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# Breaching the Gap between Clinical and Radiographic Diagnosis of Amelogenesis Imperfecta: A Review

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**Abstract:** ***Amelogenesis imperfecta encompasses a complicated group of conditions which demonstrate developmental alterations in the structure of enamel in the absence of systemic disorder or syndrome. A complex inheritance pattern gives rise to amelogenesis imperfecta. It affects the structure and appearance of enamel both in the primary and secondary dentition. Gene mutations which are responsible for deformed amelogenesis result in diverse phenotypes showing a wide spectrum of characteristics. It is important to understand the different phenotypes and associated radiographic findings related to AI to help narrow down the search for a candidate gene in order to establish a definitive molecular aetiology. Precise diagnosis of AI done by clinical and radiographic findings will help in stabilisation, restoration and regular maintenance of the patient.***

**Keywords:** Amelogenesis imperfecta, Hypoplasia, Hypomaturation, Hypocalcification, Taurodontism

## 1. Introduction

Amelogenesis imperfecta (AI) is a collective term used for a number of conditions with abnormal enamel formation. The first definition of AI was given by Weinmann et al (1945) – "as a disease caused by a primary defect in enamel" & they classified AI into two types, namely hypoplastic and hypocalcified.[1,2] Aldred MJ in 2003 gave a refined definition in order to diagnose AI –"A group of conditions, genomic in origin, which affect the structure and clinical appearance of enamel of all or nearly all the teeth, and which may be associated with morphologic or biochemical changes elsewhere in the body."[1]Amelogenesis imperfecta (AI) comprises a group of low prevalence hereditary conditions that cause alterations in the structure or morphology and chemical composition of the enamel matrix during development[2,3] The condition affects both quality and quantity of the enamel. The average global prevalence of AI is <0.5% (<1 in 200).[2,4,5]

The condition is not a single entity. It consists of a number of subtypes, characterized by their varying modes of inheritance and different clinical and radiographic appearances. According to Witkop, AI can be categorized into 14 subtypes, which makes its diagnosis extremely complex. Although AI is generally considered to primarily affect the enamel, there are numerous reports of other manifestations associated such as taurodontism or elongation of the pulp chamber due to apical displacement of the root furcation, etc.[4,6] As AI is a rare and heterogeneous condition from a clinical and genetic point of view, dental clinicians, in general, have difficulty in making a correct diagnosis about the presence of AI and the identification of its clinical subtype.[3] Hence, it is a clinician's job to diagnose the condition rightly and to manage the manifestations in early stage.

## 2. Discussion

Amelogenesis is a process in which normal enamel is synthesized during tooth development as an extracellular matrix and it occurs in two stages- Secretory and Maturation. In the secretory stage, the ameloblasts produce a partially mineralized protein matrix that will correspond to the adult enamel. In the maturation stage, the protein matrix is degraded and mineralization gets completed.[3,7]

As the name suggests, amelogenesis imperfects is a condition associated with defective amelogenesis.

For diagnostic purpose, AI represents a group of conditions,genomic in origin, that affect the morphological appearance and chemical composition of enamel of all or nearly all the teeth in a more or less equal manner.[8]

**Classification of AI-**
Witkop and Rao in 1971 divided the conditions first into three groups[1,9]

**(A)Hypoplastic**
1) Autosomal dominant hypoplastic–hypomaturation with taurodontism
   - Winter type
   - Crawford type
2) Autosomal dominant smooth hypoplastic with eruption defect and resorption of teeth
3) Autosomal dominant rough hypoplastic
4) Autosomal dominant pitted hypoplastic
5) Autosomal dominant local hypoplastic
6) X-linked dominant rough hypoplastic

**(B) Hypocalcified**
1) Autosomal dominant hypocalcified

**(C) Hypomaturation**
1) X-linked recessive hypomaturation
2) Autosomal recessive pigmented hypomaturation
3) Snow-capped teeth, autosomal dominant
4) White hypomature spots?

There is revised classification given in 1988-

Classification of amelogenesis imperfecta proposed by Witkop (1988)[1,8,9]

**Type I Hypoplastic**
IA – hypoplastic, pitted autosomal dominant
lB – 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,
IIC – snow-capped teeth, X-linked
lID – autosomal dominant?

