AMELOGENESIS IMPERFECTA IN SIBLINGS – A REPORT OF TWO CASES.

A Case Report

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ABSTRACT

Amelogenesis imperfecta (AI) is a diverse collection of inherited diseases that exhibit quantitative or qualitative tooth enamel defects in the absence of systemic manifestations. This defect is entirely ectodermal, since mesodermal components of the teeth are basically normal. The AI trait can be transmitted by either autosomal dominant, autosomal recessive, or X- linked modes of inheritance with more than one person affected in the family. This article describes amelogenesis imperfecta in two sisters with detailed clinical and radiological findings.

KEYWORDS: Amelogenesis imperfecta, hypoplastic type, familial type.

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INTRODUCTION:

Dental enamel, the 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.[1,9] Dental enamel formation is divided into secretory, transition and maturation stages. 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 cells. 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.[1,9]

The final composition of enamel is a reflection of the unique molecular and cellular activities that take place during its amelogenesis. Deviation from this pattern may lead to amelogenesis imperfecta (AI).[2] AI is also known by varied names such as hereditary enamel dysplasia, hereditary brown enamel, hereditary brown opalescent teeth.[1,11] AI is caused by mutations in genes that control amelogenesis and follows inheritance patterns of autosomal-dominant, autosomal recessive or X-linked modes of transmission.[2,6,8,10,11] Genes implicated in autosomal forms are genes encoding enamel matrix proteins, namely: enamelin and ameloblastin, tuftelin, MMP-20 and kallikrein – 4.[2]

AI results in poor development or complete absence of enamel of the teeth caused due to improper differentiation of ameloblasts. The enamel defects of this condition are clinically divided into hypoplastic, hypocalcified and hypomineralized forms.[3,5] This enamel anomaly may affect both the primary and permanent dentition.[3,6] This article describes amelogenesis imperfecta in two sisters with detailed clinical and radiological findings.

CASE REPORT – 1

A female patient of age 18 belonging to low socio economic status reported to the outpatient department of oral medicine and radiology with chief complaint of yellowish discoloration of teeth since childhood. Family history revealed presence of same type of teeth in her younger sister (Case 2), while one elder sister and two younger brothers were not affected. None of the parents and other family members from maternal and paternal side had the history of discoloration of teeth. Her primary dentition was also discolored. Patient did not have disturbance in eruption of any tooth. Medical history was non contributory.

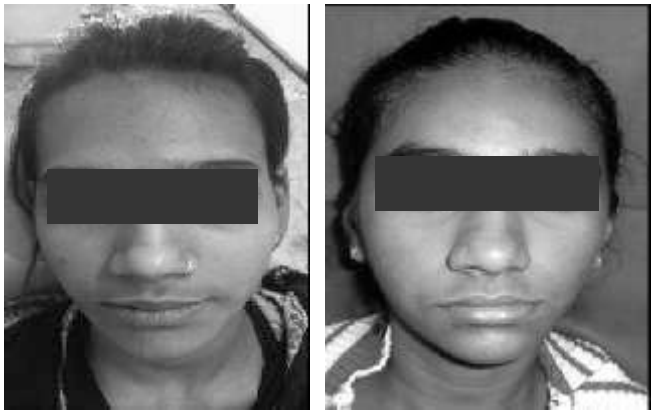

Figure1: Profile photographs patients showing bilateral facial symmetry in Case 1 & Case 2

Extraoral examination **(Figure 1)** did not reveal any

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relevant findings. On intraoral examination, all teeth were present **(Figure 2)** except 46 which had been extracted before 2 months due to caries.

