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Amelogenesis imperfecta: review and diagnosis of a case with hypoplasia and skeletal open bite

Amelogenesis imperfecta*: revisão e diagnóstico de um caso com hipoplasia e mordida aberta esquelética*

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Abstract

Introduction: To present a case of a patient presenting with the hipoplastic type of amelogenesis imperfecta, a hereditary disorder that disturbs the formation of the dental enamel both in the primary and the permanent dentition resulting in poor development or complete absence of the enamel of the teeth. **Discussion and conclusions**: *Amelogenesis imperfecta* occurs in the general population in the approximate range of 1 in 14.000 to 1 in 16.000. Dental features associated with AI include: quantitative and qualitative enamel deficiencies, pulpal calcification, taurodontism and root malformations; failed tooth eruption and impaction of permanent teeth; progressive root and crown resorption; congenitally missing teeth; and anterior and posterior open bite occlusions. The disorder may create unaesthetic appearance, dental sensitivity and loss of occlusal vertical dimension. The complexity of the management of patients with AI supports the suggestion the dental profession should have appropriate methods for the rehabilitation of rare dental disorders. The treatment of patients with AI should start with early diagnosis and intervention to prevent later restorative problems.

Keywords: *Amelogenesis imperfecta*. Enamel abnormality. Prothesis restoration.

Resumo

Objetivos*: Apresentar um caso de paciente acometido pelo tipo hipoplásico de* amelogenesis imperfecta*, doença hereditária que afeta a formação do esmalte dentário, tanto na dentição primária como na permanente, resultando em desenvolvimento deficiente ou ausência completa do esmalte nos dentes.* ***Discussão e conclusões****: A* amelogenesis imperfecta *ocorre na proporção de 1:14.000 a 1:16.000 na população em geral. Aspectos dentários associados a esse distúrbio incluem: deficiências qualitativas e quantitativas de esmalte; calcificações pulpares; taurodontismo e más-formações radiculares; falhas na erupção dentária e inclusão de permanentes; reabsorção progressiva coronal e radicular; ausências congênitas de dentes e oclusões com mordidas abertas anteriores e posteriores. A desordem pode causar aspecto antiestético, sensibilidade dentária e perda de dimensão vertical. A complexidade do tratamento de pacientes sugere que a profissão odontológica deve buscar métodos apropriados para a reabilitação em casos da ocorrência dessa doença rara. O tratamento deve começar com diagnóstico e intervenção precoces para prevenir dificuldades de restauração com a evolução dos casos.*

Palavras-chave*:* Amelogênese imperfeita*. Anomalias do esmalte. Restauração protética.*

Introduction

Amelogenesis imperfecta (AI) is a hereditary disorder that disturbs the formation of the dental enamel both in the primary and the permanent dentition, resulting in poor development or complete absence of the enamel of the teeth (1-4). Because the dentin is visible through the slim enamel, the teeth seem yellow and are well damaged. It occurs in the general population in the approximate range of 1 in 14.000 to 1 in 16.000 (5, 6).

Three types of AI are described in the literature: hypoplastic AI (Type I), hypomaturation AI (Type II) and hypocalcified AI (Type III). Enamel hypoplasia is an exclusive ectodermal disturbance related to alterations in the organic enamel matrix, which can cause white flecks, narrow horizontal bands, lines of pits, grooves and discoloration of teeth, varying from yellow to dark brown (7). Enamel hypocalcification is a defect in the mineralization process which causes the enamel to become soft and friable.

The hypomaturation variance is determined by abnormality in the final stages of the mineralization process and differs from hypocalcification in that the enamel is harder with a mottled opaque white to yellow-brown or red-brown color (8). Therefore, the entire enamel of primary as well as permanent teeth is affected. The teeth may or may not be discolored and often there is considerable occlusal wear (9). Dental features associated with AI include (6, 10): quantitative and qualitative enamel deficiencies, pulpal calcification, taurodontism and root malformations; failed tooth eruption and impaction of permanent teeth; progressive root and crown resorption; congenitally missing teeth; and anterior and posterior open bite occlusions. The disorder may create unaesthetic appearance, dental sensitivity and loss of occlusal vertical dimension. In these patients, the pulp and dentin are usually normal and the teeth are caries resistant (11).

