Research Highlights

When it rains it pours: one patient, three dental anomalies

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Abstract

Dentinogenesis imperfecta (DI) may present with a broad spectrum of dental and systemic manifestations and can be related to several anomalies. This case report describes a patient with the uncommon triad of DI Type 2 (bulbous crowns with pulp chambers obliteration), impacted upper first permanent molars (FPM) and severe hypodontia. This paper outlines the aetiology of the described dental anomalies and the available treatment options. It underlines the importance of a holistic approach, highlighting the clinical and psychological complexities in treating such patients, and demonstrates the need to seek specialist opinion and input from the interdisciplinary team. It also discusses how early intervention can resolve the functional and aesthetic problems caused by dentinal defects and thereby improve patients' quality of life.

Abbreviations

DEJ - Dentine-enamel junction
DGP - Dentin glycoprotein
DI - Dentinogenesis imperfecta
DPP - Dentin phosphoprotein
DSP - Dentin sialoprotein
DSPP - Dentin sialophosphoprotein gene
FPM - First permanent molars
IHS - Inhalation sedation
LL - Lower left
LR - Lower right
MDT - Multidisciplinary team
OVD - Occlusal vertical dimension
PEB - Post-eruptive breakdown
SSC - Stainless steel crown
UL - Upper left
UR - Upper right

Introduction

Dentinogenesis imperfecta (DI) is a rare genetic disorder characterised by abnormal dentine structure and curtailed mineralisation.[1] It can present in both the primary and permanent dentitions with a plethora of unique clinical and radiographic features. Approximately 1 in 45,000 of the general population are affected by DI, and it is slightly higher in the Black African population of South Africa.[2,3]

This autosomal dominant condition is linked to mutations in collagen type I matrix genes COL1A1 and/or COL1A2, usually classified as Type 1 DI and associated with the condition called osteogenesis imperfecta (prevalence 1 in 20,000). This disorder is also linked to the mutation of dentin sialophosphoprotein (DSPP) gene, which is responsible for expressing dentin sialoprotein (DSP), dentin glycoprotein (DGP) and dentin phosphoprotein (DPP) (non-collagenous proteins of dentine organic matrix which regulate mineralisation and hydroxyapatite formation).[3] A DSPP defect and subsequent deficient dentine

mineralisation is inherited in an autosomal dominant manner, has prevalence of 1 in 6000 to 1 in 8000 and is classified as Type 2 isolated DI.[3] Both Types 1 and 2 have very similar clinical and radiographic presentations.

Clinical presentation and associated anomalies

The affected dentition has amber/brown or grey/blue opalescent appearance (**Figure 1**) and is highly susceptible to enamel chipping due to the defective dentine-enamel junction (DEJ) bonds and rapid dentine attrition. Post-eruptive breakdown (PEB) and tooth-wear can further lead to pulpal exposures and the development of periapical pathologies, such as apical abscesses. This is most noticeable in primary teeth due to the large pulps. As a result, tooth surface loss is rapid, leading to reduction in occlusal vertical dimension.[2,4]

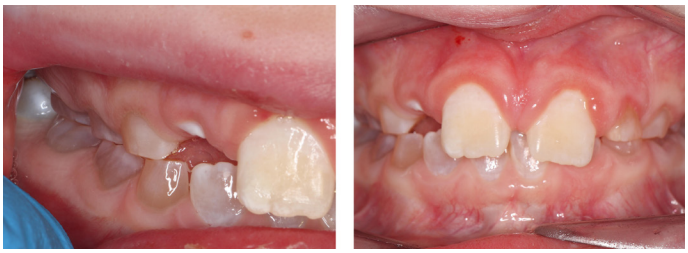

Figure 1. Tooth appearance in DI.

Depending on the classification and the severity of the condition, patients with DI can have a variety of radiographic manifestations. Crowns of the affected teeth can appear dysmorphologically bulbous with short and thin roots. In turn, the abnormal crown morphology of the second primary molar can lead to ectopic eruption and impaction of first permanent molars (FPM).[3,5,6]

Another significant characteristic of DI is tertiary dentine deposition and pulp chamber obliteration (**Figure 2**). According to the literature, this is associated with an increased attrition-related permeability of the enamel and dentine, which can then initiate pulp necrosis and occlusion of the chamber.[7,8]

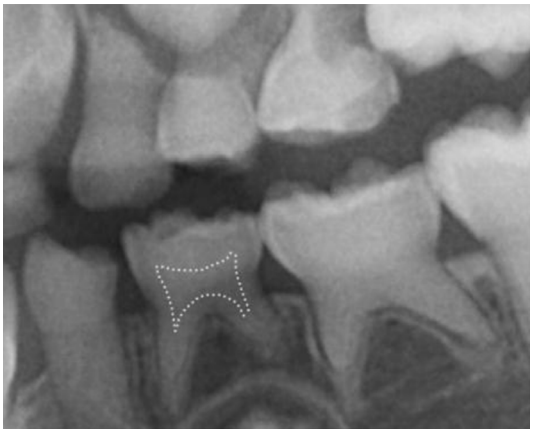

Figure 2. Section of a panoramic radiograph that shows a complete pulp obliteration in primary dentition.

