ISSN: 0975-833X

*Available online at http://www.journalcra.com*

*International Journal of Current Research*
*Vol. 11, Issue, 04, pp.2904-2907, April, 2019*

**DOI: https://doi.org/10.24941/ijcr.35043.04.2019**

**INTERNATIONAL JOURNAL OF CURRENT RESEARCH**

## RESEARCH ARTICLE

# AMELOGENESIS IMPERFECTA HYPOMATURATIVE TYPE – A RARE CASE REPORT

***[1]Anbu Meena, S., [1]Parimala, D., [2]Ramya, M., [2]Annasamy Rameshkumar, [3]Krithika, C.L. and [3]Kannan A Ashokan**

[1]Department of Oral Medicine and Radiology, SRM Dental College and Hospital, Ramapuram, Chennai, Tamil Nadu, India

[2]Department of Oral Pathology and Microbiology, SRM Dental College and Hospital, Ramapuram, Chennai, Tamil Nadu, India

[3]Department of Oral Medicine and Radiology, SRM Dental College and Hospital, Chennai, Tamil Nadu, India

**ARTICLE INFO**

*Article History:*
Received 03rd January, 2019
Received in revised form
09th February, 2019
Accepted 21st March, 2019
Published online 29th April, 2019

*Key Words:*
Amelogenesis imperfecta,
Hypomaturative Type, Enamel Defect.

**ABSTRACT**

Amelogenesis imperfect (AI) is an inheritable disease manifest as enamel defect. It affects both primary and permanent dentition. It seldom affects the dentin and pulp.Amelogenesis imperfecta shows wide range of variation starting from mild discolouration to complete abrasion as soon as the teeth erupt into the oral cavity. Enamel may show quantitative as well as qualitative defect in mineral and protein content. In this article we present a special case of amelogenesis imperfecta which shows a rare hypomaturative type with sporadic inheritance and we discuss about its mode of inheritance, types, clinical presentation, histopathological features and management.



***Citation:*** **Anbu Meena, S., Parimala, D., Ramya, M., Annasamy Rameshkumar, Krithika, C.L. and Kannan A Ashokan. 2019.** "Amelogenesis imperfecta hypomaturative type – a rare case report", *International Journal of Current Research*, 11, (04), 2904-2907.

## INTRODUCTION

Amelogenesis imperfect (AI) includes a complex group of conditions that shows developmental defect in the enamel structure without any systemic disease (Nevillee *et al*., 2009). Dental Enamel is the most hardest and mineralized tissue in the body, made up of >95% by hydroxyapatite crystals (Arora, 2018). Dental enamel is unique when compared to other mineralized structures like Dentin, cartilage and bone in the way it is non collagenous, developed from epithelium. Enamel will not undergo resorption and remodeling (Chanmougananda, 2012). Ameloblast forms Enamel and the process is termed as amelogenesis. Each and every step in amelogenesis is very important for enamel formation (Smith, 2009). Enamel composition depends purely on the molecular and cellular events in amelogenesis. The prevalence of AI ranges from 1 in 718 to 1 in 14,000 based on the population included in the study (Chaudhary, 2009). AI is an inherited enamel defect which shows clinical and genetic variation. It affects primary or permanent dentition as a whole involving all the teeth or few teeth in the dentition (Sarawathi, 2011). The dentin and root are spared and will not be involved even in the affected teeth (Chaudhary, 2009).

***Corresponding author: Anbu Meena, S.,**
Department of Oral Medicine and Radiology, SRM Dental College and Hospital, Ramapuram, Chennai, Tamil Nadu, India

Disorders of the enamel epithelium may cause alteration in the eruption mechanism resulting in the anterior open bite (Rajendran, 2009). Patients may also present with other abnormalities like congenitally missing teeth, pulpal calcifications, root and crown resorption, hypercementosis, root malformations, and taurodontism. Because of attrition and/or incomplete eruption they will have short clinical crown which will affect their chewing efficiency and also cause tooth sensitivity (Nigam, 2015).