The diagnosis and classification of *amelogenesis imperfecta* has traditionally been based on the clinical display or phenotype and the heritage form. *Amelogenesis imperfecta* may be inherited as a X-linked, autosomal predominant or autosomal recessive hereditary trait, depending on the character. In most cases, males with X-linked *amelogenesis imperfecta* experience more serious dental abnormalities than females with this kind of this circumstance.

The treatment plan for patients with AI is related to many factors including the age of the patient, the socio-economic status, the type and severity of the disorder and its intraoral manifestation (12). *Amelogenesis imperfecta* cases can tax the skills of the clinician. Numerous treatments have been described for rehabilitation of *amelogenesis imperfecta* in adults and children (13). Crowns are sometimes being used to pay for the tender enamel. Usually stainless steel crowns are used in children which may be replaced by porcelain when or once they hit maturity. In the worst case scenario, the teeth may get to be extracted and implants or dentures are required. This clinical report describes the treatment sequence of a 22-year-old individual with AI.

Case report

A 22-year-old male patient reported to the dental OPD of Navodaya Dental College & Hospital, Raichur, Karnataka, India with the chief complaint of esthetic and functional inadequacy of permanent teeth along with considerable tooth sensitivity. He was very self conscious about the appearance of his teeth. A detailed medical, dental and social history was obtained. The patient expressed extreme dissatisfaction with his appearance and his mother confirmed that the patient had been socially affected by this problem.

Clinical examination showed a convex profile. Intraoral examination revealed a constricted maxillary arch with 18 mm anterior open bite and pulp exposure was present in relation to maxillary central incisors. The enamel of all teeth was hypoplastic and yellow-brown in color. The surfaces of the teeth were rough and the enamel was either not visible or very thin over the crowns of all teeth (Figure 1).

The teeth were opaque, pigmented and had no carious lesions. There were areas where the enamel was missing, similar to hypoplasias, although it was hardened and did not break loose when probed. In spite of the loss of dental structure, there was no reduction in the vertical dimension. The dentin, where it was exposed, was brown and hypersensitive (Figures 2 and 3).

Posterior teeth on both the sides of the mouth were in a cross bite relationship (Figures 4 and 5).

Radiographically, the teeth showed normal pulpal morphology while the enamel appeared to be very thin. The enamel of the teeth appeared to have the same radiodensity as dentin. The roots of

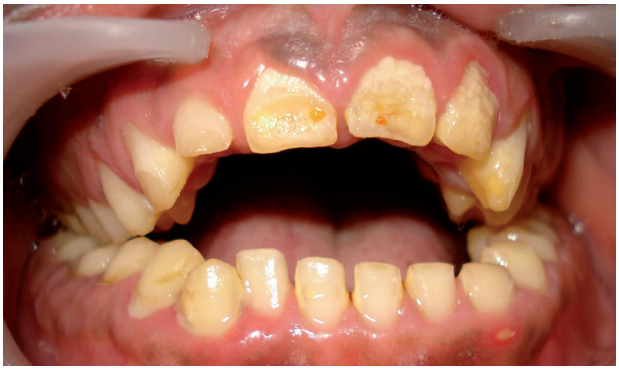

Figure 1 - Clinical appearance of *amelogenesis imperfecta* – labial view of maxillary and mandibular teeth

