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Amelogenesis Imperfecta: A Case Report and Review of Literature A Genetic Teeth Discoloration

Sujatha D[1], Dhanya M[2], Kolappan R[3]
[1]Professor, [2,3]Post-Graduate Student, Department of Oral Medicine and Radiology, The Oxford Dental College and Hospital, Bengaluru, Karnataka, India

***Corresponding author**
Dr. Dhanya. M
Email: dhanya.mukesh1@gmail.com

Abstract: Defective enamel formation is the defects occurring at the stages of enamel formation. Amelogenesis imperfect (AI) is a hereditary disorder that causes developmental disturbances in the structure of enamel. Quantitative defects in matrix formation leads to hypoplastic form of Amelogenesis imperfecta. Inadequate mineralization of matrix leads to hypocalcification and hypomaturationvariants. It is very important to diagnose AI and provide functional and esthetic management of these patients to improve the oral health related quality of life.This paper describes a case of Amelogenesis imperfecta where both the father and son were affected with hypoplastictype.
Keywords: Amelogenesis imperfecta, Hypoplastic, Enamel, Hereditary.

INTRODUCTION

Amelogenesis imperfecta (AI) is a group of hereditary diseases affecting the tooth enamel in either quality or quantity and is associated with crown malformation and abnormal enamel density [1].

Enamel formation begins with a formative stage comprising of morphogenesis and differentiation of ameloblasts and secretion of organic matrix. The synthesis of full thickness of enamel is accomplished at this stage. Calcification stage starts when inorganic salts are deposited in the developing organic matrix. Inorganic component of enamel is increased at the expense of organic components and water. Maturation refers to the progression of enamel mineralization. Any disturbances during these developmental stages can result in abnormal enamel formation.

About 14 different hereditary subtypes of amelogenesis imperfect exist, with numerous patterns of inheritance and a wide variety of clinical manifestations. An ideal classification system for Amelogenesis imperfecta has not been established. The most widely accepted classification is the one given by Witkop, which relies on the phenotype and pedigree [2]. The clinical presentations of patients with AI depend on the type of AI involved. The prevalence varies from 1:718 to 1:14 000, according to the populations studied [3].

AI has been categorized as hypoplastic, hypocalcified, hypomaturation types and hypoplastic hypomaturation type. It is estimated that hypoplastic AI represents 60 to 73% of all cases, hypomaturation AI represents 20 to 40%, and hypocalcification AI represents 7% [4, 5].

Hypoplastic AI is characterized by inadequate deposition of enamel matrix. In hypoplastic form the affected dentition exhibit thin enamel with yellowish brown discoloration, rough or smooth and glossy, square shaped crown with lack of contact between adjacent teeth. The other clinical features include flat occlusal surfaces of the posterior teeth mainly due to attrition [6, 7].

Hypomaturation AI is characterized by a defect in the maturation of the enamel's crystal structure. Affected teeth are normal in shape but exhibit a mottled, opaque white-brown-yellow discoloration. The enamel is softer than normal and tends to chip from the underlying dentin. Radiographically, radiodensity of enamel is similar to that of dentin [6, 7].

Hypocalcified AI is characterized abnormal mineralization. In this type the teeth are appropriately shaped on eruption,but the enamel is very soft and easily lost and often becomes pigmented brown to black. Radiographically, in Hypocalcified form, thickness of enamel is normal but radiodensity of enamel is less than that of dentin [6, 7].

This paper describes a case of hypoplastic Amelogenesis imperfecta where both the father and son were affected, representing sex linked inheritance pattern.

CASE REPORT

A 7-year-old boy reported to the Department of Oral Medicine with a chief complaint of decayed teeth associated with pain since a week. On examination, the entire dentition appeared yellowish in colour. The patient's parents gave a history of teeth discolouration from the time the teeth erupted. His medical history was not significant and did not include systemic features of any of the amelogenesis imperfect-related syndromes. Patient's family revealed that the father and his elder sibling had a similar problem(Pedigree Chart Fig1).

