Clinical features, dental findings and dental care management in osteogenesis imperfecta

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This article reports the case of a child diagnosed with type III osteogenesis imperfecta, assessing the clinical features observed, with emphasis on the craniofacial, oral and dental findings, and describes the dental care management rendered for this special needs patient. The issues addressed and discussed throughout the paper shows that, as far as the oral health professionals are familiarized with the disease-specific clinical manifestations and the possible implications of this condition, there are no hindrances for a successful, high-quality dental and behavioural management.
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INTRODUCTION

Osteogenesis imperfecta (OI) is an inherited disorder of the connective tissue resulting from mutations in either the collagen 1a1 gene or the collagen 1a2 gene on chromosomes 17 and 7 respectively, which encode for type I collagen chain formation. The disease causes either a decrease in collagen synthesis or the production of structurally defective collagen. As a consequence, all tissues rich in type I collagen may be affected.[1-3] The clinical manifestations most commonly observed in patients with OI include abnormal bone formation, growth deficiency, bone fragility, blue sclerae, hearing losses, skin thinness, joint laxity, hypermobility and dentinogenesis imperfecta.[2-4] The incidence of osteogenesis imperfecta is estimated to range from 6 to 20 in every 100,000 newborns, whereas its prevalence is reported to range from 4 to 10 in every 100,000 individuals.[2] Due to the variety of clinical manifestations usually associated with this disorder, several categorizations have been proposed. Four major types of osteogenesis imperfecta are often distinguished on the basis of clinical features and disease severity, according to the classification suggested by Sillence.[5]

The patient documented in this case report was diagnosed with type III osteogenesis imperfecta. This disorder can be inherited in either an autosomal dominant or an autosomal recessive pattern and is acknowledged as the most severe form of OI in children. It has been reported that, although the weight and stature are normal at birth and the limbs appear neither short nor deformed, a progressive deformity of long bones and spine occurs during childhood, which precludes bones from growing normally and results in great difficulty for independent mobility. The final stature of patients with the disorder is much shorter than average. The sclera is blue in infancy and becomes progressively normal with time. Other relevant signs and symptoms often associated with type III OI include the trend to bone fractures, possible hearing loss and dentinogene-

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sis imperfecta. There have been reports of death during childhood, while other individuals can reach the third decade of life.[1,2,3,4,5] Children with severe osteogenesis imperfecta live with several physical limitations and restrictions, but the hindrances they must deal with lessen proportionally during adolescence, when the resistance of bone structure increases. Despite the obstacles they face, individuals with OI may enjoy productive and fulfilling lives well into their adult years. In certain cases, patients confined to wheelchairs are able to walk again, but this naturally will depend on the severity of the damage accumulated along the previous years.

Pamidronate, a bisphosphonate drug, is administrated using a cyclical intravenous dosage of 7.5mg/kg/year at four- to six-month intervals. This treatment has been reported to yield biochemical, radiological and histomorphometric evidences of decreasing the rate of bone loss, in addition to reduce chronic pains and improve patient mobility.[3]

Osteogenesis imperfecta may be accompanied by dentinogenesis imperfecta (DI). Craniofacial, oral and dental manifestations are frequently observed and may be of high diagnostic importance if the physical signs and symptoms of OI are not consistent and the medical diagnosis is uncertain. Teeth affected by DI present with an opalescent grayish, brownish or yellowish hue. Although the enamel is normal in thickness and radiodensity, it is often dislodged, thus leaving the softer dentin exposed to wear, which may lead to rapid and severe attrition. The radiographic findings reveal ampullar extensions of the pulp chamber,[6] shortened roots and bulbous crowns that constrict at the cervix. Pulp chamber and root canals may be partially or totally obliterated and, in some cases, periapical radiolucency is also noticed.[2]

On histological examination, dentin is characterized by the existence of irregular dentinal tubules, amorphous areas, embedded cells and poor calcification.[2,7,8] In addition, occlusal disorders are often observed. Skeletal class III malocclusion has been described in the majority of patients diagnosed with type III and type IV osteogenesis imperfecta. Anterior and posterior open bites and crossbites have also been reported.[1]

This article reports the case of a child diagnosed with type III osteogenesis imperfecta, addressing the major clinical features observed, with emphasis on the disease-specific craniofacial, oral and dental findings, and describes the dental care management rendered for this special needs patient.

