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Amelogenesis Imperfecta: A Literature Review

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The College of Dentistry, University of Baghdad, Department of Oral diagnosis in Partial Fulfillment for the Bachelor of Dental Surgery degree

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Certification of the Supervisor

I certify that this project entitled " **Amelogenesis Imperfecta: A Literature Review**" was prepared by the fifth-year student **Duha Hikmet Hammed** under my supervision at the College of Dentistry/University of Baghdad in partial fulfilment of the graduation requirements for the Bachelor Degree in Dentistry.

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Dedication

I dedicate my graduation thesis to my father soul who wanted me to be a doctor and he couldn't see this day and share this success with me.

To the one who nursed me with love and tenderness, to the symbol of love and healing, to the white heart, my beloved mother to the source of my happiness and light in my life, my brothers ,sisters and best friends Ghofran.

ACKNOWLEDGMENT

In the first place, I would like to thank God for making my dream come true, facilitating my career, and giving me the patience and strength to continue my scientific career. Today, by the grace of God, I present to you my graduation thesis, and I hope that you will be satisfied.

I would like to express grateful thanks to dean of college of dentistry, University of Baghdad **Prof. Dr. Raghad Al-Hashimi**.

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Phrase	Abbreviation
AI	Amelogenesis Imperfecta
MMP_20	Matrix Metalloproteinase 20
E	Enamel
DEJ	Dentinoenamel junction

Introduction

Introduction

Amelogenesis imperfecta (AI) refers to a group of rare, inherited disorders characterized by abnormal enamel formation affects the entire ectodermal component. The term is typically restricted to those disorders of enamel development not associated with other abnormalities of the body **(Naik *et al*., 2011).** Hereditary brown enamel, hereditary enamel dysplasia, hereditary brown opalescent teeth are the other terminologies used for amelogenesis imperfect **(Crawford *et al*., 2007; Wright *et al*., 2011).**

Amelogenesis imperfecta affects both the primary and permanent dentitions. In the teeth affected by AI, the dentin and roots appear normal. Based on phenotypic characteristics and mode of inheritance, the classifications of AI are proposed **(Mayur *et al*., 2009; Toupenay *et al*., 2018).** Depending on phenotypic characteristics of enamel(appearance, structural and developmental defects), AI is classified into 4 main patterns: hypoplastic, hypomaturation, hypocalcified, and hypomaturation-hypoplastic. AI may be inherited as an X-linked, autosomal dominant, or autosomal recessive genetic trait, depending on the mode of inheritance **(Naik *et al*., 2011).**

The restoration of the defects created by AI improves the esthetic and functional concerns of the patient. Treatment Planning of these cases involves an interdisciplinary approach to evaluate, diagnose, and resolve esthetic problems using a combination of periodontal, prosthodontic, orthodontic and restorative treatment **(Canger *et al*., 2010).**

Chapter One

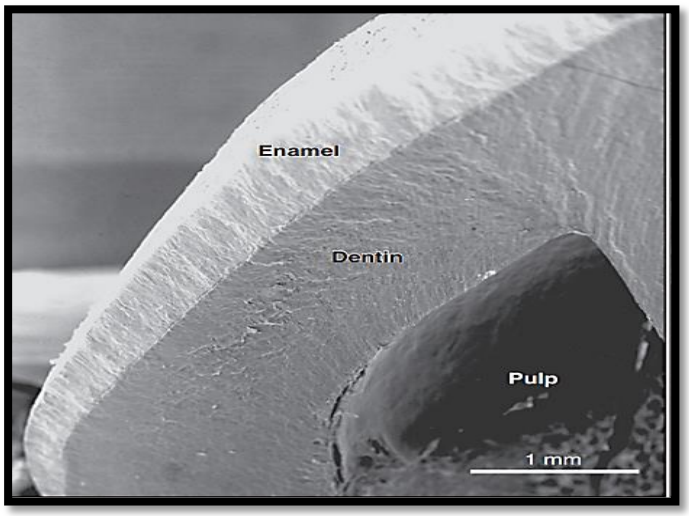
Review of Literature

Review of Literature

1.1 Enamel

Enamel is the hardest calcified matrix of the body. The cells that are responsible for formation of enamel, are the ameloblasts, are lost as the tooth erupts into the oral cavity, and hence enamel cannot renew itself. Enamel brittle is therefore an underlying layer of more resilient dentin is necessary to maintain its integrity, if this supported layer of dentin is destroyed by caries or improper cavity preparation, the unsupported enamel fracture easily **(Figure 1.1)(Nanci, 2018).**

The color of enamel covered crown ranges from yellowish white to grayish white. The translucency may be attributable to variations in the degree of calcification and homogeneity of enamel. The high mineral content make enamel extremely hard this is property that together with its complex structural organization enables enamel to withstand the mechanical forces applied during mastication **(Kumar, 2015).**



Figure(1.1): Scanning electron microscope view of tooth structures **(Nanci, 2018).**

1.1.1 Chemical Properties of enamel

The enamel consists mainly of inorganic material (96%) and only a small amount of organic substance and water (4%), organic material consists of some unique proteins, amelogenins (90%) and the non-amelogenins such as enamelin and ameloblastin (10%), The inorganic material of the enamel is hydroxyapatite crystals and also contain various such as magnesium, lead, and fluoride **(Chen and Liu, 2014).**

1.1.2 The basic structure of enamel

The building block of enamel is the enamel rod or prism which is an array of aligned carbonated apatite crystals. Typical rod crystals in mature enamel are ~50 nm wide and more than 10 μm long. The elongated crystals in each rod run parallel to one another. Each rod is also associated with an interrod substance, which consists of crystals arranged in different orientation **(Beniash *et al*., 2019).**

Rod and inter-rod enamel is formed from the tomes process of ameloblasts. Each rod is wrapped in a sheath of organic matrix called rod sheath, whereas crystals within the rod about one another, with discontinuous, organic meshwork in between. In cross-sections of human enamel, many rods resemble fish scales **(Figure1.2) (Kumar, 2015).**

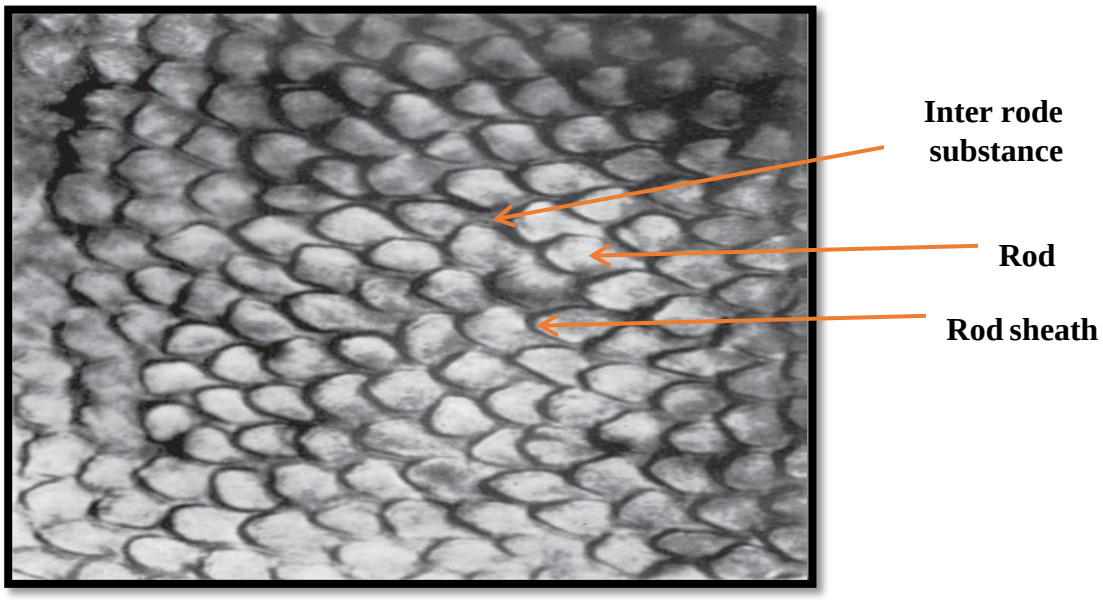


Figure (1.2): Decalcified section of enamel of human tooth germ. Rods have appearance of fish scales **(Kumar, 2015).**

1.1.3 Enamel Formation

Dental enamel formation, also called amelogenesis, involve different phase regulated by ameloblast that express an important set of genes that encode the production of enamel formation . The initiation of tooth formation starts around the 37th day of gestation **(Lacruz *et al*., 2017)**. A thickening of the stratified squamous epithelium, also called the oral ectoderm, gives rise to the dental lamina which is the foundation for the tooth germ. The tooth germ aggregates to form the tooth bud (intiation), cap stage (proliferation), bell stage (histodifferentiation and morphodifferentiation), apposition and calcification **(Almhateb, 2012).**

As development proceeds, the enamel organ assumes a bell-shaped structure with a well-defined dental papilla along its concave internal. It can be divided into four regions: the outer enamel epithelium, the stellate reticulum, the stratum intermedium, and the inner enamel epithelium **(Chen and Liu, 2014).**

The inner enamel epithelium further differentiates into ameloblasts. The ameloblasts make and secrete the enamel organic matrix, and are then calcified

with calcium phosphates to form the enamel. Unlike other calcified tissues, such as dentin and bone, there are no living cells in mature enamel. Ameloblasts are no longer present when enamel is formed. Thus, when enamel is damaged, there are no cells to carry out the repair. **(Yildirim, 2013)**.