Intraoral examination revealed mixed dentition with permanent maxillary and mandibular central incisors and first permanent molars. The teeth were yellowish brown in colour, with whitish chalky appearance in the cervical third(Fig2). Tapering of the coronal surface to the incisal edge was seen in relation to anterior teeth(Fig3). The teeth had rough surface and it was not brittle on probing.The patient's father also had similar yellowish discoloration of dentition which was slightly severe than the patient (Fig4). Deep dental caries was detected on mandibular left first and second primary molars.

Orthopantamogramshowed a full complement of developing permanent teeth. The teeth appeared deficient in the quantity of enamel(Fig5). On the basis of family history and clinicoradiographic features a diagnosis of hypoplastic type amelogenesis imperfect was given.

Since, primary mandibular left molar was decayed and showed physiological mobility; it was extracted and sent for histopathological examination. Cross-section showed lack of enamel mostly in the incisal edge area. As part of the treatment plan composite veneers were advised and to be changed to porcelain veneers or crowns at a later date. Preventive regime included regular fluoride treatment, sealants, dietary monitoring and regular reviews.

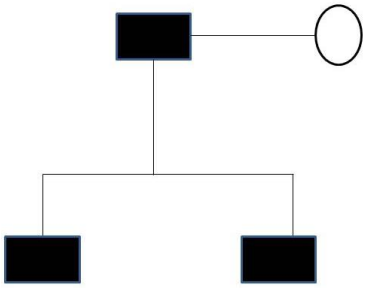

Fig-1: Pedigree chart showing the father and his two sons affected with amelogenesis imperfect

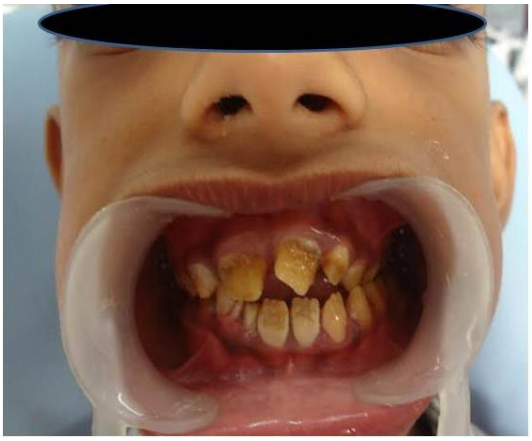

Fig-2: Diffuse yellowish brown discoloration seen on all the surfaces of all the teeth with whitish chalky apperarance in the cervical third

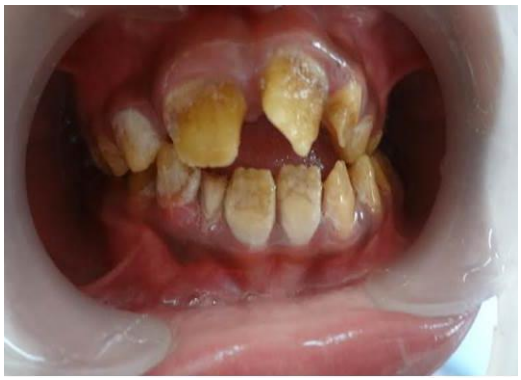

Fig-3: Tapering of the coronal surface to the incisal edge seen in relation to anterior teeth

