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Unraveling the Enigma of Amelogenesis Imperfecta: A Detailed Case Report

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Abstract:- Amelogenesis Imperfecta encompasses a range of diverse conditions characterized by structural defects in tooth enamel development, featuring intricate inheritance patterns. These conditions can manifest in both primary and permanent dentition and may be inherited through autosomal dominant, autosomal recessive, or X-linked modes of transmission. The treatment plan involves considering various factors including the patient's age, the nature and extent of the disorder, intraoral factors, and the patient's socioeconomic status. In this report, we present cases of two siblings diagnosed with hypoplastic amelogenesis imperfecta through clinical and radiographic assessments.

Keywords:- *Amelogenesis, Amelogenesis Imperfecta, Enamel hypoplasia*

I. INTRODUCTION

Dental enamel is an epithelial-derived tissue comprised of highly organized hydroxyapatite crystals that form in a defined extracellular space[1].It comprises group of conditions that show developmental alterations in the structure of the enamel[2]. The condition arises due to mutations or changes in any of the genes responsible for encoding particular enamel proteins, including the Enamelin gene (ENAM), Amelogenin gene (AMELX), Kallikrein 4 gene (KLK4), Matrix Metalloproteinase 20 gene (MMP-20), and Distal-less homeobox 3 gene (DLX3).[3].

Amelogenesis Imperfecta (AI) has been categorized based on the clinical, radiographic, and histological characteristics of the enamel anomaly and the inheritance pattern [3]. It is primarily classified into four major categories, which include hypoplastic, hypocalcification, hypo maturation, and hypoplasia-hypo maturation with taurodontism. [4] .The most common classification was given by Witkop and Sauk[2] [Table 1]

The primary clinical problems of AI include tooth sensitivity, loss of occlusal vertical dimension, dysfunction, and esthetics[5]. Diagnosis of AI is primarily based on clinical and radiological features with possible family history[4].An interdisciplinary approach is needed to evaluate, diagnose, and resolve esthetic problems using a combination of periodontal, prosthodontic, and restorative treatment[5].

II. CASE REPORT 1

31-year-old female patient reported to the department with chief complaint of yellowish discoloration of upper and lower teeth since childhood with history of discoloration of deciduous teeth and chipping of teeth since few years. Patient was a known case of right uterine myomas with hydrouteronephrosis and underwent surgery for the same. Patient's family consists of both parents and 5 siblings [1 brother and 4 sisters] in which younger sister has similar discoloration. On extraoral examination, no abnormalities were detected. On intraoral examination, yellowish discoloration of all the maxillary and mandibular teeth with square shaped crown and thinning of enamel noted [Figure 1 a,b,c] which was non-tender with no chipping off of enamel on palpation, dentin exposed 26,27,34,35,36,37,44,46,47, dentinal caries 46,47,36,37, root stumps 35,45. Based on clinical findings provisional diagnosis of Hypoplastic Amelogenesis imperfecta was given. Differential diagnosis of dentinogenesis imperfecta, dental fluorosis, dentin dysplasia was given. Panoramic radiograph was taken which revealed reduced enamel thickness and radiodensity of enamel similar to that of dentin [Figure 2]. The patient is then referred to department of conservative department for the esthetic evaluation.

III. CASE REPORT 2

25-year-old female patient reported to the department with chief complaint of yellowish discoloration of upper and lower teeth since childhood with history of discoloration of deciduous teeth and chipping of tooth since few years. No history of delayed eruption of permanent tooth and no history of early exfoliation of deciduous teeth. Patient's family consists of both parents and 5 siblings [1 brother and 4 sisters] in which elder sister has similar discoloration. On extraoral examination, no abnormalities were detected. On intraoral examination, all the maxillary and mandibular teeth appear to be yellowish in color with square shaped crown with lack of contact between adjacent teeth and flat occlusal surface of posterior teeth with thinning of enamel. Dentin exposed 31, 32,41 ,42, cuspal chip off noted irt 43,44,45,34,35 and height of mandibular teeth appeared reduced which was non-tender, hard in consistency, smooth surface [Figure 3 a, b]. Based on clinical findings provisional diagnosis of Amelogenesis imperfecta hypoplastic type was given. Differential diagnosis of dentinogenesis imperfecta, dental fluorosis, dentin dysplasia was given. Panoramic radiograph was taken and revealed

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reduced enamel thickness and radiodensity of enamel was similar to that of dentin.

