

## Treatment considerations for patient with *Amelogenesis Imperfecta*: a review

Considerações de tratamento para pacientes com amelogenese imperfeita: revisão de literatura

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### ABSTRACT

**Objectives:** *Amelogenesis imperfecta* (AI) is a group of inherited disorders primary affecting the structural of enamel. Patients with AI experience poor esthetic, excessive tooth sensitivity and compromised chewing function that dental treatments are frequently required at early age. This review describes the non-enamel implications, stage-specific management strategies and outcomes of selected restorative materials based on literature evidence.

### RESUMO

**Objetivos:** Amelogenese imperfeita (AI) é um grupo de desordens hereditárias que afetam primariamente a estrutura do esmalte dentário. Pacientes com AI apresentam estética pobre, sensibilidade dentária excessiva e função mastigatória comprometida. Nestes casos, os tratamentos dentários em idades precoces são frequentemente necessários. Esta revisão descreve as implicações não relacionadas com o esmalte dentário, as estratégias de atuação em idades específicas e ainda os resultados de tratamentos restauradores com determinados materiais e baseados em evidência científica.

### KEYWORDS

*Amelogenesis imperfecta*; Dental enamel, abnormalities; Dentistry, operative.

### PALAVRAS-CHAVE

Amelogenese imperfeita; Esmalte dentário, anormalidades; Assistência odontológica

### INTRODUCTION

*Amelogenesis imperfecta* (AI) is a hereditary defect of enamel affecting both the primary and permanent dentition [1]. By definition, AI includes only those cases where enamel defects occur in the absence of other syndromes or metabolic disorders [1]. The incidence of AI ranges from 1 in 718 to

1 in 14,000 depending on the population studied [1,2]. According to Witkop, AI can be classified as hypoplastic, hypomaturational, hypocalcified, and hypomaturational-hypoplastic with taurodontism. When clinical features and inheritance pattern are taken into consideration, 15 subtypes of AI can be distinguished. These 15 subtypes are currently the most widely used and accepted AI classification system (Chart 1).

**Chart 1** – Classification of AI proposed by Witkop (1989).

Type I	Hypoplastic
IA	Hypoplastic, pitted autosomal dominant
IB	Hypoplastic, local autosomal dominant
IC	Hypoplastic, local autosomal recessive
ID	Hypoplastic, smooth autosomal dominant
IE	Hypoplastic, smooth X-linked dominant
IF	Hypoplastic, rough autosomal dominant
IG	Enamel agenesis, autosomal recessive
Type II	Hypomaturation
IIA	Hypomaturation, pigmented autosomal recessive
IIB	Hypomaturation, X-linked recessive
IIC	Snow-capped teeth, X-linked
IID	Snow-capped teeth, autosomal dominant
Type III	Hypocalcified
IIIA	autosomal dominant
IIIB	autosomal recessive
Type IV	Hypomaturation-hypoplastic with taurodontism
IVA	Hypomaturation-hypoplastic with taurodontism, autosomal dominant
IVB	Hypomaturation-hypoplastic with taurodontism, autosomal recessive

Although AI affects only the enamel formation, it has significant consequences for the affected patients. Often AI patients experience difficulty in maintaining oral hygiene, decreased masticatory function, and a lower self-esteem, affecting their over-all quality of life [3,4]. Furthermore, most variants of AI require extensive dental treatment, which can be time consuming and often poses a significant economic burden on their family. 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.

