AMELOGENESIS IMPERFECTA – A CASE REPORT

Dental Science

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ABSTRACT

Amelogenesis imperfecta (AI) is a disorder group of hereditary development that affects the dental enamel structure which is marked by clinical alterations without association with systemic abnormalities and diseases. The AI trait can be transmitted by either autosomal dominant, autosomal recessive, or X-linked modes of inheritance. Genes implicated in autosomal forms are genes encoding enamel matrix proteins, namely: enamelin and ameloblastin, tuftelin, MMP-20 and kallikrein – 4. Here we present a case of 5 year old girl having amelogenesis imperfecta affecting the deciduous dentition which was diagnosed on the basis of classical clinical and radiographic features.

INTRODUCTION

Amelogenesis imperfecta (AI) is a clinical and genetical condition, which comprises developmental disorders that demonstrate alterations in the enamel. It represents a group of inherited disorders, which are clinically heterogeneous and exhibit tooth enamel defects in the absence of systemic manifestations. Both primary and permanent dentitions are affected[1]. This is entirely ectodermal, since mesodermal component of the teeth are basically normal[2] but the affected teeth are more resistant to dental caries.[3] The characteristic of amelogenesis imperfecta can be transmitted by either autosomal dominant, autosomal recessive or X-linked mode of inheritance.[2] The prevalence of this condition has been expected to range from 1 in 718 to 1 in 14,000, depending on the population studied.[4]

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.[5] Hypoplastic AI represents 60 – 73% of all cases, hypomaturation AI represents 20 – 40%, and hypocalcification AI represents 7%[4].

This clinical report describes the sequenced full mouth of a 5yr old patient with hypoplastic amelogenesis imperfecta along with a complete review that we diagnosed on the basis of clinical and radiographic features.

CASE REPORT

A 5-year old girl presented with the chief complaint of discolouration of the tooth and sensitivity since 1 year. Detailed medical and dental history was recorded from the parents of the patient. Dental family history revealed the presence of similar type of problem in their relatives. Medical history was non-contributory. Extra oral examination did not reveal any relevant findings. The hair, skin and nails of the patient were normal. There was no systemic problem.

On intraoral examination she had a normal complement of deciduous dentition. The thickness of enamel was reduced and was completely chipped off from some teeth exposing the dentin exhibiting a yellowish brown discoloration as shown in Fig.1. The surfaces of the teeth were smooth, and the enamel was either not visible or very thin over the crowns of all the teeth. Fig.2. The dentin, where it was exposed, was brown. The emergence pattern and timing of teeth seemed to be within the normal range. The teeth were vital, firm, and not tender to percussion.

An intraoral periapical radiographs revealed the presence of permanent tooth buds with generalized thinning of enamel on all tooth surfaces and even enamel was absent at certain areas. (Fig.3) On the basis of clinical and radiographic features, final diagnosis of hypoplastic Amelogenesis Imperfecta was made.

DISCUSSION

Amelogenesis imperfecta (AI) is a term for a clinically and genetically heterogeneous group of conditions that affect the dental enamel, occasionally in conjunction with other dental, oral and extra oral tissues.[2] It is a hereditary disorder with clinical impact on both deciduous and permanent teeth and first described in 1890.[6]

In primary dentition the development of enamel starts at seventh week of gestation involving formation and secretion of an organic matrix containing the proteins amelogenin and enamelin.[7] This is pursued by mineralisation of the matrix at nine weeks and subsequent maturation of this enamel with change in enamel protein predominantly from amelogenin to enamelin.[7]

Amelogenesis imperfecta, the hereditary defect of enamel, occurs as a result of disturbance beginning at the organic matrix stage and continuing through calcification.[8] The development of enamel is a multistep process, so the problem in any one of the steps may give rise to this defect. In general, the development of enamel can be divided into three major stages – elaboration of the organic matrix, mineralisation of the matrix, maturation of the enamel[9].

