CASE REPORT

Amelogenesis Imperfecta and Distal Renal Tubular Acidosis: A Case Report

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Abstract

Amelogenesis imperfecta (AI) is an inherited dental condition affecting enamel, which can result in significant tooth discoloration and enamel breakdown, requiring lifelong dental care. Distal renal tubular acidosis (dRTA) is a condition in which the kidneys are unable to acidify the urine to a pH < 5.5 in the presence of systemic metabolic acidosis. Management of AI and dRTA patients requires both medical and dental expertise to achieve long-term successful results. The aim of this paper is to present the dental management of a child with AI and dRTA.

Keywords: Amelogenesis imperfecta, Distal renal tubular acidosis.

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Introduction

AI represents a group of conditions, genomic in origin, which affect the structure and clinical appearance of enamel of all or nearly all the teeth, and that may be linked to morphologic or metabolic alterations in other parts of the body.[1] The reported prevalence of this condition varies from 1 in 14,000 to 1 in 4000, depending on the specific population studied.[2] AI is caused by mutations in genes that control amelogenesis such as amelogenin (AMELX), enamelin (ENAM), kallikrein4 (KLK4), enamelysis (MMP-20), and FAM83H and follows inheritance patterns of autosomal-dominant, autosomal recessive, or X-linked modes of transmission.[3]

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Classification

The most widely accepted classification of AI is based upon the predominant clinical and radiographic appearance of the enamel defect and mode of inheritance. The four main types are Hypoplastic, Hypomaturation, Hypocalcified, and Hypomaturation-Hypoplastic with taurodontism. Hypoplastic AI represents 60–73% of all cases. It refers to a deficiency in the quantity of enamel and presents with thin enamel which may be rough, smooth or pitted with normal radiodensity, lack of contact points, and anterior open bite. Hypomaturation AI represents 20–40% of cases where enamel is mottled brown or yellow with localized or diffuse opacities. Radio opacity of enamel is similar to that of dentin, and enamel tends to chip away from dentin. Hypocalcification AI represents 7% of cases. Radiographically enamel is less opaque than dentin and initially it is normal in thickness but due to its soft consistency it is easily removed soon after tooth eruption and the teeth may be light yellow to brown in color.[4]

Dental anomalies associated with AI include: quantitative and qualitative enamel deficiencies; congenitally missing teeth, pulpal calcifications, crown resorption, delayed tooth eruption, follicular cysts, hypercementosis, anterior open bite, and taurodontism.[5] It has been observed that patients with AI have significantly higher levels of social avoidance and distress than did patients without the condition. Diagnostic challenges, aesthetics, dental sensitivity, and loss of vertical dimension of occlusion due to rapid tooth degradation are all common clinical issues seen in AI patients.[6]

Many factors influence the treatment plan for these patients, including the patient's age, socioeconomic status, the kind and severity of the condition, and the intraoral situation at the time the treatment plan is formed.[7]

Renal tubular acidosis (RTA) arises from the kidney's inability to excrete enough acid or retain enough bicarbonate (HCO_3-), resulting in impaired urinary acidification. The most common type of primary RTA in childhood is distal RTA (dRTA), characterized by impaired urinary acidification or H+ ion secretion leading to hyperchloremic acidosis with persistent alkaline urine and very low urinary citrate levels. It can be hereditary (primary) or acquired (secondary) and often presents with signs and symptoms related to severe hypokalemia, including proximal muscle weakness, polydipsia, and polyuria. dRTA has a small daily loss of HCO_3- which is treated with supplemental sodium or potassium bicarbonate.[8]

A hypothesis that may link AI with RTA is that many of the dental proteins that were thought to be tissue-specific may also be expressed in non dental tissues such as renal tissues. Distal RTA can have a wide variety of systemic associations and the association with AI highlights the need for a thorough renal evaluation in children with enamel disorders.

Early diagnosis of dRTA is critical so as to maximize growth with treatment.

Although AI has been rarely reported in association with metabolic disorders such as hypocalciuria, Bartter-like syndrome, and dTRA, very few cases with dRTA have been identified till date.[9]

This case report presents the dental management of a child patient with AI and dRTA, which rarely presents in conjunction.

Case Description

A 11-year old girl reported to the Department of Pedodontics and Preventive Dentistry at Army College of Dental Sciences with the complaint of pain and intraoral swelling in left lower back tooth region, generalized sensitivity, and discolored teeth. A detailed medical and dental history was obtained followed by intraoral and extraoral examination.

The medical history revealed that the patient is suffering from dRTA which was detected at 2 years of age when she presented with weakness, lethargy and inward bowing of knees and is currently on oral supplements (tablet Sodamint 325 mg, Rantac 150 mg).

