CASE REPORT

Dentinogenesis Imperfecta or Dentin Dysplasia? A Diagnostic Dilemma.

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Abstract

Background: Dentinogenesis Imperfecta and dentin dysplasia are developmental disorder of dentin with inherited autosomal dominant mode. Both of them are present with unique clinical, histopathological and genomic features. The diagnosis is based on family history, and detailed clinical examination, while genetic diagnosis may become useful in the future once sufficient disease-causing mutations have been discovered.

Objectives: To identify the overlapping features of Dentinogenesis Imperfecta and dentin dysplasia.

Case presentation: Here, we present a case of 22 years old female patient with complaint of discolored and brittle teeth. Clinical, radiographic and histopathological investigation shows overlapping features of both dentinogenesis imperfecta and dentin dysplasia.

Results: Treatment included removal of few teeth, crown lengthening of the maxillary and mandibular teeth, and placement of anterior and posterior tooth-coloured restorative materials. Besides wear and fracture, anterior teeth often have an unesthetic appearance because of discoloration. Current treatments of choice, including composite bonding restorations and, more recently, all-ceramic restorations, are typically suggested to preserve the remaining teeth and tooth structure.

Conclusion: This case unveils both the anomalies to be part of the same continuum of the genetic event.

Keyword: Dentinogenesis imperfecta, Dentin dysplasia, Polarized microscope, Stereomicroscope

Introduction

Dentinogenesis imperfecta (DI) is a hereditary developmental disturbance of the dentin in the nonexistence of any systemic disorder.[1] However, similar clinical features may be seen in conjunction with the systemic hereditary disorder of bone. But extensive studies have proven that DI is clearly a disorder distinct from bone disorder like osteogenesis imperfecta.

It is an autosomal dominant condition affecting both deciduous and permanent teeth. Barret in 1882 first recognised this condition, followed by Robert and Schour in 1939, who coined the term 'Dentinogenesis imperfecta'.[2] The condition typically results in the appearance of opalescent teeth often referred to as Capdepont's Teeth and Tulip Teeth. Overviewing the disagreements in the classification of DI, the revised classification by Wiktop is considered the most suited. In which there are two types, Type 1 with partial or total precocious obliteration of pulp chamber and root canals with continued dentin formation, blunt short root and Type 2 with normal enamel, thin dentin, and large pulp chambers, called as the Shell teeth.[1]

Interestingly, similar clinical features of DI can be overlapped with the other developmental disturbances of dentin, such as Dentin dysplasia (DD), which was first described by Ballschmiede, in 1920 as 'rootless teeth', because of spontaneous exfoliation of multiple teeth in seven children of one family. The first concise description of the disease was published in 1939 by Rushton, who was also the first to designate it as 'dentin dysplasia.'[3] Based on the clinical features seen in DD, Witkop has suggested that these conditions can be referred to as radicular dentin dysplasia (type I) and coronal dentin dysplasia (type II).[4]

Even though by the conventional definition of DD, it should have no correlation with DI,4 but the overlapping features of the both create diagnostic confusion. Hence, we are presenting a case of developmental disturbances of dentin which shares both the features of DI and DD creating diagnostic dilemma.

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Case Presentation

A 22-year-old woman's primary concern when she arrived at the Department of Oral Pathology and Microbiology,

GDC&RI, Bangalore, was that her teeth had been discoloured and brittle since she was a young child. The patient claimed that her mother had passed on the illness to her. The patient further disclosed that the same illness affects her maternal cousin. When questioned, it was discovered that her mother and maternal relative also gave similar histories, and that her deciduous teeth were similarly discoloured.

Patient had undergone esthetic treatment in her native place 5 years back. But gave a history of treatment failure. Apart from this, her past medical history was non-contributory. The history

Table 1: Showing the common features of DD and DI in our case:

Features of present case	Features related to Dentinogenesis imperfecta	Features related to Dentin Dysplasia
Pattern of inheritance	Autosomal dominant	Autosomal dominant
Severe attrition	Present	Present
Pulpal Obliteration with crescent shaped pulpal remnants	Totally obliterated	Obliteration with crescent shaped pulpal remnants
Root length	Short and blunt	Depends on the stage
Osteodentin	Present	Not common
Periapical Radiolucencies	Not common	Present

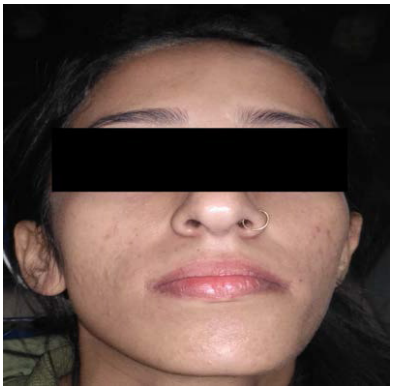

Fig. a Extra-oral examination shows no significant findings.

