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An Anomalous Alliance- Amelogenesis Imperfecta and Nephrocalcinosis

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Abstract

Amelogenesis imperfecta is a diverse group of hereditary disorders that affects the quality and quantity of dental enamel. Amelogenesis imperfecta, commonly described as an isolated trait, may be observed with a number of variable dental or systemic disorders. The relationship between Amelogenesis Imperfecta and Nephrocalcinosis was first given by MacGibbon in 1972 and entitled as 'Enamel Renal Syndrome'(ERS). The recessive mutation in FAM20A gene is responsible for this syndrome. We report a case of Enamel Renal Syndrome and thereby highlight the importance of recognizing this possible association at an early stage.

Keywords: Amelogenesis Imperfecta, Enamel renal syndrome, Nephrocalcinosis, Nephrolithiasis

Introduction

Amelogenesis imperfecta is a varied group of rare inherited disorder affecting both the primary and permanent dentitions. Based on the phenotypic nature of observed enamel defect, it may be differentiated in three main groups: Hypoplastic(HP), , Hypocalcified(HC),Hypomaturation (HM).[1] Each main clinical group is further divided into subgroups based on the mode of inheritance and clinical appearance of defective enamel.

Amelogenesis imperfecta, usually occurred as an isolated trait, but can be associated with number of varying dental and/or systemic disorders, such as nephrocalcinosis in Enamel Renal Syndrome or gingival hyperplasia in Amelogenesis Imperfecta and Gingival Fibromatosis Syndrome.[2]

The first case of this rare association between AI and nephrocalcinosis was reported by MacGibbon in 1972. This rare association has been referred as "Enamel Renal Syndrome", amelogenesis imperfecta and nephrocalcinosis syndrome, amelogenesis imperfecta and gingival fibromatosis syndrome, or Lubinsky-MacGibbon syndrome. Affected patients generally suffer from distinctive oro-dental and systemic features. Dental phenotype consisting of generalized hypoplastic amelogenesis imperfecta, yellow-brown discoloration, delayed eruption of permanent teeth. Systemic features in this syndrome include nephrocalcinosis, nephrolithiasis in enamel renal syndrome or gingival hyperplasia in gingival fibromatosis syndrome. This distinct functional and aesthetic problems in this syndrome leading to an inferior quality of life in. The renal findings in this type of association ranging from nephrolithiasis, nephrocalcinosis to chronic renal failure.[3]

This article depicts a case report of 17-year-old female patient with multiple over-retained deciduous teeth, unerupted permanent teeth associated with nephrolithiasis.

Case Report:

A 17-year-old female reported to the department of oral medicine and radiology with a chief complaint of yellowish discoloration and delayed eruption of

permanent teeth. Medical history was non-contributary. Family history revealed history of consanguineous marriage (parents were second degree cousins). On general physical examination patient was moderately built and well-nourished. The extraoral were apparently normal.

Clinical Findings:

Intraoral Examination

Soft Tissue Examination:

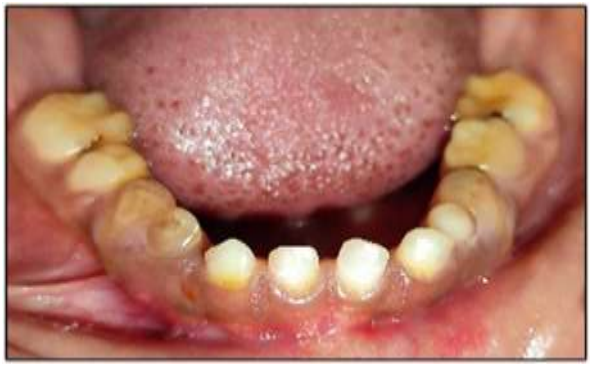
1. Pigmented gingiva with normal contour (Fig1A)

Hard tissue examination: Multiple over-retained deciduous teeth and unerupted permanent teeth with atypical findings as described below:

1. Relative microdontia with a yellowish-brown discoloration of both primary and permanent teeth (Fig A)
2. Translucent primary teeth showing marked attrition (Fig A)
3. Reduced vertical dimension at occlusion due to severe attrition (Fig1 A)
4. Esthetic thing: Gummy smile(Fig1A)
5. Both deciduous and permanent molars had flat occlusal surfaces(Fig1B)

