

Amelogenesis imperfecta with multiple impacted teeth and altered eruption pathways

- A radiographic case report

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ABSTRACT

Amelogenesis imperfecta (AI) comprises a vast group of developmental alterations in the quantity and/or quality of enamel, unaffected by systemic disorders.

In this case report we describe the typical clinical and radiographic features of AI with multiple impacted teeth and altered eruption pathways. Patients with AI benefit from early diagnosis and treatment to prevent tooth loss and loss of vertical dimension.

Optimal management of these patients requires a multi-disciplinary approach and regular dental recall appointments. Therefore it is the oral health clinician's responsibility for the timeous identification of this condition.

Keywords

Amelogenesis imperfecta, impactions, altered eruption pathways, gubernacular canals.

INTRODUCTION

Amelogenesis imperfecta (AI) comprises a vast group of developmental alterations in the quantity and/or quality of enamel, unaffected by systemic disorders. To date over 15 genetic mutations have been identified in cases of non-syndromic AI.¹⁻⁶ A recent publication hypothesised that gene mutations in *ENAM* and *AMELX* leads to endoplasmic reticulum stress in ameloblasts which in turn activates an unfolded protein response as a pathogenic mechanism for AI.⁷

Additionally, for reasons not yet fully understood, defects in acid phosphatase and *RELT*, have been implicated in AI.⁸⁻¹¹ Several other mutations have been identified in syndromic cases with AI, including *DLX3* (trichodonto-osseous syndrome) and *FAM20A* (enamel-renal syndrome, amelogenesis imperfecta-gingival fibromatosis syndrome).¹²⁻¹⁴

Four phenotypic subtypes of AI are recognised; hypoplastic, hypocalcified, hypomatured and mixed hypomatured/hypoplastic in combination with taurodontism.¹⁵⁻¹⁷ In the hypoplastic subtype, the disruption occurs in the deposition of the enamel resulting in a thin band of fully mineralised enamel or pits and grooves dispersed in normal enamel thickness.

The hypocalcified subtype encompasses presentations where the full thickness of enamel is deposited but no mineralisation occurs. Only partial mineralisation occurs in the hypomatured subtype.^{17,18} This classification is however limited, as both qualitative and quantitative enamel defects may co-present within the same patient.^{18,19}

The diagnosis of AI is largely based on characteristic clinical and radiographic findings.^{17,20} In normal circumstances enamel has a higher radiographic density when compared to dentine due to the higher mineral content.²¹ In the hypoplastic subtype, a thin band of radiopaque enamel can be distinguished from the less radiopaque dentine. With hypomatured AI the radiographic density of enamel and dentine is alike, whereas in the hypo-

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3. Tsholofelo Kungoane: Pathological diagnosis and tooth ground section - 10%
4. Christy L Davidson: Conception and design of the manuscript. Drafted and revised the work critically for important intellectual content - 30%

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calcified subtype the enamel will have a lower density than that of the dentine and may not be evident on a radiograph.¹⁷

The primary clinical concerns of AI include increased dentinal sensitivity, altered aesthetics, loss of vertical dimension, malocclusion and increased caries susceptibility.^{17,22} Other dental features that have been reported in association with AI include pulpal calcifications, root malformations (including taurodontism) and impactions of permanent teeth.^{20,23,24} In this case report we describe a patient that presented with all the typical radiographic features of AI, and additional interesting findings.

CASE PRESENTATION

A 26-year-old male patient presented with the main complaint of small and sensitive teeth. His dental history included the recent construction a maxillary partial denture for improved functionality and composite veneers on the maxillary anterior teeth for aesthetics.

No familial history of similar conditions were reported. Extraoral examination revealed recurrent Herpes labialis on the right bottom lip. Intraorally, generalised tooth wear and numerous retained primary teeth were present.

The maxillary central incisors showed veneer preparations while both maxillary lateral incisors had composite veneers. The permanent first molars displayed a rough and yellow appearance with no visible enamel (Figure 1). The mandibular right second incisor had a distal uncomplicated coronal fracture. The patient's oral hygiene was satisfactory with non-inflammatory gingival enlargement. The patient was referred for radiographic examination.

The panoramic radiograph (Figure 2) revealed a full complement of teeth however only the incisors and first permanent molars were erupted. All the remaining teeth were impacted and no supernumerary teeth were detected. Pulpal calcifications could be seen in some of the erupted and a few of the unerupted teeth. The mandibular right first molar and left second deciduous molar were carious.

