

Amelogenesis imperfecta  
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The background features a light beige grid pattern. In the top-left corner, there is a white wavy line with several brown four-pointed stars. In the bottom-right corner, there is another white wavy line with three brown four-pointed stars. On the left side, there is a stylized plant with dark green leaves and a brown circular shape behind it. On the right side, there is a stylized plant with dark green leaves and a white circular shape with a brown center behind it.

introduction

## introduction

Amelogenesis imperfecta is a congenital disorder characterized by abnormal formation of the teeth affects only the ectodermal portion of the teeth .Enamel is composed mostly of mineral, that is formed and regulated by the proteins in it. Amelogenesis imperfecta is due to the malfunction of the proteins in the enamel (ameloblastin, enamelin, tuftelin and amelogenin) as a result of abnormal enamel formation via amelogenesis



# Amelogenesis

Enamel of tooth is one of the four major tissues that make up the tooth in humans. It makes up the normally visible part of the tooth, covering the crown.

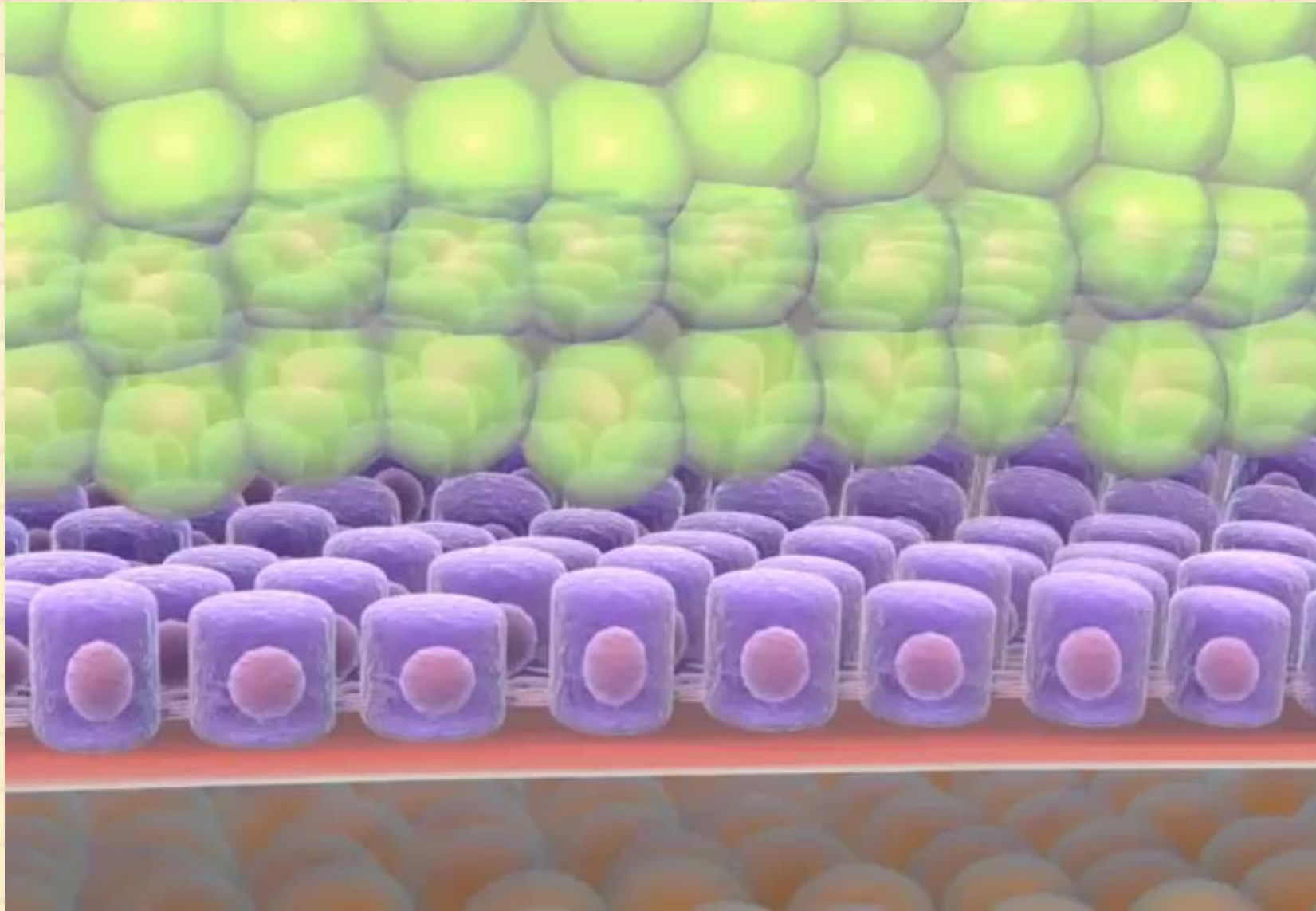
- very hard,
- white to off- white
- highly mineralized
- acts as a barrier to protect the tooth
- In rare circumstances enamel fails to form, leaving the underlying dentin exposed on the surface.



