Case Report

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Enamel Agenesis: A Rare Clinical Variant of Amelogenesis Imperfecta

Ranjit Manne[1*] Manoj GS[2] G Chandrasekhar[3] J Shubhaker[4]

[1]Professor, Department of Orthodontics and Dentofacial Orthopedics, Panineeya Institute of Dental Sciences, Hyderabad, India.
[2]PG Student, Department of Orthodontics and Dentofacial Orthopedics, Panineeya Institute of Dental Sciences, Hyderabad, India.
[3]Professor and Head, Department of Orthodontics and Dentofacial Orthopedics, Panineeya Institute of Dental Sciences, Hyderabad, India.
[4]Raeder, Department of Orthodontics and Dentofacial Orthopedics, Panineeya Institute of Dental Sciences, Hyderabad, India.

ABSTRACT

Background: Amelogenesis Imperfecta (AI) is one of the heterogeneous group of developmental disorders that effects the enamel. Amelogenesis Imperfecta is as an ectodermal disorder which causes white flecks, narrow horizontal bands, lines, pits, grooves, soft and friable enamel, yellowish to dark brown discoloration of teeth. In certain conditions of AI, a diffusely reduced thickness of enamel is considered as Enamel Agenesis. The present paper describes about the clinical, radiological and histopathological findings of a patient with Enamel Agenesis a rare variant of Amelogenesis Imperfecta.

Keywords: Enamel Agenesis, Amelogenesis Imperfecta.

INTRODUCTION

Amelogenesis imperfecta (AI) is one of the heterogeneous group of developmental disorders that effects the enamel. The congenital enamel defects that are not accompanied by other metabolic disorders other than tooth form or eruption are classified under AI.[1] Amelogenesis Imperfecta has been associated with abnormalities in dental eruption, congenitally missing teeth, anterior open bite, pulpal calcifications, dentin dysplasias, root and crown resorption, hypercementosis, root malformations and taurodontism.[2,3]

Amelogenesis Imperfecta is an ectodermal disorder which causes white flecks, narrow horizontal bands, lines, pits, grooves, soft and friable enamel and yellowish to dark brown discoloration of teeth.[4,5] In certain conditions of AI, a diffusely reduced thickness of enamel is considered as *Enamel Agenesis.* [12] According to Alvares and Souza Freitas, "this alteration is probably inhibitory in nature and causes atrophy and lack of function of ameloblastic cells, leaving a result of structural defects on enamel formation".[6]

Amelogenesis Imperfecta was first reported in 1890 but was not considered as a clinical entity which is different from dentinogenesis imperfecta until 1938[7]. Its prevalence varies widely between studies, from 1 in 14,000[1] to 1 in 4,000[8] depending on the diagnostic criteria used and the population group studied. In some reported cases, the hereditary pattern is autosomal dominant and others it is recessive. There are also reports linking AI with X chromosome. It has been suggested that alteration in the process of degradation and resorption of enamel matrix contribute to the characteristic hypomineralization of the affected enamel.[11]

Classification of AI that was proposed by Witkop and Sank in 1976[9] is widely accepted which considers the inheritance pattern of the disorder as well as its specific clinical characteristics.[1]

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*Correspondence Dr. Ranjit Manne.
Department of Orthodontics and Dentofacial Orthopedics, Panineeya Institute of Dental Sciences, Hyderabad, India.
Email: ranjit_manne@yahoo.co.in

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The molecular defect identified with the appearance of AI is related to the gene (*AMELX* gene) that codifies amelogenin, the most abundant protein of the original matrix.[10] Despite recent advances in detecting AI in a molecular level, the information related to phenotype and genotype are still at an infantile stage. Following is a case report on enamel agenesis (Witkop: Type 1G), a rare variant of Amelogenesis Imperfecta.

CASE REPORT

A 15 years old male patient visited the department of orthodontics and dentofacial orthopedics with chief complaints of esthetics, improper biting and chewing and severe teeth sensitivity. His general medical health status was revealed as normal and as per physicians advice various blood investigations (serum electrolytes, serum calcium, thyroid and lipid profiles) were performed which were normal. Patient was not associated with any other syndromes. Consanguineous marriage of his parents was reported. Paternal uncle son has similar condition of teeth. Patient had reported severe teeth sensitivity while having hot, cold, sweet and sour food. On general examination hyperextensibility of left hand index finger was seen which was not related to AI or any other medical condition.

On intraoral examination, retained deciduous teeth, few erupted permanent teeth and multiple permanent missing teeth were found. Both deciduous and permanent teeth were hypoplastic and yellowish-brown in color. The surfaces of the teeth were smooth with complete lack of enamel and severe attrition of teeth were seen. Lateral open bite was present. (Fig. 1 B-F) On radiological examination, multiple unerupted teeth submerged in the bone and complete loss of enamel was noticed. (Fig. 2)

A provisional diagnosis of Amelogenesis Imperfecta was made based on the clinical and radiographic findings. Stereomicroscopic images of ground section of a therapeutically extracted deciduous molar confirmed the absence of enamel and final diagnosis was Enamel Agenesis (type 1G) subtype of hypoplastic type of Amelogenesis Imperfecta. (Fig. 3)

Based on the clinical and radiological findings, the case was started with acrylic crowns on upper and lower anterior teeth for enhancing esthetics, to prevent tooth sensitivity, to maintain space in the anterior region and to prevent further attrition of teeth. Therapeutic extractions of all deciduous teeth were performed to facilitate natural eruption of posteriors. Upper Nance button and lower lingual arch were cemented for space maintenance in the posterior segment. (Fig .4)

