Amelogenesis Imperfecta with Taurodontism, Microdontia and Minor Thalassemia: A Case Report

Fatemeh Mazhari[1], Negar Mokhtari Amirmajdi[2]

[1] Dental Material Research Center, Faculty of Dentistry, Mashhad University of Medical Sciences, Mashhad, Iran

[2] Department of Pediatric Dentistry, Facultyof Dentistry; Mashhad University of Medical Sciences, Mashhad, Iran

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Abstract

Amelogenesis imperfecta is a group of genetic disorders that affects both the morphology and quality of tooth structure. Although the disease entity is primarily associated with abnormalities of dental and oral structures, it has been reported to be associated with a few syndromes. A 9-year-old girl with minor thalassemia referred to the Department of Pediatric Dentistry of the Mashhad Faculty of Dentistry with a complaint of sensitivity of first permanent molars. Dental findings consisted of amelogenesis imperfecta, microdontia, posterior cross bite and taurodontism. This is the first report of thalassemia accompanied with amelogenesis imperfecta. Although the patients often are non-symptomatic, the trait can be passed on to a child and if both parents carry the trait, the child could develop a more severe form of the disease; therefore, early diagnosis is important.

Key words: Amelogenesis imperfecta, Minor thalassemia, Taurodontism, Microdontia.

Introduction

Amelogenesis imperfecta (AI) is a group of genetic disorders that affects teeth. Depending on its type, both the morphology and quality of tooth structure may be altered. It can affect primary and/or permanent dentition (1). The genetic pattern responsible for the disease may be autosomal dominant, autosomal recessive, sex linked or even sporadic (2).

The primary site of involvement in AIis the dentition. However, other oral findings may also be associated with the condition. These include dental and skeletal open bite, pulpal calcification, dentin dysplasia, taurodontism, crown and root resorption, unerupted teeth, congenitally missing teeth, gingival hyperplasia and follicular hyperplasia (3). Although the disease entity is primarily associated with abnormalities of dental and oral structures, it has been reported to be associated with a few syndromes (4-6).

Thalassemia refers to genetic disorders in globin chain production. In individuals with beta thalassemia, there is either a complete absence of globin production (thalassemia major) or a partial reduction in globin production (thalassemia minor) (7).

This article presents a girl with hypoplastic AI who also suffers from minor thalassemia and microdontia accompanied with taurodontism and bilateral posterior cross bite.

Case Report

A 9-year-old female patient referred to the Department of Pediatric Dentistry of Mashhad Faculty of Dentistry with a chief complaint of sensitivity of her first permanent molars. She was first seen by a general dental practitioner for restoration of her primary teeth. Her physical examination was normal but her medical history revealed minor thalassemia. The patient was

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treated symptomatically with folic acid and ferrous sulfate supplements. She was born at full-term after a normal pregnancy and uncomplicated delivery and her post-natal period was uneventful. Moreover, she was of normal development and intellect and there was no consanguinity within the family. Family history showed the same disease with same dental manifestations in one of the older patient's sisters. The patient's mother was examined and showed unaffected permanent dentitions. Although the patient's father refused an examination, he had very small teeth without any tooth discoloration according to his wife. The patient had two healthy older brothers as well as another healthy older sister. The patient lived in a non-fluoridated area and had never taken fluoride supplements. In the intraoral examination, soft tissue had a normal appearance. The timing of tooth eruption and the emergence pattern seemed to be within the normal range. The patient's oral hygiene was poor. Dental findings included microdontia, enamel hypoplasia in all primary and permanent teeth and posterior cross bite. There was no space between teeth (Figs. 1 and 2).

The panoramic radiograph showed all permanent teeth including molars. Taurodontism was observed in four permanent first molars. There was also a thin layer of enamel with normal density covering the dentin (Fig.2).

Based on clinical and radiographic findings, a diagnosis of hypoplastic AI was made and appropriate treatment wasplanned based on the patient's needs. Treatment plan for this case consisted of oral hygiene instructions, prophylaxis, topical fluoride therapy and restorative treatment. Due to the sensitivity of the first permanent molars and their susceptibility to further structure loss caused by defective tooth texture and as there was still a long period of growth remaining, all the mandibular and maxillary first permanent molars were treated by removing caries and restoring with temporary full coverage stainless steel crowns (SSC) (3M ESPE).Due to their small size, SSCs of the second primary molars (no. 2) were used. Maxillary left primary molars underwent pulpotomy and restored with SSC. The patient refused treatment of her maxillary right primary molars and the cross bite (Fig.3).

