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CASE REPORT

Comprehensive Dental Rehabilitation of an Osteogenesis Imperfecta Child with Dentinogenesis Imperfecta Type 1

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ABSTRAK

Dentinogenesis imperfecta-Type I (DGI-Type I) ialah anomali pembentukan gigi yang diperhatikan dalam sesetengah pesakit osteogenesis imperfecta (OI). Gigi-gigi dalam anomali ini sering mengalami perubahan warna, dan enamelnya mudah pecah disebabkan oleh pembentukan struktur dentin yang tidak sempurna. Gigi-gigi yang terjejas biasanya haus dengan cepat jika keadaan ini tidak dikenal pasti lebih awal. Diagnosis awal membolehkan pelaksanaan strategi preventif dan rehabilitasi yang efektif berdasarkan umur pesakit. Perlindungan gigi-gigi yang terjejas membolehkan pelbagai fungsi pergigian, seperti mastikasi dan estetik gigi boleh dipelihara dengan baik sejajar dengan tumbesaran kanak-kanak OI. Selain itu, ia juga dapat memastikan kualiti hidup berkaitan kesihatan mulut pesakit OI juga adalah setara dengan kanak-kanak sihat.

Kata kunci: *Anomali gigi; genetik; kecacatan pembentukan gigi*

ABSTRACT

Dentinogenesis imperfecta-Type I (DGI-Type I) is a dental developmental anomaly observed in some osteogenesis imperfecta (OI) patients. Teeth in this dental anomaly often appear discoloured, and their enamel breaks easily due to the defective underlying dentine. The affected teeth are abraded rapidly if the condition is not identified early. Early diagnosis allows the proper execution of age-based preventive and rehabilitation strategies. Protection of the affected teeth enables various functions of the dentition, such as mastication and dental aesthetics, which could be maintained effectively in growing OI children. Thus ensuring the oral health-related quality of life of the patients with OI is at par with that of their healthy counterparts.

Keywords: Dental anomaly; dental developmental defects; genetics

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INTRODUCTION

Genetics plays an essential role in tooth development. Failure of proper gene expression related to tooth development may lead to anomalies ranging from tooth formation to structural defects (Suryadeva & Begum 2015). Dentinogenesis Imperfecta (DGI) is one of the inherited tooth developmental anomalies that exhibit debilitating consequences of varying degrees to the dentition. Teeth with DGI are weaker than usual, discoloured and prone to fracture (MedlinePlus 2017).

Three types of DGI are documented based on Sheild's 1973 classification, namely DGI-type I, a recognised syndromic entity related to osteogenesis imperfecta (OI); DGI-type II, similar clinical, radiographic and histological presentations as DGI-type I, but without OI; and DGI-type III, a rare but severe form of DGI without OI characterised by teeth with thin dentin layer (shell) surrounding a large pulp chamber (Leung et al. 2009). However, based on the genetic aetiology classification, DGI-type II and DGI-type III are grouped under dentin sialophosphoprotein (DSPP) gene expression-related diseases, whilst DGI-type I is recognised as a separate entity unrelated to defective DSPP gene expression (de La Dure-Molla et al. 2015).

Besides the discolouration of teeth ranging from blue-grey or yellow-brown to translucent hue, patients with DGI usually experience early post-eruptive enamel loss due to dentine exposure. The exposed dentine is easily abraded away up to the gingival margin. Thus, dental aesthetics concerns, loss of vertical facial height, ineffective masticatory function, dental caries and early tooth loss are a few of the adverse consequences of DGI (MedlinePlus 2017; Tsoukala et al. 2022). OI patients with DGI often exhibit poorer oral health-related quality of life compared to healthy children (Cachia Mintoff et al. 2022). This case report emphasised the need for the early detection of DGI in a young child with OI and the prevention of adverse consequences through a comprehensive dental care plan.