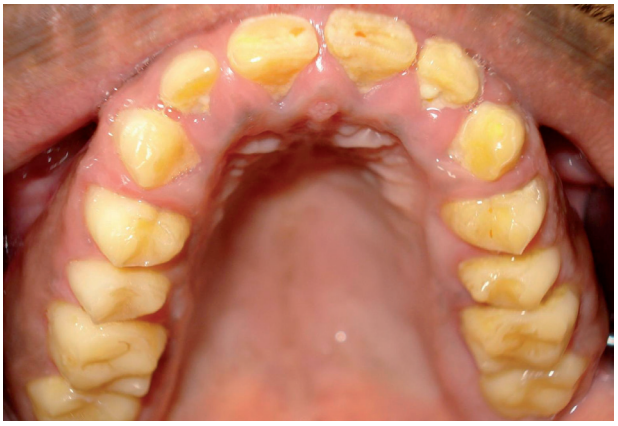

Figure 2 - Occlusal view of the maxillary teeth

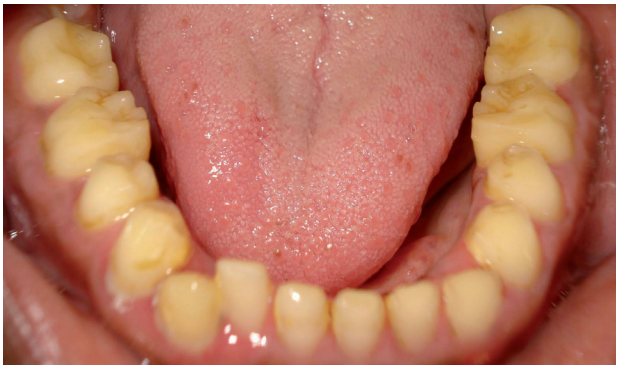

Figure 3 - Occlusal view of the mandibular teeth

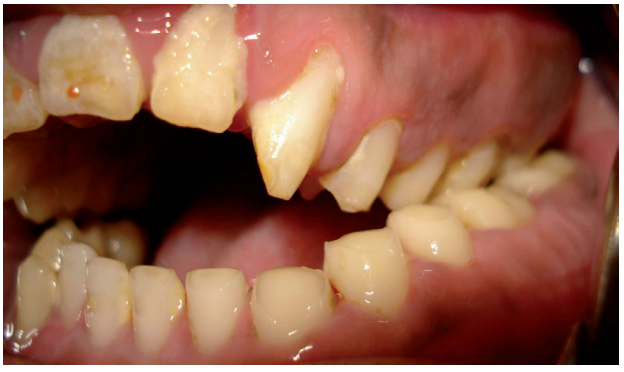

Figure 4 - Appearance of the mouth from the left side

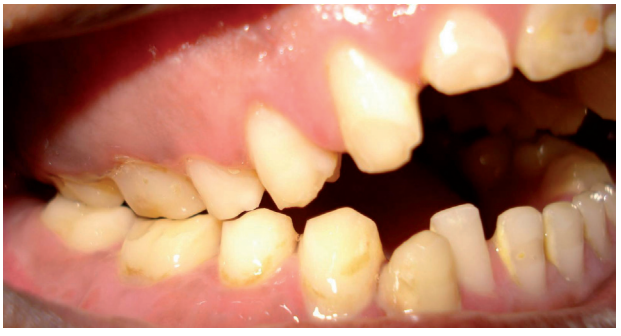

Figure 5 - Appearance of the mouth from the right side

all teeth appeared to be normal in shape and size in radiographic evaluation (Figure 6).

Intraoral x-ray images revealed the following findings. The enamel of the cusp was defective, the enamel radiopacity of the proximal surfaces was lower than normal, and the junction of enamel and dentin was obscure. There were no abnormalities in the condition of the root formation, the pulp chamber or the alveolar bone (Figures 7 and 8).

The patient's oral hygiene was unsatisfactory. The patient reported he was reluctant to brush because of the sensitivity of his teeth. No other abnormalities were observed. The patient was informed of the diagnosis and all the possible treatment options. The patient expressed little concern about the ultimate conservation of the teeth and was insistent on a full oral rehabilitation.

