

CASE REPORT

A rare case diagnosed as dentin dysplasia type II

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Abstract

According to their phenotypic features, the hereditary dentin defects in humans are categorized in two major classes: dentinogenesis imperfecta and dentin dysplasia. At its turn, the dentin dysplasia is subdivided in dentin dysplasia type I and dentin dysplasia type II, a milder clinical manifestation of the condition. Here we report the clinical and radiographic findings of dentin dysplasia type II in two members of a family, a young adult female and her mother. Except a mild shade change of the incisal margins in upper central incisors and left upper canine of the daughter no abnormal occlusal wear or crown shape change of the teeth were disclosed in both patients. However, confluent large pulp stones in the thistle-tube shaped pulp chambers and pulpal obliteration were a common finding. The condition was diagnosed as dentin dysplasia type II.

Keywords: dentin dysplasia type II, diagnosis, radiographic traits.

Introduction

Dentin dysplasia type II (DD-II) is a rare genetic disorder of dentin development with autosomal dominant trait that is characterized by inherited dentin defects and affects solely the teeth [1, 2].

According to Shields classification, DD-II, also known as coronal dentin dysplasia, belongs to a group of five hereditary dentin disorders (dentinogenesis imperfecta type I, II, and III and dentin dysplasia type I and II), which have in common the abnormal development of dentin [3].

Dentinogenesis imperfecta (DGI) has a prevalence of one in 8000 cases [4, 5]. In dentinogenesis imperfecta type I (DGI-I), the dentinal defects are simultaneously manifested with osteogenesis imperfecta, while type II dentinogenesis imperfecta (DGI-II) occurs as an isolated trait [6]. The dentinogenesis imperfecta type III (DGI-III) comprises mixed features of type I, type II and multiple pulp chamber exposures [3].

DD-II (coronal type of dentin dysplasia), traditionally considered to be rather rare, is less frequent than DD-I (radicular type of dentin dysplasia) or DGI-II, and is equally affecting both sexes. Although in different degree, both deciduous and permanent dentitions are affected clinically, histologically, and radiographically in DD-II [1, 7, 8].

Clinically, in DD-II, the deciduous teeth are amber and translucent. Histologically, the pulp chambers are obliterated by abnormal structured dentin, resembling dentinogenesis imperfecta. Though the permanent teeth are normal in coronal morphology and color, radiographically they have thistle-tube shaped pulp chambers and numerous pulp stones [1, 9].

In this paper, it is reported a case of DD-II found at the visit for usual root canal retreatment of two permanent teeth in a young adult female patient. Though the 23-year-old woman had previous endodontic treatments, the pathological condition was not diagnosed before. There are described the specific radiographic images of patient's permanent dentition and the particularities that influence the positive and differential diagnosis and the treatment outcome as well.

Case presentation

A 23-year-old woman with a noncontributory medical history was seen for a mild hypersensitivity at cold and sweets in upper left second premolar (tooth 25). The clinical inspection disclosed a deep distal caries. Previous defective root canal treatments were also observed in upper left first premolar (tooth 24) and lower left first molar (tooth 36). A routine retreatment of the root canals was started in upper left first premolar (Figure 1) and lower left first molar (Figures 2 and 3). Except the mentioned affected teeth and an almost unobservable shade change of upper central incisors and left canine (Figure 4), all permanent teeth were normal in size, shape, and color (Figure 5). Carious activity and enamel attrition were minimal. The oral soft tissues and alveolar bone had a normal status. Thorough medical investigation and current clinical laboratory tests did not disclose any associated systemic disease.

Excluding the first right mandibular molar that was extracted, the radiographic examination showed an almost full complement of permanent teeth (Figure 6). Much of the pulp chamber of all vital teeth was occupied by isolated

or confluent pulp stones (Figure 7) both in upper (Figure 8) and lower jaw (Figure 9). In some teeth, especially in molars, the pulp chamber was significantly obliterated. The anterior teeth and premolars had coronal pulp morphology resembling thistle-tube shape.

