

Amelogenesis Imperfecta type IV: A Challenge to Esthetics & Function

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ABSTRACT

Amelogenesis Imperfecta (AI) comprises a group of developmental conditions, genetic 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. It is usually inherited either as an X-linked, autosomal dominant or autosomal recessive trait. The enamel may be hypoplastic, hypomineralised or both and affected teeth may be discolored, sensitive or prone to disintegration. Diagnosis is made from the family history, pedigree plotting, and accurate clinical observation. The treatment of patients with AI must begin with early diagnosis and intervention to prevent compromised esthetics and occlusion disharmony. Herein, we present a case report of hypoplastic amelogenesis imperfecta with taurodontism and a brief review of this clinical entity.

KEYWORDS: Amelogenesis Imperfecta, Taurodontism, Panoramic Radiograph

INTRODUCTION

Amelogenesis imperfecta (AI) is a clinical entity and heterogeneous group of conditions that affect the enamel; occasionally in conjunction with other dental, oral and extraoral tissues.¹ AI is classified as: hypoplastic, hypocalcified, and hypomaturational forms. This genetic disorder is apparently associated with the malfunction of the enamel building proteins such as ameloblastin, enamelin, tuftelin, and amelogenin. Due to mutations occurring in the genes encoding these proteins, the affected teeth show disorganized enamel matrix resulting in hypoplastic/hypocalcified enamel.²

Clinical manifestations associated with AI include quantitative and qualitative enamel defects, pulpal calcification, taurodontism and root deformities, impaction of permanent teeth, root and crown resorption, congenitally missing teeth and open bite occlusion.³ Other manifestations include failure of multiple permanent teeth to erupt into the oral cavity.⁴ AI seems six times more susceptible to have impaction of permanent teeth and associated anomalies such as follicular cysts than unaffected persons.⁵ Radiographic examination plays a significant role in the identification and diagnosis of such underlying changes.

We present a case emphasizing and diagnosing the clinical conditions and the need for management of this entity.

CASE REPORT

A 20 - year- old male patient reported to the Department

of Oral Medicine and Radiology with a complaint of yellowish discoloration of his teeth which was noticed 10 years ago. He gave no history of adverse or deleterious habits. He resided in a fluoridated area since his birth and had consumed bore well water since childhood. He noticed discolouration only on his permanent teeth and his milk teeth was unaffected. He is the first born child of biologically unrelated parents and has noticed similar discolouration of teeth in his younger brother also. The pedigree chart was constructed for a better understanding of his inheritance as in Figure 1. This chart depicts that his parents had a non consanguineous marriage where none of his grandparents or his parents' siblings were affected. Only his younger brother was affected.

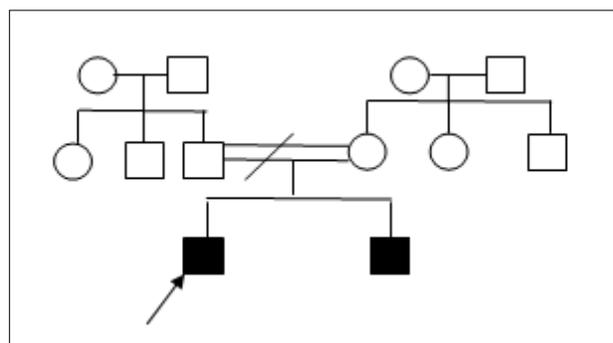


Fig 1: Pedigree chart for the case

On general physical examination, no significant changes were detected with regard to the patient's skin, hair, and nails. Intraoral examination revealed generalized yellowish brown discoloration on the buccal and lingual surfaces of maxillary and mandibular posterior teeth with

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decreased crown height. (Fig 2- Fig 4) There was also generalized diffuse chalky white appearance seen on the middle thirds of the crown in relation to 12, 11, 21, 22, 32, 42 and on cervical thirds of the crown in relation to 31 & 41. There was no chipping of enamel noted on probing.



Fig 2: Teeth in occlusion



Fig 3: Maxillary teeth with spacing seen



Fig 4: Mandibular teeth with spacing seen

On routine examination, Occlusal caries in 17, 27, 47 was noted. Angle's Class III molar relation with crossbite in the right posterior teeth and maxillary and mandibular spacing were present. (Fig 5) A provisional diagnosis of enamel hypoplasia secondary to dental fluorosis was given. Hypomaturational AI, Dentinogenesis imperfecta and Type II dentin dysplasia were considered in the differential diagnosis.



Fig 5: Teeth in occlusion

Dentinogenesis imperfecta clinically presents as yellowish brown discoloration often with distinct translucence and enamel tends to crack away from the defective dentin. Severe attrition may also be noted. Similarly, in dentin dysplasia Type II teeth demonstrate blue to amber to brown translucence. So, the differential diagnosis was drawn based on the similar clinically presented discoloration that affects the teeth seen in both the conditions.