DISCUSSION

It is difficult to diagnose AI due to its complex classification and absence of relevant data from family members.[13] The clinical features of Amelogenesis Imperfecta vary according to the type.[13,14,15] In the hypoplastic type of Amelogenesis Imperfecta, the teeth exhibit a chalky white to dark brown colour, the occlusal surfaces, and incisal edges are generally abraded, and occasionally the complete loss of the enamel is observed. In the hypocalcified form of Amelogenesis Imperfecta, the enamel exhibits a cheesy consistency and can be easily removed with a sharp explorer. The hypomaturative type of Amelogenesis Imperfecta is characterized by normal enamel thickness with the appearance of white opaque areas on the incisal surfaces. Amelogenesis imperfecta can be associated with other systemic disease or syndrome. One of them is enamel renal syndrome which is characterized by nephrolithiasis and hypocitraturia.[16] Tricho-dento-osseous syndrome (TDO) is often confused with hypoplastic type of Amelogenesis Imperfecta. Certain clinical features like nail defects, kinky hair differentiate TDO from AI.[18]

In the present case general medical health status was found to be normal. Based on the clinical, radiological and histopathological features, it was diagnosed as Enamel Agenesis (type 1G) subtype of hypoplastic-type of Amelogenesis Imperfecta. A multi disciplinary approach consisting of Oral Medicine and Radiology, Oral Pathology, Prosthodontics, Orthodontics and Endodontics may be beneficial in the treatment of AI.

In the present case, to prevent tooth sensitivity, to maintain space in the anterior region , to prevent further attrition of teeth and to enhance esthetics, acryclic crowns were given in both upper

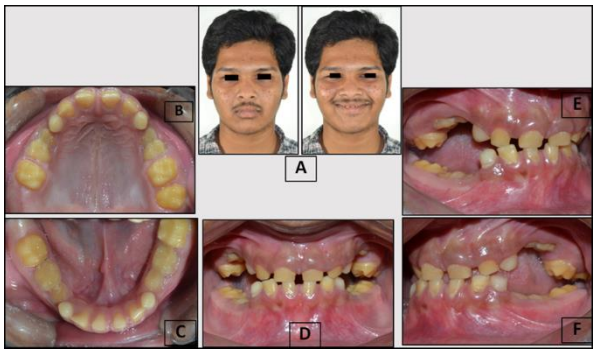

Fig 1: Showing retained deciduous teeth with few erupted permanent teeth and multiple missing permanent teeth. Severe attrition of teeth with lack of enamel. Maxillary and mandibular occlusal views (B and C); Intraoral frontal view (D); Right and left lateral views (E and F); Facial photographs (A).

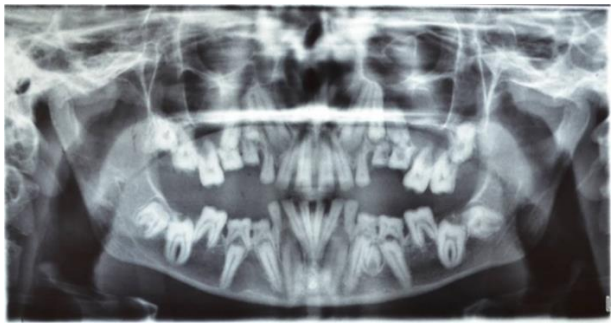

Fig 2: Pantomograph showing multiple impacted permanent teeth and retained deciduous teeth with complete lack of enamel.

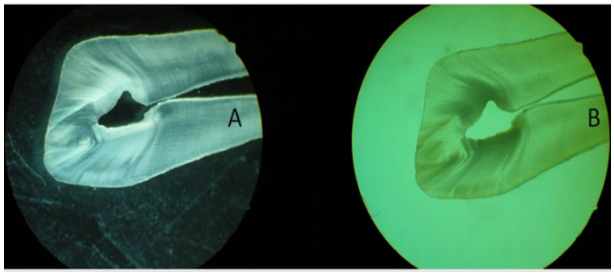

Fig 3: A photomicrograph of a ground section of extracted deciduous teeth showing complete absence of enamel.

and lower anterior region. A wait and watch policy was taken in order to allow natural eruption of submerged permanent teeth after therapeutic extractions of deciduous teeth and maintaining the posterior extraction space with the help of space maintainers.

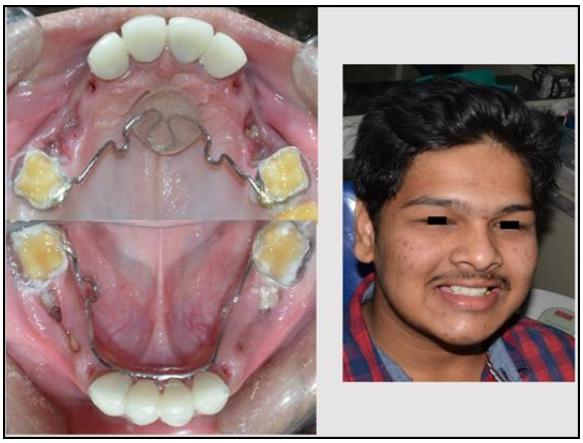

Fig 4: Acrylic crowns placed; Deciduous teeth were extracted and space maintainers were cemented.

CONCLUSION

There should be adequate knowledge of diseases and systemic disorders that effect the development and malformation of dentition, that helps in diagnosing and planning treatment of Enamel Agenesis a rare variant of Amelogenesis Imperfecta which requires a multidisciplinary approach.

CONFLICT OF INTEREST

No potential conflict of interest relevant to this article was reported.

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