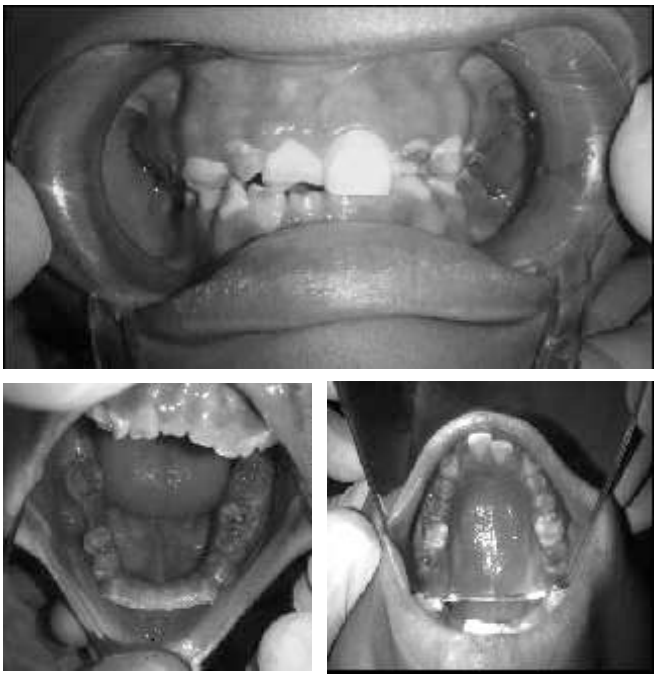
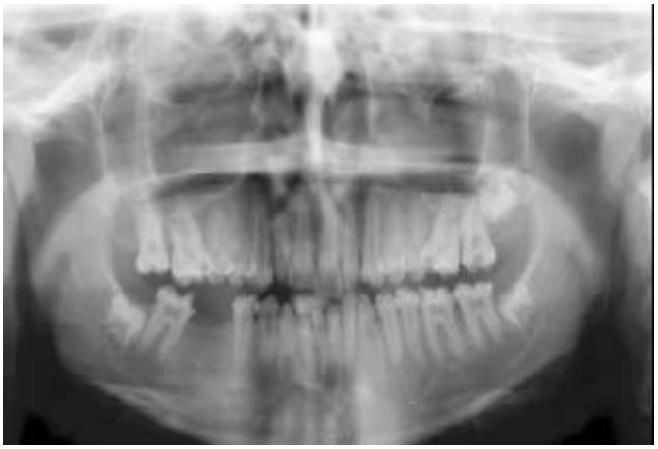


Figure2: Intraoral photographs showing mottled enamel in Case 1

The teeth, in general, exhibited a yellowish brown discoloration, with diffuse pitting present on almost all surfaces of all teeth, more prominent on labial and buccal surfaces. The surfaces of teeth were rough. The thickness of enamel was reduced exposing the dentin on almost all teeth except 11, 13, 16, 21, 23, 26.

A provisional clinical diagnosis of amelogenesis imperfecta and differential diagnosis of environmental enamel hypoplasia (dental fluorosis), dentinogenesis imperfect and dentin dysplasia were considered.



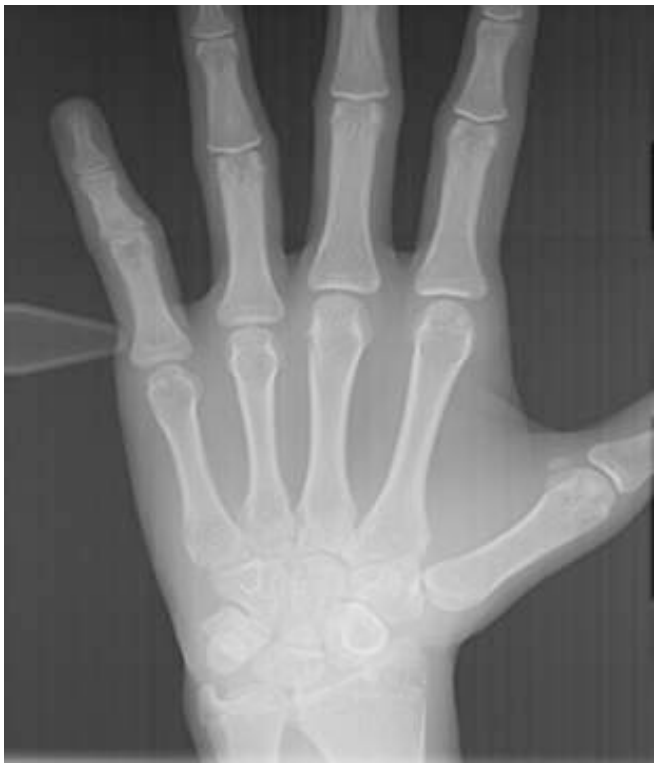


Figure 3: OPG and Hand Wrist Radiographs of Case 1. OPG shows generalized enamel structure loss

Radiographic investigations **(Figure 3)** included orthopantomogram (OPG) and hand wrist radiograph. Examination of radiographs revealed that enamel was almost half its expected thickness, but was of normal density i.e it was more radiodense than the dentin. Loss of cuspal height and open proximal contacts in posterior teeth were noted. Pulp chambers were normal with no sign of obliteration of root canals in any teeth. OPG also revealed same features in all teeth including unerupted 28, 38, 48 and absence of 18. OPG also showed normal bone and joints. Hand wrist radiograph showed normal bones, joints and skeletal maturation.

