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Amelogenesis Imperfecta: A case report

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Abstract

Amelogenesis Imperfecta(AI) represents structural developmental defect of tooth enamel having complex inheritance pattern. It represents a group of heterogenous conditions. AI has several names such as hereditary enamel dysplasia, hereditary brown enamel, hereditary brown opalescent teeth. In this disorder, the enamel is hypoplastic, hypomineralized or both. It may show autosomal dominant, autosomal recessive, sex-linked or sporadic pattern. Here, we report three cases among six children of the same family with Amologenesis imperfecta, analyse the clinical presentation, diagnostic features and clinical complications of Amelogenesis imperfecta.

Keywords: Amelogenesis imperfecta, Discoloration, Hypoplastic, Hypomaturative

Introduction

Amelogenesis Imperfecta, a group of hereditary diseases affecting tooth enamel in either quality or quantity.(1)

Dental enamel, a highly mineralized tissue has over 95% of its volume being occupied by unusually large, highly organized, hydroxyapatite crystals.(2) The formation of enamel has been highly organized, and unusual structure is thought to be rigorously controlled in ameloblasts. This has been through the interaction of a number of organic matrix molecules.(3)

The enamel transitions from a soft and pliable tissue to its final form, during organogenesis, which is almost entirely devoid of protein.(4) The final composition of enamel is a reflection of the unique molecular and cellular activities that take place during its genesis. Deviation from this pattern may lead to amelogenesis imperfecta.

Improper differentiation of ameloblasts in AI causes poor development or complete absence of enamel of the teeth. Both the primary and permanent dentition may be affected. AI encompasses a group of hereditary diseases that involve the defective formation or calcification of enamel. The occurrence of enamel defects without any sign of generalized or systemic defects is the pathognomic feature of AI. Teeth exhibit yellow to dark brown discoloration. The teeth exhibit pits and grooves and in some cases enamel may be completely absent or cheesy to hard.

Non-enamel anomalies such as delayed eruption, crown resorption, congenitally missing teeth, pulpal calcifications, dental follicular hamartomas, and gingival hyperplasia had been found to be associated with AI. Exclusion of extrinsic environmental or other factors, the establishment of a likely inheritance pattern, and recognition of phenotype and correlation with the dates of tooth formation to exclude a chronological developmental disturbance involves diagnosis.

The enamel may be hypoplastic, hypo mineralized or both and teeth affected may be discolored, sensitive or prone to disintegration either post-eruption or preeruption.(2) It may be associated with morphologic or biochemical changes elsewhere in the body.(5)

Hypoplastic AI represents 60-73% of all cases; hypomaturation AI represents 20-40%, and hypo-calcification AI represents 7%.(6) The most widely accepted classification is that proposed by Witkop and Sank in 1976. Witkop and Rao, (1971) classified AI broadly based on phenotype and style of inheritance(7) into three categories: Hypoplastic variety, hypocalcified variety, and hypo maturation variety. Aldred and Crawford in 1995.(8) classified based on a molecular defect, biochemical result, mode of inheritance and phenotype.

We present here case report of AI (hypo maturative and hypoplastic) along with a complete review that we diagnosed on the basis of clinical and radiographic features.

Case Report

A 46years, Mohd. Ashraf resident of Bhagwa near Doda, presented his 3 among 6 children to Department of Oral Medicine and Radiology with the chief complaint of yellowish discolouration and decaying teeth. Children presented to the department were namely Amna aged 20yrs, Hilma aged 18yrs, Fatima aged 14yrs, Nazia aged12 yrs, Mohd. Toyub aged 10yrs, Ashiq Ali aged 8yrs. Out of them Hilma, Mohd. Toyub, Ashiq Ali had complain of yellowish discoloration of teeth.

The children's parents did not seek any treatment previously, thinking that since the condition was not resulting in any other systemic manifestations. It was only now, when they realized that the girl Hilma, must seek a dentist for esthetics being an important part in her growing age.

On enquiry, it was revealed that deciduous teeth were also similarly discoloured, and, younger brothers Mohd Toyub & Ashiq Ali, also suffered from the same condition.

Past medical history was non-contributory. Family history except siblings was unremarkable.