Cone-beam computed tomography (CBCT) scan revealed in successive slices the real three-dimensional (3-D) spreading out of coronal pulp calcification in upper right wisdom molar (Figure 10), upper left central and lateral incisors (Figure 11), upper left canine and first premolar (Figure 12), upper left second premolar and first molar (Figure 13), upper left wisdom molar (Figure 14), lower right wisdom molar (Figure 15), and lower left second molar (Figures 16 and 17) and lower left wisdom molar (Figure 18).



Figure 1 – Postoperative periapical radiograph of the upper left first premolar (tooth 24) after the completion of root canal retreatment. Note the pulp chamber advanced obliteration in upper left canine (tooth 23) and second premolar (tooth 25), as well as the almost complete pulp chamber obliteration in upper left molars (teeth 26–27). A deep distal caries is also observed in tooth 25.



Figure 2 – Preoperative periapical radiograph of tooth 36 showing a canal filling of distal root short to the apex and a periapical radiolucency in the mesial root of the same tooth. Confluent large pulp stones in the thistle-tube shaped pulp chambers of both premolars (teeth 34–35). Almost completely obliterated pulp chamber by pulp stones in the second lower molar (tooth 37).



Figure 3 – Postoperative periapical radiograph of tooth 37 after the completion of root canal retreatment. Note the pulp chamber obliteration in both still vital lower molars (teeth 37–38).



Figure 4 – Frontal view of the occlusion showing a mild shade change of the incisal margins in upper central incisors and left upper canine.



Figure 5 – Frontal view of upper and lower arch of the patient – no occlusal wear or crown shape change of the teeth.

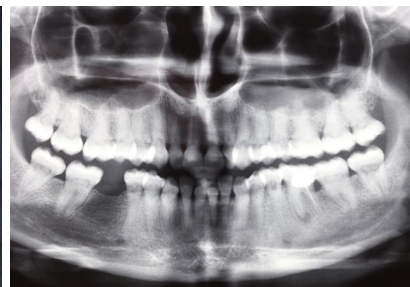


Figure 6 – Orthopantomogram of a 23-year-old female at the first visit.

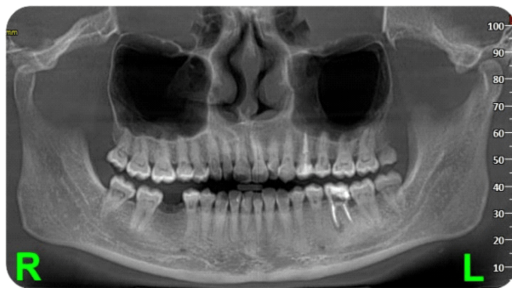


Figure 7 – Orthopantomogram of the same patient after the completion of endodontic treatment. R: Right; L: Left.

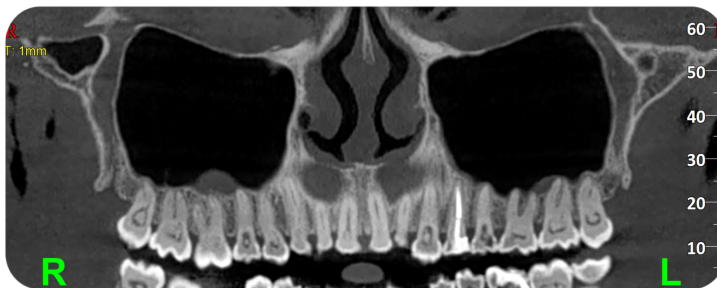


Figure 8 – Orthopantomogram: detailed view of upper teeth after the root canal retreatment in left first premolar (tooth 24). R: Right; L: Left.

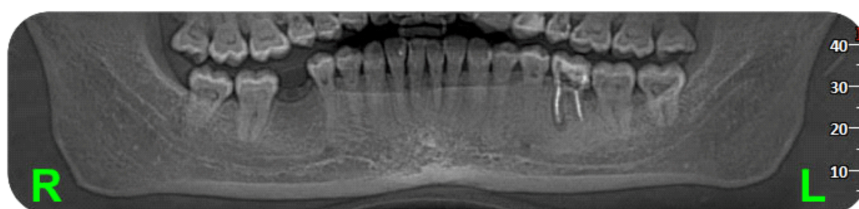


Figure 9 – Orthopantomogram: detailed view of lower teeth after the root canal retreatment in left first molar (tooth 36). R: Right; L: Left.

