Clinical Findings and Management of a Rare Case of Multivariate Type of Dentinal Dysplasia: A Case Report

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Abstract

Dentinal dysplasia is a rare autosomal dominant condition affecting both deciduous and permanent dentitions. The presentation of dentinal dysplasia can be varied and intriguing. It is subdivided into two types and shares similar characteristics with dentinogenesis imperfecta and its variations. Marked radiographic similarities have been reported between dentinal dysplasia and dentinogenesis imperfecta. We present a case of a 26-year-old man who presented at our clinic with clinical features identical to those of aggressive periodontitis, accompanied by clinical and radiographic features commonly associated with both type 1 and 2 dentinal dysplasia. As the condition had not been reported previously in the literature, we believe the term “multivariate generalized dentinal dysplasia” would describe this unique condition accurately. The treatment plan was complex, aided by biomarker evaluation to time the treatment chart, which included extraction of selected teeth, periodontal surgery involving bone grafts, splinting, local delivery of medications, laser-assisted debridement, and regular follow-up appointments. Hereditary dentinal abnormalities are rare and are observed in people of different ethnicities with varied presentations.

The treatment of these conditions is challenging and demanding, as most patients are in an active disease state. Biomarker assessment during treatment planning plays a critical role in long-term clinical outcomes.

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Introduction

Dentinal dysplasia (DD) is a rare developmental condition with an autosomal dominant trait and a prevalence rate of 1:100,000. It characteristically affects dentin formation.[1,2] The condition was first reported by Ballschmiede and Berlin in 1970 [3], but the term was coined by Rushton in 1939.[4] DD was subdivided into type 1 radicular DD and type 2 coronal DD by Wiktop Jr.[5] In type 1 DD, both deciduous and permanent dentitions are affected, and it shows severe generalized mobility and premature exfoliation. In type 2 DD, the teeth exhibit bluish discoloration, characteristically seen in the deciduous dentition, although both permanent and deciduous dentitions can be affected. Permanent teeth show characteristic “thistle-tube” pulp topography and reveal pulp stones or pulp obliteration on radiographs in some cases. Type 1 DD shares common characteristics with other hereditary dentin disorders such as dentinogenesis imperfecta type 1, 11, 111, and DD-11, according to the Shield’s classification.[6] The conditions are varied in expression, which further lead to the proposal of a new classification system by De La Dure-Molla et al.[7] Genetic testing revealed dentin sialophosphoprotein (DSPP), which is extensively expressed in the dentin. Single mRNA for DSPP encodes proteins by end proteases in relation to matrix metalloproteinases (MMP). Furthermore, despite being non-collagenous, DSPP-related peptides play an

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important role in the conversion of pre-dentin to mineralized dentin. The other proteins that play equally significant roles are dentin sialoproteins (DSP) and dentin phosphoproteins (DPP) which are encoded in DSPP.[7] Mobility is usually the chief complaint of a patient with DD, and teeth are often lost prematurely in both deciduous and permanent dentitions.[8,9] The condition shows radiographically, with varied presentations such as the presence of short or tapered roots, taurodont teeth, and crescent-shaped radiolucent lines in the apical area and pulp chamber on radiographs, as well as the absence of pulp. Generalized periapical lesions are associated with non-carious teeth. These findings are characteristic and were proposed by O'Carroll et al.[10] Although morphological similarities were reported, there were differences with respect to discoloration, with some authors reporting bluish and some reporting greyish discolorations.[8,11,12] Ciola et al. further proposed the type 3 DD, as the condition reported by Eastman was localized and not generalized.[13] A new entity of DD termed symmetric multi-quadrant isolated dentin dysplasia, proposed by Qari et al., where the causes could be environmental and non-hereditary factors, also revealed a diverse presentation, not previously reported in literature.[14] Microscopic evaluation of the extracted teeth with type 2 DD revealed normal enamel and dentin, with the dentin exhibiting a thin line adjacent to the dentino-enamel junction and dysplastic dentin masses centrally. This was associated with pulp obliteration, and in some specimens, calcifications.[15] The aim of this article is to report a rare presentation with features of both type 1 and 2 DD and to provide an overview of its diagnosis and management.

