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CASE REPORT

Dentinogenesis Imperfecta with Osteogenesis Imperfecta: A Case Report

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Abstract

Dentinogenesis imperfecta (DI) is an autosomal dominant disorder of the dentine tissue that forms the majority of the hard structure in teeth. DI may appear as a solitary dentine inherited disorder (DI-2) or in association with OI (DI-1). DI affects both primary and permanent dentitions and the teeth appear dusky blue to brownish. Osteogenesis imperfecta (OI) is an inherited autosomal dominant disorder of collagen type 1 with many clinical varieties depending on its severity. Type 1 collagen is present in bone and other body tissues such as dentine in teeth. This case report describes a 5-year-old male patient with clinical and radiological features of DI-1, the patient had multiple bone fractures over the years, the patient had multiple carious teeth affecting the primary molars, and presented largely with grayish teeth. Additionally the etiology, clinical features, differential diagnosis, and treatment options have been explored.

Keywords: Bone fractures; Deciduous dentition; Dentinogenesis imperfecta; Osteogenesis imperfecta.

Introduction

Osteogenesis imperfecta (OI) or "brittle bone disease," is caused by mutations in the collagen type I genes COL1A1 and COL1A2 or other collagen genes for other rarer types of OI, introducing production of a defective collagen type I that results in important shift in different tissues in the body.[1] Nonetheless, the defective collagen impacts bones more than other organs.[1] Human dentition can undergo many variations in terms of the number, size, shape, and structure throughout the formation of dental tissue. DI is an example of this and affects dentine.[2] Barret in 1882 was probably the first to recognize DI. Talbot is credited for the premier published report, defining the disorder as an enamel defect, as quoted by Witkop.[3] Hereditary opalescent dentin, a term first used by Skillen, Finn, and Hodges to explain the brown translucent teeth, which are opalescent and lack pulp chamber. [4-6] DI is a localized mesodermal dysplasia affecting the primary and permanent dentition together, in an autosomal dominant way with high penetrance and a low mutation rate.[7] DI is a common dental genetic disease and affects 1:8000 births.[8] The

shade of the teeth differs from brown to blue and sometimes described as amber and grey. Enamel shows hypoplastic or hypocalcified defects in about one-third of the patients and the affected patient's enamel tends to chip away from the defective dentin. The uncovered dentin may then undergo severe and rapid attrition.[7] DI classification was first suggested by Shield: type 1 associated with OI; type 2 similar to type 1 but without OI; and type 3 that is rare and found in the triracial Brandywine population of Maryland.[9] Radiographically, the teeth have bulging crowns with constricted root. At the beginning, pulp chambers are wider than normal and look like shell teeth but they obliterate with time. [9] Histologically the dentin is composed of irregular tubules, with huge areas of uncalcified matrix. Tubules tend to be bigger in diameter and less numerous in a given volume of dentin than in the normal teeth.[10] DI treatment includes the elimination of the infection source or pain, enhancement of esthetics and protection of the posterior teeth from wear. Treatment starts in infancy and usually continues into adulthood with many options including the use of crowns, over dentures and implants depending on the age of the patient and the dental status. If the diagnosis occurs early, a good prognosis can be anticipated.[11] The advantages of early treatment in the primary dentition with DI involves, good dental health while preserving the tooth structure and maintaining ideal aesthetic appearance. The dentist also ensures functionality by maintaining the vertical dimension of occlusion (VDO). Avoiding premature loss of primary teeth while maintaining arch stability are also vital. Considering the pediatric patient, the concept of dental home was embraced. This is a collaboration of the dentist with the patient, which includes a holistic oral care delivered in a concise, accessible, coordinated and family-centered way, to build a relationship with the dental team, while the importance of oral health is reinforced. [12]

Case presentation

A five-year-old Bahraini boy presented to the Dental and Maxillofacial Center at the Bahrain Defense Force Hospital with his father in October 2017. The father reported that "his son had multiple holes in his teeth and sometimes he felt pain." The patient was born at full term and was reported as "normal". At the age of one year, he presented to the trauma clinic with a history of fall that resulted in a fractured arm and left clavicle. At two years of age, the patient fell down and presented again to the trauma clinic with a fracture of the left Humerus, and Orthopedic surgeons treated the fractures. (Figure 1)

