# *A Genetic Echo in Enamel: Familial Hypoplastic Amelogenesis Imperfecta in a Young Female*

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## *ABSTRACT*

Amelogenesis imperfecta (AI) is a group of inherited developmental disorders affecting enamel formation, resulting in structural and esthetic abnormalities of teeth. This case report describes a 19-year-old female with a positive family history suggesting a hereditary pattern.

The patient was treated by a multidisciplinary approach. This report highlights importance of early diagnosis and comprehensive treatment planning that are essential in managing AI to restore function, improve esthetics, and enhance the patient's psychological well-being and quality of life.

**Keywords**: Amelogenesis imperfecta, Enamel hypoplasia, Hereditary dental disorder, Hypoplastic type, Prosthetic rehabilitation.

## *INTRODUCTION*

Amelogenesis imperfecta (AI) represents a heterogeneous group of inherited developmental disorders that primarily affect the formation of enamel in both primary and permanent dentitions [1]. It occurs in the absence of systemic disease and is characterized by quantitative and/or qualitative defects in enamel structure [2].

The process of amelogenesis involves two principal stages: formation of the organic matrix followed by its mineralization, both of which are regulated by ameloblasts [3]. Disturbances during any stage may result in defective enamel, leading to clinical manifestations such as discoloration, reduced enamel thickness, and structural weaknesses [4].

AI exhibits considerable clinical variability and may be inherited in autosomal dominant, autosomal recessive, or X-linked pattern. s [1,5]. Mutations in genes such as AMELX and ENAM are implicated in its pathogenesis [5]. Based on clinical and histological features, AI is broadly classified into hypoplastic, hypomaturation, hypocalcified, and mixed types [2]. The hypoplastic type is characterized by reduced enamel thickness and surface irregularities such as pitting and grooves [4].

> ***Take Home Message***
> *Amylogenesis imperfecta is a hereditary enamel defect that demands early recognition and comprehensive lifelong dental care to preserve function,* aesthetics, and patient well-being.

Clinically, patients may present with esthetic concerns, dental hypersensitivity, increased susceptibility to caries and occlusal discrepancies [3]. The condition may significantly impact functional efficiency and psychological well-being, necessitating early diagnosis and comprehensive management [6].

The purpose of this case report is to describe the clinical presentation, diagnosis and multidisciplinary management of generalized hypoplastic amelogenesis imperfecta, with emphasis on familial occurrence and rehabilitation strategies

## CASE DETAILS

A 19-year-old female patient reported to the Department of Oral Medicine and Radiology with a chief complaint of pain in the lower left posterior region of the jaw for the past 3–4 days

The patient was reportedly asymptomatic until approximately one year prior, when she first noted multiple decayed teeth and sought dental consultation. Root canal treatment was initiated for one tooth; however, further treatment was discontinued due to personal constraints. Over subsequent months, she experienced recurrent food impaction and intermittent pain on mastication, which she managed with medications. The recent onset of acute pain prompted her to seek further evaluation.

There was no history of consumption of high-fluoride (tube well) water. The patient maintained regular oral hygiene practices, including brushing twice daily. Medical history was non-contributory.

## CLINICAL FINDINGS

Intraoral examination revealed generalized yellowish-brown discoloration affecting all tooth surfaces. The enamel exhibited a rough, pitted surface texture with generalized spacing between teeth. Enamel chipping was evident in relation to 13, 23, 26, 34, and 44. Tenderness on percussion was present in relation to 36. Mildly increased vertical overlap was noted.

The patient's father and paternal aunt also gave history of similar dental complaints, including early onset of generalized yellowish discoloration and multiple decayed teeth suggesting a hereditary etiology (Fig.2)