Case Report

A 26-year-old man with non-contributory medical and dental history had come to the university dental clinic in the month of January,2019. He had complained of pain in the upper teeth. He was examined for pain in relation to the maxillary right first molar. Clinical examination revealed deep pockets of 9-11 mm depth in majority of maxillary and mandibular posterior teeth and generalized grade 2-3 mobility and grade 3 mobility with mandibular anterior teeth. Loss of attachment in relation to maxillary anterior teeth was > 8 mm. On the basis of preliminary findings, a provisional diagnosis of generalized aggressive periodontitis was made. Routine radiographs and orthopantamograph (OPG) were advised to substantiate the provisional diagnosis. OPG revealed generalized bone loss and sinus pneumatization (Figure 1). Furthermore, a cone-beam computed tomography (CBCT) scan was advised, which revealed generalized 'thistle-tube' like appearance of the roots (Figure 2). Pulp chambers were constricted with evidence of pulp stones in some chambers. A striking clinical feature was the presence of generalized brownish discoloration in the cervical part of all teeth.

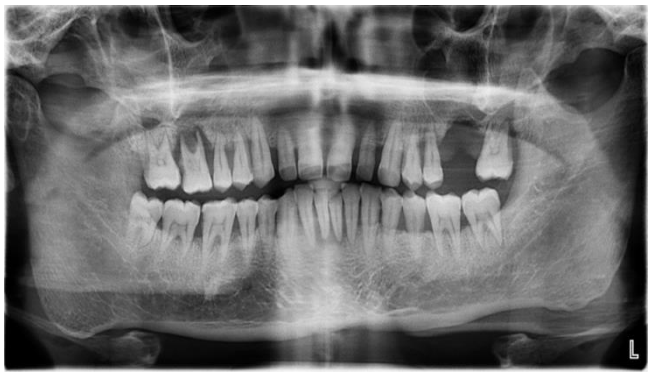

Figure 1. Pre-treatment orthopantomogram.

The patient was further asked on any familiarity with his condition with relation to any member of his family, to which he could not confirm. Treatment was planned in a sequential manner, owing to the aggressive tendency of the condition. Salivary samples at the pre-treatment stage were assessed for sialic acid (SA) levels to determine the severity of inflammation. As periodontitis was in the active state with severe inflammation, as confirmed by elevated SA levels of 8.54 mg/dl, the patient was prescribed antibiotics and analgesics (cephalexin 500 mg thrice a day for 3 days with ibuprofen 400 mg twice a day) for symptomatic treatment. The normal range of SA is 1.5-3 mg/dL. Once pain control was achieved, the maxillary right molar was extracted as an emergency treatment modality, and after a week, the maxillary anterior teeth were splinted, and mandibular anterior teeth were extracted. Oral hygiene instructions were provided, and chlorhexidine mouthwash (Curasept, Curaden AG, Kreins, Switzerland) was prescribed to the patient.