### Case report

An 18 year old male patient came to the department of oral medicine and radiology with the chief complaint of yellowish brown discolouration of his teeth. History of presenting illness revealed that his primary teeth was also discoloured. Patient Medical History was non-contributory and there was no positive family history (Figure 1). Intra oral hard tissue examination revealed generalized yellowish-brown discoloration of the teeth with normal enamel thickness. The shape and size of the teeth appeared to be normal (Figure 2). On palpation the surface was rough and there was no softness or chipping of the enamel evident. Sensitivity, Caries, Anterior Open bite, Attrition and other complication of AI were absent in this patient.

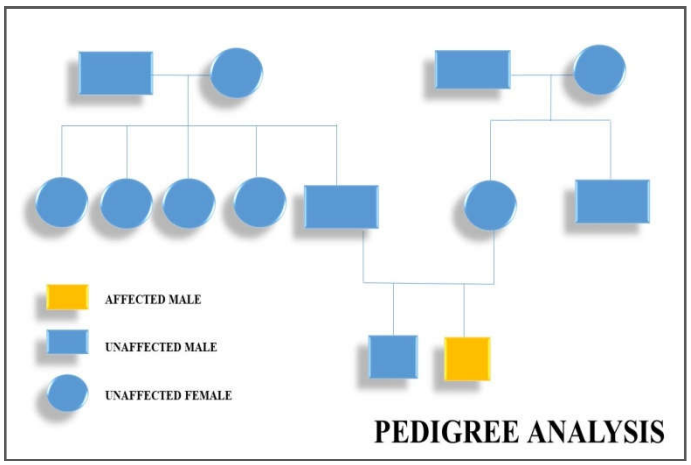


**Figure 1. Illustrate the Pedigree Chart of the patient depicting all other members of the family were normal suggesting Sporodic type of inheritance**

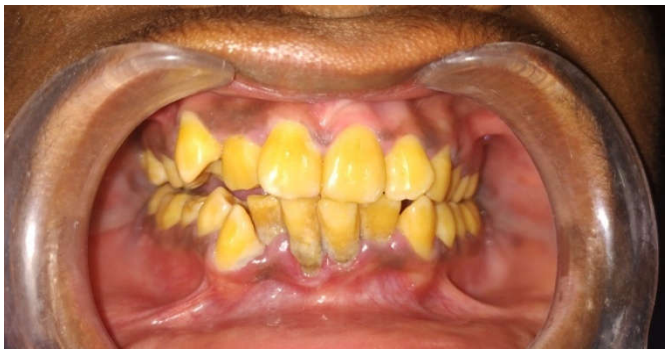

**Figure 2. Shows Yellowish brown discolouration of enamel with normal thickness. The shape and size of the teeth shows no abnormalities**

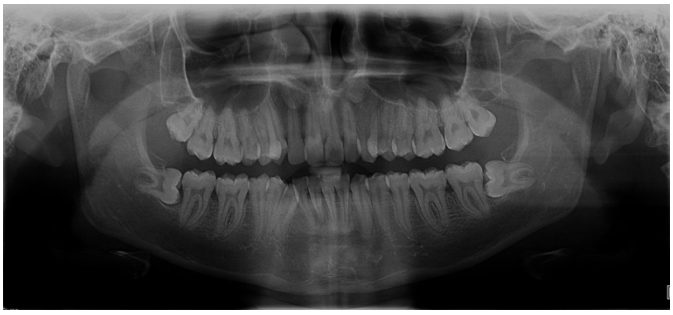

**Figure 3. OPG shows normal enamel thickness with normal dentin ,pulp chamber and root canal spaces with no signs of obliteration and horizontally impacted lower third molars, 38 and 48**

On examining the soft tissue of the periodontium revealed the presence of chronic generalized gingivitis. On Radiographic investigations, the OPG showed normal enamel thickness with radiodensity same as that of dentin with a normal pulp chamber and root canal spaces with no signs of obliteration and also revealed the presence of horizontally impacted lower third molars, 38 and 48 (Figure 3). A provisional diagnosis of hypomaturative type of AI was given with a differential diagnosis of enamel hypoplasia, dentinogenesis imperfecta, dentin dysplasia, regional odontodysplasia. Following which horizontally impacted third molar 48 was extracted and sent for histopathological investigation in which ground section of the teeth revealed irregular orientation of the enamel rods in some areas (Figure 4). Incremental lines are not prominent in the occlusal aspect. Increased number of enamel lamellae and tufts were seen (Figure 5). Enamel spindles are seen in moderate number.

