

## Dentinogenesis Imperfecta Associated with Osteogenesis Imperfecta Type I

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### ABSTRACT

*Dentinogenesis imperfecta (DI) belongs to a group of genetically conditioned dentin dysplasia and is characterized clinically by an opalescent amber appearance of the dentin. The teeth with DI show a grayish-blue to brown hue with dislodged enamel, dysplastic dentine with irregular dentinal tubules and interglobular dentine, short roots and pulpal obliteration, which all may lead to rapid and extensive attrition. DI can appear as a separate disorder or with osteogenesis imperfecta (OI). Osteogenesis imperfecta (OI) comprises a heterogeneous group of disorders characterized by bone fragility, frequent fractures, and low bone mass. A case of DI associated with OI are presented in this paper with a purpose to present the dental and skeletal characteristics of DI associated with OI in mild form.*

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facial profile was found to be straight with normal mouth opening.



**Fig. 1: Blue sclera**

### INTRODUCTION

Dentinogenesis Imperfecta (DI) is a hereditary developmental condition that affects the structure and composition of dentine. It occurs in both syndromic and non-syndromic forms<sup>1</sup>. Osteogenesis imperfecta (OI) is one of the syndrome in which dentinogenesis imperfecta is seen. The incidence of OI varies between 6 and 20 in 100,000 newborns and its prevalence is 4-10 in 100,000 individuals. DI has been reported in more than 50% of patients suffering from OI<sup>2</sup>. Here we report a case of dentinogenesis imperfecta associated with osteogenesis imperfecta.

### CASE REPORT

A 19 year old female came to the department with a complaint of brown discoloration and decayed teeth. Her past medical history revealed that she was asthmatic since last 15 years and used inhaler containing steroid and long acting beta agonist, Albuterol Sulfate daily once. Other medical and also family history was insignificant. Past dental history revealed presence of similar brown discoloration in deciduous teeth. She got her both right and left lower back teeth removed 5 years back as they were grossly decayed from a private dental clinic. General Physical examination showed that she was short statured with height of 138 cm and weight was 39 kg and her eyes had pale blue sclera, while other features were normal. On extraoral examination, her

facial profile was found to be straight with normal mouth opening. Intraoral examination revealed normal soft tissues along with the presence of 17, 16, 15, 14, 13, 12, 11, 21, 22, 23, 24, 25, 26, 27, 31, 32, 33, 34, 35, 37, 38, 41, 42, 43, 44, 45, 47, 48 with missing 36 and 46. There was alteration in size of maxillary and mandibular anterior teeth due to attrition and alteration in shape of all teeth due to chipping off of enamel. The colour of teeth was bluish gray opalescence at incisal and middle 1/3<sup>rd</sup> and cervical 1/3<sup>rd</sup> showed white opacity. Pitting of enamel was evident on cervical 1/3<sup>rd</sup> of teeth which was present in rows parallel to incisal and occlusal surface (Fig.2). Proximal caries was present in both mesial and distal aspect of 12, 21, 22 and in distal aspect of 16, 26. In 37, 47 buccal surface caries was present. On exploration with explorer, enamel was hard in consistency with no tendency of chipping off of enamel. Teeth were neither tender nor mobile. Class I canine relation was present with the anterior deep bite.



**Fig. 2: Bluish gray opalescence hue of teeth**

A panoramic radiograph (Fig. 3) showed presence of normal complement of permanent teeth except 36, 46, 18, and 28. Teeth had bulbous crowns with cervical constriction and small pulp chambers lacking pulp horns. Roots were short and slender. Radiodensity of enamel was more than dentin. Crown completion with incompletely formed roots with large pulp canal of 38, 48 were seen. 16, 26 showed proximal radiolucency involving enamel, dentin and pulp.



**Fig. 3: Orthopantomogram**

Based on the typical findings of blue sclera, bluish gray opalescence hue of teeth, chipping off of enamel, bulbous crowns with cervical constriction and small pulp chambers lacking pulp horns a diagnosis of dentinogenesis imperfecta type I was made. Further, the Patient was referred to orthopedic department for skeletal survey which revealed no abnormalities except the short stature.

Treatment plan included extraction of severely destructed 26, root canal treatment of 16, 12, 21, 22, 31, 32, 41, 42 and providing cast crown restoration to achieve complete mouth rehabilitation.

## DISCUSSION

Dentinogenesis imperfecta (DI) is hereditary developmental condition, characterized by opalescent and translucent dentin due to a mesenchymal defect affecting both the primary and permanent dentition.

Incidence of DI is 1 in 6000 - 8000. History of dentinogenesis imperfecta dates back to 19<sup>th</sup> century when WC Barrett first recognized it in 1882<sup>3</sup>. Talbot in 1893 was the first to publish a report describing it as an enamel defect but later in 1908 Fargin-Foyelle and Malassez recognized that the defect is primarily due to abnormal dentin. In 1933, Skillen, Finn and Hodges first used the term 'hereditary opalescent dentin' and in 1939, the term dentinogenesis imperfecta was coined by Robert and Schour.<sup>4</sup>

DGI has been classified by Shields into three types, type I, DGI associated with osteogenesis imperfecta, type II, DGI without OI and type III, brandy wine type, which is a rare variety characterized by shell teeth, with very little dentin and multiple pulp exposures in the primary teeth.<sup>5</sup> Our patient falls under type I classification that is dentinogenesis imperfecta associated with osteogenesis imperfecta.

