

PROSTHODONTIC MANAGEMENT OF PATIENTS WITH AMELOGENESIS IMPERFECTA: A CASE REPORT WITH REVIEW OF LITERATURE

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ABSTRACT:

Amelogenesis imperfecta (AI) represents a group of developmental conditions, genomic in origin, which affect the structure and clinical appearance of enamel of all or nearly all the teeth. The enamel may be hypoplastic, hypomineralised or both and teeth affected may be discolored, sensitive or prone to disintegration. The condition presents problems of socialization, function and discomfort but may be managed by early intervention, both preventively and restoratively, with treatment continued throughout childhood and into adult life. When an individual with amelogenesis imperfecta presents with malocclusion, it is an orthodontic concern as etching is compromised. Presented here is a case of AI treated in a multidisciplinary approach. The patient was rehabilitated with full-mouth zirconium oxide ceramic fixed bridges. Adaptation of the temporomandibular joints and masticatory muscles to the bridges was carefully observed over 3 years. At the end of this follow-up period, the patient was satisfied with the esthetics, function and phonation of her prostheses.

Keywords: Amelogenesis imperfecta (AI), enamel abnormality, prosthetic rehabilitation, porcelain.



INTRODUCTION:

Amelogenesis imperfecta (AI) is a hereditary defect of enamel affecting both the primary and permanent dentition [1]. The formation of enamel is a multistep process, and enamel defects can occur at any one of those steps. By definition, AI includes only those cases where enamel defects occur in the absence of other syndromes or metabolic disorders [1]. It is a clinically and genetically diverse group of conditions caused by mutations in genes critical for normal enamel

formation, mineralization, and maturation. The incidence of AI ranges from 1 in 718 to 1 in 14,000 depending on the population studied. Changes in color, thickness, hardness, and smoothness have been observed in the enamel of teeth affected by AI, depending on the type and severity of the disorder.^[1,2]

According to Witkop, AI can be classified as hypoplastic, hypomaturation, hypocalcified, and hypomaturation-hypoplastic with taurodontism [1]. In hypoplastic AI, the teeth are yellowish

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brown in color, rough in texture, and widely spaced. In hypomaturation AI, the clinical crowns are of normal size and contact adjacent teeth, but the mottled, brown-yellow enamel is soft. In hypocalcified AI, the enamel layer may be of normal thickness, but is rough and soft and wears away quickly following tooth eruption. In hypomaturation-hypoplastic AI with taurodontism, the enamel is mottled white-yellow-brown in color and is thin at the areas of hypomaturation. The permanent molars associated with this condition have taurodontism. In addition, other teeth may also have enlarged pulp chambers.^[1,2,3]

Although AI, by definition, affects only the enamel formation, it has multiple consequences for affected patients. Often these patients experience difficulty in maintaining oral hygiene, decreased masticatory function, and a lower self-esteem, which significantly affect their over-all quality of life ^[4]. Furthermore, most variants of AI require extensive dental treatment, which can be time consuming and often poses a significant economic burden on their family. The clinical management of a growing child with AI at any given developmental stage may present great challenges to the patient, their parents, as well as to the oral health professionals involved. Clinicians must therefore consider treatment alternatives to balance the patient's esthetics and functional needs, the status of patient's growth and development, the financial implications for the patient's family, and the long-term prognosis.

Treatment of AI depends on the individual's specific diagnosis and phenotype. Case reports have presented different strategies including: the use of glass ionomer cements, composite resin, stainless steel crowns, lab-fabricated crowns, and even multiple extractions necessitating an overdenture. Unfortunately, research on long-term follow outcomes of patients with AI is particularly scarce. The majority of evidence relies on case reports that present treatment modalities and outcomes of only a few AI patients with or without an additional description of their family members. It is surprising to note that there is currently no standard of care established for managing patients with AI, especially during the mixed dentition stage.^[5,6]

Genetic Etiology: The different clinical manifestations of AI have specific gene mutations associated with each phenotype. Mutations in four candidate genes have been proven to cause AI: amelogenin, enamelin (ENAM), kallikrein4 (KLK4) and enamelysin (MMP-20). Mutations in the AMELX gene encoding for the amelogenin-protein cause most of the X-linked hypoplastic AI (Kim *et al.*, 2004). Depending on the specific mutation, the phenotype associated with AMELX mutation can be smooth hypoplastic, hypocalcified, or hypomaturation. The ENAM mutations encoding for the enamelin protein result in an autosomal dominant or recessive hypoplastic AI with the phenotype ranging from relatively minor, localized enamel pitting to severely hypoplastic enamel ^[1,5].