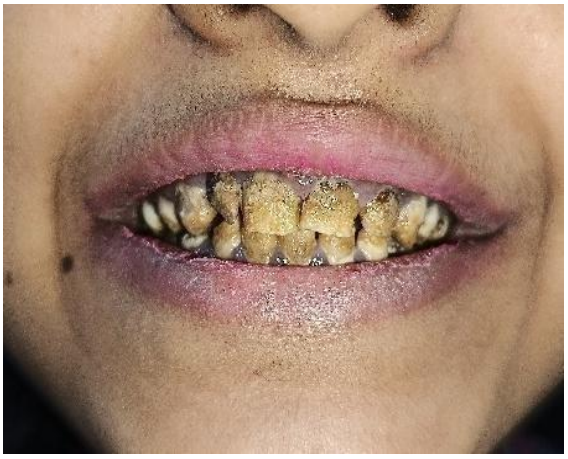

*Fig.1(a): Intraoral Presentation, Labial View*

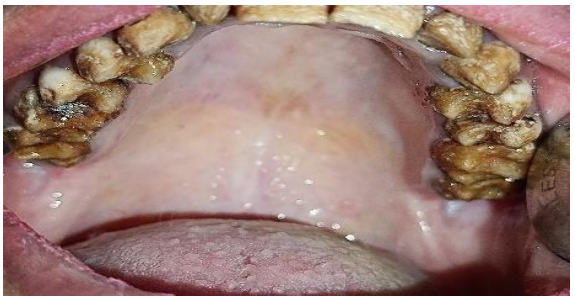

*Fig.1(b): Intraoral Presentation, Maxillary Labial, Buccal & Occlusal View*

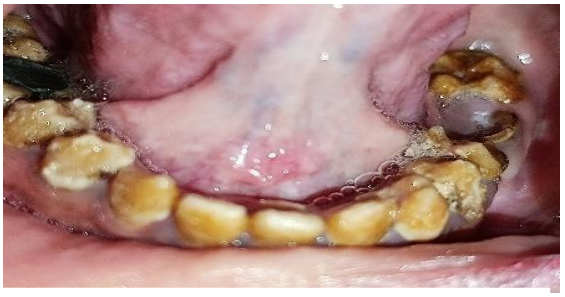

*Fig.1(c): Intraoral Presentation, Mandibular Labial, Buccal & Occlusal View*

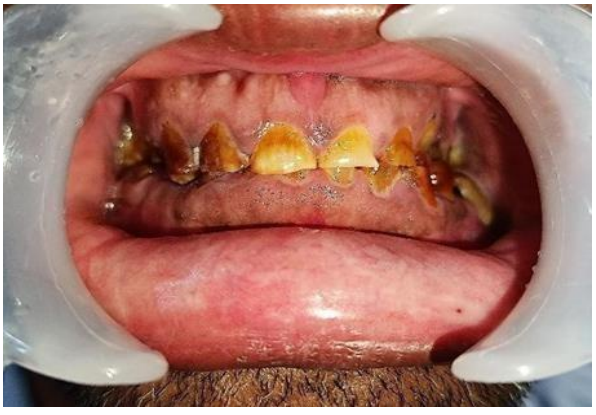

*Fig.2- Intraoral view of Patient's Father's Teeth*

## *RADIOGRAPHIC FINDINGS*

Radiographic examination (Fig.3) revealed a total of 30 teeth. The enamel layer appeared markedly thin and poorly demarcated from dentin. Pulp chambers and root canal morphology were within normal limits. Generalized widening of the periodontal ligament space and interruption of the lamina dura were observed in relation to teeth 35, 36, and 46, with associated periapical radiolucencies.

Additional findings included impacted 43, dilaceration of the mesiobuccal root of 16, deep carious lesions in 16, 35, and 36, and mild proximal caries in multiple teeth.

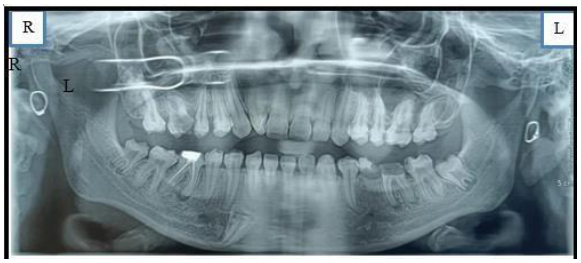


*Fig.3 - Radiographic presentation (panoramic view of jaws)*

## *DIAGNOSIS*

Based on clinical and radiographic findings, a final diagnosis of generalized hypoplastic pitted type amelogenesis imperfecta was established, associated with pulpal and periapical pathologies.

**Treatment plan:**

- Phase I (Emergency Phase): Management of acute pain
- Phase II (Etiotropic Phase): Not required
- Phase III (Surgical Phase): Extraction of non-restorable teeth (35, 36)
- Phase IV (Restorative Phase): Endodontic treatment followed by full-mouth prosthetic rehabilitation with fixed crowns and replacement of missing teeth using fixed partial denture
- Phase V (Maintenance Phase): Oral prophylaxis and periodic follow-up.

