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CASE REPORT

Clinical manifestations and dental management of dentinogenesis imperfecta associated with osteogenesis imperfecta: Case report

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KEYWORDS

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Abstract Dentinogenesis imperfecta (DI) associated with osteogenesis imperfecta (OI) is a genetic disorder that affects the connective tissues and results in dentine dysplasia. This case report discusses the systemic and dental manifestations of OI and DI in a 4-year-old child, with moderate presentation of both disorders, who was treated at King Fahd Military Medical Complex in Dhahran. Dental treatment included the use of strip and stainless-steel crowns under local anesthesia, as well as behavior modification techniques. Rigorous home care instructions, including reinforcement of the oral hygiene practice and avoidance of any episode that may lead to bone fracture, were discussed with the parents. The case was reevaluated at 3-month follow-up visits, wherein the medical and dental histories were updated, the child's growth was monitored, periodic clinical and radiographic examinations were performed, and the oral hygiene was evaluated via the debris index score and caries risk assessment. Further treatment of the permanent dentition may be needed in the future.

Introduction

Dentinogenesis imperfecta (DI) is an inheritable disorder of tooth development that occurs during the histodifferentiation stage. DI results in structural defects in dentin formation in the deciduous or both the deciduous and permanent teeth. The incidence of DI is 1 in 8000. It can be subdivided into three basic forms: Shields types I, II, and III. Shields type I occurs with osteogenesis imperfecta (OI) due to a defect in type I collagen. Shields type II (also known as hereditary opalescent dentin) is the most common type of DI, and it is not associated with OI. Shields type III is very rare and was first diagnosed in

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a population in Brandywine, Maryland, USA (Shields et al., 1973). Shields types II and III of DI are caused by a defect in dentin sialophosphoprotein gene (DSPP) (Malmgren and Lindskog, 2003; Kim and Simmer, 2007).

OI, also known as "brittle bone disease," is a genetic disorder that affects the connective tissues. A person with OI experiences recurrent, multiple bone fractures. Different types of OI have been recognized, but most are due to mutations in the COL1A1 and COL1A2 genes, which encode the pro-alpha 1 and 2 polypeptide chains of type I collagen. Ligaments, sclera, bone, and dentin are mainly affected (Niyibizi et al., 2000). Abnormalities frequently seen in patients with OI include hearing loss, blue sclera, weak joints, easily bruising, deficient growth, short stature, DI, asthma, and spinal curvature (Marini, 1988). The incidence of OI is 1 per 20,000–30,000 live births (Sillence, 1981).

In 1979, Sillence et al. classified OI into basic types I–IV. Type I is an autosomal dominant mild form of OI. Patients present at preschool age with blue sclera and hearing deficits in 50% of cases. Type II is an autosomal recessive form, in which patients present with blue sclera and perinatal death. Type III, considered to be the most severe form of the disease, is an autosomal recessive form characterized by a progressive short stature and blue sclera that normalize with age. Type IV is an autosomal dominant form with moderate severity. Patients present with normal sclera and hearing; bowing bones and vertebral fractures are also common findings. Most cases (90%) are classified as type I or IV, with or without involvement of the teeth. Types V–VII have since been added to the original classification system. These types do not have type I collagen mutations, but the patients show microscopically abnormal bone and a similar phenotype (Sillence et al., 1979; Elnagdy et al., 2012).

About 50% of children and adults with OI have dental involvement of varying degree and severity (Santili et al., 2005). Although both dentitions may be affected, the deformity is generally more severe in the primary teeth (Waltimo et al., 1996; O'Connell and Marini, 1999). Teeth with DI have certain features, including amber bulbous crowns or gray-brown discoloration, constricted cementoenamel junctions, narrow roots, partial or total obliteration of the pulp chambers, and root canals with evidence of periapical radiolucencies. The enamel is normal, but may shear rapidly due to deficient dentinoenamel junction, resulting in dentine attrition and loss of the vertical dimension (Waltimo et al., 1996).

