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Case Report

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Case Report: Child with Amelogenesis Imperfecta

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Abstract

We present a patient with Amylogenesis imperfecta (AI) and unexplained bone and blood abnormalities, challenging existing genotype-phenotype models. The present patient is a 10-year-old girl who was diagnosed with amylogenesis imperfecta with great difficulty and limitations. She underwent temporary composite treatment in a short time and returned to the embrace of her family and society with a beautiful appearance and great self-confidence.

Keywords: Amelogenesis imperfecta, Composite treatment, Child.
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Introduction

Dental anomalies are an important group of dental morphological variations whose prevalence is different in different populations.[1] These anomalies are related to dental development that are caused by genetics, systemic, local or traumatic diseases.[2] These disorders may occur at any stage of tooth development. Disorder in the initial stage leads to hypodensia or extra teeth, disorder in the morphological differentiation stage leads to abnormality in the size and shape of the teeth, and disorder in the stages of tissue differentiation, deposition and mineralization leads to hypoplasia or hypo mineralization such as amylogenesis imperfecta.[2-4] Its exact cause is unknown, but mutations in genes that regulate enamel matrix proteins may play a role in its occurrence by affecting skeletal development. AI is a rare genetic disorder that affects both children and adults. This disorder represents a group of inappropriate developmental conditions of genomic origin that affect the structure and clinical appearance of tooth enamel more or less equally and may be associated with morphological or biochemical changes.[1-5] Various treatments have been proposed for children with amelogenesis imperfecta, but no definitive treatment guidelines have been proposed so far.[5] The current study introduces a 10-year-old child with AI, who has made an acceptable recovery with temporary composite treatment.

Case Introduction

The patient is a 10-year-old girl who was referred to the clinic by a pediatric specialist with a complaint of extensive malformation and untidy of upper and lower teeth, as well as very poor dental hygiene. In the initial examinations, the child had no history of any special disease and no family history of this disease was reported in his family. Also, extraoral examinations did not show disease-related findings. No excessive skin stretching or highly flexible joints were observed. In the first examination, due to the untidy of the teeth and improper occlusion, the patient was very depressed and sad, and even for the examination of the teeth, she was not willing to show them and laugh (she even cried a little), she had low self-confidence, and according to her parents, she became withdrawn and had very limited social contact.

In the examination of the teeth, extensive caries in the upper and lower teeth, complete discoloration and sensitivity of the teeth, diastema between the anterior teeth, small and grooved teeth, rough tooth surfaces, enamel hypoplasia and improper occlusion and improper placement of the teeth on was also observed (Figure 1-3). An oral panoramic radiograph was requested for the patient. In the radiographic examination, the teeth were normal in terms of dentin and root shape, the thickness of the enamel was reduced (its consistency was normal in the probe examination) and all the series of permanent teeth were present and the eruption of the teeth had not been completed. So, dental X-rays showed thin, pitted enamel consistent with AI. After taking the history, performing the examinations and taking the necessary x-rays, the diagnosis of amelogenesis imperfecta of the hypoplastic type was given to the patient. Diagnosis of autoimmune disease is largely clinical and involves observing changes in the shape and structure of the teeth, such as smaller teeth, brown discoloration of the teeth, malocclusion, and tooth sensitivity, which can be confirmed by standard radiographs. Blood tests or genetic testing are usually not required.[4, 5]

Necessary explanations were provided to the patient's parents, and after obtaining their informed consent and agreement, treatment measures were started. Due to the age of the patient and the lack of development of complete both jaws growth and incomplete eruption of the teeth, it was not possible to perform the final treatment and change the occlusion for the patient, and therefore it was decided that in order to control the current condition and especially to improve the patient's appearance, to improve the condition and health of psychological, increasing social relations and most importantly to restore the smile to this beautiful girl, temporary treatment should be used with the necessary considerations, especially strict oral and dental hygiene. Due to the lack of complete bonding of adhesive materials on hypoplastic enamel, it was

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decided to use non-bonded zirconia permanent prosthetic treatments and complete occlusion reconstruction for her at the appropriate age (17-18 years).

At first, impression was done for the patient and a cast was designed, and suitable spaces and mock-ups were formed with composite, and then this mock-up was transferred to the patient's mouth using a silicone index (a-silicone), and then the teeth were repaired as follows.

Teeth were isolated by Split Dam method and hypoplastic enamel surfaces were removed. In order to ensure proper cleaning of enamel hypoplasia, at first burr and sandblaster with aluminum oxide particles of 30 microns and pressure of 1.5 bar were used, and then subgingival cord was used. 37% phosphoric acid was used for 15 seconds to etch the teeth. Then, after applying bonding for 15 seconds, composite was used by layering method and using dentin composite with canister ratio of 0.8 to ensure good tooth bonding. Each tooth was treated with Tokuyama Plephic Astria color OA_1 (for dentine and to cover the discoloration of the teeth) and white enamel (for enamel and coordination of the teeth). In order to ensure a good bond between the composite and the tooth, Fulari SE Portex bonding was used. SE Portex bonding contains chlorhexidine and has the power to clean proteases enzyme that are not cleaned by etching, and therefore increases their bonding power. The teeth were reconstructed in two sessions of about 7.5 hours, and after a week, a session was held for occlusion check-up and final polishing (Figure 4, 5). Then 1 and 3 months after the treatment, follow-up visits were given to the patient and necessary recommendations were provided. According to the observations and reports of the parents, in this short period of time, in addition to the excellent improvement in the appearance of the teeth, the patient's mood, social communication, satisfaction, and quality of life of the patient and her family have significantly improved, and the patient's beautiful smile has become fully revealed (Figure 6).