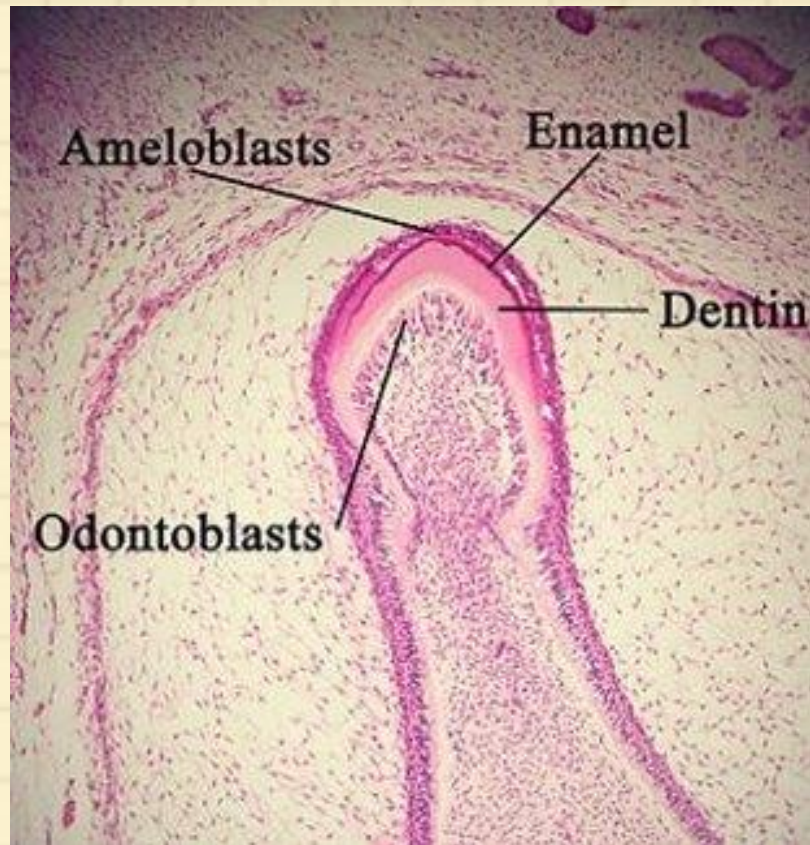
# Development



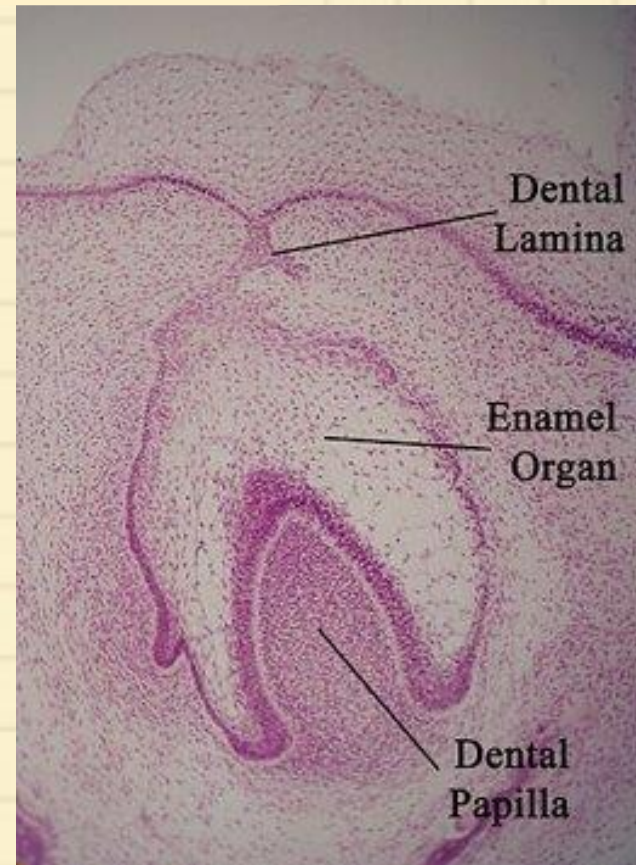
- Human enamel forms at a rate of around 4  $\mu\text{m}$  per day.
- beginning at the future location of cusps.
- around the third or fourth month of pregnancy.
- creation of enamel divided into two stages :
- The first stage, called the secretory stage, involves proteins and an organic matrix forming a partially mineralized enamel.
- The second stage, called the maturation stage, completes enamel mineralization.







Histologic slide showing enamel formation



Histologic slide showing a developing tooth. The mouth would be in the area of space at the top of the picture.



# Development



- ❖ enamel, unlike many other tissues of the body, has no way to regenerate itself. After destruction of enamel from decay or injury, neither the body nor a dentist can restore the enamel tissue. Enamel can be affected further by non-pathologic processes.
  
- ❖ Enamel is covered by various structures in relation to the development of tooth:
  - Nasmyth membrane or enamel cuticle, structure of embryological origin is composed of keratin which gives rise to the enamel organ.
  - Acquired pellicle, structure acquired after tooth eruption is composed of food debris, calculus, dental plaque (organic film).

# Amelogenesis imperfecta

## Etiology

Intrinsic enamel defect that affects all teeth of both dentitions Results from defective amelogenin genes on X and Y chromosomes and also chromosome 4 At least 16 variants noted based upon inheritance pattern, enamel qualities, and radiographic features.

# Amelogenesis imperfecta

The development of enamel or amelogenesis is completed in three main stages:

1. The formative stage (Characterized by deposition of the matrix ) Defect in the formative stage lead to hypoplastic (type I).

Characteristics of hypoplastic AI

1. Enamel of reduced thickness
2. Pitting and grooves
3. Hard and translucent enamel
4. Radiographically, the enamel contrasts normally from dentine.
5. 60\_70 % of AI are hypoplastic type.



