

## Case Report

## Dentinogenesis imperfecta type II: an affected family saga

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**Abstract:** Dentinogenesis imperfecta (DI) type II or hereditary opalescent dentin is inherited in simple autosomal dominant mode with high penetrance and low mutation rate. It generally affects both the deciduous and permanent dentitions. DI type II corresponds to a localized form of mesodermal dysplasia, observed in histodifferentiation. Early diagnosis and treatment are therefore, fundamental, aiming at obtaining a favourable prognosis since late intervention makes treatment more complex. We present two cases of DI type II with the disease affecting three generations of a family in India, and briefly highlight the molecular basis of this disease. (J. Oral Sci. 49, 241-244, 2007)

**Keywords:** dentinogenesis imperfecta; type II; dentin; permanent dentition.

## Introduction

Dentinogenesis imperfecta type II is a genetic oral disease and was probably first recognized by Barret in 1882 (1). The term 'dentinogenesis imperfecta' was coined by Robert and Schour in 1939. We present two cases of DI type II in a family affected by this disease for the last three generations, as an attempt to make the general dentist and patient aware of the disease at an early stage. For better understanding and treatment planning of the disease, it should be detected at deciduous dentition level, and the dentist should understand the relevance and clinical

implications of genetic diseases in the oral cavity.

## Case Report

A 22 year old male visited the College Hospital with his 20 year old brother with complaints of pain and chipping of teeth. History revealed gradually chipped teeth of primary and permanent dentitions, due to lack of diagnosis of the condition and thus no proper treatment. On eruption, both dentitions were intact, varying from white to grayish in colour. Deciduous teeth were shed off prior to normal shedding age.

A family pedigree was revealed on their maternal side with affected mother, maternal grandfather, aunts and male maternal cousin. Maternal uncles were not affected and neither were their children (Fig. 1). The mother and aunts due to lack of awareness of the disease, and thus no timely treatment, were denture wearers since 20-25 years of age. They were born to a non-consanguineous couple. No history of any type of bone abnormality was associated

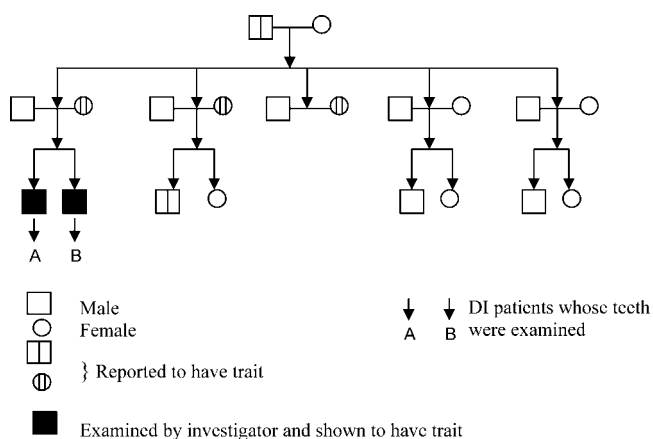


Fig. 1 Pedigree of dentinogenesis imperfecta in the patient's family.