CASE REPORT

An 8-year-old male patient sought treatment at the clinics of the *Center of Formation of Human Resources Specialized in Dental Care for Special Patients*, at Ribeirão Preto School of Dentistry, University of São Paulo. An overview of the patient's files revealed that he was brought for the first time in 1998, at the age of 4.

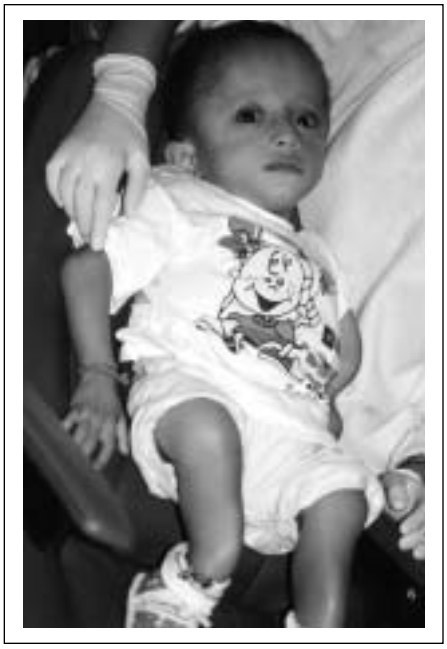

Figure 1. Appearance of the patient at the age of 4 years

The past medical history revealed that the child had sustained bilateral clavicle fractures during the intrauterine period and was born with normal weight and stature by cesarean section. At birth, the child presented with fractured right superior and inferior limbs, sternum and diaphragm deformities, ears with low implantation and blue sclerae and was diagnosed with type III OI.

The genetic investigation showed an autosomal dominant pattern of inheritance. The patient's maternal grandparents are second-degree cousins and his maternal great-grandparents are first-degree cousins. However no reports of previous cases had ever been documented in his family.

During childhood, the patient experienced repetitive pneumonia and sustained several bone fractures after minor traumas. According to the medical records, the patient presented osteopenia, osteosclerosis, macrocephaly, conductive hearing loss ranging from mild to moderate, right thoracic scoliosis, accentuated sacrococcygeus angulation, loss of vertebral bodies height with increased biconcavity, bilateral curvatures of the forearms, thighs and legs, and low stature and weight (83 cm and 17 kg, respectively). The coloration of his sclerae was becoming progressively less bluish (Figure 1).

When the patient first came to our service in 1998, at the age of 4 years, he had multiple carious lesions, ranging from moderate to severe degrees, in the teeth 51, 52, 53, 61, 62, 63, 64, 74, 75, 84 and 85. Generalized gingivitis and heavy supragingival and subgingival plaque