IV. DISCUSSION

Amelogenesis Imperfecta is a group of hereditary diseases which affects tooth enamel[6]. The average global prevalence of AI is less than 0.5% [4].It affects both primary and permanent dentition , but has more predilection in permanent dentition and to incisors and first molars[3]. Other anomalies that can accompany AI include delayed tooth eruption, congenital absence of teeth, anterior open bite, taurodontism, pulp calcifications, dentin dysplasias, resorption of both roots and crowns, hypercementosis, root abnormalities, malocclusion, and gingivitis. [3].

Hypoplastic AI occurs due to defect in enamel matrix formation .It shows reduced enamel thickness, pitting and grooves ,hard and translucent enamel[7]. The radiographic signs include a square crown, a relatively thin radiopaque layer of enamel, low or absent cusps, and multiple open contacts between the teeth with normal enamel density. A " picket fence " appearance of the anterior teeth is also noted [8].In both our cases it was diagnosed hypoplastic amelogenesis imperfecta which was asymptomatic and showed reduced enamel thickness and radiodensity of enamel similar to that of dentin in radiograph .

The hypocalcified variant of AI is marked by a crown of normal size and shape but with enamel that is relatively soft and prone to rapid wear[6]. It occurs due to defect in enamel calcification [6]. Radiographically enamel thickness is normal but its density is even less (more radiolucent) than that of dentin[8].

Hypo maturation AI have normal thickness but have a mottled appearance, slightly softer and wears off but not as severe as hypocalcified[7]. Radiographically shows a normal thickness of the enamel, but its density is the same as that of dentin[8].

The hypoplastic-hypomaturation type with taurodontism presents a combination of hypomaturation and hypoplasia[7]. In this variant, the enamel is thin, shows a mottled yellow to brown discoloration, and is pitted. Molar teeth often display taurodontism, while other teeth have enlarged pulp chambers[6].

The differential diagnosis include Dental fluorosis , enamel hypoplasia ,Molar-Incisal Hypo mineralization (MIH)[9]. Early identification of AI, along with appropriate management, is essential for preventing the ongoing deterioration of the dentition and minimizing the potential negative psychological effects on the patient[10].

The management for individuals with AI can be categorized into three distinct stages: The initial phase, referred to as the temporary phase, involves interventions during primary and mixed dentition, the transitional phase follows, spanning from the eruption of all permanent teeth until adulthood and the permanent phase focuses on treatment and maintenance in adulthood[11]. During the primary dentition phase, the treatment objectives involve ensuring optimal conditions for the emergence of permanent teeth, as well as supporting the normal growth of facial bones and temporomandibular joints[11]. To achieve this, various approaches can be employed, such as the placement of stainless-steel crowns, polycarbonate crowns, resin-modified glass ionomers (RMGI), prefabricated crowns (including stainless steel crowns with or without esthetic facings), or direct composite resin when primary molars erupt. These interventions serve to prevent the development of caries and mitigate the wear and tear of defective enamel.[11] In the mixed dentition phase, the treatment goals shift toward preserving tooth structures, maintaining tooth vitality, reducing tooth sensitivity, maintaining the vertical dimension, and enhancing esthetic outcomes.

In the permanent dentition stage, the ultimate treatment aims include reducing tooth sensitivity and restoring the vertical dimension of occlusion, thereby enhancing functionality and esthetic appearance. Management include full mouth rehabilitation which can be done combined with a multidisciplinary approach comprising prosthodontics, periodontics, orthodontics, and endodontics[11]. Periodontal care encompasses procedures such as crown lengthening and gingival reshaping for addressing short clinical crowns and gingival hyperplasia. Orthodontic interventions may be employed to close gaps between teeth and rectify anterior open bites. Root canal therapy becomes necessary when pulp exposure occurs due to extensive attrition or significant tooth reduction. In cases of severe malocclusion, orthognathic surgery may be recommended as a potential treatment option.[11].