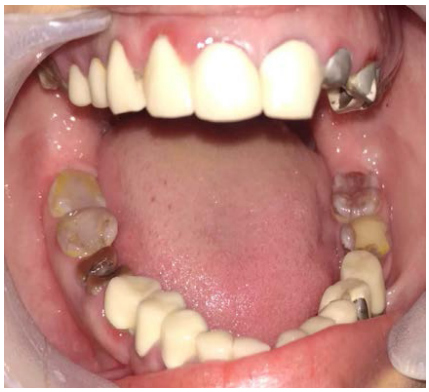

Fig. b Intra-oral examination reveals presence of fixed prosthesis from 15 to 25 and 35 to 45. Loss of ceramic facing irt 22 to 25. Grossly decayed irt 46. Clinically missing 36. Tooth coloured restorations irt 37, 47, 48

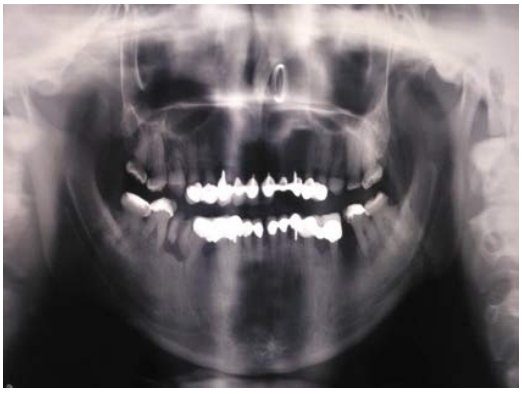

Fig. c Radiograph shows appearance of thin to absent enamel, with few teeth showing short, blunt, conical malformed roots and periapical radiolucency

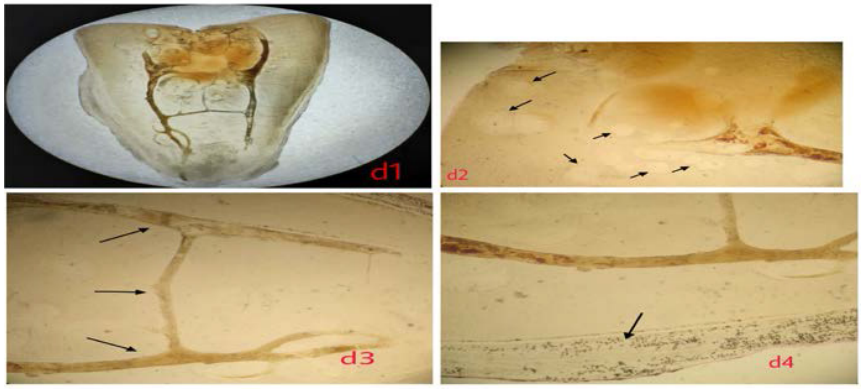

Fig. d Microscopic ground section;
d1: root stump with dentin and cementum,
d2: radicular areas showed whorls of tubular atypical dentin, d3: obliterated pulp chamber due to dentin deposition,
d4: obliterated pulp chamber due to dentin deposition

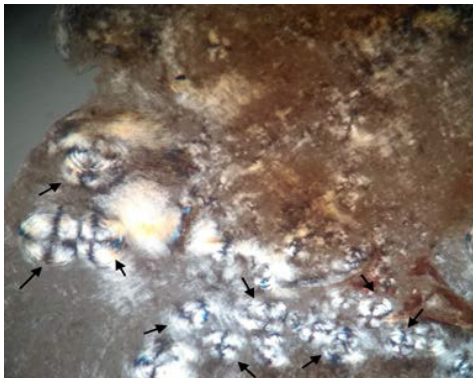

Fig. e Polarizing microscopic view of lava flowing around boulders appearance of atypical dentin

did not reveal any eruption disturbances. From a functional point of view, she had been avoiding hard food substances in recent days. Extraoral examination was nothing of significant (Fig a).

On intraoral examination, it was found that she had a normal complement of teeth. The thickness of enamel was reduced and was completely chipped off from some teeth exposing the dentin (Fig b). The emergence pattern and timing of teeth seemed to be within the normal range. No open bite was present. Upon further examination, occlusion was satisfactory and no loss of vertical dimension. Presence of fixed prosthesis from 15 to 25 and 35 to 45. Loss of ceramic facing 22 to 25. Grossly decayed 46. Clinically missing 36. Tooth coloured restorations irt 37, 47, 48. Examination of the periodontium revealed the presence of chronic, generalized, marginal, and papillary gingivitis, with calculus deposition and unsatisfactory oral hygiene.