Intraoral examination revealed that the patient's oral hygiene was unsatisfactory and she exhibited Chronic marginal gingivitis. It was observed that enamel thickness of all teeth was reduced with dentin exposure (Fig. 1). The patient also 54 55 64 84. Deep dental caries extending into the pulp in relation to 74 75 (Fig. 2) and loss of enamel in all teeth excluding 1st permanent molars. The patient was in mixed dentition stage (Fig. 3). There was no history of consanguineous marriage or presence of similar abnormalities in the family, including grandparents. Attrition of the enamel was evident on her primary teeth resulting in a reduction in the occlusal vertical dimension.[2]

The clinical and radiographic features and the family history were consistent with a diagnosis of type IV AI (hypoplastic-hypomaturation with taurodontism).

The treatment objectives were to provide emergency care followed by preventive and restorative therapy. The treatment aimed at pain relief, elimination of tooth sensitivity, improvement of esthetics, prevention of further loss of tooth structure, modification of the child's attitude, and behavior toward dental treatment and improving her periodontal health.

The parents were informed of the diagnosis and the treatment modalities available. The treatment alternatives, expected clinical longevity of the restorations and the length of the treatment period were also explained in detail to the child and her parents. After thorough consideration of the treatment options, the following treatment plan was devised-

- Oral prophylaxis, oral hygiene instructions, and topical fluoride application
- Endodontic therapy in relation to 74 75
- Restoration with Glass Ionomer Cement in relation to 54 55 64 84 (GC Gold Label Fuji IX)
- Direct composite Resin Restorations (3M ESPE Filtek Z250 Universal Restorative) on permanent teeth including maxillary and mandibular incisors and first permanent molars (Fig. 4)
- Restoration of primary molars with stainless steel crowns (3M ESPE) (Fig. 5).

Follow-up

The patient was recalled at 3 months interval.

Discussion

AI is a condition that comprises genetically and clinically diverse groups of hereditary disorders that primarily affect the quantity, structure, and composition of enamel. AI has many forms and affects the well-being of children at a very early stage of development. It is a rare disorder, and general dentists, hygienists, and dental assistants do not often meet patients with AI.[10]

