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Case Report of Rare Syndrome Associating Amelogenesis Imperfecta and Nephrocalcinosis

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Abstract

Amelogenesis imperfecta (AI) is a group of hereditary disorders that affect the quality and/or quantity of dental enamel. AI also named enamel-renal syndrome, is an extremely rare syndrome which is characterized by hypoplastic amelogenesis imperfecta (hypoplastic dental enamel) and nephrocalcinosis (precipitation of calcium salts in renal tissue). Patient presented oral clinicopathological manifestations, includes permanent dentition, alterations in the tooth shape, reduction of the enamel thickness and yellow discoloration with many teeth pulpal exposed, delayed tooth eruption, and intrapulpal calcifications. Nephrocalcinosis is often asymptomatic but can progress during late childhood or early adulthood to impaired renal function.

Key words: *Amelogenesis imperfecta, nephrocalcinosis, autosomal recessive.*

INTRODUCTION

Amelogenesis Imperfecta is a rare hereditary disorder that disturbs the normal 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 [1,2]. Dental enamel formation is a highly specialized process. The mechanisms of dental enamel formation requires specific all critical molecular participants [3]. defects in the genes encoding enamel proteins can cause syndromic and isolated AI. Identifying the genes that cause isolated AI narrows the focus to components that are most specialized for dental enamel formation. AI is most commonly has an autosomal dominant pattern of inheritance but autosomal recessive, sex linked and sporadic inheritance cases can also occur spontaneously in one or more members of the same family [4]. It affects both the deciduous and/or permanent dentition and amelogenesis imperfecta may affect all or only some of the teeth in the primary and/or permanent dentition [5]. AI also occurs as an integral and diagnostic feature of number of syndromes. Amelogenesis imperfecta associated with nephrocalcinosis primarily affects dental enamel and other finding like unerupted teeth, anterior open bite, pulpal calcifications, root and crown resorption, cementum deposition, truncated roots, and taurodontism have been reported [6]. Precipitation of calcium salts in the renal tissue is one of the important factors in nephrocalcinosis, which is associated with AI is a rare syndrome has been reported in just a few families [7].

CASE REPORT:

A 20 years old male patient referred to the department of Dentistry, BLDE University's Shri BM Patil Medical College & RC complains of discolouration, defective shape and size of teeth, sensitivity of teeth, with a history of involvement of both dentitions and delayed eruption of teeth. Patient suffered from pneumonia and urinary tract infection during his child hood age, also as positive family history to his grandfather defective teeth and renal problem. No history of consanguineous of the patient's parent marriage. Extra oral examination shows concave profile with competent lips. On intraoral examination shows the oral hygiene was good with mild marginal gingivitis. Dental examination shows all teeth in both the jaws are present. The color of the teeth was yellowish brown with altered shape and size with hypodontia was detected. Both jaw teeth shows gross attrition, and pulpal exposure in the posterior and lower anterior teeth (fig 1).
Radiovisiography and panoramic view, the bone structure and trabeculation were normal. The teeth number 18, 45 is impacted. The entire teeth shows altered crown pattern of crown with normal root appearing. Pulp chamber are large with pulp stones in most of the teeth seen. Multiple periapical abscess due pulp exposure is seen (fig 2&3). Renal ultrasound showed bilateral nephrocalcinosis (fig 4).

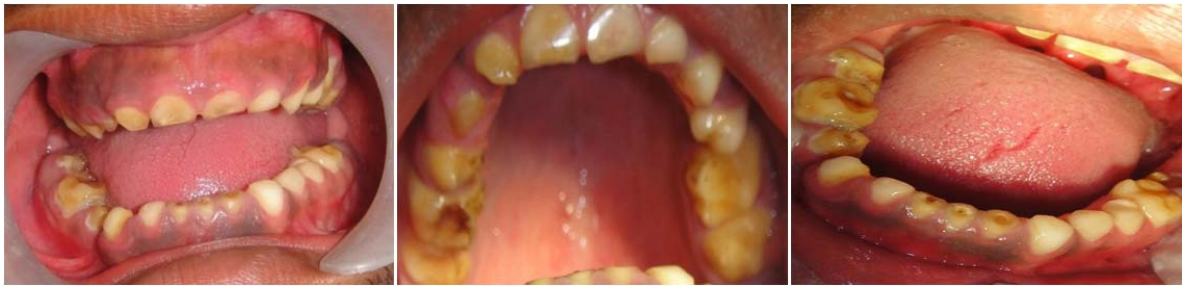

Figure 1. Clinical photograph showing all permanent dentition, alterations in the tooth shape, reduction of the enamel thickness and yellow discoloration with many teeth pulpal exposed.

