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"Enamel Imperfection"....Perfection with Diagnosis & Management: A Case Report

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Abstract

Amelogenesis imperfecta (AI) is a term for clinically and genetically heterogeneous group of conditions affecting the dental enamel, occasionally in conjunction with other dental, oral and extraoral tissues. It is usually inherited either as an X-linked, autosomal dominant or autosomal recessive trait. The enamel may be hypoplastic, hypomineralised or both and affected teeth may be discolored, sensitive or prone to disintegration. Diagnosis is based on the family history, pedigree plotting, meticulous clinical and radiological observation. Dental radiographs of AI teeth provide important information to the clinician with respect to the degree of enamel mineralization to design an appropriate treatment plan. The treatment of patients with AI should start with early diagnosis and intervention to prevent latter restorative problems. Herein, we present a case report of hypomaturation amelogenesis imperfecta with oligodontia and taurodontism who were provided with functional and esthetic rehabilitation.

Keywords: Amelogenesis imperfecta; Enamel; Genetic; Hypomaturation.

Introduction

Amelogenesis imperfecta (AI) is a term for clinically and genetically heterogeneous group of conditions affecting the dental enamel, occasionally in conjunction with other dental, oral and extra oral tissues.[1] It is an exclusively ectodermal disorder, because the mesodermic components of the teeth are not altered. It can affect both the primary and permanent dentition. The prevalence of this condition is 1:718 to 1:14000, depending on the population studied. Its etiology is related to the alteration of genes involved in the process of formation and maturation of the enamel.[2] Its genetic inheritance pattern can be autosomal dominant, recessive, X-linked and sporadic.[3] The genetic origin of the autosomal forms is still unknown, although the cause of X-linked form is definitely related to defects in the amelogenin gene, which is the principal protein related to the formation of human dental enamel.[4] This genetic alteration can be divided into 3 main types: hypoplastic, hypo-calcified and hypo-maturation, according to the clinical characteristics of the enamel, which reflect the stage of formation at which the enamel was affected. Each type can be subdivided into subtypes depending on the mode of inheritance, as well as on the clinical and radiographic aspect of the enamel defect, although in some cases characteristics overlap, making classification difficult.[5] The hypoplastic types are characterized by a deficiency in the quantity of enamel, which can be expressed clinically through a fine enamel, or with grooves and pits on its surface. The hypocalcified types show enamel that has low mineralization, manifested clinically by pigmented, softened and easily detachable enamel. The hypomaturation types are associated with anomalies of the maturation stage during the formation of the enamel, resulting in opaque and porous enamel.[2,3]

This article describes a case of amelogenesis imperfecta hypomaturation type, associated

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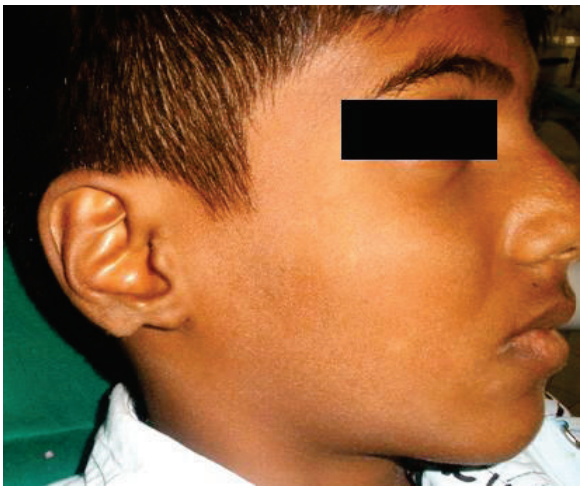
Figure 1: Right ear lobe showing bifurcation

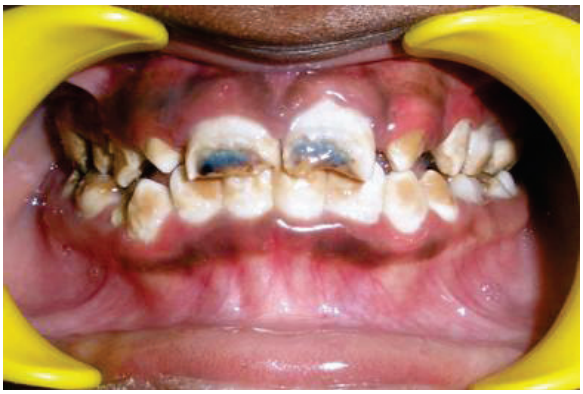


with various dental manifestations, clinical diagnosis and management to enhance the functional and esthetic harmony.