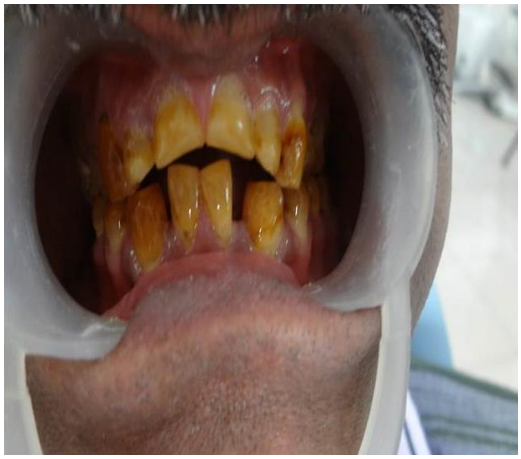

Fig-4: Yellowish brown discoloration of the father's dentition

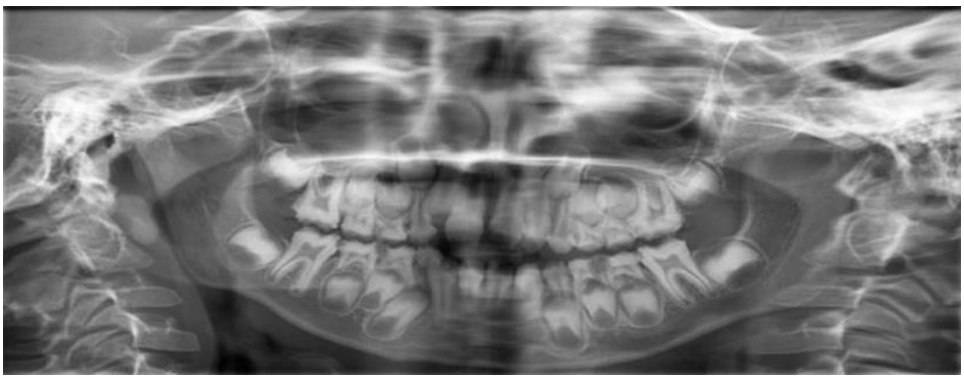

Fig-5: OPG showing a thin radiopaque enamel. There is obliteration of the pulp chamber especially in the posterior teeth

DISCUSSION

During organogenesis, the enamel changes from a soft and pliable tissue to its final form, that 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. Amelogenesis imperfecta encompasses a complicated group of conditions that demonstrate developmental alterations in the structure of the enamel in the absence of a systemic disorder [3].

The AMELX gene is associated with the enamel protein amelogenin, which constitutes up to 90% of enamel matrix. AMELX-associated variants of AI are X-linked with 14 different mutations currently known. Because of the effect of lionization, the male and female phenotypes are variable but often associated with the genotype. The male phenotypes include both the diffuse smooth hypoplastic as reported in the present case and it is also seen in hypomaturation variants [2].

The other genes which has been reported to be responsible for AI include ENAM gene,MMP-20 gene,KLK4 gene,DLX3 gene and AMBN gene [7].

Management for Amelogenesis imperfectapose great challenges to a dentist. Clinical problems associated are poor esthetics, chipping and attrition of enamel, exposure of dentine causing sensitivity, poor oral hygiene, gingivitis and dental caries.

Individuals with AI often concerned about poor dental esthetics. Clinically, they often present with a complain of tooth sensitivity and extensive tooth attrition. Hence it is necessary to provide appropriate dental treatments throughout the developmental stage. Proper oral hygiene and preventive treatments are essential so as to prevent caries, gingivitis, and calculus formation which may exacerbate existing problems. Cooperation and motivation of both the patient and parents are equally important for the successful outcome of AI management because the dental treatments can extend over many years and long-term success depends on regular attendance for dental procedures and the maintenance of optimal of oral health care [8].

The management of individuals affected by AI has been described as three stages inthe literature which include the temporary phase that is undertaken during the primary and mixed dentition period; the second stage is the transitional phase done after the eruption of all the permanent teeth and the last phase of treatment is the permanent phase which is undertaken in the adulthood [8].

The differential diagnosis of AI include fluorosis, tetracycline staining. In the present case, patient's family history ruled out fluorosis and tetracycline staining where tetracycline is deposited in hydroxyapatite crystals and chelation of calcium occurs resulting in yellow discoloration.

CONCLUSION

AI is a genetic developmental disorder that can affect the oral-health related quality of life and causes psychological concerns. The dentist should diagnose AI as early as possible in order to render proper and prompt treatment. Current literature pertaining to AI focuses on the molecular genetics. Oral rehabilitation can provide a good prognosis provided there are fewer complications. Patients should be counselled and motivated and must be taught how to maintain good oral hygiene, which goes a long way in maintaining the dentition.

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