1.1.3.1 The stages of amelogenesis

1- Enamel matrix deposition

Ameloblast synthesize and secrete enamel matrix 30 % hydroxyl apatite crystals and 70 % water and protiens, such as amelogenin, ameloblastin, enamelin, and enzymes like enamelisin, also called Matrix Metalloproteinase 20 (MMP_20), these proteins are responsible for creating and maintaining an extracellular environment favorable to mineral deposition **(Nanci, 2018).**

2- Mineralization of the Enamel matrix

When the E. matrix reach the full thickness, mineralization will be started, This process involved additional minerals with the removal of organic material and water to reach 96% mineral content. This minerals makes the initial E. crystals that formed in first stage to grow wider and thicker due to the deposition of large amount of hydroxy apatite crystals **(Almehateb, 2012).**

1.1.4 Ameloblast

It is the cell responsible for amelogenesis and according to the functions performed the life cycle of ameloblasts can be divided into Pre-secretory stage (Morphogenic stage, Organizing/differentiating), secretery stage (formative stage), post secretory stage (maturative stage, protective stage and desmolytic stage) **(Figure 1.3)(Maji, 2017; Nanci, 2018).**

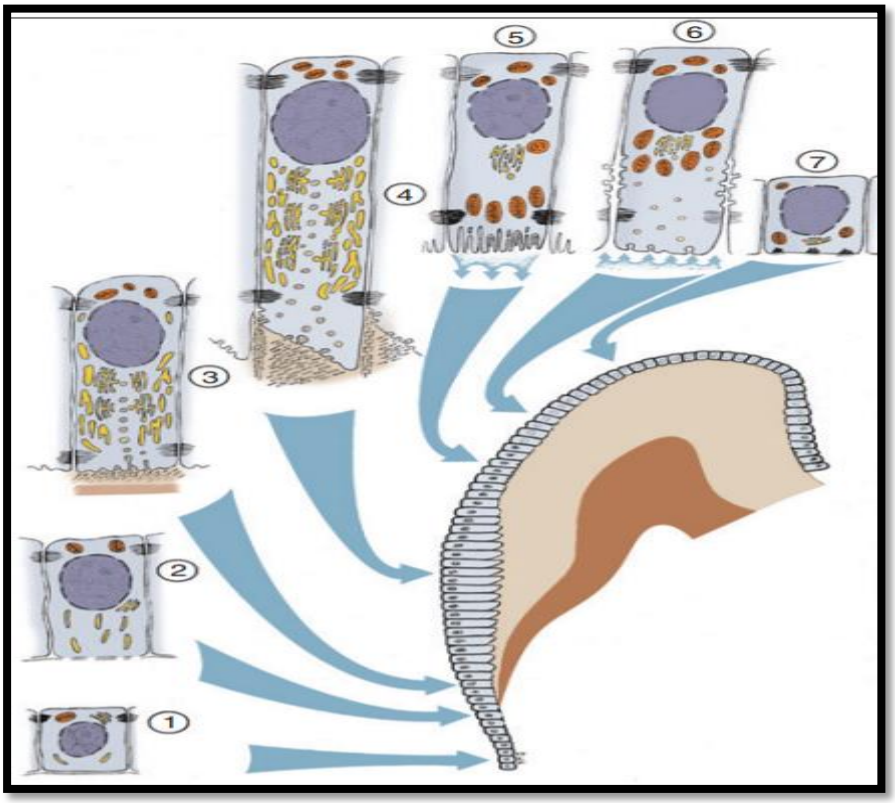


Figure (1.3): Schematic representation of the various functional stages in the life cycle of ameloblasts as would occur in a human tooth. 1, Morphogenic stage; 2, histodifferentiation stage; 3, initial secretory stage (no Tomes' process); 4, secretory stage (Tomes' process); 5, ruffle-ended ameloblast of the maturative stage; 6, smooth-ended ameloblast of the maturative stage; 7, protective stage **(Nanci, 2018).**

1-Morphogenic stage

Before the ameloblasts are fully differentiated and produce enamel, they interact with the adjacent mesenchymal cells, determining the shape of the dentinoenamel junction (DEJ) and the crown. During this morphogenic stage, the cells are short and cuboidal, with large oval nuclei that almost fill the cell body **(Kumar, 2015).**

2-Differentiating stage or organizing stage

During this stage, inner enamel epithelium cell undergo differentiation to ameloblasts as a prerequisite for enamel formation. This stage also name organizing stage because during this stage, the ameloblast exert organizing influence on dental papilla cell which are adjacent to them and help to

differentiation to odontoblasts, The polarity of the cells is reversed as the Golgi complex shifts from a proximal to distal position **(Maji, 2017).**

3-Formative or secretory stage

In this stage, ameloblasts perform the function of secretion of enamel matrix and partial mineralization. The ameloblast which are fully differentiated starts secretory function only after layer of dentin is deposited. The secretery ameloblasts are structurally suited for synthesis and secrtion of enamel protiens in which become columnar in shape with tomes' process which responsible for formation of enamel rods and inter rod substance **(Chiego, 2018).**

4-Maturative Stage

Before maturation stage the ameloblasts enter a brief transitional stage. Their height is decreased and protein synthesizing organelles are reduced.The overall number of ameloblasts is reduced by programmed cell death (apoptosis) **(Robinson, 2014).**

During the maturative stage, ameloblasts helps in the mineralization and maturation of enamel. Ameloblasts enter into the maturative phase only after the desired thickness of enamel matrix is laid down. In this stage, ameloblasts have to introduce the inorganic material necessary for maturation (by ***Ruffled-ended ameloblasts)*** and also reabsorb proteins and water to provide space for the minerals (***Smooth-ended ameloblasts)*** **(Maji, 2017).**

5-Protective stage

When the enamel has completely developed and has fully calcified, the ameloblasts cannot differentiated from the cells of the stratum intermedium and outer enamel epithelium which fuse together to form the reduced enamel epithelium. The function of reduced enamel epithelium is to protect the mature enamel by separating it from the C.T, until the tooth erupts **(Chatterjee, 2014).**

6-Desmolytic stage

The reduced enamel epithelium proliferates and seems to induce atrophy of the connective tissue separating it from the oral epithelium, so that fusion of the

two epithelia can occur . It is probable that the epithelial cells elaborate enzymes that are able to destroy connective tissue fibers by desmolysis. Premature degeneration of the reduced enamel epithelium may prevent the eruption of a tooth **(kumar, 2015).**

1.2 Enamel defects

Generalized developmental abnormalities of enamel may be attributed to genetics, systemic influences i.e. nutritional deficiencies or metabolic disorders, or may be idiopathic. They may also be caused by local factors such as trauma or infection. One of the main enamel defects is Amelogensis Imperfecta **(Almehateb, 2012).**

1.2.1 Amelogensis Imperfecta

Amelogenesis imperfecta (AI) is a heterogeneous group of genetic conditions characterized by defects in the formation of enamel in all teeth of both dentitions **(Roma and Hegde, 2016).** This condition causes teeth to be unusually small, discolored, pitted or grooved, and prone to rapid wear and breakage. The prevalence varies from 1:700 to 1:14,000, according to the populations studied **(Gadhia *et al*., 2012).**

1.2.1.1 Causes of Amelogenesis imperfecta

Amelogenesis imperfecta presents large variability in its clinical expression. Mutations have been reported in different genes. Some of them encode for enamel proteins, either structural (amelogenin, enamelin, ameloblastin) or enzymatic (kallikrein 4, MMP20); some others encode for transcription factors , cellular proteins, cellular receptor and calcium carrier **(Toupenay *et al*., 2018).**