Case report

A 12 year old boy reported to the Department of Pedodontic & Preventive Dentistry with the complaint of pain in upper front teeth along with esthetic and functional inadequacy of his teeth. The patient resided in a non-fluoridated area since his birth. His maternal and postnatal history was not significant and there was no relevant family history. His dental history revealed restoration of mandibular right & left first permanent molar and maxillary right permanent molar. The patient's hair, skin and nails were normal. Patient's right ear lobule showed bifurcation (Fig 1). Intraoral examination showed mixed dentition phase with dental caries in relation

Figure 2: Dental caries in 11, 21 & Missing 12, 22



to 11 & 21, missing teeth 12, 22 (Fig 2) and retained deciduous teeth (65,75,85) (Fig 3 & 4). Both deciduous and permanent teeth were opaque, pitted, with yellow-brown pigmentations. The occlusal aspects of posterior teeth were devoid of enamel. The interdental contacts were preserved. The enamel was easily detached on probing in relation to 11, 21, 16, 26, 75, 85, 36, and 46. Panoramic radiograph (Fig 5) revealed multiple missing teeth (12, 22, 35, 45, and 28) and thin enamel contrasting little with underlying dentin. Taurodontism was found in relation to 16, 26, 36 and 46. Intraoral radiograph revealed thin enamel contrasting with underlying dentin and pulp involvement due to caries in relation to 11 and 21 (Fig 6). Histology of the extracted 65 under light microscopy (10X) showed extremely thin enamel and flat dentino-enamel junction. (Fig 7). It's also evident that increased number of dental lamella and irregular pattern of enamel rod arrangements due to decreased maturation

Figure 3: Retained 55

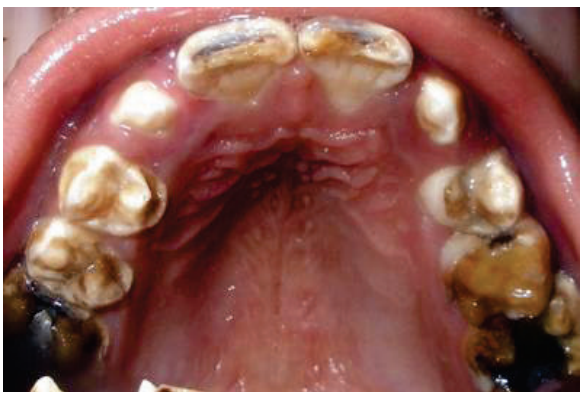


Figure 4: Retained 75, 85

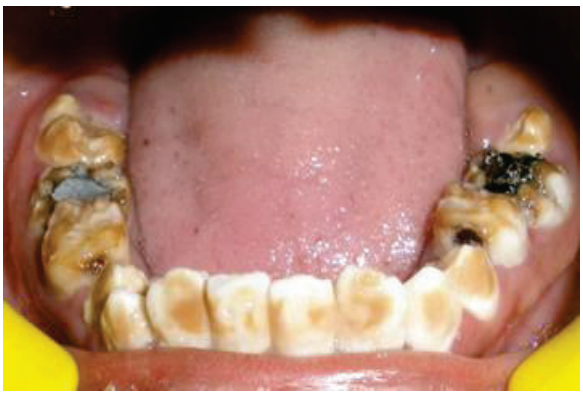


Figure 5: OPG Showing multiple missing teeth (12, 22, 35, 45, 28) & taurodontism in relation to 16, 26, 36, 46

