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CASE REPORT

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Imperfect osteogenese: child case report

Osteogênese imperfeita: relato de caso infantil

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ABSTRACT

Osteogenesis Imperfecta leads to alterations in type 1 collagen fiber, apart from causing bone fracture, blue sclera and other related deformities. As few medical records are available in the field of dentistry regarding these alterations, having a better understanding of this medical disorder and its dental management has become a matter of extreme relevance if one is to provide adequate treatment for patients suffering from this medical condition. This paper reports the case of a 2-year old patient with Osteogenesis Imperfecta who received treatment as part of the *Acolher Project* – PNE run by the Federal Fluminense University in Rio de Janeiro, Brazil.

KEYWORDS

Imperfect osteogenese; Oral pathology; Pediatric dentistry.

RESUMO

Osteogênese Imperfeita leva a alteração na fibra colágena tipo I, ocasiona fraturas ósseas, escleras azuladas e outras deformidades. Essa alteração apresenta poucos registros científicos no ramo da odontologia sendo extremamente importante para as áreas de Pacientes Especiais, Odontopediatria e Odontologia Hospitalar no entendimento dessa desordem e o manejo odontológico. Este estudo relata o caso de uma paciente de 2 anos de idade com Osteogênese Imperfeita atendida no Projeto Acolher-PNEUFF

PALAVRAS-CHAVE

Osteogênese imperfeita; Patologia bucal; Odontopediatria.

INTRODUCTION

Osteogenesis Imperfecta (OI) is a rare disease that is characterized by bone fragility, recurrent fractures with secondary deformities, premature deafness, blue sclera and dentinogenesis imperfecta1. OI is associated with mutations in the genes of type 1 collagen fiber or in genes responsible for the processing of type 1 collagen protein. The occurrence of alterations in the collagen fiber is the key element to look for when it comes to the pathophysiology of OI. The frequency ratio of such alterations ranges from 6-7:100000 individuals diagnosed with the disease [1]. OI is related to a group of hereditary alterations, most of which are regarded autosomal dominant derived from various mutations in one of the genes that codify the alpha chains – COL1A1 and COL1A2 of type 1 collagen. Mutations in one of these genes can be identified among 80-90% of OI patients [2].

Type I of OI is the term employed to characterize patients with low degree of deformities, normal stature, fewer fractures, and with neither dentinogenesis imperfecta nor greater deformities of the long bones. Type II of OI is the most serious condition that often results in the death of the affected patients during perinatal period. Type III of OI is the typical case that is generally described in the literature, and which is characterized by patients that exhibit moderate to severe condition of the disease,

triangular facies, low stature, deformities of the long bones and dentinogenesis imperfecta. The condition exhibited by the rest of the patients is denominated Type IV of OI; this last group of patients is largely heterogeneous in the sense that they exhibit differences in terms of both degree of severity and clinical characteristics [3] as shown in Table 1.

The clinical symptoms presented by patients with OI also include an increase in elasticity with capsular ligament laxity, increased tendency to bleed, triangular facies and alterations in the skin. Other less frequent symptoms may include inguinal hernia, late congenital anomalies and mental retardation [4]. Some syndromes, including the Marfan Syndrome (MFS) and the Ehlers-Danlos Syndrome, may exhibit an association with Osteogenesis Imperfecta. The Marfan Syndrome (MFS) is an autosomal dominant disorder of the connective tissue with a prevalence ratio of 1:5000-10000 newborn babies. This disorder is derived from mutations in the FBN1 gene that codifies fibrillin-1, which is an important component of the microfibrils of the extracellular matrix. MFS is a multisystemic disease and its main manifestations occur in the skeletal, ocular and cardiac regions, with the latter being the main cause of morbimortality related to this pathology [5]. The Ehlers-Danlos Syndrome (EDS) is the term given to a group of typically rare disorders related to the connective tissue. This disorder is characterized by hereditary alterations in the genes that affect the synthesis and processing of different forms of collagen. The global prevalence ratio of EDS is 1:5000, and it has no specific racial ou gender predominance [6].