Panoramic radiograph revealed generalized loss of enamel with increased radiodensity in dentin and widened pulp chambers. 28 was vertically impacted. There was also apically positioned pulpal floor and bifurcation of roots close to the apex in 28 and also in 16, 17, 18, 26, 27, 36, 37, 38, 46, 47 and 48 suggestive of taurodontism. There was shortening in length of distal roots of 36 and 46. (Fig 6)



Fig 6: Digital panoramic radiograph with taurodontism seen on the maxillary and mandibular posteriors

Dentinogenesis imperfecta was excluded from the diagnosis as radiographically the teeth will have normal enamel thickness with thin dentin and enlarged pulp chambers with cervical constriction which were not clearly present in the current case. Similarly, type II Dentin Dysplasia was also excluded as radiographically bulbous crowns, cervical constriction with thistle tube shaped pulp chambers will be noted. Therefore on the basis of history, clinical findings and radiographic findings, a final diagnosis of AI Type IV A (Hypomaturational- Hypoplastic AI with taurodontism) were made.

DISCUSSION

AI is a developmental condition of the dental enamel that is characterized by hypoplasia and/or hypomineralisation. AI can be autosomal dominant, autosomal recessive, sex-linked and sporadic inheritance patterns. The enamel may be hypoplastic, hypomineralised or both, and teeth affected may be discoloured, sensitive or prone to disintegration either post eruption.

Many classifications of AI have formed since the original division into hypoplastic and hypocalcified types in 1945.⁶ Out of them some were exclusively based on the phenotype (appearance), others have used the phenotype as the primary discriminant and the way of inheritance as a secondary factor in diagnosis.¹ The most commonly used classification was proposed in 1989 by Witkop⁷, and revised by Nusier in 2004. Based on enamel appearance and hypothesized developmental defects, AI is categorized as 4 patterns: Type I hypoplastic, Type II hypomaturation, Type III hypocalcification and Type IV hypomaturation-hypoplastic with taurodontism.

During organogenesis, the enamel transitions from a soft and pliable tissue to its final form, this is almost entirely devoid of protein. Mutation or alteration in any of the following genes encoding specific enamel proteins such as Enamelin gene (ENAM), Amelogenin gene (AMELX), Kallikrein 4 gene (KLK4), Matrix Metalloproteinase 20 gene (MMP20) and Distalless homeobox 3 gene (DLX3) have been linked with AI. (8) The final outcome of enamel is a reflection of the peculiar molecular and cellular activities that take place during its genesis. Deflection from this pattern may lead to AI.^{8,9} Studies showed that AI is family linked and can be inherited as autosomal dominant, autosomal recessive, or X linked dominant, and X linked recessive.^{8,9}

Despite both deciduous and permanent dentition are involved in AI, it is affected more commonly in permanent teeth than primary teeth and more routinely in incisor teeth and first molars in both upper and lower jaws.

Witkop⁷ and Witkop and Sauk¹¹ stated that while some kindreds exhibiting taurodontism were associated with amelogenesis imperfecta or the Tricho Dentosseous syndrome indicated as an autosomal dominant mode of inheritance or as an isolated trait. Ackerman et al.¹² stated that the morphology of the roots of teeth was primarily determined genetically but that it may be modified environmentally. He also stated the pathogenesis of taurodont formation included as an unusual developmental pattern; a delay in calcification of the pulp chamber floor; an odontoblastic deficiency; a change in Hertwig's epithelial root sheath, with an apparent failure of the epithelial diaphragm to invaginate at the normal horizontal level; and a late or incomplete union of the horizontal flaps of the epithelial diaphragm.

In patients with hypoplastic amelogenesis imperfecta, the basic alteration centers on an inadequate deposition of

enamel matrix. The *amelogenin* gene is a tooth-specific gene expressed in pre-ameloblasts, ameloblasts, and in the epithelial root sheath remnants. The phenotype in hypoplastic is expressed by thin enamel and spacing between the teeth, or in some pedigrees by rough, irregular or randomly pitted enamel. If the prime defect is in the amount of enamel matrix produced, the enamel will be hard, normally translucent and not subject to significant attrition. The buccal surfaces of the teeth are affected more severely, and the pits may be arranged in rows or columns. Staining of the pits may occur. The enamel between the pits is of normal thickness, hardness, and coloration. Radiographically, the teeth exhibit a thin peripheral outline of radiopaque enamel. Often, unerupted teeth exhibiting resorption are seen.⁸

In hypomaturation amelogenesis imperfect affected patients, the enamel matrix is laid down and begins to mineralize. There is also a defect in the maturation of the enamel's crystal structure. The *enamelin* (*ENAM*) gene is a tooth-specific gene expressed chiefly by the enamel organ, and, at a low level, in odontoblasts. The human *ENAM* gene is located on chromosome 4 (4q13.3). The *ENAM* gene was therefore supposedly a candidate gene for autosomal dominant type of AI. Mutation in the matrix metalloproteinase 20 gene (*MMP-20*) in the region 11q22.3 – q23, has been described as being associated with the autosomal recessive pigmented hypomaturation amelogenesis imperfecta. Though affected teeth are normal in shape, they exhibit a mottled opaque white-brown yellow discoloration. The enamel often fractures from the underlying dentin and is soft enough to be penetrated by a dental explorer. Anterior open bite and unerupted teeth exhibiting resorption are uncommon. Radiographically, the affected enamel exhibits a radiodensity that is similar to dentin.⁹

