

# Dentinogenesis Imperfecta Type 2: A Case Report

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## ABSTRACT

Dentinogenesis imperfecta (DI) is a development disorder involving the dentin. DI also known as hereditary opalescent dentine corresponds to a localized form of mesodermal dysplasia, observed in histodifferentiation. Dentinogenesis imperfecta causes esthetic as well as functional problems. Early diagnosis may prevent patients from these problems and provide a life without nutritional deficits and psychosocial distress. The diagnosis is based on family history, pedigree construction and detailed clinical and radiographic examination. Also, late intervention makes the treatment more complex. The present case describes an eight-year-old child with type II Dentinogenesis Imperfecta and briefly highlights the clinical and histological basis of this disorder.

**KEYWORDS:** Amelogenesis Imperfecta, Dentinogenesis Imperfecta Type1, Dentinogenesis Imperfecta Type2

## INTRODUCTION

Dentinogenesis imperfecta (DI) or hereditary opalescent dentin was first described in the late 19th century.<sup>1</sup> It is an autosomal dominant disorder characterized by abnormal dentine formation affecting either the primary or both the primary and secondary dentitions. Non-syndromic DI is reported to have an incidence of 1 in 6,000 to 1 in 8,000, whereas the incidence of Dentin dysplasia (DD) type I is 1 in 100,000.<sup>2</sup>

Shields et al. proposed three types of dentinogenesis imperfecta: DI type 1 is associated with osteogenesis imperfect (OI). DI type 1 and type 2 are similar clinically, radiographically and histologically, except that DI type 1 is with osteogenesis imperfecta; DI type 3 is only found in the triracial Brandywine population of Maryland.<sup>1</sup> DI type 2 and DI type 3 have thus been suggested as differential expression of the same gene. The Shields' system does not account for the molecular etiologies of the hereditary dentine defects.<sup>2</sup> Therefore, a revised classification was proposed where DI is classified as 1 and 2. Both types are not associated with OI. DI1 corresponds to DI type II and DI2 corresponds to the DI type III of Shields classification, respectively. There is no substitute for DI type I in this revised classification.<sup>1</sup>

In all three subtypes of DI, teeth of both dentitions are affected with variable clinical appearance. Teeth are opalescent with the color ranging from blue to gray.<sup>3</sup> The enamel tends to crack away from the defective dentin in about one- third of the patients. The dentin so exposed then tends to undergo rapid attrition.<sup>1</sup> Radiographically, the teeth show bulbous crowns with constricted short roots. Initially, pulp chambers may be abnormally wide

and resemble "shell teeth," but they progressively obliterate.<sup>1</sup>

Histologically, the dentin is irregular with reduced dentinal tubules. The characteristic scalloping of the dentinoenamel junction, which is thought to mechanically lock the enamel and dentin, is decreased; leading to loss of enamel. Generally, the structure of the mantle dentin is normal, with coarse and branched tubules in circumferential dentin. The total number of tubules is reduced.<sup>1</sup>

## CASE REPORT

An 8-year-old boy reported with a chief complaint of broken upper front tooth and discolored teeth. Patient's medical history was non-contributory. The family history suggested that the patient's father suffered from a similar condition of discolored teeth.

A detailed history of the chief complaint revealed that 2 days back the patient had a fall while running and suffered a sudden blow on the face. There was no abnormality noted on general and extra-oral examination. Intra-oral hard tissue examination showed Ellis class II fracture with respect to 11. Generalized yellowish gray discoloration of teeth was noted (Figure 1). 55,64,65 were grossly destructed. 85, 75 showed distocclusal caries and 16 had occlusopalatal caries. The patient also had an open bite.

Radiographic examination showed features characteristic of DI. There was a complete obliteration of pulp space with all deciduous teeth. 11, 12, 21, 22 and all permanent second molars 17, 27, 37, 47 showed widened pulp space. Cervical constriction with partial obliteration of pulp was noted with all 16, 26, 36, 46. (Figure 2)

*How to cite this article:*

Khot K, Madne S, Deshmane S, Bagri-Manjrekar K, Warke D. Dentinogenesis Imperfecta Type 2: A Case Report. Int J Dent Med Res 2015;1(6):72-75.



Figure 1: Generalized yellowish gray colour of teeth and 11 showing an Ellis class II fracture.



Figure 2: Orthopantomograph showing complete obliteration of pulp space in all deciduous teeth, partial obliteration of pulp in 16, 26, 36, 46 and widened pulp space in 11,17, 21, 27, 37, 47.

The family history, as well as clinical and radiographic examination, were consistent with the diagnosis of Dentinogenesis imperfecta type II (Shield's Classification). On histopathological examination, the dentin was composed of irregular tubules, with large areas of the uncalcified matrix. Tubules were larger in diameter and thus less numerous than normal (Figure 3A, B). Some areas showed complete absence of tubules. (Figure 3C, D) This further confirmed the diagnosis of Dentinogenesis imperfecta.