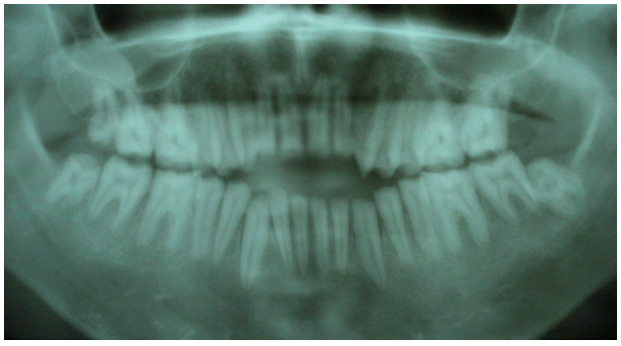

Figure 6 - Panoramic radiograph of the patient

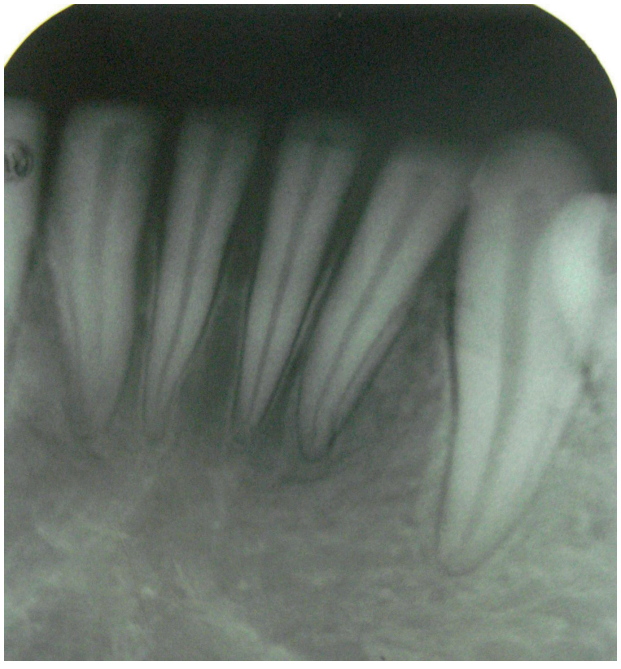

Figure 7 - Radiographic appearance of the mandibular permanent incisors

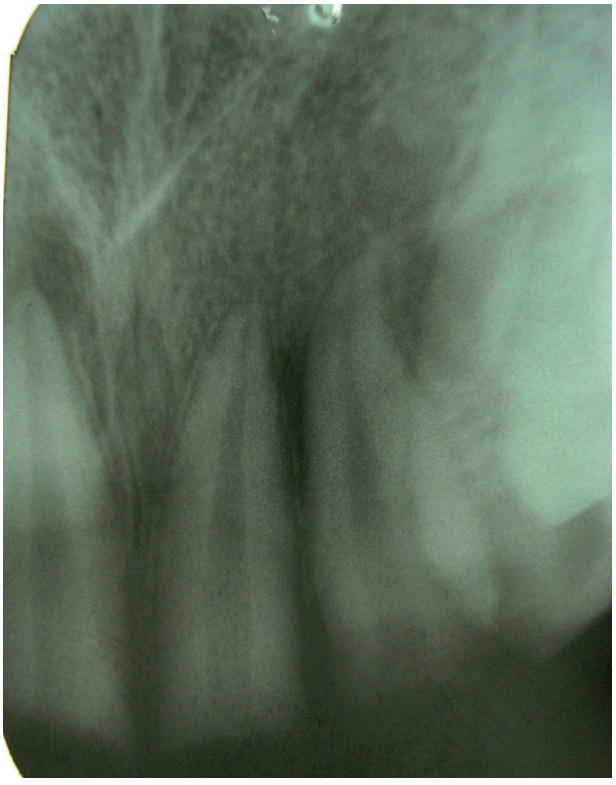

Figure 8 - Radiographic appearance of the maxillary permanent incisors

A treatment plan was developed with the following aims: to reduce the reported sensitivity of the teeth, to improve the esthetics and to restore masticatory function. Using diagnostic models, along with clinical and radiographic findings, the treatment plan was formulated. The patient was placed on an intensive oral hygiene program because of his inadequate oral hygiene. Following periodontal treatment, the patient was referred to Department of Conservative & Endodontics for pulpal management of maxillary central incisors and then to Department of Orthodontics for management of open bite and cross bite. Full mouth rehabilitation was considered after the orthodontic treatment.

Discussion

Enamel hypoplasia is considered to be due to local or systemic disturbances in the developmental enamel (14). Local factors causing enamel defects include trauma, inflammation, tumors of the jaws, and radiation hazards. The present patient had no

history of exposure to radiation or traumatic injury and no osteomyelitis or tumor of the jaw. Since enamel defects were present both in the deciduous and all permanent teeth, traumatic injury, apical periodontitis of the deciduous teeth, and inflammation were ruled out as causes. Systemic factors causing enamel defects include a variety of conditions: nutritional deficiencies, maternal disorders, perinatal and neonatal disturbances, infancy and early childhood disorders, genetic disorders, avitaminosis D and hypervitaminosis D, inborn errors of metabolism and endocrinopathies.

The differential diagnosis of defects in the dental enamel must be based on clinical and, if possible, laboratory data. This diagnosis can be the key to discovering genetic and systemic diseases, and also local aggressor factors that occur during dental development (15). Diagnosis and subsequent classification are mainly based on their morphological characteristics and on family inheritance (16) making routine clinical observations extremely important.

Although there are reports of a great tendency of AI patients to show impaction of permanent teeth and other anomalies associated with a delay in eruption, such as follicular cysts (17), radiographic images did not reveal any associated pathological alteration in this case.