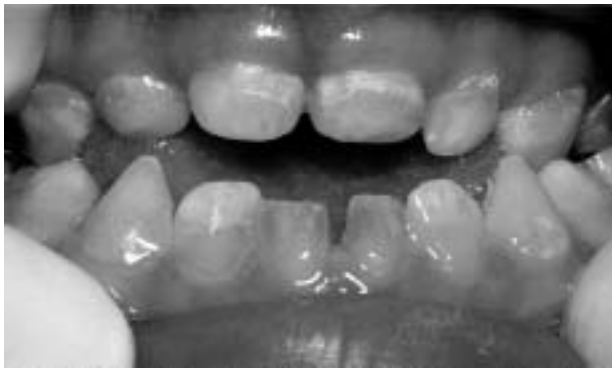

Figure 2. Primary dentition affected by dentinogenesis imperfecta

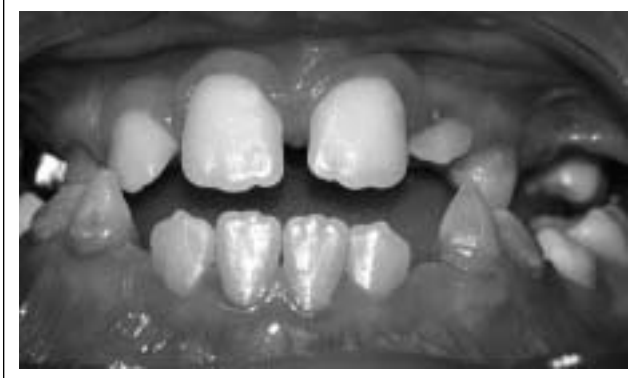

Figure 3. Intraoral clinical appearance of the mixed dentition

deposits denoted defective oral hygiene habits. The primary dentition exhibited typical signs of dentinogenesis imperfecta, such as fractured or dislodged enamel with exposed dentin, attrition, discoloration ranging from a yellowish to a brownish hue, microdontic appearance of affected mandibular central incisors, anterior open bite and bilateral posterior crossbites (Figure 2).

Since the patient was at high caries risk and activity, he was enrolled in an individual-targeted prevention program aimed at improving his oral health status. These measures comprehended elementary instruction on dental care, diet counseling on low-sucrose intake and training of the parents on mechanical removal of dental plaque. The treatment strategies included caries excavation and temporary sealing of carious cavities with glass ionomer cement, use of a disclosing agent to reveal dental plaque, professional prophylaxis and topical applications of 0.12% chlorhexidine gluconate and 1.23% acidulated phosphate-fluoride gel. The goals of this phase of the therapy were mainly to motivate the patient, stimulate his commitment with the treatment and provide him with an effective psychological conditioning.

The surgical/endodontic/restorative treatment was then performed at sequential weekly appointments and all procedures were accomplished under local anesthesia and rubber dam. Pulpectomy was performed on tooth 52, pulpotomy was performed on teeth 75 and 85, and the residual roots of teeth 54, 55 and 65 were extracted. The rehabilitation of decayed and severely damaged teeth was provided by means of resin composite or amalgam restorations and stainless steel crowns. Counseling on mechanical removal of dental plaque and dietary habits were strongly reinforced at each visit. Once the restorative procedures were completed, the patient was required to return for follow-up recalls, but, regrettably, he failed to attend for the scheduled visits and did not show up for a long period.

In 2002, four years later, the child was readmitted at the clinic for dental care. The intraoral examination

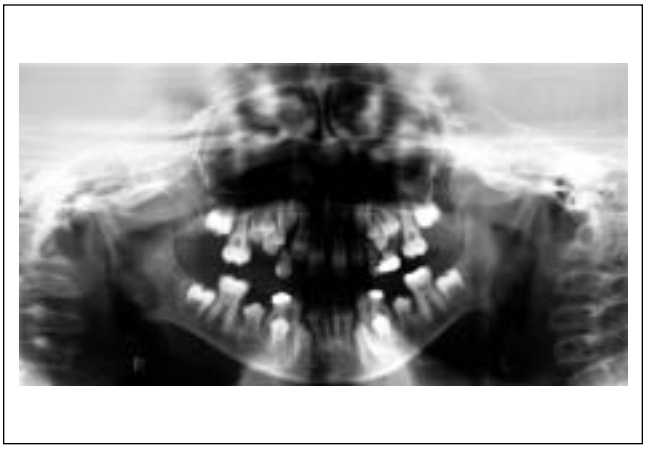

Figure 4. Panoramic radiograph of the patient at the age of 10 years, showing typical features of dentinogenesis imperfecta, such as pulpal calcifications in the permanent first molars. Notice the restorative/rehabilitative treatment accomplished on both dentitions.

revealed that, although his motor skills had improved remarkably, inflamed, red gingiva and supragingival/subgingival plaque deposits, denoted a persistent poor oral hygiene. The occlusal surfaces of the permanent first molars were decayed and demanded either amalgam or resin composite fillings. The primary teeth exhibited characteristics of dentinogenesis imperfecta, whereas the permanent dentition was clinically normal (Figure 3), even though the radiographic examination revealed the existence of pulpal calcifications (Figure 4). The patient presented Angle class III molar relationship with anterior open bite and bilateral posterior crossbites (Figure 5).

