

CASE REPORT

Amelogenesis Imperfecta - A Case Report with Genetic Transmission

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ABSTRACT:

Amelogenesis imperfecta (AI) is a hereditary disorder expressing a group of conditions that cause developmental alterations in the structure of enamel. AI is a serious problem that reduces oral health-related quality of life and causes some physiological problems. The treatment of patients with AI may upgrade the quality of life and reinforce their self-esteem. The AI trait can be transmitted by either autosomal dominant, autosomal recessive, or X-linked modes of inheritance. It is necessary to diagnose the case and provide durable functional and esthetic management of these patients in order to improve the quality of their lives. We present a case of amelogenesis imperfecta affecting the dentition of a 36 year old male which was inherited by his 3 year old daughter.

Key words: *Amelogenesis Imperfecta, hereditary disorder, developmental*

INTRODUCTION

Amelogenesis imperfecta (AI) is a hereditary disorder that expresses a group of conditions that cause developmental alterations in the structure of enamel. Generally both the primary and permanent dentitions are diffusely involved. In the affected teeth, the dentin and root form are usually normal. These teeth are more resistant to decaying. No racial predilections of the AI have been reported.¹

Classifications of AI are primarily based on phenotype and mode of inheritance. The most commonly used classification was proposed in 1988 by Witkop, and revised by Nusier in 2004. Based on enamel appearance and hypothesized developmental defects, AI is classified as 4 patterns: hypoplastic, hypomaturation, hypocalcified, and hypomaturation-hypoplastic.¹

The primary clinical problems of AI are tooth sensitivity, loss of occlusal vertical dimension, dysfunction, and esthetics. Restoration of these defects is important not only because of esthetic and functional concerns, but also because there may be a positive psychological impact for the patient. Treatment planning for patients with amelogenesis imperfecta is related to many factors: the age and socioeconomic status of the patient, the type and severity of the disorder, and the intraoral situation. An interdisciplinary approach is necessary to evaluate, diagnose, and resolve esthetic problems.²

The disease is genetically and clinically heterogeneous, and autosomal dominant, autosomal recessive and X-linked inheritance were established in an extensive study of Swedish families with AI. However, there is no apparent correlation between the phenotype and the mode of inheritance.³

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The aim of this paper is to present a case of amelogenesis imperfecta affecting a 36 year old male showing genetic transmission to his daughter, with an insight on the genetic aspects of this disease.

CASE REPORT

A 36-year-old male patient presented with a chief complaint of discoloured teeth. Medical history was non contributory. Extraoral examination did not reveal any relevant findings.

The patient's oral hygiene was poor and he presented hyperemic and edematous gingiva.

The teeth appeared brown with a roughened surface with generalised extrinsic and intrinsic stains [Figure 1]. There was loss of contact between the teeth. Vertical dimension of the face was also decreased. There was no anterior open bite. The mandibular teeth showed spacing. Severe attrition was seen in both maxillary and mandibular teeth [Figure 2].

The panoramic view showed all 32 teeth which were fully erupted. There was no retention of teeth, none of the teeth were missing. No pulp calcifications or external resorption was seen [Figure 3].

Radiographically, the enamel layers of all teeth were absent giving a tapered appearance.

Pulp chambers had normal size and shape. The lamina duras were normal.

Incidentally his daughter had the same complaint of discolouration [Figure 4]. Her maxillary and mandibular incisors were discoloured and attrited [Figure 5]. Radiographic examination revealed loss of enamel layer, [Figure 6] similar to that seen in her father.

A diagnosis of amelogenesis imperfecta (hypoplastic type) was made.

DISCUSSION

Amelogenesis imperfecta (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 on the population studies. Hypoplastic AI represents 60-73% of all cases, hypomaturation AI represents 20-40%, and hypocalcification AI represents 7%.⁴

Clinical presentation of the AI varies according to its type. In the hypomaturation type, the affected teeth exhibit mottled, opaque white-brown yellow discoloured enamel, which is softer than normal. In radiographs, the thickness of enamel is normal. 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. In the hypoplastic type, the enamel is well-mineralized but its amount is reduced. Clinically grooves and pits will be realized on the surface of the fine enamel. The rough pattern of hypoplastic type, exhibits thin, hard, and rough surfaced enamel. The tooth is tapered towards the incisal/ occlusal face and has open contact points. Radiographs exhibit a thin peripheral outline of radiodense enamel, and low or absent cusps.⁴ Clinical and radiographic appearances of the teeth of our case were in accordance with hypoplastic type.

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.⁵

AI may be associated with some other dental and skeletal developmental defects or abnormalities, such as crown and root resorption, attrition, taurodontism, delayed eruption and tooth impaction, dens in dente, pulp stones, anterior open bite and agenesis of teeth. In our case, tooth impaction, delayed eruption, crown resorption, pulp stones, and tooth agenesis were present. Up to date, 15 cases of delayed eruption, 14 cases of pulp stones, 9 cases of tooth agenesis have been reported.⁶

According to Seow^[7] pulp stones are formed as the result of the external local irritation because of the thin enamel layer and attrition.

