REVIEW ARTICLE

Amelogenesis Imperfecta: A Review

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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 entity can present a variety of clinical presentation varies from hypoplastic, hypomaturative to hypocalcified which are the result of various genetic mutations. AI can present with a vast variety of features in single entity, so detailed knowledge of genetic mutations regarding AI, diagnostic, radiographic features, and different treatment modalities are mandatory while dealing these cases. We are presenting a review article on AI, mainly focused on its clinical presentation, genetic background, and its treatment modalities.

Keywords: Amelogenesis imperfecta, dominant, enamel, recessive

INTRODUCTION

Amelogenesis imperfecta (AI) is an inherited disorder which affects only the ectodermal portion of the teeth, i.e., enamel with the variable occurrence of approximately 1/700-1/14,000.[1-3]

Various classifications have been proposed based on the phenotype,[4,5] based on the clinical, microradiographic and histopathological findings,[6] based on the phenotype and mode of inheritance,[1,3,7-10] based on molecular defect, biochemical result, mode of inheritance, phenotype,[11,12] and based on molecular defect sub-classification of the AMELX conditions.[13] Most commonly accepted classification is the one being proposed by Witkop 1988[1] whichclassified AI into mainly 4 types, i.e., hypoplastic, hypomaturative, and hypocalcified type based on developmental stages of enamel and hypoplastic-hypomaturation with taurodontism [Table 1].[1]

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Hypoplastic AI occurs due to defect in enamel matrix deposition, i.e., the first stage of enamel formation. Clinically patient presents with thin enamel with yellowish-brown, rough or smooth, flat occlusal surfaces of the posterior teeth due to attrition, and with/without grooves and/pitting. The enamel will be thin, well mineralized and do not chip. Radiographically thin enamel but normal radiodensity will be seen. Defects in matrix formation with a disturbance in the differentiation or viability of ameloblasts will be seen in histology section.[14-16] Different types of hypoplastic AI are described in Table 2.

In hypomaturative AI Enamel, matrix protein is normal, but a defect in the maturative process of enamel's crystal structure so enamel matrix is immature. Clinically mottled yellowish brown with opaque discolored/snow colored crown can be seen, but normal in size and shape of the crown with Soft, poorly mineralized and discolored enamel which is easy to penetrate by the dental probe. The thickness of enamel may be normal, but radiodensity is less than of dentin radiographically. Microscopically alterations in enamel rod and rod sheath structures will be

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appreciated.[14-16] Different types of hypoplastic AI are described in Table 3.

In hypocalcified type, the enamel matrix is laid down appropriate, but deficient calcification processes causing proper shape on eruption but very soft and friable enamel. On eruption, the enamel may be yellowish brown or orange, but it often becomes stained brown to black with calculus deposition. With time, coronal enamel is chipped more than a cervical portion. There are two types, i.e., diffuse autosomal dominant (AD) and diffuse autosomal recessive (AR), with more severity in AR. Radiographically enamel is less radiopaque then dentin.[14-16]

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.[14-16] Different types of hypoplastic AI are described in Table 4.

Table 1: Different type of AI[1]

Hypoplastic	IA: Hypoplastic, pitted AD
	IB: Hypoplastic, local AD
	IC: Hypoplastic, local AR
	ID: Hypoplastic, smooth AD
	IE: Hypoplastic, smooth XLD
	IF: Hypoplastic, rough AD
	IG: Enamel agenesis, AR
Hypomaturation	IIA: Hypomaturation, pigmented AR
	IIB: Hypomaturation, XLR
	IIC: Snow-capped teeth, AD
Hypocalcified	IIIA: AD
	IIIB: AR
Hypomaturation-hypoplastic with taurodontism	IVA: Hypomaturation-hypoplastic with taurodontism, AD
	IVB: Hypoplastic-hypomaturation with taurodontism, AD

AI: Amelogenesis imperfecta, AD: Autosomal dominant, AR: Autosomal recessive, XLD: X-linked dominant, XLR: X-linked recessive

GENETIC MUTATION IN AI

The trait of AI can be transmitted by an AD, AR, or X-linked mode of inheritance. Multiple genes are found to be altered.[17-53] Different gene mutation and their resulting defects in AI are summarized in Table 5.