Fig 1A: Pigmented gingiva and decreased vertical dimensional at occlusion

Fig 1B: Microdontia with yellowish-brown discoloration



Significant Radiographic findings: Cone Beam Computed Tomography (CBCT) revealed

1. Unerupted canine, premolars, second and third molars in both the jaws
2. Thickness of enamel was reduced
3. Pericoronal radiolucencies with sclerotic border was noted around non-erupted teeth
4. Complete root formation with radicular dilaceration noted with distal root of mandibular permanent second molars of both the sides
5. Multiple pulp stones in the pulp chamber

Volume 7, Issue 2; March-April 2024; Page No 240-245

Fig 2 A: Multiple impacted teeth, pericoronal radiolucencies

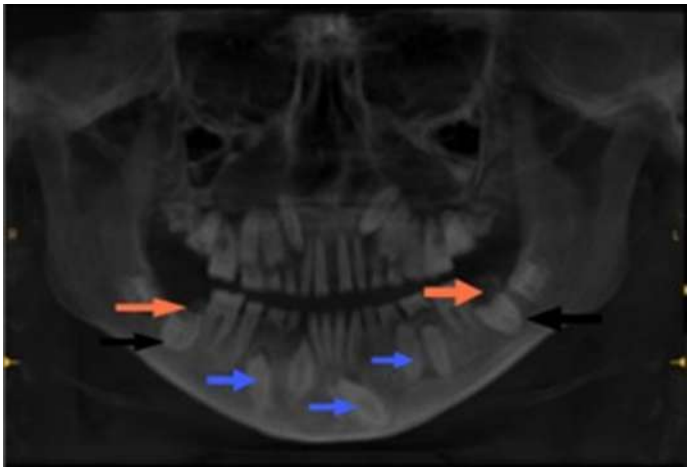
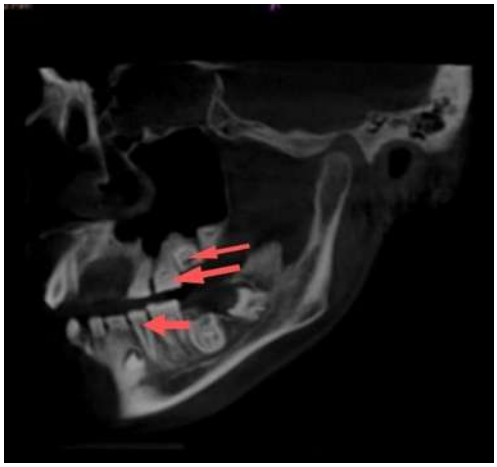


Fig 2B: Multiple pulp stones and root dilaceration



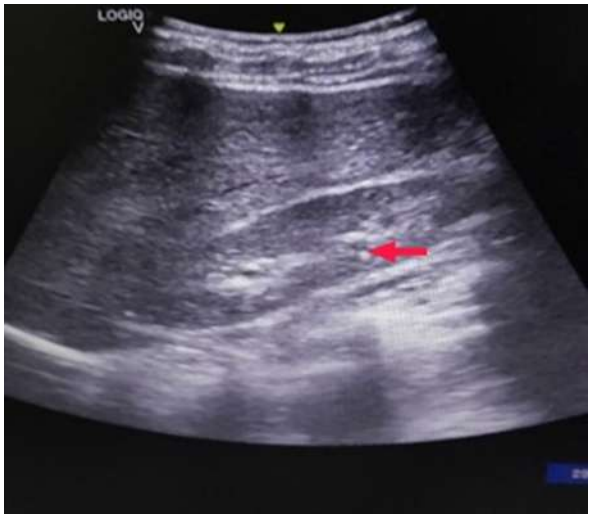
The provisional diagnosis of Amelogenesis Imperfecta were given based on the clinical and radiographic findings.

These findings fulfil the oral diagnostic criteria given by De la Dure-Molla et al. for ERS and further investigation (USG) were done to confirm the same.[2]

An ultrasound examination were performed, which demonstrated multiple hyperechoic foci in the medulla of right kidney with largest measuring 6mm, suggesting nephrolithiasis.

Volume 7, Issue 2; March-April 2024; Page No 240-245

Fig 3: Hyperechoic foci in the medulla of right kidney



Laboratory findings:

A 24-hour urine spectrophotometric analysis revealed very low calcium level to 1.71mg/dl (Average range 5-30mg/dl) indicating hypocalciurea.

Other biochemical and haematological findings were within normal range (Serum sodium 137mEq/lit, serum potassium 4.89mEq/lit, serum chlorides 101mEq/L, Alkaline phosphatase 105U/L, Urinary sodium 121mmol/L, Urinary potassium 56.65mmol/L. blood urea 17 mg/dl, serum creatinine 0.7 mg/dl).

