Dentinogenesis imperfecta type II: approach for dental treatment

Dentinogênese imperfeita tipo II: abordagem de tratamento odontológico

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Resumo
Introdução: A dentinogênese imperfeita (DI) é uma desordem hereditária no desenvolvimento da dentina, que afeta tanto a dentição decidua quanto a permanente. A DI apresenta como características dentes escurridos e translúcidos que vão do cinza ao marrom ou âmbar. O esmalte pode se separar facilmente da dentina quando submetido ao estresse oclusal. Radiograficamente há evidencias de constrição cervical, raiz curta e polpas reduzidas, sendo os canais menores do que o comum ou completamente obliterados. A escolha do tratamento pode ser decidida com base no caso, considerando-se a idade da criança, grau de perda de tecido dentário e de cooperação do paciente.

Objetivo: O objetivo deste relato de caso foi descrever o tratamento odontológico precoce realizado em uma criança afetada pela DI tipo II. Relato do caso: O tratamento envolveu procedimentos básicos de prevenção. Molares deciduos foram desgastados, de tal forma que a estrutura remanescente do dente foi coberta com resina composta para proteger a dentina exposta. Selante resinoso foi aplicado em todos os primeiros molares permanentes. Mordida cruzada posterior foi tratada com a expansão do arco superior. Conclusão: O tratamento precoce restaurou a dimensão vertical do paciente resultando em estética e função aceitáveis para os dentes permanentes completarem sua erupção.

Descritores: Dentinogênese imperfeita; anormalidades dentárias; assistência odontológica.

Abstract
Introduction: Dentinogenesis imperfecta (DI) is a hereditary dentin development disorder that affects both primary and permanent dentitions. The DI characteristics are discolored and translucent teeth ranging from gray to brownish-blue or amber. The enamel may split readily from the dentin when subjected to occlusal stress. Radiographically there are evident of cervical constrictions, short root and pulp chambers, and the root canals are smaller than normal or completely obliterated. The dental treatment choice can be decided on a case-by-case basis, considering the degree of dental tissue loss, and child age and cooperation. Objective: The aim of this case report was to describe the early dental treatment performed in a child affected by DI type II. Case report: The treatment involved basic preventive procedures. Primary molars were worn to such an extent that the remaining tooth structure was covered with composite resin to protect the exposed dentin. Resin-based sealant was applied in all first permanent molars. Posterior cross bite was treated with the expansion of the upper arch. Conclusion: The early treatment restored the patient’s vertical dimension resulting in acceptable esthetics and function for the permanent teeth to complete their eruption.

Descriptors: Dentinogenesis imperfecta; tooth anomaly; dental care.
INTRODUCTION

Dentin is a major structural component of tooth and serves as the framework upon which enamel and cementum form. Dentin supports enamel via mechanical stress distribution through well-organized dentinoenamel junctions. Heritable dentin defects are rare diseases that affect primarily the organic dentin matrix. They have been classified into three types of dentinogenesis imperfecta (DI-I, DI-II and DI-III), and two types of dentin dysplasia (DD-I and DD-II) both of which present autosomal dominant transmission that affects the primary and permanent dentition.

According to Shields et al. (1973), the DI type I is associated with osteogenesis imperfecta. DI type II is found in patients with dentition abnormalities alone and no bone disease. DI type III is the Brandywine form, named for the city Brandywine, Maryland, where there was a large population of patients with this disorder. DI has a reported incidence range from 1:6000 to 1:8000 (for all types of DI) in the United States. Clinically, the color of affected teeth ranges from gray to brownish-blue with a translucent opalescence. Radiographically, the teeth show bulbous crowns, cervical constriction and short roots. In DI type II the pulp chambers and root canals are partial or completely obliterated, while in DI type III the teeth present viable pulp chambers leading to what is referred to as “shell teeth”. Due to the lack of support of the poorly mineralized underlying dentin, the enamel frequently fractures leading to rapid wear and attrition of the teeth.

In general, patients with DI require complex treatment that should take into account the degree of tooth destruction, age and cooperation of the patient. Early treatment prevents the abrasion of the erupted tooth (causing vertical dimension loss) and also offers the recovery of function and improves esthetic appearance, avoiding possible physiologic disorders.

This case report aimed to describe the approach for dental treatment performed in a child affected by the DI type II to avoid tooth and occlusion deterioration.

CASE REPORT

This study was approved by the Ethics Committee of São Paulo State University (Protocol number 21/07), and all volunteers gave their written consent to participate. A white male child, aged 6 years (proband, individual III:15) was diagnosed with dentinogenesis imperfecta type II. The patient’s main complaint was poor esthetic and difficulty with chewing. The family heredogram revealed affected father, paternal aunt and her three daughters, two of them, the individuals III.7 and III.8 (ages 24 and 22-year-old, respectively), revealed radiographically obliterated pulp chambers (Figure 1). No history of any type of bone abnormality was associated with any of the family members that presented the same disorder, which was confirmed by clinical and radiographic examination.

The proband showed mixed dentition, amber and opalescent coloring teeth (Figure 2), mainly in the lingual face of mandibular incisors (Figure 3). There was no enamel on the occlusal surface of the second primary molars that showed extensive attrition of the dentin and caries lesions leading to a marked reduction of the crown height. In addition, the child presented bilateral posterior and canine cross-bite.

Panoramic radiography images showed wide pulp chambers and root canals of recently erupted teeth (34, 35, 44 and 45) and not erupted teeth (37 and 47). A tendency to pulp chamber closure...
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The patient vertical dimension was restored to obtain acceptable function and esthetics when permanent teeth complete their eruption.

