CASE REPORT

Dentin dysplasia: diagnostic challenges

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SUMMARY

Dentin dysplasia (DD) is a rare autosomal dominant disorder associated with disturbance of the dentin. While the crowns appear clinically normal, on radiography, the pulp spaces appear partially or completely obliterated, with short blunted roots, and multiple periapical radiolucencies affecting the apparently sound teeth. Clinical signs include spontaneous abscess formation or increased tooth mobility which can lead to exfoliation. DD can therefore have a significant impact on the patient's dentition, and treatment is often challenging. Shields' classification of dentin disorders has been recently criticised for failing to consider differential variations and expressions of these disorders. This paper describes a case of a 23-year-old woman with previously undiagnosed DD, who presented with clinical and histological features belonging to several of these diseases, thus highlighting the potential diagnostic challenges faced with Shields' classification.

BACKGROUND

Dentin is a mesenchymal cellular mineralised tissue comprising approximately 70% inorganic material, 20% organic material and 10% water. The organic content consists of 90% type I collagen and 10% non-collagenous proteins and lipids. Dentin acts as a protective layer for the pulp and supports the overlying enamel and cementum.¹

Genetic disorders of the dentin have been widely reported and are broadly categorised into two main categories; dentinogenesis imperfecta (DI) and dentin dysplasia (DD). DD is a rare genetic disorder that is associated with disturbance of dentin formation. It was first described by Ballschmiede in 1930² who referred to it as 'rootless teeth'. Rushton later termed this clinical presentation as DD in 1939.³

A clinical classification of genetic disorders of dentin was proposed by Shields in 1973 and is commonly used worldwide.⁴ According to Shields' classification, DI is divided into three subgroups (types I–III); DD is classified into two types, where type I affects radicular dentin and type II affects coronal dentin. Type I is estimated to affect 1 patient in every 100 000 while type II has an unknown prevalence and it is thought to be lower than type I.¹ Shields' classification of dentin disorders has been recently criticised as it was based solely on clinical phenotypes and lacked any molecular genetic information concerning the inherited disorders of dentin. The clinical and radiographic features of DD can vary, with some case reports describing clinical signs that belong to both types I and II, thus making it challenging for clinicians

to make an accurate diagnosis.⁵ Genetically, inheritance is generally autosomal dominant with complete penetrance,⁶ although some case reports report no family history.

DD typically affects both primary and permanent dentitions. Coronally, the dentition appears normal, although some authors have described the presence of brown/yellow opacities.⁷ Clinical signs and symptoms may relate to spontaneous abscess formation or tooth mobility. Due to short roots, premature exfoliation of the teeth may occur either spontaneously or following minor trauma. Radiographically, root morphology varies, from roots appearing short and blunted with a marked apical constriction to complete agenesis (table 1).

In type I DD, pulpal tissue is often replaced by mineralised tissue, with partial or complete obliteration of the pulp chamber leading to a crescent-shaped pulpal remnant parallel to the cemento-enamel junction.⁶ In type II, the pulp chambers may be enlarged: often referred to as 'thistle tube' appearance. In both types of DD, pulp stones may be present.⁸

Periapical lesions or abscesses often develop spontaneously, occurring in the absence of caries or other pathology.⁹ The proposed aetiology of this relates to the strangulation of the pulp by narrowing of the apical constriction due to abnormal dentin formation, leading to pulpal necrosis. Dental eruption tends to be normal, although a few cases of delayed eruption have been reported in the literature, and teeth may be malaligned.¹⁰

Histologically, the enamel and dentin adjacent to the amelodentinal junction appear normal. Apical to the thin layer of normal coronal dentin are large, calcified, dentin masses which nearly obliterate the pulp chamber and canals. The dysplastic dentin masses have atypical dentin tubules, which are blocked by numerous calcified bodies containing tubular dentin referred to as denticles. Deeper layers of dentin show atypical tubular dentin with an amorphous structure. In type I DD, the affected dentin has a 'waterfall' or 'stream flowing around boulders' appearance, occurring due to a cycle of odontoblast death, recruitment of new odontoblasts and further dentin production, then death of these odontoblasts.