Mutations have been reported in the *KLK4* and *MMP-20* genes which code for the kallikrein and enamelysin proteinases cause a hypomaturation AI that is transmitted as an autosomal recessive trait . These gene mutations, however, account for only a quarter of all AI cases .

The hypomaturation-hypoplastic with taurodontism AI (AIHHT) is a variation of tricho-dento-osseous syndrome (TDO). The principal clinical features of TDO include kinky hair at birth, osteosclerosis, brittle nails, enamel hypoplasia, and taurodontism. Mutations in the distal-less homeobox 3 (*DLX3*) genes cause TDO . Price *et al.* reported that AIHHT is a distinct condition and not due to a *DLX3* mutation. . However, Dong *et al.* reported a case of TDO syndrome caused by a 2-bp deletion in the *DLX3* that was classified as AIHHT . This 2-bp mutation was later identified to be the causative factor of a family with TDO. It is likely that the clinical diagnosis of a family in Dong's study should have been TDO instead of AIHHT. As more family members with AIHHT and TDO are investigated, the genotype and phenotype correlation of families with *DLX3* mutations may be better demonstrated.^[5,6,7]

Recently, Lee *et al.* identified mutations in family with sequence similarity 83 member H (*FAM83H*) gene responsible for autosomal dominant hypocalcified amelogenesis imperfecta. Unlike other genes that cause AI, *FAM83H* does not encode an enamel matrix protein. Its location inside the ameloblast and its function are completely unknown.

Mutations of *FAM83H* gene account for another 25% of the AI cases, indicating that more AI candidate genes still need to be identified. Identification of mutation genes and cataloging mutations under different types of AI will provide a better understanding of enamel anomalies.^[8,9]

Clinical of AI: Although AI primarily affects enamel formation, a variety of clinical implications may also be present, such as low caries susceptibility, rapid attrition, excessive calculus deposition, and gingival hyperplasia^[6].

The severity of clinical problems varies with each type of AI. Low caries susceptibility has been reported in children with severe hypoplastic and hypomineralized AI^[6,7]. While Sundell stated that the bacteriological and salivary data in the AI patients were inadequate to explain low caries susceptibility, it was suggested that additional investigations which focus on determining the difference of oral microflora between affected and unaffected individuals would be informative. Sundell also speculated that atypical crown morphology with less dramatic fissures, loss of proximal contacts, and rapid attrition commonly associated with hypoplastic AI teeth may contribute toward low caries susceptibility^[8,9].

Rapid and excessive calculus formation has been reported as a common finding related to the hypomaturation and hypocalcified types of AI^[10]. In a review, Wright speculated that the factors contributing to excessive calculus accumulation may include: a rough

enamel surface, altered salivary flow rate, composition, oral hygiene abilities occurring secondary to dental sensitivity, and altered oral microflora [10]. However, there was no evidence provided to support his theory. Sundell did observe that the saliva secretion rate, pH and buffer capacity from AI individuals corresponded to children without AI. Moreover, the gingival condition and oral hygiene among patients with AI were reported to be poor. It can be assumed that atypical tooth morphology and poor oral hygiene may accelerate plaque accumulation or increase tooth sensitivity, posing challenges for dental care providers.[10,11]