OTHER FEATURES ASSOCIATED WITH AI

AI may be associated with some other dental and skeletal developmental defects or abnormalities, such as crown and root resorption, attrition, microdontia, taurodontism, delayed eruption and tooth impaction, dens in dente, pulp stones, anterior open bite, and agenesis of teeth.[54-56]

A study by Seow 1995 in 23 subjects with AI showed a significant acceleration of dental age in AI children of

Table 2: Variation of hypoplastic[14-16]

Type	Variant	Mode of inheritance	Characteristic features	Radiographic features
IA	Generalized pitted	AD	Pinpoint to pinhead-sized pits which are scattered across the surface of the teeth affecting buccal surface more severely, stained and arranged in rows or columns. The enamel between the pits is of normal thickness, hardness, and coloration and normal contact between teeth	Normal radiographic contrast of enamel and dentin
IB	Localized pitted	AD	The affected teeth may demonstrate either horizontal rows of pits, a linear depression or one large area of hypoplastic enamel surrounded by a zone of hypocalcification. Middle third of the buccal surfaces of the teeth is mainly affected, leaving incisal and occlusal surface intact	Normal radiographic contrast of enamel and dentin
IC	Localized pitted	AR	It is more severe and typically demonstrates the involvement of all teeth in both dentitions	Normal radiographic contrast of enamel and dentin
ID	Diffuse smooth	AD	The crown with thin, hard, glossy and smooth enamel, altered shape, opaque white to translucent brown color, anterior open bite and open contact between teeth	The teeth exhibit a thin peripheral outline of radiopaque enamel
IE	Diffuse smooth	XLD	Male: Diffuse thin, smooth and shiny enamel in both dentitions, yellowish brown, altered the shape of crown, open bite and open contact between teeth Female: Vertical furrows of thin hypoplastic enamel, alternating between hands of normal thickness	Male: A peripheral outline of radiodense enamel Female: The banding often is detectable with dental radiographs
IF	Diffuse rough	AD	The thin enamel denser then smooth type, hard and rough-surfaced, white to yellow-white, taper teeth toward the incisal-occlusal surface, open contact points and Anterior open bite	Thin peripheral outline of- radio-dense enamel
IG	Enamel agenesis		Total absence of enamel, causing shape of crown by rough dentin, yellow-brown hue which taper toward the incisal-occlusal surface, open contact points and anterior open bite	No peripheral enamel overlying the dentin with absence of eruption of many teeth with significant resorption

AD: Autosomal dominant, AR: Autosomal recessive, XLD: X-linked dominant, XLR: X-linked recessive

Table 3: Variation of hypomaturative AI[14-16]

Type	Variant	Mode of inheritance	Characteristic features	Radiographic features
IIA	Diffuse pigmented	AR	The enamel surface is mottled, agar-brown and soft enough to be punctured by a dental explorer	The affected enamel exhibits a radiodensity that is similar to dentin
IIB	Diffuse pigmented	XLD	Male: The deciduous teeth are opaque with translucent mottling while the permanent teeth are opaque yellow-white and may darken with age The enamel tends to chip and often call be pierced with a dental explorer point Female: Vertical bands of white opaque, translucent enamel are random and asymmetric	Male: The contrast between enamel and dentin is reduced Female: The bands are not detectable
IIC	Snow-capped	XLD/ XLR	Features are similar in male and female, i.e., a zone of white opaque enamel on the incisal or occlusal one-quarter to one-third of the crown affecting both deciduous and the permanent dentitions	The contrast between enamel and dentin is reduced
Type IID	Snow-capped	AD	Similar to snowcapped X-linked	The contrast between enamel and dentin is reduced

AD: Autosomal dominant, AR: Autosomal recessive, XLD: X-linked dominant, XLR: X-linked recessive, AI: Amelogenesis imperfecta

Table 4: Variation of hypomaturative AI with taurodontism[14-16]

IVA	Hypomaturation - hypoplastic with taurodontism	AD	Hypomaturation is a major defect than hypoplastic The enamel appears as mottled yellow-white to yellow-brown with pits on the buccal surfaces	The enamel appears similar to dentin in density. Large pulp chambers with varying degrees of taurodontism can be seen
IVB	Hypoplastic - hypomaturation with taurodontism	AD	Hypoplasia is a major defect	Decrease in the thickness of the enamel remaining similar to the hypomaturation - hypoplastic variant

AD: Autosomal dominant, AR: Autosomal recessive, XLD: X-linked dominant, XLR: X-linked recessive, AI: Amelogenesis imperfecta

approximately 1.13 ± 0.78 years compared with control children. The study also found a six-fold increase (26.1% vs. 4.3%) in the tendency of AI patients show impaction of the permanent teeth and associated anomalies such as follicular cysts.[54]

Koruyucu, *et al*. 2014 had conducted studies in 31 cases of AI and found out that the main complaints of patients with AI were dissatisfaction with the appearances of their teeth, extreme dental sensitivity, the presence of dental caries and other dental anomalies such as open bite and orthodontic problems. The study concluded that the patients with AI may have a high caries index, open bite, delayed tooth eruption, pathologic root resorption, pulpal calcifications, taurodontism, hypodontia, cross bite deep bite, prognathism, and retrognathism.[56]