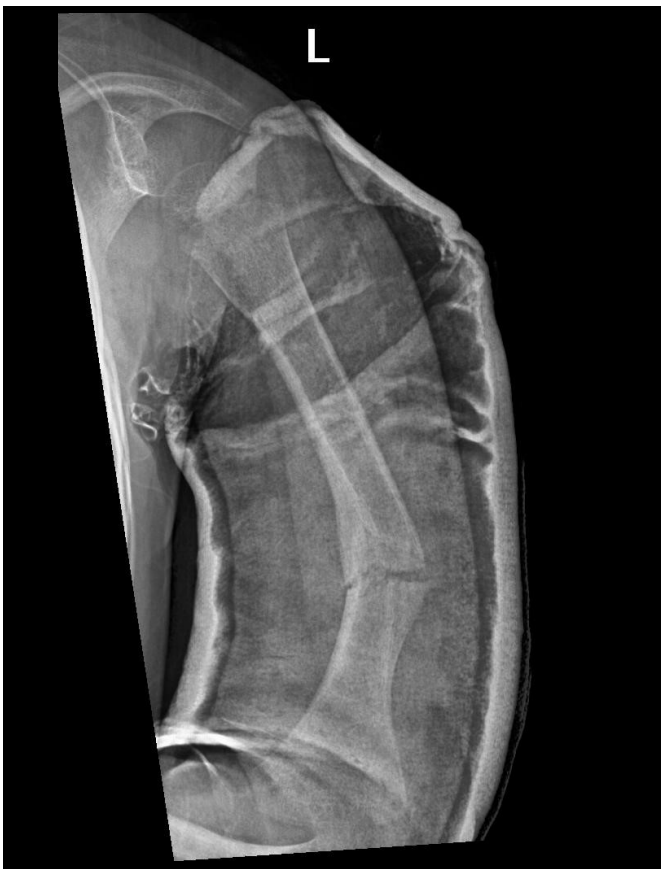

Figure 1: Radiograph showing left Humerus fracture

The patient was referred to the pediatric clinic for further investigations to rule out any underlying diseases including OI. Genetic testing showed a heterozygous mutation in the COL1A2 gene related to OI and inheritance as an autosomal dominant manner. Family history revealed consanguinity among the parents, however the parents did not present with OI. The patient has three sisters and one brother. One of his sisters was found to have translucent teeth, which was similar to that seen in the patient. His other siblings were examined and no abnormalities were detected, the patient's father added that no such changes were reported in the distant family (Figure 2).

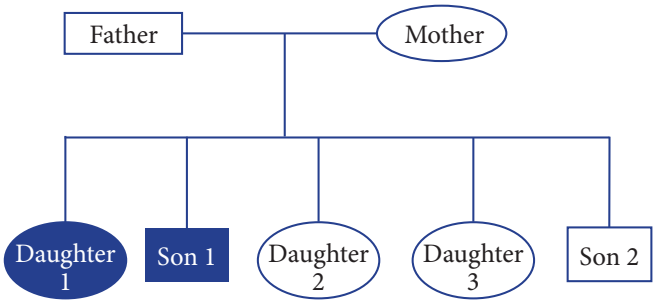

Figure 2: Family Pedigree showing the parents and their five children, two were diagnosed with OI (shaded) and three were normal

On general physical examination, the patient had blue sclera (Figure 3). On extra-oral examination no facial swellings or asymmetry was observed, lymph nodes were non-palpable and the temporomandibular joint appeared normal. The patient's weight and height were well within the normal range. No skeletal deformities were noted. Vital signs were well within the normal values.

