ABSTRACT

Amelogenesis imperfecta (AI) is a diverse collection of inherited diseases that exhibit qualitative or quantitative tooth enamel defects in the absence of systemic manifestations. Also known by varied names, such as hereditary enamel dysplasia, hereditary brown opalescent teeth, this defect is entirely ectodermal, since mesodermal components of the teeth are basically normal. This article details a case of AI along with complete review which presents in his twin siblings with clinical, radiological and histopathological report.

Keywords: Amelogenesis imperfecta, Mutation, Enamel protease, Inherited disease.


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INTRODUCTION

Dental enamel, the most highly mineralized structure in the human body, is formed within a unique, extracellular matrix derived through the synthesis and secretion of proteins by the ameloblast cells. Dental enamel differs from other mineralized tissues, such as bone, cartilage and dentine, in that it is noncollagenous, originated from epithelium, and does not undergo resorption and remodeling.1

Dental enamel formation is divided into secretory, transition and maturation stages.2 During the secretory stage, enamel crystals grow primarily in length. The crystallites lengthen at a mineralization front formed near the secretory surfaces of the ameloblast cell. During the maturation stage, mineral is deposited exclusively on the sides of the crystallites, which grows in width and thickness to coalesce with adjacent crystals. The arrangement of ions in dental enamel crystals closely approximates that of calcium hydroxyapatite. Sketched out in the protein matrix, the hydroxyapatite crystal architecture continues to grow in width and thickness due to a progressive protein hydrolysis.2 The main structural proteins in forming enamel are amelogenin, ameloblastin and enamelin. These proteins are proteolytically cleaved following their secretion. Some of the cleavage products accumulate in the enamel layer, while others are either degraded or reabsorbed by ameloblasts.2

AI represents a group of conditions, genomic in origin, which affect the structure and clinical appearance of the 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.3 In its mildest form, AI causes discoloration, while in the most severe presentation the enamel is hypominalerized causing it to be abraded from the teeth shortly after their emergence into the mouth.4 Both the primary and permanent dentitions were affected. Enamel findings in AI are highly variable, ranging from deficient enamel formation to defects in the mineral and protein content.5 The four main types of AI were described as follows: Hypoplastic, hypominalerized, hypomaturation and with taurodontism.6

In this paper, we present a case showing the clinical, radiological and histopathological features of this entity.

CASE REPORT

A 24-year-old male patient came to our department of oral medicine and radiology with the chief complaint of small, yellowish and unesthetic appearance of all the teeth (Fig. 1A) and came for the rehabilitation of the same. History revealed that his primary dentition was also discolored and gave a familial history of his twin siblings (younger sisters) having the same problem and their parents had consanguineous marriage. The patient’s medical history revealed no systemic abnormalities. On intraoral examination patient had many retained deciduous teeth and permanent teeth had loss of enamel with yellowish discoloration and severe attrition leading to loss of vertical height of the face. His twin siblings were also examined who also presented with the same findings (Figs 2A to D).

On radiographic investigation, full mouth IOPA (Fig. 1B) revealed that all the teeth had relatively thin radio-paque layer of enamel with normal pulp chamber and root canal spaces with no signs of obliteration. Loss of cuspal height in the posterior teeth with open contacts was noted. OPG (Fig. 1C) revealed the same features in the teeth with impacted permanent teeth in relation to 13, 14, 12, 23, 24, 25, 34, 33, 42, 43, 44 and 45 and retained deciduous teeth in relation to 52, 53, 54, 55, 63, 64, 65, 73, 74, 75, 82, 83, 84 and 85 with characteristic picket fence appearance of the teeth due to loss of contacts and small square-
shaped crowns. The radiographic investigations of his siblings also revealed the same features. Correlating his history, clinical and radiological features, we came to a provisional diagnosis of Type I hypoplastic amelogenesis imperfecta. Following which a single tooth was extracted and sent for histopathological evaluation (Fig. 1D) and the ground section of the tooth showed normal dentin and cementum with absence of enamel.

The patient was referred to the department of prosthodontia for complete rehabilitation, where they planned to remove the existing primary teeth and impacted teeth. With the use of the remaining permanent teeth as the abutment, fixed prosthesis was given.

