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Amelogenesis Imperfecta and Gingival Hyperplasia in a 21-Year-Old Female Patient: A Case Report

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KEYWORDS

Abstract:
Enamel Renal Syndrome (ERS), is a rare genetic condition that is characterized by hypoplastic amelogenesis imperfecta, failed tooth eruption, intra-pulpal calcifications, gingival enlargement, and in some patients, nephrocalcinosis. Patients with ERS are likely to seek dental care first due to the retention of primary teeth and failure of permanent tooth eruption. Herein we report a case exhibiting the orodental hallmarks (amelogenesis imperfecta and gingival hyperplasia) of ERS without the renal symptoms, in a young female patient.

Introduction

Amelogenesis imperfecta (AI) is a group of hereditary disorders that affect the development of tooth enamel. Gingival hyperplasia, on the other hand, is a medical condition characterized by an overgrowth of gum tissue. Although these conditions typically occur independently, some documented cases show that they can appear together, suggesting the possibility of a syndrome.(Farias et al., 2021) (de la Dure-Molla et al., 2014a)One such syndrome is Enamel Renal Syndrome (ERS). It is a rare autosomal recessive genetic condition that is characterized by hypoplastic AI, failed tooth eruption, intra-pulpal calcifications, gingival enlargement, and in some patients, nephrocalcinosis. Patients with ERS are likely to seek dental care first due to the retention of primary teeth and failure of permanent tooth eruption. This syndrome exhibits FAM20A (FAMily with sequence similarity 20A) gene mutation (a Golgi-associated secretory pathway pseudokinase). (Wang et al., 2014) (de la Dure-Molla et al., 2014b) (Hassib et al., 2020)

ERS is characterized by nephrocalcinosis, nephrolithiasis, gingival fibromatosis and hypoplastic amelogenesis imperfecta Laboratory alterations concerning renal affection may not always be present. The renal phenotype, typically silent during childhood, is characterized by reduced calcium, phosphate, and citrate excretion with subsequent nephrocalcinosis.(Sharma et al., 2022) Another syndrome with a FAM20A mutation is the amelogenesis imperfecta gingival fibromatosis syndrome (AIGFS), which also exhibits AI and gingival enlargement.(Roomaney et al., 2023) (Nasrallah & Berro, 2024)

According to a study by de la Dure-Molla et al. in 2014, patients with ERS exhibit a unique orodental phenotype, including generalized hypoplastic AI in both primary and permanent teeth, pulp stones, delayed tooth eruption, hyperplastic dental follicles, and varying degrees of gingival hyperplasia with calcified nodules. (de la Dure-Molla et al., 2014a)

This case report details the presentation, diagnosis, and management of a patient with both amelogenesis imperfecta and gingival hyperplasia. The study was conducted after obtaining an informed consent from the patient and was approved by the ethics committee of the Faculty of Oral and Dental Medicine (FODM), Future University in Egypt (FUE).

Case Presentation

A 21-year-old female patient presented to the outpatient clinics of the Dental Hospital, FUE, with concerns about the appearance of her teeth and gums. The patient reported having discolored and easily chipped teeth since childhood. Additionally, she expressed difficulty maintaining oral hygiene due to excessive gum tissue covering her teeth. The patient gave a negative family history of a similar condition. A complete blood picture (CBC) was requested from the patient which came back normal.

Extraoral examination revealed no significant abnormalities. Intraoral examination revealed generalized enamel defects with discoloration ranging

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from yellow to brown of the erupted teeth, attrition, and semi-lunar notching of the incisal edges of her incisors. Generalized gingival enlargement, partially obscuring teeth without any signs of inflammation or bleeding was also noted. (figures 1)

Panoramic radiographs revealed normal alveolar bone development multiple unerupted teeth with prominent follicles, and pulp calcifications. (figures 2)

The decayed, non-restorable teeth were extracted. Gingivectomy followed by osteotomy (crown lengthening) was performed (figure 3). The excised tissue was sent for histopathological assessment which documented the presence of hypertrophic fibrous tissue with an overlying hyperplastic epithelium. The postoperative course was uneventful.

