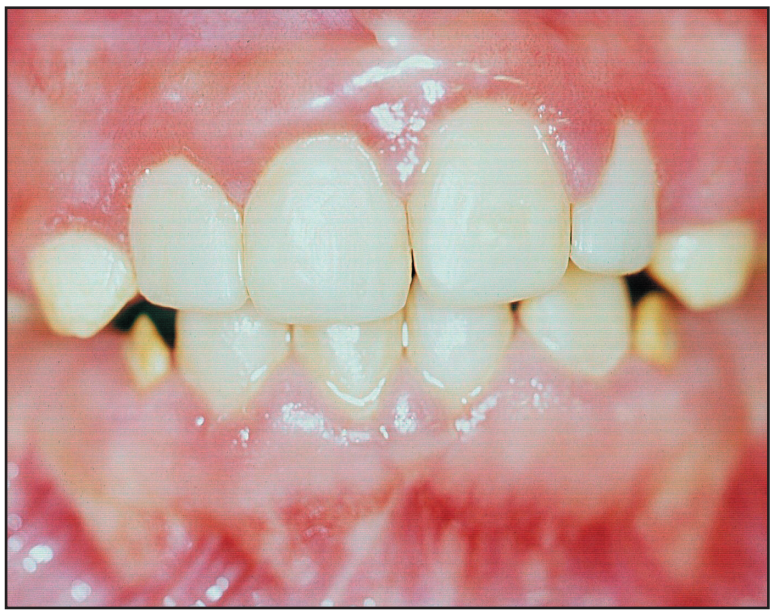

Figure 5. Frontal view after treatment.

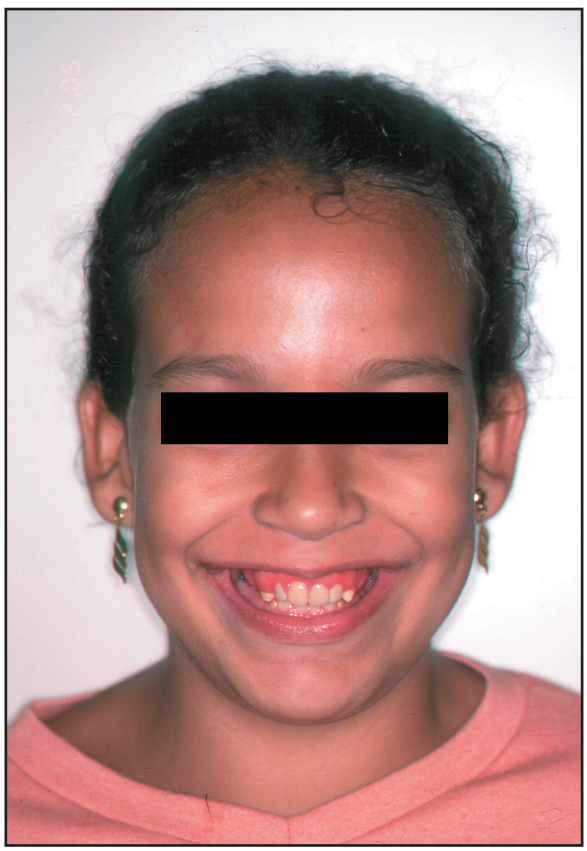

Figure 6. Patient smiling after treatment.

While undergoing restorative treatment, the patient was also examined by an orthodontist to evaluate the anterior open bite. Considering that the patient was still in the transitional growing period and had a tongue thrusting habit, the orthodontist decided to use a removable appliance. Although the patient did not undergo any speech therapy, she wore the appliance properly, and the problem was solved. One study stated that, since the anterior open bite associated with AI is supposed to be of skeletal origin, a multi-segment Le Fort 1 intrusion osteotomy becomes often necessary.[15] This suggests that the patient should be routinely examined by an orthodontist to evaluate whether the correction of the anterior open bite remains stable or if she needs to undergo an additional treatment.

The results achieved suggest that limiting AI treatment in the transitional stage to such simple methods and delaying more invasive treatment until adulthood is highly beneficial to the young dental patient.

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