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Oral Pathology

HYPOPLASTIC TYPE OF AMELOGENESIS IMPERFECTA – A REPORT OF TWO CASES

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ABSTRACT Amelogenesis imperfecta (AI) represents a group of developmental conditions of the dental enamel, which are genomic in origin, affecting the structure and clinical appearance of enamel, characterised by hypoplasia and/or hypomineralisation that shows autosomal dominant, autosomal recessive, sex-linked and sporadic inheritance patterns. Diagnosis is based on thorough family history, pedigree chart frame, and meticulous clinical observation. Genetic analysis also serves as a significant research tool. Treatment of Amelogenesis Imperfecta is important mainly because of social psychological attitude in the patient, apart from which aesthetic and functional aspects are considered that necessitates a multidisciplinary approach. This article showcases two case report of amelogenesis imperfecta- diffuse smooth hypoplastic type with a review of the literature.

KEYWORDS : Amelogenesis imperfecta, developmental disorder, enamel discoloration, hypoplastic enamel.

INTRODUCTION:

Amelogenesis imperfecta (AI) is a heterogeneous group of genetic disorders characterized by defects in tooth enamel formation in the absence of any generalized or systemic diseases. The name hereditary amelogenesis imperfecta was suggested by Weinmann *et al.* in 1945. The terms aplasia and hypoplasia were used earlier. The condition has been classified into four categories with multiple subtypes based on the mode of inheritance and phenotypic presentation. The hypoplastic type is the most frequent form of AI (61.2%), followed by hypomaturation AI (32.2%), hypocalcified AI and mixed hypomaturation / hypoplastic AI (3.2%). [1,2] Major enamel matrix proteins (amelogenin, enamelin, and ameloblastin) are suggested to contribute to the enamel formation of teeth. These proteins are secreted by ameloblasts during the secretory stage, which are important for the growth of enamel crystal.[3] Increased sensitivity to hot and cold, nonaesthetic appearance due to discoloration of the anterior teeth and impaired masticatory function are the common symptoms experienced by the patient affected by Amelogenesis imperfecta.

In this article, we present two case reports showcasing a hypoplastic type of hereditary Amelogenesis imperfecta with a review of literature.

CASE REPORT 1:

A 23 years old male patient reported with the chief complaint of discoloration in his teeth since childhood. The patient revealed a similar history of discoloration even in his primary dentition and a familial history of one of his twin sibling having a similar complaint. Parents had a consanguineous marriage. The pedigree chart was framed as in Figure 1. Patient's medical history revealed no systemic abnormalities.

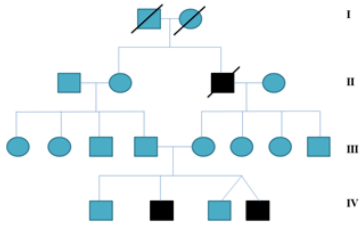

FIGURE 1: Pedigree chart of case report 1

On intra-oral examination, hard tissue findings revealed generalized diffuse brownish yellow discoloration of teeth. Teeth appeared smooth and glossy with no pitting or roughness. Attrition was present in posterior teeth in relation to 16, 17, 26, 27, 36, 37, 46, 47 region. Angles class I malocclusion with anterior open bite was elicited (Figure 2). Investigatory procedures included OPG showing a thin peripheral outline of radio-opaque enamel in all the teeth. Loss of crown structure involving enamel was evident in relation to 16, 17,26,27,36,37,46,47 region (Figure 3). Impacted 48 was extracted and ground sectioned, viewed under a light microscope and compared with the unaffected normal tooth (Figure 4.a).

Microscopic features revealed an increase in number of enamel lamellae and tufts. The enamel rods exhibited a wavier course in a few areas. Gnarled enamel was also absent. The incremental line of retzius was not prominent in the occlusal aspect. The dentinoenamel junction appeared flat without scalloping (Figure 4.b). Figure 4.c reveals a ground section of a normal tooth, where regularly arranged enamel rods are seen along with the normal thickness of enamel. Hypercementosis of the affected tooth was also evident (Figure 4.e). On the basis of the complete history of the patient, clinical, radiographic, and histopathological features the diagnosis of hypoplastic, diffuse smooth, AI was confirmed. Patient was advised for a full mouth veneering.

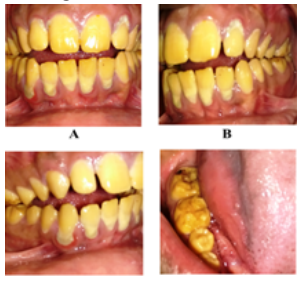

FIGURE 2: Case report 1 - Patients Occlusion showing anterior open bite (A); Diffuse brownish yellow discoloration of teeth (B & C); Attrition of molar teeth (D).