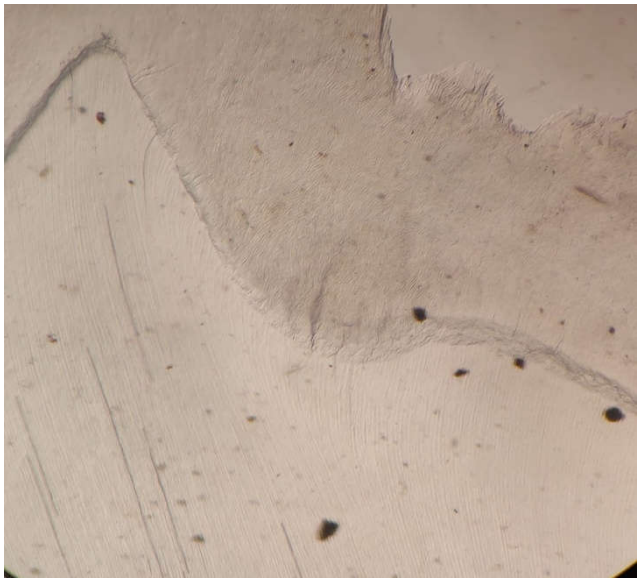

**Figure 4. Reveal ground section of the teeth with irregular orientation of the enamel rods**

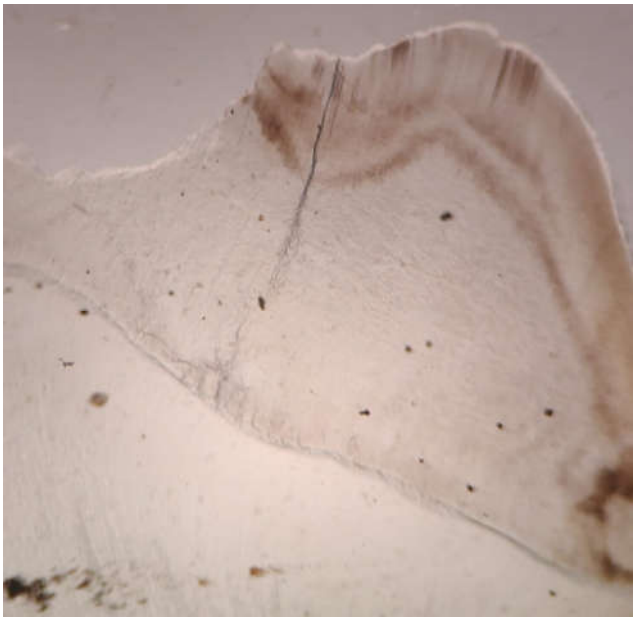

**Figure 5. Depicts increased number of enamel lamellae and tufts**

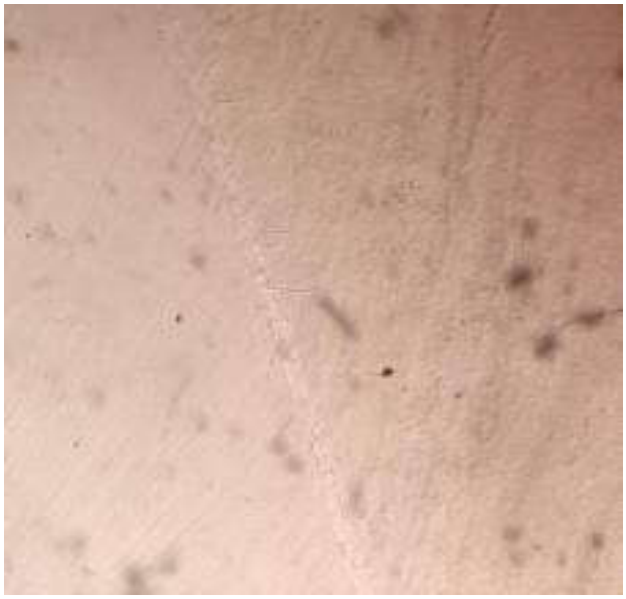

**Figure 6. Represents dentino-enamel junction is devoid of scalloping**

**Table 1.**

| | | |
|---|---|---|
| Type I hypoplastic | IA | Pitted autosomal dominant |
| | IB | Local autosomal dominant |
| | IC | Local autosomal recessive |
| | ID | Smooth autosomal dominant |
| | IE | Smooth X-linked dominant |
| | IF | Rough autosomal dominant |
| | IG | Enamel agenesis, autosomal recessive |
| Type II hypomaturation | IIA | Pigmented autosomal recessive |
| | IIB | Hypomaturation |
| | IIC | Snow capped teeth, X-linked |
| | IID | Autosomal dominant? |
| Type III hypocalcification | 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 dominant |

**Table 2.**

| Hypoplastic form | Hypomaturation form | Hypocalcified Form |
|---|---|---|
| Reduction in the thickness of enamel matrix with normal mineralization | Defect in the mineralization process with normal matrix formation | Defect in the quality of the mineralization process with normal quantity of matrix formation |
| CLINICAL APPERANCE | | |
| Reduced thickness of enamel | Normal thickness of enamel | Normal thickness of enamel with loss of translucency |
| The colour appears normal with translucency to a yellow to dark brown colour depending on the thickness of enamel and dentin | Colour may be affected by staining from the oral environment. Mottled appearance to yellow-brown or redbrown discoloration | Colour may be affected by staining from the oral environment. Teeth appear more dark |
| Reduction in tooth size Rough, irregular or pitted enamel | Enamel is hypomineralized and prone to attrition | Enamel is hypomineralized and exhibits a soft cheesy consistency. Easily broken down |
| RADIOGRAPHIC FEATURE | | |
| Enamel and dentin appears normal | Enamel has similar radiodensity as dentin | Enamel is less radiopaque than dentin |

Most part of dentino-enamel junction is devoid of scalloping (Figure 6). Reduced amount of gnarled enamel is present. A treatment plan consists of oral prophylaxis and prosthetic treatment was framed. Esthetics was the patient major concern so patient was advised for prosthetic management of the discoloured tooth.

