LUPINE PUBLISHERS

Interventions in Pediatric Dentistry Open Access Journal

DOI: 10.32474/IPDOAJ.2022.08.000279

ISSN: 2637-6636

Review Article

Amelogenesis Imperfecta in Children

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Received: September 28, 2022 **Published:** October 10, 2022

Abstract

Amelogenesis imperfecta or enamel defect refers to a group of genetic conditions in which tooth enamel cannot form properly. Evolutionary disorders of Amelogenesis imperfecta disrupt the structure and appearance of enamel in both deciduous and permanent teeth. People with this defect have small, yellow, or brown teeth that are prone to damage or breakage.

Keywords: Amelogenesis imperfecta; enamel defect; genetic condition; deciduous and permanent teeth; teeth discoloration

Introduction

Amelogenesis imperfecta (AI) is a group of hereditary defects of tooth enamel that exhibit clinical and genetic diversity which affects both the deciduous and permanent dental systems [1]. The incidence of this condition is estimated to be 1 in 14000 individuals [2]. In the mild form, it changes the tooth color, while in the severe form; the hypomineralized enamel wears out shortly after growth. Various changes are seen in tooth enamel, from lack of enamel formation to defects related to mineral and protein content [3]. In addition to enamel defects, abnormalities in dental eruption, anterior open bite, and taurodontism may be seen [4]. In patients with AI, their chewing function is impaired due to dental sensitivity, and crown shortening clinically due to attrition or incomplete tooth eruption is seen [5]; however, the teeth are normal in terms of dentin and root shape [6]. Enamel hypoplasia manifests in a variety of ways depending on the type of AI, with pitting and grooves defects [2]. Therapeutic goals include reducing the pain of tooth sensitivity, maintaining tooth structure as much as possible, and improving the teeth appearance due to the psychological impact of this disorder in reducing self-esteem [7]. Amelogenesis imperfecta is phenotypically divided into 4 types: hypoplastic (type I), hypomaturation (type II), hypocalcified (type III), and hypomaturation-hypoplasia with taurodontism (type IV).

Causes of Enamel Defects

This disorder results from mutations in the ENAM, AMELX, or MMPO2 genes. In one study, Lagerstrom and colleagues stated that mutation of AMELX had been attributed as a cause of Amelogenesis imperfecta [8]. Kim and colleagues found out the Mutations in MMP20 could lead to autosomal recessive hypomaturation Amelogenesis imperfecta [9]. Besides, another research revealed that Autosomal recessive inheritance has also been documented for ENAM mutations [10]. These genes are responsible for making the proteins needed to make tooth enamel. A mutation in any of these genes can block the production of protein, and lead to the formation of enamel that is thin or soft [11]. Mutated genes can be passed from parents to their children, or the mutation can cause the disorder in a person without a family history.

Classification

Type I- Hypoplastic

It is characterized by a defect that occurs in the amount of formed enamel. In other words, the enamel is strong but not as enough strong as normal teeth. The teeth are small and may be white, yellow, or brown, and the enamel is relatively thin. Besides, the teeth may have random pits and grooves [11,12]. Most cases of enamel formation defects are of type 1.

Type II- hypomaturation

This type of AI is commonly associated with an open bite. The creamy white to yellow-brown color can be seen on the rough-surfaced teeth. On the other hand, the teeth may be sensitive. The enamel has a normal thickness, but it is very soft so that the teeth can be full of stains and have a mottled appearance. Unfortunately, teeth tend to be chipped off; or even they can be scraped easily [11,12].

Type III- Hypocalcified: In this type, the defect occurs in the early stages of tooth enamel formation. Tooth enamel can be normal in thickness, but it is very soft and tends to be chipped away or scraped off. Tooth discoloration can be seen in white, yellow, or brown; and the tooth surface may be rough. Besides, teeth may react to cold and heat and become sensitive. The prevalence of this type of defect is less than type 1 and type 2 [11,12].

Type IV- hypomaturation/hypoplasia/taurodontism

This type of Amelogenesis imperfecta usually is characterized by a small size of teeth. The color may range from white to yellow-brown. Teeth appear to be mottled or spotted, and the surface can be covered by pits. The enamel is thinner with areas that have less density [11,12].

Amelogenesis Imperfecta vs. Dentinogenesis Imperfecta

AI and DI are both genetic disorders of tooth development. DI is a type of dentin dysplasia that causes teeth to be discolored (blue-gray or yellow-brown color) and gives a translucency appearance to the tooth [13]. In both conditions, teeth are also weaker than normal, making them prone to rapid wear, breakage, or even loss. These problems can affect both primary teeth and permanent teeth. Dentinogenesis imperfecta is caused by mutations in the DSPP gene (dentine sialophosphoprotein gene) [14]. Although genetic factors could be the main contributor factors, any environmental or systemic disturbance that inhibits calcium metabolization can also result in unusual dentine [15,16].