Correlating history, clinical features and radiographic features diagnosis of Type 1 hypoplastic amelogenesis imperfecta was considered and patient was referred to the department of prosthodontia for full mouth rehabilitation.

CASE REPORT – 2

A female patient of age 16 years belonging to low socio economic status reported to the outpatient department of oral medicine and radiology with

chief complain of yellowish discoloration of teeth since childhood. Family history revealed presence of same type of teeth in one of her elder sister (Case 1) while the other elder sister and two younger brothers were not affected. None of the parents and other family members from maternal or paternal side had the same history of discoloration of teeth. Her primary dentition was also discolored. Patient did not have disturbance in eruption of any tooth. Medical history was non contributory.

Extraoral examination **(Figure 1)** did not reveal any relevant findings. On intraoral examination **(Figure 4)** all the teeth were present. The teeth, in general, exhibited a yellowish brown discoloration, with diffuse pitting present on almost all surfaces of all teeth, more prominent on labial and buccal surfaces. The surfaces of teeth are rough. The thickness of enamel was reduced exposing the dentin on almost all teeth.

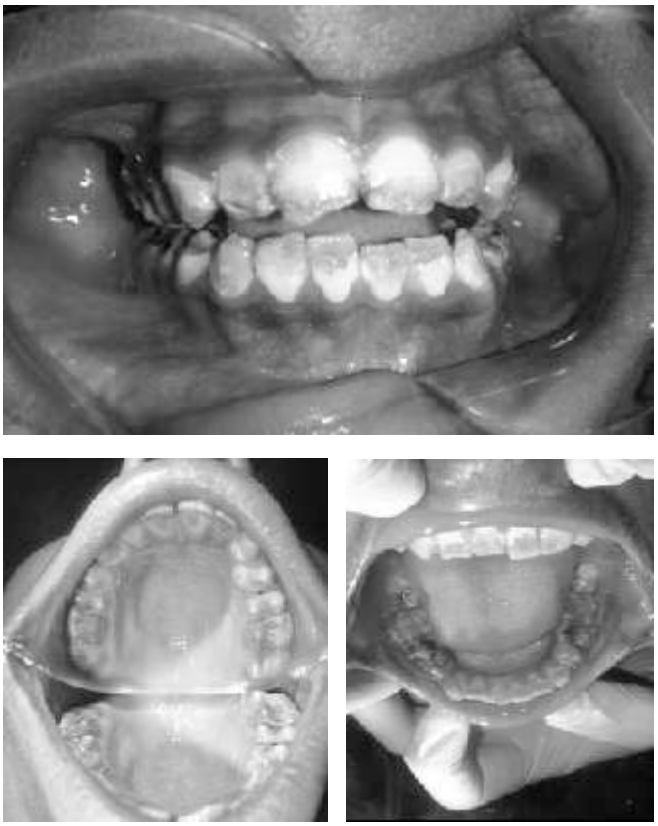


Figure 4: Intraoral photographs showing mottled enamel in Case 2

A provisional clinical diagnosis of amelogenesis imperfecta and differential diagnosis of environmental enamel hypoplasia (dental fluorosis), dentinogenesis imperfect and dentin dysplasia were considered.

Radiographic investigations **(Figure 5)** done included orthopantomogram (OPG). Examination of OPG revealed a missing 18 and normal pulp chambers and root canals with no sign of obliteration in any teeth. Multiple teeth showed loss of enamel structure. OPG also showed normal bone and joints. There was presence of unerupted 28, 38 and 48.

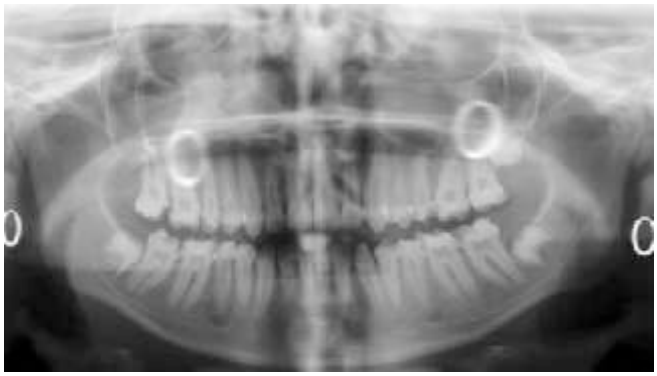


Figure 5: OPG of Case 2 showing generalized enamel structure loss

Correlating history, clinical features and radiographic features diagnosis of Type 1 hypoplastic amelogenesis imperfecta was considered and patient was referred to the department of prosthodontia for full mouth rehabilitation.

