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Case Report

Amelogenesis Imperfecta: A Case Report and Review of Literature

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Abstract: Amelogenesis Imperfecta (AI) refers to a group of hereditary developmental disorders that impact the structure and appearance of enamel on all or nearly all teeth in a relatively uniform manner. AI poses significant challenges by reducing oral health-related quality of life and causing various physiological issues. Here, we present a case report of the Hypoplastic type of AI, diagnosed based on characteristic clinical and radiographic findings.

Keywords: Amelogenesis Imperfecta, Hypoplastic teeth, Discoloration, Hypomaturative.

INTRODUCTION

Enamel is regarded as the hardest and most mineralized tissue in the human body, with 85% of its volume composed of hydroxyapatite crystals [1]. Amelogenesis Imperfecta is a group of hereditary disorders that affect the enamel, impacting its quality or quantity, and are associated with crown malformations and abnormal enamel density [2].

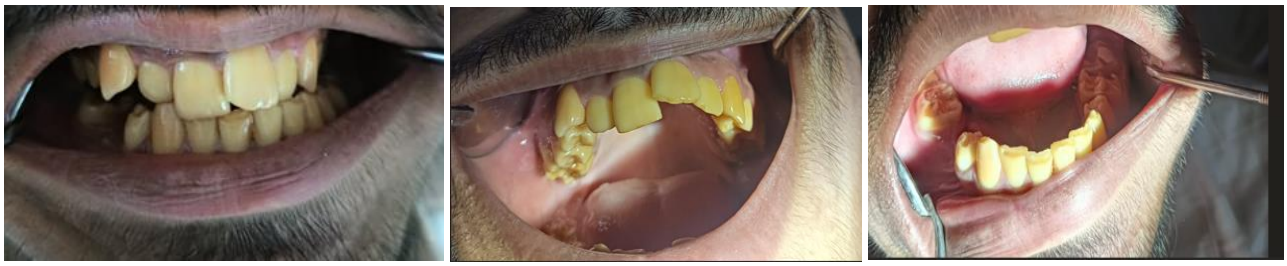
Amelogenesis Imperfecta is a genetic condition that affects the structure, physical characteristics, and clinical appearance of enamel, often leading to a negative impact on a patient's self-esteem. These cases are rare, occurring in about one in a million, and require careful, precise management due to the soft and fragile nature of enamel, which is prone to chipping. This case report highlights clinical approach focused on managing such unique cases in both medical and dental fields with an interdisciplinary strategy, aiming to strengthen the maxillofacial structure.

CASE REPORT

A 19-year-old male patient visited the Department of Oral Medicine and Radiology with a primary complaint of yellowish discoloration and poor appearance of his teeth, which he had observed for the past 9–10 years. The patient, an unmarried and well-educated medical student, mentioned a family history of similar dental problems. His mother, three siblings, maternal aunt, uncle, and three maternal cousins exhibited the same dental characteristics. He had no significant dental or medical history. Upon extra-oral examination, his mouth opening was normal with no temporomandibular joint issues, and he was otherwise healthy with no comorbidities.

Intraoral examination revealed reduced enamel thickness on the teeth, with some teeth having completely chipped enamel, exposing the underlying dentin. The upper and lower teeth showed amber-yellow discoloration and signs of attrition. Tooth surfaces were rough with minimal remaining enamel, though the remaining enamel was hard and showed no further chipping. The teeth had

short clinical crowns. The emergence pattern and timing of the teeth appeared normal. The occlusal plane was uneven due to worn-down posterior teeth, and crowding was observed between certain teeth. The exposed dentin had a brown color. Periodontal examination indicated healthy gingiva with no abnormalities (Fig. 1). Radiographic evaluation, including a full-mouth Orthopantomogram (OPG) and Intraoral Periapical Radiograph (IOPA), confirmed findings consistent with Amelogenesis Imperfecta (Fig. 2).



A **B** **C**

Figure 1: Showing preoperative clinical photograph of patient; A: Frontal view; B: Maxillary Arch view; C: Mandibular Arch view

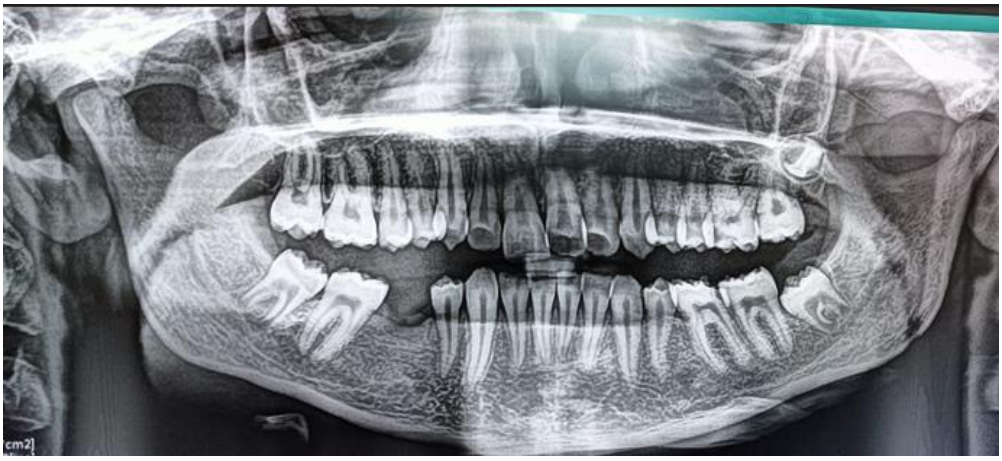


Figure 2: Showing pre-operative orthopantomogram of patient

The Orthopantomogram (OPG) revealed generalized thinning of enamel across all tooth surfaces, with complete enamel absence in some areas, while the pulp chamber and root morphology appeared normal. The OPG also showed impacted teeth 28 and 38, and a thistle tube appearance in teeth 11, 12, and 21. Additionally, high pulp horns were observed in teeth 36 and 37.