Dentinogenesis imperfecta (DI), also referred to as hereditary opalescent dentin or odontogenesis imperfecta, is a kind of anomaly that occurs when the odontoblasts – the cells responsible for the synthesis or production of dentin, fail in their differentiation, producing a dentin with abnormal structure, leading to the development of opaque teeth with brownish or gray-blue color [7]. Evidence shows that, likewise OI, type I DI is caused by mutations in the genes of type I collagen, while the other types of DI are associated with mutations in the gene that codifies dentin sialophosphoprotein (DSPP) - found to be the most abundant non-collagenous protein of this tissue, which plays an important role in the biomineralization of the dental tissue. With the aid of radiography, the teeth affected by DI may be seen to display a tendency towards pulp canal obliteration over time, in addition to the occurrence of cervix cervical constriction as well as the presence of short and thin roots. Periapical alterations can be observed as a result of the severe exposure and attrition of the dental tissue [8].

As part of the dental treatment, the suggested recommendations include the preservation and re-establishment of the vertical dimension of occlusion; the protection of the posterior teeth from attrition; and aesthetic restoration. The use of steel dental crown for the posterior deciduous teeth can be considered a preventive mechanism against severe abrasion of the dental structure. For the permanent teeth, one may decide to reconstruct the anterior teeth with either composite resin or metalo-ceramic and the posterior teeth with the dental crown fused to the molars, as well as prosthetic rehabilitation. One should be mindful, however, that these teeth cannot be used as pillars as the roots tend to undergo fracture with the exertion of pressure. Overdentures are also recommended in severe cases and endodontic treatment should be undertaken without much delay when the need arises.

Teeth with periapical rarefaction and radicular fractures must be extracted; the extraction may be difficult as a result of the dentin fragility. Based on the guidelines of ADA (1997) for the dental treatment of OI patients with pins, plates ou screws, the administration of antibiotics for infection prophylaxis is not indicated [9].

The difficulties associated with dental treatment – which have already been pointed out by Nowak (1979), such as the extreme bone fragility of the jaw teeth which results in (jaw/ mandibular/ alveolar bone) fractures; and the possibility of dislocation of the temporomandibular articulation; as well as the condylar fractures in the movements involving the maximum opening of the mouth, have contributed towards providing guidance, especially, to surgical procedures regarded indispensable for the restoration and maintenance of the dental health of patients [10].

In most cases, patients seek dental treatment when they experience pain or for aesthetic purposes; this renders the procedure more invasive and complicated, once they have not been initially subjected to less invasive procedure for prevention. What makes this present case a special one has to do with the age of the patient, being extremely young, coupled with the absence of carious lesions; these factors enable one to work on preventive procedures.

The use of a multidisciplinary team is essentially indispensable in cases involving the general health of the patient. Indeed, the interaction of health professionals of different speacialties, including pediatry, genetics, radiology, pediatric dentistry and orthodontia, helps to join forces and knowledge so as to cater for the holistic needs of the patients and/or for the improvement of their quality of life and well-being [9].

The present paper seeks to report the dental management of patients with Osteogenesis Imperfecta through a clinical report of an infant patient.

Table 1 - Types of osteogenesis imperfecta

Type I	Type II	Type III	Type IV
Minimum to moderate bone fragility	Severe bone fragility	Moderate to severe bone fragility	Heterogeneous
Normal stature	Perinatal or neonatal death	Low stature	

CLINICAL REPORT

A 2 year old female patient leucoderma turned up at the *Acolher-PNE Project* run by the Federal Fluminense University accompanied by her legal guardian for the assessment of her dental condition.

A judicious anamnesis was conducted with the patient's guardian, where a wide range of data was collected. The data collected included: the gestational period, interferences suffered during pregnancy, any medications used by the mother during pregnancy, the mother's age at the time when she got pregnant, the eating and deleterious habits of the patient, the presence of pain in any region of the oral cavity, medications used by the patient as well as data on oral hygiene. In addition, the patient was subjected to clinical, extra physical (Figure 1) and intra oral examinations, where the following was detected: absence of lesions in the oral and perioral tissues, presence of soft tissues with normal consistency and coloring, and the absence of caries and periodontal disease. The patient displayed teeth with brownish coloring as well as the absence of elements in her oral cavity (Figure 2 and 3); these features underscored the diagnosis of Odontogenesis Imperfecta. The patient exhibited the elements 54,52,51,61,62,64,74,72,71,81,82,84 in her oral cavity.