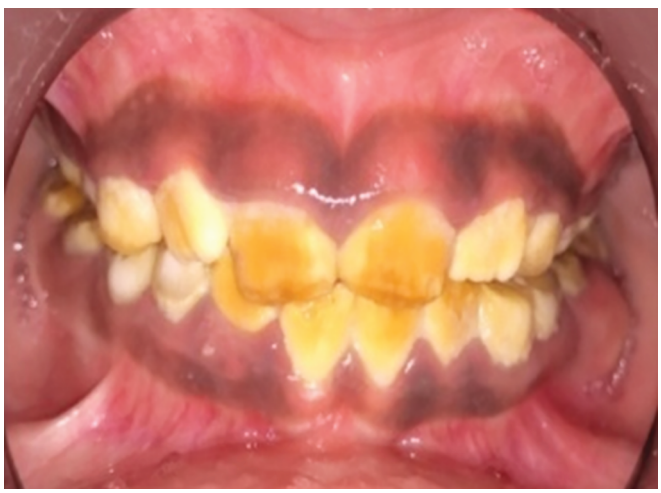

Fig. 1: Pretreatment intraoral view

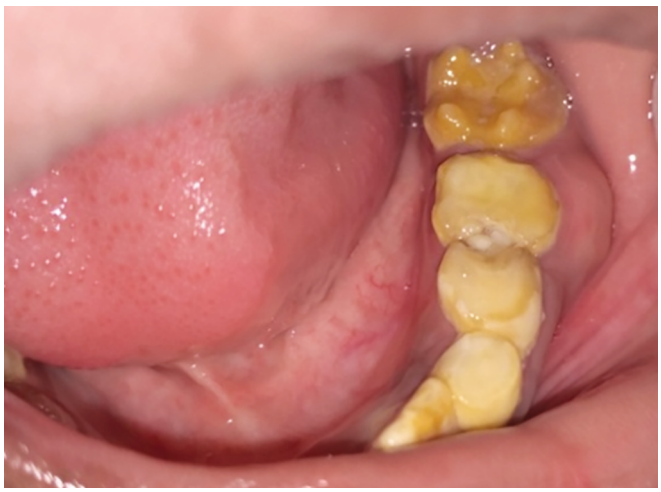

Fig. 2: Pretreatment view showing abscess WRT 75

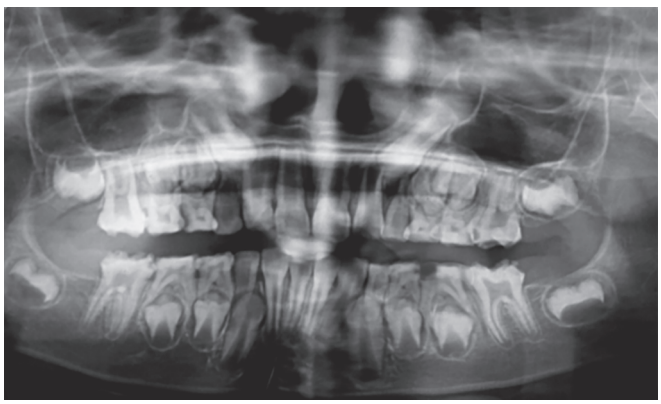

Fig. 3: Pretreatment orthopantomogram

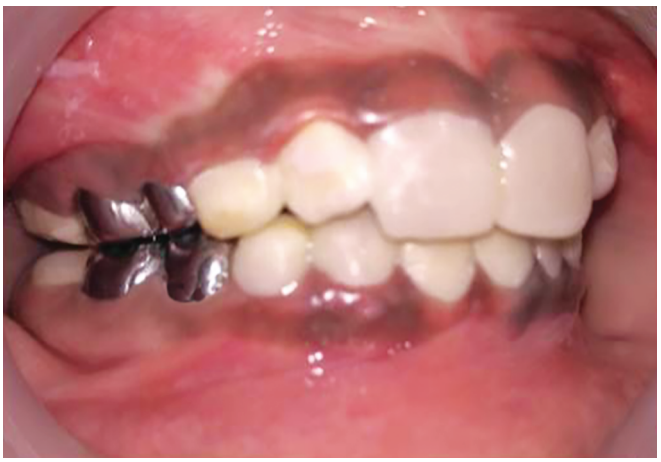

Fig. 4: Post-treatment intraoral view showing full mouth rehabilitation

Clinically, the intricacy of the etiological causes, the range of clinical presentations, and the paucity of evidence-based therapeutic guidelines make the treatment of AI difficult.[2]

The abnormalities related to AI affect the children and their parents in an extensive manner. Thus, early and radical interventions must be done so as to reduce the effects such as tooth sensitivity and improve normal chewing ability, and enhance the esthetic appearance of teeth to reduce negative social consequences.[4]

Primary distal type 1 RTA is the most common type of RTA found in children nowadays. In children, dRTA most often presents as a hereditary condition with hyperchloremic metabolic acidosis, hypokalemia, nephrocalcinosis/nephrolithiasis, polyuria, rickets, and growth retardation. Thus, early diagnosis of dRTA is important.[9]

There are several challenges to overcome while managing a pediatric patient with AI. The child's age, dental anxiety, peer pressure, and parental expectations are just a few problems which can make the dental treatment difficult. A multidisciplinary team involving general dental practitioners, pediatric dentists, orthodontists, restorative colleagues, therapists, and hygienists can be helpful in treating this group of patients.[11]

To enhance oral health in children with AI, early diagnosis and timely preventive care are critical. Often, pediatric dentists are the first to see children with AI and the therapy must involve an overall comprehensive plan that will includes a rough draft of future treatment needs. Complete oral rehabilitation of patients with AI is critical not only for aesthetic and functional reasons, but also for its psychological implications.[2]

Restorative treatment for AI for primary and permanent teeth consist of crowns and composite veneers. If there is a need for pulp therapy, it must be completed before the crown is placed. SSCs are the ideal material for posterior teeth, especially primary posterior teeth and zirconia, and porcelain are used for crowns on permanent anterior teeth. The final phase of the treatment should fulfil emotional and aesthetic demands so that the young patient can gain social self-confidence.[4]

Conclusion

AI is a condition which affects children from an early stage of their lives. If diagnosed and treated early, it can significantly improve quality of life for the patient. As the present case reports, a patient with AI is often not just affected by this inherited disorder affecting oral health but may also suffer from various other systemic disorders such as dRTA thus suggesting a link between the two disorders. Therefore, it is exceedingly important to perform a complete renal evaluation in patients with AI. Further research is however required to fully understand the various defects which are found in the different types of AI for the development of a standardized multidisciplinary approach.

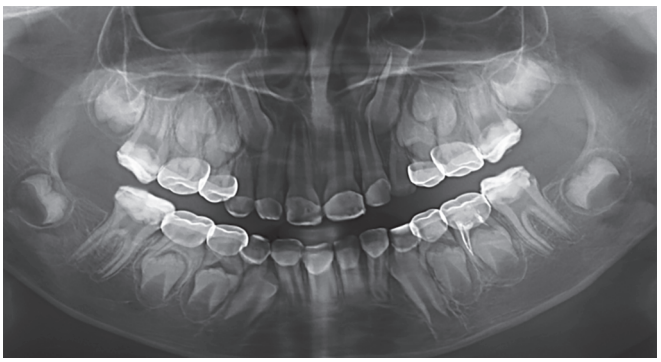

Fig. 5: Post-treatment orthopantomogram

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