CASE REPORT

A 3-year-old boy diagnosed with OI Type III was referred to the paediatric dental clinic for dental assessment. The boy's parents were concerned about their child's worn and discoloured teeth. Lately, the boy has complained of sensitive teeth, especially when eating cold food. The boy appears potentially cooperative for dental assessment with a Frankl scale score of 2, and he has no prior exposure to dentistry.

The boy's bone deformities and growth retardation were detected intra-uterine through an antenatal ultrasound. The OI diagnosis was confirmed at birth, and the boy had the first fracture of his left femur at the age of 7 months. After that, the boy had a further eight recurrent fractures to the left femur (Figure 1). All the fractures were treated conservatively with hip spica cast applications. Currently, he is on 3-monthly cyclic therapy of intravenous 5 mg pamidronate and a daily oral dosage of vitamin D3 (200 IU) and calcium (100 IU). Besides OI, the boy also has bronchial asthma, which is treated with budesonide and salbutamol inhalers as and when required.

The boy has blue sclerae and is of short stature with spinal scoliosis and mild bowing of legs. Antero-posterior left femur radiograph showed a proximal diaphysis fracture with an exuberant callus and multiple transverse lines noted at both proximal and distal metaphyses secondary to pamidronate therapy (Figure 2). The boy's lateral skull radiograph also showed wormian skull bones.

Extraorally, the boy has bitemporal bossing with a convex facial profile and competent

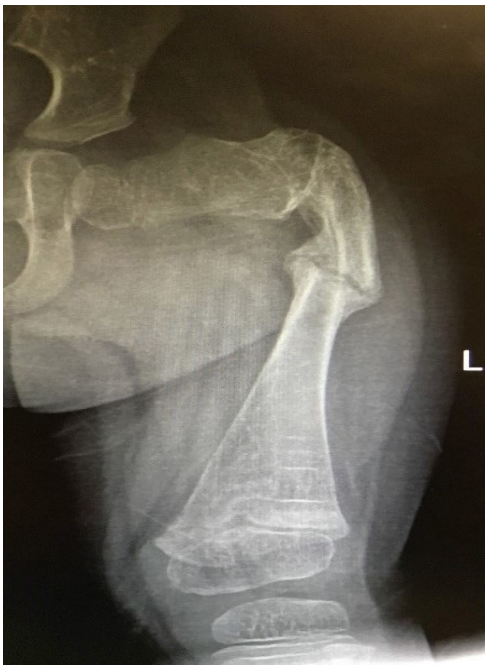

FIGURE 1: Anterior-posterior view of the left femur showing fracture at the proximal

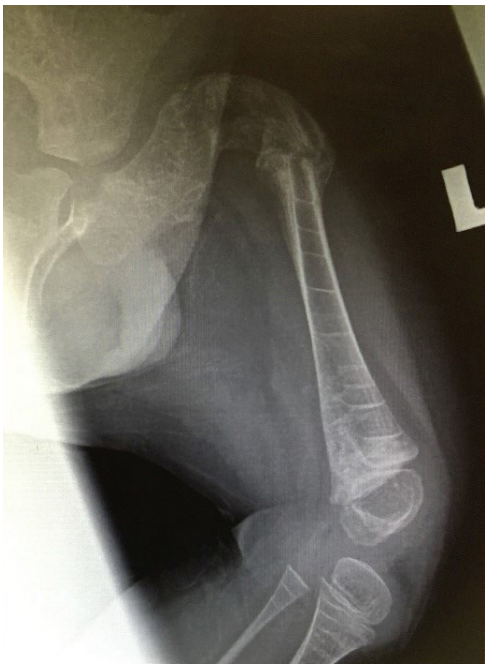

FIGURE 2: Anterior-posterior view of the left femur showing multiple transverse lines

lips. Intraorally, all his primary teeth were present and showed signs of tooth wear. Mild generalised gingivitis was noted with healthy oral mucosa. The oral hygiene was fair, with a plaque score of 30%. Plaque deposition was predominant near the cervical margins of the teeth. As the child was experiencing tooth sensitivity, he began to avoid cold foods and drinks. However, he has no issue with toothbrushing, and his mother supervises his brushing. He uses children's fluoridated toothpaste with 550 parts per million (ppm) fluoride. Attempts to obtain a dental panoramic tomograph or periapical radiograph failed because of the boy's small stature and intolerance to radiograph film placement inside his mouth.