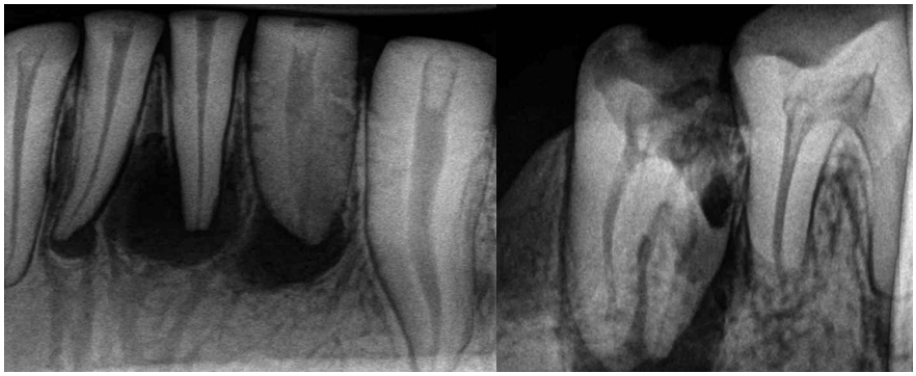

Figure 2. Radiovisiography shows reduced enamel, coronal intrapulpal calcifications, pulpal exposed, multiple periapical abscess

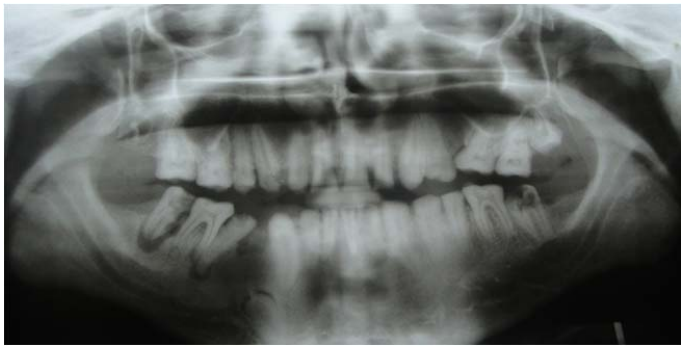

Figure 3 Panoramic radiographs showing all permanent teeth with coronal intrapulpal calcifications, multiple periapical abscess and 44 unerupted teeth had large well-defined pericoronal radiolucencies.

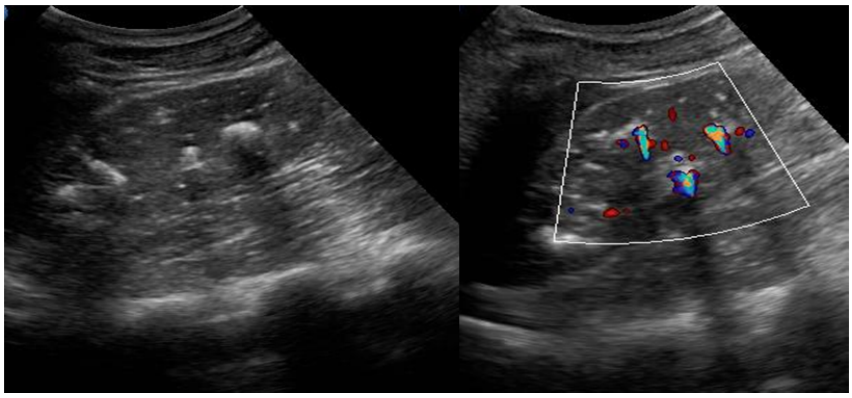

Figure 4. Renal ultrasound presenting nephrocalcinosis

DISCUSSION

AI is a serious of problems resulting in poor oral health-related quality of life. AI affects the structure and clinical appearance of the enamel of all or nearly all the teeth in a more or less equal manner [8]. The morbidity and severity of AI with nephrocalcinosis is more and most of the cases were detected at the age of adulthood [9]. In our case AI was detected and then ultrasound of kidney was done. Mac-Gibbon et al reported in these cases renal function was stable from birth till adulthood but the progressive renal failure was observed [10]. Lubinsky et al and Hall et al described AI and nephrocalcinosis in siblings with normal serum calcium and phosphate level. The AI and nephrocalcinosis syndrome has also reported in consanguineous and non-consanguineous families. The common characteristics are the presence of thin or absent enamel, presence of intrapulpal calcifications, delayed tooth eruption bilateral, nephrocalcinosis and normal plasma calcium [11,12]. To avoid morbidity with nephrocalcinosis associated with AI, kidney ultrasound should be performed in all AI patients. Witkop et al & Peter et al reported in these cases AI patients' histological appearance of teeth with hamartomatous pericoronal proliferation [13,14]. Most of the research showed the molecular basis of AI. X-linked genetic defects were identified in autosomal forms. Only mutations in the amelogenin (AMEL X) gene have associated with the various X-linked AI forms [15]. Hotton et al reported that tissue non-specific phosphatase alkaline (TNSALP), the calcium sensing receptor (CaSR) and calbindin 28 kDa are proteins involved in the calcium and phosphate metabolism. These TNSALP, CaSR, calbindin 28kDa are present both in the kidney and teeth, defective mutations in above gene and protein result in the hypophosphatasia characterised by defective bone and teeth mineralization [16].

In summary, further research is necessary to clarify genetic defect in this syndrome, with two uncommon conditions factors, AI and nephrocalcinosis. The early diagnosis provided by the oral symptoms leads to a better renal prognosis.

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