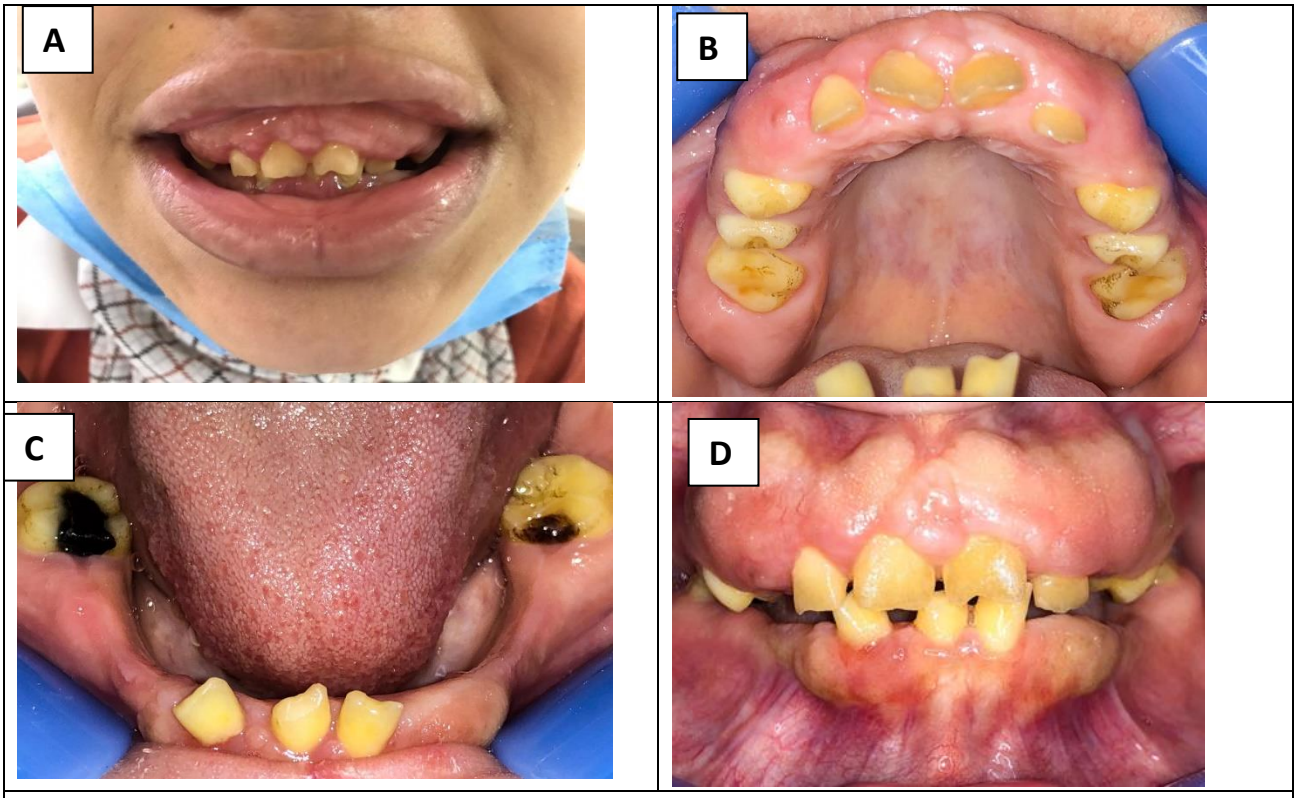

Fig. 1: A,B,C and D, clinical presentation of the case showing enamel hypoplasia and gingival enlargement.