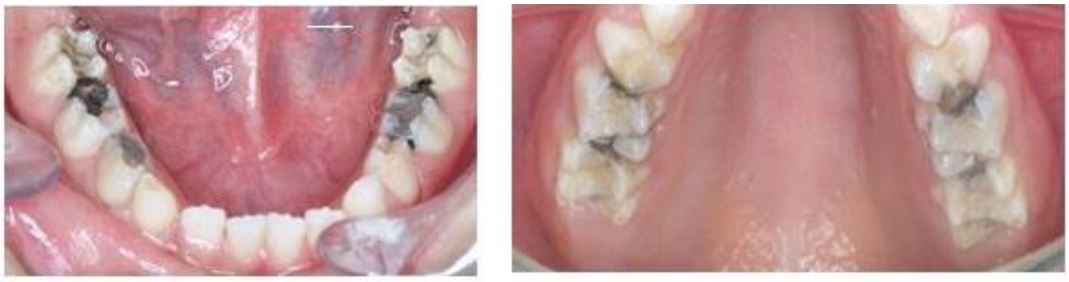

Figure 1. Intraoral photographs of the patient before treatment
a) Occlusal view of the mandibular arch, b) Occlusal view of the maxillary arch

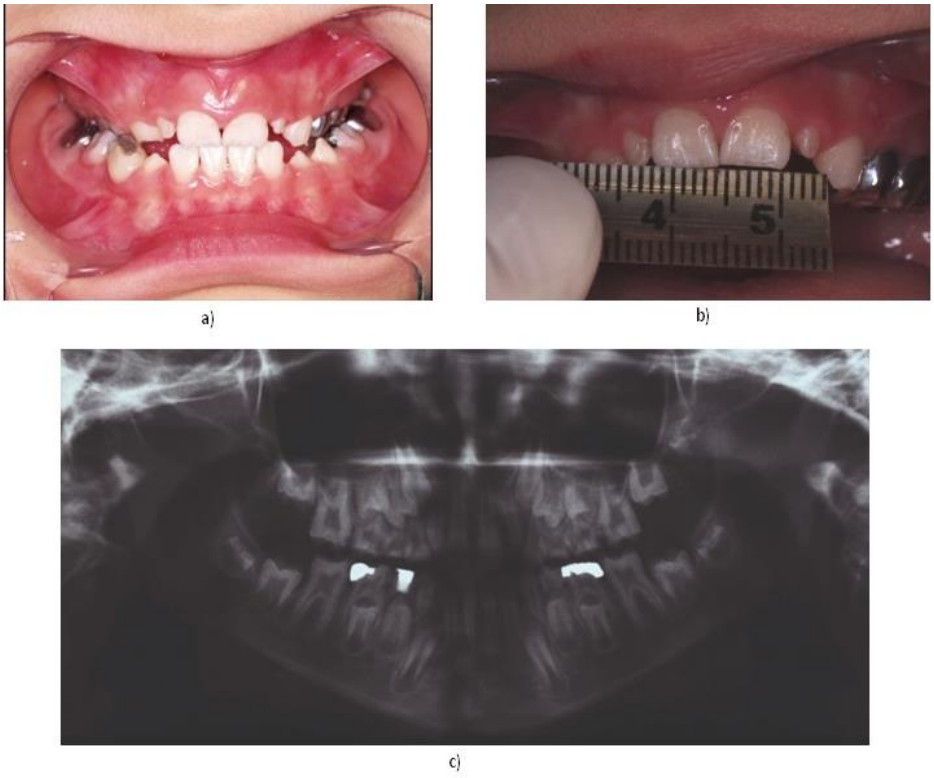

Figure 2. Dental anomalies present in the patient
a) Posterior cross bite b) Microdontia c) Taurodontism and enamel hypoplasia