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[10]. Table-1

TYPE I	Hypoplastic
IA	Hypoplastic, pitted autosomal dominant
IB	Hypoplastic, local autosomal dominant
IC	Hypoplastic, pitted autosomal recessive
ID	Hypoplastic, smooth autosomal dominant
IE	Hypoplastic, smooth X-linked dominant
IF	Hypoplastic, rough autosomal dominant
IG	Enamel agenesis, autosomal recessive
TYPE II	Hypomaturation
IIA	Hypomaturation, pigmented autosomal recessive
IIB	Hypomaturation
IIC	Snow capped teeth, X-linked
IID	Autosomal dominant?
TYPE III	Hypocalcification

IIA	Autosomal dominant
IIB	Autosomal recessive
TYPE IV	**Hypomaturation-hypoplastic with taurodontism, autosomal dominant**
IVA	Hypomaturation-hypoplastic with taurodontism, autosomal dominant
IVB	Hypoplastic-hypomaturation with taurodontism, autosomal dominant

Clinical features of patients with AI depend on the type which is involved. AI has been classified on the basis of clinical, radiographic, and histologic appearance of the enamel defect and the mode of inheritance of the trait.[10] AI has been categorized as hypoplastic (autosomal dominant/autosomal recessive/x-linked dominant) the enamel is well mineralized but its amount is reduced and clinically, grooves and pits will be realized on the surface of the fine enamel, hypocalcified (autosomal dominant/autosomal recessive) shows pigmented, softened, and easily detachable enamel, hypomaturation types (autosomal recessive/x-linked recessive/autosomal dominant) and hypoplastic-hypomaturation type the affected teeth exhibit mottled, opaque white-brown or yellow discoloured enamel, which is softer than the normal.[10,11]

A variety of symptoms can be presented with AI. The most substantial findings comprise extensive loss of tooth tissue, tooth sensitivity, excessive attrition leading to short clinical crowns, spacing in the anterior region of the dentition, normal or tight proximal contacts in the posterior region, and a general enamel caries resistance.[11]

Literatures have investigated that mutations in five genes have been associated with amelogenesis imperfecta. Each gene can be mutated in a variety of ways, often creating diverse and distinct phenotypic patterns.[8] Mutations in the amelogenin gene (AMELX) cause X-linked amelogenesis imperfecta, while mutations in the enamelin gene (ENAM) cause autosomal-inherited forms of amelogenesis imperfecta.[12] Some reports involve that kallikrein-4 (KLK4) & MMP-20 cause the mutation of this gene which has been associated with the autosomal recessive, pigmented hypomaturation variant of amelogenesis imperfecta. and DLX3 genes is in a group of genes that code for a number of proteins that are critical for craniofacial, tooth, hair, brain and neural development, mutation of this gene is associated with the hypoplastic-hypomaturation variants of amelogenesis imperfecta with taurodontism.[8]

AI may be inherited in an X-linked manner or as an autosomal dominant or recessive trait. However, there are cases where the diagnosis of AI remains tentative in apparently sporadic cases of enamel defects. Ultimately, it is anticipated that molecular genetic tools will allow more precise diagnosis.[2]

Treatment planning for patients with amelogenesis imperfecta is related to the age, socioeconomic factors, the type, and severity of the disorder and intraoral structures. An interdisciplinary approach may be required to evaluate, diagnose, and resolve the esthetic problem.[13] The unbeaten management of AI during childhood requires the cooperation, preventive counselling, emotional support and motivation of the patient and parents.

Figure 1. – Multiple Brownish discoloration of primary dentition

Figure 2. The incisal edges of the anterior teeth are worn and chipped along with the attrition of the molar occlusal surfaces showing the thinness of the enamel.
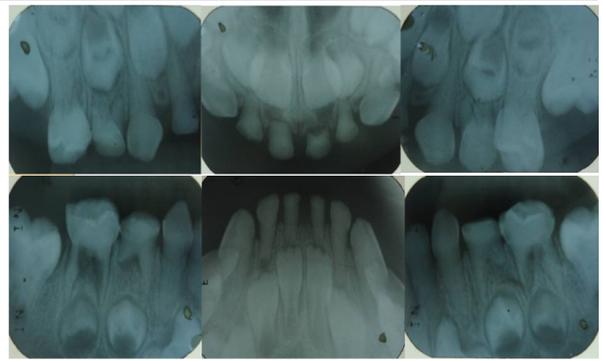

Figure 3. – Full mouth periapical radiographs of Maxilla & Mandible showing the thin of enamel over the tooth surfaces. The radiodensity of dentin on some surfaces is not differentiated from enamel.

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