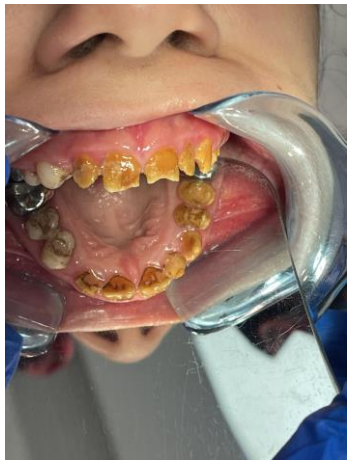

Figure 1. Diagnosis of AI

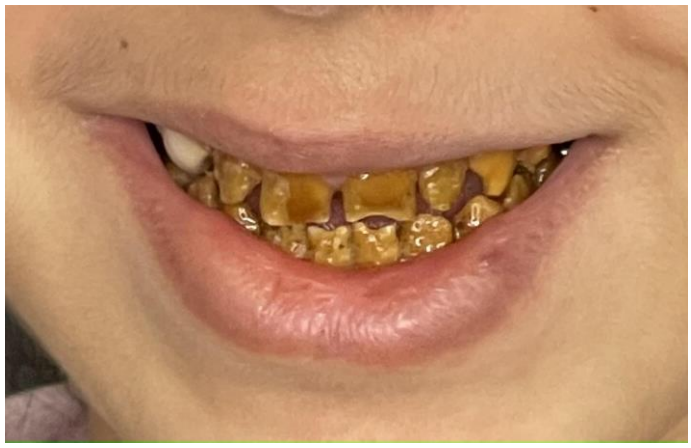

Figure 2. Diagnosis of AI

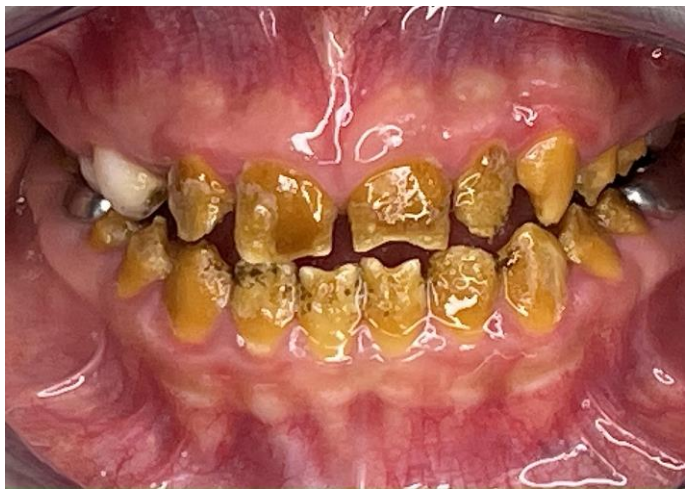

Figure 3. Diagnosis of AI

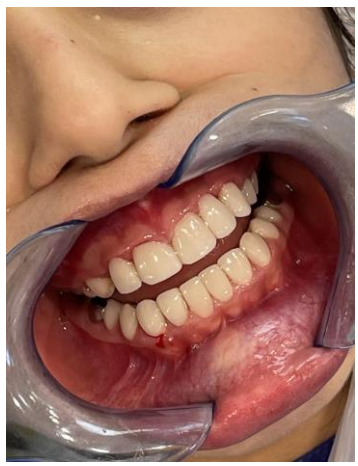

Figure 4. Treatment of AI

Figure 5. Treatment of AI

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Figure 6. Total Treatment of AI

Discussion

Acquired immunodeficiency (AI) is a rare genetic disorder of dental enamel. It causes teeth to be abnormally small, discolored, pitted or grooved, prone to rapid wear and fracture, and other abnormalities.[1, 2] It is seen in both primary and permanent teeth.[3] Review studies have highlighted the association of acquired immunodeficiency with skeletal dysplasia. Based on this association, AI is divided into four categories: hypoplastic (type I), hypomaturity (type II), hypocalcification (type III), and hypomaturity/hypoplasia/tarodontism (type IV).[4] The disorder can be inherited as an X-linked genetic trait, autosomal dominant or autosomal recessive, depending on its subtype.[5, 6]

The most disorder in AI is related to the structure of tooth enamel. Naturally, tooth enamel is composed of highly organized hydroxyapatite crystals that occupy 95% of the matrix volume. Ameloblasts are responsible for the secretion, formation and maturation of enamel matrix using matrix molecules such as enamelin, amelogenin and ameloblastin.[6] It is believed that the diversity of enamel abnormalities in Al reflects the time difference in the occurrence of disorders during AI. Defects that form during the formation of enamel bonds can create a layer of defective enamel that easily separates from the underlying dentin.[7, 8]