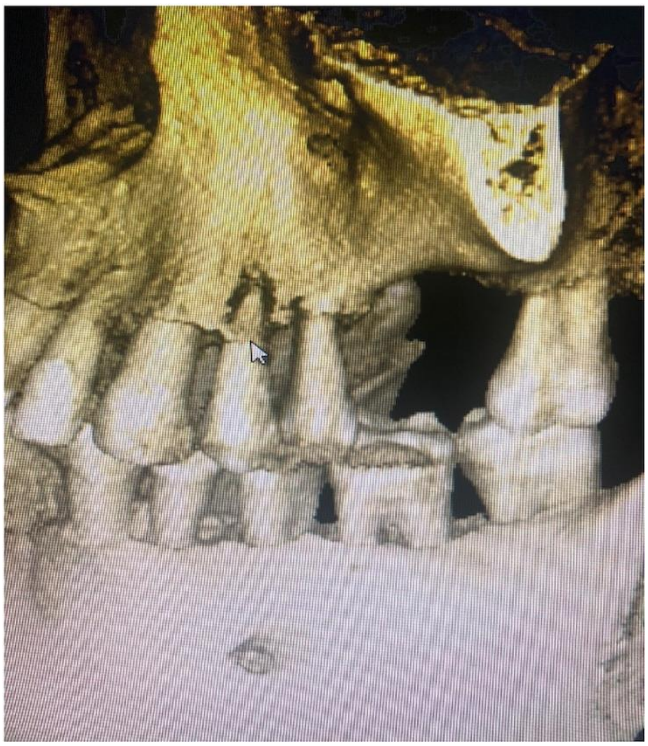

Figure 2. Intraoperative cone-beam computed tomography image.

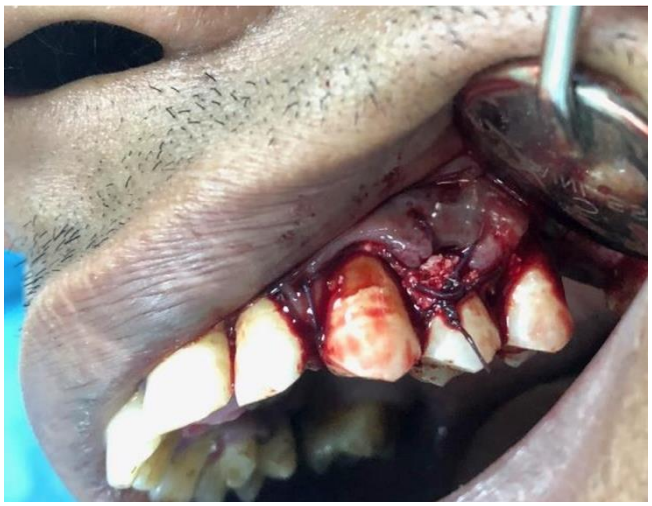

Figure 3. Periodontal flap surgery with placement of mucograft, bone graft, and splint.

The patient was recalled after a 2-week interval, and routine scaling and root planing procedures were performed. Teeth in late grade 1 and early grade 2 mobility were splinted (Ribbond Ultra-2 mm, Ribbond, Inc. Seattle, Washington, USA). The patient was recalled after one week for subgingival laser decontamination using a diode laser (980 nm, 200 μm width fiber optic tip, Dr Smile, Lambda Spa, 36040 Brendola, Italy). The patient was recalled again after one month, and scaling was performed and salivary samples were reassessed for SA levels.

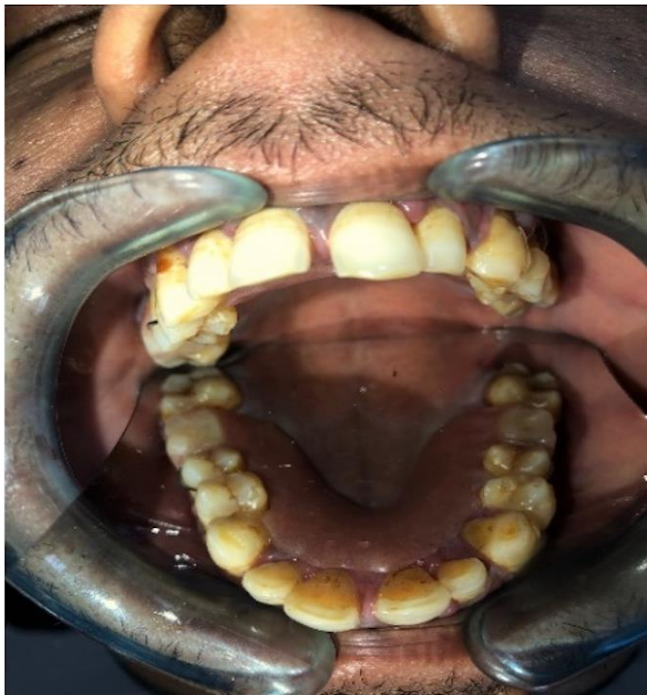

Figure 4. Placement of removable partial denture.