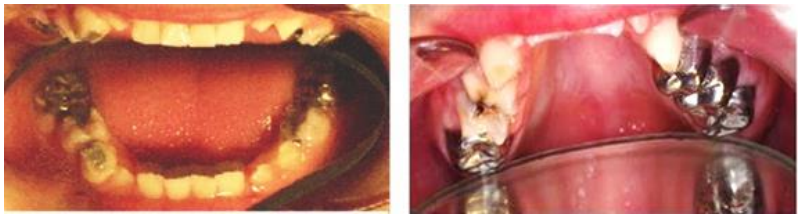

Figure 3. Intraoral photographs of the patient after treatment (The mandibular primary molars had already been restored by a general dental practitioner) a) Occlusal view of the mandibular arch, b) Occlusal view of the maxillary arch

Discussion

AI is a disease with hereditary origin that affects the enamel of primary and permanent teeth. It is reported to have an incidence of 1 in 718 to 1 in 14,000, depending on the population studied (8).

It may cause different signs and symptoms depending on the type of the disease. In the hypoplastic type, the enamel is well-mineralized but its thickness varies from thin and smooth to normal thickness with grooves, furrows and/or pits. The enamel is hard but because of its thinness it is more susceptible to fracture.Radiographs exhibit thin radiodense enamel. The hypocalcified type shows softened and easily detachable enamel. Radiographically, enamel has a contrast similar to or even less than that of the dentin. In the hypomaturation type, the affected teeth exhibit enamel with a mottled, opaque white-brown-yellow color, which is softer than normal but not as soft and easily abraded as in the hypocalcified type. In radiographs, the thickness of enamel is normal, but its density is the same as that of the dentin (9).

In this case, as the enamel had normal hardness but its thickness was less than normal and it was pitted, the diagnosis of pitted hypoplastic type was made.

Association of AI with a group of dental anomalies such as dental and skeletal open bite, pulpal

calcification, dentin dysplasia, taurodontism, crown and root resorption,unerupted teeth, congenitally missing teeth, gingival hyperplasia and follicular hyperplasia has been reported (3). The patient had a posterior cross bite and taurodontism, but other mentioned anomalies were not detected. Also, microdontia was observed. In some types of AI the teeth with thin enamel are also small in size and the interproximal contacts are open; however, in this case there was a true microdontia and the teeth appeared smaller than expected, maxillary laterals were peg-shaped and the contacts were closed. The closed contacts were probably due to a smaller than normal maxilla, which also had resulted in a cross bite. For restoration of the permanent teeth with SSC, even SSCs no. 2 of second primary teeth was slightly loose for them and required a greater amount of crimping to fit.

AI occasionally occurs in conjunction with syndromes such as Kohlschutter syndrome[5], Platyspondyly with AI (10), nephrocalcinosis (1), Jalili syndrome (11), tricho-dento-osseous (2) and Heimler syndrome (4). None of the signs and symptoms of the mentioned syndromes were detected in the presented case. However, medical history revealed presence of beta-thalassemia minor. Based on genetic and clinical features, thalassaemias are classified as homozygous, heterozygous or compound heterozygous. Thalassemia minor is the heterozygous form of the disease, which is mild and usually asymptomatic; the only manifestation is hypochromic microcytic anaemia. The homozygous form of b-thalassemia named thalassemia major exhibits the most severe clinical symptoms with marked orofacial deformities (12). This patient received folic acid and ferrous sulfate supplements. One of her older sisters also suffered from thalassemia minor and AI. However, the presence of AI was only reported by the mother (according to her mother's statement, the older sister's teeth had the same appearance as the patient).

The treatment plan for cases of AI is related to many factors such as the severity of the disorder, the age of the person, the socioeconomic status and the intraoral condition. A conservative option is restoring with direct composite that causes the least tissue loss due to preparation methods. Another relatively conservative approach is amalgam restoration which causes less structure loss than full coverage restorations (13). In this case, because of generalized pitting on the teeth surfaces and teeth sensitivity, complete coverage was considered. These teeth required full coverage with preformed crowns until the end of growth when precision cast crowns can be provided. Another consideration in patients with AI is esthetics and the consequent psychological complications (13). In this case,as the anterior teeth were almost normal in color and shape except for a few pits on labial surfaces, esthetics was not a concern for the patient. The treatment plan focused on restoring the posterior primary and permanent teeth for resolving the sensitivity and preventing tooth loss.

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Corresponding Author:
Negar Mokhtari
Faculty of Dentistry
Vakilabad Blvd, Mashhad, Iran
Tel: 0098-915-3169692
Fax: 0098-511-6090082
E-mail: alazkoo@gmail.com