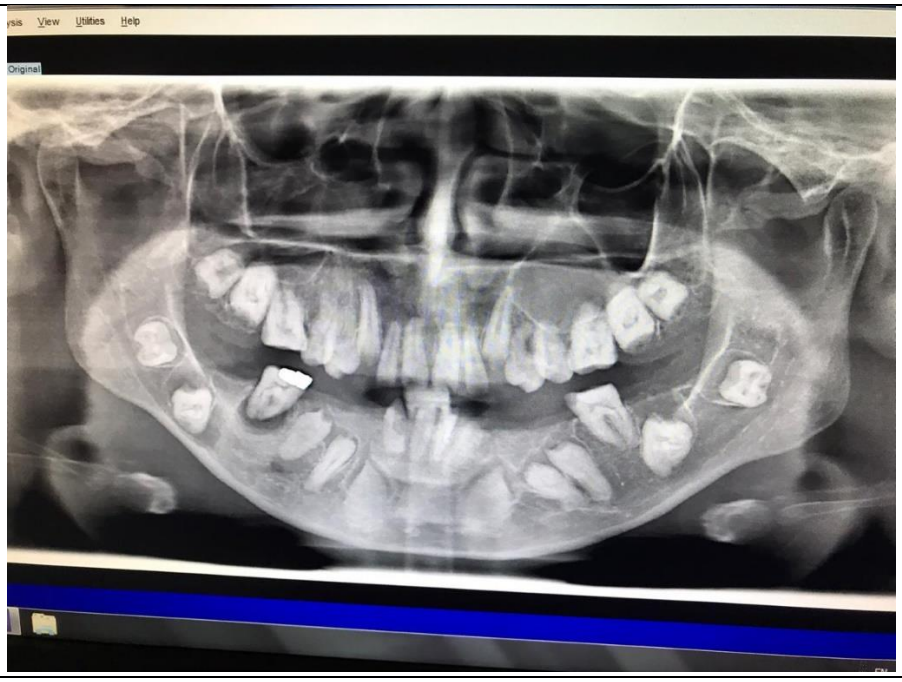

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Fig. 2: Panoramic radiograph showing multiple unerupted teeth some of which exhibit a prominent follicle. Note: pulp obliteration in some teeth.

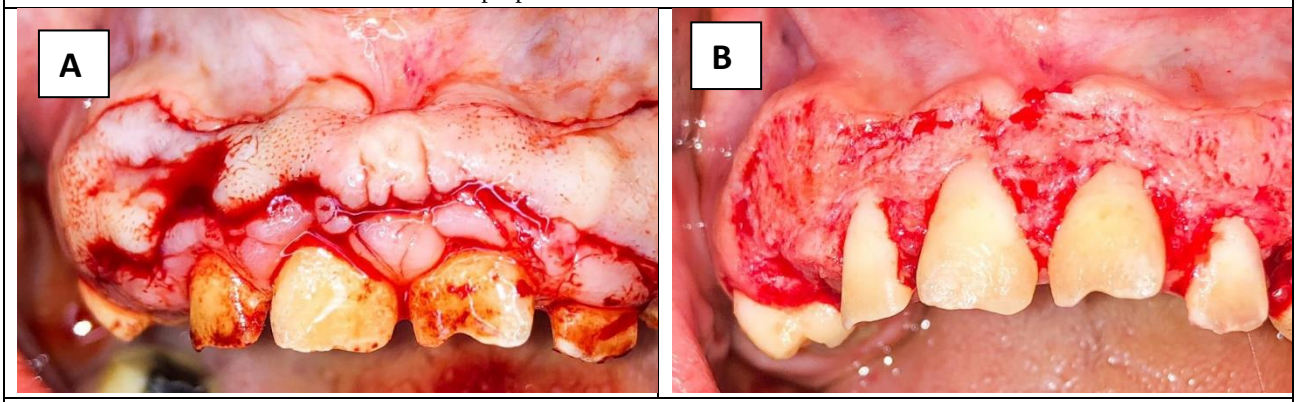

Fig. 3: Surgical procedure gingivectomy (A) and osteotomy (B)

Discussion

ERS is a rare genetic condition that often results in a unique oral phenotype and can sometimes have severe systemic effects. When dentists identify such cases, they refer them to a nephrologist for further evaluation and testing. This highlights the importance of recognizing the oral profile, as the dentist is often the first healthcare professional to come into contact with the patient. (Nasrallah & Berro, 2024) (Roomaney et al., 2023) (Sharma et al., 2022)

Although the pathognomonic oral profile developed by de la Dure Molla et al.,2014, helps identify those potentially having ERS, it is important to note that not all features are necessary for diagnosis.(de la Dure-Molla et al., 2014b) In this case report, the patient presented with AI and gingival hyperplasia, hallmarks of ERS. Diagnosing ERS can be challenging because the onset and severity of symptoms can vary significantly. While AI manifests early in childhood, nephrocalcinosis might not present till later on in life. (Farias et al., 2021) (Nasrallah & Berro, 2024)

This case report describes a 21-year-old woman presenting with a combination of discolored, chipped teeth, excessive gum tissue, and unerupted teeth. The clinical and radiographic findings suggest a diagnosis of ERS. According to Farias MLM et al, 2021, the diagnosis of ERS is based on orodental abnormalities and kidney function impairment. However, since renal changes occur late, the presence of the characteristic oral phenotype is enough to diagnose this syndrome, even in the absence of other symptoms. (Farias et al., 2021)

Conclusion:

This case underscores the importance of considering rare syndromes like ERS when encountering patients with AI. Early diagnosis and a collaborative approach involving dentists and nephrologists can ensure comprehensive care and potentially prevent future complications.

Conflict of interest

None to declare

Ethics approval

The patient signed an informed consent and the study was approved by the ethics committee at the FODM, FUE

Acknowledgement

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