Patients with AI are also affected by their poor esthetics, tooth sensitivity, and decrease of occlusal vertical dimensions through loss of tooth structure [3]. AI patients may experience compromised chewing function due to tooth sensitivity and the short clinical crowns caused by attrition and/or incomplete eruption. Unfortunately, restorative treatment for patients with AI is not often provided at an early age due to issues relating to tooth sensitivity, difficulty in managing extensive treatment needs, and even cost. There is a tendency to adopt a “wait-and-see” policy, often resulting in the development of deep bite and deleterious tooth wear. The resultant deep bite, short clinical crowns and altered mesiodistal dimensions of teeth complicate treatment considerably.[3,6,10]

Growth and Development: AI is a diverse group of genetic disorder primarily

affecting the quality and/ or quantity of enamel. Non-enamel-related manifestations may also occur, including an open bite malocclusion, accelerated dental development, high prevalence of dental impaction, congenital missing teeth, crown and/or root resorption, pulp calcification, and associated abnormalities^[3,6,7] Open bite malocclusion has been reported to be associated with AI.

Children with AI may also exhibit accelerated tooth eruption when compared to the unaffected population. Seow found that all subjects with AI regardless of variants showed a significant acceleration of dental age of approximately 1.13 + 0.78 years compared with children in the control group [3].

Oral pathologic findings have also been reported to be associated with AI. Seow found that patients with AI had a (26.1% vs. 4.3%) higher tendency than the unaffected group to have impacted permanent teeth and associated anomalies, such as follicular cysts. The observed impacted teeth among hypocalcified and hypomaturational AI types primarily involved canines [6,7,9].

Therefore, they speculated that there may be specific AI types that have taurodontism but were not included in the population studied. Whether these concurrent anomalies developed as a direct consequence of the molecular defect responsible for the enamel malformation or as a result of unknown secondary factors remains to be

determined. The presence of these abnormalities has apparent implications on the clinical management of these patients. Early screening for these abnormalities should be done so that interceptive treatment can be rendered to prevent further damages to the developing affected dentition [6,9,11,12].

Treatment Considerations: Individuals with AI often experience concern over poor dental esthetics, tooth sensitivity, and extensive tooth attrition. Because of this, it is necessary to provide appropriate dental treatments throughout the developmental stage. During any given phase of treatment, strict oral hygiene instruction and preventive treatments are equally essential in order to prevent caries, gingivitis, and calculus formation which may exacerbate existing problems. The successful management of AI requires the cooperation and motivation of both the patient and parents because the dental treatments can extend over many years and long-term success depends on regular attendance for dental procedures and the maintenance of optimal of oral health care.

The management of individuals affected by AI has been described as three stages in the literature [11].

a. Temporary phase — undertaken during the primary and mixed dentition

b. Transitional phase – when all permanent teeth have erupted and continue till adulthood

c. Permanent phase – occurs in adulthood.

In the primary dentition, the dental treatment of affected children aims to ensure favorable conditions for the eruption of the permanent teeth as well as for the normal growth of the facial bones and the temporomandibular joints^[11]. Upon eruption of the primary molars, stainless steel crowns are placed to prevent the development of caries and the attrition of defective enamel, while maintaining adequate space and vertical dimension of occlusion. In the primary anterior teeth, polycarbonate crowns, resin modified glass ionomers (RMGI), prefabricated crowns (stainless steel crowns with or without esthetic facing) or direct composite resin can be used as alternative restorations. When a more conservative approach is desired, RMGI is recommended in occlusal non-stress bearing areas because of its fluoride releasing and chemically retentive ability, while composites resin provide acceptable resistance to occlusal wear in stress bearing tooth surfaces [11,12].

In the mixed dentition, the treatment goals are to preserve tooth structures, maintain tooth vitality, decrease tooth sensitivity, establish correct interproximal and occlusal function, and improve esthetics. However, rehabilitation in the mixed dentition is complex, since teeth have different eruption sequence, and definitive treatment cannot be rendered until complete eruption of the permanent dentition. During this stage, there is often a need to reestablish the vertical dimension of occlusion. Stainless steel crowns on the permanent first molars are often recommended because they

provide sufficient and stable vertical dimension of occlusion. Casting onlays bonded onto the posterior teeth and composite resin restorations on occlusal surface have also been used as conservative approaches to increase vertical dimension of occlusion [13,14]. Several treatment modalities have been reported to improve dental esthetics. Direct or indirect composite resin veneers may be used to mask the discoloration and improve the crown morphology and contact with adjacent teeth. Also, full-coverage adhesive composite resin or polycarbonate crowns have also been advocated [14,15].