DISCUSSION

Amelogenesis imperfecta encompasses a complicated group of conditions that demonstrate developmental alterations in the structure of the enamel in the absence of the systemic disorder.7 The prevalence of this condition has been expected to range from 1 in 718 to 1 in 14,000, depending on the population studied, hypoplastic AI represents 60 to 73% of all cases, hypomaturation AI represents 20 to 40% and hypocalcification AI represents 7%.8

Weinmann et al in 1945, initially subdivided amelogenesis imperfecta in hypoplastic and hypocalcified types.9 Following which many classifications evolved with at least 10 subtypes, characterized by clinical features and mode of inheritance. Witkop and Sauk in 1976 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 (hypomaturatin), and also recognized the combined defects (Table 1).3

AI may be inherited in an X-linked manner or as an autosomal dominant or recessive trait.10 However, there are cases where the diagnosis of AI remains tentative in apparently sporadic cases of enamel defects. Ultimately, it is anticipated that molecular genetics tools will allow more precise diagnosis.

The formation of enamel is highly organized and unusual structure is thought to be rigorously controlled in ameloblasts through the interaction of a number of organic matrix molecules that include enamelin (ENAM; 4q21.), amelogenin (AMELX; Xp22.3-p22.1.), ameloblastin (AMBN; 4q21.), tuftelin (TUFT1; 1q21.), amelotin (AMELOTIN 4q13), dentine sialophosphoprotein (DSPP; 4q21.3.), and a variety of enzymes, such as kallikrein 4 (KLK4; 19q13.3–q13.4.) and matrix metalloproteinase 20 (MMP20; 11q22.3–q23.).11

The different patterns of inheritance correspond with different genomic sites. Xp22.3–p22.1 (AMELX, AIH1) is associated with the X-linked form. AmelX null mice have no amelogenin and display ‘distinctly abnormal teeth’ with ‘disorganised, hypoplastic enamel’.12 Another gene on the

Figs 1A to D: (A) Clinical picture, (B) IOPA of the upper incisors, (C) OPG, (D) ground section of the extracted tooth
long arm of the X chromosome at the AIH3 locus must also be involved in enamel formation.\textsuperscript{13} 4q11–q21 (AIH2, ENAM 4q21) is associated with both autosomal dominant and autosomal recessive inheritance patterns and within this 4q13.3 has been identified as being associated with an autosomal recessive inheritance.\textsuperscript{14-16} A transgenic animal model overexpressing ameloblastin has produced an Al-type phenotype.\textsuperscript{17} Wang et al 2004 showed a relationship between the regulation of ameloblast differentiation and components of the bone morphogenetic protein (BMP) pathway.\textsuperscript{18} This suggests that there is at least the potential for mutations in this pathway to account for some cases of amelogenesis imperfecta.\textsuperscript{18} So far no mutation in the amelotin gene has been related to amelogenesis imperfecta.\textsuperscript{19}

Amelogenesis imperfecta is sometimes associated with syndromes like, amelogenesis imperfecta with taurodontism, trichodentoosseous syndrome, AI with nephrocalcinosis and cone-rod dystrophy with AI. The commonest differential diagnosis which should be kept in mind during the clinical assessment is dental fluorosis. The variability of this condition, from mild white ‘flecking’ of the enamel to profoundly dense white coloration with random, disfiguring areas of staining and hypoplasia, requires careful questioning to distinguish from AI. Fluorosis may present with areas of horizontal white banding corresponding to

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Figs 2A to D: (A and B) Clinical photo and OPG of the first twin sister, (C and D) clinical photo and OPG of the second twin sister
periods of more intense fluoride intake and may show the premolars or second permanent molars to be spared (chronological distribution). In the latter case, the history will often reveal excessive fluoride intake either in terms of a habit, such as eating toothpaste in childhood, or related to a local water supply.²¹

Histologically, a ground section of the involved showed very thin enamel, composed of lamillations of irregularly arranged enamel prisms.²¹ The treatment of these patients is usually done in two phases, temporary phase followed by transitory phase.²² In infancy, the primary dentition is protected by the use of preformed metal crowns on posterior teeth. Either polycarbonate crowns or composite restorations are used on anterior teeth.²³ The eruption of the permanent dentition, beginning at 6 years of age, presents a particularly difficult period. Some of the forms of AI present with hypersensitive teeth or with teeth that crumble, and both presentations provide a very real disincentive to good oral hygiene and are very difficult to restore. In this stage crown restorations along with orthodontic correction of the malaligned teeth and if required surgical correction of anterior open bite which is seen in many AI patients should be carried out.

CONCLUSION

The dentist has to diagnose AI as early as possible to offer early management and balance the decision for early intervention and long-term survival of the restorations. Dental practitioners should consider the social implications for these patients and intervene to relieve their suffering. Thus, this article is an attempt to improve the clinician’s knowledge about the clinical and radiological diagnosis of AI as well as its intervention required for such a condition.

REFERENCES


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