DISCUSSION

Amelogenesis imperfecta (AI) encompasses a complicated group of conditions that demonstrate developmental alterations in the structure of enamel in the absence of systemic disorder. The prevalence of this condition range from 1 in 718 to 1 in 14,000, depending on the population.[1,2,11]

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 into three categories: hypoplastic variety, hypocalcified variety, and hypomaturation variety. Later in 1989 Witkop gave the classification shown in table 1.[1,2]

Table - 1

TypeI	Hypoplastic
IA	Hypoplastic, pitted autosomal dominant
IB	Hypoplastic, local autosomal dominant
IC	Hypoplastic, local autosomal recessive
ID	Hypoplastic, smooth, autosomal dominant
IE	Hypoplastic, smooth X- linked dominant
IF	Hypoplastic, rough autosomal dominant
IG	Enamel agenesis, autosomal recessive
TypeII	Hypomaturation
IIA	Hypomaturation, pigmented autosomal recessive
IIB	Hypomaturation, X- linked recessive
IIC	Snowcapped teeth, autosomal dominant
TypeIII	Hypocalcified
IIIA	Autosomal dominant
IIIB	Autosomal recessive
TypeIV	Hypomaturation- hypoplastic with taurodontism
IVA	Hypomaturation-hypoplastic with taurodontism, autosomal dominant
IVB	Hypoplastic-hypomaturation with taurodontism, autosomal dominant

Hypoplastic AI represents 60-73% of all cases; hypomaturation AI represents 20-40%, and hypocalcification AI represents 7% of all cases.[4,5,11]

The first clear descriptions of X-linked hypoplastic AI were those of Schulze and Lenz and Schulze, who recognized the different manifestations in affected males and females. This was confirmed and expanded upon by Schulze in a monograph, detailing families from a geographically discrete area in Germany. The inheritance pattern of X-linked disorders dictates that male-to-male transmission cannot occur. Conversely, all female offspring's 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] The cases in this report had hypoplastic autosomal recessive (Type IC) type AI.

AI is sometimes associated with syndromes like, AI with taurodontism, trichodentoosseous syndrome, AI with nephrocalcinosis and cone-rod dystrophy with AI.[6,7] The commonest differential diagnosis which should be kept in mind during the clinical assessment is environmental enamel dysplasia (dental fluorosis) and dentinogenesis imperfecta. Dental fluorosis may present with areas of horizontal white banding corresponding to periods of more intense fluoride intake and may show the premolars or second permanent molars to be spared

(chronological distribution). The variability of this condition, from mild white flecking of enamel to profoundly dense white coloration with random, disfiguring areas of staining and hypoplasia, requires careful questioning to distinguish from AI. 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.[1,4] In the present cases, there is no such history is given by patient. Also the OPG shows the affected enamel formation in unerupted third molars and shows normal bones, joints and skeletal maturation in hand wrist radiograph. Dentinogenesis imperfecta can be differentiated from AI in our cases by the presence of bulbous crowns and narrow roots, the relatively normal density of any remaining enamel, and on radiographs the obliteration of pulp chambers and root canals is seen, in the absence of marked attrition.[7]

Treatment planning for patients with AI is related to many factors: the age and socioeconomic status of the patient, the type and severity of the disorder. The complexity of the condition requires an interdisciplinary approach for optimal treatment outcomes.[3] The treatment approach should be ideally be developed keeping in mind the specific AI type and underlying defect. The treatment of these patients has been usually done in two phases, temporary phase followed by transitory phase. Adhesive restorative techniques, over dentures, fixed partial dentures, full porcelain crowns, porcelain fused to metal crowns, and inlay/onlay restorations constitute the contemporary treatment modalities.[4]

CONCLUSION

A dentist should diagnose Amelogenesis imperfecta (AI) as early as possible for timely intervention and proper treatment planning for 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 clinicians knowledge about clinical and radiological diagnosis of AI as well as the intervention required for such a condition.

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