All the teeth showed brownish-grey discolouration with moderate to mild attrition on the upper anterior and posterior molar teeth, respectively. The lower anterior teeth showed severe attrition. However, many teeth had enamel breakdowns, showing the dentine without pulp exposure (Figures 3-5). No evidence of active caries noted on the affected teeth. Some of the teeth have white speckled discolouration appearance resembling enamel hypomaturation.

In the current case, progressive deterioration of the dentition is expected to occur over time. Therefore, the immediate phase of treatment had been executed with proper preventive strategies, including the restoration of affected teeth with stainless steel crowns and strip crowns (Figures 6-8), as well as monitoring of the future developing dentition. As the patient was only potentially cooperative and required multiple dental treatments, all the restorative treatment was performed under general anaesthesia.

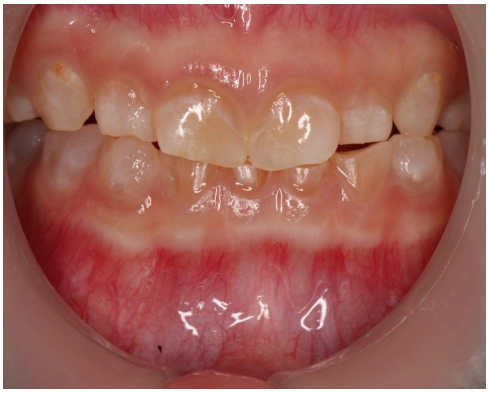

FIGURE 3: Anterior view of the dentition before treatment

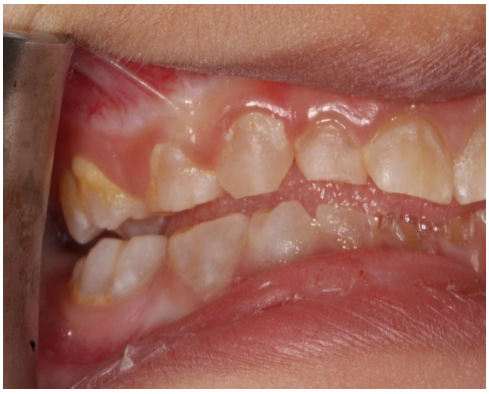

FIGURE 4: Right lateral view of the dentition before treatment

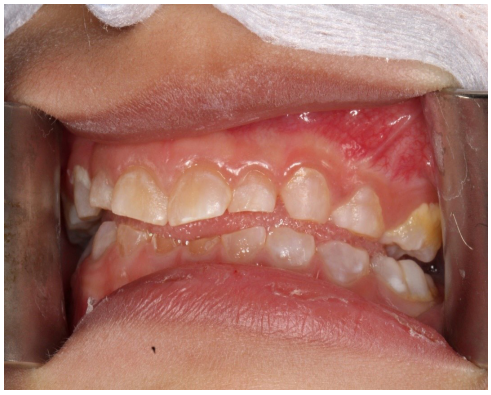

FIGURE 5: Left lateral view of the dentition before treatment

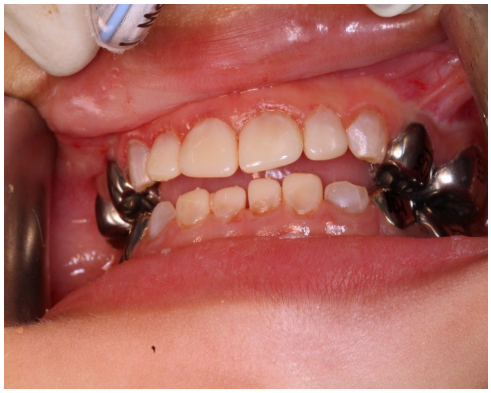

FIGURE 6: Anterior view of the dentition after treatment

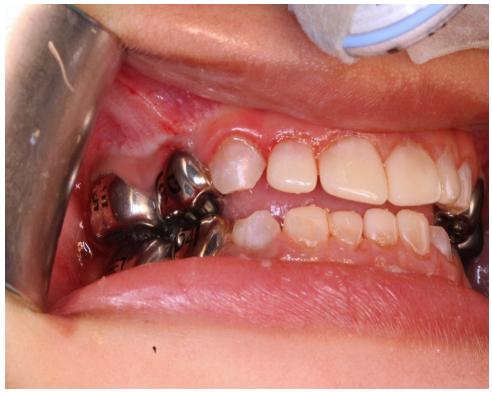

FIGURE 7: Right lateral view of the dentition after treatment

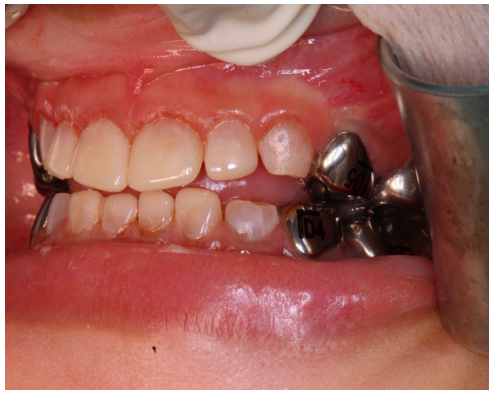

FIGURE 8: Left lateral view of the dentition after treatment

DISCUSSION

OI, known as "brittle bone disease," is a rare genetic disorder commonly related to mutations of COL1A1 and COL1A2 genes that encode Type I collagen in most cases (Subramaniam et al. 2023). However, other diverse gene mutations related to OI have also been identified (Valadares et al. 2014). Because of the impaired collagen synthesis, collagen-rich structures, such as ligaments, sclera, lungs, heart, skin, joints, bones and teeth, are affected at varying severities (NIAMS 2022; Subramaniam et al. 2023). The