## DISCUSSION

AI denotes a group of disorders with genetic origin, and it will affect the enamel structure and clinical appearance of some or all the teeth in a same manner, and in addition to it there may be biochemical or morphologic changes elsewhere in the body (Witkop, 1988). Witkop and Sauk classification of AI is depicted in Table 1 (Aldred, 2003). The hypoplastic type is most common in females and it contributes to about 60%–73% of cases. The hypomaturation type is common in males and it was see in 20%–40% of cases. Hypocalcified AI is rare and it affects7% of individuals affected by AI. (Arora, 2018). AI shows autosomal dominant, autosomal recessive, sex linked inheritance patterns, as well as sporadic in few rare cases. X-linked form of AI shows the typical pattern of X-linked inheritance. Heterozygous females have 50% chances of transmitting the affected gene to children of both the sex. It affects males and females in distinctly different ways. Males display the condition fully (Fischman, 1967). Autosomal dominant form of AI affects one or more people in every generation of a family. The expression of AI may look similar or it may look entirely different among the members of the same family. Autosomal recessive form of AI is more common in population where intermarriage within the family is more common. Sporadic cases of AI may arise because of one of many causes. It may be due to new mutation or different expression with or without incomplete transmission of dominant gene. It is mandatory to examine other family members in such situation for proper diagnosis and eliminate any other dental defect caused because of non-genetic causes (Crawford, 2007). In the process of amelogenesis, soft extracellular matrix made of protein is converted into hard mineralized tissue called enamel and this process is controlled by molecular and cellular events which is mediated by specific genes. Any genetic mutation will reflect eventually as a defect in the enamel (Lakshman, 2016). It include enamelin, amelogenin, ameloblastin, and tuftelin, amelotin, dentin sialophosphoprotein, kallikrein and matrix metalloproteinase 20 (Chanmougananda, 2012). Different types of AI with its clinical and radiographic appearance is depicted in Table 2 (Roma, 2016), Hypoplastic AI is due to defect in the secretory stage of enamel and it results in thin but mineralized enamel in mild cases to entire loss of enamel in severe cases. Hypomineralized AI is due to defect in the maturation stage and it results in full thick enamelbutis weak and fails prematurely. The hypomineralized type is classified into hypomaturation and hypocalcified AI. In which hypomaturation is due to defect in protein removal from the enamel matrix and result in brittle enamel and hypocalcified is due to defect in calcium transport and result in soft enamel (Smith, 2017).

