Amelogenesis Imperfecta with Class III Malocclusion: A Case Report

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ABSTRACT

Amelogenesis imperfecta (AI) is a group of the genetic disorder that affects the morphology and quality of the tooth structure. Although the disease entity is primarily affected with the abnormality of dental and oral structures, it has been associated with a few syndromes. Although the patients often are non-symptomatic, the trait can be passed on to a child and if both parents carry the trait, the child could develop a more severe form of the disease; therefore, early diagnosis is important. It reduces the oral health-related quality of life and causes physiological problems. Here we present a case report of a patient hypoplastic hypomaturation, autosomal dominant type of AI based on the family history, pedigree chart, clinical, radiographical, and histological features.

Keywords: Amelogenesis imperfecta, Enamel hypoplasia, Malocclusion, Microdontia

INTRODUCTION

Amelogenesis imperfecta (AI) encompasses a heterogeneous group of developmental disorders that demonstrates alterations in the enamel. It is characterized by clinical and genetic heterogeneity in the absence of systemic abnormalities or diseases.¹ AI is also known by varied names such as hereditary enamel dysplasia, hereditary brown enamel, and hereditary brown opalescent teeth.²

AI encompasses a complicated group of conditions that demonstrate developmental alterations in the structure of the enamel in the absence of a systemic disorder. The prevalence of this condition has been expected to range from 1 in 718 to 1 in 14,000, depending upon the population studied.³ Hypoplastic AI represents 60-73% of all cases, hypomaturation AI represents 20-40%, and hypocalcification AI represents 7%. No racial predilections of the AI have been reported. Both primary and permanent dentitions are usually affected.⁴

AI is caused by mutations in genes that control amelogenesis and follows inheritance patterns of autosomal dominant, autosomal recessive or X-linked modes of transmission. There are also patients for whom family history cannot be identified, but where a mutation is present.

The inheritance pattern of X-linked disorders dictates that male to male transmission cannot occur. Conversely, all female offsprings of the affected male must be affected. Affected females have a 50% of passing on the trait to the offspring of either sex. Mutations in the amelogenin gene (AMELX) cause X-linked AI, while mutations in the enamelin gene (ENAM) cause autosomal-inherited forms of AI. Recent reports involve kallikrein-4 (KLK4), matrix metalloproteinase gene (MMP-20) and distal-less homeobox 3 (DLX3) genes in the etiologies of some cases.⁵

Witkop and Sauk listed the varieties of AI, divided according to whether the abnormality lay in a reduced amount of enamel (hypoplasia), deficient calcification (hypocalcification), or imperfect maturation of the enamel (hypomaturation), and also recognized the combined defects.⁶ Clinically, AI appears as an alteration of enamel formation resulting in hypoplasia, hypocalcification, and hypomaturation. Enamel hypoplasia results in a decreased quantitative enamel
formation. The enamel in hypocalcification appears normal but poorly mineralized while hypomaturation results in an abnormal mineralization in the final stages of tooth formation. The most common form, the hypoplastic type, is deficient in normal enamel.\(^7\) The crowns of the teeth appear blanched, snow-capped, yellow brown, pitted or grooved.

Diagnosis involves exclusion of extrinsic environmental or other factors, establishment of a likely inheritance pattern, recognition of phenotype and correlation with the dates of tooth formation to exclude a chronological developmental disturbance. Furthermore, dental radiography in form of orthopantomogram (OPG) and full mouth intraoral radiographs plays a vital role in diagnosing the difference in density of enamel in AI patients and normal patients along with dentin thickness, pulp canal and root length.\(^8\)

**CASE REPORT**

A 26-year-old Male patient reported to the Department of Oral Medicine and Radiology, with a chief complaint of discolored teeth since childhood and an associated complaint of difficulty in chewing hard food. Patient gave a positive history of discoloration with deciduous and permanent dentition; there were no complaints of unusual sensitivity or thermal changes, in response to food or drinks. There were no systemic symptoms associated. His main concerns were difficulty in mastication and his appearance; hence he visited the dental outpatient department for treatment. Past dental history revealed that the patient had undergone the extraction due to dental caries 4 years back, which was uneventful. Past medical history was not significant. Patient is residing in a non-fluoridated area since his birth. After taking a brief family history regarding the condition, the following pedigree chart was constructed (Figure 1).

On extra-oral examination, the patient had no facial asymmetry and the patient’s hair, skin and nails were normal (Figure 2).

On intra-oral examination, he had a normal complement of teeth with clinically missing 22, 26 and 37 (Figures 3 and 4).

Intra-oral photograph showed generalized brownish stains, hypoplastic enamel without pitting and anterior open bite (Figure 5).

The teeth in general exhibited yellowish brown discoloration with diffuse sloughing on labial surfaces of anterior teeth that was soft in consistency. The thickness of enamel was found to be reduced on 17, 16, 15, 11, 21, 23, 42, 46 exposing the dentin. He had high arched U-shaped palate, reduced vertical dimension and anterior deep bite with Class III malocclusion.