Mutations in AMELX, ENAM, MMP20 and FAM83H genes can cause AI.[9] Al can have different inheritance patterns depending on the type of altered gene. In most cases, they are caused by mutations in the FAM83H gene and are inherited through an autosomal dominant pattern.[10] It may also be inherited through an autosomal recessive pattern, which can be caused by a mutation in the ENAM or MMP20 gene. In about 5% of cases, Al is caused by a mutation in the AMELX gene and is inherited through an X-linked pattern. Other cases of AI are caused by new gene mutations and occur in people who do not have a family history of the disorder.[10-12]

The exact incidence of amelogenesis imperfecta is uncertain and related estimates range from 1 in 700 people in northern Sweden to 1 in 14,000 people in the United States, of which about 40% have the predominant hypoplastic type.[1, 12, 13] The autosomal dominant and recessive forms of this disorder affect men and women in relatively equal numbers, but the X-dominant type of this disorder affects men twice more than women.[11, 12] The probability of AI disease in siblings of an affected person is less than 10% and usually does not follow a specific pattern.[13]

AI is identified by defective or missing tooth enamel. The secondary effects of this disorder may include cracking, early decay, or tooth loss and susceptibility to various diseases of the tissues surrounding the tooth (periodontal), such as the gums, ligaments, and alveolar bones where the tooth root is located.[14] Also, teeth become sensitive when exposed to heat or cold. In the hypoplastic type, the individual's teeth have small to normal crowns so that the upper and lower teeth do not overlap and do not touch each other. Their color varies from yellowish white to brownish yellow. The thickness of tooth enamel ranges from thin and polished to normal with numerous grooves, lines or holes. In the type of hypo maturation, the teeth are usually white, yellowish or brown, which may be accompanied by sensitivity, pain and burning. Tooth enamel is generally of normal thickness, but is prone to chipping or scratching. In the hypocalcified type, patients have white, yellowish to brown rough teeth, which are probably associated with sensitivity, pain and burning. Although tooth enamel is generally of normal thickness, it is prone to chipping or scratching. In the type of hypo maturation/hypoplasia/taurodontism, the teeth are smaller than normal and their color varies from white to yellow-brown. The teeth are mottled or stained in appearance. Tooth enamel is thinner than normal and parts of them are pitted.[2, 4-6]

Diagnosis of AI is usually made by visual examination, family history and x-ray findings at the time of tooth eruption. Using a simple manual tool to detect different types of Al at the

age of one to two years can be helpful. Normal lab tests do not rule out a syndrome; follow-up is needed for late-onset symptoms.[15] X-rays can reveal the presence of teeth that never erupted or have been absorbed by them, as well as the contrast between enamel and dentin. The most common differential diagnosis for this disorder is dental fluorosis, which can range from pale white streaks to deep white discoloration with malformed areas of pigment and hypoplasia. A history of excessive fluoride intake due to habits such as toothpaste use in childhood or high fluoride levels in local water supplies can aid in the diagnosis.[15] Other differential diagnoses include Ehlers-Danlos syndrome, which is less likely to be confirmed in this case because of the lack of joint flexibility and skin elasticity.[16]

There is no specific and fixed guideline for the treatment of these patients. The treatment of this disease depends on the type of AI, the age of the affected person, the type and severity of the tooth enamel abnormality. Clinical treatment aims to access the beautiful appearance of damaged teeth, reduce dentine sensitivity, preserve tooth structure and optimize chewing performance. Clinicians should consider systemic evaluations for patients with AI and atypical features.[17]

Pediatric veneers and resin-based bonding should be preferred in primary teeth. In permanent teeth, non-invasive or minimally invasive dentistry should be the first choice to use a treatment gradient from minimally invasive options to prosthetic treatments.[18] Preventive measures, types of veneers, implants, full crown restoration and a type of prosthesis that covers defective teeth and corrects open bite are known as excellent treatments for this disorder. Hypoallergenic toothpaste can prevent painful sensitivity to heat and cold, and genetic counseling is recommended for families of children with AI.[18, 19]

In the present patient, according to the age conditions and also the growth rate of the jaws and teeth, temporary composite treatment was used with success and full satisfaction, so that while solving the physical and mental problems of the child, the social problems and obstacles and especially the educational obstacles of the patient were reduced until at the age of above, permanent prostheses should be used for definitive treatment. It is hoped that the treatment used in all similar children can be used successfully.

Limitation

Lack of genetic confirmation limits definitive syndrome classification.

Ethical Considerations

This article is a part of the results of the thesis for the degree of Doctor of General Dentistry, which has been registered with the Ethics Committee of Golestan University of Medical Sciences under the ethics code (IR.GOUMS.REC.1403.170). Verbal and written consent was obtained from the child's parents. No additional costs were imposed on them and their right to discontinue the study was guaranteed.

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Conflict of Interest

The authors declared that they have no conflict of interest.

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