

## AMELOGENESIS IMPERFECTA: REPORT OF TWO CASES

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**ABSTRACT:** Amelogenesis Imperfecta (AI) represents a group of genetic conditions, which affect the structure and clinical appearance of enamel all or nearly all the teeth. AI patients have a serious problem that reduces oral health related quality of life and causes some physiological problems. Herewith are presented cases of cousin sisters with AI. Both of them had hypoplastic type of AI which is diagnosed on the basis of classical clinical and radiographic features

**KEYWORDS:** : Amelogenesis Imperfecta (AI), Hypoplastic, Genetic

## INTRODUCTION

Amelogenesis imperfecta (AI), a group of hereditary diseases affecting enamel in either quality or quantity, is associated with crown malformation along with abnormal enamel density<sup>1</sup> The prevalence varies from 1:700 to 1:14 000, according to the populations studied<sup>2</sup>

Clinical features are depend on the type of AI involved. AI has been classified on the basis of clinical, radiographic, and histologic appearance of the enamel defect and the mode of inheritance of the trait. AI has been categorized as 1) Hypoplastic (autosomal dominant /autosomal recessive/x linked dominant), 2) Hypocalcified (autosomal dominant/ autosomal recessive) 3) Hypomaturational types (autosomal recessive/x linked recessive/autosomal dominant) and 4) Hypoplastic hypomaturational type.<sup>1,2</sup> Hypoplastic AI is most common representing 60 to 73% of all cases, hypomaturational AI represents 20 to 40%, and hypocalcification AI represents 7%.<sup>3</sup>

Hypoplastic form of AI is characterized by thin enamel with yellowish brown colour, rough or smooth and glossy, square shaped crown, lack of contact between adjacent teeth, flat occlusal surfaces of the 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.<sup>2,4,5</sup>

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. Radiographically, in hypocalcified form, thickness of enamel is normal but radiodensity of enamel is less than that of dentin. Histologically, in hypocalcification type shows defects of matrix structure and mineralization.<sup>2,4,5</sup>

Hypomaturational 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 coloured cloudy white/yellow/brown/ snow-capped. Radiographically, radiodensity of enamel is similar to that of dentin. Histologically, in hypomaturational type shows alterations in enamel rod and rod sheath structures.<sup>2,4,5</sup>

## Case reports

**Case 1:** A 21- year-Female patient came with a complaint of yellow discoloration of teeth since 12-13 years. Patient was giving history of discoloured anterior deciduous teeth. Medical history was not significant but family history revealed similar problem of teeth in many member of family. (Fig.1)

Patient had permanent dentition with no missing teeth. Upper and lower posterior teeth showed yellowish brown discoloration with attrition. (Fig.2, Fig.3 and Fig.4). Consistency of enamel in affected teeth was hard and chipping of enamel. On the basis of clinical examination, provisional diagnosis of hypoplastic AI was made and panoramic radiograph was advised which showed presence of a thin layer of enamel with radiodensity of enamel more than dentin along with open contact area (Fig.5). Final diagnosis of hypoplastic AI was made.

**Case 2:** A 25- year-Female patient came with a complaint of discoloured teeth since childhood. Patient gave history of brownish coloured deciduous teeth. When permanent teeth erupted they were also small size, brown coloured with space between teeth. On intraoral examination, patient had permanent dentition except 63 as 23 was impacted. Proximal caries was seen with 36, 47. Root piece with 15 and grossly destructed 16,46. All teeth yellow-brown discoloured, proximal open contact area in

Case 1



Fig.1. Extra oral Photograph



Fig.2: Intraoral-frontal Photograph



Fig.3: Intraoral-Maxillary occlusal Photograph



Fig.4: Intraoral-Mandibular occlusal Photograph



Fig.5. Orthopantomograph ( OPG) of the patient

posterior teeth(Fig.7, Fig.8 and Fig.9). Soft tissue examination showed Sinus associated with 47 on attached gingiva. On the basis of clinical examination, provisional diagnosis of hypoplastic AI was made. Panoramic radiograph was advised.

Radiological examination: Panoramic radiograph showed periapical radiolucency associated with 47 and 36, severe bone loss with 47. Severe root resorption seen with 63 along with impacted 23. Posterior teeth showed open contact area. (Fig.10).

Careful history was taken and family chart was prepared. (Fig.11).Final diagnosis of hypoplastic AI was made.

Discussion

During amelogenesis, the enamel transitions from a soft and pliable tissue to its final form, this is almost entirely devoid of protein. The final composition of enamel has unique and it is a result of molecular and cellular activities that take place during its genesis. Deviation from this pattern may lead to AI.<sup>3,6</sup> Mutation or alteration in the genes encoding specific enamel proteins such as Enamelin gene (ENAM), Amelogenin gene (AMELX), Kallikrein 4 gene (KLK4), Matrix Metalloproteinase 20 gene (MMP.20), and Distal-less homeobox 3 gene (DLX3) has found to be associated with AI.<sup>7</sup> Studies showed that AI is familial similar to our two cases and can also be inherited as autosomal dominant, autosomal recessive, or x-linked dominant and x-linked recessive.<sup>1</sup>

Case 2



Fig.1. Extra oral Photograph



Fig.2: Intraoral-frontal Photograph



Fig.3: Intraoral-Maxillary occlusal Photograph



Fig.4: Intraoral-Mandibular occlusal Photograph



Fig.5. Orthopantomograph ( OPG) of the patient

Although both deciduous and permanent dentition are affected in AI, it is affected more commonly in permanent teeth than primary teeth and more commonly in incisor teeth and first molars in both upper and lower jaws.<sup>8</sup> According to various studies of AI, many non-enamel anomalies has been reported with AI, e.g.: Delayed tooth eruption, congenitally missing teeth, anterior open bite, taurodontism, pulpal calcifications, dentin dysplasias, root and crown resorption, hypercementosis, and root malformations. Malocclusion and gingivitis may found in association with AI.<sup>1,4</sup>

Patients chief complaints associated with AI were non-aesthetic appearance, extensive loss of tooth structure, dental sensitivity, and loss of vertical dimension.<sup>4,8</sup> Treatment planning for patients with AI is

related to many factors such as age and socioeconomic status of the patient, the type and severity of the disorder, and the intraoral situation at the time the treatment is planned.

Treatment plan includes removal of surface stains, reducing sensitivity, maintaining vertical dimension of occlusion, and the aesthetics with adhesive techniques/overdentures/porcelain-fused-to metal crowns/ fixed partial dentures are used for better aesthetics and restoring its function. Preventive aspects include dietary advice, fluoride supplements, and oral hygiene instructions.<sup>1,3</sup>

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