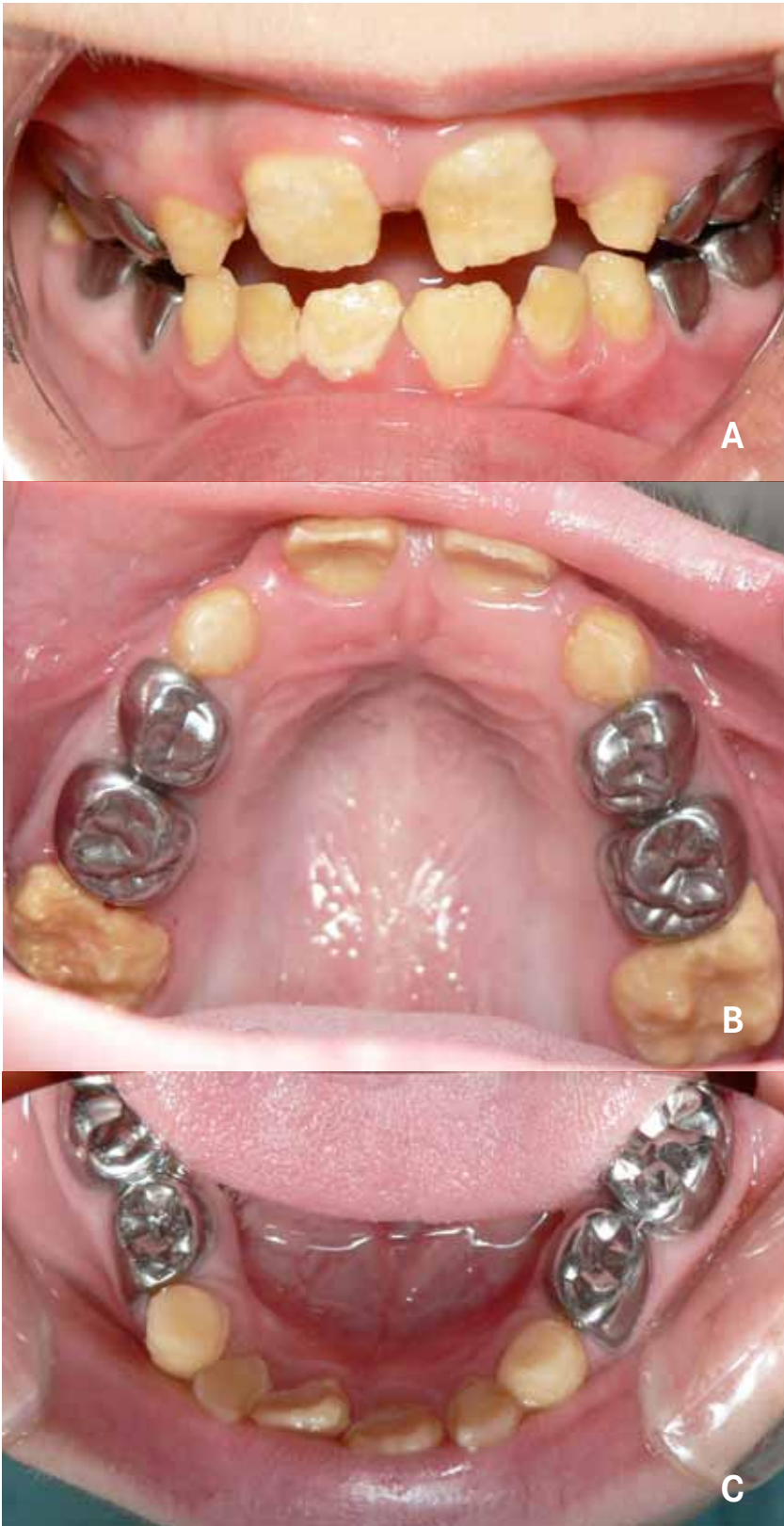
## CLINICAL IMPLICATION OF AI

Although AI primarily affects enamel formation, a variety of clinical implications may also be present, such as low caries susceptibility,