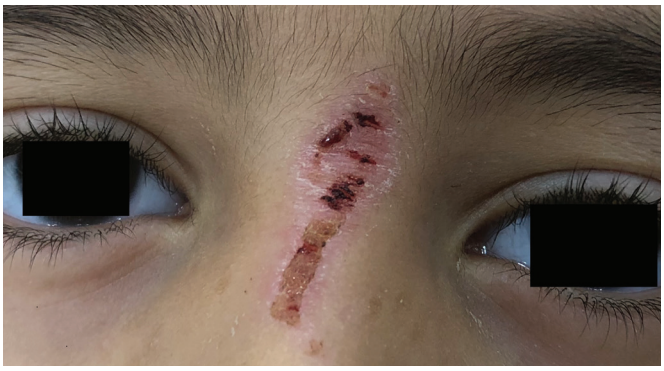

Figure 3: Clinical photograph of the patient with blue sclera

Intra-oral examination revealed normal soft tissue, poor oral hygiene, multiple carious teeth affecting 64,74,84 and badly broken down 52,62 with generally grayish teeth. Photographs and intra-oral periapical radiographs were taken, but it was difficult to take extra-oral radiographs due to the limited patient cooperation. The case was diagnosed as DI with OI (Type 1) and chronic pulpitis in relation to 52, 62, 64, 74, 84. A structured treatment plan was drawn for the dental management of the patient. Firstly, preventive measures like diet advice, oral hygiene instructions, oral prophylaxis, fluoride application and regular follow-up towards dental caries. Secondly, conservative management and/or extraction under local anesthesia, possible relative analgesia or general anesthesia according to the patient's cooperation level. Thirdly, regular follow-up was scheduled every quarterly. The diagnosis and possible sequel of the primary and permanent dentitions were explained to the patient's father. Official communication with the patient's Pediatrician was instituted for collaboration. Through several visits, the above mentioned treatment plan was executed. Teeth were treated under inhalation sedation and local anesthesia with full-metal crowns were placed on 64, 74, 84 using halls technique. Badly broken down teeth 52,62 were extracted under inhalation sedation and local anesthesia (Figure 4).

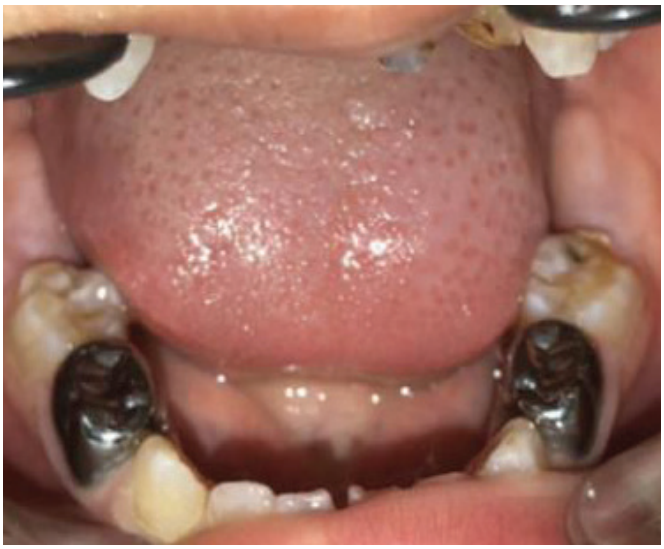

Figure 4: Clinical photograph showing mandibular (lower) primary teeth with stainless steel crowns on (74, 84)

Discussion

One of the most significant challenges for the pediatric dentist is to provide adequate treatment to achieve functional and esthetic restorations in diseases like DI. There are no reports specific to the incidence of OI in the Kingdom of Bahrain. Initial diagnosis and treatment are vital for obtaining a good prognosis, delay in intervention would deplete the available the treatment options.[13] Torija et al included covering the posterior teeth with stainless steel crowns and anterior teeth with stainless steel crowns with open face composite restorations.[14] Restorative treatment in pediatric patients include, glass ionomer with fluoride-releasing and chemically attaching materials are suggested for

occlussally non-stressed areas.[15] Most of the cases presented for treatment in early stages of DI have had good prognosis, patients were able to maintain both aesthetics and function of their dentition. In this case report, the patient was diagnosed with DI with OI (Type 1), pediatric team and dental team followed the multidisciplinary approach. Patient and his father were introduced again to oral hygiene measures, preventive measures, the importance of topical fluoride. Multiple stainless steel crowns for teeth 64,74,84 were placed using halls technique. Badly broken down teeth 52,62 were extracted under inhalation sedation and local anesthesia. Appointment for review was given every 4 months. When the patient presented to the clinic for his first follow-up appointment, the patient was doing well, reported no symptoms, his father reported that the patient was eating better. Oral hygiene instructions were enforced and topical fluoride was applied. The goal was two pronged i.e., therapeutic and preventive measures.

Conclusion

Oral rehabilitation for patients with DI is challenging. It requires a wide range of dental specialties to achieve esthetic and functional stability. This is critical because it contributes to the patient's psychological status and quality of life.

Conflict of interest

The authors declare no conflicts among them for this publication.

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