Based on history and clinical examination a provisional diagnosis of Dentinogenesis imperfecta was proposed along with a differential diagnosis of Amelogenesis imperfecta, Environmental enamel hypoplasia, Dentin dysplasia, Regional odontodysplasia, Tricho-dentoosseous syndrome. After the complete examination patient was advised for orthopantamogram radiograph (Fig c).

The radiographic examination revealed thin to absent enamel, with few teeth showing short, blunt, conical malformed roots and periapical radiolucency. Based on the clinical and radiographic findings, a provisional diagnosis of dentinogenesis imperfecta was made. Patient consent was explained about the diagnosis and treatment outcome. Informed consent was obtained prior to the treatment onset.

The root stumps of the lower right permanent first molar were extracted due to poor prognosis and submitted for histopathological examination.

The root stumps were observed under stereomicroscope to assess the gross anatomy revealing brown to amber discoloured dentin.

The microscopic examination of ground section (Fig d) shows root stump with dentin and cementum (d1). The enamel is not seen. radicular areas showed whorls of tubular atypical dentin (d2) with obliterated pulp chamber due to dentin deposition (d3). The central portion of the root showed calcified tubular dentin, and atypical osteodentin, increase layer of cellular cementum (Fig d4) and fused denticles, giving the root the appearance of a "lava flowing around boulders"

Under polarizing microscopy (Fig e), lava flowing around boulders appearance of atypical dentin was observed along with focal areas showing hypercementosis with increase in thickness of cellular cementum.

Discussion

Both DD and DI are the developmental disturbances of dentin, which occurs during the period of histodifferentiation. The pathogenesis of DD is still unknown. Logan et al[5] proposed that it is the dentinal papilla that is responsible for the abnormalities in root development. They suggested that multiple degenerative foci within the papilla become calcified, leading to reduced growth and final obliteration of the pulp space. Wesley et al[6] proposed that the condition is caused by an abnormal interaction of odontoblasts with ameloblasts leading to abnormal differentiation and/or function of these odontoblasts.

The gene that causes defect in DI has been identified as Dentin Sialo phosphoprotein (DSPP) which maps to chromosome 4q21.3. This protein constitutes about 50% of the non-collagenous component of dentin matrix and believed to have a crucial role in converting predentin into mineralized dentin. The gene product is a precursor protein that is cleaved into two dentin-specific proteins, one as Dentin Sialoprotein (DSP) and other as Dentin Phosphoprotein (DPP).[7] Both are involved primarily for the development of the dentin. Hence mutation of the protein causes the developmental disturbances.

Malmgren and colleagues showed some evidence of a genotype-phenotype correlation, with one DSPP missense mutation being associated with a more severe phenotype in one family[8] Dean and colleagues, noting the phenotypic similarity of dentinogenesis imperfecta, to that in the primary dentition in dentin dysplasia, hypothesized that these conditions may be due to different alleles of the same gene.[9] Recent studies showed that out of eight mutations, seven are associated with DI and the eighth is known to produce dentin dysplasia type II. So both can show overlapping features, as evident in our case (Table 1).

The recent reconceptualization of Shields Types 2 and 3 along with DD Type 2 under the common umbrella of DSPP mutations offers a clearer and more unified framework for understanding these disorders. By recognizing them as part of a spectrum, clinicians can better diagnose, treat, and manage the dental and developmental challenges faced by affected individuals. This shift towards genetic understanding has the potential to refine both clinical practice and patient care, offering more personalized approaches to treatment and management of Dentinogenesis imperfecta and related disorders.

Soon after the diagnosis the treatment should be initiated in both the cases. Cast metal crowns on the posterior teeth and jacket crowns on the anterior teeth have been used with considerable success, although care must be taken in the preparation of the teeth for such restorations. Caution must also be exercised in the use of partial appliances which exert stress on the teeth, because the roots are easily fractured. Experience has further shown that fillings are not usually permanent because of the softness of the dentin. In case of tooth exfoliation rehabilitation must be done with suitable material.

Conclusion

As Dentinogenesis imperfecta and Dentin dysplasia have some common mutation factors, they may create a diagnostic confusion for the dentist with similar clinical and histological features. In both the cases early diagnosis and intervention is the key to prevent loss of enamel followed by subsequent loss of dentin and to achieve better prognosis.

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