These genes provide instructions for making proteins that are essential for normal tooth development. These proteins are involved in the formation of enamel, which is the hard, calcium-rich material that forms the protective outer

layer of each tooth. Mutations in any of these genes alter the structure of these proteins or prevent the genes from making any protein at all. As a result, tooth enamel is abnormally thin or soft and may have a yellow or brown color. Teeth with defective enamel are weak and easily damaged *(***Table 1.1) (Gadhia *et al*., 2012).**

Table (1.1): Causes of amelogenesis imperfecta. The genes are listed on the left side of the column. Mutations or alterations in the expression of these genes result in the various phenotypes associated with amelogenesis imperfecta as described in the columns on the right side **(Gadhia *et al*., 2012).**

Genes	Phenotypes associated with amelogenesis imperfecta
ENAM	Variable hypoplasia ranging from local pitting to marked, generalised enamel thinning. A variety of hypoplastic phenotypes depending on the specific mutation and its effect on the protein. Murine *ENAM* null mouse failed to show any true enamel.
AMELX	Abnormal maturation and mineralization defects. Distinctly abnormal teeth with disorganized hypoplastic enamel. Variable phenotype ranging from hypoplasia to hypomaturation/hypomineralisation.
KLK4 and *MMP20*	Defects in the final crystallite mineralisation or maturation of the enamel. The murine *Mmp20* null mouse exhibits both hypoplastic and hypomineralised defects Murine *Klk4* null mouse exhibits hypomaturation defects
AMELOTIN	No mutation in the amelotin gene has been related to amelogenesis imperfecta
FAM83H	Autosomal dominant hypocalcified amelogenesis imperfecta. Normal enamel thickness with decreased mineral content.

1.2.1.2 Classification of amelogenesis imperfecta

Clinical researchers usually classify AI into four main types of which 17 subtypes are recognized. The most commonly used classification of AI was proposed by **Witkop (1988**) which was later revised by **Nusier (2004).**

The main types are based on clinical appearance, radiographic appearance and enamel thickness, and the subtypes are based on mode of inheritance. The main types are: hypoplastic (type I); hypomaturation (typeII); hypocalcified

(typeIII); and hypomaturation/hypoplasia/taurodontism (type IV). AI may be inherited as an X-linked, autosomal dominant, or autosomal recessive genetic trait, depending on the mode of inheritance **(Table 1.2) (Sabandal and Schäfer, 2016; Novelli *et al*., 2021).**

Table (1.2): Witkop classification of amelogenesis imperfecta **(Novelli *et al*., 2021).**

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	Hypoplastic-hypomaturation with taurodontism, autosomal recessive

1-Hypoplastic AIH (type I)

Inadequate deposition of enamel matrix with normal mineralization in which that quantitative defect in enamel matrix formation. About 60-73% of all the cases of amelogenesis imperfecta are the hypoplastic type **(Toupenay *et al*., 2018).**

Clinical features

1-Hypoplastic AIH (type I) consists of quantitative alteration of enamel with localized or generalized reduced thickness.

2-Teeth are yellow to light brown

3-Surface is rough with pits or larger area defects. Severe hypoplastic phenotype leads to morphological anomalies seen on radiographic examinations.

4-No pain is associated with this AI, although some slight thermal sensitivity may sometimes be reported

5-Because of reduced enamel thickness in some cases, abnormal contour and absent interproximal contact points may be evident **(Gadhia *et al*., 2012).**

Radiographically

Thin enamel but normal radiodensity will be seen, Enamel can be distinguished from the underlying dentin. **(Figure 1.4)(Shivhare *et al*., 2016; Regezi *et al*., 2016)**

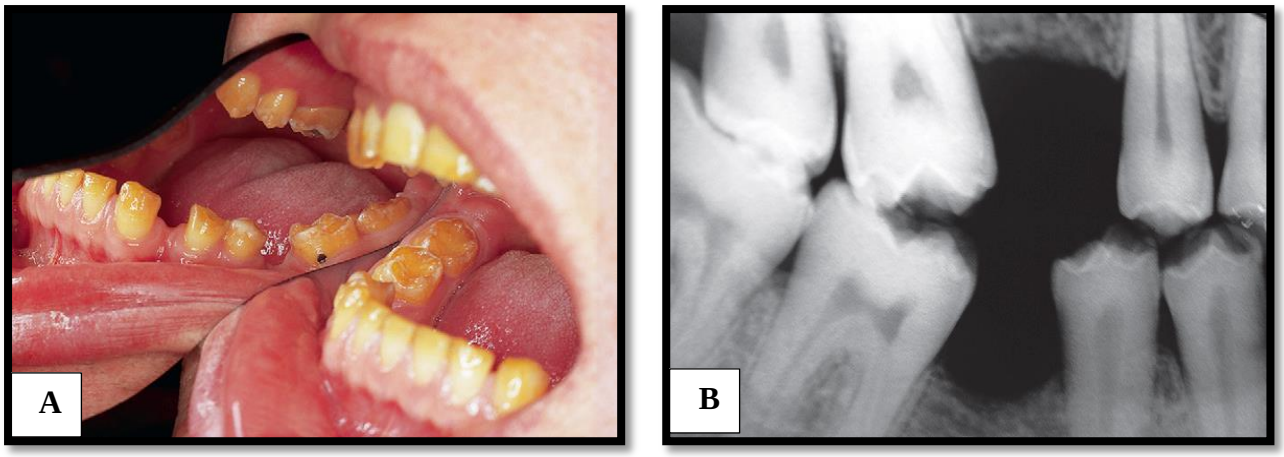


Figure (1.4): Hypoplastic AI **A**: Clicincal image **B**: Radiographical image**(Regezi *et al*., 2016).**

Subtype classification of hypoplastic AI according to the clinical appearance

1-Hypoplastic generalized pattern affects the entire dentition. Pin point pits are presents on the buccal surface of the teeth and later on these pits may be stained.

The enamel between these pits is of normal thickness, hardness and colouration **(Figure 1.5) (Chi *et al*., 2017)**

2-Hypoplastic localized pattern affects only some teeth in the oral cavity. Large defects on the buccal middle third of the teeth are seen **(Figure 1.6) (Tyagi., 2019)**.

3-Hypoplastic smooth pattern exhibit smooth surface which is thin, hard and glossy**(Figure 1.7) (Chi *et al*., 2017).**

4-Hypoplastic rough pattern the enamel is thin, hard and rough **(Figure 1.8) (Chi *et al*., 2017).**

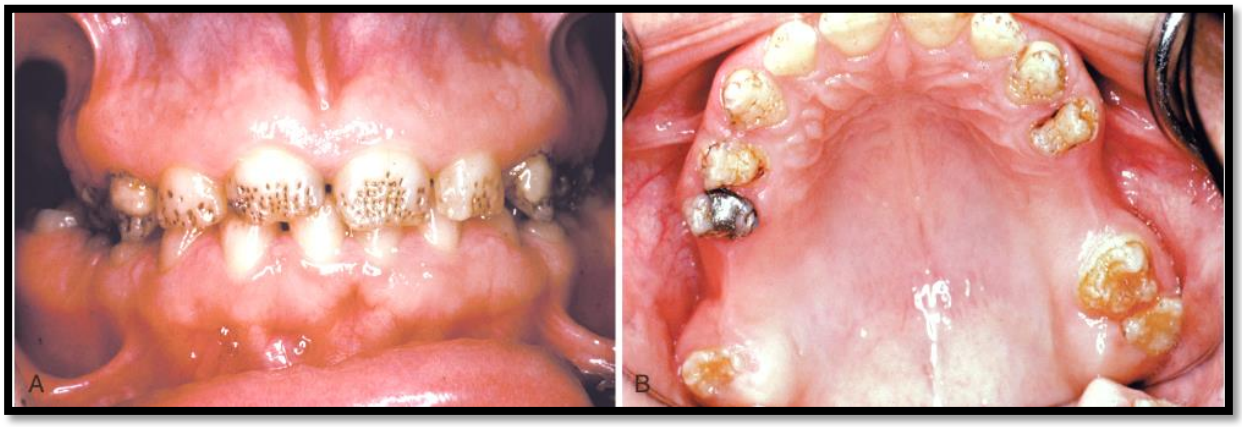


Figure (1.5): Hypoplastic Amelogenesis Imperfecta, Generalized Pitted Pattern(**(Chi *et al*., 2017).**