**Histopathological Findings:** Histologically, in hypoplastic type, there is deficiency in enamel matrix formation where as in hypocalcification type, there is deficiency in structure of enamel matrix and also defect in mineralization is noted. Altered enamel rod and rod sheath structures were evident in hypomaturative type of AI histologically in many studies (Mehta, 2013).

**Differential diagnosis:** Many forms of enamel dysmineralization were also noted and they exhibit a characteristic pattern depend upon the stage of amelogenesis when the insult was noted. It can be differentiated from AI by the fact it will cause defect in all the teeth in same way and it will have familial inheritance. Another important thing to be considered is Fluorosis but it will not uniformly affect all the

teeth usually premolars and second permanent molars remains unaffected. One should also note the fluoride intake history for proper diagnosis (American Academy on Pediatric Dentistry Council on Clinical Affairs, 2008).

**Treatment:** Heritable developmental dental disorders have remarkable negative effects in the affected persons and their family. American Academy of Pediatric Dentistry had given guidelines for the treatment of heritable dental development anomalies including AI. (American Academy on Pediatric Dentistry Council on Clinical Affairs, 2008). Factors such as age, socio-economic status, type of AI and its severity have to be considered in treatment planning. Because AI is a rare developmental enamel disorder, there is no proper evidence-based clinical recommendation (Toupenay, 2018). The major concern of patients with AI were discolouration and poor appearances of their teeth., severe tooth sensitivity, dental caries and dental orthodontic problems (Koruyucu, 2014). The dentin and pulp are mostly spared and remain unaffected so restoration of the enamel defect is greatly enforced and proper oral hygiene maintenance is mandatory for good prognosis (DeSort, 1983). Treatment primarily aim to preserve the remaining tooth structure wherever possible and prevent further loss of tooth. It is essential to improve mastication efficiency and appearance because this will have greater impact on patient's psychology and confidence (Nigam, 2015). Management directed at three aspects of treatment includes prevention, restoration, and esthetics. Preventive aspects include dietary advice, regular use of fluoride mouthwashes, topical fluoride applications, and oral hygiene instructions. Restoration are based upon the type of dentition. GIC restorations should be advised in the primary dentition, direct and indirect composite resin veneers in the mixed dentition while porcelain veneers, full crowns, extractions of excessive defected teeth followed by fixed, or removable prosthesis should be advocated in permanent dentition based on number of teeth affected, patient age, and economical status. Resin composite veneer are generally preferred in AI patients to preserve tooth structure because it needs only a minimal preparation. Because of recent advancement in aesthetic dentistry, it is now able to achieve proper function and aesthetics in AI patients (Leevailoj, 2017).

**Conclusion**

Being a developmental disorder that is of more esthetic concern, the genetic basis of the disorder must be explained to the patients and education should be given. Considering the complexity of the condition and complications that could be encountered during treatment it makes it mandatory for the clinician to use various strategies that will improves the quality of life of the patient.

## REFERENCE


Aldred, M.J., Savarirayan, R., Crawford, P.J. 2003. Amelogenesis imperfecta: a classification and catalogue for the 21st century. *Oral diseases*. Jan;9(1):19-23.

American Academy on Pediatric Dentistry Council on Clinical Affairs. Guideline on oral heath care/dental management of heritable dental development anomalies. Pediatric dentistry. 2008;30(7 Suppl):196.

