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Case Report Open Access

Hypoplastic Smooth Autosomal Dominant Amelogenesis Imperfecta: A Case Report

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Abstract

Enamel Hypoplasia or Amelogenesis Imperfecta (AI) can be considered as the incomplete or defective formation of the organic enamel matrix of teeth. It is also considered as a complicated group of developmental disturbances that demonstrates structural defects and also affects the clinical appearance of enamel of the teeth. It is an exclusive ectodermic disturbance which is characterized by narrow horizontal bands, white flecks, lines of pits and grooves, showing yellow to dark brown discoloration of teeth. AI reduces oral health-related quality of life and causes some physiological problems and also affects the aesthetics of the individual. The present case shows one of the type of AI which was diagnosed on the basis of typical clinical and radiological features.

Keywords: Amelogenesis imperfecta; Enamel hypoplasia; Hypoplastic teeth

Introduction

Enamel Hypoplasia may be defined as an incomplete or defective formation of the organic enamel matrix of the teeth. When Hypoplasia is related to a hereditary cause it is called as Amelogenesis Imperfecta (AI) [1].

AI is a disorder of tooth characterized by developmental alterations in the structure of the enamel and affects all the teeth on both dentitions or acquired ones, involving one or more teeth. It is a complex inheritance pattern of dental enamel which causes teeth to be unusually small, discoloured, pitted and grooved [2-4].

It is an inherited disorder related to the alteration of the gene involved in the formation & maturation of the enamel [5]. The dentin and root of affected teeth are usually normal and these teeth are more resistant to decay [6].

The exact incidence of AI is uncertain but estimates in the population vary widely between 1:718 and 1:14,000 in western population [7].

Case Report

A 23 year old male patient presented with a chief complaint of yellowish brown discoloration of all the teeth since childhood. Past dental history revealed the same discolouration of his primary dentition. Familial history of his younger sister having the same problem was also noted. History did not reveal any eruption disturbances. From functional point he was unable to chew hard food. No systemic abnormalities were found. On an intraoral examination retained deciduous teeth, reduced enamel thickness and yellowish discoloration of permanent teeth was also appreciated. Enamel was chipped off i.e 22, 23, and 24 (Figure 1).

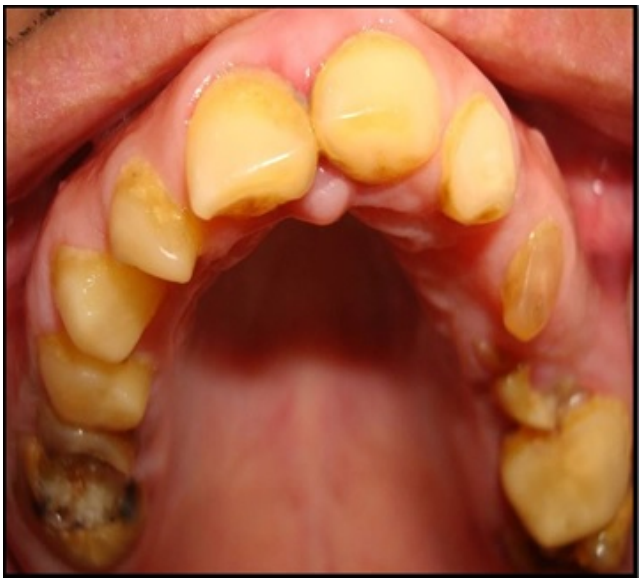

Figure 1: Showing yellowish brown discoloration with reduced thickness of enamel and smooth glossy surface.

The vertical height of the face seems to be reduced due to several losses of mandibular teeth. The enamel was found to be diffuse, thin, smooth, and glossy with anterior open bite and numerous open contact points. The colour of enamel varies from brown to yellow brown. Carious exposure was also appreciated i.e. 16 and 46 (Figure 2).

Radiographic analysis (IOPA) revealed thin radiopaque layer of enamel with normal pulp chamber, loss of cuspal height in all anterior and posterior teeth and open contact (Figure 3).