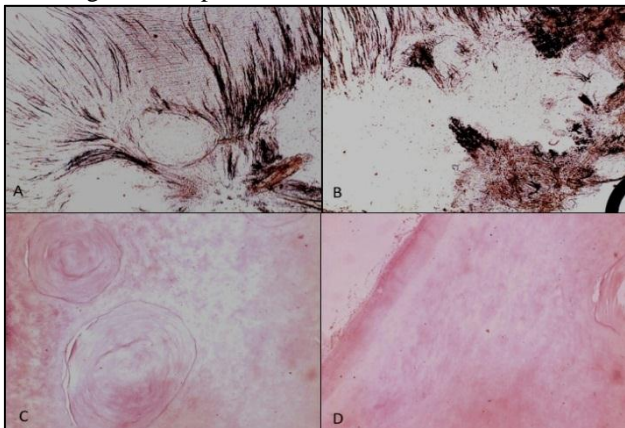


Figure 3A & B: Decalcified section showing dentin composed of irregular tubules with large areas of uncalcified matrix. Tubules are larger in diameter than normal. Figure 3C & D: H & E section showing some areas completely devoid of dentinal tubules.

The treatment was planned in two stages:

- An initial phase during mixed dentition period-during this stage 55, 64, 65 were extracted, and 16 was filled.
- A long-term prosthetic rehabilitation in the permanent dentition which included stainless steel crowns for all posterior teeth and polycarbonate resin crowns for all anterior teeth.

Patient was recalled every 3 months for a checkup.

## DISCUSSION

Dentinogenesis imperfecta (DI) is one of the most common genetic disorders affecting the structure of dentin. The term 'dentinogenesis imperfecta' was coined by Robert and Schour in 1939.

Dentinogenesis is a highly ordered process in which the organic predentine matrix is progressively mineralized by ectomesenchyme derived cells called odontoblasts. Dentine phosphoprotein, secreted from cellular processes of odontoblasts, acts as a nucleator of hydroxyapatite crystals during the mineralization process. Mesodermal dental abnormality is leading to overproduction of dystrophic dentine results in obliteration of the pulpal cavities. It is caused by mutations in the DSPP gene, which maps to chromosome 4 (gene map locus 4q21.3). It encodes a protein called Dentin sialophosphoprotein which constitutes about 50% of the non- collagenous component of dentin matrix.<sup>2</sup> The genetic basis for this clinical heterogeneity is unknown.

Like amelogenesis imperfecta, the classification of the disorders of dentin is gradually evolving as the result of these recent molecular genetics finding. Two systems, one by Witkop and other by Shields, were well accepted but not totally satisfactory.<sup>(3, 4, 5, 6)</sup> (Table 1)

Shields	Clinical presentation	Witkop
Dentinogenesis imperfecta I	Osteogenesis imperfecta with opalescent teeth	Dentinogenesis imperfecta
Dentinogenesis imperfect II	Isolated opalescent teeth	Hereditary opalescent teeth
Dentinogenesis imperfect III	Isolated opalescent teeth	Brandywine isolate

Table 1: A new classification of heritable human defects 11

The classification system for dentinogenesis imperfecta proposed by Shield et al. (1973), is briefly summarized.<sup>2</sup>

**Dentinogenesis imperfecta type I:** Individuals with DI-I also have osteogenesis imperfecta. The teeth of both dentitions are typically amber coloured, translucent with significant attrition. Radiographically, there is dentinal hypertrophy leading to pulpal obliteration and presence of short, constricted roots. Expressivity is variable, even within an individual.

**Dentinogenesis imperfecta type II-** The dental features of DI-II are similar to those of DI-I, but osteogenesis

imperfecta is not a feature. Bulbous crowns with marked cervical constriction are a typical feature.

**Dentinogenesis imperfecta type III-** This is a form of DI, known as the Brandywine isolate, found in a tri-racial population from Maryland. There are variable clinical features similar at times to those seen in DI-I and DI-II; but the primary teeth show multiple pulp exposures. Radiographically, they often manifest as "shell" teeth *i.e.* teeth which appear hollow due to hypotrophy of the dentine.<sup>(7,8,9)</sup>

The disadvantage of this classification is that it does not account for the molecular aetiologies for hereditary dentine defects.

The most recent classification adopted by the Mendelian Inheritance in Man (MIM) database is based on that of Shields but excludes DGI with osteogenesis imperfecta. Thus, the entity once termed DI-II by Shields has now become DI-I (MIM 125490), while the classification of DI-III (MIM 125500), DD-I (MIM 125400) and DD-II (MIM 125420) is unchanged.<sup>2</sup>

Genetic research has confirmed that Osteogenesis imperfecta with opalescent teeth is a separate disease from Dentinogenesis imperfecta. Osteopontin which is a bone glycoprotein is also expressed in dentin. However, there is no association between a type of polymorphism at the osteopontin locus and dentinogenesis imperfecta. Hence, the revised classification has been proposed.