V. CONCLUSION

This case report sheds light on the rare genetic disorder Amelogenesis imperfecta. Through thorough analysis of the patient's clinical presentation, diagnostic procedures, and treatment options, we have gained valuable insights into the challenges posed by this condition. By sharing this information, we hope to contribute to the medical community's understanding of Amelogenesis imperfecta and aid in the development of more effective interventions for affected individuals. Continued research and collaboration are essential to further unravel the complexities of this disorder and improve the quality of life for those impacted by it.

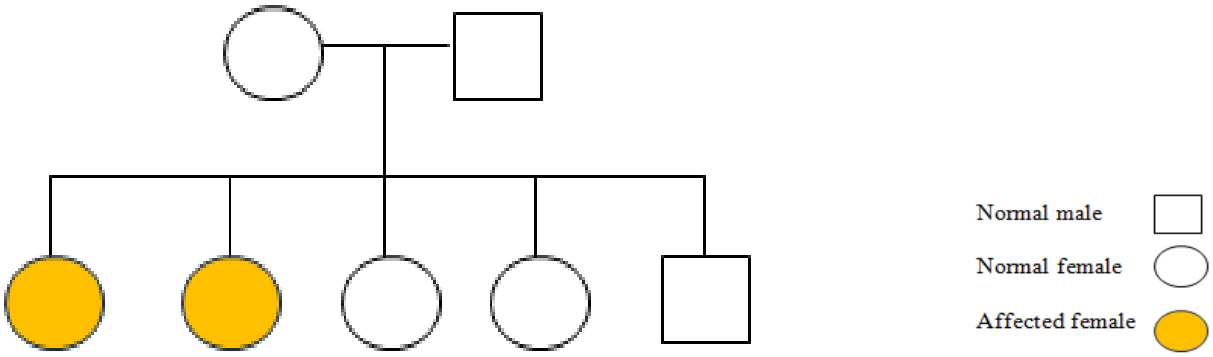

CHART NO. 1 PEDIGREE CHART

Table 1 Classification of Amelogenesis Imperfecta According to Witkop (1989)

Type I	**Hypoplastic**
IA	Hypoplastic, pitted autosomal dominant
IB	Hypoplastic, local autosomal dominant
IC	Hypoplastic, local autosomal recessive
ID	Hypoplastic,smooth,autosomal dominant
IE	Hypoplastic , smooth X-linked dominant
IG	Enamel agenesis ,autosomal recessive
Type II	Hypomaturation
IIA	Hypomaturation, pigmented autosomal recessive
IIB	Hypomaturation, X linked recessive
IIC	Snow-capped teeth, autosomal dominant
Type III	Hypocalcified
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

