

Atypical Case of Amelogenesis Imperfecta and Generalized Impacted Dentitions: Dental Management and Follow Ups

Original
Article

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ABSTRACT

Background: Odontogenesis is a complicated process that ends with normal dentition formation followed by ordinary eruption time. Many genes regulate tooth development; however, any alteration in the odontogenic mechanism can lead to malformed teeth formation or inappropriate eruption behavior.

Aim: We present a 17-year-old patient who complained of unerupted teeth with amelogenesis imperfecta (AI).

Methods: The patient was subjected to full medical and dental histories taking and a comprehensive dental management with long-term follow-ups were performed. A blood sample was taken and whole exome sequencing (WES) was performed for patient.

Results: WES data excluded variants in any of the known causative genes of AI or delayed eruption. This may point to either the presence of a deep intronic variant which requires the use of a higher technology like whole genome sequencing or a new unraveled gene for AI and delayed eruption. Although the unknown genetic etiology of the condition.

Conclusion: The provided dental procedure was satisfactory to the patient. The long-term follow-ups helped in better case evaluation. Inherited disorders remain always challenging in diagnostic and management aspects.

Key Words: Amelogenesis imperfecta, Delayed eruption, Impacted dentition, Orthotraction.

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INTRODUCTION

Teeth eruption is a normal physiological process occurs spontaneously at suitable chronological age. The eruption process is influenced by the maturation development of jaw bones and teeth. The theory of dental eruption is based on the interrelation between adaptive powers of periodontal ligament with active motion of dental follicles to resorb the deciduous overlying root or bone tissues (Kjaer, 2014). Multiple genes are involved in the traditional mechanism of dentition eruption and affect phenotypic presentation of teeth in the oral cavity (Fatemifar *et al.*, 2013).

Pathological eruption behavior has been described being delayed or premature. The pattern of eruption itself could be localized or generalized according to local, systemic or genetic factors (Kjaer, 2014). Delayed eruption is inherited with many syndromes like mainly Cleidocranial Dysplasia (OMIM# 119600, 216330), osteoglophonic dysplasia syndrome (OMIM# 166250) and GAPO syndrome (OMIM# 230740) (Hanisch *et al.*, 2018).

Delayed eruption was also reported in association with hereditary structure teeth defect as amelogenesis imperfecta and dentinogenesis imperfecta cases (Hassib *et al.*, 2020).

Inherited amelogenesis imperfecta (AI) is a rare inherited disorder occurs during amelogenesis which may affect both primary and permanent dentitions. AI could be exhibited as an isolated trait or associated with syndromes. AI is manifested in various forms according to its phenotypic presentation. A new classification included the type of AI, mode of inheritance and molecular data was proposed by (Bloch-Zupan *et al.*, 2023). So far, more than 70 genes have been reported in patients with the inherited AI (Bloch-Zupan *et al.*, 2023).

The incorporation of surgery with orthodontic management is of great interest to solve the problematic impacted teeth which affects esthetic and function (Maceno *et al.*, 2018).

This study presents a 17 years old male with generalized amelogenesis imperfecta and unerupted dentitions. An ortho-traction to expose impacted teeth with full mouth rehabilitation was performed to the affected patient. In addition, a 6-years follow up was assessed.

CASE PRESENTATION:

This study was approved by the Reviewer Scientific Board of the Oro-dental Genetics Department, National Research Centre (NRC), Cairo, Egypt and the Medical Research Ethics Committee of NRC (Approval: 19268). The patient's mother signed a written informed consent to accept the dissemination of the data. Patient was subjected to meticulous clinical and dental examinations and photo and pedigrees were taken as well.

A 17-year-old male patient complaining of discolored teeth of the lower jaw and completely edentulous upper jaw was examined at the outpatient clinic of Oro-dental Genetic Department, Centre of Excellence, NRC. The parents of the patient were first cousin (Figure 1A). The clinical examination revealed bilateral clinodactyly of the 5th finger, on examining joint clinically; there was no hyperextensibility. Clinical skin examination revealed no skin laxity. Blood laboratory analyses for calcium, phosphorus and alkaline phosphatase were within the normal range. Abdominal ultrasonography revealed normal sonography. Extra-oral examination demonstrated thick lips, short philtrum, broad chin, bilateral well developed nasolabial fold (Figure 1B) and fused ear lobules to neck skin (Figure 1C). The dental examination exhibited many unerupted teeth and the present dentition had amelogenesis imperfecta. The upper jaw was completely edentulous and some of the lower anterior teeth were not present as well (Figure 1D). Panoramic radiograph showed multiple bony unerupted teeth, as well as loss of enamel layer (Figure 1E). Cone beam CT displayed the level of soft tissue and hard tissue impacted dentitions (Figure 1F). The preliminary diagnosis went to an amelogenesis imperfecta disorders.

