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CASE REPORT

Enamel Renal Syndrome in a Bahraini Child: A Rare Case Report

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Abstract

Enamel renal syndrome is considered a rare phenomenon found in patients diagnosed with amelogenesis imperfecta when it is associated with nephrocalcinosis, resulting in severe renal damage as age progresses. We report the case of a patient with typical features of amelogenesis imperfecta and nephrolithiasis. This case presentation highlights the role of a general dental practitioner in the recognition of such unique syndromes.

Keywords: Amelogenesis Imperfecta; Nephrocalcinosis; Amelogenesis Imperfecta Nephrocalcinosis; Renal Insufficiency; Kidney Calculi.

Introduction

Amelogenesis imperfecta (AI) is a developmental condition that results in defective structure and clinical appearance of the enamel, associated with mutations in *AMEL* (amelogenin), *ENAM* (enamelin), *MMP20 (*matrixmetalloproteinaise20), K*LK4* (kall ikrein-4), and *FAM83H* genes.[1,2] The qualitative defect in the enamel is either hypoplastic, hypomineralized, or hypomature, depending on its clinical presentation and the stage of amelogenesis. Primary dentition, permanent dentition, or both can be affected by this disorder. It has a prevalence that varies from 1:700 to 1:14000 and possibly exhibits a genetic pattern of autosomal dominant, autosomal recessive, sex-linked, and sporadic inheritance.[3] AI is associated with many syndromes and serves as a good diagnostic feature in spotting such syndromes. One such syndrome is the enamel renal syndrome (ERS) – a condition characterized by the presence of AI along with nephrocalcinosis. It has a combination of dental features like AI, unerupted teeth, anterior open bite, pulpal calcifications, root and crown resorption, cementum deposition, truncated roots and taurodontism, and renal features like nephrocalcinosis, nephrolithiasis, and other functional abnormalities.[4,5] This condition is known by different names such as McGibbon syndrome, Lubinsky syndrome, Lubinsky-MacGibbon syndrome, AI and nephrocalcinosis, AI and gingival fibromatosis syndrome, or enamel renal gingival syndrome.[2,6] MacGibbon reported the first ERS in two brothers in 1972.[7] The cause for this syndrome was found to be a mutation in the *FAM20A* gene.[2] However, the actual prevalence of this syndrome is

still unknown, except for one study that emphasized its rarity.[2,8] The affected patients generally suffer from functional and esthetic problems, leading to an inferior quality of life. The severity of the associated renal disorders has been described as ranging from no complications to chronic renal failure.[9]

Case Presentation

An 8-year-old Bahraini male came to the Dental and Maxillofacial Center (Bahrain Defence Force Hospital) with his father complaining of teeth sensitivity on eating and drinking and was unhappy about the yellowish appearance of his teeth. The patient had a medical history of alpha thalassemia-2 trait, multiple chronic flank pain incidents, and recurrent urinary tract infection (UTI). Personal history revealed that he had two siblings, and his younger sister (5 years old) also had similar dental complaints. A written consent was taken from the patient's father for obtaining images and publication of the case report.

On examination, no abnormalities were found extra-orally. However, intra-oral examination revealed gross plaque accumulation, erythematous edematous gingiva with bleeding tendency on probing, generalized irregular morphology, and thin and rough enamel which was more yellowish than normal. Furthermore, the patient's dentition was at a mixed stage, indicating a delay in the eruption pattern along with the development of malocclusion due to the presence of an open bite anteriorly and posteriorly on the right side (Figures 1A and B).

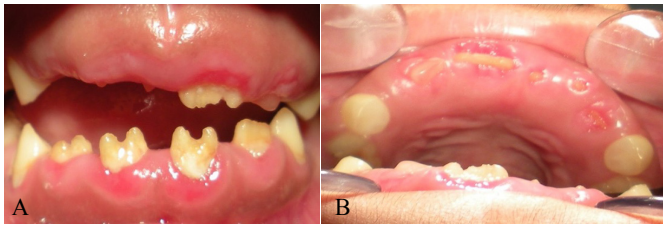

Figure 1: (A) Frontal view showing erythematous gingiva and irregularly shaped incisors.

(B) Maxillary occlusal view showing the delay in eruption of teeth and the fibrous gingiva.

Radiographic Assessment

A dental panoramic tomogram (DPT) (Figure 2) was obtained for the assessment and evaluation of teeth. It showed a clear absence of variable radiodensity between the enamel and dentin, a delayed pattern of tooth eruption, pulp stones (marked by arrows in the figure), and potential impaction of some teeth.

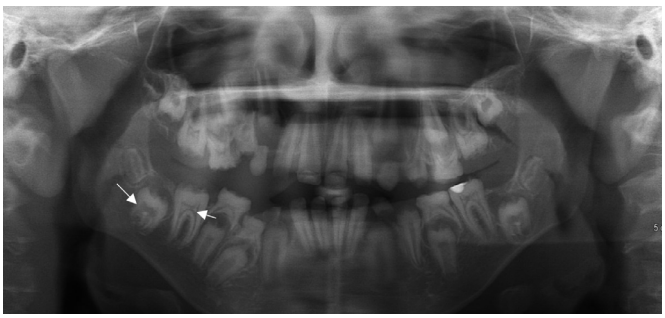

Figure 2: DPT showing a lack of contrast between enamel and dentin in all the teeth and intra-coronal pulp calcification (white arrows) in a few teeth.

Following DPT, the patient was referred to the medical block for further medical investigations. A renal ultrasound was obtained which showed moderate to marked dilatation of the pelvicalyceal system mounting to grade 3 hydronephrosis in the right kidney while the left kidney showed moderate dilatation of the pelvicalyceal system mounting to grade 2-3 hydronephrosis. Both showed increased echo pattern. Neither masses nor stones were detected.