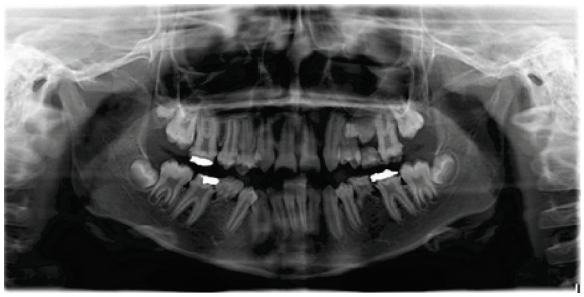


Figure 6: IOPA showing pulpal involvement of 11 & 21

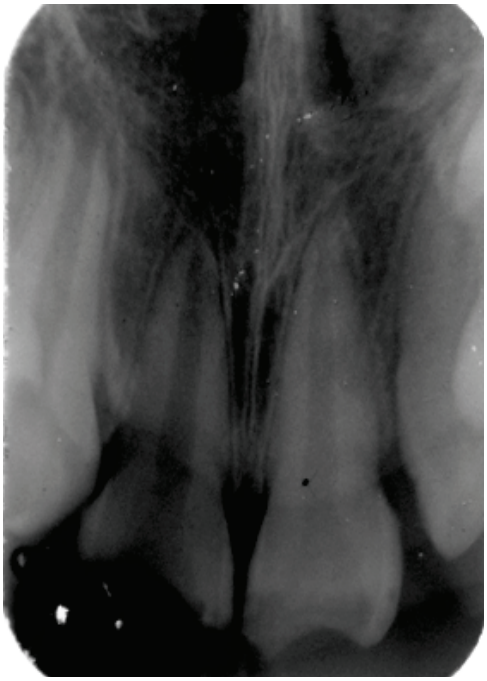


Figure 7: Histology of extracted 65 under Light Microscopy (10X) showing thin enamel and flat dentino-enamel junction

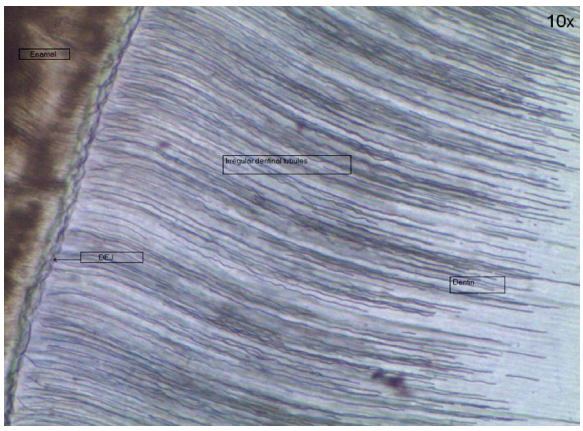


Figure 8: Histology of extracted 65 under Light Microscopy (10X) showing enamel lamella and irregular arrangement of enamel rods

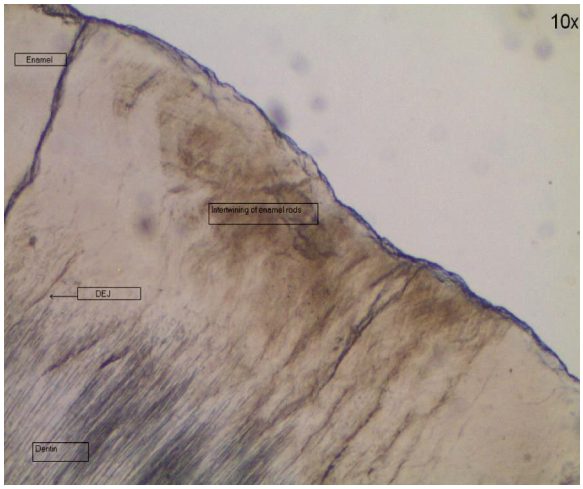


Figure 9: Crown placement in relation to 11, 21 and restoration of lower incisors with composites