**Type III Hypocalcified**
IIA – autosomal dominant
IIB – autosomal recessive

**Type IV -Hypomaturation–hypoplastic with taurodontism**
IVA – Hypomaturation–hypoplastic with taurodontism, autosomal dominant
IVB – Hypoplastic–hypomaturation with taurodontism, autosomal dominant

Modified classification of AI[10]

| Inheritance | Phenotype | Related gene |
|---|---|---|
| Autosomal dominant | Generalized pitted | - |
| Autosomal dominant | Localized hypoplastic | ENAM |
| Autosomal dominant | Generalized thin | ENAM |
| Autosomal dominant | Diffuse hypocalcification | FAM83H |
| Autosomal dominant | Localized hypocalcified | FAM83H |
| Autosomal dominant | With taurodontism | DLX3 |
| Autosomal recessive | Localized hypoplastic | - |
| Autosomal recessive | Generalized thin | ENAM |
| Autosomal recessive | Diffuse hypomaturation | WDR72 |
| Autosomal recessive | Pigmented hypomaturation | MMP20,KLK4,C4orf26 |
| X-linked | Generalized thin | AMELX |
| X-linked | Diffuse hypomaturation | AMELX |
| X-linked | Snow-capped hypomaturation | - |

## 3. Diagnostic Methods

1) **Clinical :** Taking proper family history, plotting pedigree chart, clinical identification and meticulous recording play crucial role in clinical diagnosis of AI.[8]
2) Radiographic: Extra- oral radiographs may reveal the generalised enamel condition, teeth shapes unerupted teeth or resorbing teeth. Intra-oral radiographs will reveal the relative contrast between the enamel and dentin even better. This will provide the insight into degree of mineralization.[8,11]
3) Histopathologic: The histopathologic alterations seen in AI are not seen in routine preparations. There is loss of enamel as sectioning of paraffin embedded specimens causes decalcification of the teeth. To overcome this, ground section of nondecalcified sections are made.[5,8]
4) Genetic: This is based on genome studies. Because of higher cost, this is presently only a research tool.[2,3,10,13,14]

### 3.1 Clinical and radiographic features

**1) Hypoplastic type**
The enamel of the affected teeth fails to develop to its normal thickness. Hence, the colour of the underlying dentin gives yellowish brown colour to the teeth. There are two patterns seen- Generalized and localized. In generalized hypoplastic AI, Pinpoint- to- pinhead- sized pits are seen across the surfaces of the teeth which do not correlate with a pattern of environmental damage[2,4,5] The buccal surfaces are more severely affected. The pits may be arranged in rows or columns. Staining of the pits may occur.[5,8]The reduced enamel thickness may also cause a loss of contact between adjacent teeth. The occlusal surfaces of the posterior teeth may become flat. The enamel thickness, hardness and colour between the pits is normal.[10,11,15] In localized hypoplastic AI, horizontal rows of pits, a linear depression or one large area of hypoplastic enamel are seen. Mostly, the altered area is located in the middle third of the buccal surfaces of the teeth. The incisal edges or occlusal surfaces are usually spared[5,8]. Both primary and secondary dentitions or only the primary dentition may get affected. Autosomal recessive type is more severe as it involves all the teeth in both the dentitions. Hypoplastic AI is most easily identified on radiographicimaging.[10,11,15]

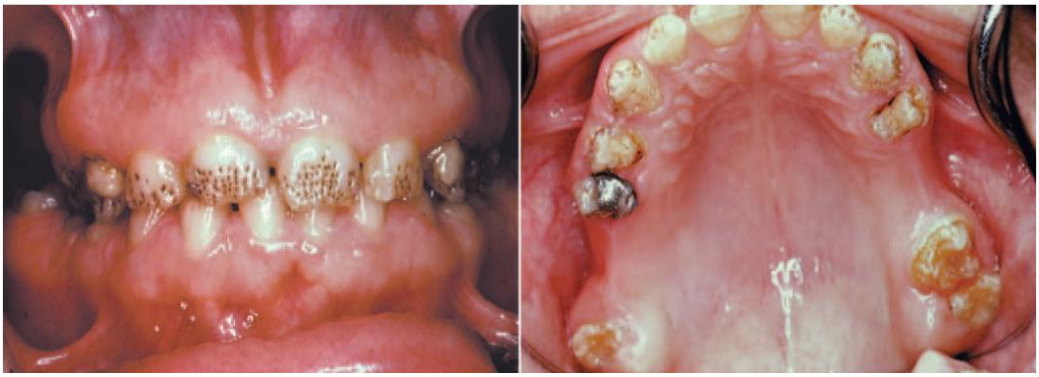

**Figure 1:** Hypoplastic Amelogenesis imperfecta- Numerous pits seen on buccal and occlusal aspects. The thickness of the enamel in between the pits is normal.[10]