Renal computed tomography (CT) (Figure 3 A and B) was then performed, and it showed bilateral moderate hydronephrosis with thinning out of the renal parenchyma, more pronounced in the right kidney. The left and right kidneys had a parenchymal thickness of about 11-13 mm and 7-11 mm, respectively. Multiple tiny renal microcalculi of 2-3 mm were seen bilaterally.

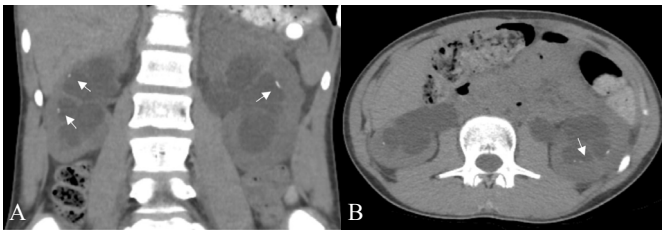

Figure 3: Renal CT - (A) Coronal view showing multiple microcalculi (white arrows) in both kidneys.

(B) Axial view showing kidneys with multiple microcalculi (white arrows).

The renal function test showed elevated creatinine 161 umol/L, uric acid 435 umol/L, urea 10.9 mmol/L, calcium 2.56 mmol/L, and inorganic

phosphate 1.56 mmol/L levels, indicating abnormal renal function.

A provisional diagnosis of ERS was made based on the clinical appearance and renal manifestations. Dentinogenesis imperfecta (DI), ectodermal dysplasia (ED), and non-syndromic amelogenesis imperfecta (NSAI) were all considered as differential diagnoses. DI was ruled out in this case as the teeth in DI have blue to brown discoloration and a "shell teeth"-like appearance radiographically.[10] ED was also excluded as it usually occurs in combination with one or more ectodermal structure defects such as partial or total alopecia, dystrophic or hypertrophic nails, etc., which was not found in our case.[11]

Full mouth rehabilitation was done with pre-formed metal crowns, considering the patient's chief complaint of poor esthetics and sensitivity. He was scheduled for regular follow-up visits at the center. He was also referred for further medical examination and management regarding his renal condition to avoid the risk of progressive renal failure.

Discussion

A general dental practitioner may come across many cases of AI; however, it is very difficult to ensure an accurate diagnosis due to its complex classification and the likely absence of relevant data from family members. The clinical features of AI vary depending on its type. In the hypomineralized type, the enamel is of normal thickness but not fully mineralized. Enamel hypomineralization can be seen due to the differences in translucency due to which the enamel might appear opaque, creamy white, or have yellow/brown discoloration. Opacities may be diffuse and/or demarcated. Defects can be localized or generalized. In the hypoplastic type, the thickness of the enamel is reduced, resulting in irregularly shaped teeth that may be pitted, thinner, or smaller in size. The hypoplastic type of AI is most commonly associated with this syndrome.[9]

A renal ultrasound is recommended in cases with such oral findings. Other relevant oral findings include localized aggressive periodontitis, gingival fibromatosis, and soft tissue calcifications, as described by Kantaputra et al,[6] who coined the term enamel-renal-gingival syndrome. The other orodental clinical features typical of ERS, as per De la Dure-Molla et al are listed in Table 1.[2]

Table 1: Orodental clinical features of enamel renal syndrome

Clinical and radiological features
Generalized thin, hypoplastic, or absent enamel
Primary and permanent teeth affected
Flat cusps on posterior teeth
Intrapulpal calcifications
Delayed tooth eruption, altered eruption pathway
Impacted posterior teeth with hyperplastic follicle (hamartoma-like)
Root dilaceration of impacted teeth
Gingival fibromatosis (of variable severity)
Gingival and dental follicle ectopic calcifications on biopsies
Semi-lunar shape of central incisor edge (additional feature)
Crown resorption of nonerupted teeth (additional feature)
Anterior open bite (additional feature)
Root hypercementosis and inter-radicular dentin dysplasia (additional feature)
Supernumerary teeth (additional feature)

In our case, nephrocalcinosis was diagnosed after the clinical and radiological diagnosis of AI. Nephrocalcinosis may persist and go undetected until the patient starts developing recurrent UTIs, pyelonephritis, or passes a stone. In such conditions, the ultrasound image of nephrocalcinosis is identical to other causes of medullary nephrocalcinosis. As a precautionary measure, the patient was referred to a nephrologist for ultrasound and CT of the kidneys and bladder, which revealed bilateral tiny renal microcalculi. Therefore, the present case was diagnosed with ERS, which is a rare autosomal disorder known to be associated with a mutation in the *FAM20A* gene. However, the mutation test was not done in the present case as the father was unwilling.

Based on the findings of CT and other diagnostic modalities, the patient was informed about the high risk of renal stone formation and was referred to a nephrologist for the necessary preventive treatment. Long-term follow-up is necessary for these patients to avoid serious renal dysfunctions.

An adequate understanding of the disease by the dental and medical teams is indispensable for making the correct diagnosis and ensuring appropriate treatment. Dentists are often the first to see such patients with oral problems as they manifest clearly. In this case, when it was observed that AI was present along with generalized fibrous gingiva that raised the doubt of some systemic involvement, the patient was further investigated with the help of a multidisciplinary team, predominantly the nephrologists. This led to the confirmed diagnosis of ERS.

Conclusion

It is very essential to consider a referral to a nephrologist for renal examination and excluding the presence of rare renal disorders like nephrocalcinosis. Obtaining a thorough medical history, performing relevant investigations, and proper referrals are imperative for the identification of such rare disorders. This can definitely augment patient care and widen the clinicians' insight when they come across any developmental abnormalities of the enamel and dentin.

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