The familial pattern of AI strongly suggests an X-linked inheritance, where all the male children of affected mothers will be affected. Affected women have a 50% probability of transmitting this inherited trait to their female offspring. The phenotypic manifestation of the hypomaturation form can differ according to sex. Males have teeth that are normal in shape and size, with irregular opaque white pigmentation and females can show discreet vertical bands of pigmentation of the enamel, although transillumination is necessary for it to show up (18).

In studies made of the prevalence of anterior open bite in patients with *amelogenesis imperfecta*, it was noted that it occurred in 24% in the affected group compared with only 2% in the general population. The coexistence of the two conditions can be due to a pleiotropic action of the AI genes, influencing the growth of the craniofacial skeleton (19). Disorders of the enamel epithelium can also cause alterations in the eruption mechanism, resulting in the anterior open bite (20). However, Witkop and Sauk (21) suggested that this malocclusion was of a dentoalveolar nature, due to the patient inserting his tongue in a reaction to protect against aggressor thermal stimuli, resulting in local interference that would prevent alveolar growth. In the present case, the patient – presenting AI and anterior open bite – did not mention any sucking habit or complaints of pain, which would call for a reaction to protect through tongue intervention, nor did he show tongue intervention when in repose. This almost rules out the possibility of the malocclusion resulting from a local mechanical interference.

Comparing the cephalometric radiographs of AI patients with a control group, other discrepancies in the facial skeleton are found, such as a reduction in the angle of the mandibular plane, increased anterior facial height and a reduction in the posterior portion of the skull base, suggesting that the open bite may be of skeletal origin and not due to a disorder in the dental eruption mechanism (20). Investigations into the embryonic development of the craniofacial complex suggest that this and the dental enamel have a common origin. In AI, therefore, it is possible that the gene acts in cells derived from the neural ridge, causing subsequent anomalies in the dental enamel and in the skull (19).

According to Seow (16), the main clinical problems of AI are esthetics, dental sensitivity, and loss of occlusal vertical dimensions. However, the severity of dental problems experienced by the patients varies with each type of AI. *Amelogenesis imperfecta* represents a group of hereditary alterations in human dental enamel which is of particular interest to the pediatric dentist. Due to the clinical implications resulting from this pathology, these professionals usually have the first contact with the patient and the opportunity to establish their diagnosis, thereby allowing prompt preventive treatment to be given and intercepting any aggravation of the clinical manifestations of this disorder.

There are several alternatives for treatment of AI. The most predictable and durable esthetic option is to restore the affected teeth with complete crowns. Though this treatment option is an invasive one involving removal of substantial tooth structure, it is still more conservative than other considered alternatives which involve extraction of remaining teeth and placement of removable prosthesis. This treatment option, however, requires meticulous oral hygiene.

Treatment is usually combined to meet biologic, restorative and esthetic requirements imposed by

short clinical crowns (22). Several materials and methods are available to the dental practitioner as far as restorative modalities are concerned. However, several limitations may exist in man of these modalities and hence should be critically reviewed prior to deciding a treatment plan (22, 23). The most important disadvantage of restoring with full veneer crowns is the aggressive removal of tooth structure (12). A more conservative option is ceramic veneers. But the esthetic and functional result in such cases is questionable (8). Developing proper occlusion is probably the most important factor in full mouth rehabilitation. Canine-protected occlusion was developed in the final restorations to decrease lateral forces on the posterior dentition (22, 23).

Several studies for AI patients have illustrated the use of composite resin restorations, sealants, and other bonded resins, polycarbonate crowns, stainless steel crowns, and space maintainers to restore a mutilated dentition that may only be the result of severe attrition.

Conclusion

The treatment of patients with AI should start with early diagnosis and intervention to prevent later restorative problems. The restoration of aesthetics and function in patients with *amelogenesis imperfecta* may be achieved with a dedicated team approach. The use of modern dental materials and a justifiable reliance on the predictable artistic abilities allows both aesthetic and durable restorations. This will boost the patient's self-confidence and also increases his responsibility to maintain adequate levels of oral hygiene.

Conflict of interest

The author declared no conflict of interest in the present manuscript.

Informed consent statement

The patient signed an informed consent, kept in the records in the archives of the Navodaya Dental College & Hospital.

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