In case of hypocalcified amelogenesis imperfecta, the enamel matrix is laid down appropriately but no significant mineralization occurs. However, *AMELX*, *AMBN*, *ENAM*, *KLK4*, and *MMP-20* were excluded from a causative role in two families with autosomal dominant hypocalcified AI, suggesting that this type of AI is caused by the mutation of a gene that is either not known or not considered to be a major contributor to enamel formation. This causes the teeth that are appropriately shaped on the eruption, to be very soft and easily lost. On eruption, the enamel is yellow brown or orange, but it often becomes stained brown to black and exhibits rapid calculus deposition. With years of function, much of the coronal enamel is removed, except for the cervical portion that is occasionally calcified better. Unerupted teeth and anterior open bite are also associated with this entity. Radiographically, the density of the enamel and dentin are similar. Before eruption the teeth are normal in shape after a period of function much of the cuspal enamel is lost, with the occlusal surface becoming the most irregular.⁸ In the present case, this clinical type was also considered as a differential diagnosis, as clinically the patient exhibited generalized yellowish brown discoloration with decreased vertical height.

In the hypomaturational-hypoplastic pattern, the predominant defect is one of enamel hypomaturational in which the enamel appears as mottled yellowish white to yellow-brown. Enamel pits are seen frequently on the buccal surfaces of the teeth. Mutation within the human *DLX3* gene homeodomain is associated with this type of AI. The *DLX3* gene is a member of the family of homeobox genes that are homologous to the *distalless* (*Dll*) gene of *Drosophila*, known to be expressed while development of the chondrocranium, dermatocranium, sensory organs, brain, limbs, and appendages, and in the processes of osteogenesis and hematopoiesis. Radiographically, the enamel appears similar to dentin in density, and large pulp chambers may be seen in single rooted teeth in addition to varying degrees of taurodontism.^{8,9} On diagnosing taurodontism in the maxillary and mandibular posterior teeth radiographically with a clinical presentation similar to that of hypomaturational-hypoplastic pattern in our current case, a final diagnosis of Amelogenesis Imperfecta Type IV A was considered.

Diagnosis should be drawn based on the family history, pedigree plotting, meticulous clinical and radiological observation. Dental radiographs of AI teeth provide vital information to the clinician related to the degree of enamel mineralization to design an appropriate treatment plan.¹⁰ Evaluation of enamel density changes in AI teeth where the enamel has a radiopacity similar to or less than that of dentin that is considered mineral deficient.⁵ In the present case, the panoramic radiograph was taken to compare the changes in radiodensity of enamel and dentin. In the process, it also revealed taurodontism on the maxillary and mandibular posterior which helped in categorizing the type of AI where taurodontism is exhibited thus helping to arrive at a final diagnosis of AI.

Treatment planning for patients with AI depends on many factors: the type and severity of the disorder, the intraoral situation at the time the treatment is decided and the age and socioeconomic status of the patient. Treatment should be comprehensive and should include removal of surface stains, reducing sensitivity by providing topical 1.1% neutral sodium fluoride toothpaste twice a day. Every 3 months, oral hygiene recalls followed by topical 5% sodium fluoride varnish applications should be performed. Maintenance in the vertical dimension of occlusion should be attained to provide an adequate interocclusal space. As for the esthetics, adhesive techniques/overdentures/ porcelain fused to metal crowns/ fixed partial dentures/ full porcelain crowns/ inlay/ onlay prostheses are used for the prosthetic treatment of the patients. Meanwhile, full porcelain restorations are becoming increasingly popular, because of their esthetics, excellent biocompatibility, and improved physical properties.^{11,12}

Full porcelain restorations are much - preferred treatment modality for the patients with AI.¹³ Due to the advances in the field of esthetic dentistry, especially in bonding to dentin, these help practitioners to restore function and esthetics to an acceptable level. In the current case upon

meticulous evaluation of the clinical and radiographic features, dentin bonded resin composite restorations for the anterior teeth with full crown veneers for the posterior teeth were preferred to reassure the mechanical durability, recover esthetics and protect the residual dentin.

CONCLUSION

AI is a serious problem that can result in reduced oral health-related quality of life and cause some physiological problems. In the most severely affected patients, teeth can present alterations in enamel thickness, color, and shape, all which compromise aesthetic appearance and masticatory function. From this point of view, people with AI need extensive treatment. The dental rehabilitation of a young patient must be done with regard to the growing potential of the jaws and the periodontal health. Based upon the complexity of this condition and considering the complications encountered during treatment, we can conclude that, with proper isolation and careful handling, direct-bonded resin composite restorations provide an excellent conservative transitional treatment for protection of AI-weakened teeth. The resin composites used for restoring the teeth have excellent color stability and no marginal discolorations.

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