Thus, based on previous findings, the diagnosis was modified to enamel renal syndrome with associated amelogenesis imperfecta, nephrolithiasis, and hypocalciurea.

Discussion:

Amelogenesis imperfecta (AI) is a diverse group of hereditary disorders that affects the quality and quantity of dental enamel. Most of the cases are inherited, either as an X-linked, autosomal dominant or autosomal recessive trait, and there is a possibility that this condition can occur spontaneously in one or more members of the same family. Amelogenesis imperfecta can occur as an isolated trait or in association with multiorgan syndromes such as, cone rod dystrophy, platyspondyly, nephrocalcinosis, hypothalamo–hypophyseal insufficiency and Kohlschutter syndrome. The association of Amelogenesis imperfecta and nephrocalcinosis syndrome has been reported in few families. There have been reported cases of these syndrome in consanguineous families, which suggests an autosomal recessive pattern of inheritance.[4]

The first report of this syndrome was described by MacGibbon in 1972 in a pair of siblings, The sister being diagnosed as having Amelogenesis Imperfecta and nephrocalcinosis syndrome by screening after her old brother died at the age of 26 as result of complications arising from nephrocalcinosis. The sister developed multiple urinary infection, hypertension and renal failure. The characteristics common to all cases appears to be multiple impacted teeth, delayed eruption of permanent teeth, yellowish discolouration of both primary and permanent teeth with thin or absent enamel, bilateral nephrocalcinosis in medullary distribution, normal plasma calcium, 25(OH) vitamin D3, and alkaline phosphatase.[5]

This rare combination of Amelogenesis imperfecta and nephrocalcinosis (NC) may be due to contiguous gene syndrome or pleiotropism. Jaureguiberry et al. identified autosomal recessive FAM20A mutations as the cause of ERS. This FAM20A gene affects the process of mineralization by inhibiting the process in some organ while allowing it to occur in bones and teeth. The homozygous mutation in FAM20A leads to increase in the promoter activity and decrease of the inhibitory activity on the growth of oxalate crystals, so as to allow mineralization of the gingiva, kidneys, lungs, and dental follicles.

The main laboratory alteration in ERS were: Hypocalciuria, elevated serum creatinine level,

Volume 7, Issue 2; March-April 2024; Page No 240-245

reduced phosphate excretion and hypocitraturia. These alterations predispose to the formation of stone formation or nephrocalcinosis. [6]

I.A Roomaney et al describe the case series of four unrelated patient with ERS from Sub-Saharan Africa with notable addition of some features such as odontoma, pneumatization of frontal sinuses, thinning of alveolar ridges, enlarged inferior turbinate and thinning of cortical of condyle.[7]

V manoj et al reported a case of ERS with sialolith of submandibular gland duct which was the first case in literature.[8]

Sabina Pena B et al, reported a case of ERS in two patients which exhibited two additional undescribed clinical features such as hypertrichosis and hearing loss. The presences of hearing loss and hypertrichosis suggest a putative function of FAM20A in cells of cochlea and epithelial cells of hair follicles, supporting future studies of FAM20A in the physiological and pathological conditions involving inner ear and hair development.[9]

More recently a case of Amelogenesis imperfecta, cleft lip and palate and polycystic kidney disease was described by Suda et al., in a consanguineous family and in this case nephrocalcinosis was secondary to polycystic kidney disease.[10]

Clinical diagnosis is based on the association of oro-dental features and renal findings. Whilst the oral phenotype is evident in childhood, the renal involvement is clinically silent at this age and requires further investigation for detection therefore oral phenotype is pathognomic and sufficient to clinically diagnose ERS even in absence of other co-segregating problems.

Conclusion:

Unrecognized and untreated nephrocalcinosis is associated with significant morbidity and for this reason children with apparently Autosomal recessive Amelogenesis Imperfecta should at least have a renal ultrasound performed to exclude such pathologies. Also, an adequate knowledge of diseases that involve both dental and medical factors is indispensable for making the correct diagnosis and ensuring comprehensive treatment. The present cases bring forth the necessity of a thorough medical history and systemic examination, including renal ultrasound and renal function tests in all patients with Amelogenesis imperfecta.

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Volume 7, Issue 2; March-April 2024; Page No 240-245

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Volume 7, Issue 2; March-April 2024; Page No 240-245