# hypoplastic AI



Hypoplastic pitted amelogenesis imperfecta

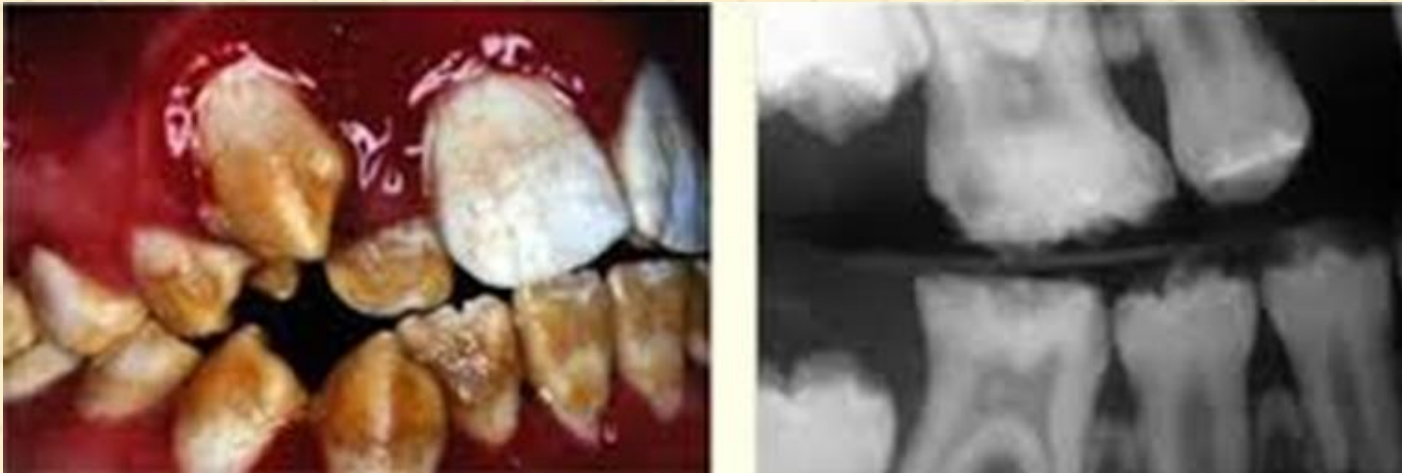


Hypoplastic pitted amelogenesis imperfecta

## Radiographic features

### 1. Hypoplastic

- shape the enamel appears thinner
- it maybe seen only in cusp tips or in sever cases totally absent
- since the calcification of the enamel is not distort radiodensity of the enamel is normal therefor shows normal contrast with underlying dentin



hypoplastic amelogenesis imperfecta in radiograph



# Amelogenesis imperfecta



hypoplastic AI is subdivided into four other pattern :

1. Generalized ( affect the entire dentition )
2. Localized ( affect some teeth in the oral cavity)
3. Smooth the Enamel thin hard and gloosy.
4. rough he enamel is thin hard and rough .



# hypocalcified AI



2. Calcification stage (Characterized by matrix mineralization ) Characteristics of hypocalcified AI

- Defect in enamel calcification
- Enamel of normal thickness
- Appears opaque or chalky
- Weak in structure
- Teeth become stained and rapidly wear down
- Radiographically, enamel is less radio-opaque than dentine.
- 7% of AI are hypocalcified type

## Radiographic features

### 2. Hypocalcified

1. As the enamel matrix is not defective the crown shape is not altered
2. normal enamel thickness
3. problem is in the calcification which makes the radiodensity of the enamel reduced it may appear similar to the underlying dentin



hypocalcified amelogenesis imperfecta diffuse yellowish brown discoloration



## hypo maturation AI

### 3. Maturation stage (Characterized by matrix maturation ) Characteristics of hypo maturation AI

1. Enamel of normal thickness but mottled in appearance
2. Slightly softer than normal and vulnerable to tooth wear, but not as severe as the hypocalcified type
3. Radiographically, similar radiodensity as dentine
4. 20\_40% of AI are hypo maturation type.





# hypomaturation AI

hypomaturation AI subdivided into two pattern

1. Pigmented pattern

Has a mottled and brown appearance



## hypomaturation AI



### 2. Snow capped

Zone of opaque white enamel is seen on incisal or occlusal edges

. Hypomaturational snow capped amelogenesis imperfecta

## Radiographic features



### 3. hypomaturation

The radiographic features are similar to hypocalcified type the enamel thickness is normal but the radiodensity is reduced



## type IV AI

1. Mixed hypomaturation and hypoplasia appearance
2. Taurodontism: body and pulp chamber enlarged, and the floor of pulp chamber and furcation is moved apically down the root



combination of hypoplastic and hypomaturation  
amelogenesis imperfecta



Taurodontism seen with permanent  
mandibular molar

# Genetics

Several gene expression is needed for enamel formation where the relevant matrix proteins & proteinases are transcribed for regular crystal growth & enamel mineralization.