Genetic basis

Dentin sialophosphoprotein (*DSPP*) at chromosome 4q12-21 has been identified to control and contribute to dentin mineralisation. Mutations in the *DSPP* gene have been associated with three isolated dentinal disorders mentioned in Shields' classification: types II and III DI and type II DD.^{5 11}

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Table 1 Differences between Shields' classification of dentin dysplasia (DD)

Classification	Dentin dysplasia type I	Dentin dysplasia type II
Incidence	1 in 100 000.	Unknown—thought to be lower than DD type I.
Clinical presentation	Normal but can vary in appearance with brown/yellow opacities present.	▶ Normal crown colour, shape and height.
Radiographic presentation	▶ Partial or complete obliteration of the pulp. ▶ Roots are shorter (absent in severe cases), merged with an apical conical aspect, with sharp construction. ▶ Periapical radiolucencies are frequent without any other associated pathology.	▶ Pulp chambers described as 'thistle tube'; enlarged pulp chambers with thin root canals. ▶ Pulp stones are common.
Causative genes	Genetically heterogeneous including <i>SSUH2</i> , <i>VPS4B</i> , <i>SMOC2</i> .	<i>DSPP</i> .

This association implies that the three diseases are a severity variation of the same pathology. A recent review paper proposed a newer classification of dentin disorders that separates syndromic and isolated forms considering the variability of expression because of gene variants, with the main objective of simplifying the diagnosis.¹¹ The new classification groups the *DSPP* disorders under a single entity of 'Dentinogenesis imperfecta (DI)', characterised by grey to brown clinical crowns, shortened and bulbous crowns, shorter and thinner roots, with pulpal alterations. The severity can range from mild to severe, with Shield type II DD being considered a mild form, characterised by no or very few signs in permanent teeth except for the 'thistle-like' appearance of the pulp (table 2). The molecular bases of DD type I remains incompletely understood. Recessive mutations in *SMOC2* and dominant mutations in *VPS4B* and *SSUH2* have been reported in single families.^{12–14} Further research is required to determine the exact role of these genes in dentin formation.

This article describes a previously undiagnosed case of DD type I/radicular DD, illustrating the typical clinical, radiographic and histological features of this rare disorder.

CASE PRESENTATION

A 23-year-old woman was referred to the restorative department at the Leeds Dental Institute by her general dental practitioner for an opinion with regards to multiple recurrent and spontaneous abscesses. The patient had previously undergone extensive haematological investigations, and interventions which included non-surgical and surgical endodontics to address the multiple recurrent spontaneous abscesses of unknown origin.

On presentation, the patient's chief complaint was discomfort associated with the slightly mobile upper left first molar. She also complained of functional difficulties associated with an anterior open bite with few contacting occlusal units.

Medically, the patient had mild asthma, which was well controlled. There were no known allergies. Family history revealed no known abnormalities of this nature affecting other family members. On further enquiry, there were no cosegregating health problems.

Intraoral examination revealed a moderately restored dentition with the presence of minimal soft plaque deposits posteriorly. The teeth appeared to have bulbous crowns with no

opacities present (figure 1). There was a class III incisal relationship with an incomplete overbite. Contacts in the intercusp position were only present on the LR7 and UR6, UL7 and LL7, UR3 and LR3. The UL5, LL5 and LR5 were absent, while the ULE was retained and ankylosed. The UL6 had a vertical root fracture and a draining sinus buccally.

An orthopantomogram (OPT) was taken which revealed the presence of bulbous crowns and short, malformed roots, with multiple periapical radiolucencies associated with the following teeth: LR6, LR7, UR6, UL6, UL8 and LL4, LL6, LL7. The following teeth were endodontically treated: UR6, UR5, UR4, UR1, UL1, UL4, UL6, LL6, LL7, LR6, LR7 (figure 2).