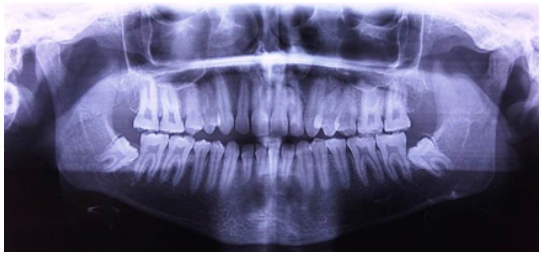

FIGURE 3: OPG of case report 1, revealing a thin peripheral outline of radio-opaque enamel in all the teeth

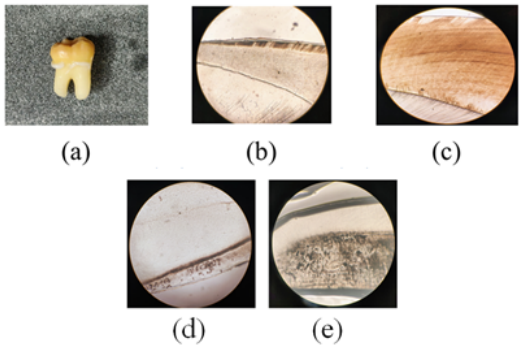

FIGURE 4: Case report 1 - Extracted tooth 48 (a); the ground section of affected and the normal tooth (b & c) respectively; normal root cementum (d); affected tooth showing hypercementosis (e).

CASE REPORT 2:

A 26 years old male patient reported with the chief complaint of dirt and deposits in his teeth for the past 6 months. Patient revealed a history of similar complaint with his elder sibling. Patient's medical history revealed no systemic abnormalities. Intraoral examination revealed yellowish discoloration of teeth exhibiting pits and grooves on middle third of the crowns with an anterior open bite. An intra-oral peri-apical radiograph revealed that enamel density was not appreciable. There was absence of contact between enamel and cementum indicating pattern III CEJ (Gap junction). Partial loss of coronal portion of the tooth was seen suggestive of attrition of the tooth. Mandibular left premolar (35) with a poor prognosis was extracted and sent for the ground sectioning and histological examination was suggestive of Hypoplastic type of Amelogenesis Imperfecta. Patient was advised for oral prophylaxsis and prosthetic treatment for upper anteriors.

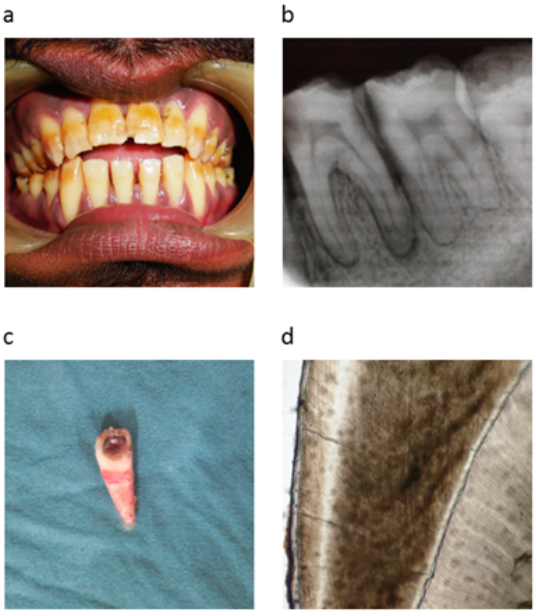

FIGURE 6: Case report 2 - Patient dentition showing enamel discoloration with anterior open bite (a); Loss of enamel evident in IOPA (b); Ground section of extracted tooth 35 (c & d)

DISCUSSION:

Amelogenesis imperfecta (AI) is a hereditary condition affecting both the primary and permanent dentitions that result in an abnormal quantitative/qualitative enamel formation. The defects that occur during the secretory phase result in improper matrix secretion leading to production of defective enamel (hypoplasia) which makes the enamel fragile and thin. The defects occurring during the maturation stage result in a normal volume of enamel but with insufficient mineralization (hypomineralization). The classifications of AI are based on clinical appearance, radiographic and histological findings and enamel thickness: **hypoplastic** (type I); **hypomaturation** (**typeII**); **hypocalcified** (typeIII); and **hypomaturation** – **hypoplasia with taurodontism** (type IV) and the subtypes are based on mode of inheritance and gene mutation. The hypocalcified type, the commonest type is characterized by enamel of reduced thickness and with incomplete matrix mineralization which happens in the secretory phase. The hypomature type shows a normal thickness of enamel, but is opaque and brittle because of an irregularity that occurs during the hydroxyapatite crystals development in the maturation phase.[4] There are three major structural proteins involved in AI: amelogenin *(AMEL),* ameloblastin (*AMBN*), enamelin *(ENAM).* Amelotin (*AMELOTIN*), dentine sialophosphoprotein and a variety of enzymes such as kallikrein 4 (*KLK4*) and matrix metalloproteinase 20, also contributes to the mutation in some cases of AI.