CLINICAL PHOTOS

- *Case 1*

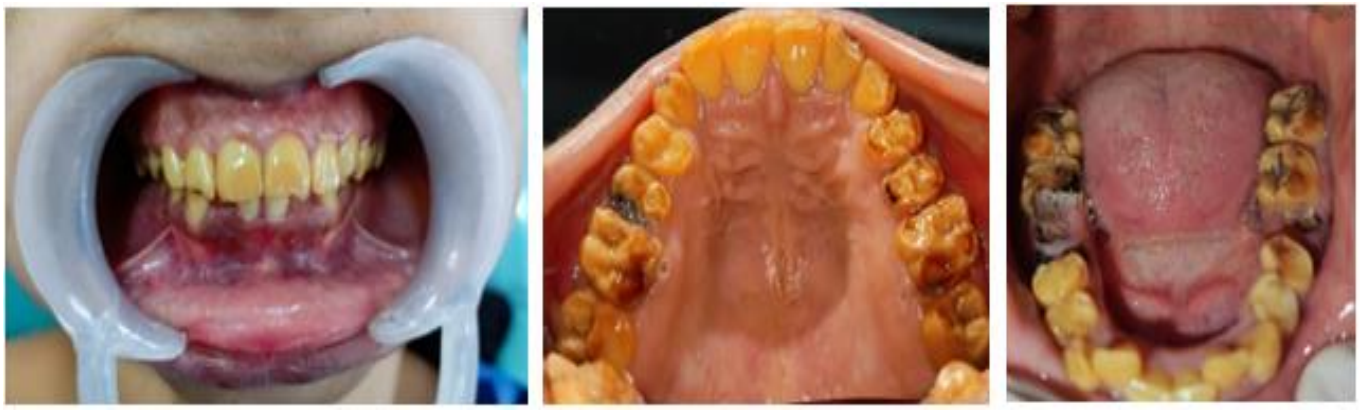

Fig 1 (A)Generalized Yellowish Discoloration on Maxillary and Mandibular Teeth
(B)Maxillary Occlusal View (C)Mandibular Occlusal View

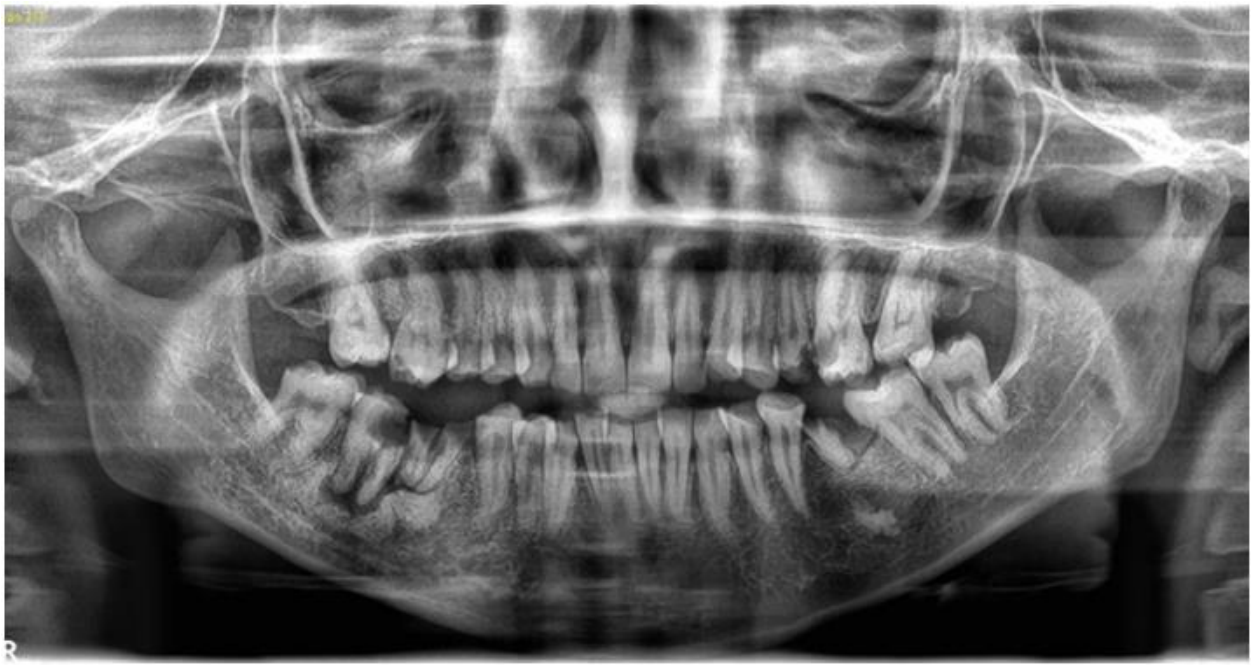

Fig 2 Panoramic Radiograph Reveals Reduced Enamel Thickness and Radiodensity of Enamel Similar to that of Dentin.