In cases with a history of multiple bone fractures, OI should be differentiated from suspected child abuse. To this end, it is vital that the clinician obtains good medical and family histories. Although the incidence of OI is rare, the presence of blue sclera, abnormal teeth, hearing problems, osteoporosis, wormian bones, joint laxity, and short stature can be considered as positive findings of OI, as their presence in child abuse is rare or uncommon. If the child has DI, the clinician should rule out the diagnosis of OI (Gahagan and Rimsza, 1991; Kruse et al., 1997; Lund et al., 2000).

This case report describes the clinical manifestations of a 4-year-old boy with DI and OI, as well as the child's medical and dental treatments with 18 months of follow-up after comprehensive dental management at King Fahd Military Medical Complex in Dhahran, KSA. The study was approved by the appropriate ethics committee related to King Fahd Military Medical Complex. Subjects gave their informed consent to participate in the study.

Case report

A 4-year-old boy was referred by a general dentist for comprehensive dental treatment. The child was diagnosed with OI at 19 months of age at King Fahd Military Medical Complex in Dhahran, KSA. Since 3 months of age, he experienced repeated and multiple fractures, especially recurrent fractures of the left femur, with frequent cast applications (Fig. 1a and b). Wormian skull bones (i.e., small puzzle-like pieces of intrasutural skull bones) were seen in his lateral skull radiograph at 19 months of age (Fig. 2). He had a mild bowing abnormality, with curved thin bones in both legs (Fig. 3). However, no bowing of the legs was detected at 4 years of age (Fig. 4). The patient was born after a full-term pregnancy, and his immunizations were up to date. He was crawling at 8 months, walking at 15 months, and weaned at 2 years. No delay in speaking was noted. He was his mother's fourth live birth, and no member of his family has the same disability. Systemic findings were unremarkable (Table 1). The child was on cyclic therapy with pamidronate (10 mg by intravenous infusion over 6 h OD), oral vitamin D3 (400 IU, OD), and oral calcium (200 IU) every 3 months.

Past dental history included extraction of tooth #74 under local anesthesia due to a dentoalveolar abscess and after the use of augmentin (156 mg/5 cc TID for 7 days), and pulpotomy of tooth #84 with negative behavior. Orofacial clinical examination revealed mild bitemporal bossing with a convex facial profile, competent lips, and eyes with normal sclera (Fig. 5a and b). There was no evidence of rib fracture or upper rib abnormality. Lymph nodes were palpable on both sides. The tongue, lingual frenum, soft and hard palates, and tonsils were all normal. Intra-oral examination showed a dentoalveolar abscess related to tooth #64 in U-shaped upper and lower arches, with generalized marginal gingivitis. The debris index score was 32 (Fig. 6a and b). The patient had a mesial step terminal plane with a class I canine relationship on both sides. The midline was coincident, with a 50% overbite and 2 mm overjet (Fig. 7a–c). The teeth were brown-gray in color, with constricted bulbous crowns and attrition.

Radiographic examination included full-mouth radiographs, two posterior bitwings, four periapical radiographs for the posterior teeth, and two periapical radiographs for the anterior teeth. OPG could not be performed because the child was too small to fit in the OPG machine. Radiographically, the primary teeth appeared to have short constricted roots, dentine hyperatrophy, and partially obliterated pulp. The presence of the developed permanent teeth buds was also evident. The child's dental age by radiography was similar to his chronological age. Evaluation of the periapical X-rays revealed normally developing occlusion, no supernumerary teeth, and the presence of unerupted upper and lower first permanent molars and permanent canines (Fig. 8).

The patient was treated in the dental clinic under local anesthesia (xylocaine 2% with epinephrine 1:100,000). Behavior modification techniques included the tell-show-do technique, positive reinforcement, distraction, and (sometimes) voice control. Treatment was divided into 10 visits. Teeth #55, 54, 65, 75, 84, and 85 were protected with stainless-steel crowns. Tooth #64 was extracted due to the presence of a dentoalveolar abscess. Teeth #51, 52, 61, 62, 71, 72, 81, and 82 were covered with celluloid strip crowns. Teeth #53, 63, 73, and 83 had