The patient's high-risk condition and the need for a comprehensive individual-based treatment were thoroughly discussed with the parents. The reasons for his dropout few years before were addressed and possible shortcomings of the treatment plan settled at that time were identified and overcome. Much time and effort were invested in highlighting that homecare oral hygiene should be supported by professional care pro-

Figure 5. Appearance of the patient at the age of 10 years. Increased resistance of bone structure and muscle strength is evident. A class III skeletal pattern, with retrusive tendency of the maxilla is still evident.

Figure 6. The patient's motor skills have improved remarkably and he is now able to handle manual activities, including handcraft.

vided at the clinics and the parents were encouraged to monitor closely the child's attempts at toothbrushing and assist him in plaque removal.

Every three months (and later bi-annually) appointments were scheduled and patient returned for revaluation of the restorative/rehabilitative treatment, as well as for monitoring tooth wear, onset of new lesions and general oral hygiene status. On account of the difficulty on toothbrushing clearly evidenced during the treatment, professional prophylaxis and topical applications of chlorhexidine gluconate and fluoride were done at each visit.

After 2 years of follow-up, the patient has maintained good oral hygiene. New caries lesions or severe gingivitis were not detected in the latest routine examination. Tooth wear has been monitored and the child will be referred to an orthodontist opportunely.

The patient has been following a medical treatment routine with bi-annual intravenous pamidronate dosages and daily-administrated calcium supplements tablets. The resistance of bone structure, muscle strength and the motor skills enhanced with the medication, and his general health conditions improved remarkably. Most importantly, his self-esteem and motivation increased proportionally, which prompted him to find out new abilities and interests. Despite the important limitations and impairments derived from the disease, he has been dedicated to a series of manual activities, including handcrafts (Figure 6). This is undisputable evidence that having a severe disability and being disabled may be two very different things.

DISCUSSION

The prevalence of dentinogenesis imperfecta has been shown to range from 8 to 40% in patients with type I osteogenesis imperfecta, from 43 to 82% in type III OI, and from 37 to 100% in type IV OI[2]. The primary dentition is more severely affected than the permanent.[2] Our clinical findings are consistent with those reported in the literature, since the primary teeth presented with typical signs of DI - discoloration ranging from a yellowish to a brownish hue and easily detachable enamel, whereas the permanent teeth lacked clinically detectable alterations. The radiographic examination revealed the presence of calcification nodules in the pulp chamber of the permanent molars, which is, according to O'Connel and Marini,[1] a radiographic evidence of dentinogenesis imperfecta often observed, even in teeth clinically unaffected by DI. Likewise, normally colored teeth and lack of radiographic signs do not necessarily indicate absence of DI on histological examination.[2]

Mild forms of OI are sometimes difficult to be diagnosed. Therefore, for patients in whom the diagnosis is uncertain, clinic/radiographic examination of oral conditions is wise and may be a helpful approach. DI-specific oral manifestations are commonly observed in patients with OI and the evidence of disturbances in dental development can be crucial for establishing the OI diagnosis.[2]

Although the enamel of teeth affected by DI has normal structure and normal or infrequently decreased mineral content,[2] it tends to crack away from tooth surface, thereby exposing the softer dentin that undergoes

rapid and severe attrition.[4] Enamel dislodgment may be attributed to the fact that the dentinoenamel junction (DEJ) in teeth of patients with OI is smooth.[9] Nevertheless, sometimes, the DEJ appears with normal scalloping and the disruption is assumed to occur in a low-mineralization dentin zone.[1,4] Indeed, the reasons for enamel shearing are still unclear, but a low density of apatite crystals in dentin as well as abnormal orientation of collagen fibers combined with the enamel irregularities, have also been suggested as possible factors involved in enamel detachment.[9]

Children diagnosed as having OI should be seen by a dentist as soon as possible after the eruption of the primary anterior teeth in order to determine whether DI involvement exists. Early dental care of patients with DI is strongly recommended to refrain them from undergoing painful symptomatology or loss of vertical dimension, as a result of an accentuate wear of tooth structure. Extensively worn or damaged teeth should receive prosthetic crowns for protection of the remnant dental complex, reestablishment of the vertical dimension and esthetic improvement.