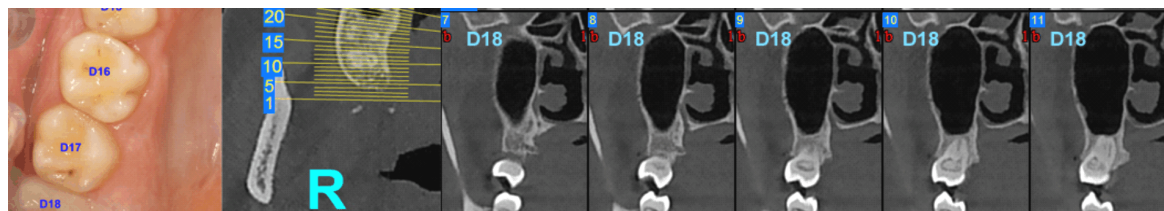


Figure 10 – CBCT scan of upper right wisdom molar (tooth 18). Note the presence and size of the pulp stones in all the successive slices. CBCT: Cone-beam computed tomography; R: Right.

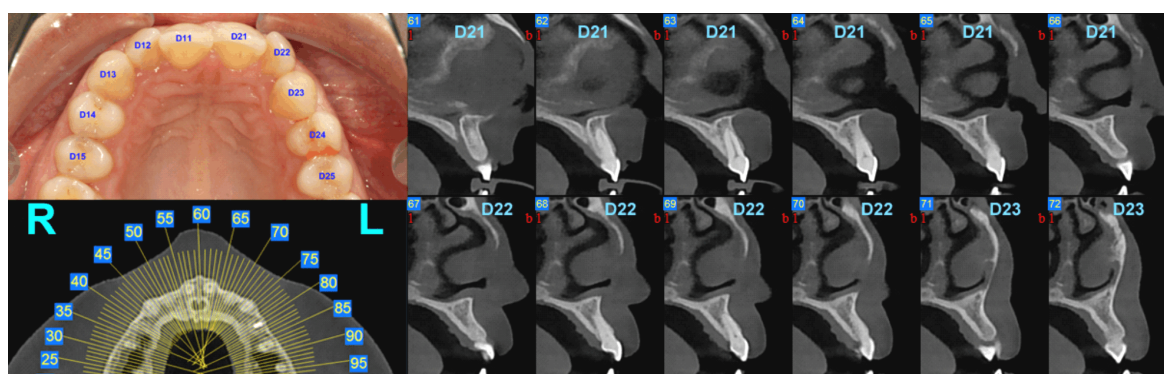


Figure 11 – CBCT scan of upper left central (tooth 21) and lateral incisors (tooth 22) showing in successive slices the extension of pulp chamber obliteration. CBCT: Cone-beam computed tomography; R: Right; L: Left.

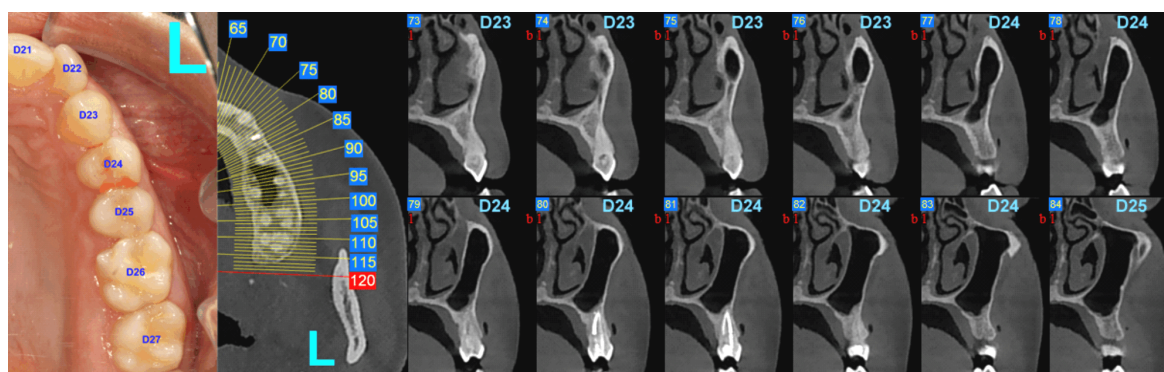


Figure 12 – CBCT scan of upper left canine (tooth 23) and first premolar (tooth 24) showing in successive slices the extension of pulp chamber obliteration. CBCT: Cone-beam computed tomography; L: Left.

