Case Report

Enamel renal syndrome: A case report with review of literature

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Abstract Amelogenesis imperfecta (AI) is a developmental, inherited disorder affecting dental enamel. Preterm and low birth weight children are prone to many serious medical problems during the neonatal period, which may affect the development of oral tissues. We report a case of AI presenting with renal disease and thereby highlighting the importance of early diagnosis of this possible association to prevent renal failure and death of the patient.

Keywords: Amelogenesis imperfecta, enamel renal syndrome, nephrocalcinosis

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INTRODUCTION

Tooth development involves different molecular processes that are very critical at different stages. Genetic diseases that disrupt key processes during tooth formation result in dental phenotypes characteristic of when and where the defective gene is normally expressed. Such disruptions during crown formation lead to the inherited defects of enamel and dentine.[1]

Developmental defects of enamel are regions of enamel with altered quality and quantity as a consequence of insults to the enamel organ at the time of enamel formation.[2] Amelogenesis imperfecta (AI) describes a group of inherited disorders that affects the quality and quantity of primary and/or permanent enamel and which may be associated with morphologic or biochemical changes elsewhere in the body. AI may affect all or only some of the teeth in the primary and/or permanent dentition.[3]

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Preterm births which occur before 37-week gestation comprise approximately 6% of all live births in developed Western nations. Changes in dental enamel are one of the most noticeable oral effects of preterm birth and may classically present as enamel hypoplasia which is defined as a quantitative loss of enamel, or as enamel opacity which is defined as a qualitative change in the translucency of the enamel.[4]

The combination of AI and nephrocalcinosis (NC) may suggest a contiguous gene syndrome or pleiotropism. One hypothesis suggests that there is an underlying abnormality in the interstitial matrix, which leads to dystrophic calcification in the kidney and abnormal enamel production in the teeth. Another hypothesis suggests that many of the dental proteins that were thought to be tissue-specific may also be traced in nondental tissues and the role of these proteins in

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calcium and phosphate metabolism and renal function needs further research. The genetic basis of AI and NC syndrome is yet to be established.[5]

CASE REPORT

A 21-year-old male patient presented with a chief complaint of discolored and sensitivity of teeth. The patient did not report any abnormal oral habits. The findings of his general physical examination were normal. There was no contributing family history. Medical history revealed preterm birth and renal disease. An intraoral soft tissue examination showed the gingiva with a normal contour. A hard tissue examination revealed retained deciduous teeth and permanent teeth with atypical findings [Figures 1 and 2]. Some of the permanent teeth were widely spaced, with a yellowish brown discoloration. The enamel layer was very thin clinically and posteruption wearing of the teeth was evident. The upper central incisors were severely attrited and the molars had flat occlusal surfaces. An ultrasound examination of his kidneys demonstrated multiple hyperechoic foci in the medulla of both the kidneys, suggesting bilateral NC [Figures 3 and 4]. Based on the clinical findings, provisional diagnosis of enamel renal syndrome was made with differential diagnosis of mottled Enamel and Barbet-Biedly syndrome.

Root canal treatment for multiple teeth was carried out, following which prosthesis was given. Retained deciduous teeth were extracted. Postoperative 6 months follow-up of the patient revealed no sensitivity in the treated teeth.

DISCUSSION

Enamel is an ectodermal protective covering of the teeth. Fully formed enamel is the most highly mineralized extracellular matrix known, consisting of approximately 96% mineral (hydroxyapetite calcium phosphate crystals) and 4% organic material and water.[1] Tooth enamel develops in three major stages: matrix deposition, initial mineralization and final maturation. Ameloblasts are

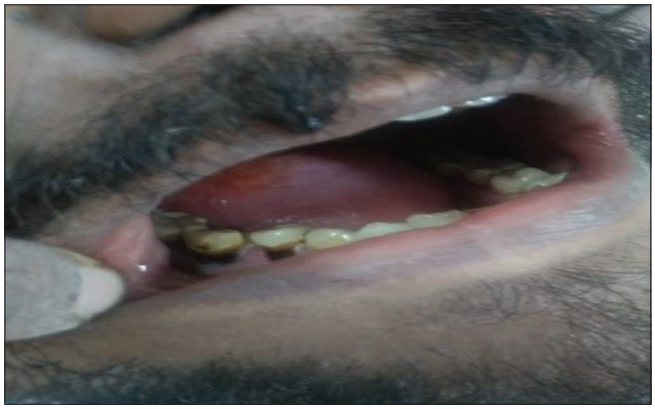

Figure 1: Intraoral picture revealing snowcapped teeth (enamel hypoplasia) with exposure of dentin