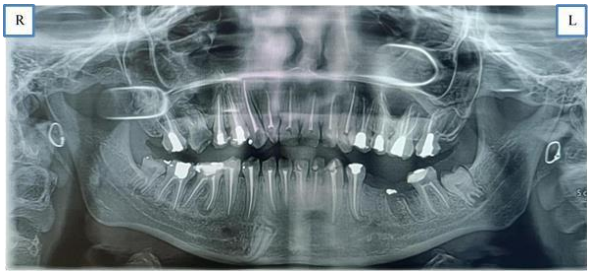


*Fig. 4 - Post surgical and endodontic treatment radiograph.*

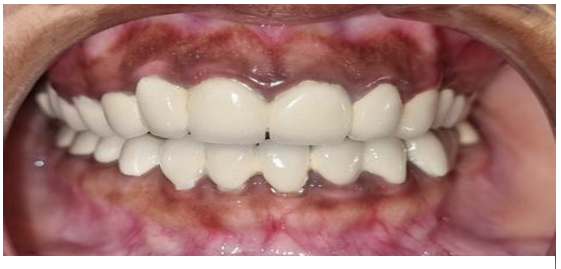

*Fig. 5 Post prosthetic treatment intraoral (labial and buccal) view*

***Post prosthetic treatment intraoral view:***

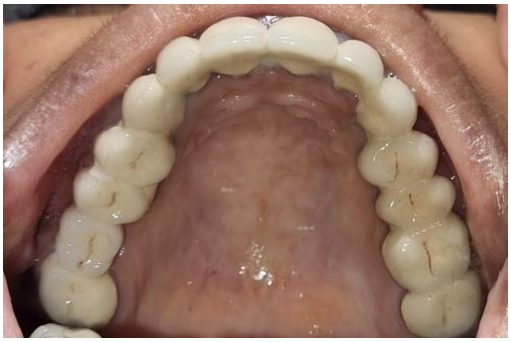

Fig. 6(a) – *Maxillary occlusal and palatal view (through intraoral mirror)*

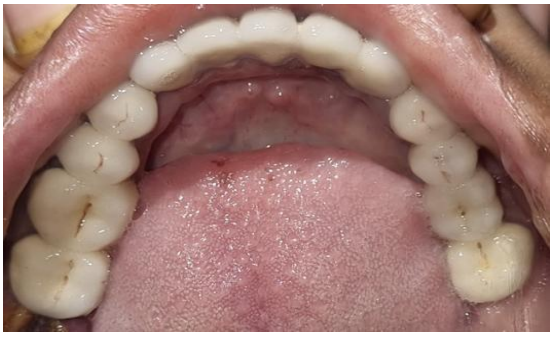

Fig. 6(b) – *Mandibular occlusal and lingual view*

## *DISCUSSION*

Amelogenesis imperfecta (AI) represents a clinically and genetically heterogeneous group of enamel defects with variable phenotypic expression, often presenting diagnostic and therapeutic challenges [7]. The present case demonstrated features consistent with the hypoplastic variant, characterized by reduced enamel thickness and generalized pitting [4].

The observed enamel defects, including discoloration, chipping, and spacing, can be attributed to defective enamel matrix formation, resulting in compromised structural integrity and increased susceptibility to attrition and caries [8].

The presence of a positive family history further supports the hereditary nature of the condition [7,9]. Differential diagnoses such as dental fluorosis and dentinogenesis imperfecta were considered; however, the generalized involvement and familial pattern favored diagnosis of AI [4]. Patients with AI are predisposed to dental hypersensitivity and early pulpal involvement, as seen in this case [8].

Management requires a comprehensive multidisciplinary approach, incorporating preventive, restorative, endodontic, and prosthetic interventions [11]. Long-term success depends on patient compliance, maintenance protocols, and addressing psychosocial concerns [10].

## *CONCLUSION*

This case highlights the clinical challenges associated with amelogenesis imperfecta and underscores the importance of a tailored, multidisciplinary treatment approach. Evidence suggests that effective management involves preventive care during early stages, conservative restorations during adolescence, and definitive prosthetic rehabilitation in adulthood. With appropriate treatment planning, patient compliance, and regular follow-up, favorable long-term functional and esthetic outcomes can be achieved.

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