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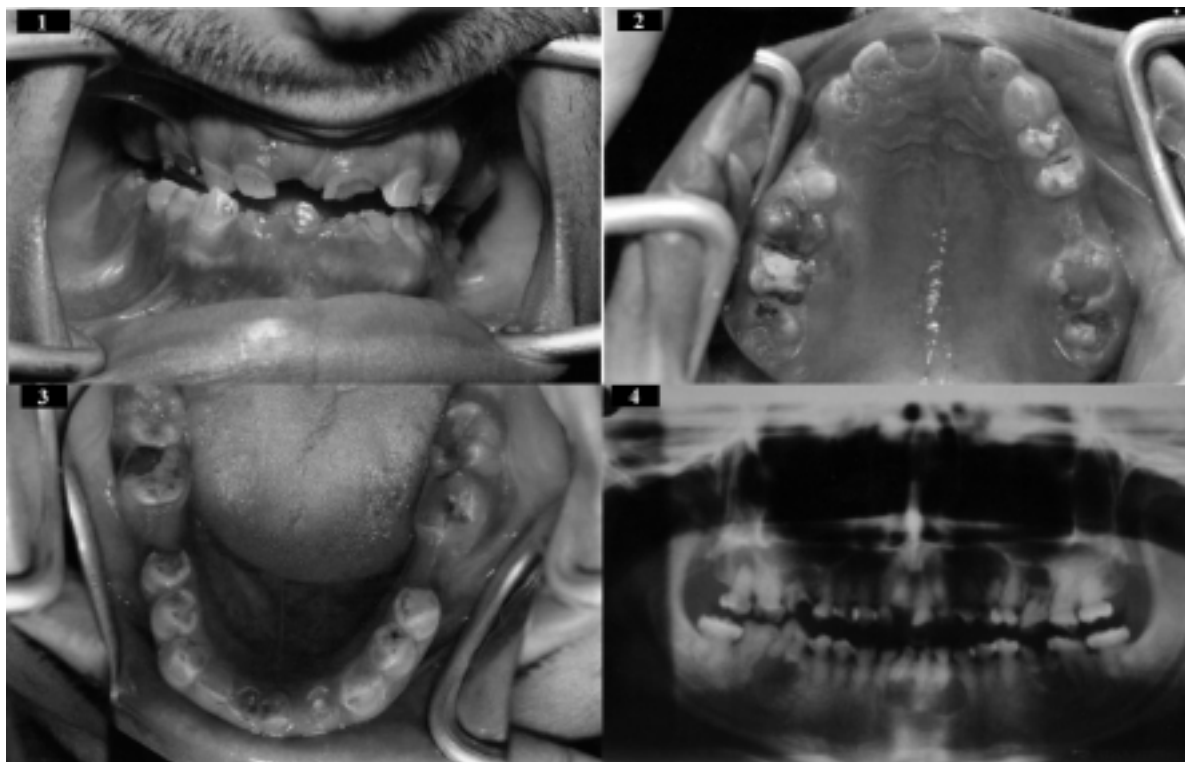


Fig. 2 (Case I)

1) Anterior view of the dentitions, revealing loss of enamel and vertical dimensions, 2) Intraoral view of the maxillary arch, 3) Intraoral view of the mandibular arch, 4) Panoramic radiograph of case I: teeth with pulp obliteration, bulbous posterior crowns and short, malformed roots.

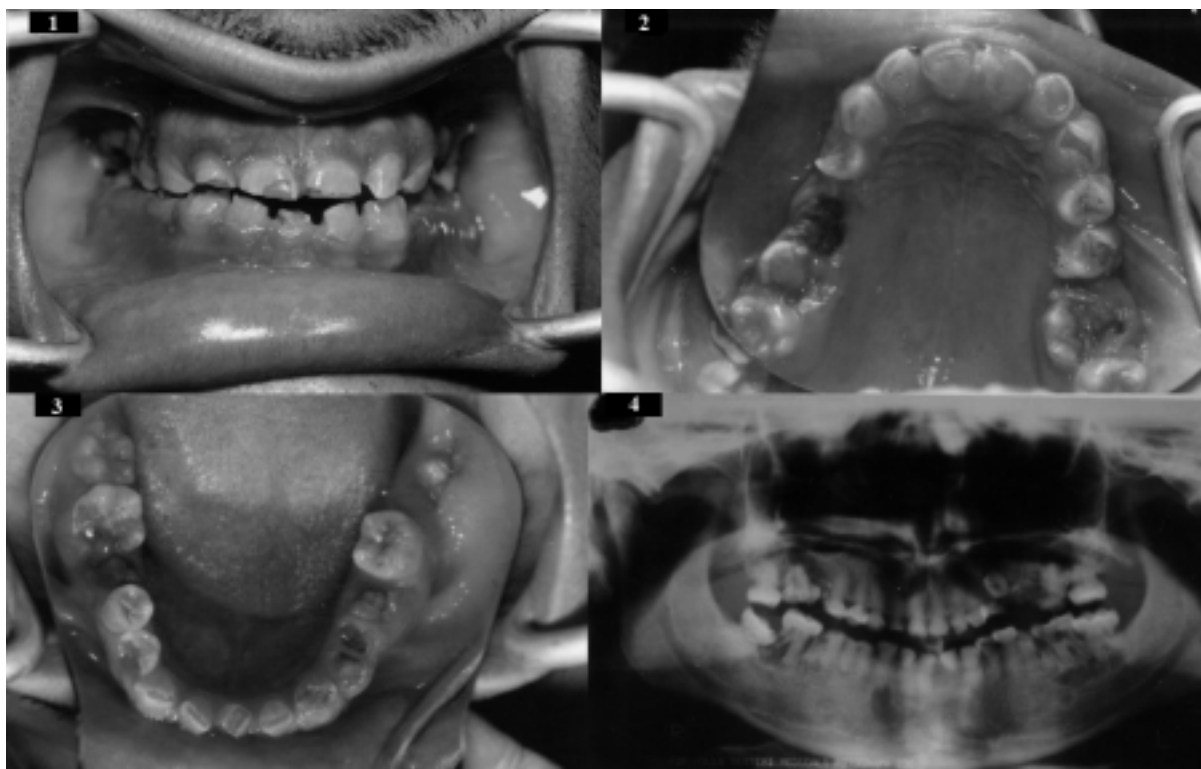


Fig. 3 (Case II)