When the patient documented in this article sought treatment for the first time, at four years of age, he had almost all primary teeth decayed and/or severely damaged. When he returned for treatment four years later, occlusal caries lesions had developed in the permanent maxillary and mandibular first molars. However, these teeth do not appear to be more susceptible to caries than normal teeth, and thus high-caries activity would be more likely due to inappropriate oral hygiene habits.[10,11]

O'Connel and Marini[1] have reported that class III dental malocclusion occurred in 70% to 80% of types III and IV of the OI population, with a high incidence of anterior and posterior crossbites and open bites. Accordingly, the patient described in this report had a class III skeletal pattern due to a maxillary skeletal retrusion. The orthodontic and surgical procedures for correcting malocclusions in OI patients are very difficult because of the easy fracturing trend. Therefore, caregivers of OI children must be properly instructed that the maintenance of sound primary dentition is particularly important to ensure normal alignment of the permanent teeth and minimize the need for extensive orthodontic treatment.

Regarding dental care management of children with osteogenesis imperfecta, it is noteworthy that most clinicians appear unwilling of providing assistance to these individuals, because of the fear of causing maxillary and mandibular fractures. It is relevant to emphasize that, in the majority of the cases, no particular precautions are needed other than those that would be taken anyway when dealing with a pediatric patient. However, OI children should naturally be handled with tenderness, avoiding impetuous movements and excessive force. During extractions, the tooth should be carefully luxated using small amplitude movements and minimal force. The child must be made himself/herself comfortable in the dental chair. The use of pillows, foam pads, rolls or similar devices improves the patient's comfort. Most of these children have normal intellectual development and are able to cooperate with the professional.

The routine administration of prophylactic antibiotics should not be indicated for dental treatment on the basis of the presence of pins, plates, screws or orthopedic rods.[1,12] The risk/benefit and cost/effectiveness ratios fail to justify a prophylactic antibiotic regimen as routine procedure and, indeed, it is likely that many more oral bacteremias are spontaneously induced by daily events than are dental treatment-induced.[12] However, it is intended neither as the standard of care nor as a substitute for clinical judgment, since it is impossible to make recommendations for all conceivable situations in which bacteremias originating from the oral cavity may occur. So, dentists must exercise their own clinical judgment with each individual situation and decide whether or not antibiotic prophylaxis is appropriate.[12] Most of patients suffering from osteogenesis have undergone surgical interventions for insertion of metal bars or rods to align and strengthen the fractured bones. When dealing with patients with certain health conditions, the likelihood of developing secondary infections by transient bacteremias after dental procedures should always be considered. In these cases, prophylactic premedication before highly invasive dental procedures (e.g. extractions and periodontal surgeries) may be an advisable approach. Yet, any perceived potential benefit of antibiotic prophylaxis must be weighed against the known risks of antibiotic toxicity, allergy, and development, selection and transmission of microbial resistance.

The findings of an earlier study[1] have raised the possibility that patients with osteogenesis imperfecta may also be at risk for developing an allergic response to latex.

This article discusses some of the most relevant features regarding the dental care management of children with OI. Is our expectation that the issues addressed throughout this paper may somehow be helpful to elucidate the syndrome-specific dental findings and conditions and, perhaps most importantly, provide dental professionals with research-based information that can make them more acquainted with this condition.

Coping with the problems of a severe disorder like OI may be overwhelming. In many ways, parenting a child that suffers from such a debilitating and life-threatening bone disease is certainly a challenging, lifelong experience. The physical disabilities, limitations and medical problems of these children are so demand-

ing that, sometimes, dental care is understandingly not considered a priority. However, parents must be instructed and made conscious that defective oral hygiene habits lead to dental problems that will inherently bring additional and avoidable pain, discomfort and complications for their children. When a supportive, motivational oral care management is rendered along with the family partnership, noticeable and lasting outcomes in oral health should be expected for these special patients.

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