

Amelogenesis Imperfecta – A Case Report

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Abstract: Amelogenesis Imperfecta is a developmental disorder of genomic origin associated with structural enamel formation. Although AI is considered as a single disease entity, it actually represents a group of heterogenous conditions, with diverse structural defects of enamel resulting in a range of clinical phenotypes. It is characterized by clinical and genetic heterogeneity in the absence of systemic abnormalities or diseases. This case report presents with a case of AI as an example of a hereditary disorder.

Key Words: Amelogenesis Imperfecta, Dental anomaly, Yellowish discoloration

I. Introduction

Amelogenesis Imperfecta is a term for a clinically and genetically heterogenous group of conditions that affect the dental enamel, occasionally in conjunction with other dental, oral and extraoral tissues. AI is typically characterized by generalized enamel defects in both primary and permanent dentition. It might also be associated with morphological or biochemical changes elsewhere in the body. ^[1]

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%. 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. The inheritance pattern of X-linked disorders dictates that male to male transmission cannot occur. Conversely all female offsprings of an affected male must be affected. Affected females have a 50% probability of passing on the trait to the offspring of either sex. ^[2] Many classifications of AI have evolved since the original division into hypoplastic and hypocalcified types in 1945. Some have been exclusively based on the phenotype, others have used the phenotype as the primary discriminant and the mode of inheritance as a secondary factor in diagnosis. ^[3]

II. Case Report

A 12 year old female patient reported with the complaint of yellowish discolored teeth for the past 5 years.



Figure 1: facial view

Patient had positive behavior assessment with a non-contributory medical history. Familial history revealed consanguineous marriage.



Figure 2: discolored upper anteriors

Extraorally patient had straight profile, no facial asymmetry, competent lips and midfacial hypoplasia. On clinical examination patient had normal soft tissue findings. Patient had permanent dentition with yellowish discoloration in upper anteriors, clinically missing 36, grossly decayed 46 and generalized attrition. Panoramic radiograph showed the presence of a thin layer of enamel with radiodensity of enamel more than dentin.



Figure 3: OPG shows thin layer of enamel with radiodensity



Figure 4: composite restoration done in relation to 12, 13, 22 & 23

On the basis of clinical and radiographic features, final diagnosis of hypoplastic AI was confirmed. As a treatment protocol, composite restoration in 12, 13, 22 and 23. Root canal treatment in 46 with Guttapercha followed by stainless steel crown were done. Parent and patient were counselled and review done periodically.

III. Discussion

Amelogenesis Imperfecta 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.^[4] AI is a developmental condition of the dental enamel that shows autosomal dominant, autosomal recessive, sex-linked and sporadic inheritance patterns, as well as sporadic cases.^[5]

Diagnosis involves exclusion of extrinsic environmental or other factors, established of a likely inheritance pattern, recognition of phenotype and correlation with the dates of tooth formation to exclude a chronological developmental disturbance.^[6] Dental enamel is a highly mineralized tissue with over 95% of its volume occupied by unusually large, highly organized hydroxyapatite crystals. The formation of this 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, amelogenin, ameloblastin, tuftsin, amelotin, dentine sialophosphoprotein and a variety of enzymes such as Kallikrein4 and matrix metalloproteinase 20.^[7]

AI may be inherited in a 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.

AI affects the enamel of all the teeth of the affected individuals within kindred, in a more or less equal manner, without reference to chronology, occasionally in association with other generalized conditions.^[8]

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 a short clinical crown, spacing in the anterior region of the dentition, normal or light proximal contacts in the posterior region and a general enamel caries resistance. Abnormal tooth eruption, morbid root and coronal resorption, congenitally missing teeth, malocclusion, anterior open bites, pulpal calcification, dentin dysplasia, hypercementosis, root malformation and taurodontism have been ordinarily repeated. It should not be omitted from quoting the surface irregularities and the crown discoloration mainly of yellowish brown shade.^[9]

There is a growing acceptance that a classification of enamel defects based primarily or exclusively on phenotype is problematical. For this reason the mode of inheritance and underlying genomic change are more important discriminators. This is particularly so in relation to genetic counselling of affected individuals and their families. The supportive clinical case needed by these individuals is substantial both in terms of clinical and emotional demands.^[10]

Treatment is an ever based on the principles of prevention before intervention. The progression of treatment during childhood has been described as a temporary phase followed by a transitory phase. In infancy, the primary dentition is protected by the use of preferred metal crowns on posterior teeth. Either polycarbonate crown or composite restorations are used on anterior teeth.^[11] The eruption of permanent dentition beginning at six 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 presentation provide a very real disincentive to good oral hygiene and are very difficult to restore.

Children with AI are not without malocclusion and the anterior open bite seen in some cases of AI requires consideration of surgical as well as restorative management.^[12]

The management of AI is often complex, takes a significant amount of tissue (more commonly from childhood to early adulthood) but its positive psychological effect on a wounded self-esteem is priceless, thus replacing the counselling therapies that could be otherwise needed in addition to the dental approach.^[13]

IV. Conclusion

Diagnosis and treatment of AI patients require lengthy, comprehensive and multidisciplinary approach which should aim to successfully address all dental, occlusal, developmental, skeletal and soft tissue problem. AI presents with problem of socialization, function and discomfort, which may be managed by early vigorous intervention, both preventively and restoratively. It is highly significant that a strict recall system and a good oral hygiene protocol are applied.

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