In the permanent dentition, the final treatment objectives are to diminish tooth sensitivity and to restore vertical dimension of occlusion, function, as well as esthetics. The final treatment often starts as soon as clinical height of the crown and the gingival tissue have been stabilized and the pulp tissues have receded. Full mouth rehabilitation combined with a multidisciplinary approach may be advantageous [12].

Prosthodontics, periodontics, orthodontics, and endodontics may be necessary. Treatment could also include orthognathic surgery. Crown lengthening and gingival recontouring may be indicated in case of short clinical crowns and gingival hyperplasia. Orthodontic treatments may be used to close interdental spaces prior to restoration and correct the anterior open bite malocclusion. Bouvier *et al.* reported an AI case that underwent orthodontic treatment successfully

without any problems arising from the placing of brackets on the performed stainless steel crown and polycarboxylate crowns [11]. Root canal therapy is indicated when pulp exposures are caused by severe attrition or tooth reduction. Orthognathic surgery may be indicated in case of severe malocclusion. Consultation with the appropriate specialists may help in developing a comprehensive treatment plan for each individual. [11]

The multidisciplinary team should be in close collaboration in terms of planning the immediate, transitory and long-term phases of treatment. The orthodontist and pediatric dentist play important roles with regard to mixed dentition. They help guide the prosthodontist and oral surgeon, who might be needed to treat the permanent dentition. In addition, the role of the periodontist includes maintaining the health and structure of the masticatory system during and after AI treatment. The speech therapist plays an important role in restoring the system and in long-term management. [1,5,8,11,14]

CASE DETAIL:

A 19-year-old woman was referred to our private dental clinic with the chief complaint of discolouration and unpleasant esthetics. Detailed dental, medical and social histories were obtained from the patient. Her general medical history was nonsignificant. She was born normally at term after an uneventful pregnancy. She was healthy and her general appearance was normal. A renal

ultrasound scan was normal and showed no evidence of nephrocalcinosis. Laboratory findings, including serum electrolytes, calcium, phosphate, urea, creatinine, alkaline phosphatase and parathormone levels, were all normal; blood pressure was also normal. However, plasma calcium levels in her mother and father were slightly increased. Although the parents were not affected by AI, the patient's brother was affected. Figure1-6

She presented with an unaesthetic smile due to discoloring and pitted enamel caused by amelogenesis imperfecta. She had moderate discomfort to "cold" drinks, but no other complaints. She had a history of orthodontics to align and straighten her teeth prior to restorative treatment. No history of periodontal disease and all probing less than 3.0 mm. JVA revealed no TMJ concerns. The case was relatively conservative as I only removed the soft enamel and provided enough room for the overlying ceramic. The most difficult thing was trying to create smooth, continuous margins since the enamel was pitted and rough. I utilized IPS Empress bonded to place with All-Bond 2 and Variolink Veneer cement. I restored the anterior first to establish aesthetics and then followed up by restoring the molars. Figure7-15

The patient was instructed in the maintenance of interproximal gingival health using dental floss . Routine panoramic radiographs were taken after treatment and annually during follow-up for 3 years .

The patient was followed at 3, 6 and 12 months and then annually with visual and radiographic examinations. During the first year, hygiene and long-term outcome were assessed. The patient acknowledged improved function and esthetics, and was pleased with the results .