Unlike amelogenesis imperfecta (a genetic disorder which presents with abnormal formation of the enamel), hypodontia (one to six teeth congenitally missing) is rarely associated with DI. Instead, there are several external influences that can damage the early dental tissues, the dental lamina, which are associated with missing teeth: infection, medications, chemotherapy, radiation therapy, dental trauma, endocrine abnormalities and intrauterine disturbances.[1,4]

This case report highlights the importance of a holistic approach to treating patients with hereditary enamel and dentinal defects, as the variety of clinical and systemic presentations and a substantial psychological factor present a myriad of implications and challenges for both the patient and the clinician.

Case

Written informed consent was obtained from the patient and the patient's parents. A 7-year-old boy presented to the University of Manchester Dental Hospital Department of Child Dental Health with his mother.

> *He was concerned about the aesthetics of his upper anterior teeth, in particular the translucent, yellow/brown appearance of the primary dentition and receding gingiva in the upper anterior region.*

Maternal history was significant for a diagnosis, as mother claimed that her teeth had a similar appearance and she has been diagnosed with DI Type 2. The patient's medical history included well-controlled asthma, mild allergic rhinitis and eczema. The mother denied the child having any previous sclera discolourations, bony fractures, bone pain or joint problems, which are all features of osteogenesis imperfecta, which is commonly linked to DI Type 1. The behaviour assessment showed a severe dental anxiety/phobia.

Extra-orally, the child had a normal stature and the sclera of his eyes were white. An intra-oral examination revealed an early mixed dentition which was clinically and radiographically caries-free. Oral hygiene was satisfactory. Bilateral partially erupted and ectopic upper first permanent molars were noted, with associated severe resorptive cavities distal to the upper right and left second primary molar teeth (URE and ULE). Primary teeth showed classic amber discolouration with signs of attrition (**Figure 3**). He also had a class 2 division 1 incisor relationship with an increased overbite.

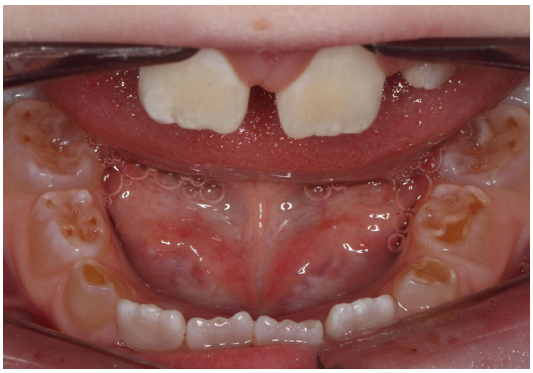

Figure 3. Primary dentition with signs of attrition and PEB.

A DPT radiograph revealed that 6 permanent teeth (UR4, UR5, UL4, UL5, LL5 and LR5) were developmentally absent. The primary molars were small and bulbous with pulp canal obliteration and canal thinning. Roots were short and blunted. UR6/UL6 were ectopic with resorption of the distal aspects of URE/ULE (**Figure 4**).

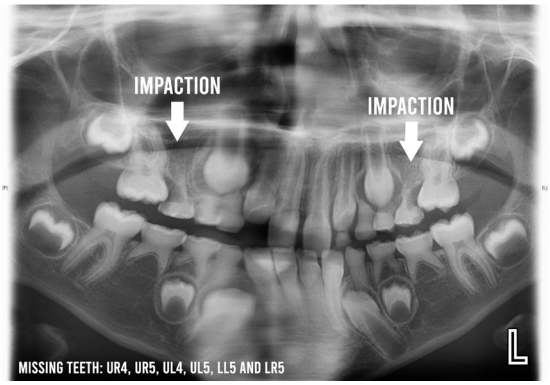

Figure 4. Panoramic radiograph of the case patient with DI.

As the patient had a normal stature, white sclera and no history of any joint issues or bony fractures, DI Type 1 related to osteogenesis imperfecta was disregarded.

Based on the patient's history, clinical and radiographic examination, the following diagnoses were made:
1. DI Type 2
2. Impacted upper first permanent molar teeth.
3. Hypodontia: developmentally absent UR5, UR4, UL4, UL5, LL5, LR5

All diagnoses were discussed with the patient and his mother. It was also explained that a further assessment with a multidisciplinary team (MDT) was necessary due to the complexities and rarity of these three anomalies being present simultaneously.