Whole Exome Sequencing (WES)

Genomic DNA was extracted from peripheral blood samples of the patient using Qiagen Blood DNA Kit (Qiagen, Hilden, Germany). WES was then performed using SureSelect Human All Exome 50 Mb Kit (Agilent, Santa Clara, CA, USA) and analyzed on Illumina NovaSeq 6000 (Illumina, San Diego, CA, USA).

RESULTS

WES did not reveal pathogenic, likely pathogenic or variants of uncertain significance in any of the known genes of AI or delayed eruption. Therefore, we suspect either the presence of an overcome deep intronic variant in such genes or may be the patient's phenotype is caused by a variant in a new undiscovered gene yet for AI or delayed eruption.

Dental management of the patient:

The predesigned treatment plan was fit to solve the main chief complaint of the patient which was an impacted discolored dentition. Based on the panoramic view and the cone beam CT (CBCT) which revealed many soft tissue impactions of certain upper teeth, surgical orthodontic traction was the optimum suggested dental management. Surgical windows were done to expose soft tissue impacted teeth (Figure 2A, B). Slight locked bone was removed around the offending tooth if needed. Mini titanium screws were placed bilaterally in the lower canine areas in the opposing arch for intermaxillary anchorage (Figure 2C). The bonding of bands and brackets were challenging due to enamel layer loss. Certain modifications were added to the proposed treatment plan to achieve better management as follow; Tooth #21 received an endodontic treatment, then; a hole was performed in the crown to place a ligature wire for pulling the tooth out to overcome bracket adhesion failure. Routine banding, orthodontic brackets and elastics were used to exert the applied force for teeth motion (Figure 2D). The patient was scheduled every three weeks to change elastics and assess the case. After six months, the teeth were in the oral cavity (Figure 2E). Fixed porcelain fused to metal bridge was fabricated to compensate esthetic and function (Figure 2F). Yearly follow up was scheduled to assess the work. After six years follow up, there was no complaint from the patient, and he was completely satisfied by the outcome (Figure 2A-F).

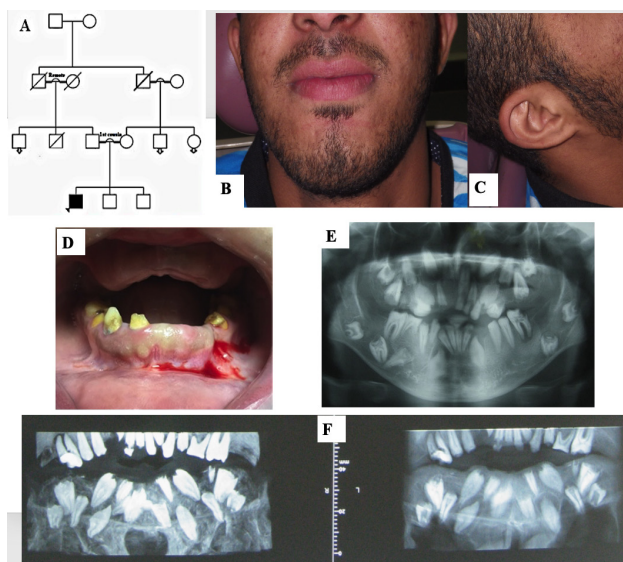


Figure 1: A) Pedigree of the affected patient, B) Profile photo shows short philtrum, thick lips, broad chin and bilateral nasolabial folds, C) Photo shows fusion of ear lobule to the neck, D) Intraoral photo shows completely edentulous upper jaw and teeth structure defects with clinically missing dentition in the lower jaw, E) Panorama showing impacted permanent teeth, delayed eruption, wide pulp chamber and loss of enamel layer, F) Cone beam CT views show the impacted dentitions in soft and hard tissues.