Once the salivary SA levels were decreased to 2.45 mg/dl, periodontal flap surgery using a bone graft and mucograft (Geistlich Bio-Oss granules 0.25 mm-1 mm, Princeton, NJ, USA; Geistlich Mucograft, 20 mm × 30 mm, Princeton, NJ, USA) (Figure 3) was performed in the premolar regions. The patient was prescribed a hyaluronic acid gel (Gengigel, Oral science, Ottawa, Canada) for topical use during the maintenance period. The patient was recalled after a month for re-evaluation. SA levels were reassessed to evaluate the severity of periodontitis. The levels of SA further dropped to 1.78 mg/dl, after which the final phase of the treatment plan, that is, prosthetic rehabilitation, was initiated. Although the severity of periodontitis was contained, considering the patient's current diagnosis, conservative prosthetic treatment was planned. A removable partial denture (Figure 4) was fabricated for the missing teeth, and the patient was instructed to schedule a recall visit at the clinic every two months for a period of 6 months. Routine scaling and local delivery of hyaluronic acid were performed at recall visits, followed by radiography to assess the current condition of the patient.

Discussion

The permanent teeth in type 1 and type 2 DD exhibit contrasting clinical features. In type 1, the primary sign is tooth mobility, which might lead to premature exfoliation.[11] Clinically, teeth appear normal. Radiographically, roots are shorter, sometimes absent, joined or with apical constriction. Periapical lesions are frequent without evidence of caries. In terms of discoloration, some studies have reported bluish to brown discoloration in the incisal region.[14]
In type 2 DD, the teeth are normal in shape, color, and height. Radiographically, they exhibit a thistle-tube appearance of the pulp and root anatomy, with a large pulp chamber and thin and slender roots. Pulp stones are also frequently seen.[16]

Teeth with dentinogenesis imperfecta (DGI) type 11 exhibit an opalescent discoloration, sometimes brownish in color, and decreased height due to extensive attrition caused by hypo mineralized dentin. Attrition can result in occlusal facets or complete absence of the coronal structure. Radiographs reveal short and thick roots with either complete obliteration of the pulp chamber or root structure. Periodontal diseases are commonly observed in relation to non-carious teeth.

Dentinogenesis imperfecta type 111, also termed the “Brandywine isolate,” was seen in a population in Maryland. Clinically, it affects both the permanent and deciduous dentitions, with bluish or opalescent discoloration of the dentitions, and most importantly, the presence of generalized attrition. Radiographically, there is pulpal enlargement, which results in reduction in volume of the dentinal tissue. The characteristic feature is the allelic relation between DGI-11 and DGI-111.[16]