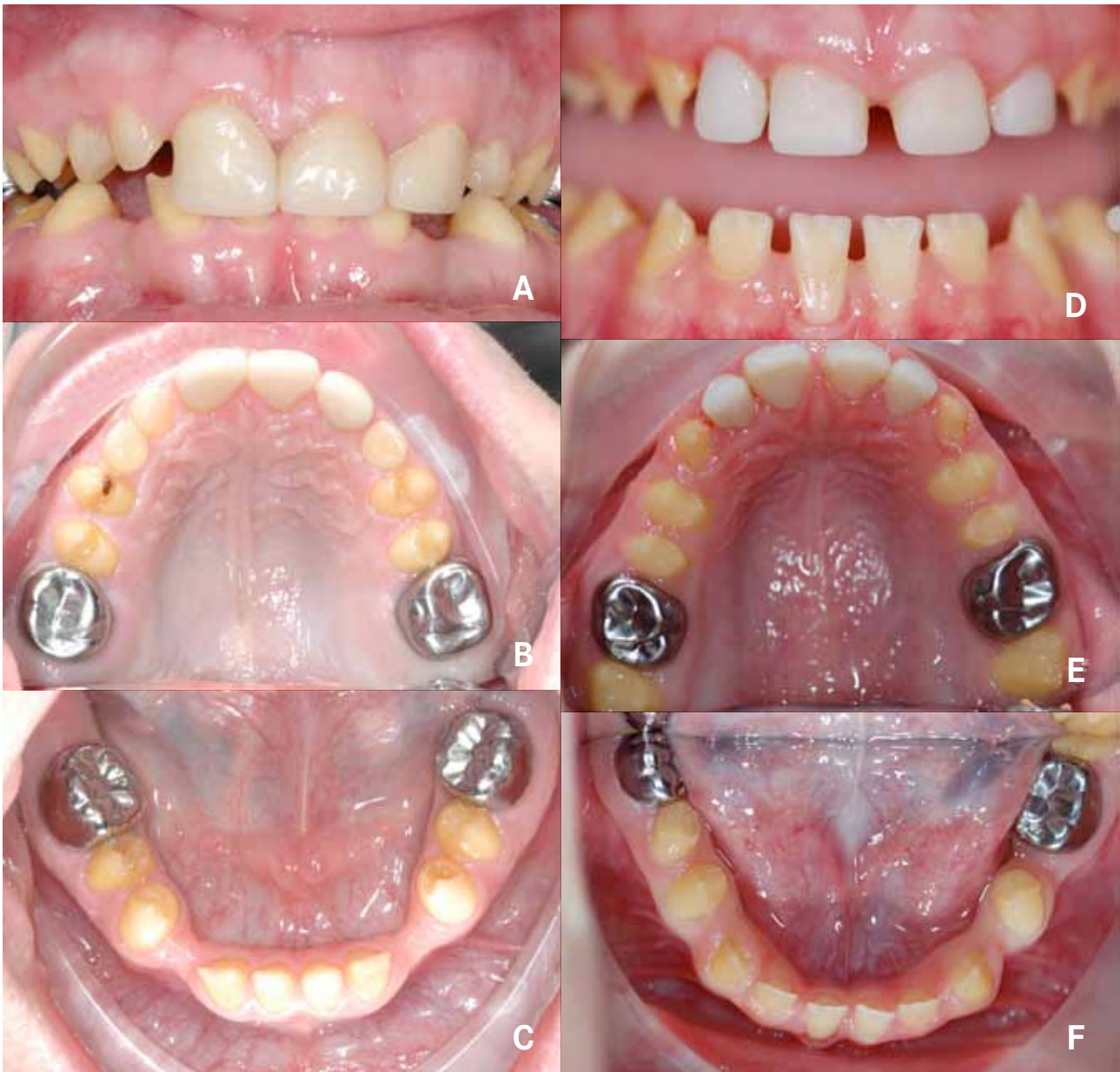
rapid attrition, excessive calculus deposition, and gingival hyperplasia [5-7]. The severity of clinical problems varies with each type of AI. Low caries susceptibility has been reported in children with severe hypoplastic and hypomineralized AI [8]. While Sundell stated that the bacteriological and salivary data from patients with AI were inadequate to explain low caries susceptibility [8], it was suggested that additional investigations focusing 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]. Rapid and excessive calculus formation has been reported as a common finding of the hypomaturation and hypocalcified types of AI [6]. 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 [6]. However, there was no evidence provided to support his speculation. Sundell did observe that the saliva secretion rate, pH and buffer capacity from AI individuals corresponded to children without AI [8]. Moreover, the gingival condition and oral hygiene among patients with AI were reported to be poor [3,5].