DISCUSSION

DI was first reported by Talbot\(^8\) (1893), however, Roberts, Schour\(^9\) (1939) coined the term Dentinogenesis Imperfecta in 1939. Many cases of DI and DD have been investigated in the last decades and the results have been compiled in the OMIM (Online Mendelian Inheritance in Man, which can be accessed at: http://www.ncbi.nlm.nih.gov/omim)\(^10\). To access each DI type, the proper MIM number should be used: DI-I (166240), DI-II (125490) and DI-III (125500).

The presented DI type II case showed a more intense discoloration in the mandibular incisors teeth compared to the maxillary incisors. This fact was not previously reported. It was observed that the reported proband showed cross bite with vertical dimension loss due to the attrition of the crowns, dentin exposure and fast abrasion due to the lower dentin mineral content. Caries were observed in the primary teeth of the reported child probably because both poor oral hygiene habits and prolonged use of feeding bottle, as mentioned by his mother.

There are no guidelines on restorative treatment in teeth affected by DI. The treatment should begin as early as possible, considering the degree of tooth destruction and patient cooperation. The objectives for early treatment, in primary and permanent dentition with DI, are the prevention or ending of dental abrasion and the recovery of masticatory function and esthetic. Two treatment modalities have been proposed to our patient to reach these objectives in primary dentition: first, composite resins were placed in primary molars to protect exposed dentin, although the adhesion seems to be less effective than on normal substrates due to poor hybrid layer created\(^11\); second, an orthodontic appliance to correct the posterior cross bite, to recover the functionality and vertical dimension loss. Sealant was applied in the first permanent molars because high-risk children present more chance of developing caries and sealant placement appears to be effective in preventing dental caries development\(^12\). The patient and parents were informed about the need of good dental hygiene and regular follow up dental visits to monitor dental arches growth and tooth eruption.

The two major noncollagenous dentin matrix proteins, dentin sialoprotein (DSP) and dentin phosphoprotein (also known as phosphophoryn) are encoded by a single gene termed dentin sialophosphoprotein (DSPP)\(^9,13\) (MIM 125485) which is located on 4q21.3\(^13\). Another protein has been identified from the proteolytic cleavage of DSPP. Dentin glycoprotein (DGP) has been proposed to play a role in dentin biomineralization\(^14\). Many studies have associated DI-II to mutations in the DSPP gene\(^15-18\). Shields et al.\(^2\) (1973) described DI type III (presented in a triracial population of Brandywine, Maryland\(^19\)), distinct from DI type II. Interestingly, with the advent of the molecular genetics, Shields’ previous classification of DI could possibly be changed. However, Witkop\(^19\) (1975) indicated that these two types

Figure 2. Frontal view of a 6-year old male proband diagnosed with Dentinogenesis Imperfecta (DI) type II, showing amber and opalescent teeth.

Figure 3. Lingual face of a 6-year old male proband diagnosed with Dentinogenesis Imperfecta (DI) type II. Mandibular incisors showed amber and opalescent dentin.

Figure 4. Radiograph of the proband (individual III:15) showing wide pulp chambers and root canals of recently erupted teeth (such as 37 and 47) and not erupted teeth (such as 34, 35, 44 and 45). Pulp chamber obliteration is observed in permanent mandibular incisors and first molars.
could be the same because of a surname, which was common to individuals with both disorders, as well as clinical similarities. Utilizing automatic sequencing, Kim et al.18 (2005) demonstrated that the probands from two unrelated kindred (one with DI-II and another with DI-III) had identical mutations in the sequence of the DSPP gene (nucleotide 1191 from Guanine to Timine in exon 3, changing aminoacid 18 from Valine to Phenylalanine). Therefore, the authors have proposed that “hereditary opalescent dentin” or “DI type II” should be used to describe both DI type II and type III phenotypes. The molecular genetics findings of the causative factor of DI in those families studied by Kim et al.18 (2005) seems to support Witkop’s19 (1975) idea that the DI types II and III were caused by a different expression of the same gene21.

To date, 13 mutations have been documented in DSP (10 of them was reviewed by Wang et al.20 (2009), and the other 3 by Lee et al.1 (2011)). Because 4 cases of DI were caused by the c.52G>T, p.V18F (cDNA nucleotide 52 mutated from G to T, leading to the altered protein: aminoacid 18 mutated from Valine to Phenylalanine) mutation in the first nucleotide of exon 3[1,21,22], it seems that this region of the gene is a mutational “hotspot”. Regarding DPP, 12 mutations have been found in the exon 5 of the DSPP gene, 4 of them were described by Nieminen et al.23 (2011) who investigated kindred of different ethnic origins, 4 were found by Song et al.21 (2008), 2 mutations were identified by Wang et al.20 (2009) and the 2 other mutations in the DPP were found by Lee et al.1 (2011). It is worth to mention that the molecular genetics study, in order to identify the genetic cause of DI-II in the present family, is still in progress.

Considering that DI-II is an uncommon dental anomaly, the authors believe that the case described illustrates the occurrence of this disease in different parts of the world, affecting people of different ethnics. Furthermore, as the dental treatment of individuals with DI is very difficult6,7,26, presenting the dental treatment of a child with this disease could be useful to other clinicians.

In conclusion, the authors provided adequate treatment eliminating caries, restoring function and using preventive measures to improve the oral hygiene of the proband and the follow-up to prevent or to treat potential alterations related to DI-II.

REFERENCES


CONFLICTS OF INTERESTS

Os autores declaram não haver conflitos de interesse.

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