From previously documented studies, the role of DSPP in the conversion of premineralized dentin to dentin has been fairly understood. The presence of DSP, dentin glycoprotein (DGP), and DPP is crucial in the process of dentinal defects. Although genomic studies have tried to correlate allelic expressions in immediate family members with hypo mineralization of dentin, the etiology is not clearly understood. DSPP, which is among the evaluated dentinal proteins, is required and essential for dentin maturation and formation. Most DSPP mutations that contribute to non-syndromic dentinal defects, namely, DD-1, DG1-11, and DGI-111, are evident because of the repetition of sequences within DPP.[16] DPP plays an important role in the mineralization of dentin matrix. Genomic sequencing of DPP is needed to better understand the role of these markers in dentinal defects.[17] Von Marschall et al.[18] had proposed that unlike DGI-1, a manifestation of osteogenesis imperfecta that is syndromic and related to gene mutations of COLIA1/2, there is a possibility that non-syndromic autosomal diseases like DGI-11, DGI-111, and DD-1 are linked to chromosome 4q. On the basis of another hypothesis, it was assumed that DD-11 might be caused by an allele gene of DGI. The involvement of a cluster of genes called Small integrin binding ligand N-linked glycoprotein (SIBLING), which is associated with DSPP, is believed to be responsible for the mineralization of bone cells by modulating MMP.1[8,19] Furthermore, McDougall, based on a linkage analysis, hypothesized an allelic relation between DD-11 and DGI-11.[20] To date, thirty-nine pathological variants have been described in relation to the DSPP gene, encoded within the peptide region of the DSP and DPP genes.[20]
The reason to use SA to time the treatment was that it plays an important role in regulation of host innate immunity. Moreover, resident microorganisms based in biofilms evade the immune/inflammatory response activated by host cells.[21] The release of SA is executed by the sialidase enzyme present in bacteria or host-derived neuraminidase, which results in collateral damage to the host tissues.[22] This activity made SA a good choice among the biomarkers to be selected for periodontal disease activity.

One of the characteristic clinical features exhibited by our patient, although not definitive, was a very slight hearing loss in relation to the left ear, which was reported only by Xiao et al.[23] The patient was referred to the department of otorhinolaryngology, and a slight sensorineural hearing loss was reported. Some studies documented in the literature have associated loss of hearing with g.49 C>A and g.1197 G>T mutations (MIM 605594), although some syndromic conditions do mimic DGI with severe microdontia, opalescent teeth, and rootless molars, such as Goldblatt syndrome, Ehlers-Danlos syndrome, Schimke immunoosseous dysplasia, and brachio-skeletal-genital syndrome.[24]

Our patient exhibited features from various types of dentinal defects reported in the literature, which could not be attributed to any single condition, as reported by Grewal et al.[25], making it difficult to make a concrete diagnosis. Moreover, the case presented with clinical and radiographic features of both type 1 and 2 DD (dual), with multiple clinical presentations of varied nature such as discoloration of the teeth, aggressive periodontitis, and generalized spacing of teeth (multivariate). We feel that this condition probably would have resulted from mutations in the existing allelic sequence or from habits, as reported by Qari et al.[14], thereby expressing clinical and radiographic features from varied types of DD.

Although an inter-disciplinary treatment approach was adopted, maxillomandibular atrophy will occur eventually because of the unpredictable nature of the course of the disease.[23] Periodic use of fluoride and dietary advice is essential. Periapical curettage and retrograde tooth filling might be advised in teeth involved Endodontically or with combined periodontal-endodontic lesions.[26,27] This form of treatment is not recommended for teeth with short roots.

Investigating the family history might bring into light the possible genomic transitions. Differential diagnosis with emphasis on DGI-11, DGI-111, DD-11, and DD-1 should be considered. In this particular study, a detailed biomarker evaluation would have defined the treatment period more appropriately. The evaluation of the biomarker SA provided a fair degree of success in maintaining the stability of the periodontium. Periodic supportive therapy paired with timely intervention ensures a fair prognosis in maintaining oral health status. The prognosis of DGI/DD depends primarily on the age of the patient and the treatment that was instituted.[28] A prompt diagnosis can reduce the treatment period, thereby reducing patient stress and providing good esthetic results. This, coupled with appropriate genetic counseling, will reduce the undue side effects of dentinal defects at a later stage.

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All authors have made substantive contribution to this study and/or manuscript, and all have reviewed the final paper prior to its submission.

Declaration of Patient consent

The authors certify that all patient consent forms have been obtained. The patient has given his consent for information to be published in the journal. The patients have understood that his name or contact details will not be published in the journal. All personal matter related to the patient will be strictly kept confidential. In the event that the journal requires the patient consent to be submitted, the details will be provided.

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Declaration of Interest

The are no conflicts of interest.

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