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 (Figure 1 A-C, 2 A-F, 3A-C, 4 A-C).

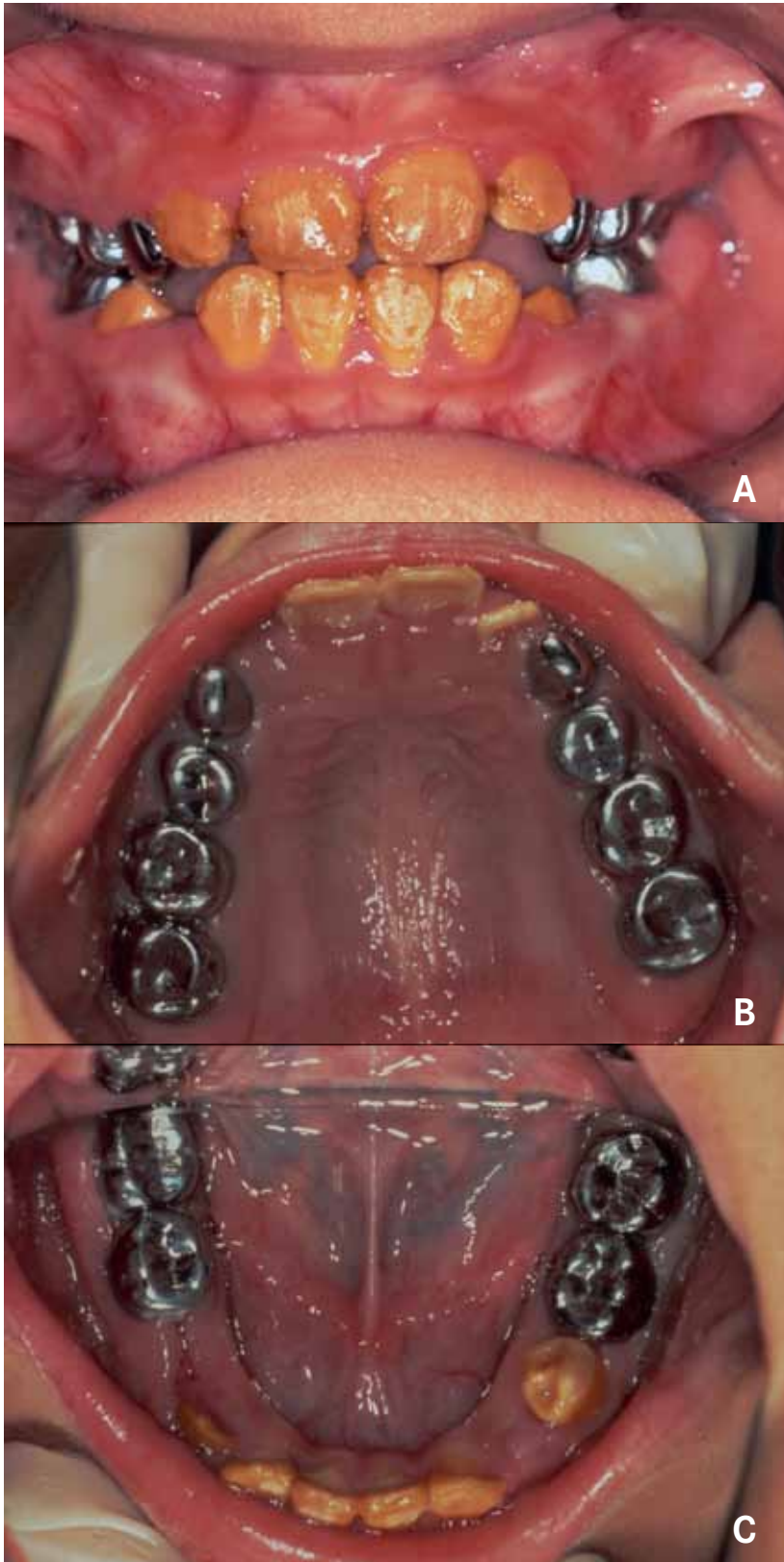
Unfortunately, restorative treatment for patients with AI is not often provided at an early age due to issues related to tooth sensitivity, difficulty in managing extensive treatment needs, and even cost. There is a tendency to adopt a "wait-and-see" policy, often resulting in the development of deep overbite and deleterious structural lost.