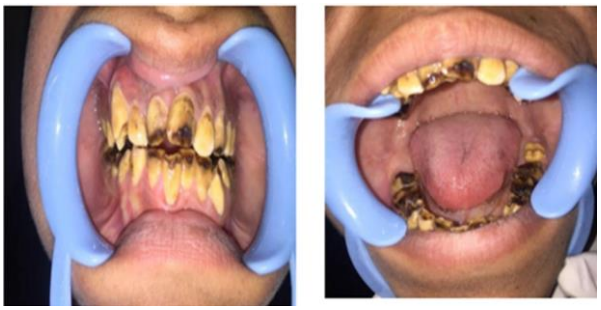

Fig. 1: Generalized yellowish brown discoloration with chipped out enamel surface

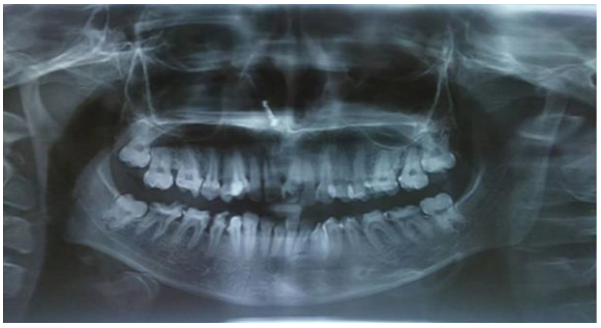

Fig. 2: Orthopantomogram showing thin layer of enamel over tooth surfaces

Clinical Examination: Intraoral examination showed Hilma had permanent dentition with no missing teeth. All the anterior and posterior teeth were affected with yellowish brownish discoloration with attrition. The thickness of enamel was reduced on the teeth and was completely chipped off from some teeth exposing the dentin. The surfaces of the teeth were rough with diffuse pitting present on the exposed tooth surfaces, more prominent on the labial and buccal aspects. The emergence pattern and timing of teeth seemed to be within the normal range. Extra-oral examination did not reveal any relevant findings. On palpation, by probing resistance was felt and tooth material is soft in consistency with mild flaking of residual enamel.

Radiographic imaging: OPG showed generalized thinning of enamel on all tooth surfaces and enamel was even absent in certain areas with normal pulp chamber and root morphology. Grossly decayed 47, 45 and proximal carious 46,36 present. Radiolucency seen on the proximal aspect of 35, 36, 45, 46, 47 suggestive of dentinal caries.

As per the patient's history and clinical findings, a provisional diagnosis of hypoplastic, rough autosomal dominant AI was proposed along with a differential diagnosis of environmental enamel hypoplasia, dentinogenesis imperfecta, dentin dysplasia.

Treatment: Esthetics along with functional limitations was the reason, patient's parents brought her to the hospital for treatment**.** The patient was referred to the department of Conservative dentistry and Endodontics for restoration of esthetics and function of affected teeth.

Discussion

AI represents a group of conditions and is genomic in origin. AI affects the structure and clinical appearance of the enamel of all or nearly all the teeth in a more or less equal manner. AI is a developmental condition of the enamel that is characterized by hypoplasia or hypomineralization. AI shows autosomal dominant, autosomal recessive, sex-linked, and sporadic inheritance patterns, as well as sporadic cases.[6]

Classification: In general AI has been classified as Hypoplastic, Hypocalfied, Hypomaturtion, Hypoplastic-hypomaturion type. There are numerous classification systems and the most widely accepted is proposed by Witkop and Sank in 1976, which considers the inheritance pattern of the disorder, as well as its specific clinical characteristics.

Aldred and Crawford, 1995[10] Classification based on: (a) Molecular defect (when known) (b) Biochemical result (when known) (c) Mode of inheritance (d) Phenotype

Witkop and Rao, (1971) classified based on phenotype and style of inheritance.[7]

Three broad categories: Hypoplastic, hypocalcified, hypomaturation.

a. Hypoplastic
- Autosomal dominant hypoplastic hypomaturation with taurodontism
- (subdivided into a and b according to author)
- Autosomal dominant smooth hypoplastic with eruption defect and resorption of teeth
- Autosomal dominant rough hypoplastic
- Autosomal dominant pitted hypoplastic
- Autosomal dominant local hypoplastic
- X-linked dominant rough hypoplastic

b. Hypocalcified
- Autosomal dominant hypocalcified

c. Hypomaturation
- X-linked recessive hypo maturation
- Autosomal recessive pigmented hypo maturation
- Autosomal dominant snow-capped teeth
- White hypomature spots?