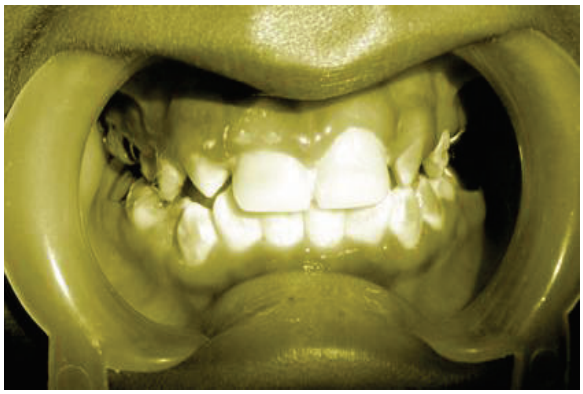


Figure 10: Stainless steel crown placement in relation to 16 & 26

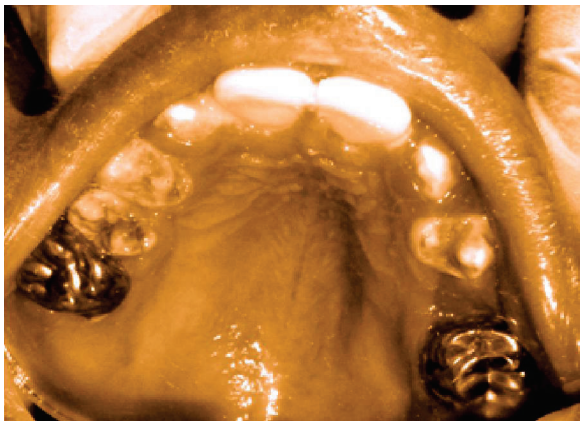
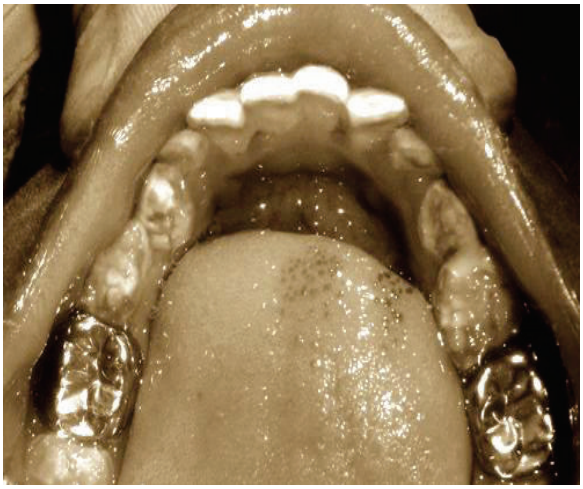


Figure 11: Stainless steel crown placement in relation to 36 & 46



were noted (Fig 8). Clinical, radiological and histological findings lead to the final diagnosis of hypomaturation type of amelogenesis imperfecta. Treatment included thorough oral prophylaxis, root canal treatment of 11 & 21 followed by esthetic crown placement, composite resin restoration of lower anteriors, stainless steel crown in relation to 16, 26, 36 & 46 and periodic topical fluoride application (Fig 9, 10, 11). The patient is presently under regular follow up since one year with emphasis on diet control and oral hygiene. The follow up period was uneventful with all the restorations being intact.

Discussion

Many classifications of AI have evolved since the original division into hypoplastic and hypocalcified types in 1945. Some have been classified exclusively based on the phenotype (appearance), others have used the phenotype as the primary discriminant and the mode of inheritance as a secondary factor in diagnosis. The most recent classification was given by Aldred *et al* in 2003 based on the mode of inheritance as phenotype (clinical and radiographic), molecular defect and biochemical result.[1]

The differential diagnosis of defects in the dental enamel must be based on clinical and, if possible, laboratory data. The diagnosis can be the key to discovering genetic and systemic diseases, and also local aggressor factors that occur during dental development.[6] Molecular and biochemical methods have shown differences in the protein content and in the composition of the enamel in the various types of AI.[7]Despite of these advances made in the diagnosis of this disorder, these sophisticated techniques are not routinely available. Diagnosis and subsequent classification are mainly based on their morphological characteristics and on family inheritance[2] making routine clinical observations extremely important.

