# Amelogenesis Imperfecta: A report of two cases

Case Report

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**ABSTRACT**

Amelogenesis Imperfecta (AI) is defined as a group of hereditary developmental defects of the dental enamel affecting both primary and permanent dentition. AI is known by various names such as hereditary enamel dysplasia, hereditary brown enamel, hereditary brown opalescent teeth. This defect involves entirely ectodermal components. AI may be inherited in an X linked manner or by autosomal dominant , autosomal recessive, or sporadic inheritance patterns. Witkop and Sauk listed the varieties of AI, based on whether the abnormality lay in reduced amount of enamel (hypoplasia), deficient calcification (hypocalcification), or imperfect maturation of the enamel (hypomaturation). They have also recognized the combined defects of enamel. Treatment planning involves an interdisciplinary approach of periodontal, prosthodontic and restorative treatment.

**KEYWORDS**: enamel, hereditary, ectodermal defect.

## INTRODUCTION:

Tooth development is a highly regulated complex biological process, which is dependent on multiple factors for its ultimate success.[1]

If any of these developmental processes are disturbed or over-exaggerated, changes can occur within the developed tooth's structure & clinical changes or alternations will be evident[1]

The etiology of dental developmental anomalies (or defects) is complex and can involve multiple causative agents such as local factors (e.g., trauma), genetic influences, or environmental insults (e.g., alcohol or drug exposures during the fetal period, febrile illnesses).[1]

An abnormality where the pathology starts in the embryonic stage of human life, before the formation of dentition is known as developmental disturbance.

Amelogenesis imperfecta is a group of hereditary diseases affecting the tooth enamel in either quality or quantity, is associated with crown malformation and abnormal enamel density.

It affects both the clinical and structural appearance of some or all of the dentition. Amelogenesis Imperfects is also known as Hereditary enamel dysplasia, Hereditary brown enamel, and Hereditary brown opalescent teeth .

AI exists in isolation or associated with other abnormalities in syndromes. It may show autosomal dominant, autosomal recessive, sex-linked and sporadic inheritance patterns.

Diagnosis is based on the family history, pedigree plotting and meticulous clinical observation. Genetic diagnosis is presently only a research tool.

Rehabilitation of the entire dentition with amelogenesis imperfecta (AI) tends to pose a great challenge to the clinician. Most of the cases of amelogenesis imperfecta are reported to be associated with skeletal and dental deformities which results in severe sensitivity of the dental tissue.

## CASE REPORT:

A 11 year old female patient came to the department of Oral Medicine and Radiology, Ahmedabad Dental College and Hospital with a chief complain of discoloration of all the teeth region since 1 year. Patient was relatively asymptomatic before 1 year then she noticed discoloration of teeth. Her medical history revealed that she had tuberculosis before 4 years

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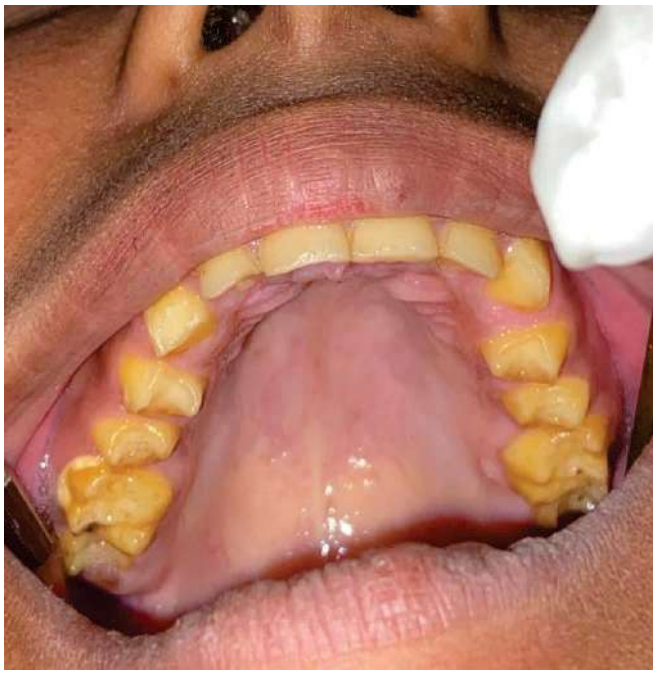
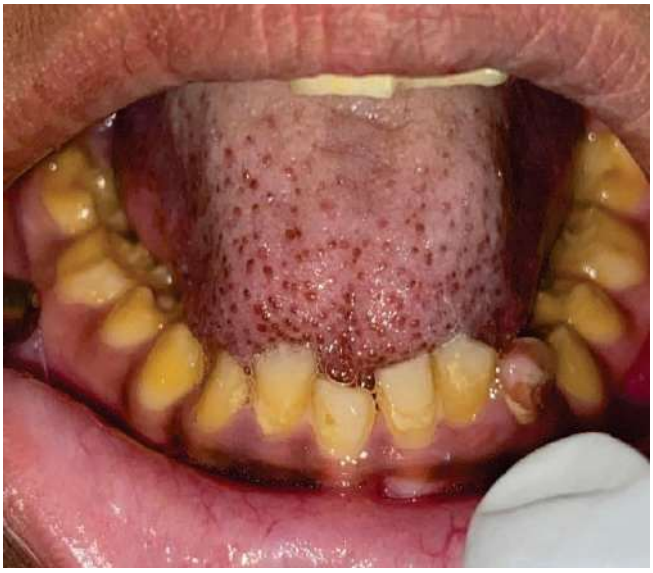
and had Completed the entire treatment for the same.