The carious mandibular right deciduous molars were associated with periapical radiolucencies. A residual root was visible in the maxillary left first premolar area. Radiographically, there was no distinction between the enamel and dentine in the erupted and unerupted teeth. The impacted teeth demonstrated corticated radiolucent tubular or canal-like structures connecting the dental follicle to the alveolar ridge which resembled eruption pathways.



Figure 1. Intraoral photograph showing a lack of enamel and a rough and yellow appearance of the maxillary first molars (black arrows) and multiple missing teeth.

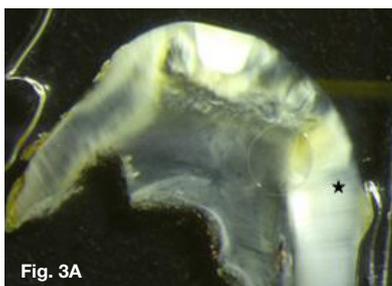


Fig. 3A

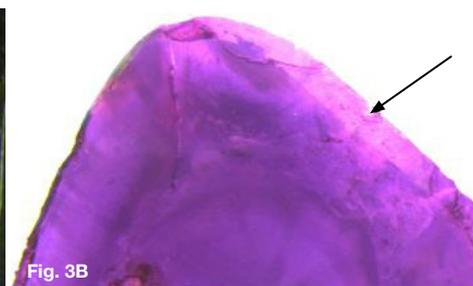


Fig. 3B

Figure 3. A: Ground section of the mandibular right second deciduous molar showing dentine with dentinal tubules, pulp with pulpal chamber and minimal representation of enamel. B: Haematoxylin and Eosin section of the crown showing the enamel (arrow head).

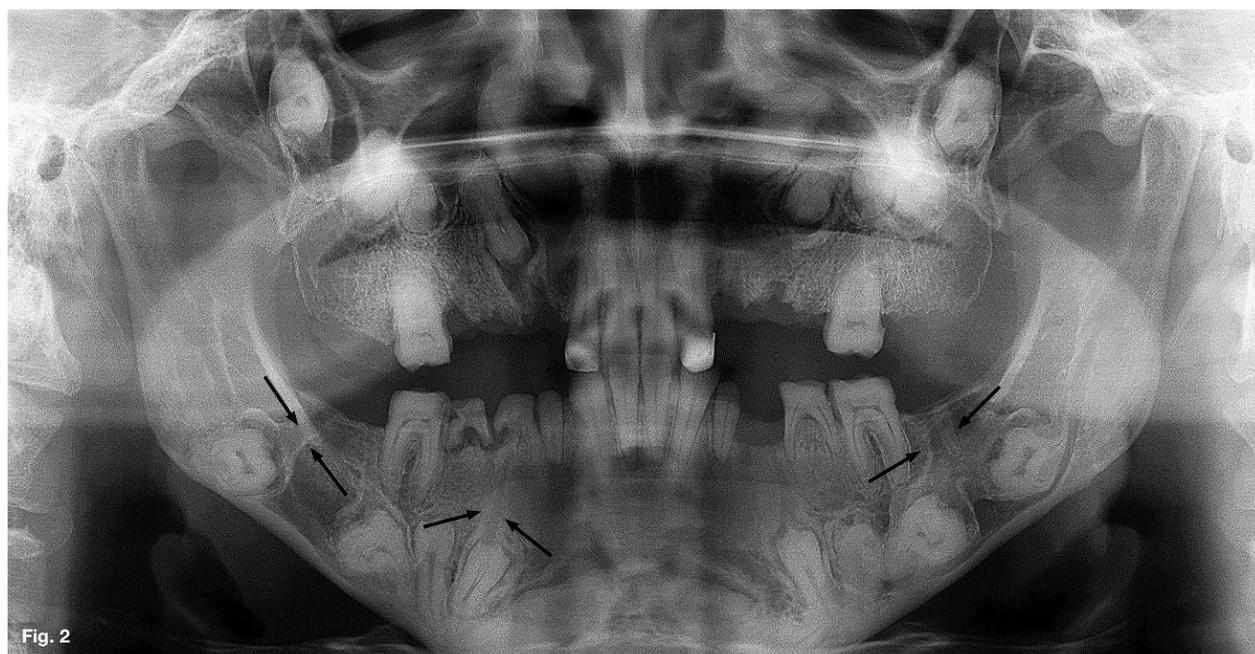


Fig. 2

Figure 2. Panoramic radiograph. Presence of numerous impacted teeth showing no visible enamel. Corticated canal-like structures connecting the dental follicle to the alveolar ridge (black arrows).

The mandibular right deciduous molars were extracted and submitted for histological examination. The ground sections (Figure 3) showed molar teeth with minimal representation of enamel. The dentine was of normal architecture with a straight dentine-enamel junction. Both teeth showed focal external root resorption.