DI type II is an entity clearly distinct from osteogenesis imperfecta (OI) with opalescent teeth and affects only the teeth. The teeth are opalescent with the color ranging from bluish gray to brown to yellowish. The dentin is abnormally soft, providing inadequate structural support to the overlying enamel. The exposed soft dentin undergoes rapid and severe functional attrition. Despite the exposure of dentin, teeth are not prone to caries.

Depending on the age of the patients, the teeth exhibit varying stages of obliteration of the pulp chambers. The cementum, periodontal ligament, and supporting bone are normal.<sup>2</sup>

Histologically, the enamel in DI is normal. The mantle dentin, a narrow zone of dentin immediately beneath the enamel, remains nearly normal, whereas the remaining dentin is severely dysplastic. Within the dysplastic dentin are focal areas of an amorphous matrix with globular and interglobular areas of mineralization. The dentinal tubules are disoriented, irregular, widely spaced and larger than normal.<sup>3</sup>

Reduced number of odontoblasts and a tubular area in the dentin with reduced mineralization, are consistent findings.<sup>4</sup> Interglobular dentin and pulpal inclusions are also seen in most cases. Biochemically, dentin shows a collagen defect and a primary defect in the calcifying matrix.<sup>1</sup> Takagi and Sasaki suggested that in DI type 2, dentine is deficient in the phosphorous ion, which is important in the early stage of odontoblastic differentiation and its mineralization. Susuki et al.

published a case describing DI type 2 with absent enamel prisms and abnormal mantle dentin.<sup>1</sup>

Dentinogenesis imperfecta should be differentiated both clinically and radiographically from Amelogenesis imperfecta (AI), Regional odonto dysplasia, Dentin dysplasia (DD), Tetracycline staining, Irradiation to jaws or chemotherapy during root development, Congenital erythropoietic porphyria and Dental Fluorosis.

Amelogenesis imperfecta like DI, is also a hereditary disorder. Unlike DI, the teeth exhibits increased sensitivity and on radiographs enamel is less radio-dense than dentine. The pulp chamber and root canals are usually not sclerosed.<sup>2</sup>

Regional odontodysplasia is a localised anomaly restricted to a single tooth or a group of contiguous teeth while in dentinogenesis imperfecta all the teeth are involved. In Regional odontodysplasia, the involved teeth either exhibit delayed eruption or do not erupt at all. The pulp chamber is very large giving a pale hazy image to the affected teeth, which is termed as ghost teeth.

Dentin dysplasia also produces crowns with altered colour and occluded pulp chambers. But the finding of a 'thistle tube' shaped pulp chamber in single rooted-tooth strengthens the possibility of dentin dysplasia. The crowns in dentin dysplasia are usually of normal shape, size and proportion while in dentinogenesis imperfecta teeth have bulbous shaped crowns with a constriction in the cervical region. If the roots are short and narrow, the condition is likely to be dentinogenesis imperfecta. On the other hand, normal appearing roots are present in dentin dysplasia type II or practically no roots at all in dentin dysplasia type I. Ultrastructural studies will help to understand the pathogenesis of the different types of heritable dentin defects as well as diagnosis of this disease.<sup>10</sup>

Congenital erythropoietic porphyria is a condition due to an error of porphyrin metabolism. This rare deficiency causes haemolytic anaemia, photosensitivity, blistering of the skin, and deposition of red-brown pigments in the bones and teeth. Mostly it is caused due to Rhesus incompatibility. The discolouration on the neck of the tooth ranges from yellow to greenish brown and grey to black. The enamel hypoplasias are usually located in the coronal third of the teeth.<sup>2</sup>

Tetracyclines have the ability to chelate calcium ions and to be incorporated into developing teeth, cartilage and bone which result in discolouration of both the primary and permanent dentitions. This permanent discoloration varies from yellow or grey to brown depending on the dose or the type of the drug received in relation to body weight.

Dental Fluorosis - Ingestion of drinking water containing fluoride at levels greater than 1 ppm during the time crowns are being formed may result in enamel hypoplasia or hypo calcification or fluorosis. Mild to moderate fluorosis ranges clinically from white enamel spots to mottled brown and white discolorations. Severe fluorosis

appears as pitted, irregular and discoloured enamel. No pitting is seen in dentinogenesis imperfecta and also the crown has no opalescent appearance. Pulp obliteration is seen in dentinogenesis imperfecta which is absent in dental fluorosis.<sup>2</sup>

The importance of restoring dental defects associated with DI is obvious. Treatment varies according to the age, the severity of the problem and presenting complaint. Many treatment modalities have been suggested: simple, removable appliances, over dentures, stainless steel crowns without or with acrylic facing, jacket crowns and pin-retained cast gold “thimbles” under acrylic resin crowns. Basically, preventional intervention is the key to diagnosis and maintenance of oral health in this disorder.

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Source of Support: Nil  
Conflict of Interest: Nil