SYNDROME ASSOCIATED WITH AI

AI with taurodontism is found to be associated with Trichodentoosseous (TDO) syndrome. TDO syndrome is an AD condition characterized by splitting of the superficial layers of the nails, Kinky or tightly curled hair, bone sclerosis of long bones and skull base, zones of provisional calcification in the long bone, taurodontism, and enamel hypoplasia that occurs with hypomaturation/hypocalcification defects.[57]

DIAGNOSTIC PRINCIPLES BEHIND AI

Accurate diagnosis requires a proper clinical history, clinical examination so that the presence of certain systemic diseases that may show generalized enamel hypoplasia can be excluded, identification of modes of inheritance determined by family pedigrees chart and proper radiographic interpretation. Accurate diagnosis enables genetic counseling in an early phase, and precautionary steps can be taken as an early step to prevent further dental complications for the patient and even for upcoming siblings in the future. Histological confirmation can be done, but required extraction of the affected tooth, which is not a good idea in all cases except where prognosis of that tooth is poor.

MANAGEMENT

Management directed at three aspects of treatment includes prevention, restoration, and esthetics. Most of the time patient report to the dentist when the dental complication like dentinal sensitivity or dental caries would have been started, so restoration should be undertaken as a first step.[56-62]

Preventive aspects include dietary advice, regular use of fluoride mouthwashes, topical fluoride applications, and oral hygiene instructions. Oral hygiene can be

Table 5: Different gene mutation in AI[17-53]

Gene	Mutation of the gene results	References
Amelogenin (AMELX)	X-linked AI	[17] [18] [19] [20] [21] [22] [23] [24]
Enamelin (ENAM)	AD AI	[25] [26] [27] [28] [29]
Ameloblastin (AMBN)	AD hypoplastic AI	[30] [31] [32]
KLK4	AR AI	[33]
MMP20	AR AI	[34] [35] [36] [37] [38]
DLX3	AD AI hypoplastic-hypomaturation with taurodontism	[39]
WDR72	AR hypomaturation amelogenesis	[40] [41]
FAM83H	Hypocalcified AI with AD AI	[42] [43] [44] [45]
C4orf26	AR AI	[46]
SLC24A4	AR hypomaturation AI	[47] [48]
ITGB6	AR AI, pitted hypomineralized AI	[49] [50] [51]
LAMB3	Hypoplastic AD AI	[52] [53]

AD: Autosomal dominant, AR: Autosomal recessive, AI: Amelogenesis imperfecta

difficult for these patients due to the sensitivity while brushing so warm water for tooth brushing can be advised. Along with preventive measures, long-term clinical follow-up term is mandatory.[56-62]

Restorative aspects can be divided at the time of dentition, i.e., during primary dentition, direct composite veneers for anterior teeth, GIC and stainless steel crowns for primary molars can be advised while in the mixed dentition stainless steel crowns, onlays for permanent molars, composite or GICs for primary and permanent teeth and for permanent incisors, direct or indirect composite veneers should be advised.[56-62]

The esthetic treatment modality also decided based on dentition, i.e., minimal intervention GIC restorations should be advised in the primary dentition, direct and indirect composite resin veneers in the mixed dentition while porcelain veneers, full crowns, extractions of excessive defected teeth followed by fixed, or removable prosthesis should be advocated in permanent dentition based on number of teeth affected, patient age, and economical status.[56-62]

Still no standard formula, current protocol, or guideline for successful treatment the accurate treatment should be planned based on the type of AI, severity and oral health habits of the patient. Except for mildly affected teeth, restorations with amalgam are usually unsuccessful due to fracture of the weak enamel margins. Adherent materials such as glass ionomer cements and composite resins are better retained compared to amalgam restorations. However, in cases of hypocalcified AI where the enamel is very weak and bonding of the restoration is questionable, full coverage is required. In the primary and early mixed dentition, stainless steel crowns are effective restorations.[56-62] The most effective method for dentinal sensitivity is full coronal coverage using stainless steel crowns on the posterior teeth, especially in mixed dentition and primary dentition.[56-62]

In constructing steel crowns, a conservative technique of tooth separation using separating elastics prior to the insertion of the crowns is recommended. Glass ionomer cements are likely to be better luting agents for the crowns compared with zinc phosphate if there are large areas of exposed dentin. Anterior open bite can be treated with different treatment modalities ranging from orthodontic banding or orthognathic surgery. Orthodontic treatment can be performed only after once all restorative treatment is finished.[14,56-62]

CONCLUSION

AI is an inherited disorder affecting enamel. This entity more often causes psychological stress in patients due to poor esthetics. A proper history, clinical evaluation, genetic mapping, radiological interpretation followed by best suitable treatments are the necessary steps to make them smile. Dental practitioners should be aware of all possible treatment for AI as one treatment which is successful in one type, may fail in others.

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