The legal guardian of the patient presented a medical report from the Fernandes Figueira Medical Institute proving that the patient has been diagnosed with Osteogenesis Imperfecta. The following data were found in the medical report: shortening of the long bones and alterations of the left foot bones, pronounced shortening of the femur in a bilateral fashion (Figure 4) and a malformation of the right foot (Figure 5). There was also a report of a fracture of the right femur with intra-uterine consolidation.

The legal guardian of the patient was asked to present a periapical and panoramic radiography of the patient's incisors so as to assess the intra-bone situation of the dental elements.

The following was done during the second medical consultation: a treatment plan was devised, prophylaxis was undertaken using prophylactic paste (Figure 6) and neutral fluor (fluoride) gel was topically applied.

Figure 1 - Blue sclera.

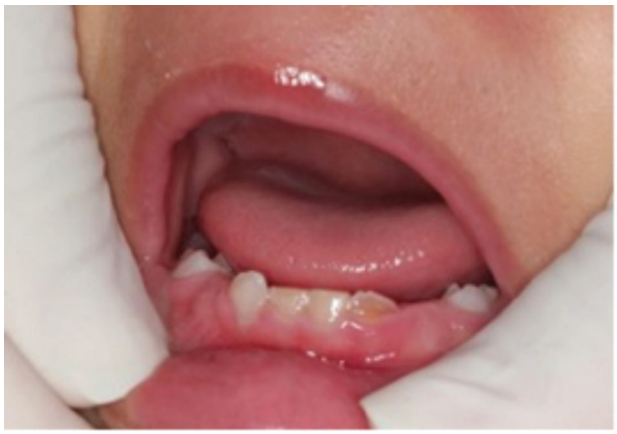

Figure 2 - Lower arch.

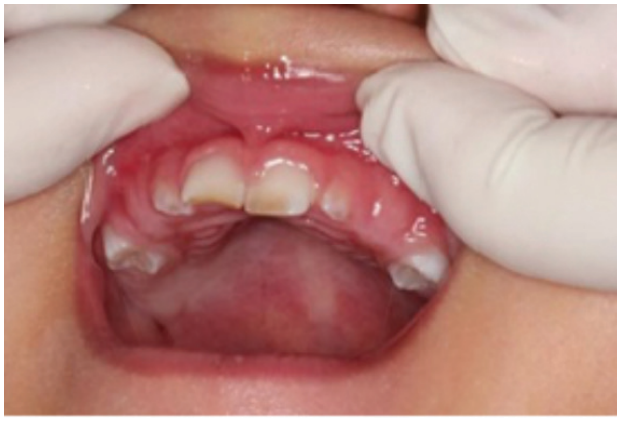

Figure 3 - Upper arch.

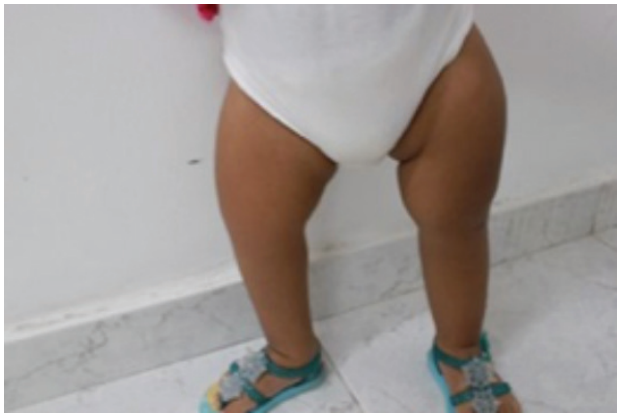

Figure 4 - Arched lower limbs.

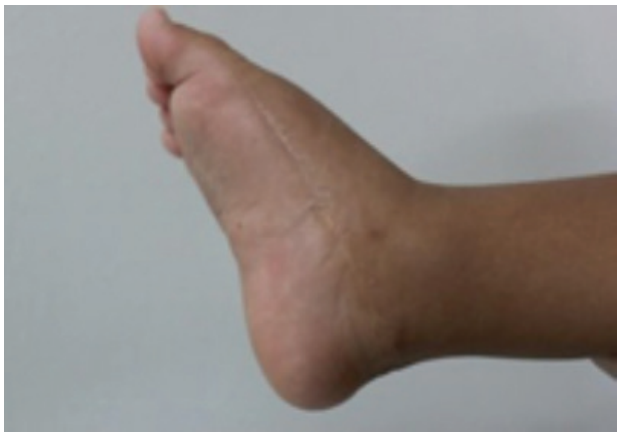

Figure 5 - Surgical Scar.