These features were in keeping with AI. The patient was referred to the post-graduate prosthodontics department for further management and dental rehabilitation. Genetic testing was declined by the patient. The patient was also referred to a physician for renal analysis to rule out enamel-renal syndrome.

DISCUSSION

The most apparent features observed on the panoramic radiograph were the numerous impacted teeth and the presence of canal-like eruption pathways connecting the dental follicle to the alveolar ridge. Patients with AI are six times more likely to have impactions of permanent teeth and associated anomalies such as dentigerous cysts.^{19,23}

Tooth eruption is a complex developmental process influenced by genetic regulation and growth of the jaws.²⁵ The dental follicle orchestrates tooth eruption through the regulation of specific genes regulating bone resorption coronally and deposition apically.^{26,27} Disturbances in this process can be attributed to follicular enlargements or disruptions in the molecular signals of the eruption process itself.^{19,23}

Gubernacular canals are bony canals thought to occur as a result of the gubernacular cord. This cord is a fibrous strand remnant of the dental lamina that connects the reduced enamel epithelium surrounding the developing tooth, with the overlying oral epithelium. It secretes epidermal growth factor mediator that promotes osteoclastogenesis, thereby directing tooth eruption through the eruptive pathway.²⁸⁻³¹

The gubernacular cord was initially only described in association with permanent teeth that have a primary predecessor but later found to also be associated with permanent molars that do not have primary predecessors.^{28,32,33} However, the existence of the gubernacular cord and canal remains controversial. Nishida et al. reported the gubernacular canal as "corticated tracts of low density, contiguous with the dental follicle of an unerupted tooth".³⁴

A recent study on cone beam computed tomography investigated the presence and morphology of these canals in relation to eruption disturbances and pathologic conditions associated with impacted and unerupted permanent teeth. They found that a disturbed eruption pattern could occur with either the absence or an alteration in the shape of the gubernacular canal.²⁹

These eruption pathways have also been observed in cases of primary failure of eruption disorder where a fault in the eruptive mechanism, with no mechanical obstruction, results in impaction.^{27,35} The panoramic radiograph of the presented case shows evidence of

altered eruption pathways, suggesting a fault in the eruptive process.

In this case report the patient had presented with non-inflammatory gingival enlargement. This finding has previously been reported in the literature as an association with AI.^{36,37} From 1990 to 2014, there have been 8 AI case reports (5 of which originated from South Africa) with dental follicular-like hamartomas and generalised gingival enlargement.^{24,38-44} These reported cases demonstrate a form of AI together with enlarged dental follicles associated with numerous impacted teeth.^{24,36,38-44} Some of these cases with enlarged follicles were diagnosed as dentigerous cysts³⁶ or odontogenic fibromas.³⁸ However, one should consider that the latest WHO classification states that pericoronal radiolucencies with central odontogenic fibroma features associated with impacted teeth, should be considered hyperplastic follicles rather than neoplastic.⁴⁵

In this case report, the patient presented with AI, gingival hyperplasia, multiple impactions, hyperplastic follicles and diffuse intrapulpal calcifications. These features have previously been reported in association with *FAM20A* mutation.^{13,46} However to our knowledge this is the first reported case showing altered eruption pathways of impacted teeth in AI.

Patients with AI benefit from early diagnosis and treatment to prevent consequences of tooth loss and loss of vertical dimension. The increased dentinal sensitivity is caused by the exposed dentinal tubules and can be managed with fluoride application or coverage depending on the extent of enamel disruption. The patients also present with increased caries susceptibility that require preventative dental procedures and regular follow up appointments. The altered aesthetics can be managed with crowns or veneer preparations. Bond strength to the altered enamel should be evaluated on a case to case basis. Direct resin restorations have shown success in the improvement of aesthetics but long term follow-up is needed to validate the longevity of the treatment success.⁴⁷ A recent review found that indirect restorative techniques have a better prognosis in the case of AI as compared to direct restorative techniques.⁴⁸

The optimal management of these patients requires a multidisciplinary approach and regular dental recall appointments.⁴⁹ Therefore, it is the oral health clinician's responsibility for the timeous identification of this condition. A limitation of this study is that the true nature of the dental follicle and associated canals can only be speculated, as there was no indication for surgical removal of the impacted teeth. These lesions will be monitored going forward.

Declaration

Authors Nel, Yakoob, Kungoane and Davidson declare no conflict of interest.

Ethical considerations

All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1975, as revised in 2008 (5).

Informed consent was obtained from all patients for being included in the study.

This article does not contain any studies with human or animal subjects performed by the any of the authors.

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