Based on the clinical and radiographic findings, the diagnosis was confirmed as the hypoplastic type of Amelogenesis Imperfecta. A comprehensive full-mouth rehabilitation plan was developed, involving collaboration with the endodontic and prosthetic departments to restore functional occlusion, improve esthetics, and prevent further tooth loss. The treatment plan was explained to the patient in detail, and informed consent was obtained. The initial phase of treatment included oral prophylaxis and endodontic therapy for tooth 26. The panoramic radiograph revealed a thin layer of enamel, with the enamel's radiodensity being higher than that of the dentin [Figure 2]. Thus, the final diagnosis of hypoplastic Amelogenesis Imperfecta was confirmed.

DISCUSSION

The primary challenge for a dentist in managing enamel defects lies in accurately diagnosing the condition, which necessitates a thorough investigation of family history and the use of auxiliary tests. This case involves the hypoplastic type of Amelogenesis Imperfecta, which, in this instance, is attributed entirely to a genetic mutation rather than hereditary factors. Amelogenesis Imperfecta may occur in isolation or in conjunction with other syndromes, and it can result from a single gene mutation or various chromosomal abnormalities. Such cases are rare and are not frequently reported.

The hallmark of Amelogenesis Imperfecta (AI) is defective enamel without any associated systemic conditions. AI is a heritable disorder that primarily affects the ectodermal component of the teeth, while the mesodermal component remains

normal. This condition can follow an autosomal dominant, autosomal recessive, or X-linked inheritance pattern [3]. The most widely accepted classification of AI was proposed by Witkop in 1988 and later modified by Nusier in 2004 [4]. AI is categorized into four types: hypoplastic, hypo maturation, hypocalcified, and hypo maturation-hypoplastic.

Clinical signs and symptoms of Amelogenesis Imperfecta (AI) often include dentinal sensitivity, loss of vertical dimension, and compromised esthetics. Treatment planning for AI requires a thorough diagnosis and an interdisciplinary approach, involving periodontal, prosthodontic, and restorative treatments. The primary goal in treating a patient with AI is to protect the entire stomatognathic system and restore the affected hard tissues. To date, the most common treatment options revolve around either full coverage restorations or adhesive restorations [5].

Amelogenesis Imperfecta (AI) encompasses a group of hereditary disorders resulting from genetic mutations in genes such as amelogenin, enamelin, and kallikrein-4 [6]. In some cases, particularly the X-linked form, the defect is due to mutations in the amelogenin gene (AMELX). In dominant cases of AI, mutations in the enamelin gene (ENAM) are implicated in the pathological process [7].

The hypocalcified type of Amelogenesis Imperfecta (AI) is characterized by pigmented, softened, and loose enamel. Radiographically, this type shows normal enamel thickness but reduced density, which is less than that of dentin. In contrast, the hypoplastic type involves normal enamel maturation but with reduced thickness [8]. Further analysis of AI patterns, particularly the hypoplastic type, is essential, as it pertains to the case report discussed.

In our case, the patient exhibited impacted and maligned teeth with labially inclined 11, labially placed 13, and labially inclined 21, along with amber-colored dentin. According to various reports, approximately 80% of patients with Amelogenesis Imperfecta (AI) have impacted teeth [9–11]. Seow's observations also indicate that individuals with AI are more frequently associated with impacted permanent teeth [11].

Several authors have recommended porcelain-fused-to-metal restorations as a treatment modality for patients with Amelogenesis Imperfecta (AI). Despite advancements in technology that have made full ceramic restorations increasingly popular, porcelain-fused-to-metal crowns remain well-accepted among practitioners due to their superior esthetics, bonding ability to dentin, and effectiveness in restoring both form and function [12, 13]. Providing laminate veneers for such patients may be less advantageous. In the present clinical scenario, full-mouth porcelain-fused-to-metal restorations were chosen for their enhanced stability, mechanical durability, satisfactory esthetics, and protection of the remaining dentin.

Restoring the smile in individuals with Amelogenesis Imperfecta (AI) requires meticulous accuracy, patience, and skill, alongside comprehensive collaboration across various dental specialties. A multidisciplinary approach to treatment planning involves addressing function, aesthetics, and vertical dimension. Several factors must be considered during this planning, including the patient's age, oral hygiene status, quality of life, periodontal health, internal tooth anatomy, remaining tooth structure, and orthodontic considerations [14, 15].

CONCLUSION

Restoring the smile in individuals with Amelogenesis Imperfecta (AI) demands meticulous accuracy, patience, and skill, along with comprehensive collaboration among various dental specialties. A multidisciplinary approach to treatment planning address's function, aesthetics, and vertical dimension. Key factors to consider include the patient's age, oral hygiene status, quality of life, periodontal health, internal tooth anatomy, remaining tooth structure, and orthodontic needs [14, 15].

Conflicts of Interest: There are no conflicts of interest.

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