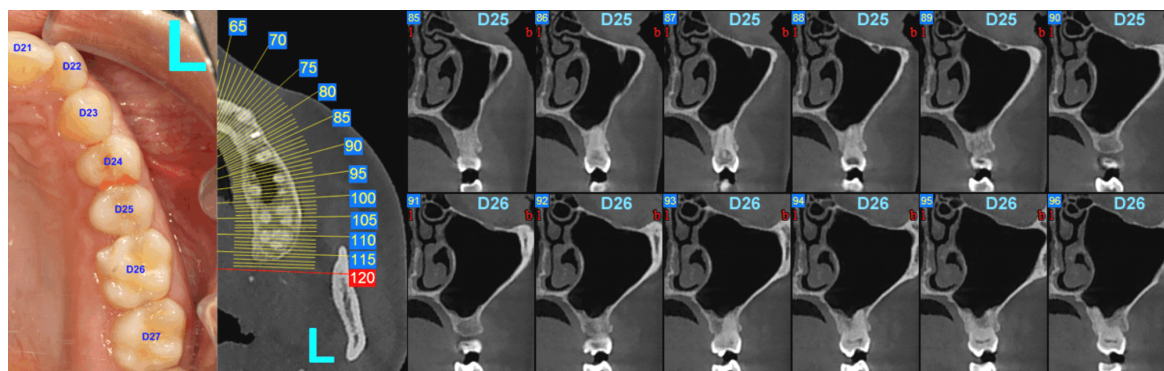


Figure 13 – CBCT scan of upper left second premolar (tooth 25) and first molar (tooth 26) showing in successive slices the extension of pulp chamber obliteration. CBCT: Cone-beam computed tomography; L: Left.

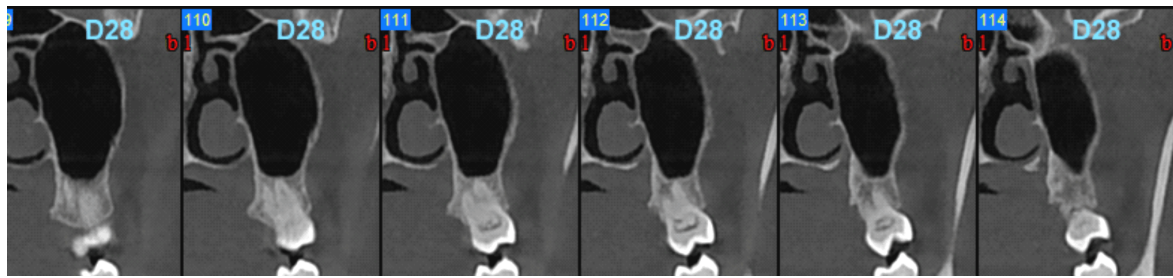


Figure 14 – CBCT scan of upper left wisdom molar (tooth 28) showing in successive slices the extension of pulp chamber obliteration. CBCT: Cone-beam computed tomography.

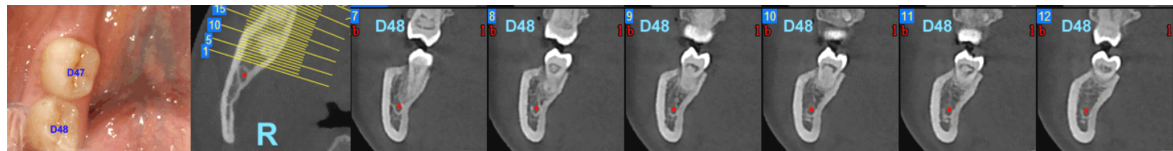


Figure 15 – CBCT scan of lower right wisdom molar (tooth 48) showing in successive slices the extension of pulp chamber obliteration. CBCT: Cone-beam computed tomography.