Arora, K., Bhat, D.V., Mitra, M., Saha, S. 2018. Early oral rehabilitation of amelogenesis imperfecta: A case series. *International Journal of Pedodontic Rehabilitation*. Jul 1;3(2):80.

Chanmougananda SC, Ashokan KA, Ashokan SC, Bojan AB, Ganesh RM. Literature review of amelogenesis imperfecta with case report. *Journal of Indian Academy of Oral Medicine and Radiology*. 2012;24(1):83.

Chaudhary M, Dixit S, Singh A, Kunte S. Amelogenesis imperfecta: Report of a case and review of literature. Journal of oral and maxillofacial pathology: JOMFP. 2009 Jul;13(2):70.

Crawford PJ, Aldred M, Bloch-Zupan A. Amelogenesis imperfecta. *Orphanet journal of rare diseases.* 2007 Dec;2(1):17.

DeSort KD. Amelogenesis imperfecta: The genetics, classification, and treatment. *The Journal of prosthetic dentistry.* 1983 Jun 1;49(6):786-92.

Fischman SL, Fischman BC. Hypoplastic amelogenesis imperfecta: report of case. *The Journal of the American Dental Association.* 1967 Oct 1;75(4):929-31.

Koruyucu M, Bayram M, Tuna EB, Gencay K, Seymen F. Clinical findings and long-term managements of patients with amelogenesis imperfecta. *European journal of dentistry.* 2014 Oct;8(4):546.

Lakshman B, Revathy V, Suganthi G, Prem Kumar S. A Unique Case of Amelogenesis Imperfecta. *Pediatr Dent Care.* 2016; 1(109):2.

Leevailoj C, Lawanrattanakul S, Mahatumarat K. Amelogenesis Imperfecta: Case Study. Operative dentistry. 2017 Sep; 42(5):457-69.

Mehta DN, Shah J, Thakkar B. Amelogenesis imperfecta: Four case reports. *Journal of Natural Science, Biology, and medicine.* 2013 Jul;4(2):462.

Neville BW, Damm DD, Allen CM, Bouquot JE. Text book of oral and maxillofacial pathology. *Central odontogenic Fibroma.* 2009:726-7.

Nigam P, Singh VP, Prasad KD, Tak J. Amelogenesis Imperfecta: A Case Report and Review of Literature. *International Journal of Scientific Study.* 2015 Jan 1;2(10):146-9.

Nigam P, Singh VP, Prasad KD, Tak J. Amelogenesis Imperfecta: A Case Report and Review of Literature. *International Journal of Scientific Study.* 2015 Jan 1;2(10):146-9.

Rajendran R. Shafer's textbook of oral pathology. Elsevier India; 2009.

Roma M, Hegde S. Amelogenesis Imperfecta: A Review of the Literature. *Journal of Pharmaceutical Sciences and Research.* 2016 Sep 1;8(9):1042.

Sarawathi VN, Ghousia S, Shashibhushan KK, Poornima S. Amelogenesis imperfect-A case report. *J Oral Health Res.* 2011;2(4):106-10.

Shivhare P, Shankarnarayan L, Gupta A, Sushma P. Amelogenesis Imperfecta: A Review. *Journal of Advanced Oral Research.* 2016 Jan;7(1):1-6.

Smith CE, Poulter JA, Antanaviciute A, Kirkham J, Brookes SJ, Inglehearn CF, Mighell AJ. Amelogenesis imperfecta; genes, proteins, and pathways. *Frontiers in physiology*. 2017 Jun 26;8:435.

Toupenay S, Fournier BP, Manière MC, Ifi-Naulin C, Berdal A, de La Dure–Molla M. Amelogenesis imperfecta: therapeutic strategy from primary to permanent dentition across case reports. BMC oral health. 2018 Dec;18(1):108.

Witkop CJ. Amelogenesis imperfecta, dentinogenesis imperfecta and dentin dysplasia revisited: problems in classification. *Journal of Oral Pathology & Medicine.* 1988 Nov 1;17(9-10):547-53.


*******