**Figure 1** – Hypocalcified AI.



**Figure 2** – Hypoplastic AI.



**Figure 3** – Hypomaturational AI.



**Figure 4** – Hypocalcified- hypomaturational AI.

## GROWTH AND DEVELOPMENT

Non-enamel-related manifestations may also occur, including open bite malocclusion, disturbances in eruption (accelerated dental development, increased prevalence of dental impaction), congenital missing teeth, pathologic crown and/or root resorption, pulp calcification, and taurodontism [3,7,9-11]. Open bite malocclusion has been reported to be associated with AI. Ravassipour et al. stated that, among 54 AI patients, 42% of AI affected individuals and 12% of unaffected family members had dental or skeletal open bite malocclusion when compared to a general Caucasian population (5%). The severity of enamel phenotype did not necessarily correspond to the presence or severity of the open bite malocclusion. They stated that the etiology of open bite malocclusion in the different types of AI is heterogeneous. It includes factors such as abnormal eruption of posterior teeth, modifying genes affecting both the dental and craniofacial developmental processes and environmental factors [11]. 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 [9].

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 [9]. Collins stated that the frequency of dental anomalies also varied in different types of AI. Hypoplastic AI frequently exhibited delayed eruption, crown resorption, and pulp calcification (Figure 3). The prevalence of taurodontism was similar in individuals with AI and individuals without AI [10]. 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 [10]. 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.

## PSYCHOSOCIAL IMPACT

The first study that attempted to objectively characterize the psychosocial impact on AI patients was conducted by Coffield and colleagues. They found that subjects with AI had higher levels of social avoidance, distress and higher levels of dysfunction. The relationship of AI status to fear of negative evaluation, lower mastery and self-esteem was age-dependent. Additionally, subjects with AI tended to show an increase in mastery and self-esteem scores with age [4]. One can speculate that AI patients could have had comprehensive dental treatment that helped them cope with their condition. In a retrospective study, Lindunger and Smedberg stated that all AI patients had positive reactions to their prosthodontic treatment [12]. Interestingly, nearly half of AI patients preferred restorative treatment done before the age of 16 [12]. Therefore, it can be argued that early diagnosis and timely intervention may increase psychosocial well being and self-esteem of AI patients, especially during the pre-adolescent and adolescent stages.

## SPECIFIC TREATMENT CONSIDERATIONS

The management of individuals affected by AI has been described as three stages in the literature [13].

- Temporary phase — undertaken during the primary and mixed dentition
- Transitional phase – when all permanent teeth have erupted and continue till adulthood
- 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 [14]. 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.

In the mixed dentition, the treatment goals are to preserve tooth structures, maintain tooth vitality, decrease tooth sensitivity, vertical dimension, and improve esthetics. When permanent first molars and anterior teeth erupt, orthodontic and prosthetic assessment is essential. 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. As for permanent molars, stainless steel crowns are often recommended because they provide sufficient and stable vertical dimension of occlusion. In addition, 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 [15,16]. Several treatment modalities have been reported to improve dental esthetics [17]. 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 [18,19].

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 [20]. Prosthodontics, periodontics, orthodontics, and endodontics may be necessary. Treatment could also include orthognathic surgery [21]. 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. 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.

