Amelogenesis imperfecta: Four case reports

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. AI is a serious problem that reduces oral health-related quality of life and causes some physiological problems. We presented here four case reports of AI (Hypoplastic and Hypomaturation) which we diagnosed on the basis of classical clinical and radiographic features.

Key words: Amelogenesis imperfecta, enamel hypoplasia, hypoplastic teeth

INTRODUCTION

Amelogenesis imperfecta (AI), a group of hereditary diseases affecting the tooth enamel in either quality or quantity, is associated with crown malformation and abnormal enamel density. The prevalence varies from 1:700 to 1:14 000, according to the populations studied.[3] Clinical features of patients with AI 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 hypoplastic (autosomal dominant/autosomal recessive/x-linked dominant), hypocalcified (autosomal dominant/autosomal recessive), hypomaturation types (autosomal recessive/x-linked recessive/autosomal dominant) and hypoplastic-hypomaturation type.[1,2] Hypoplastic AI represents 60 to 73% of all cases, hypomaturation AI represents 20 to 40%, and hypocalcification AI represents 7%.[3]

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 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.[2,4,5]

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.[2,4,5]

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.[2,4,5]

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.[1]

**CASE REPORTS**

**Case 1**

A 17-year-old male patient presented with a complaint of yellow discoloration of his teeth since 8 to 9 years. Patient was giving positive history of discolored anterior deciduous teeth; however, medical history and family history was unremarkable. Patient had permanent dentition with no missing teeth. All the anterior and posterior teeth were affected with brownish discoloration; while attrition present with posterior teeth. Height of the clinical crowns of upper and lower posterior teeth were reduced. In 11, 14, 21, 22, 24, 34, 42, pitting were present. Consistency of enamel was hard. Chipping of enamel was not present in any teeth [Figure 1]. Panoramic radiograph was taken which showed presence of a thin layer of enamel with radiodensity of enamel more than dentin [Figure 2]. On the basis of clinical and radiographic features, final diagnosis of hypoplastic AI was made.

**Case 2**

A 12-year-old male patient came with a complaint of yellow brown discoloration of his upper anterior teeth since 4-5 years. His parents were aware of discolored deciduous teeth. Family history was unremarkable. Patient had mixed dentition present with deciduous molar root pieces. On examination, brownish discoloration was present in 21, 11, 36, 46 and chalky white discoloration was present in relation with all other teeth. Incisal edges

![Figure 1: Clinical photos showing Amelogenesis Imperfecta (Case 1 - Case 4)](image1)

![Figure 2: OPG-panoramic view of patients showing radiographic features of Amelogenesis Imperfecta (Case 1 - Case 4)](image2)
of 33, 34, 43, 44 were thin with dentin exposed. Enamel was softer in consistency with chipping of enamel on probing [Figure 1]. On the basis of clinical examination, provisional diagnosis of hypomaturative AI was made. Panoramic radiograph was advised which showed presence of a thin layer of enamel with radiodensity of enamel more than dentin and taurodontism of 16, 26 [Figure 2]. Final diagnosis of hypoplasic-hypomaturative AI with taurodontism of 16, 26 was made.

Case 3
A 22-year-old female patient came with a complaint of yellow discoloration of teeth since 12-13 years. Patient was giving history of discolored anterior deciduous teeth. Medical history and family history was unremarkable. Patient had permanent dentition with no missing teeth. Upper and lower posterior teeth showed yellowish brown discoloration with attrition [Figure 1]. Consistency of enamel in affected teeth was hard and chipping of enamel not present in any teeth. On the basis of clinical examination, provisional diagnosis of hypomurative AI was made and panoramic radiograph was advised which showed presence of a thin layer of enamel with radiodensity of enamel more than dentin [Figure 2]. Final diagnosis of hypomurative AI was made.