The patient's mixed dentition showed general enamel defects, as much in their deciduous as in their permanent teeth, suggesting that this malformation did not occur due to aggressor agents during a certain period of formation of the dental germ. The clinical aspect can be suggestive of fluorosis, but the patient lived in an area where the fluoride levels in the water supply were below optimum levels (< 0.7 ppm F) and also from the mother's finding his primary dentition showed general enamel defects. According to Fejerskov *et al*,[8] primary teeth exhibit less dental fluorosis than their permanent successors. During pregnancy, the placental barrier seems to protect the foetus against excessive fluoride intake, even where the mother's intake is high. Only minor differences are seen in the fluoride level in blood of prenatal babies in high and low-fluoride areas. In a study by Fantaye *et al*,[9] no fluorosis noted in primary incisors, as enamel mineralization is completed more or less at the time of birth. In accordance to the above literature, fluorosis can be excluded as cause for the enamel alterations in these teeth. Furthermore, the patient did not show any systemic disease which could cause general enamel hypoplasia, such as renal or endocrine disturbances involving calcium metabolism.[2]

Although there was no relevant family distribution and he is the first person to be affected, the morphological characteristics of the enamel corresponds with the genetic anomaly known as hypomaturation AI. According to Seow (1993),[2] this AI variant shows fine enamel of an opaque white coloring

which can be detached. The interdental contacts are preserved and the radiographic image denotes thin enamel contrasting little with the underlying dentin. 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.[10] In accordance with these literature, the male patient in this report exhibited similar morphological and radiographical characteristics.

It is common for AI patients to receive little or no oral healthcare during childhood. Pitted enamel surfaces may predispose AI teeth to plaque accumulation, but spacing between the teeth may reduce the susceptibility to interproximal caries. In the ground section, the prominences of enamel lamella pave way for the progression of caries. In our patient, the presence of these features enhanced the caries attack. In a study by Collin *et al*, 22 AI patients were assessed for the prevalence of anomalies and observed 1 congenitally missing teeth, 6 showed delayed eruption, 9 crown resorption and 4 pulpal calcifications. The enamel density was also assessed and the mean density in hypocalcified AI group was lowest and the hypomaturation variety showed no significant changes on radiograph.[11] Non-enamel dental anomalies like taurodontism, elongation of pulp chamber due to apical displacement of root furcation and pulp calcifications occur with increased frequency in these patients when compared with their unaffected siblings.[12] Among the above mentioned oral features congenitally missing teeth, taurodontism were observed in our patient.

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. It should be managed by early vigorous intervention, both preventively and restoratively, with treatment continued throughout childhood and into adult life. In this patient, root canal therapy and esthetic crown replacement of the upper central incisor had been done. The lower anteriors were restored labially with composite resin. No tooth structure was removed in order to prevent unnecessary tooth sensitivity that may arise. The structural grooves were used as retentive element to aid adhesion of the restorative material. All the permanent first molars that had been restored before were protected with preformed metal crowns. Since he was in the mixed dentition stage and premolars, canine were still erupting, permanent restoration was not planned until complete eruption. Due to congenitally missing mandibular second premolar, its predecessor i.e., primary second molar is retained till natural exfoliation time to avoid space loss. Restorative treatment requires long- term analysis and good oral care practice. Hence a regular follow up is being maintained to check the longevity of the restoration and proper oral hygiene practice.

Conclusion

Amelogenesis imperfecta represents a group of hereditary alterations in human dental enamel is of particular interest to the pediatric dentist because, in addition to the clinical implications that may result from this pathology, we, are the first to 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.

Diagnosis is based on the family history, pedigree plotting, meticulous clinical and radiological observation. Dental radiographs of AI teeth provide important information to the clinician with respect to the degree of enamel mineralization to design an appropriate treatment plan. [13]Genetic research is presently only a research tool. Differential diagnosis includes dental fluorosis, enamel hypoplasia and molar incisal hypomineralisation (MIH). Hence there is a need to balance the decision for early intervention and long time survival of the restorations to prevent latter problems.

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