Genetics

Early developing enamel matrix is rich in protein, but it successively loses its protein, and finally

becomes highly mineralized. The matrix proteins are a heterogeneous group which are generally separated into amelogenins (major component) and amelins. Amelins have been suggested to function as nucleation sites for the hydroxyapatite crystals, while amelogenins are thought to regulate the rate, size and pattern of hydroxyapatite crystal growth. Two separate genes have been isolated for human amelogenin, one on the X chromosome, and one on the Y chromosome.⁸

To date, mutations in gene encoding the protein amelogenin have been shown to cause some X-linked recessive forms of AI. This locus has been designated AIH1. The heterogeneity that characterizes this group of disorders has limited further progress to identification of additional loci by linkage analysis, however the underlying genes have not been identified. For example a second locus for X-linked recessive AI, (AIH3) has been mapped to chromosome Xq24-q27.1 Similarly, an autosomal dominant, local hypoplastic form of AI, (AIH2) has been mapped to a 4mb region of human chromosome 4q11-q21 that encompasses the gene encoding the ameloblast specific protein ameloblastin, AMBN.

Recently a gene encoding a second ameloblast specific protein, Enamelin has been mapped to the AIH2 critical region within 15kb of AMBN.⁹

The enamel gene, ENAM, has been mapped with different techniques to the same region on chromosome 4q as AIH2 and AMBN, suggesting that this region could contain a cluster of genes encoding enamel proteins. A splice site mutation in ENAM was recently found to be associated with autosomal dominant AI, where the patients presented with smooth hypoplastic enamel.¹⁰

Treatment

Treatment for amelogenesis imperfecta includes replacement of the missing teeth and protecting the existing dentition with crowns and bridges. Though AI is unsightly and painful at times the oral rehabilitation has good prognosis, esthetically and functionally, if the situation is not complicated with too many anodontia or impacted teeth. Recently, new materials called second-generation laboratory

composites, poliglases or ceromers have been developed. They have different filler components that improve wear resistance, physical properties and their use has expanded into posterior intracoronal, full crown and even fixed partial denture restorations.

CONCLUSION

Amelogenesis imperfecta (AI) represents a group of developmental conditions, genomic in origin, which affect the structure and clinical appearance of enamel of all or nearly all the teeth in a more or less equal manner and which may be associated with morphologic or biochemical changes elsewhere in the body. We have presented a case of amelogenesis imperfecta affecting a 36 year old male and his 3 year old daughter emphasizing its clinical features and genetic transmission.

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FIGURE 1: Brownish teeth with a roughened surface with generalised stains.



FIGURE 4: Discoloured maxillary central incisors.

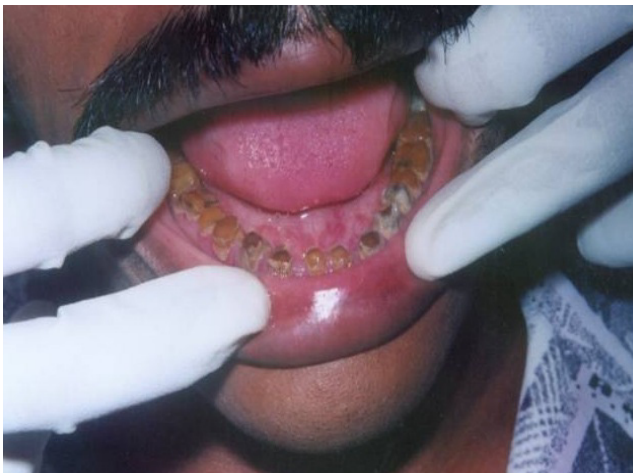


FIGURE 2: Discoloured and attrited mandibular teeth.



FIGURE 5: Discoloured mandibular central incisors.



FIGURE 3: OPG showing loss of enamel layers of all the teeth.

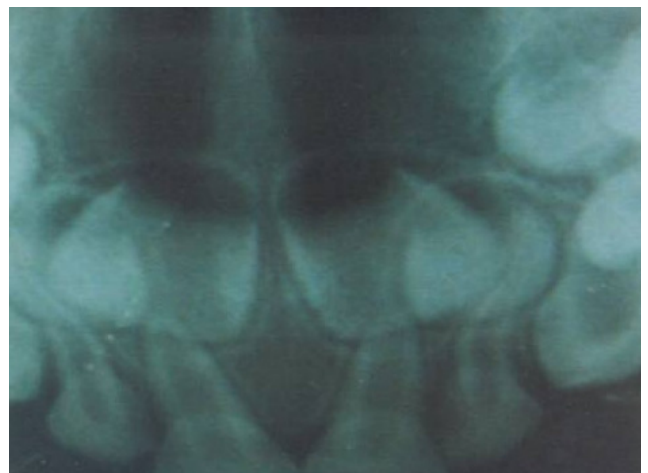


FIGURE 6: IOPA showing loss of enamel layer.