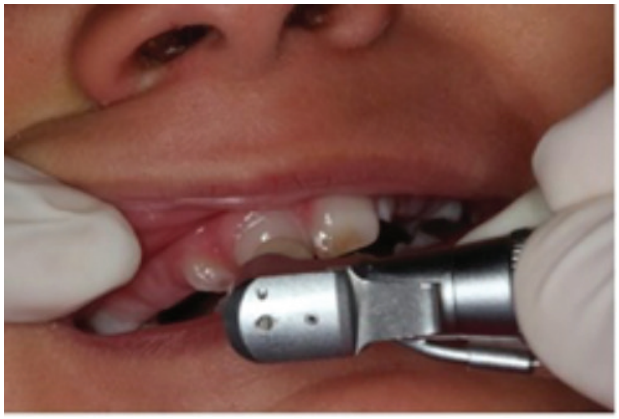

Figure 6 - Prophylaxis with Prophylactic paste.

DISCUSSION

The patient displayed bone fractures and bone malformation; these features provided the basis for the diagnosis of Osteogenesis Imperfecta (OI). Osteogenesis Imperfecta is a rare disease characterized by bone fragility, recurrent fractures with secondary deformities.1 Through clinical assessment, a brownish coloring was noted in the dental elements - a typical feature observed in patients with OI. The term applied to this abnormal coloring condition is Dentinogenesis Imperfecta (DI). DI is a genetic disease that affects teeth development, leading to grayish blue or yellowish brown discoloring with excessive translucency. The teeth become weak and suffer from rapid abrasion [11].

Dentition management in the presence of DI depends on gravity and on whether the enamel fracture occurs and results in excessive degradation of the teeth. The placement of dental crown is the recommended treatment in such cases, since intra-crown restorations are not maintained in teeth suffering from DI which are affected by the loss of enamel and excessive loss of dentin [12].

In primary dentition, stainless steel dental crowns in the molars can be used in order to avoid dental abrasion and to maintain a vertical dimension of occlusion. The aesthetic aspect of the teeth can be improved by using composite coatings or composite strip crowns. However, in cases whereby the dental treatment of the infant patient is sought at a later period the teeth may have suffered from abrasion up to the gum level, and the only treatment option available in such cases is to provide full dental prostheses. Children often adapt well to full dental prostheses though these prostheses need to be revised regularly and the prostheses should be removed as the child becomes older. When the dental abscesses develop, the pulp canal therapy fails to produce successful results, as such the removal of the affected teeth becomes necessary. In much younger children, where cooperation is limited or the level of treatment required is greater, general anesthesia may be necessary in order to facilitate treatment. In some cases, the parents or their wards oeremoval of sources of pain or infection only [13].

It is worth noting that the earlier one pays the needed attention to hygiene habits, norms of conduct and diet, the smaller the risks of experiencing dental problems in the future [14]. A diet rich in sucrose not only affects one's dental health but may also provoke changes in metabolism, such as diabetes, hypertension and cardiovascular changes [15].

As many of the professionals in the field are not used to dealing with this kind of systemic complication, the legal guardian of the patient decided to bring her to the *Acolher-PNE Project* run by the Federal Fluminense University - a reference center for the treatment of children with special needs. The patient's guardian was found to be extremely attentive and interested in receiving orientations on how to maintain the patient's oral hygiene; this is reflected in the absence of caries and periodontal disease in the patient's dental structure. As aforemetioned, the case report presented in this work is about a female baby aged 2 years; hence, the follow-up of the patient's condition is of extreme relevance as it enables one to observe the modifications of her condition over the course of the growth of the infant.

Owing to her dental and physical conditions, the patient requires a constant follow-up by dentistry professionals so as to ensure that the dental fragility does not transform into a problem for the patient in the form of dental fractures or dental abscesses which are much common among this group of patients due to extreme calcifications of the endodontic canal.

In the future, a thorough evaluation will be carried out regarding the use of metallic and ceramic dental crowns, removable prostheses or even implants in adult age. Above all, careful considerations will be given to the patient's limitations, including the care in managing the patient's oral cavity and the head in order to avoid bone fractures.

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