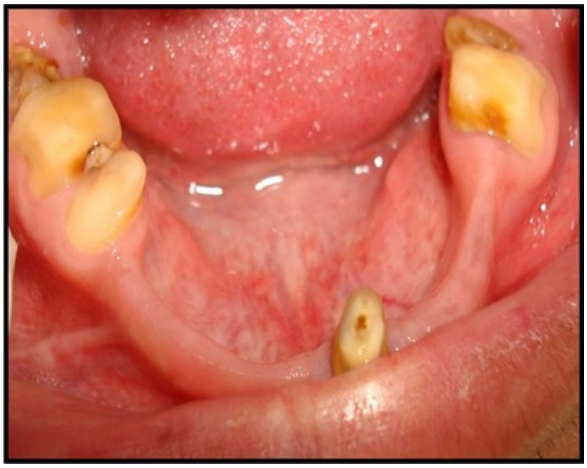

Figure 2: Showing tooth discoloration and missing teeth.

Presence of a thin layer of enamel, multiple impacted permanent teeth and few retained deciduous teeth were also noted on analysing panoramic radiograph.

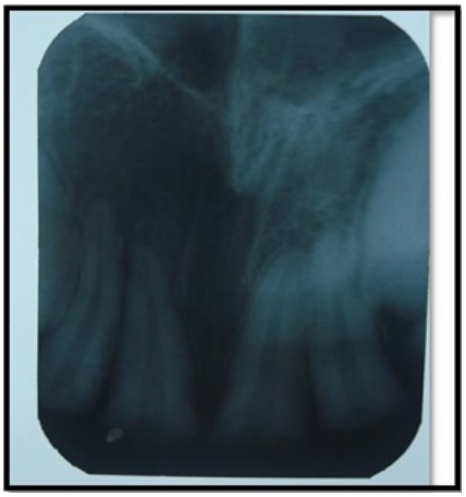

Figure 3: IOPA revealed that all the teeth had thin radiopaque layer of enamel with normal pulp chamber

The tooth specimen was sent for histopathological examination which showed enamel deficiency but normal complement of dentin and cementum was present. On correlating the history, typical clinical and radiographic features the diagnosis of Hypoplastic Smooth Autosomal Dominant Amelogenesis Imperfecta was given (Figure 4).

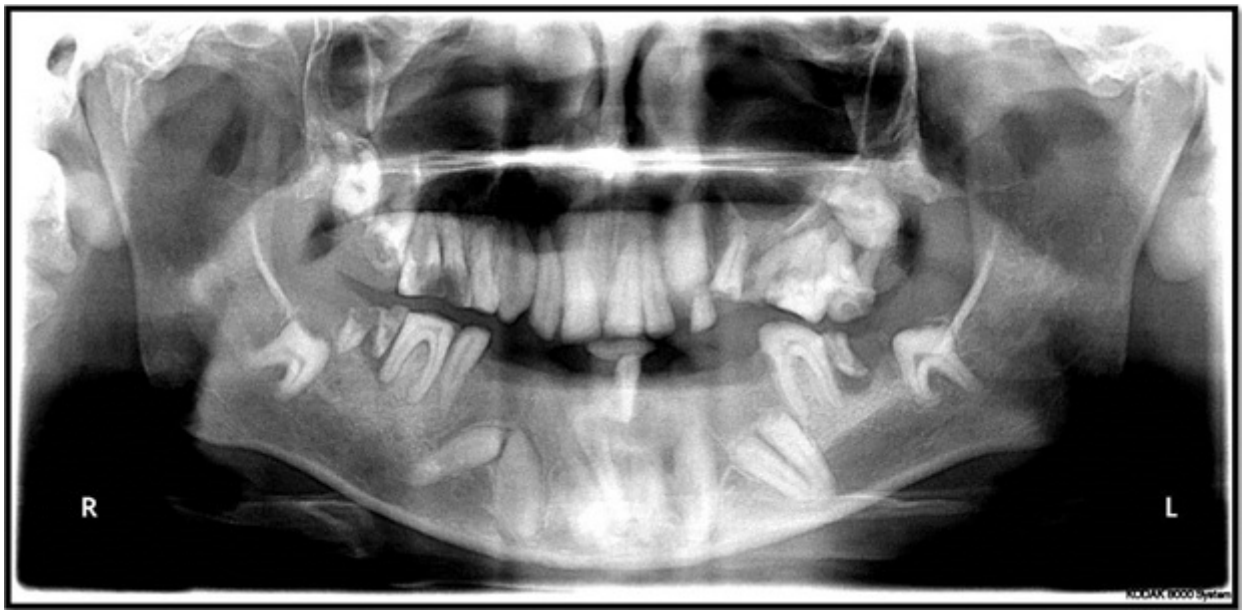

Figure 4: OPG revealed multiple impacted teeth present without enamel capping.