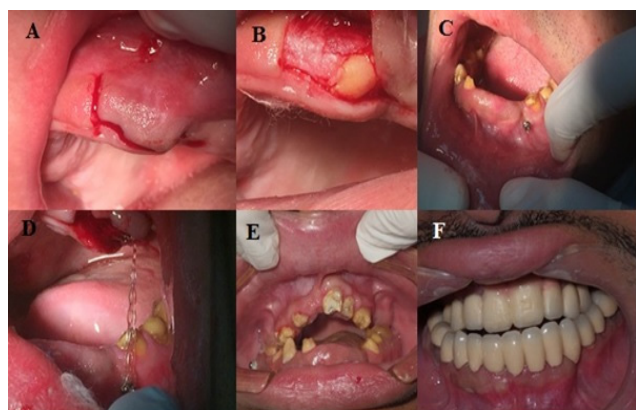


Figure 2: A) surgical gingival incision, B) Tooth exposure on the ridge, C) Screw placement onto buccal bone (blue arrow), D) Anchorage by elastic placement to move tooth, E) Teeth appears in the jaws after orthodontic traction, F) Fixed prosthodontics restoration for restoring function and esthetic.

DISCUSSION

The prevalence of localized impacted teeth worldwide ranges from 5.6 to 18.8% (Patili and Maheshwari, 2014). The generalized non erupted dentition was presented mainly in GAPO and osteoglophonic dysplasia syndromes (Wilson and Jain, 2020).

The universal incidence of amelogenesis imperfecta is 1:700 to 1:14,000. AI has been present with various forms according to the amelogenesis stage defect. However, the differentiation being the dental anomaly is isolated or part of a syndrome needs a meticulous diagnosis besides sophisticated laboratory investigations to reach the accurate diagnosis (Crawford et al., 2007).

The combined AI with delayed eruption may be reported in enamel renal syndrome (OMIM#204690) and dental anomalies with short stature disorder (OMIM#601216) (Hassib et al., 2020; Nawaz et al., 2023). Conversely, the case herein remains unique as the generalized retarded dentition of the upper arch with the global AI was not previously reported according to our knowledge.

The use of exome sequencing (ES) has become the gold standard technique to identify pathogenic variants and reach a definitive diagnosis for cases with atypical phenotypes or new syndromes (Marwaha et al., 2022). Nevertheless, ES data of our patient revealed negative result which denoted that this case might be a new syndrome with unknown unreported gene or a deep intronic variant not detected by the ES.

Surgical intervention accompanied with orthodontic traction is widely used to exposed impacted teeth. The active traction force applied to the offending dentition helps in tooth eruption and alignment in the dental arch.

It is considered very successful method for multiple retarded eruptions (Einy et al., 2022). Moreover, the use of titanium screws for intermaxillary anchorage to induce teeth eruption facilitates and accelerates the process (Kuroda et al., 2007). Herein, we could not be able to apply orthotraction to all the impacted upper dentitions except for the teeth just beneath the gingiva. Therefore, we succeeded to expose many teeth.

The clue of success in surgical active traction is having free dentition without ankylosis. The knowledge of the causative factors of impaction is ultimately important for designing the treatment plan and achieving the speculated outcome. CBCT was performed to confirm that the unerupted teeth were not ankylosed.

In genetic aspect, active orthodontic traction with screws procedure was favorable with cleidocranial dysostosis cases. This technique was effective to expose the teeth in the oral cavity, as well as, align the dentitions (Kuroda et al., 2007). At the contrary, GAPO and osteoglophonic dysplasia with ankylosed and inverted teeth or thickening of alveolar ridge avoid the impacted teeth to be managed (Silveira et al., 2006; Kuthiroyly et al., 2017).

The resort to use the surgical orthodontic traction with titanium screw was the suggested dental management for this case. Despite the unknown background of this new disorder, a trial was firstly done and ended by a worthy dental manipulation and good patient contentment.

CONCLUSION

The genetic disorders remain always challenging onto diagnostic and management aspects. The good understanding of the different treatment availabilities, the treatment plan strategies, as well as the ideal dental manipulation remain the main objectives to achieve a successful outcome. It is important in unusual cases to extend the follow-up periods for better assessment and validation.

CONFLICT OF INTEREST

There are no conflicts of interest.

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