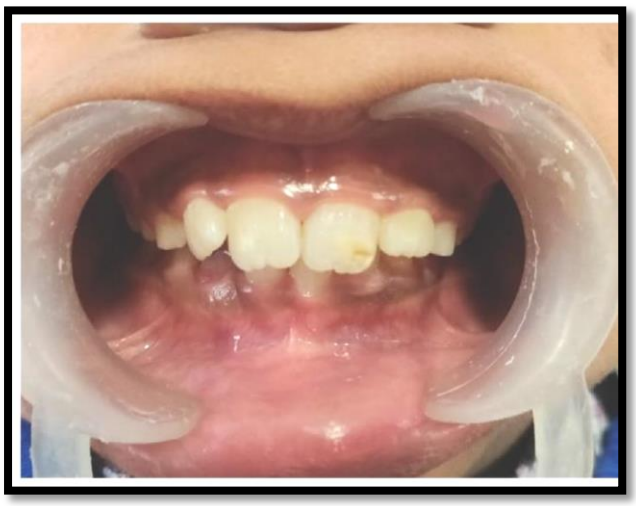


Figure (1.6): Hypoplastic Amelogenesis Imperfecta, localized Pattern **(Tyagi., 2019).**

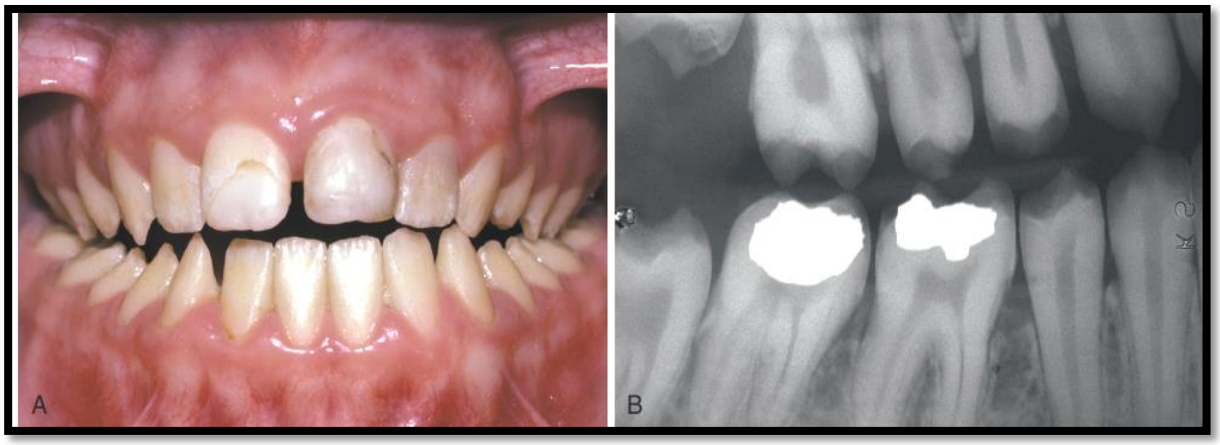


Figure (1.7): Hypoplastic Amelogenesis Imperfecta, Smooth Pattern **(Chi *et al*., 2017).**

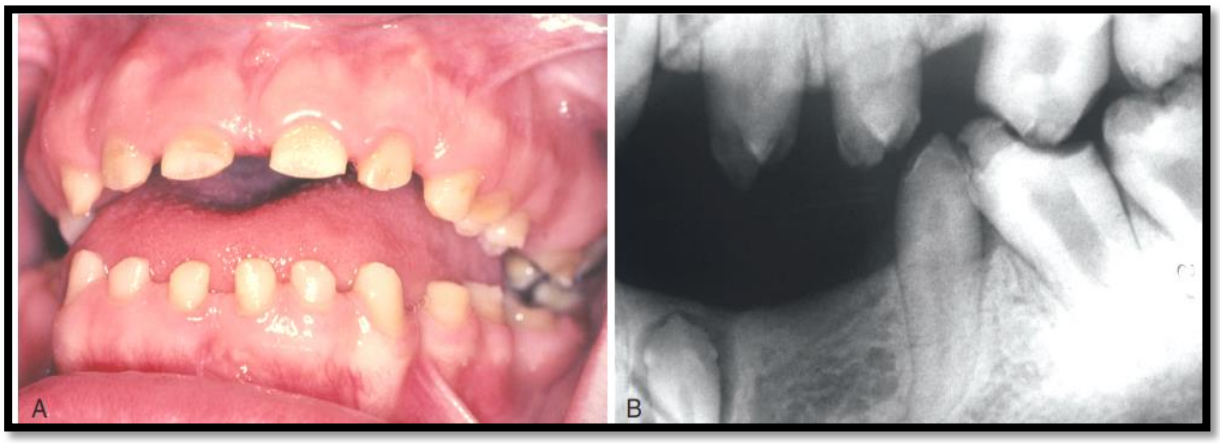


Figure (1.8): Hypoplastic Amelogenesis Imperfecta, Rough Pattern **(Chi *et al*., 2017).**

Classification of hypoplastic AI according to the mode of inhertance (Sabandal and Schäfer, 2016).

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.

2-Hypomaturation AI (typeII)

Hypomaturation AI (typeII) consists of a defect in matrix protein degradation. In enamel, which is the most calcified structure in the organism, proteins must be degraded and removed to achieve final crystal growth. So there is normal matrix formation and defective mineralization and occur during the maturation stage, therefore there is a qualitative defect in mineralization. Hypomaturation type represents 20-40% of all the cases of amelogenesis imperfecta **(Toupenay *et al*., 2018).**

Clinical features

1. The color of teeth here varies from creamy opaque to marked yellow/brown.
2. The surface of the teeth appear soft and rough leading to sensitivity due to dentinal exposure but not as severe as the hypocalcified type.
3. The enamel thickness is normal but often chips off and abrades away easily (**Figure 1.9)(Gemimaa and Arvind, 2014; Roma and Hegde, 2016).**

Radiographically

Contrast between enamel and dentin is lost, similar radiodensity as dentine **(Gadhia *et al*., 2012).**

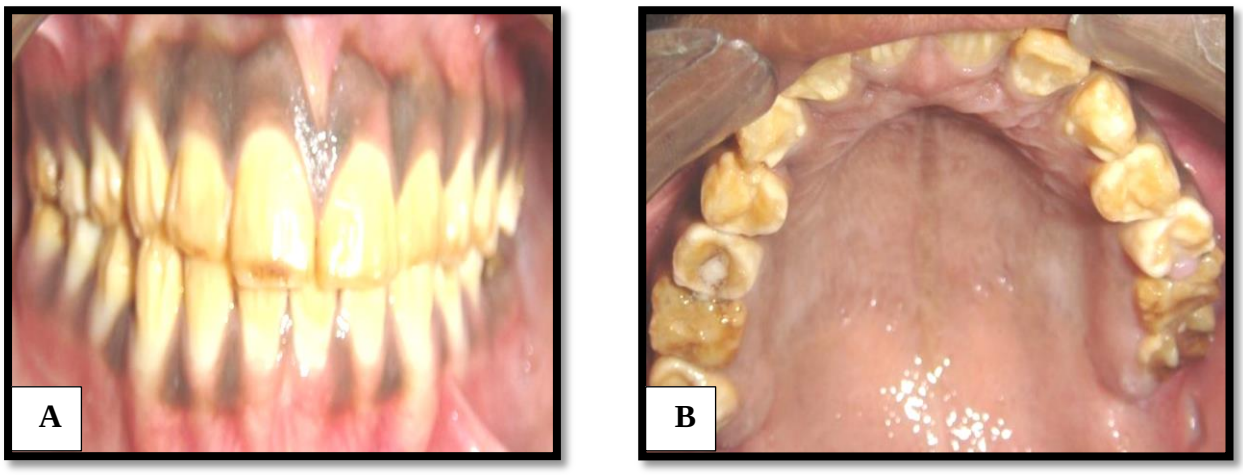


Figure (1.9): Hypomaturation AI **A**:Creamy opaque, yellow/ brown. **B:** Occlusal view of hypomaturation enamel **(Gemimaa and Arvind, 2014).**

Subtype classification of hypomaturation AI according to the clinical appearance (Claudio *et al*., 2021).

1- The pigmented pattern : the enamel has mottled and brown appearance
2- Snow- capped pattern: the enamel has zone of opaque white on incisal and occlusal edges.

Classification of hypomature AI according to the mode of inheritance (Patel *et al*., 2013).

Type IIA: Hypomaturation, pigmented autosomal recessive.

Type IIB: Hypomaturation, X-linked recessive.

Type IIC: Hypomaturation, snow-capped teeth, X-linked Type.

Type IID: Hypomaturation, snow-capped teeth, autosomal dominant.