Due to the severe impaction of both upper FPMs, the patient underwent interceptive extractions of the URE and ULE to facilitate the eruption and mesial drifting of FPMs (**Figure 5**).[10,11] This treatment pathway was chosen as the second primary molars had a poor prognosis with severe unrestorable resorption. The procedure was carried out under inhalation sedation (IHS) due to high levels of dental anxiety. Six months post-extraction, the clinician will review the patient to assess the eruption of current permanent dentition and to consider restorative options for the missing teeth.

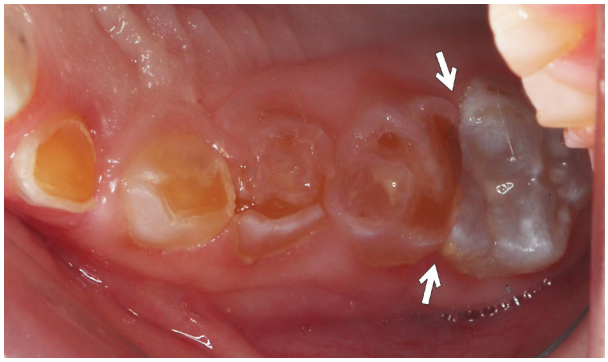

Figure 5. Impacted upper first permanent molars.

Discussion

Management of patients with DI presents multiple challenges: both clinical aspects of care and the patient's psychological wellbeing need to be taken into account. Poor dental aesthetics and increased dentine sensitivity, as well as hypodontia and ectopic eruption, bring a plethora of challenges to both the patient and the practitioner.
Patients with similarly complex conditions are very likely to be affected by dental anxiety/phobia[1,12] as they require more frequent dental and hospital visits. Such situations call for the application of different behaviour management techniques: psychological (e.g. behaviour shaping) and pharmacological (e.g. IHS, intravenous sedation, general anaesthetic). The aim here is to successfully complete the treatment whilst avoiding a negative experience and fostering a positive relationship with the dental team.

> *The clinical concern when treating DI is to prevent tooth wear and dentine sensitivity by protecting the enamel through early placement of fissure sealant, stainless steel crowns (SSCs) or cuspal coverage restorations on both primary and permanent molars.*

In the described case the placement of SSCs on lower primary molars was avoided, as this would have likely hastened their exfoliation[13] and the aim was to maintain primary teeth for as long as possible due to lack of successors.

Affected anterior teeth can be restored functionally and aesthetically with direct and indirect composite/porcelain restorations. Further oral hygiene support is crucial in preventing pulpal bacterial ingress, pulpal necrosis and periapical abscesses.[1,4]

Impacted FPMs occur as the result of abnormal teeth eruption.[14,15] If impaction is left untreated, FPMs can erupt with malocclusion, such as mesial tipping and rotation.[1] Early treatment is imperative to correcting the path of eruption and preventing resorption of adjacent primary molars. Treatment options for impacted teeth include monitoring for spontaneous correction, placement of separator or active appliance and interceptive extraction of the second primary molar.[9-11,12-15] Early loss of URE and ULE was unfortunate due to the missing successor teeth; however, due to the extent of the resorption, both teeth had a hopeless prognosis and extraction was deemed the most appropriate option.

This case highlights the complexity of simultaneously managing three dental anomalies and emphasises the importance of seeking specialist advice from an interdisciplinary team in similarly intricate cases. In this particular case, the team would include consultants from Paediatric Dentistry, Restorative Dentistry and Oral Surgery. Restorative specialists could consider retaining and restoring primary molar teeth going into adulthood, in order to maintain the vertical dimension of affected dentition, as well as considering prosthetic replacement of the developmentally absent or prematurely lost teeth. Orthodontic and oral surgery specialists may consider more advanced restorative options for managing the spaces caused by hypodontia, for example, additional orthodontic extractions or crowns, bridges and dental implants once growth has completed.[4] As patients with DI are more likely to have molar-cross-bite and Class 3 malocclusions an orthodontic treatment should be well-planned due to the higher risks of root resorption.[16]

Conclusion

The presented case report describes a holistic and systematic approach to treating a unique patient with three dental anomalies: DI Type 2, impacted FPMs and hypodontia. Early clinical identification, close monitoring and long-term comprehensive MDT-integrated management of each anomaly is crucial for prevention of potential complications. Furthermore, this approach to treatment can improve the compromised aesthetics and function, thereby helping the overall mental and physical wellbeing of the patient, as the treatment plan is likely to be carried out throughout their entire life.

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Contribution statement All authors contributed equally to the work. All authors made substantial contributions to the conception or design of the work, or the acquisition, analysis or interpretation of data for the work. All authors drafted the work or revised it critically for important intellectual content and gave final approval of the version to be included in Inspire

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