- AMELX and ENAM encode extracellular matrix proteins of the developing tooth enamel
- KLK-4 and MMP20 encode proteases that help degrade organic matter from the enamel matrix during the maturation stage of amelogenesis.
- SLC24A4 encodes a calcium transporter that mediates calcium transport to developing enamel during tooth development.
- Mutations in the ENAM gene are the most frequent known cause and are most commonly inherited in an autosomal dominant pattern.
- About 5% of amelogenesis imperfecta cases are caused by mutations in the AMELX gene and are inherited in an X-linked pattern.



- Recessive genetic disorders occur when an individual inherits two copies of an abnormal gene for the same trait, one from each parent.
- Parents who are close relatives have a higher chance than unrelated parents to both carry the same abnormal gene
- Dominant genetic disorders occur when only a single copy of an abnormal gene
- X-linked genetic disorders are conditions caused by an abnormal gene on the X chromosome and manifest mostly in males.
- A male cannot pass an X-linked gene to his sons
- Males with an abnormal gene for an X linked dominant disorder are more severely affected than females and often do not survive.



# diagnosis

- family history
- pedigree plotting (a diagram of a family health history tree)
- clinical observations
- radiographic assessment.
- It is important to identify other causes of defects in enamel, both extrinsic and intrinsic, to exclude these during diagnosis of AI like past medical history (tetracycline staining), dental fluorosis, enamel hypoplasia, trauma, or molar-incisor hypomineralisation (MIH).

The examination of the teeth should be done to check

- discoloration
- Changes in shape
- Hardness of the enamel



# Epidemiology



The exact incidence of amelogenesis imperfecta is uncertain. Estimates vary widely, from 1 in 700 people in northern Sweden to 1 in 14,000 people in the United States. The prevalence of amelogenesis imperfecta in non-human animals has not been explored, however its presence has been noted.

# Treatment



The main objectives of treatment is :

pain relief, preserving patient's remaining dentition, and to treat and preserve the patient's occlusal vertical height.

The teeth is protected by use :

- Preformed metal crowns on the posterior teeth
- Esthetic restoration like ceramic crown on the anterior teeth
- In case of loss of mature tooth structure by attrition overdenture are provided



# Case Report

A 12-year-old boy was referred to the Department of Pediatric Dentistry, university of Baghdad for examination, evaluation and treatment of gross attrition and considerable sensitivity of his teeth.



Frontal view (pretreatment)



Occlusal view of the maxillary arch (pretreatment mirror view)



Occlusal view of the mandibular arch (pretreatment mirror view)





Frontal view of the left and right sides (posttreatment)





Occlusal view of the maxillary arch (posttreatment mirror view)



Occlusal view of the mandibular arch (posttreatment mirror view)