## TREATMENT MODALITIES FOR AI

With the advance in techniques and increase in availability of various dental materials, many studies have shown the use of glass ionomer cements, composite resin veneers, porcelain veneers, stainless steel crowns, lab-fabricated crowns, and/or over dentures can restore the affected teeth [3,20,22-25]. The treatment approach should consider the specific AI type and underlying defect. In the patients with hypoplastic AI, enamel is usually sufficient for bonding so composite resin restoration may be successful masking discoloration and improving crown morphology [26]. In patients with hypocalcified AI, there is insufficient enamel for bonding. Glass ionomer cements and composite resin restorations might initially be successful in these cases, but the long term-prognosis is guarded as the hypocalcified enamel may fracture, causing defective margins and broken restorations. Full coverage restorations are commonly recommended for hypocalcified AI. In case of hypomaturation AI, the defective enamel contains excessive organic matter that overtime becomes porous and stained; the defective enamel should be removed before placement of restorations.

## INTRACONORAL RESTORATION

Direct restorative materials, such as amalgam, glass ionomer cements, composite resins, resin-modified glass ionomer cements, have been advocated for restoring teeth with AI. Amalgam can be used to restore small lesions in posterior teeth especially with mildly affected



hypoplastic AI. However, amalgam restorations are usually unsuccessful in severely affected teeth with AI due to fractures in defective enamel margins. Seow found that adhesive materials such as glass ionomer cements and composite resins are better retained in small restorations compared to amalgam restorations [3]. 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 [14]. Unfortunately, very little evidence exists to support the long-term use of glass ionomer cements and resin modified glass ionomer cements.

Composite veneers and composite resin restorations have been advocated to mask discoloration and improve dental esthetics. Composite resin restorations can be placed with minimal tooth preparation or no tooth preparation to preserve tooth structure and is a favorable treatment option for partially erupted teeth. Rada reported composite resins provided satisfactory esthetics and durability [23]. However, a high failure rate associated with insufficient bonding between the composite resin restoration and enamel among variants of AI has been reported in the past [26]. Several approaches have been documented to improve bonding strength between composite resin restoration and enamel. Venezie et al. found that pretreatment of enamel surfaces affected by AI with five percent sodium hypochlorite resulted in an improvement of bonding strength in vitro [27]. On the other hand, Sonmez et al. further investigated this in the clinical setting and found that pre-treatment with sodium hypochlorite had no significant effect on the success of the adhesive restoration in the intraoral condition. Due to this lack of consensus in the literature, there is still a necessity to evaluate the effectiveness of intraconoral restorations of teeth with AI.

## EXTRACORONAL RESTORATION

Although bonding onto the hypoplastic enamel is feasible, sufficient enamel must be

available for bonding. In certain types of AI such as hypocalcified type, enamel fracture frequently results in defective margins of the restoration. Therefore, full coverage restorations may be a more effective method to provide maximal protection, esthetics, and function.

Stainless steel crowns are reported to be the most effective and efficient restoration in managing tooth sensitivity and restoring severely broken down primary molars and permanent molars in children [3,24]. Stainless steel crowns can be placed with minimum preparation, preserving tooth structure as much as possible in preparation for a more definitive treatment in the future. However, stainless steel crowns do not have perfectly adapted margins and as such their long-term use on the permanent teeth must be carefully considered. In order to obtain custom-made margins, casting crowns are a viable alternative for posterior teeth [15]. In a study comparing stainless steel crowns and casting crowns in restoring the permanent first molars affected by AI or teeth with severe enamel defects, no significant differences in quality or longevity of the restorations were found after up to 24 months of follow-up [28]. The long-term effect of using stainless steel crowns on the permanent first molars is unknown and needs further investigation.

Casting onlays have also been suggested to protect the remaining tooth structure on the posterior teeth, and may be fabricated without tooth preparation [15,16]. These restorations can be used before the teeth are fully erupted. It may be a concern that the placement of supraoccluding restorations will result in an increase of the vertical dimension and alter occlusion negatively. In a study of 12 children, age 7-to 18-years old, affected by either *Amelogenesis imperfecta* or dentinogenesis imperfecta, patients were able to re-establish their occlusion within 3 months after restorations being cemented [16]. To date, the long-term evaluation of the efficacy of such alteration of occlusion by restoration is necessary.