Case 4
A 14-year-old male patient came with a complaint of yellow discoloration of teeth since 5-6 years. Past dental history was suggestive of discolored deciduous teeth. Medical history and family history was unremarkable. Anterior as well as posterior teeth of all segments showed brownish discoloration [Figure 1]. Height of the crowns of upper and lower posterior teeth were reduced. Consistency of enamel and dentin was hard. Chipping of enamel was not present in any tooth. On the basis of clinical features, provisional diagnosis of hypomurative type of AI was made. Panoramic radiograph advised to confirm the diagnosis which showed presence of a thin layer of enamel with radiodensity of enamel more than dentin [Figure 2]. On the basis of clinical and radiographic features, final diagnosis of AI of hypomuration type was made.

DISCUSSION
During organogenesis, 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 is a reflection of the unique molecular and cellular activities that take place during its genesis. Deviation from this pattern may lead to AI.\(^3\)\(^5\)\(^6\) Mutation or alteration in any of 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) have been linked with AI.\(^7\) Studies showed that AI is familial and can be inherited as autosomal dominant, autosomal recessive, or x-linked dominant and x-linked recessive.\(^3\)

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.\(^8\)

According to various studies of AI, many non-enamel anomalies have been found in association 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 also have been found in association with AI.\(^1\)\(^4\)

Various studies showed that oral complaints associated with AI are unaesthetic appearance, extensive loss of tooth structure, dental sensitivity, and loss of vertical dimension.\(^6\)\(^8\) No standard formula for successful treatment of AI has been demonstrated. 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, and the intraoral situation at the time the treatment is planned. It includes removal of surface stains, reducing sensitivity, maintaining vertical dimension of occlusion, and the esthetics with adhesive techniques/overdentures/porcelain-fused-to metal crowns/fixed partial dentures/full porcelain crowns/inlay/onlay restorations are used for better esthetics of the patients. Preventive aspects in the primary and mixed dentition include dietary advice, fluoride supplements, and oral hygiene instructions. Topical fluoride application can be done during the permanent dentition. Restorative aspects in the primary dentition includes giving glass ionomer restorations and stainless steel crowns, in the mixed dentition stainless steel crowns.\(^1\)\(^3\)\(^7\)\(^8\)

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Osteochondroma of mandibular condyle: A clinico-radiographic correlation

Abstract
Osteochondroma (OC) of temporo mandibular joint is a rare, slow growing, benign tumor that causes a progressive enlargement of the condyle, usually resulting in facial asymmetry, temporo mandibular joint (TMJ) dysfunction, limited mouth opening and malocclusion. Pain is rarely associated with this tumor. OC is composed of cartilaginous and osseous tissues. Radiographically, there is unilaterally enlarged condyle usually with an exophytic outgrowth of the tumor from the condylar head. We present a rare case of osteochondroma of right mandibular condyle in a 45-year-old male who reported with painless swelling over TMJ area and progressive limited mouth opening. Panoramic radiograph and computed tomography (CT) was performed for better evaluation of the pathological condition. This paper describes the clinico-radiographic features and differential diagnosis of OC.

Key words: Mandibular condyle, mandibular dysfunction, neoplasm, osteochondroma, temporo mandibular joint

INTRODUCTION
Osteochondroma (OC) is defined as an osteocartilagenous exostosis with cartilage capped exophytic lesion that arises from the bone cortex. It is one of the most common benign tumor of the axial skeleton, but is rarely associated with the facial bones. It has been described in the head, cranial base, jaw, maxillary sinuses, condyle, ramus, body, coronoid process and symphyseal mandibular region. The embryonic development of the temporo mandibular joint (TMJ), by the endochondral ossification, makes this area the most frequent facial site for OC. It represents approximately 35% to 50% of all benign tumors, and 8% to 15% of all primary bone tumors.

CASE REPORT
A 45-year-old male patient reported to the department with complaint of painless hard slow growing swelling on the right TMJ area and asymmetrical face since two years. He noticed reduced mouth opening and deviation of the jaw while opening and closing the mouth [Figure 1a]. There was no history of any trauma or ear infection. The medical history was non-contributory. The clinical examination revealed non-tender bony hard oval swelling on right TMJ measuring around 2 cm × 2 cm [Figure 1b], reduced TMJ movements, facial asymmetry with approximately 8 mm deviation of the midline to the right side, severe malocclusion, unilateral posterior cross bite on the right.