- *Case 2*

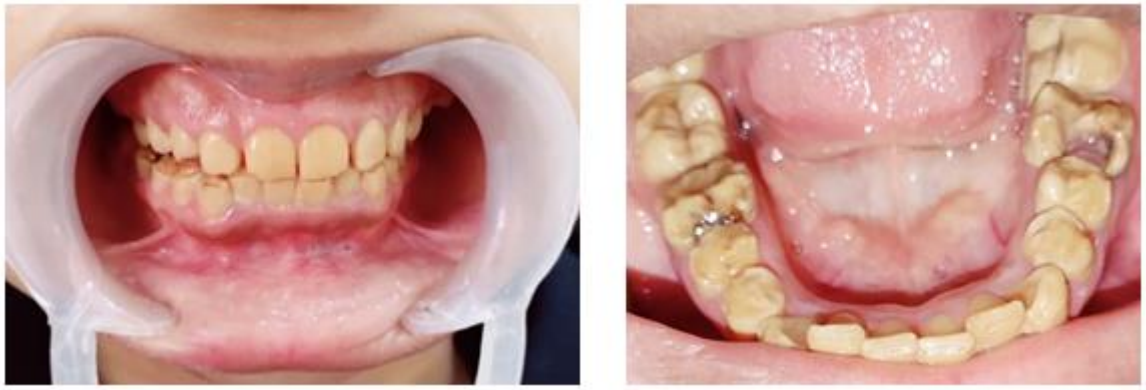

Fig 3 (A) Generalized Yellowish Discoloration on Maxillary and Mandibular Teeth (B)Mandibular Occlusal View

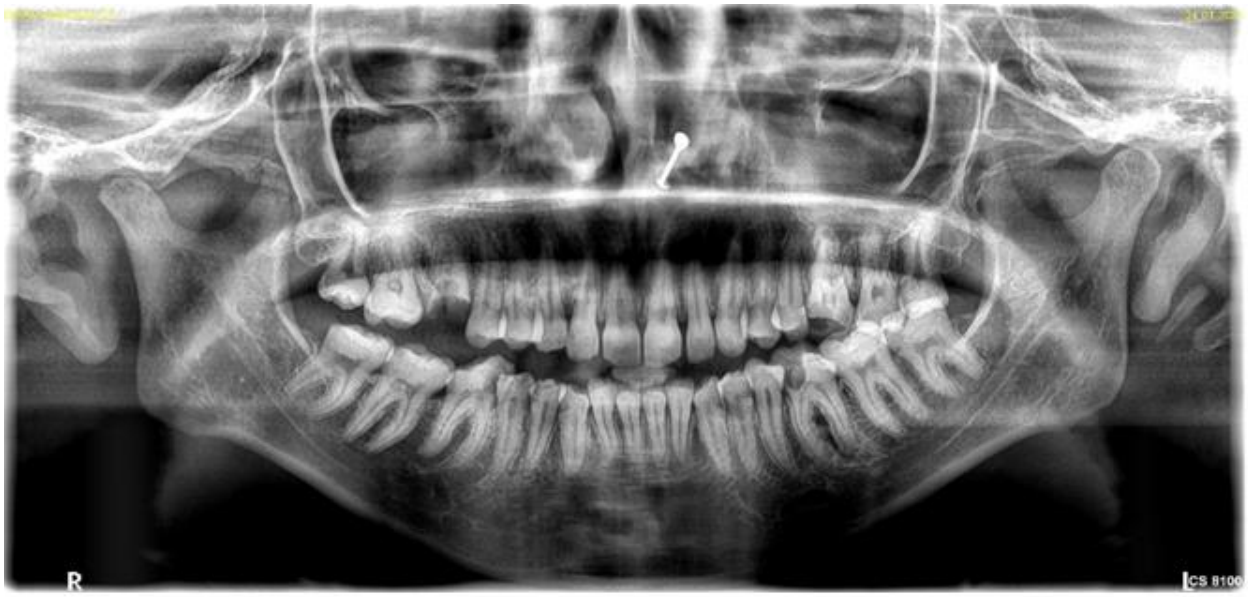

Fig 4 Panoramic Radiograph Reveals Reduced Enamel Thickness and Radiodensity of Enamel Similar to that of Dentin.

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