classical classification of OI with four types (Type I-IV) based on the clinical Sillence classification has evolved because of newer discoveries of genes and different inheritance patterns. Currently, as many as 22 types of OI have been identified (Panzaru et al. 2023). Key clinical features of OI include multiple bone fractures, blue sclera and varying degrees of bone deformities. OI-type I is often the mildest form. OI-type II and III are the severe forms, where newborns with OI-type II usually die a few weeks after birth. Newborns with OI-type III usually survive with multiple recurrent bone fractures and severe skeletal deformities. OI-type IV often displays mild to moderate symptoms (Rare Diseases 2021). The boy in the current case has OI-type III with a history of recurrent bone fractures, bone deformities, short stature, spinal scoliosis, blue sclerae and DGI.

DGI is not clinically present in all OI patients, with reported DGI prevalence ranging from 19% to 62.5% based on the respective studies (Nguyen et al. 2021). Regarding different types of OI, DGI has been reported in the range of 8 to 40% in OI-type I, 43% to 82% in OI-type III, and 37% to 100% in OI-type IV (Leung et al. 2009). The severity of dentine involvement depends on the quantity and quality of the Type I collagen fibrils formed. In DGI related to OI-type I, there is a reduction in the quantity of structurally normal collagen; therefore, the dentine is only mildly affected. Meanwhile, in OI-type III and IV, qualitative and quantitative changes in collagen synthesis result in severe forms of DGI (Shetty et al. 2011). DGI can affect both the primary and permanent dentitions. The primary teeth are more severely affected than the permanent ones (Kaur et al. 2019). The discolouration observed in DGI teeth ranges from yellow-brown to opalescent-grey-type teeth. Teeth with yellow-brown discolouration often exhibit more severe attrition than opalescent-grey teeth (Nguyen et al. 2021). In the current case, the boy's teeth have brownish-grey discolouration, indicating an intrinsic defect in the tooth development.