At a subsequent review, the patient presented with a new buccal sinus between the UL7 and UL8. The UL8 was tender to percussion. A small volume cone beam computed tomography (CBCT) of the suspected area revealed periapical pathology affecting the palatal root of the UL8 (figure 3).

INVESTIGATIONS

An OPT, small volume CBCT of the UL8 area and histopathology analysis of the UL8 tooth were undertaken.

DIFFERENTIAL DIAGNOSIS

Based on the initial clinical and radiographical findings, a diagnosis of DD type I/radicular DD was made.

TREATMENT

Initial management involved reinforcement of oral hygiene and toothbrushing instruction, followed by supra/subgingival periodontal scaling. Upper and lower impressions were taken, and study models were articulated on an average value articulator to aid further treatment planning.

The UL6 and UL8 were subsequently extracted under local anaesthesia. The UL8 tooth displayed a bulbous unrestored crown, with marked cervical constriction. The roots appeared thin and partially resorbed.

Figure 1 Extra/intraoral views of the patient's dentition.**Table 2** Proposed new classification of isolated dentin disorders

Shields' classification of dentin disorders ⁴	Proposed classification of isolated dentin disorders ¹¹
Dentin dysplasia type I	Radicular dentin dysplasia
Dentin dysplasia type II	Dentinogenesis imperfecta—mild
Dentinogenesis imperfecta type II	Dentinogenesis imperfecta—moderate
Dentinogenesis imperfecta type III	Dentinogenesis imperfecta—severe

Figure 2 Orthopantomogram revealing the presence of bulbous crowns with multiple periapical radiolucencies associated with a number of teeth.

Selective occlusal adjustment, following trial adjustment on articulated study models, was carried out to increase the number of occlusal contacts and improve function.

Histopathological analysis was carried out on the extracted UL8 which revealed an abnormal dentin structure, displaying irregular and reduced dentinal tubules coronally (figure 4). This appearance was significantly different in comparison to the dentin structure of a normal tooth (figure 5). The pulp cavity was obliterated in part and appeared necrotic focally, with the presence of pulp stones (figure 6). Radicular dentin appeared atubular and amorphous. The characteristic 'stream flowing around boulders' of type I DD was not present, possibly due to a mild phenotype expression. Genetic counselling was offered to the patient but was declined at this moment in time.

OUTCOME AND FOLLOW-UP

The patient reported a significant improvement in comfort and chewing efficiency following extraction of the UL6 and UL8 and selective occlusal adjustment.

DISCUSSION

DD is a rare genetic condition that affects dentin formation. This case exhibited most features commonly found in patients with type I DD, which include clinically normal crowns, presence of short root morphology, abnormal radicular dentin structure,

Figure 3 An axial view showing the presence of a periapical radiolucency associated with palatal root of the upper left third molar tooth.

Figure 4 Histological images of the extracted upper left third molar displaying an abnormal and irregular dentin structure with a significant reduction in dentin tubules.

multiple spontaneous periapical radiolucencies and partial pulpal obliteration. This case also exhibited a class III malocclusion with an incomplete overbite present. Malocclusion is not considered a specific feature of DD, but has been reported.¹⁵ The lack of any cosegregating health problems was a strong indication that this was an isolated form of the disease.