Amelogenesis imperfecta should be carefully diagnosed clinically as there are different reasons for yellowish tooth discoloration such as poor oral hygiene, aging, dental fluorosis, turners teeth, dentinogenesis imperfecta, and dentin dysplasia. Patient's history of tooth discoloration provides convenient information regarding the etiology and thereby helps in ruling out other related conditions. The eruption of the affected teeth is often delayed and a tendency for tooth impaction exists. Proper clinical examination and inspection of the intrinsic and extrinsic stains using a scratch test helps in better diagnosis. Radiological interpretation of AI in hypoplastic type usually presents a square crown showing open contacts, relatively thin radio-opaque layer of enamel, low or absent cuspal elevations. The appearance of anterior teeth in radiograph resembles a picket fence - type appearance. In cases of advanced abrasion, the appearance of obliterated pulp chambers usually resembles to that of dentinogenesis imperfecta.[5]
Amelogenesis imperfecta being a genetic disease exist in isolation or in interelation with other features in syndromes. It can be either a single gene defect or appear from a microdeletion / chromosomal defect. *Cone-rod dystrophy* is a syndrome associated with AI showing linkage to 2q11. *Jalili syndrome* refers to the coexistence of cone rod dystrophy (CRD) and AI, due to a mutation of the CNNM4 gene. *Taurodontism*, an inherited anomaly of the dentine, is considered to be a feature of hereditary amelogenesis imperfecta type IV- *Hypomaturative-hypoplastic with taurodontism AI (AIHHT),* one of the phenotypes present in patients diagnosed with *tricho–dento–osseous syndrome (TDO).* Therefore, the mutant genes in hereditary amelogenesis imperfecta could affect cells other than the ameloblasts or several modifications in genes may be involved. TDO is due to a DLX3 mutation and primarily affects hair, bones, and teeth showing enamel hypoplasia, and severe taurodontism. *Nephrocalcinosis* is also said to be in association with amelogenesis imperfecta. *Hamartomatous atypical follicular hyperplasia (EDHFH)* is a rare syndrome similar to central odontogenic fibroma in multiple impacted teeth and shows generalized enamel dysplasia with features of hypoplastic AI having open-bite malocclusion, gingival overgrowth, hypodontia, pulpal calcifications and aberrant root formation of the unerupted teeth.[13] A *skeletal anterior open bite* is seen in approximately 50% of patients with AI of either X-linked or autosomal inheritance. Such an association is also seen in both our cases which itself is considered as a syndrome that is said to carry ENAM or AMGX mutations. The disturbances of the enamel epithelium can also cause defects in the eruptive mechanism, eventually resulting in an anterior open bite. Studies have proved that deficiencies in the formation, migration and the proceeding development of neural crest cells may lead to a number of congenital anomalies of craniofacial complex.[6] Clinical investigations have shown that vertical dysgnathia increases the anterior maxillomandibular distance resulting in improper placement of tongue from its normal rest position and thus, an anterior oral seal can be produced. Literature search reveals that anterior open bite was seen most commonly in the hypocalcified type of AI, less in the hypoplastic type, and completely absent in the hypomaturation type.[7,8] However in both our case reports, anterior open bite was exhibited in hypoplastic type of AI.

Management of amelogenesis imperfecta requires multispeciality treatment modalities. The first study conducted by Coffield and colleagues, attempted to objectively characterize the psychosocial impact on AI patients which showed that subjects with AI had higher levels of social avoidance, distress and higher levels of dysfunction. Early diagnosis and prompt intervention increases psychosocial wellbeing and self-esteem of AI patients, particularly during the pre-adolescent and adolescent stages. As in most cases, aesthetic correction along with psychological counselling is of prime importance. The age of the patient determines what type of treatment plan to be followed which are categorized as temporary and permanent phase.[1] Primary and mixed dentition will require management on caries prevention, composite restorations or prefabricated crowns (Temporary phase). For permanent dentition (Permanent phase) treatment objectives should be on decreasing tooth sensitivity, full mouth rehabilitation, and treatment for malocclusion such as crown lengthening, orthodontic treatment and orthognathic surgery. [9,10] Renner et al. reported a case of AI treated by maxillary overdenture which is also an alternate treatment procedure to a complex prosthodontic problem. The increased sensitivity of teeth affected by AI results in poor oral hygiene in patients leading to generalized gingivitis which requires periodontal treatment like scaling and root planning. In both our case presentation, proper treatment protocol was advised and patients was found satisfactory.

Thus, accurate diagnosis along with the identification of modes of inheritance using family pedigrees chart accompanied by proper radiographic and histological interpretation is essential to commence an early phase of treatment.

CONCLUSION:

Early identification of Amelogenesis imperfecta aids in faster intervention by maintaining good oral hygiene and restoring the functional and aesthetic requirements. Thus, this article gives an insight on the thorough family and clinical history along with radiological and histological findings which provides better knowledge to treat AI accordingly.

CONFLICTS OF INTEREST: NIL

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