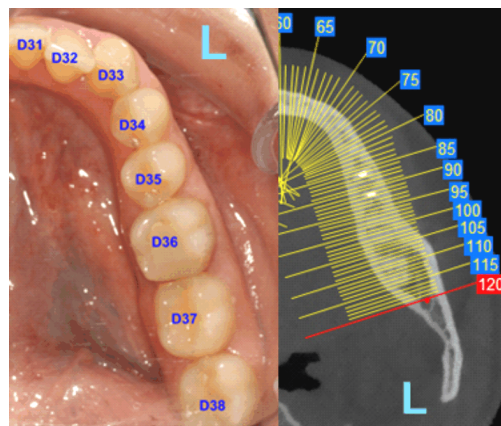


Figure 16 – Direction of CBCT slices for detection of pulp stones in lower left second (tooth 37) and wisdom (tooth 38) molars. CBCT: Cone-beam computed tomography; L: Left.

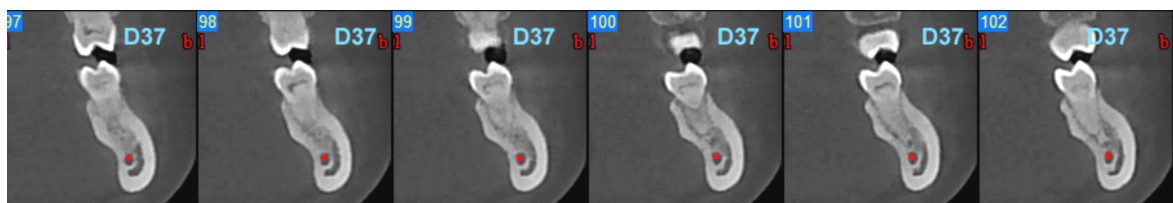


Figure 17 – CBCT scan of lower left second molar (tooth 37) showing in successive slices the extension of pulp chamber obliteration. CBCT: Cone-beam computed tomography.



Figure 18 – CBCT scan of lower left wisdom molar (tooth 38) showing in successive slices the extension of pulp chamber obliteration. CBCT: Cone-beam computed tomography.

The radiography of her 45-year-old mother (Figure 19), who was the solely member of her family available for examination, showed multiple affected teeth by confluent pulp stones and pulp chamber obliteration.



Figure 19 – Orthopantomogram of the patient's mother (45-year-old) showing except the previous endodontic treatments multiple affected teeth by confluent pulp stones and pulp chamber obliteration.

Discussions

The permanent teeth in DD-II are normal in shape and size. The root growth usually is also habitual and the final root size is normally preserved in deciduous and permanent teeth as well [10].

Unlike the DGI where the crown color is amber-brown in both deciduous and permanent dentitions in DD-II only permanent teeth are normal colored, whereas the deciduous teeth may be bluish-grey opalescent, yellow, grey-amber or brown, resembling DD-I, pulpal dysplasia, and hereditary opalescent dentin [7, 10–12].

Accordingly, it was not surprising that the young adult patient who asked for endodontic retreatment has not been previously informed she has a dentin defect. Actually, the first diagnosis of the condition was established in our office.

Commonly, in DD-II, the teeth are free of decays [13]. Therefore, the dental status of the young female respected the general rule as she has only one extracted tooth and two other formerly endodontically treated teeth.

Though radiographically the periapical bone usually is normal, the teeth might give various responses to pulp testing and seldom toothaches were reported in non-carious teeth [13]. Sometimes, there were also encountered apical radiolucencies. It has to be underlined that if the thermal test may be inconclusive, the electric ones in these circumstances are definitely negative [13].

Histologically, in DD-II, the coronal dentin of deciduous teeth appears normal whereas in the roots a superficial layer of normal dentin covers without transition an extremely dense amorphous and atubular dentin. The permanent teeth have a relative normal structure of coronal dentin. The root dentin is more frequent amorphous, atubular and has globular appearance. Unlike the unaffected teeth in DD-II, amorphous calcified conglomerates in the form of numerous true pulp stones are frequently found in the pulp tissue. Progressively with age, the pulp chamber in permanent teeth become obliterated [7, 10, 13, 14].