On extraoral examination, the face was bilaterally symmetrical. The skin was normal. The lymph nodes were not palpable. There was no anesthesia or paresthesia noted over the face. The facial expressions were normal. There was no pain or swelling on face, no pus discharge.

On intraoral examination, all teeth were yellowish brown in color. There was generalized wearing of occlusal and incisal surfaces. Enamel was reduced in thickness which got chipped off on probing exposing dentin in some areas. Patient gave history of similar appearance of deciduous teeth. The surfaces of the teeth were rough. The emergence pattern and timing of teeth seemed to be within normal range.

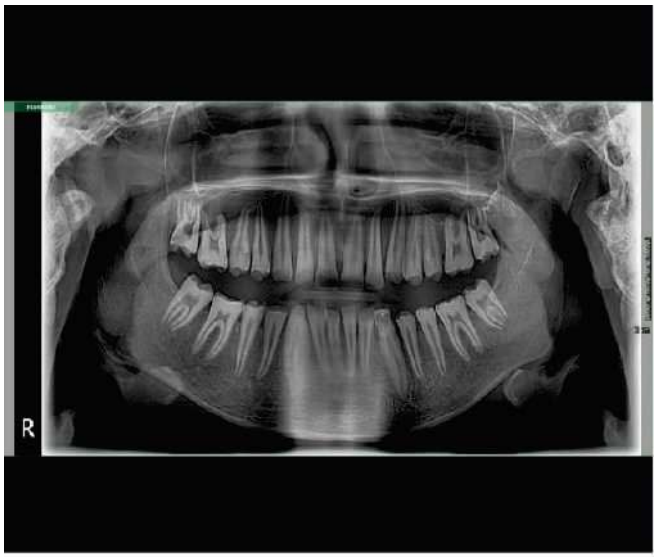
Gingiva was pink, firm and resilient. Clinical photos showing Amelogenesis Imperfecta are as below:





On the basis of history, clinical findings, a provisional diagnosis of Amelogenesis Imperfecta was made. The differential diagnosis included were Environmental enamel hypoplasia, Dentinogenesis Imperfecta, Tetracycline stains. Patient was advised for Orthopantomograph.

Orthopantomograph shows all permanent teeth and retained 73. Dentin and pulp appears normal with no signs of pulp obliteration. Enamel is thin in all teeth over occlusal, incisal and interdental aspects but it is more radiodense than dentin.

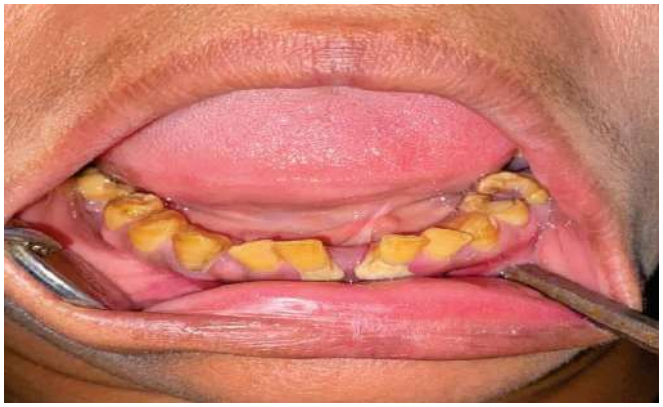


**Case 2**

A 36 year old female patient came to the department of Oral Medicine and Radiology, Ahmedabad Dental College and Hospital with a chief complain of discoloration of all the teeth region since 15 days. Patient was relatively asymptomatic before 15 days then she noticed discoloration of teeth.