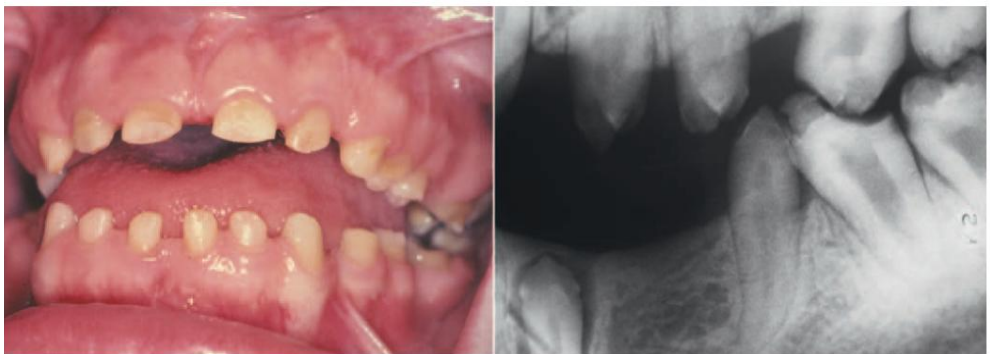

**Figure 2:** Small, yellowish brown teeth with open contacts and severe attrition

Radiographically, impacted tooth is seen and reduced radiopacity of enamel is noted.[10,11]

**2) Hypomaturation type**

In this type of AI, the enamel is mottled but is of normal thickness. The enamel density of the enamel is less and is softer in consistency than dentin. It may break away from the crown causing wearing of teeth.[5,8]This type is associated with enamel that chips out easily but does not establish massive loss upon eruption. The colour may vary from cloudy white, yellow or brown. The appearance where the teeth look like they have caps of white, opaqueenamel. It is called as "snow-capped" teeth.[10,11,15]

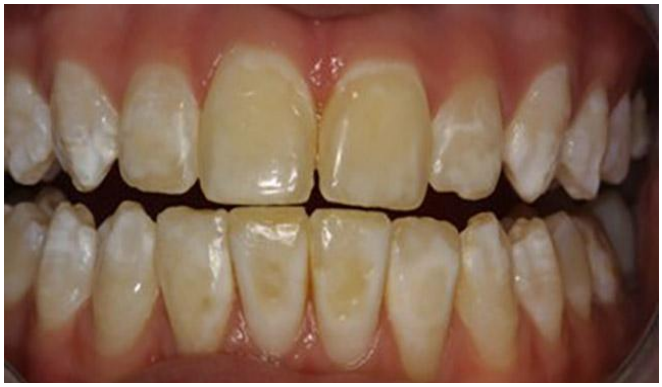

**Figure 3:** Hypomaturation Amelogenesis imperfecta- "Snow-capped" appearance

**3) Hypocalcification type**

It is a more common type than hypoplastic variety. [10]The thickness of the enamel is normal when they erupt and the teeth are of normal size and shape. As the enamel is poorly mineralized, it fractures easily after it comes into function. The enamel is called as "cheesy enamel" which is lost rapidly and diffusely except for the residual band in the cervical portion of the teeth.[5,8] The teeth wear off easily, sometimes to the level of gingiva. The hypocalcified enamel has increased permeability and gets stained and darkened easily.[10,15]

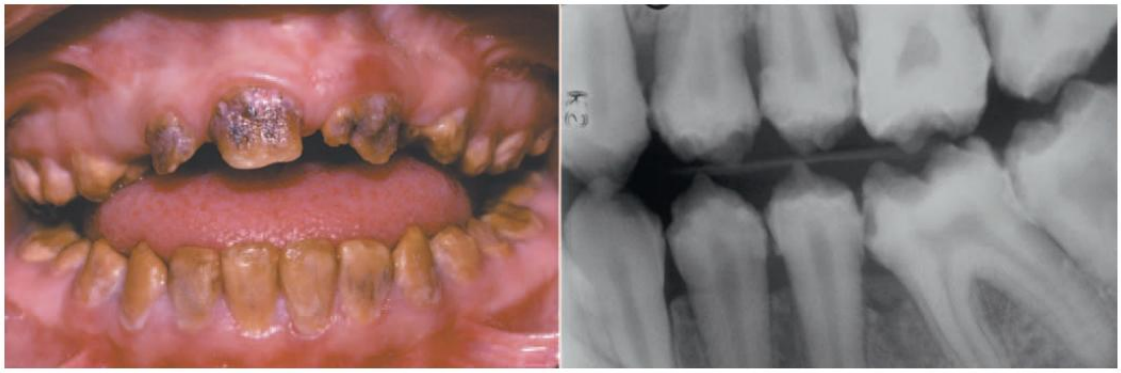

**Figure 4:** Hypocalcified Amelogenesis imperfecta- Diffuse yellowish brown discoloration

Radiographically, loss of enamel causing reduced density of enamel.

**4) Hypomaturation or hypoplastic with Taurodontism**

This type has enamel hypoplasia in combination with hypomaturation. The primary and secondary dentitions are diffusely involved. In case of hypomatiration-hypoplastic type, the primary defect is hypomaturation in which enamel appears mottled yellow-white to yellow-brown. Pits are seen on buccal aspects of the teeth. Radiographically, enamel appears same as dentin in density with large pulp chambers

may be seen in case of single-rooted teeth in addition to varying degrees of taurodontism[11,15].