3-Hypocalcified AI (Type III)

Qualitative defect occurs when the enamel is insufficiently mineralised and soft with normal matrix formation. on comparison with hypomaturation variety, the degree of mineralization is markedly reduced. Is associated with defects in calcification and appears in enamel with normal thickness at the time of eruption. Hypocalcified type represents 7% of all the cases of amelogenesis imperfecta. Because of the poor mineralization, the enamel rapidly wears down and X-rays show less opacity **(Möhn *et al*., 2021).**

Enamel mineral content is reduced causing pain while masticating, and brushing. Gingivitis and periodontal diseases have been described, with large amounts of dental calculus. Teeth are very sensitive to temperature and brushing **(Toupenay *et al*., 2018).**

Clinical features

1. the crowns of the teeth in such cases appears to be opaque white to yellow-brown, soft rough enamel surface.
2. dental sensitivity and very poor aesthetic
3. Due to severe hypomineralization, there may be early loss of enamel.

4. The thickness of enamel appears to be normal at eruption that often chips and but, tends to abrade easily post eruptively.

5. There may be delayed eruption of teeth.

6. An anterior open bite of skeletal origin may be seen.

7. Accumulation of a large amount of supragingival calculus is evident **(Figure 1.10A)(Regezi *et al*., 2016; Roma and Hegde 2016).**

Radiographically

Normal enamel thickness (immediately after tooth eruption) but less radiopacity compare to dentine **(Figure 1.10B) (Gerdolle *et al*., 2015; Sabandal and Schäfer, 2016).**

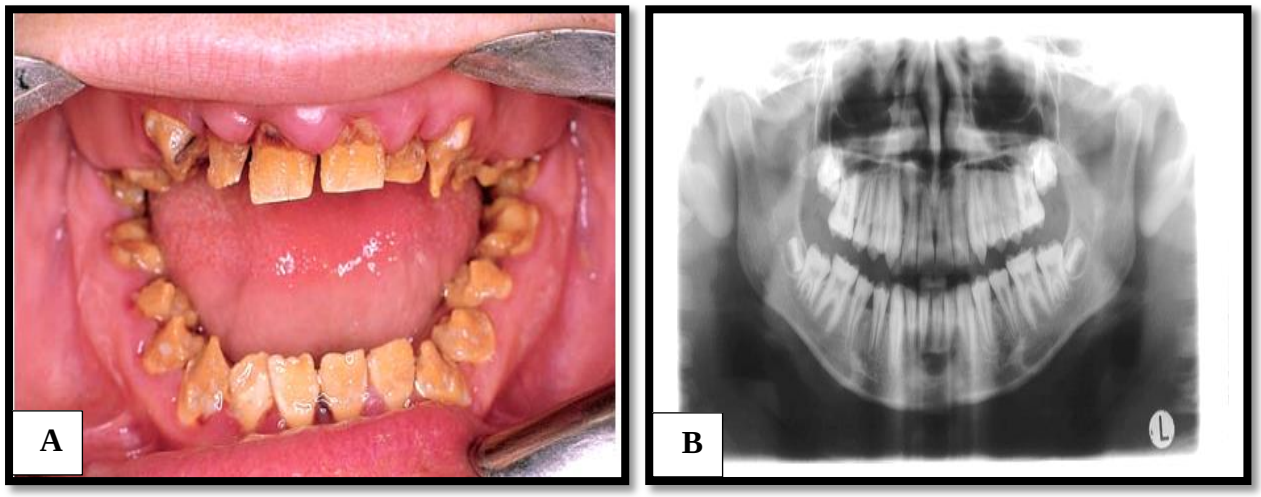


Figure (1.10): Hypocalcified Amelogenesis imperfacta **A:** clinical image (**Regezi *et al*., 2016) B:** radiographic image **(Sabandal and Schäfer, 2016).**

Classification of hypocalcification AI according to mode of inheritance (Sabandal and Schäfer, 2016).

Type IIIA: Autosomal dominant.

Type IIIB: Autosomal recessive.

4-Hypomaturation-Hypoplastic with Taurodontism (Type IV):

This type of amelogenesis imperfecta exhibits enamel hypoplasia in combination with hypomaturation. The deciduous and the permanent dentitions are diffusely involved **(Shivhare *et al*., 2016).** Historically, two patterns have been recognized that are similar but differentiated by the thickness of the enamel

and the overall tooth size. Hypomaturation-Hypoplastic type represents 21% of all the cases of amelogenesis imperfecta (**Adorno-Farias *et al*., 2019).** There are two subtype according to the predominant defect:

1. Hypomaturation-hypoplastic pattern, the predominant defect is one of enamel hypomaturation in which the enamel appears as mottled yellow-white to yellow-brown. Pits are seen frequently on the buccal surfaces of the teeth.

2. Hypoplastic-hypomaturation pattern, the predominant defect is one of enamel hypoplasia in which the enamel is thin but also hypomature **(Chi *et al*., 2017).**

Clinical features

1. Mixed hypomature and hypoplastic appearance
2. The enamel is thin, mottled yellow to brown. Molar teeth exhibit taurodontism, and other teeth have enlarged pulp chambers
3. Taourodontism common feature
4. Teeth appear smaller than normal and they lack proximal contact
5. The crown show pitting and tend to have hypomineralized area **(Figure 1.11A) (Visram and McKaig, 2006; Adorno-Farias *et al*., 2019).**

Radiographically

The enamel contrast is normal to slightly greater than dentin, and shows large or bulbous pulp chambers which appear taurodontic **(Figure 1.11B) (Roma and Shreya, 2016; Adorno-Farias *et al*., 2019).**

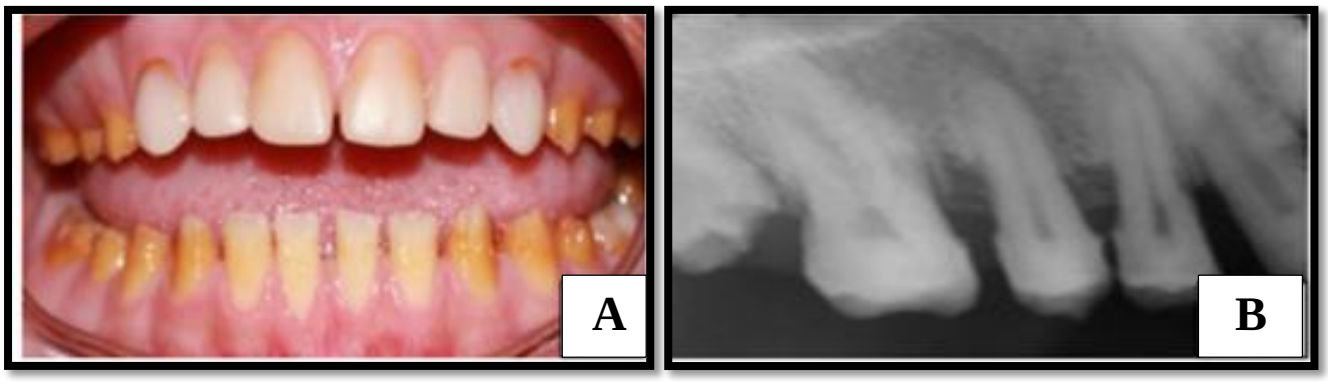


Figure (1.11): Hypomaturation-Hypoplastic AI **A:** Clinical image **B:** Radiographic image **(Adorno-Farias *et al*., 2019).**

Classification of hypomaturation and hypoplastic AI according to the mode of inheritance (Sabandal and Schäfer, 2016).

IVA: Hypomaturation–hypoplastic with taurodontism, autosomal dominant.

IVB: Hypoplastic–hypomaturation with taurodontism, autosomal recessive

1.2.1.3 Diagnosis of amelogenesis imperfecta

The severity of AI can vary significantly between patients and often it is difficult to make a diagnosis of the phenotype from clinical examination alone. In some cases the different phenotypes described may coexist in the same patient and on the same tooth. Clinical presentation can range from mild discolouration, slight pitting and minimal post eruptive breakdown of enamel to severe discolouration, pitting or signifcant tooth surface loss due to rapid post eruptive breakdown of hypomineralised enamel **(Patel *et al*., 2013).**

A dentist can identify and diagnose amelogenesis imperfecta on the basis of the patient's family history and the signs and symptoms present in the affected individual. Extraoral X-rays (X-rays taken outside the mouth) can reveal the presence of teeth that never erupted or that were absorbed. Intraoral X-rays (X-rays taken inside the mouth) show contrast between the enamel and dentin in cases in which mineralization is affected. Genetic testing is available for the genes AMELX, ENAM, and MMP20 **(Crawford *et al*., 2007).**

Accurate diagnosis enables genetic counseling in an early phase, and precautionary steps can be taken as an early step to prevent further dental complications for the patient and even for upcoming siblings in the future. Histological confirmation can be done, but required extraction of the affected tooth, which is not a good idea in all cases except where prognosis of that tooth is poor **(Shivhare *et al*., 2016).**