1) Anterior view of the dentitions, revealing serious loss of enamel and vertical dimensions, 2) Intraoral view of the maxillary arch, 3) Intraoral view of the mandibular arch, 4) Panoramic radiograph of case II: teeth with pulp obliteration, bulbous posterior crowns and short, malformed roots.

with any of the family members.

Clinical examination of case I revealed serious loss of enamel and vertical dimension. Teeth revealed hard, brown dentin and most were with worn incisal and occlusal surfaces upto gingival level. Orthopantomograph (OPG) revealed periapical radioluscencies associated with several teeth. Third molars had intact enamel and pulp chambers were obliterated (Fig. 2).

Clinical examination of case II revealed most teeth with intact crowns, pale grayish enamel and hard brown dentin. OPG showed few periapical radioluscencies. Posterior teeth had bulbous crowns as compared to their short, malformed roots (Fig. 3).

Both cases showed no associated bone abnormality.

## Investigations

Orthopantomograph of the patient were done.

## Treatment

Case I: Treatment is difficult due to loss of vertical dimension leading to a severe posterior deep bite. For bite opening an acrylic plate has been given.

Case II: Composite veneers have been placed in the anterior teeth and root canal treatment followed by metal crowns with acrylic copings in the posterior teeth.

Main hindrances in treatment are the extreme fragility of the teeth and sclerosed canals. Based on the patient's history, family history, dental history, clinical and radiographic examination, the conclusion of DI type II was made. For further knowledge and research, DNA studies of the cases and their affected family members have been put into action; the results of which are awaited yet.

## Discussion

Dentinogenesis imperfecta (DI) was probably first recognized by Barret in 1882 (1). The term was coined by Robert and Schour in 1939. Shields divided dentinogenesis imperfecta into DI type I, II and III defect. Witkop named the types as dentinogenesis imperfecta, hereditary opalescent dentin, and brandywine isolate. It is a localized form of mesodermal dysplasia observed in histodifferentiation (1-3). DI type II has a reported incidence of 1:8,000 births. Genetically DI type II is transmitted as an autosomal dominant Mendelian trait with almost 100% penetrance. It probably represents a basic defect in structural or regulatory protein. The defective DI type II causing gene has been identified as dentin sialophosphoprotein (DSPP) and has been mapped to chromosome 4q21.3. The gene product is a precursor protein that is cleaved into 2 dentin-specific proteins, dentin sialoprotein (DSP) and dentin phosphoprotein (DPP). The genetic basis for this clinical

heterogeneity is unknown (4-8). Examination of the family pedigree revealed that in each instance an affected child had an affected parent. This indicated complete penetrance of the gene in the family. Though gene penetrance was high in the family yet gene expression was somewhat variable. Variation in clinical expression of this defect among family members has been reported, especially in relation to tooth colour and attrition pattern (1). DI is characterized by opalescent teeth with marked attrition and short roots constricted in cervical regions. Affected teeth have thickened dentin and a resultant obliteration of the pulp chambers (9). The discoloration in the primary dentition can be classified as yellow/brown or opalescent gray. The yellow/brown DI is more prevalent and more prone to attrition than the opalescent gray (10). This disorder may involve either primary or permanent dentitions, or both. Severity of the trait in individual teeth varies with age at which the particular tooth develops. Thus these cases have been reported as an attempt on our part to create awareness among general dentists so that they make common masses aware of these genetic oral diseases. The essential aim of treating this disorder is to prevent the abrasion of the erupted tooth and to establish proper vertical dimension (2). This will enable proper and early diagnosis and treatment so as to intercept attrition and preserve function, aesthetics and normal growth and improve aesthetics of the teeth. It is also said that those with DI in primary dentition maybe absent of DI in the permanent dentition. But, interestingly, those with DI in the permanent dentition always had DI in the primary dentition (10). Severe DI can be treated at two stages in deciduous teeth. If diagnosed early, stage I is around 18-20 months and stage II can be around 28-30 months (11). This will also help the disease to be curbed at its onset in deciduous dentition and pave the way for a healthier permanent dentition. At the same time, it reinforces the importance of a proper history taking of the patients.

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