Clinical Features

Type I/hypoplastic AI: Hypoplastic form of AI is characterized by thin enamel with yellowish-brown

color, rough or smooth and glossy, square-shaped crown, lack of contact between adjacent teeth, flat occlusal surfaces of posterior teeth due to attrition, and with/without grooves and/pitting. Radiographically, in hypoplastic type, there is a presence of thin radiopaque layer of enamel with normal radiodensity. Histologically, in hypoplastic type, defect is in enamel matrix formation.[5] In about 50% of cases, the anterior open bite is noticed as a result of a decreased crown height.[2,6,9]

Type II/hypomaturation AI: Hypomaturation form of AI is characterized by normal thickness of enamel but softer than normal but harder than hypocalcified type and may crack away from the crown, mottled-colored cloudy white/yellow/brown/snow capped. Radiographically, radiodensity of enamel is similar to that of dentin. Histologically, in hypomaturation type, alterations in enamel rod and rod sheath structures had been noted in various studies.

AI may be allied with some other dental and skeletal developmental defects or abnormalities. They are crown and root resorption, attrition, taurodontism, delayed eruption, and tooth impaction, dens in dente, pulp stones, anterior open bite, and agenesis of teeth.[6]

AI is sometimes associated with syndromes such as AI with taurodontism, tricho-dento osseous syndrome, AI with nephrocalcinosis, and cone-rod dystrophy with AI.[10]

Type III/hypocalcified AI: This variety of AI appears as opaque white to yellow-brown discoloration with soft and rough enamel surface. Dentin sensitivity and open bite are common, as well as heavy calculus formation. Hypocalcified form of AI is the most common type and is characterized by normal size and shape of crown, softer enamel which wears down rapidly and can be removed by a prophylaxis instrument, and become pigmented-dark brown colored. Radiographically, in hypocalcified form, thickness of enamel is normal but radiodensity of enamel is less than that of dentin. Histologically, in hypocalcification type, defects of matrix structure and mineralization is seen.[2,6]

In hypoplastic-hypomaturation with taurodontism, the enamel is thin, mottled yellow to brown, and pitted. Molar teeth exhibit taurodontism and other teeth have enlarged pulp chambers.

Differential Diagnosis: Extrinsic disorders of tooth formation, chronological disorders of tooth formation, and localized disorders of tooth formation should be considered in the differential diagnosis. The differential diagnosis considered most probable is dental fluorosis. The variability of this condition, from mild white "flecking" of enamel to profoundly dense white coloration with random, disfiguring areas of staining and hypoplasia, entails careful interrogation to distinguish from AI. Fluorosis may present with areas of horizontal white banding corresponding to periods of more intense fluoride intake. Premolars or second molars are normally spared (chronological distribution). The history often reveals excessive fluoride intake in terms of a habit, such as eating toothpaste in childhood or related to a local water supply.[2]

Management of Amelogenesis Imperfecta

The 3 principles of management of patients with AI are

1. Alleviation of pain and anxiety
2. Restoration and maintenance of remaining dentition with regard to esthetics.
3. Maintenance restoration of occlusal vertical height
4. Management of young patients with AI is preferably done in 3 phases:

a. Temporary Phase, undertaken during primary or mixed dentition. Posterior teeth are generally restored with composites or polycarbonate crowns.
b. Transitional Phase, when all permanent teeth have erupted this phase continues till adulthood. Anterior teeth in adolescents may be restored using porcelain veneers.
c. Permanent Phase, which lasts through adulthood. Anterior teeth may be restored with porcelain jacket crowns. Anterior open bite may at times require surgical management.

Role of Dentist

The dentist has to diagnose the condition as early as possible to offer early intervention 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 diagnosis as well as intervention required for such a condition.

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. Accurate diagnosis and appreciation of associated clinical problems in each case enable the institution of early preventive measures and management techniques using a multidisciplinary approach.

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