The exact pathophysiology of DD is unknown. It has been hypothesised that an alteration in the epithelial part of tooth germ during root formation causes a premature invagination of Hertwig's epithelial root sheath and apical constriction.¹⁶ This, however, has not been proven. Alternatively, it has also been proposed that the DD is caused by atypical interactions of odontoblasts with ameloblasts leading to an abnormal function of the ectomesenchymal-derived odontoblasts.¹⁷

While the diagnosis of DD is largely based on clinical and histopathological findings, it can potentially be challenging to clinicians in primary care due to the low incidence rate of this disease. Indeed, lack of previous clinical experience could contribute to the delay in reaching a correct diagnosis, as highlighted by this case. This can result in patients being incorrectly referred and investigated, which embarks them on a 'diagnostic odyssey', before a correct diagnosis is reached. This can take several years or decades before the correct diagnosis is established, which can lead to considerable uncertainty and stress for the patient and their families. This can also be expensive for the healthcare system and prevents timely access to treatment. The 2016 annual report of the chief medical officer in the UK recommended the establishment of referral pathways for rare disorders to secondary care, with a strong emphasis in using genomics in screening for diseases.

Genomics can be expected to have an increasing role within the National Health Service in differentiating syndromic variants of dentin disorders. Diagnosed patients who are planning on starting a family should also be offered genetic counselling since this hereditary condition has a 50% chance that a child born will be affected. This was made available to the patient but was declined at this time.

Figure 5 Histological images of a control tooth displaying normal dentin structure with patent dentin tubules.

Figure 6 The pulp cavity appears partially obliterated and necrotic focally, with the presence of pulp stones.

Management of DD is complex. Careful assessment of psychological effect on diagnosed patients is vital. The prospect of premature loss of teeth can often be distressing and difficult to cope with. An empathetic approach is warranted, with further support required in younger patients. A referral to a psychologist may be useful prior to embarking on complex restorative treatment. The main goal is to maintain the dentition for as long as possible and to eliminate any source of infection or pain. In cases with DD, obliteration of the pulp chamber may make conventional endodontic treatment challenging. Nonetheless, this should be still the first option in treating symptomatic teeth with a patent root canal system.^{18 19} If orthograde endodontic treatment is not possible due to lack of patency, then a retrograde approach may be warranted, although this might not be feasible in teeth with short roots due to likely increased mobility and subsequent tooth loss. Symptomatic teeth with complete root agenesis should be extracted.

The general aims of restorative treatment include:

- Prevention: regular monitoring and reinforcement of oral hygiene instruction to prevent primary dental disease, with the main focus to retain the dentition for as long as possible.
- Intervention: removal of potential sources of infection to alleviate pain and discomfort.

Patient's perspective

'Ever since being a young child, I have had repeated problems with my teeth, requiring regular visits to the dentists, multiple courses of antibiotics and a lot of pain killers, as this seemed to be the only solution. For years I have been referred for numerous investigations and treatments, which have been unable to identify the underlying cause. It was very frustrating not knowing what was causing the problems I was having, and it felt like there was nothing else that could be done. After so many years of being sent from pillar to post, it is a great relief to finally have a diagnosis, which will hopefully help with the future management of the condition.'

Learning points

- Clinicians are faced with potential diagnostic challenges with the current classification of dentin disorders.
- Adult patients with suspected dentin disorders should be referred to a specialist in restorative dentistry in order to provide appropriate care and management.
- Clinical genomics play a vital role in screening for rare diseases and future family planning.
- The establishment of referral pathways for rare disorders to secondary care can optimise the delivery of patient care.

- Restoration: restore function and appearance when teeth are lost.

Should teeth be lost, the options for replacing missing teeth include a removable prosthesis, resin-bonded bridges or dental implants. Dental implants should only be considered in patients who have ceased dentoalveolar growth.

This case highlights the inherent difficulties associated with Shields' classification of dentin diseases as clinical signs vary in presentation depending on severity and can belong to several of the diseases within the classification, thus making it very challenging for general practitioners to correctly diagnose patients with inherited dentin disorders. With ongoing advances in clinical genetics, various authors have suggested that Shields' classification has now become obsolete as it fails to separate syndromic and isolated forms, or to account for variability of expression due to gene variants.^{5 20 21} Nonetheless, restorative management of such patients should be conservative, with strong emphasis on prevention and retention of their natural dentition for as long as possible.

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