Besides defective and discoloured teeth, DGI patients may also present with enamel that easily breaks off from the defective dentine because of an altered enamel-dentine junction. In most cases, the enamel structure is normal; however, in some instances, the enamel is either hypomineralised or hypoplastic (Sapir & Shapira 2001). The enamel breakdown causes rapid attrition of dentine, leading to shortening of crown height, loss of occlusal vertical dimension, tooth sensitivity, and pulp exposure (Kaur et al. 2021). Crowns of DGI teeth are usually bulbous with marked cervical constriction. Radiographically, the teeth show normal enamel radiodensity and thickness with early pulp chamber obliteration (Akhlaghi et al. 2016).

Dental caries experiences in patients with DGI with normal enamel structures are reported to be similar to or lower than those of their healthy counterparts (Nguyen 2021). Lower caries prevalence in these patients is because of the rapid attrition of the exposed defective dentine after enamel breakdown that prevents the colonisation of bacteria within the abnormal and reduced dentinal tubules. Hence, rapid attrition protects the

dentine against caries (Devaraju et al. 2014). In the current case, the DGI teeth have normal enamel structure, but evidence of attrition is clinically visible without any pulp involvement.

In managing DGI in OI patients, early detection of patients' dental status is important to prevent adverse consequences. Therefore, seeing these patients early in their dental development is essential. Establishing a good rapport with our medical counterparts dealing with OI cases is vital for early patient referral. In the present case, the orthopaedic team referred the patient early for dental assessment. Therefore, proper assessment treatment could be instituted for the patient. The main treatment objective for cases of DGI is to maintain the affected teeth in occlusion through restorations with either direct adhesive material or stainless steel crowns. The restorative treatments protect teeth from rapid attrition, pulp pathology and early tooth loss due to extraction.

In DGI, progressive deterioration of the dentition is expected to occur over time if no treatment is carried out. The treatment phases for DGI can be divided into immediate, interim and definitive phases. In the current case, the immediate phase of treatment has been executed with proper preventive strategies, restoration of affected teeth and monitoring of the developing dentition. As the patient was only potentially cooperative and required multiple dental treatments, all the restorative treatment was carried out under general anaesthesia in one visit. Both composite strip crowns and stainless steel crowns were used to restore the teeth. These restorations were able to fulfil the treatment objective for the patient. Furthermore, proper case management enables the maintenance of dental aesthetics, phonetics, masticatory efficiency and the occlusal vertical dimension of the patient.

Besides teeth restoration, it is also essential to institute good preventive strategies for DGI patients, such as proper toothbrushing guidance with fluoridated toothpaste, periodic fluoride application, dietary counselling and follow-up. The status of adhesive restorations should be monitored for chipping or cracks because the adhesion of composite to DGI teeth is often compromised. Furthermore, OI patients are also on bisphosphonate therapy to lower their bone loss. There is a risk of developing bisphosphonate-induced osteoradionecrosis in cases where teeth with DGI need extraction.

As for the long-term plan, the severity of DGI in permanent dentition should be assessed early. Defective surfaces can be covered with tooth-coloured restorative material such as composites, glass ionomer cements or stainless steel crowns. To maintain proper functional dentition, a multidisciplinary approach involving the paediatric dentist, oral surgeon, orthodontist, and restorative dentist is essential in managing DGI cases in children and adolescents.

CONCLUSION

Early identification of dental conditions in OI patients is vital to prevent adverse sequelae that lead to debilitating dentition in severe cases. Maintaining deciduous teeth in functional occlusion can improve patients' masticatory function, dental aesthetics, phonetics, and occlusal vertical dimension and prevent malocclusion.

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