Control view after 24 months

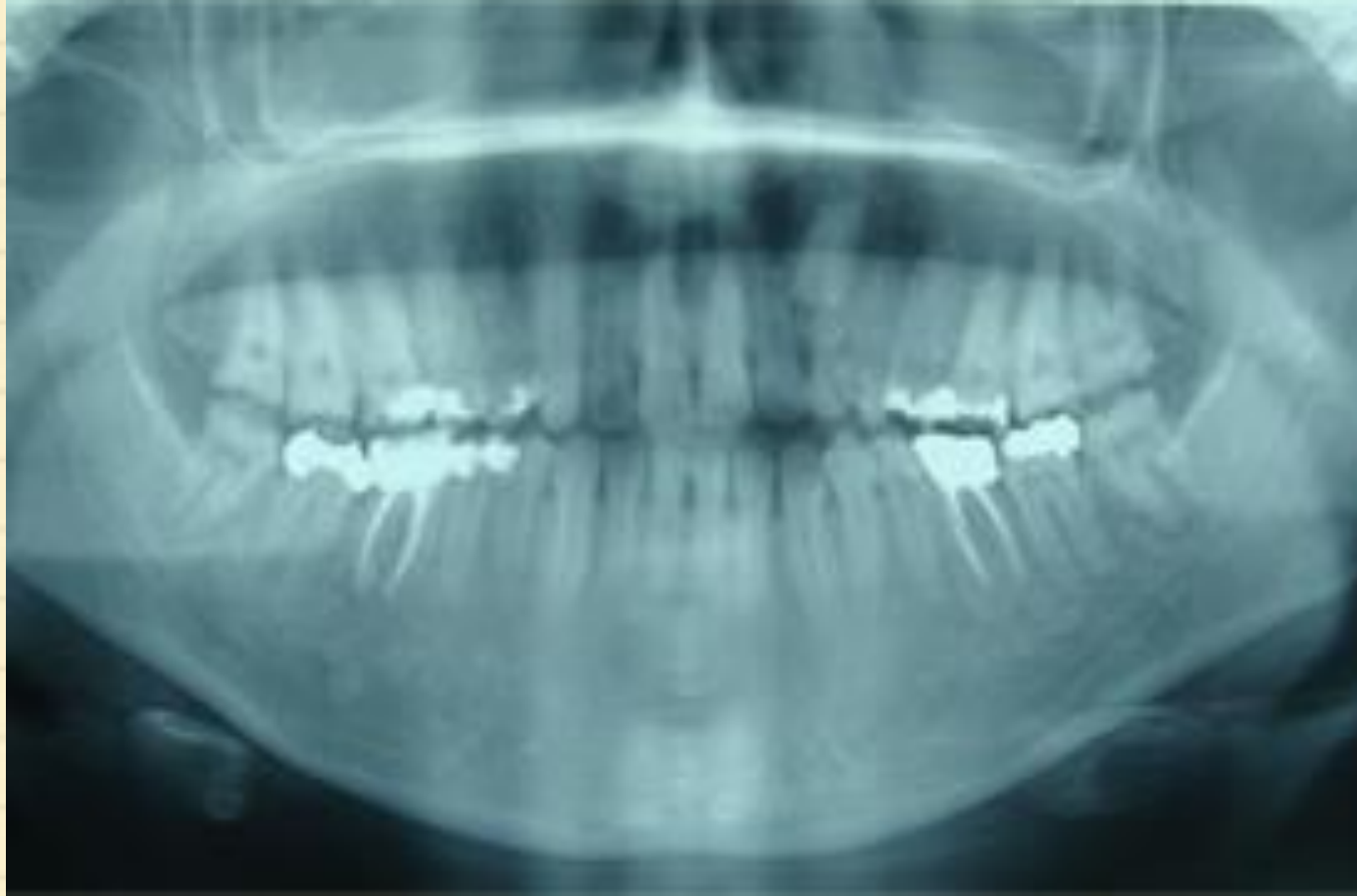
# Case Report

A 19-year-old woman was referred to Baghdad university's department of prosthodontics with the chief complaint of discolouration and unpleasant esthetics.





Intraoral views before treatment show (a) discoloured teeth with rough surfaces and irregular defects, (b) class I molar occlusion on the right and (c) class III molar occlusion on the left.



Panoramic radiography before treatment reveals a very thin enamel layer, normal roots and pulp chambers, as well as earlier root canal therapy and fillings.



Intraoral view after preparation of the teeth.



post treatment intraoral (a) and radiographic (b) views showing restored teeth





The patient was very satisfied with the results in terms of both esthetics and function. Photographs taken after treatment: (a) from facial profile and (b) from right side.

# Conclusion

Several investigators have suggested a classification system for amelogenesis imperfecta, based on the phenotype and pedigree combined with scanning electron microscopic examination, biochemical methods, and molecular genetics. Thus the dentist has to diagnose the condition as early as possible to offer early intervention and balance the decision for early intervention and long-term survival of the restorations. Dental practitioners should consider the social implications for these patients and intervene to relieve their suffering.



Thank You

ANY QUESTION?