Acrylic resin crowns or poly carbonate crowns were used to restore the anterior teeth. The acrylic resin or polycarbonate crowns are

esthetically acceptable and less expensive. Even when there is no sufficient enamel available for bonding, acrylic crowns or polycarbonate crowns are likely to be helpful for anterior teeth, particularly in the mixed dentition [19]. When the enamel defects involve proximal surfaces or decreased bonding is expected, these crowns offer reasonable esthetics and retention due to their full coverage and the use of cement. However, their use in AI teeth has not been systemically evaluated.

For AI patient with a deep overbite, composite resin bonded restorations and veneers are not recommended as a treatment option because incisal loading creates negative impact that leads to restoration failure. Little information in the literature reports the use of stainless crown in the anterior teeth in children with deep overbite. Rosenblum reported a case using stainless steel crowns, which have been pre-fitted and then professionally veneered in a patient with 100% overbite. Rosenblum stated that this technique increased the longevity of the veneer and produced a better fitting crown [24].

Porcelain fused to metal (PFM) crowns for the anterior and posterior teeth are the restoration of choice for AI patients and have been reported to be successful but only in affected adults [25]. Ceramic crowns are also considered for anterior restorations because of its esthetics [20]. However, those types of crowns require significant tooth reduction. Thus, the use of PFM and ceramic crowns in young permanent teeth is not recommended due to the presence of large pulp and short clinical crowns. Obtaining radiographs prior to restorative treatment is essential to determine when these restorations can be placed.

## OVERDENTURE

An overdenture is supported both by soft tissues and the roots of teeth or modified teeth. Renner et al. reported a case which was successfully treated by maxillary overdenture. The overdenture provides a simplified solution to a complex prosthodontic problem that is

both reversible when growth and development demand that the prosthesis be altered and usually requires minimal intraoral modifications for its fabrication [22].

## ORTHODONTIC TREATMENT

Patients with AI often need orthodontic treatment due to dental and/or skeletal problems. The challenge of using fixed orthodontic appliances is to manage the defective enamel and determine whether the defective enamel can withstand the force that applied during the treatment and when removing the appliances [29]. To date, many methods such as using plastic brackets, glass ionomer cement base adhesives, and traditional banded appliances can be used to improve appliance retention but the evidence is weak. Moreover, the lack of uniformity of enamel means that the second and third order bends which are part of a pre-adjusted appliance prescription are not fully expressed and more detailing bends at the finish stage of orthodontic treatment are needed [29]. Achieving perfect occlusion is not always the treatment goal at the end of the orthodontic treatment as the main principle is often to place the teeth in a position that facilitates the placement of restorations. Research on orthodontic treatment for patient with AI relies on case reports that present orthodontic treatment progress of AI patient. 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 crown and polycarboxylate crowns [13].

## TREATMENT OUTCOME

Treatment of AI depends on the individual's specific diagnosis and phenotype. Unfortunately, research on long-term follow up of restorative 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. Based on reported assessments of AI cases, it has been found that the treatment modalities of patients with AI vary, and their prognoses are generally unpredictable. Lindunger and Smedberg assessed the outcome of the prosthodontic management of adult AI patients and they found that 213 restorations in 15 patients were rated as acceptable to excellent, with one exception [12]. Periodontal status in AI patients did not differ from patients without AI, but hypomineralized AI patients tend to have high scores in periodontal parameters. The overall results showed that the restorations performed well, and that all the patients had positive reactions to prosthodontic treatment. While this retrospective study has shown positive results of prosthodontic rehabilitation for patients with AI, research on long-term prognosis of restorative outcomes of AI patients is still particularly scarce and deserves further investigations.

## CONCLUSIONS

*Amelogenesis imperfecta* is a group of inherited disorders that pose diagnostic and restorative treatment challenges for dental care providers. There is currently no standard of care established for managing patient with AI although multidisciplinary approach may be advantageous. 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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