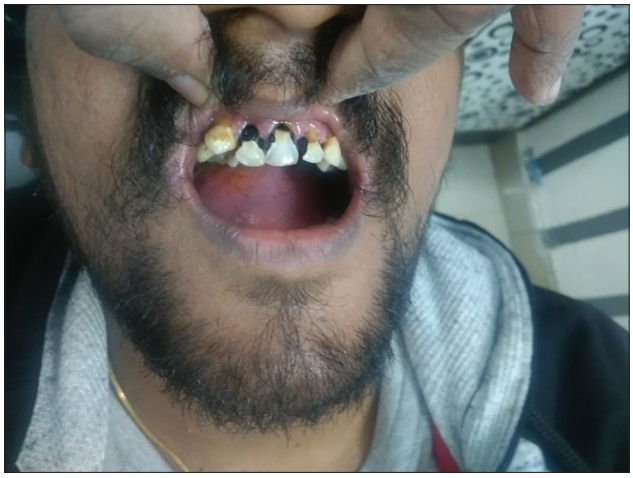

Figure 2: Anterior teeth showing enamel hypoplasia with exposure of dentin

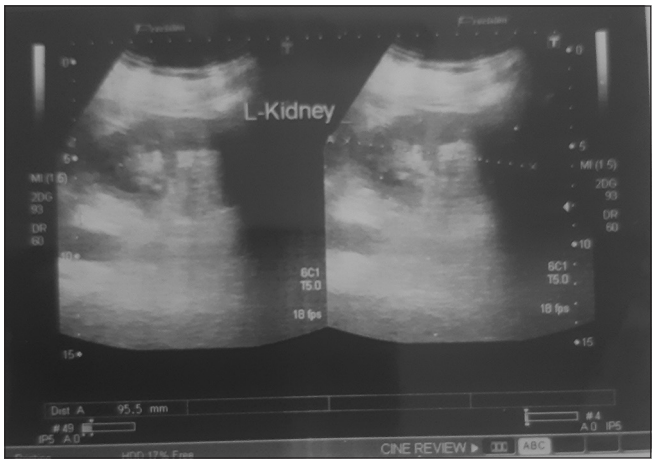

Figure 3: Left kidney showing calcinosis

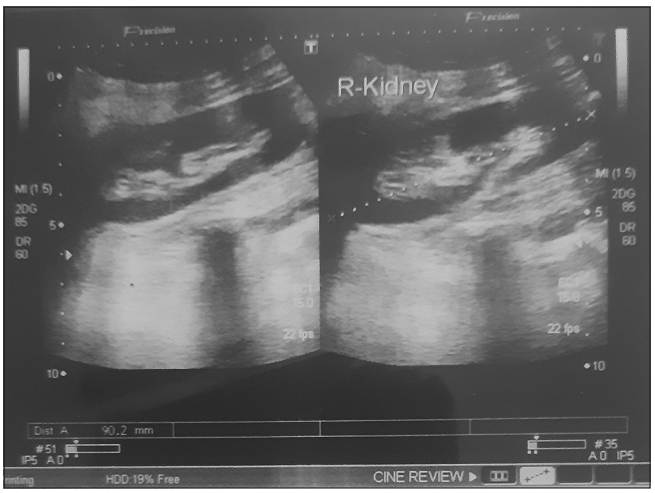

Figure 4: Right kidney showing calcinosis

extremely sensitive to external influences, with more than ninety different factors known to be associated with disturbance in enamel formation. Although the causes are diverse, the most commonly reported factors include serious disease during the first 3 years of life, hypocalcemia, renal disorders, nutritional deficiencies and viral infections associated with high fever.[6]

AI is a group of inherited anomalies of dental enamel. Since its first classification as hypoplastic and hypocalcified type in 1945, several classifications have evolved. Four major types were recognized based on phenotype (hypoplastic, hypocalcified, hypomaturation and hypomaturation–hypoplastic) and then subdivided into 15 subtypes based primarily on phenotype and secondarily on the mode of inheritance.[7]

Conditions associated with AI are Tricho-Dento-Osseous Syndrome, Cone-Rod Dystrophy and AI, Kohlschutter Syndrome, McGibbon Syndrome, Vitamin D deficiency rickets, Auto Immune polyendocrinopathy. AI with NC or McGibbon Syndrome is rare and possibly under-diagnosed. The first report of this syndrome was in 1972 by McGibbon. The common characteristics are, the presence of enamel agenesis, intrapulpal calcifications, delayed tooth eruption, and unexplained bilateral NC. The condition is usually asymptomatic in the early stages. Patients may present with episodes of urinary tract infections, hematuria, polyuria, and polydipsia. The major long-term complication in patients with medullary NC is renal failure.[1]

The present case describes the occurrence of AI in the association of renal diseases. The case is presented with bilateral medullary NC but did not present with renal failure. NC, which is the precipitation of calcium salts in the renal tissue, is frequently associated with chronic renal failure and renal tubular acidosis (RTA). RTA is a group of disorders in which renal excretion of acid is reduced out of proportion to any reduction of glomerular filtration rate, and as a result metabolic acidosis sets in. Medullary NC is usually associated with RTA as either its cause or consequence.[8] Kalyvas *et al*. in their review of literature listed 16 cases of jaw enlargements in dialysis patients, of which 14 patients presented with diffuse swelling of both the jaws and three patients had localized swelling of the mandible.[8] In our case, the enamel alterations were generalized affecting both lower and upper teeth.

The combination of AI and NC may suggest a contiguous gene syndrome or pleotropism. One hypothesis suggests that there is an underlying abnormality in the interstitial matrix, which leads to dystrophic calcification in the kidney and abnormal enamel production in the teeth. The involvement of two separate but closely linked genes has also been suggested. Another hypothesis suggests that many of the dental proteins that were thought to be tissue-specific may also be expressed in nondental tissues and the role of these proteins in calcium and phosphate metabolism and renal function needs further research. The genetic basis of AI and NC syndrome is yet to be established.[7]

Our case also revealed a history of preterm birth. It was previously thought that enamel defects were limited to the primary dentition only as the permanent teeth have not yet begun their formation at the time of the preterm birth. However, recent studies have indicated that the effects of birth prematurity may extend into the permanent dentition as well.[4]

CONCLUSION

Our case is illustrate that AI can be associated with renal disease hence, it is mandatory to alert the dentists who see patient with hypoplastic AI, that they must obtain medical history with particular reference to renal system and should refer the patient for medical examination including renal function tests and ultrasonography to detect NC. It is very important because of the morbidity associated with unrecognized and untreated renal disease. The early diagnosis provided by the oral symptoms leads to a better renal prognosis.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient (s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initial s will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of interest

There are no conflicts of interest.

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