Radiographically, deciduous teeth in DD-II are similar to DD-I and dentinogenesis imperfecta, with extremely short roots and pulp chamber obliteration occurring post-eruptive and showing only some chevron-shaped horizontal radiolucencies that represent still uncalcified pulp tissue. Unlike the deciduous teeth, in permanent ones the radiographic image is modified resembling a flame-shape. Due to the abnormal enlargement and extension of pulp chamber toward the root often the image is mimicking a thistle-tube shape. Radiopaque areas resembling pulp stones are also present. However, sometimes the coronal pulp can be free of calcifications [7, 10–12].

The orthopantomogram of reported patient revealed in all teeth confluent large pulp stones in the thistle-tube shaped pulp chambers and pulpal obliteration. Additionally, the CBCT images allowed the 3-D visualization of the real volumetric trait that was generated during the process of pulp chamber obliteration.

DD-II is caused by mutations of the dentin sialophosphoprotein (*DSPP*) gene located on the long arm (q) of chromosome 4 (4q21.3) that codes two major non-collagenous proteins of dentin matrix, dentin sialoprotein and dentin phosphoproteins. By linkage analysis, along with DD-II, DGI-II and DGI-III have also been mapped to chromosome 4q21 [6, 15, 16].

DSPP is the most recent member of the acidic secretory calcium-binding phosphoprotein (*SCPP*) gene family sharing a common 5' region, which appeared during biological evolutions of species in reptiles and mammals descending from the ancestral *SPARC*-like 1 gene [17].

Since dentin sialophosphoprotein is required for normal dentin formation, *DSPP* gene expression is mandatory tooth-specific. The largest part of *DSPP* mutations generating the aforementioned non-syndromic dentin defects, such as DD-II, DDI-II and DDI-III, occur within the repetitive coding sequences of dentin phosphoprotein (DPP) [17, 18].

DPP is highly phosphorylated and varies among species individuals. In humans, the size of DPP domain belonging to *DSPP* is between 770 and 902 amino acids. To better understanding of DPP role in biomineralization and inherited dentin defects, the sequencing through the DPP repetitive region is highly required [17].

However, a thorough comprehensive genetic analysis of dentin sialoprotein and dentin phosphoprotein in Chinese families recently revealed an associated phenotypic continuum of different *DSPP* gene variants, due to their expanded spectrum [19].

Based on the phenotype similarity of deciduous teeth in DD-II and DGI-II an attractive hypothesis suggests that the causal gene of DD-II might be an allele gene of the dentinogenesis imperfecta, which was already located on chromosome 4q13-q21 [10].

In non-syndromic subdivisions of dentinogenesis imperfecta (DGI-II, DGI-III) and milder dentin dysplasia (DD-II), there were found mutations in one allele of the *DSPP* gene. However, the loss of a single *DSPP* allele causes no dental phenotype [20].

Accordingly, it was forwarded the assertion that DD-II and DGI-II might be various phenotypic expressions of the same developmental dental disorder rather than different entities [6, 21].

Unlike DGI-I, a syndromic dental symptom of osteogenesis imperfecta, which is related to mutations of *COL1A1/COL1A2* genes DGI-II, DGI-III and DD-II, that also affect dentin, are non-syndromic autosomal dominant genetic diseases linked to chromosome 4q and small integrin-binding ligand *N*-linked glycoprotein (SIBLING) family [20].

A clinical and genomic investigation of the most important dentin defects in seven Chinese families found DD-II in only one family as compared with DGI-II in five families and DGI-III in one family. The laboratory analysis revealed variants of *DSPP* in six out seven families. One variant (c.52G>T) was present in two out of six families, whereas each of the remaining four families were characterized by different variants, as follows: c.2684delG and three novel variants, such as c.52-2A>G, c.1874-1877delACAG, and c.3509-3521del13bp [19].

There were described four out of 19 mutations in the *DSPP* gene that provoke DD-II, one of them is a missense mutation located in the signal peptide and the other represent frameshift mutations of DPP coding region. The largest part of these mutations consists in a single nucleotide substitution in the 5' end of *DSPP* near intron/exon junctions [8].

Two family studies in patients affected by DD-II showed in one family an Arg68Trp missense mutation in dentin sialoprotein part of the gene, whereas in the other family was found an Ala15Val missense mutation in the last residue of signal peptide [22].