DISCUSSION:

The term "amelogenesis imperfecta" describes a diverse group of hereditary conditions primarily affecting the quality and/or quantity of dental enamel. The affected teeth show a soft enamel of normal thickness that chips and wears easily and has a radiodensity similar to that of dentin ^[2,3]. The results of clinical and radiographic evaluations indicated that the patient in the present case had hypomaturation form AI. All the teeth are misshapen, and spotted. Occlusion and vertical opening are rapidly affected by attrition. The insufficiency of the enamel makes the teeth extremely sensitive to contact and thermal stimuli. These problems combine to make early diagnosis essential and immediate treatment a necessity, even for the youngest patients ^[4,6]

Zirconium oxide-based restorative materials have excellent mechanical properties and low bacterial adhesion and they are biocompatible. This allows several applications in restorative dentistry, one of which is as a core material for ceramic crowns and fixed partial dentures. In addition, studies have reported that zirconium oxide restorations represent promising prosthetic treatments for posterior

regions. The present case report describes the prosthetic rehabilitation of an adolescent patient with AI. The maxillary and mandibular right and left third molars were fully erupted; therefore, the permanent restorative material, zirconium oxide ceramic, was selected as a suitable replacement for the defective structures because of the attractive mechanical properties described above.^[15,16]

Previous studies have reported several methods for determining type of AI using combinations of clinical, radiographic, histologic and genetic criteria. Histologic evaluation using staining and SEM confirmed the diagnosis of AI. Although, SEM revealed a thin enamel layer with irregular structure, a thick layer of primary dentin had formed around the coronal pulp to resist traumatic forces.^[16,17]

Patients with this anomaly often seek treatment because of an unpleasant appearance, impaired mastication and social embarrassment. Lack of uniformity of the occlusal plane, congenitally missing teeth, tooth sensitivity, poor dental esthetics and decreased vertical dimension may create prosthodontic challenges^[18,19].

In the present case, the main complaints were tooth discoloration and generalized sensitivity. The patient was dissatisfied with her dental appearance and concerned about the long-term condition of her teeth. The aim was to satisfy her desire for esthetic improvement and not primarily to change her occlusal scheme. Therefore, full-mouth zirconium oxide

ceramic rehabilitation of the patient was provided without changing the occlusal vertical dimension. The occlusion was adjusted according to the patient's centric relation, as this was the only reproducible position. Canine-guided occlusion was provided, as the maxillary and mandibular canines came into contact on the working side while the posterior teeth were disoccluded.^[16,19,20]

After prosthetic rehabilitation, both facial appearance and occlusion were improved. Long-term dental management consisted of regular clinical and radiographic reviews at 3, 6, 12, 24 and 36 months. Function, phonation and esthetic expectations of the patient were met. In the radiologic and clinical examination, no problem was seen in soft tissue or in maintenance of the restorations. Function, parafunction, aging and stress fatigue affect the longevity of restorations in the oral environment. Therefore; continuous follow-up of at least 5 years will be needed to assess the success of such restorations in the oral cavity.^[5,8,18,19,20]

CONCLUSION:

In the presented case report, esthetic and functional rehabilitation of hypoplastic AI was performed with the use of zirconium oxide ceramic restorations. The treatment plan for cases of AI is related to many factors: the age of the patient, the socioeconomic status of the patient, the type and severity of the disorder, and the intraoral situation at the time the treatment is planned. The patient's functional and esthetic expectations were

achieved and no problem was detected at the 3rd annual clinical visit. The cumulated evidence on outcomes of alternative restorations for each type of AI is critically needed. With such evidence,

clinicians may then select more favorable approaches to treat individual AI patient and to optimize their patient's oral health and long term prognosis.

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FIGURES:



Figure1. Pre-operative of a young woman that presented with Amelogenesis Imperfecta



Figure2. Pre-operative left site



Figure3. Pre-operative right site



Figure 7. Post-operative views after full mouth rehabilitation using IPS Empress



Figure4.maxillary occlusal



Figure 8. Post-operative views after full mouth rehabilitation using IPS Empress



Figure 5. mandibular posterior before tooth preparation



Figure 9.post-operative maxillary anterior



Figure 6.Orthopantomogram of the patient.



Figure 10.post-operative left site



Figure 11. post –operative right site



Figure 12. Definitive crowns after cementation in maxillary



Figure 13. Definitive crowns after cementation in mandibular



Figure 14. Post-operative views after full mouth rehabilitation using IPS Empress (posterior maxillary)



Figure 15. Post-operative views after full mouth rehabilitation using IPS Empress (posterior mandibular)