1.2.1.4 Associated dental features with AI

AI may be associated with some other dental and skeletal developmental defects or abnormalities, such as crown and root resorption, attrition,

microdontia, taurodontism, delayed eruption and tooth impaction, dens in dente, pulp stones, anterior open bite, cross bite and agenesis of teeth **(Shivhare *et al*., 2016).**

Anterior open bite is the result of defects in the eruptive mechanism, secondary to the disturbances of the enamel epithelium. Other common causes of anterior open bite are tongue thrusting and thumb sucking habits seen in AI patients **(Koruyucu *et al*., 2014).**

1.2.1.5 Clinical consideration

The AI can have variety of presentation. This condition can impact both primary and permanent dentition . The clinical implications of AI are significant for patients and clinicians. They include esthetic involvement, hypersensitive teeth, mastication impairment and psychosocial affections and others as showed in **table (1.3)(Gerdolle et al., 2015; Acosta-de Camargo *et al*., 2021).**

Because AI is a rare and heterogeneous condition from a clinical and genetic point of view, dentists, in general, have difficulty in making a correct diagnosis regarding the presence of AI and the determination of its clinical subtype **(Adorno-Farias *et al*., 2019).**

Table (1.3): The most common clinical consideration related to AI **(Geridolle *et al*., 2015).**

Compromised periodental health
Asymmetric gingival contour
Hypersensitive tooth
High risk of cavities
Discolored and pitted surface
Diminutive teeth with short clincal crown height
Malformed tooth
Congenital missing tooth
Pulp calcification
Taurodontisim
Anterior and posterior open bite
Multiple posterior diastemata
Loss of vertical dimension of occlusion
Impaired esthetics

1.2.1.6 Management (Treatment) of AI

Objectives of the treatment (Toupenay *et al*., 2018).

1. Pain prevention and treatment
2. Protection of dental tissue integrity in order to maintain occlusal function and limit dental biofilm retention
3. Restoration of smile aesthetics
4. To alleviate the sensitivity

Management directed at three aspects of treatment includes prevention, restoration, and esthetics. Most of the time patient report to the dentist when the dental complication like dentinal sensitivity or dental caries would have been started, so restoration should be undertaken as a first step **(Shivhare *et al*., 2016).**

Treatment options available to restore patients with AI vary considerably depending on several factors such

1. Age of the patient
2. Patient motivation
3. Periodontal condition, endodontic status
4. Loss of tooth structure, severity of disorder
5. Socioeconomic status and most importantly the patient's availability for treatment and cooperation **(Patel *et al*., 2013).**

1.2.1.6.1 Treatment options of AI

1-Direct composite

Composite resins can be used in mild cases to veneer the surface of the teeth or for more extensive build ups in more advanced cases . The advantage of direct composite restorations are that they do not require complex aboratory procedures, the treatment is reversible and it is relatively quick. Sound tooth structure is preserved, as they require very minimal bevelling preparation of the teeth or no preparation at all. It is also a relatively inexpensive treatment **(Novelli *et al*., 2021).** Direct restorative materials, such as amalgam, glass ionomer

cements, composite resins, resin-modified glass ionomer cements, have been advocated for restoring teeth with AI **(Figure 1.12)(Chiung *et al*., 2013; Abd Alraheam and Donovan, 2020).**

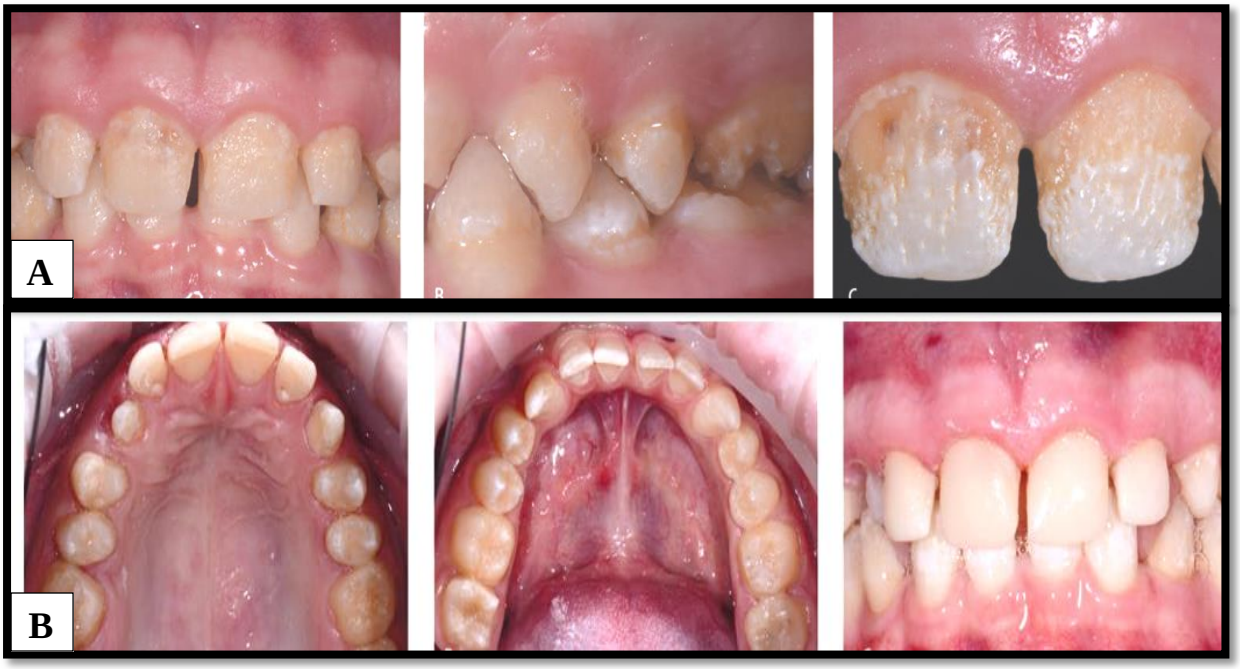


Figure (1.12): **A:** Patient intraoral photographs showed hypoplastic AI before treatment **B:** Patient intraoral photographs after composite rehabilitation**(Abd Alraheam and Donovan, 2020).**

2-Indirect composite

Indirect composite onlays or crowns can be used to restore posterior teeth or anterior teeth where extensive tooth tissue loss has occurred and moisture control is difficult to achieve for direct buildup of teeth with composite (**Figure1.13) (Preissner *et al*., 2013).**

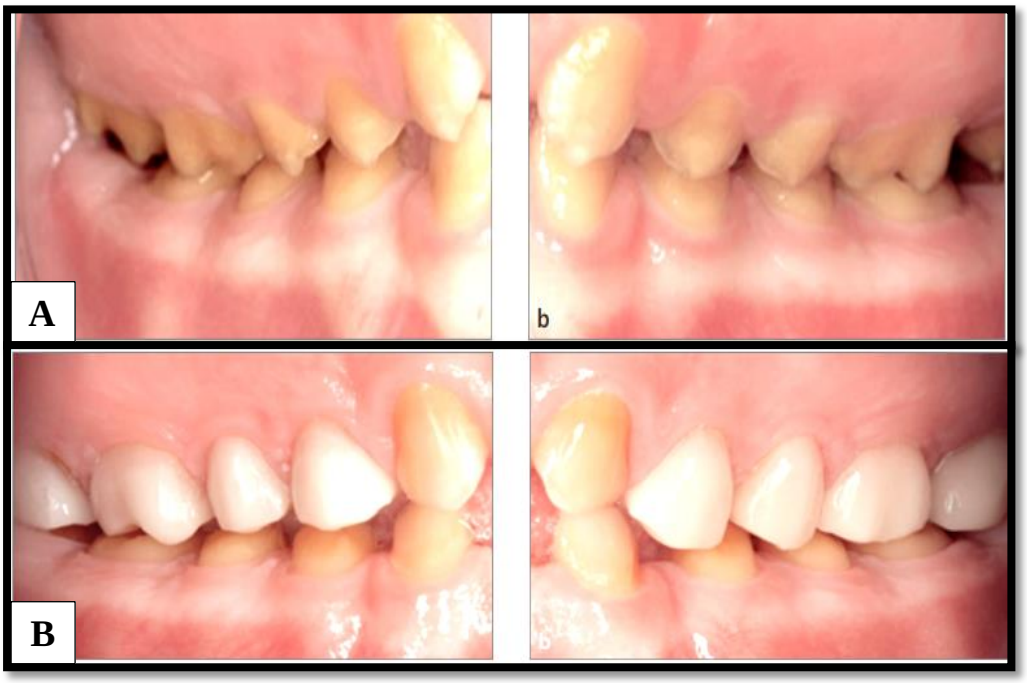


Figure (1.13): A: before treatment with composite resin crowns **B:** final situation maxillary posterior teeth with no preparation composite resin crowns **(Preissner *et al*., 2013).**