Regarding the mutations that shift the reading frame by deletions and insertions, they are located in the 3' end of *DSPP* but do not generate a dominant dental phenotype. Since the investigation of genotype–phenotype correlation was not finalized, the dentin expression of *DSPP* mutations is still considered currently a matter of debate [8]. However, it is thought that there is a correlation between the genotype missense mutation of *DSPP* and the phenotype severity [22, 23].

Excepting DD-II, these mutations in the *DSPP* gene are also involved through dominant negative effects in generating other dentin abnormalities, such as DGI-II and DGI-III. The intimate mechanism seems to rely upon the exon skipping rather than intron retention [8]. It seems that a complete loss of a single copy of *DSPP* gene might cause recessive forms of DD or DGI. However, is unlikely to generate a phenotypic expression of dentin disorders. Though the mutation rate seems to be very low, its real value is still not known [7, 8].

Due to the increased number of *DSPP* mutations that were found in non-syndromic DGI-II, DGI-III and DD-II, it is thought that dentin dysplasia occur when mutant odontoblasts produce an amount of *DSPP* less than the 50% secretion level of normal ones, but definitely higher compared to dentinogenesis imperfecta [20].

There were found two classes of mutations that disrupt the odontoblasts ability to secrete and mineralize the dentin matrix. The first class is based on missense mutations within the first 18 amino acids of *DSPP* gene, whereas the second one changes the gene character from acidic to hydrophobic [22].

It is established that the mutations in *DSPP* result in retention of mutant proteins within the odontoblasts in

all non-syndromic DGI-II, DGI-III and DD-II. However, since the dominant negative effects of such proteins, related to their carboxy-terminal acidic repeat domain, proved to be lower in dentin dysplasia it is thought that in this non-syndromic dentin condition the odontoblast secrete fewer mutant proteins than in dentinogenesis imperfecta [20].

A better understanding of hereditary etiology in DD-II is of paramount clinical significance since allows an early diagnosis of the condition, before the obliteration of pulp chamber and root canals [8].

The diagnosis of DD-II relies on detailed patient history, careful clinical evaluation and radiographic image revealing the thistle-shape of pulp chamber, numerous pulp stones and even pulp chamber obliteration.

In deciduous dentition, the differential diagnosis of DD-II with DGI is more difficult because due to the amber color and obliterated pulps, both clinical and radiographic findings are consistent conventional feature of DGI.

Investigating the family history of previous case reports considered this dentin disorder conditioned by patient's genetic background. Particularly, in DD-I, sometimes was also associated the formation of apical cysts, sinus tracts and recurrent abscesses without an obvious etiology [13]. Actually, it has to be highlighted that in such circumstances a differential diagnosis with DGI is mandatory [6].

Since in our report, excepting her mother, other family members were unavailable to be evaluated, the family history is incomplete. However, the mother's permanent teeth showed also thistle-tube deformities of the pulp chambers and numerous large pulp stones.

The literature also mentions rare clinical cases manifested by tooth mobility or apical radiolucencies and abscesses [7]. Due to the degree of root canal obliterations, the routine orthograde endodontic treatment is frequently a challenge and the solely conservative alternative can be the root apex resection followed by retrograde filling [13, 24]. Unfortunately, in common practice, habitually no other treatment procedures were suggested but extraction [7].

As a rule, taking into account that usually in DD-II the permanent teeth are not affected by dental decay, regular tooth brushing and periodic control are sufficient, excepting seldom the need of treatment for the specific aforementioned symptoms. However, an early diagnosis resulting in genetic counseling can be of utmost benefit for patients and their relatives.

✚ Conclusions

Dentin dysplasia type II is a rare genetic disorder of dentin development. In permanent teeth commonly the diagnosis is a challenge without radiographic examination since clinically the coronal shape, size and color are normal. The condition is recognized due to the thistle tube shape of pulp chambers and confluent large pulp stones that finally result in complete pulpal obliteration. An early diagnosis is highly beneficial when an orthograde root canal treatment is indicated as in case of advanced pulp calcification the endodontic access is definitely compromised. The genetic counseling is also recommended.

Conflict of interests

The authors declare that they have no conflict of interests.

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