Discussion

Amelogenesis imperfecta is a hereditary disorder that interferes with normal enamel formation and related to the alteration of the gene involved in the formation and maturation of enamel. It has been found that AI is familial condition and can be inherited as an autosomal dominant, autosomal recessive, or x-linked dominant and x-linked recessive type [8,5].

AI can be classified into four patterns as hypoplastic, hypomaturation, hypocalcified, and hypomaturation-hypoplastic type [3]. This classification was based on enamel appearance and hypothesized developmental defects which was proposed in 1988 by

Witkop, and revised by Nusier in 2004 [8,3]. Clinical picture of AI is varied and also depends on the type of AI involved [3,8]. AI can be ruled out on the basis of clinical, radiographic, and histological appearance of the enamel [8,9]. Hypoplastic AI represents 60 to 73% of all cases, hypomaturation type represents 20 to 40%, and hypocalcification type represents 7% only [3,10].

In Hypoplastic type enamel is well mineralized but its amount is reduced. It can be further subdivided in rough and smooth types. Generally in hypoplastic variety enamel becomes thin and gives yellowish-brown discoloration of tooth. Texture of tooth becomes rough or smooth and surface appears glossy [3]. The shape of the crown becomes squarish with loss of contacts between adjacent teeth. The rough pattern of hypoplastic type exhibits thin, hard and rough surface of enamel. On radiographic analysis, presence of thin radiopaque layer of enamel with normal radiodensity was appreciated [9,11,12]. The clinical and radiographical appearance of the present case was consistent with the smooth pattern.

The Hypomaturation type of AI exhibits normal thickness of enamel and the enamel is comparably softer so it can be pierced by an explorer and can be lost by chipping away from the underlying dentin. Radiodensity of enamel is analogous to that of dentin which can be analysed radiographically. Histologically alterations in enamel rod and rod sheath structures can be appreciated [1,11,12].

The least common type of AI is Hypocalcified form which is characterized by pigmentation, enamel is so soft that it can be lost soon after eruption and leaves a crown composed of only dentin. On radiographic examination normal thickness of enamel is seen but radiodensity is reduced somehow to that dentin. Defects of matrix structure and mineralization is seen histologically [9,11,12].

According to Witkop classification there are mainly four types of AI. The fourth variety of AI is associated with syndrome like taurodontism and called as "hypoplastic-hypomaturation with taurodontism". This is seen in Tricho-Dento-Osseous Syndrome In this form the enamel is thin, mottled yellow to brown, and pitted. Molar teeth reveal taurodontism and other teeth have enlarged pulp chambers [1,8].

Molecular studies have shown that the aetiology of AI is related to mutation and alteration in genes like Enamelin (*ENAM*), Amelogenin (*AMELX*), Kallikrein 4 (*KLK4*), Matrix Metalloproteinase 20 (*MMP-20*), and Distal-less homeobox 3 genes (*DLX3*) which are involved in the process of formation and maturation of the enamel. The different pattern of inheritance corresponds with different genomic sites [13,14].

The success of AI cases depends on many multidisciplinary approaches. As the aesthetic appearance is the prime consideration of AI patient, the treatment planning depends on several factors like age, socioeconomic status, type and severity of the disorder etc. After analysing the benefits and limitations of each technique the successful treatment can be planned for AI patient [14,15]. Advance techniques and increase in availability of various dental material like glass ionomer cements, porcelain veneers, stainless steel crowns, composite resin veneers, lab-fabricated crowns, over dentures can be used to restore the affected teeth [16,17]. The treatment approach should consider the specific AI type and underlying defect. In the patients with hypoplastic AI, enamel is usually sufficient for bonding so composite resin restoration may be the treatment of choice as it masks discoloration and improves crown morphology [17].

In clinical practice it is difficult to differentiate between Amelogenesis Imperfecta and Enamel Hypoplasia. During diagnosing such cases clinician always keep in mind the differential diagnosis of Dental Fluorosis also. Apart from clinical picture the careful questioning and history of patient act as an important parameter to distinguish between these two conditions.

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