Related Disorders

Symptoms of the following disorders can be similar to those of AI. Many Syndromic conditions affect enamel formation, but for our purposes, comparisons of the following syndromes may be useful for a differential diagnosis.

Tricho-dento-osseous (TDO) Syndrome

TDO syndrome is one of a group of congenital disorders known as ectodermal dysplasia. The condition primarily affects hair and teeth. A person with TDO syndrome may have AI as well. All individuals with this syndrome have dental abnormalities that affect both the primary and permanent teeth. The tooth enamel has the appearance of hypoplasia and hypomineralization [17]. As a consequence, the enamel may be abnormally thin, soft, and pitted, and usually has discoloration [18]. Teeth hypersensitivity has been reported [19]. Both the primary and secondary molars may be abnormally shaped, and pulp chambers may be abnormally large (taurodontism) [20,21]. Also, many teeth may have unusually short, open roots [17,18]. Due to improper tooth development, TDO patients may suffer from high rates of dental caries causing a dental abscess [18,22] which leads to swelling and pain. Some affected individuals may also have widely spaced teeth. Decreased tooth width, premature, attrition, or delayed tooth eruption has also been reported [18,22,23]. Children with this disorder may have to wear dentures. Hypoplastic-hypomaturation subtype is common in TDO patients, but hypomaturation-hypoplastic is less common in individuals with TDO [24].

Axenfeld-Rieger syndrome (ARS)

This syndrome is a rare genetic disorder characterized by dental abnormalities and eye anomalies. The main oral feature is oligodontia, which varies from a single missing tooth to multiple missing teeth [25,26]. Dental abnormalities are including hypodontia, Microdontia, mild craniofacial abnormalities, and cone-shaped teeth. Other abnormalities are enamel hypoplasia, taurodontism, delayed eruption, and shortened roots [27,28].

Management Strategies

AI has a very negative effect on the chewing function, psyche, and appearance of the patients. Therefore, it could make them avoid appearing in a social gathering and lead to increasing stress and decreasing their self-confidence. Dental treatment in these patients will be challenging [29]. There is no standard formula for successful treatment in these patients [30]. Treatment choices depend on several factors: age, the severity of the disorder, socioeconomic status, and level of patient cooperation [31]. Prevention aspects in the period of deciduous and mixed teeth include dietary recommendations, fluoride supplements, and health instructions. Topical fluoride can be applied during the period of permanent teeth. Restorative aspects in the period of deciduous teeth include the restoration of glass ionomer and SCC (30). Oral hygiene is difficult for these patients due to allergies while brushing. Using lukewarm water to rinse the mouth can help. Regular use of fluoride mouthwash can help reduce allergies and caries [32]. Root canal therapy and cosmetic veneers are effective for decayed teeth to achieve both function and beauty [33]. Treatment of these patients in childhood and early adolescence focuses on the prevention and preservation of deciduous and permanent teeth.

There are few studies on long-term follow-up of patients treated with different therapies. Non-invasive and reversible treatment with composite resin should be considered before other destructive treatments. The use of composite provides beauty that is very important to patients; It also preserves tooth tissue. Some dentists avoid composites restorations due to their staining, and technical sensitivity. Staining can be treated with regular polishing. If necessary, the surface layer can be removed, and the restoration restored with a new layer. If the fractured edge occurs in composite restorations, it can be easily restored. In young people, the exposed dental structure at the gingival margin can be easily covered

by restorative restorations. These advantages make the use of composite more economical and biologically more cost-effective than other aggressive and expensive restorations. Composite resins are the first choice of restorative material for all patients, especially patients in early adolescence. Invasive techniques including a full-coverage crown will be helpful for the patient in older ages when the pulp has receded, and the gingival surface is fixed [34]. AI causes psychological distress for the patient due to aesthetic disorders. Therefore, proper diagnosis and treatment not only can improve the patient's quality of life and motivate the patient to maintain oral hygiene in the long run, but also it can be effective in maintaining improved dental conditions.

Conclusion

Amelogenesis imperfecta (AI) is a hereditary disorder that affects the enamel of primary and permanent teeth. Affected teeth appear with discoloration. They are more susceptible to dental caries, tooth attrition, calculus accumulation, and gingival hyperplasia. AI patients have similar oral complications including teeth sensitivity, poor dental aesthetics, and decreased occlusal vertical dimension. They may have other dental anomalies such as open bite, multiple impacted teeth, congenitally missing teeth, and taurodontism. Amelogenesis imperfecta is divided into 4 types: hypoplastic, hypomaturation, hypocalcified, and hypomaturation-hypoplasia with taurodontism. Mutations in several genes are responsible for causing Amelogenesis imperfecta. There are a great number of alternatives for the treatment of Amelogenesis imperfecta. Analyzing the benefits and limitations of each technique will help the professional decide on the best treatment plan.

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DOI: 10.32474/IPDOAJ.2022.08.000279