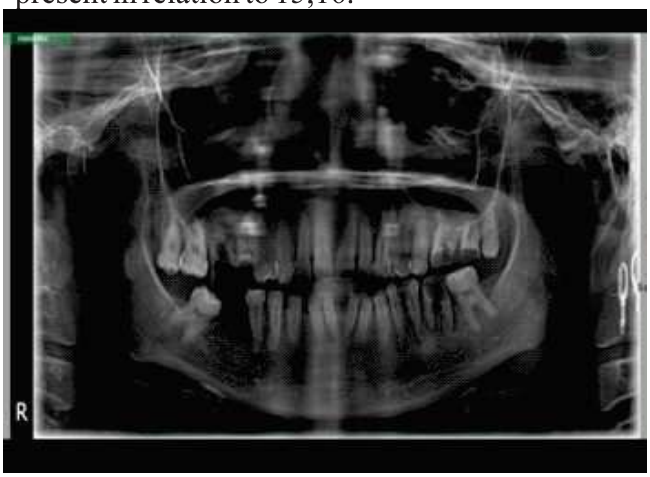
On extraoral examination, the face was bilaterally symmetrical. The skin was normal. The lymph nodes were not palpable. There was no anesthesia or paresthesia noted over the face. The facial expressions were normal. There was no pain or swelling on face, no pus discharge.

On intraoral examination, there was root piece present in relation to 15, 16. There was Ellis Class 1 fracture in relation to 11. There were grossly carious teeth in relation to 26, 27, 36. There was proximal caries in relation to 34, 35. All teeth were yellowish brown in color. There was generalized wearing of occlusal and incisal surfaces. Enamel was reduced in thickness which got chipped off on probing exposing dentin in some areas. Patient gives history of similar appearance of deciduous teeth. The surfaces of the teeth were rough. The emergence pattern and timing of teeth seemed to be within normal range. Gingiva was pink, firm and resilient.



On the basis of history, clinical findings, a provisional diagnosis of Amelogenesis Imperfecta was made. The differential diagnosis included were Environmental enamel hypoplasia, Dentinogenesis Imperfecta, Tetracycline stains. Patient was advised for Orthopantomograph.

Orthopantomograph shows all permanent teeth except 46, 38,48. Dentin and pulp appears normal with no signs of pulp obliteration. Enamel is thin in all teeth over occlusal, incisal and interdental aspects but it is more radiodense than dentin. There are grossly carious teeth in relation to 26, 27, 34, 35 involving enamel, dentin and pulp with associated periapical radiolucencies approx of size 3mm X 2mm. Also, there is radiolucency in crown of 36 involving enamel, dentin and pulp with periapical radiolucency approx. of size 4mm X 4mm with corticated borders. There is root piece present in relation to 15,16.



## DISCUSSION:

Amelogenesis Imperfecta is a developmental, often inherited disorder, affecting dental enamel. It is usually occurs in the absence of systemic features and comprises of diverse phenotypic entities [2]

Disorders like AI often present in childhood and patients' complaints occur in the early stages of their lives. Here, our first case presented at the age of 11years whereas second case presented at the age of 36 years. [6]

It may show autosomal dominant, autosomal recessive, sex-linked and sporadic inheritance patterns. The autosomal dominant and recessive forms of the disorder affect males and females in equal numbers. The X-linked dominant type of the disorder affects twice as many males as females. The X-linked recessive type affects only males. The mutated genes can be passed on from parents to their children, or the mutation can develop in people with no family history of the disease. In both of our cases, there was no relevant family history [7]

Various studies showed that oral complaints associated with AI are unaesthetic appearance, extensive loss of tooth structure, dental sensitivity, and loss of vertical dimension. In both of these cases, patients presented with a complaint of discoloration of teeth [2]

According to various studies of AI, many non-enamel anomalies have been found in association with AI, e.g., Delayed tooth eruption, congenitally missing teeth, anterior open bite, taurodontism, pulpal calcifications, dentin dysplasias, root and crown resorption, hypercementosis, and root malformations. Malocclusion and gingivitis also have been found in association with AI. In both of these cases, no other anomaly had been associated.[2]

## CONCLUSION

There are various proposed classifications of AI based on the phenotype and pedigree combined with scanning electron microscopic examination, biochemical methods, and molecular genetics.

The dental practitioners should 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. Also, the social implications for these patients should be considered.

Thus, this presentation is an attempt to improve the clinician's knowledge about the clinical diagnosis as well as intervention required for such a condition.

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