![Pedigree chart of the case](image1.png)

![Frontal profile picture of the patient](image2.png)

![Intra-oral photograph showing upper arch (maxilla) with roughly malformed enamel and clinically missing 22,26](image3.png)
Based on these clinical findings a provisional diagnosis of AI was given with a differential diagnosis of dentinogenesis imperfecta and dentin dysplasia.

The patient was subjected to radiographic investigations. An intra-oral periapical radiograph in relation to 25 revealed relative thickening and apparent blunting of the root apex, separated from periapical bone by normal appearing periodontal ligament space suggestive of hypercementosis (Figure 6).

An OPG showed missing 22, 26 and 36, reduced enamel thickness with normal dentin. The enamel appeared more radiodense than dentin, with normal pulp chamber and root canal spaces without any signs of obliteration. There was a loss of cuspal height with open contacts in posterior teeth and roots in the lower arch were elongated with bulbous appearance (Figure 7).

The patient underwent extraction of 25 and the tooth was subjected to histological examination. Ground section showed areas of hypo calcification adjacent to dentino enamel junction (DEJ), obliteration of DEJ was seen at some areas (Figure 8).

The diagnosis of hypoplastic hypomaturation, autosomal dominant type of amelogenesis imperfecta with partial anodontia was confirmed on the basis of family history, pedigree chart, clinical, radiographic, histological features and according to classification given by Witkop and Sauk.9

Figure 4: Intra-oral photograph showing lower arch (mandible) with roughly malformed enamel and clinically missing 36

Figure 5: Intra-oral photograph showing generalized brownish stains, hypoplastic enamel without pitting and anterior open bite

Figure 6: Photograph of IOPA of upper anterior region showing 23, 24, 25& 27 with thin enamel & normal dentin, pulp canal and root length, relative thickening and apparent blunting of root apex, separated from periapical bone by normally appearing periodontal ligament space suggestive of hypercementosis

Figure 7: Photograph of OPG showing reduced thickness of enamel with loss of cuspal height, open contacts in posterior teeth and elongated roots in lower arch with bulbous appearance.

Figure 8: Ground section showing areas of hypocalcification adjacent to dentino-enamel junction (DEJ) with obliteration of DEJ in some areas.
The treatment proposed for him included oral prophylaxis, restoration of malformed teeth, crown placement following root canal treatment, esthetic rehabilitation with fixed full cast crown of porcelain fused to metal.3,10 The patient was asked to follow-up once in 6 months.

**DISCUSSION**

AI is a developmental, often inherited disorder affecting dental enamel. Mutation or alteration in any of the genes encoding specific enamel proteins such as ENAM, AMELX, KLK4, MMP-20, and DLX3 gene have been linked with AI.11,12 The genetic pattern responsible for the disease may be autosomal dominant, autosomal recessive, sex-linked or even sporadic.

The pedigree chart constructed for the present case showed a vertical as well as horizontal distribution of the AI. The patient’s father, his younger brother and sister, were affected by similar condition. But his mother, brother, and his mother’s family side were not affected. This indicated autosomal dominant type of enamel hypoplasia.

The predominant clinical manifestations reported are enamel hypoplasia, hypomineralization (subdivided into hypomaturation and hypocalcification); or a combined phenotype, which is seen in most of the cases. Other anomalies reported are congenitally missing teeth, delayed tooth eruption, taurodontism, dentin dysplasias, pulpal calcifications, hypercementosis, Class III malocclusion, anterior open bite and posterior cross bite.

The clinical presentation of the AI varies according to its type. In the hypomaturation type, the affected teeth exhibit mottled, opaque white-brown yellow discolored enamel, which is softer than normal. In radiographs, the thickness of enamel is normal, but the density is the same as that of the dentin.13

The hypocalcified type shows pigmented, softened, and easily detachable enamel. Radiographically, enamel thickness is normal, but its density is even less than that of the dentin. The enamel is well-mineralized, but its amount is reduced. Radiographs exhibit a thin peripheral outline of radiodense enamel and low or absent cusps.

In hypoplastic-hypomaturation type with taurodontism, the enamel is thin, mottled yellow to brown, and pitted. The molars exhibit taurodontism and other teeth have enlarged pulp chambers. Compared with the dentinogenesis imperfecta our patient did not complain of sensitivity of teeth since the dentin is intact.

The absence of bluish brown/amber translucency and obliteration of the pulp chamber helped us to differentiate it from dentin dysplasia.

AI presents with problems of socialization, function, and discomfort which should be managed by early vigorous intervention, both preventively and restoratively. Hence, a multidisciplinary approach with a team consisting of an orthodontist, prosthodontist and endodontist should be considered for treatment of the same.14

**CONCLUSION**

AI is a heterogeneous developmental disorder present with severe dental anomalies. It is the responsibility of the dentist to diagnose the condition as early as possible and to consider the social implications for these patients and intervene to relieve their suffering. Thus, in this article an attempt was made to improve the clinician’s knowledge about the clinical and radiographic diagnosis; as well as the intervention required for such a condition.

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