In case of hypoplastic-hypomaturation type, the primary defect is one of enamel hypoplasia in which thickness of enamel is less but enamel is also hypomature. Except for the decrease in thickness of enamel, this type radiographically resembles hypomaturation-hypoplastic type.[5,11,15]

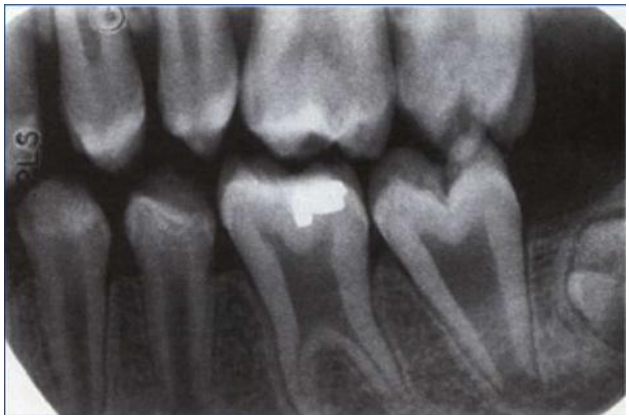

**Figure 5:** Taurodontism seen with permanent mandibular molars with generalized reduced enamel density

### 3.2 Radiographic features

Radiographic findings play adjunctive role in diagnosis of AI.

In case of hypoplastic amelogenesis imperfecta, which has a square crown, relatively thin radiopaque layer of enamel, low or absent cusps with many open contacts between the teeth. This peculiar feature is said to have "picket fence" appearance. The density of the enamel is normal.[5,8,10,15]

In case of hypomaturation type, there is normal thickness of the enamel, but the density is similar to that of adjacent dentin.

In case of hypocalcified type, the thickness of enamel is normal, but the density is even less (more radiolucent than the dentin). Advanced abrasion may cause obliteration of the pulp chambers.[10,15]

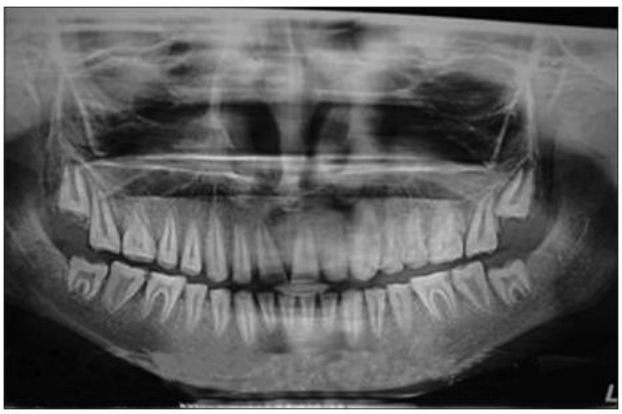

**Figure 6:** Square shaped teeth are seen due to severe attrition. Loss of contact between the adjacent teeth causes "picket-fence" appearance[9,11]

**Syndromes associated with AI**[10,11,16]**-**

AI is considered as a syndrome itself as it often associated with many other abnormalities. There are some known syndromes which consists AI (dental finding) as one of the features.

**1) Tricho-dento-osseous syndrome-**

Clinical findings-Amelogenesis imperfecta with taurodontism, kinky (steel wool) hair, osteosclerosis and brittle nails. Other findings include shortened mandible and an obtuse angle.

**2) Kohlschütter-Tönz syndrome-**

Clinical features-Amelogenesis imperfecta, epilepsy,mental retardation. It is also called as Amelo-cerebro-hypohidrotic syndrome. It is a rare inherited syndrome characterized by epilepsy, psychomotor delay or regression, intellectual disability & amelogenesis imperfecta (abnormal formation of tooth enamel). It is known as a type A ectodermal dysplasia.

**3) Enamel-renal syndrome-**

Clinical features- AI with nephrocalcinosis . Pulp stones may also seen.

**4) Amelogenesis Imperfecta with cone and rod dystrophy-**[17]

Clinical features- Photophobia, pendular nystagmus, reduced central vision starting in the first two years of life night vision difficulties by the end of first decade. First permanent molars seen with taurodontism.

## 4. Conclusion

Amelogenesis imperfecta is a group of inherited disorders which are clinically heterogeneous and exhibit tooth enamel defects in the absence of systemic manifestations. Factors to consider during diagnosis of AI include family history, pedigree plotting (a diagram of a family health history tree), clinical observations, radiographic assessment and meticulous recording. As primary and secondary both dentitions may get affected, early diagnosis helps in proper management of the patient.

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