3-Porcelain veneers

These restorations are popular in the anterior region because they can achieve excellent aesthetic results with a relatively conservative tooth preparation when compared to a full coverage crown**(Patel *et al*., 2013)** composite resin veneers may be used to mask the discoloration and improve the crown morphology and contact with adjacent teeth (**Figure 1.14)(Chiung *et al*., 2013; Acosta-de Camargo and Mangles, 2021).**

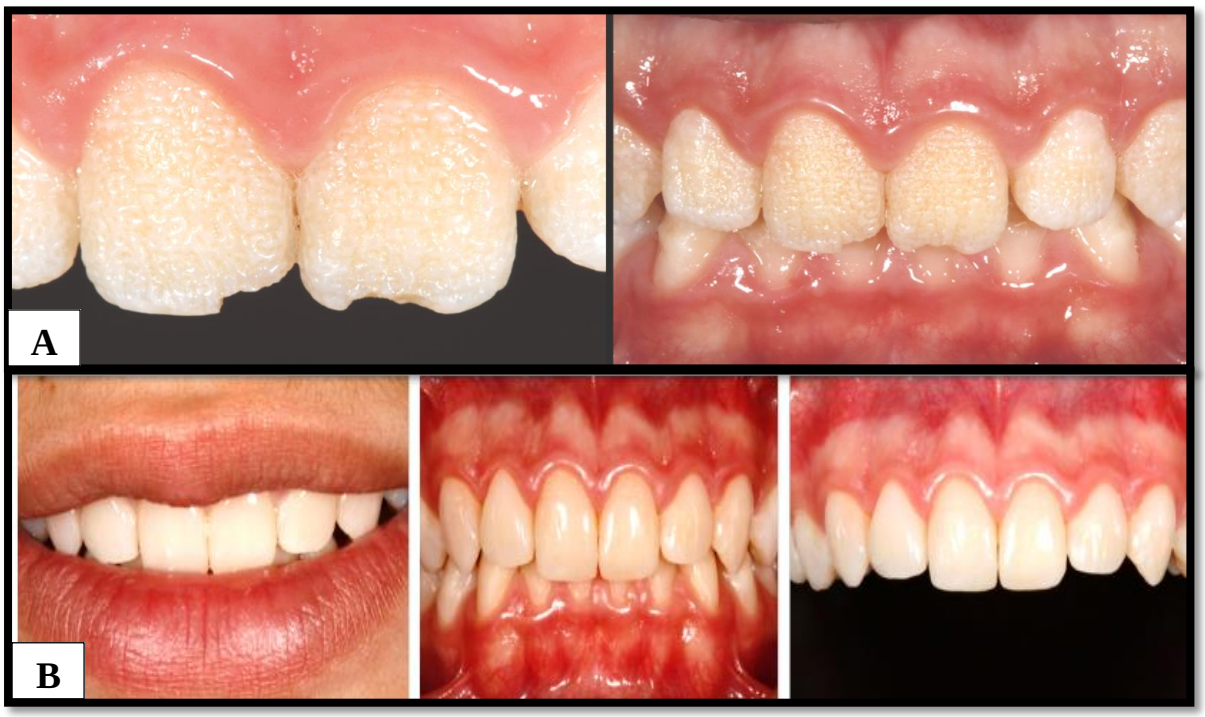


Figure (1.14): **A:** Frontal view of Hypoplasic Amelogenesis imperfect before treatment **B:** Frontal view of completed treatment with direct resin veneers **(Acosta-de Camargo and Mangles, 2021).**

4-Dentures

Treatment of patients with AI has included extractions and the fabrication of complete or partial dentures when unaesthetic appearance, increased sensitivity, extensive loss of tooth structure and vertical dimension and un restorable tooth **(Verma *et al*., 2019).**

Overdenture has been suggested for children as it is alterable to accommodate their growth process. Overlay denture can be used as a provisional or permanent prosthesis in some patients and can provide reversible and relatively inexpensive option **(Figure 1.15)** (**Ghodsi *et al*., 2012).**

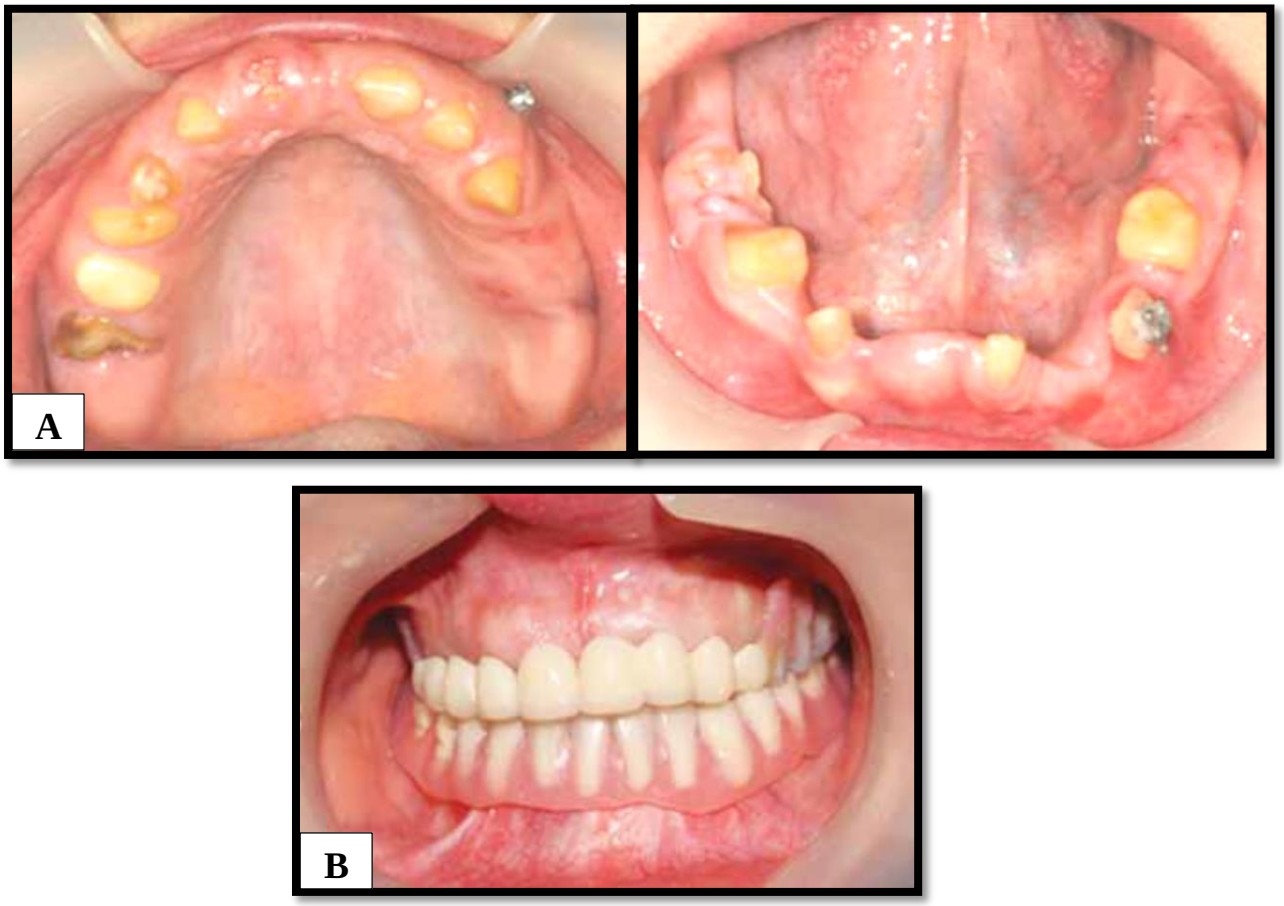


Figure (1.15): A: Intraoral view of the patient with amelogenesis imperfecta before treatment **B:** intraoral view after treatment with Partial overlay and overdenture (**Ghodsi *et al*., 2012).**

5-Implants

In advanced cases of AI where the teeth are unrestorable and the patient is seeking a fixed option, dental implants can be considered. Careful planning is essential and timing of extractions with respect to implant placement is very important to preserve bone, which will resorb away relatively quickly following tooth extraction **(Patel *et al*., 2013).**

6-Bleaching and microabrasion

In patients with AI, preservation of tooth structure is vital and minimally invasive treatment options must be considered where possible. Microabrasion using an acidic slurry containing 18% hydrochloric acid and pumice is often effective in removing superficial stains and improving the appearance of the teeth **(Figure 1.16)(Patel *et al*., 2013).**

An alternative conservative approach is the use of long term bleaching or tooth whitening since amelogenesis imperfecta have inherited intrinsic stains. Intrinsic stains cannot be removed easily by regular prophylactic measures, it requires special agents that can penetrate the matrix of enamel and dentine by using hydrogen peroxide and it is represent minimally invasive treatment and satisfactory aesthetic improvement has been reported **(Yano *et al*., 2021).**

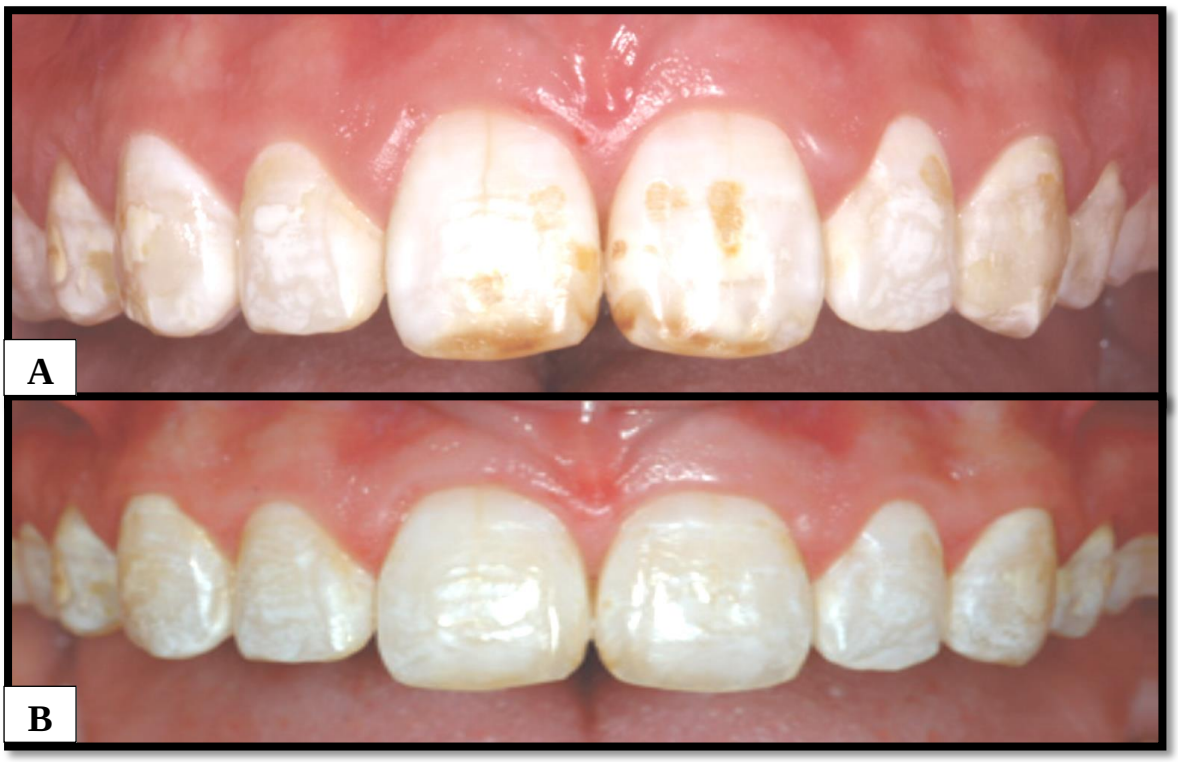


Figure (1.16): A: Intraoral view of the patient with amelogenesis imperfect before treatment **B:** intraoral view after treatment with microabrasion (**Patel *et al*., 2013).**

7- Crown lengthening surgery

Often patients with AI have reduced clinical crown height due to loss of tooth structure resulting from enamel chipping away and tooth wear. This tooth surface loss may be compensated for by dentoalveolar compensation leaving a 'gummy' appearance to the patient's smile. The extensive loss of tooth structure renders the clinical crown height inadequate for the placement of restorations and so clinical crown lengthening is required **(Figure 1.17)(Kalaivani *et al*., 2015).**

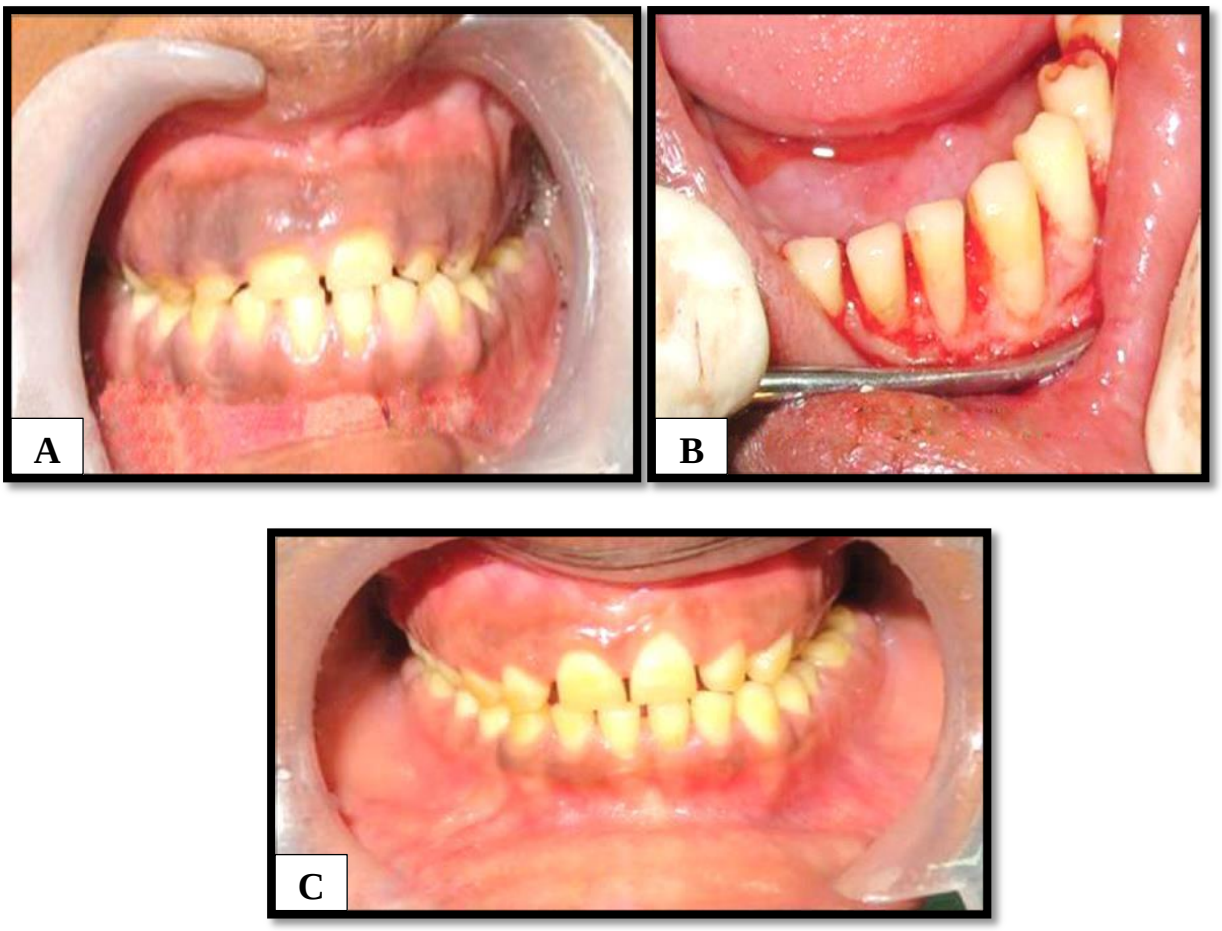


Figure (1.17): A: Intraoral view of the patient with amelogenesis imperfecta showing short clinical crowns **B:** Intraoperative view after ostectomy **C:** Postoperative healing after crown lengthening surgery **(Kalaivani *et al.*, 2015).**

Chapter Two
Conclusion

Conclusion

Amelogenesis imperfecta is a rare disorder that affects the form and function of teeth and has a major impact on the social acceptance of the patients particularly during adolescence.

One of th**e** greatest challenges faced by the clinician is the total rehabilitation with Amelogenesis Imperfecta. The enamel formation is affected in multiple ways. The color, thickness and resistance of enamel is reduced to a great extent. Both deciduous and permanent dentition are affected with AI, but more commonly seen in permanent teeth.

Early intervention and multidisciplinary treatment plan should be carried out for indicated situation while implementing careful evaluation, case selection, treatment planning and precise surgical procedures in order to meet the aesthetic and functional demands of the patient with the modern technology.

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