## Etiopathogenesis

DI is an inherited disorder which affects dentin. Dentine is composed of a mineral phase of hydroxyapatite (70%), an organic phase (20%) and water (10%). The organic phase is composed primarily of type I collagen (85%) and the remaining, non-collagenous protein is dominated by two proteins, DSPP (dentin phospho protein) and dentin sialoprotein (DPS). Dentine formation or dentinogenesis is highly ordered process in which the organic predentine matrix is progressively mineralized by ectomesenchymally-derived cells called odontoblasts. Predentine or unmineralised region contains type I collagen. This collagenous component of the matrix provides the correct three-dimensional structure into which the mineral component of dentine is deposited. Dentine phosphoprotein is secreted from cellular processes extending from the odontoblasts which acts as a nucleator of hydroxyapatite crystals during the mineralization process. Disturbances in the secretion of these proteins causing disturbance in the proper shape and placement of dental matrix crystals of apatite leads to dentinogenesis imperfecta. Mutations in the genes encoding the major protein constituents of dentine underlie to this hereditary dentine defect.<sup>6,7</sup>

DGI-I is a syndromic form of DGI associated with osteogenesis imperfecta which is an autosomal dominant condition, results from mis-sense mutations affecting either of the two genes encoding type I collagen (COL1A1 and COL1A2).<sup>8</sup> DGI type II and DGI type III is caused by mutations in the dentine sialophosphoprotein gene (DSPP), which is located within human chromosome 4q22.1 and consists of 5 exons spanning approximately 8343 bp.<sup>9</sup>

## Clinical Features

Clinical manifestation of dentinogenesis imperfecta associated with osteogenesis imperfecta is manifested in tissues rich in type I collagen, which is a result of

impaired collagen synthesis. Therefore characteristic clinical signs and symptoms includes bone fragility, deformity of the spine and long bones, short stature, blue sclera, and bluish gray opalescence hue of teeth.<sup>10</sup> Extraoral features include Growth deficiency, Joint laxity, Hearing loss, Blue sclera and Short stature. Silence classified OI based on clinical, genetically, and radiographic features into four groups. Type I is a mild form of OI. Type II is the lethal form of OI, even during the prenatal and perinatal period. Type III patients show progressive limb deformation. Patients with type IV of OI are those who show moderate to severe phenotypes and do not fit into any of the first three categories. There are three other new but uncommon types of OI. Hence, patients affected by these types do not demonstrate DI and blue sclera; OI types V-VIII are called syndromes resembling OI.<sup>11</sup>

Intraorally, primary teeth are more severely affected than is the permanent dentition. The color of the teeth varies from opalescent gray or brown to yellow, and both upper and lower dentitions are involved with commonly seen enamel fractures. Midface hypoplasia, class III malocclusion, unilateral or bilateral crossbite, ectopic eruption of the first or second permanent molars and missing of second premolar are more prevalent in this patients.<sup>12</sup>

### Clinical Differential Diagnosis

Clinically, dentinogenesis imperfecta should be differentiated from amelogenesis imperfecta, Congenital erythropoietic porphyria, Dentin dysplasia, hypophosphatemia. Hypocalcified forms of amelogenesis imperfecta initially develop normal enamel thickness but the poorly calcified enamel is soft and friable and is rapidly lost by attrition leaving dentine cores. It is differentiated from dentinogenesis imperfecta as on radiographs enamel is less radio-dense than dentine. Congenital erythropoietic porphyria is a rare condition resulting from an inborn error of porphyrin metabolism. This deficiency leads to haemolytic anaemia, photosensitivity, blistering of the skin, and deposition of red-brown pigments in the bones and teeth. There are also enamel discolourations and hypoplasias due to neonatal haemolytic anaemias. Dentinogenesis imperfecta is differentiated from congenital erythropoietic porphyria as discolouration in congenital erythropoietic porphyria ranges from yellow through to green, brown and grey to black is usually found at the necks of teeth and the enamel hypoplasias are usually located in the coronal third of the teeth. Both DI and DD can produce crowns with altered colour and occluded pulp chambers. The finding of a 'thistle tube' shaped 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. Hypophosphatemia causes mobility of teeth as seen in dentinogenesis imperfecta but it is differentiated from dentinogenesis imperfecta as due to aplasia or marked hypoplasia of cementum is seen in hypophosphatemia not in dentinogenesis imperfecta<sup>13,7</sup>.

### Radiographic Features

Radiographically, the crowns of the teeth are bulbous with marked cervical constrictions, and pulp chambers may be obliterated. Roots are short and slender. The thickness and radiodensity of the enamel are normal.<sup>14</sup>

### Radio-graphical Differential Diagnosis

Regional odontodysplasia, Vitamin D-dependent rickets and vitamin D-resistant rickets should be differentiated from DI. Regional odontodysplasia is a localized anomaly restricted to a single tooth or a group of contiguous teeth while in dentinogenesis imperfecta all the teeth are involved. Also the involved teeth either exhibit delayed eruption or do not erupt at all. Pulp chamber is very large giving a pale hazy image to the affected teeth, which is termed as ghost teeth.<sup>15</sup>

Vitamin D-dependent rickets is characterized by yellowish to brown enamel, chronic periodontal disease, large quadrangular pulp chambers and short roots. It can be differentiated from dentinogenesis imperfecta as there is loss lamina dura seen vitamin D-dependent rickets which is not seen in DI.<sup>7</sup>

### Histopathology

Histologically the dentin is composed of irregular tubules, often with large areas of uncalcified matrix. The tubules tend to be larger in diameter and less numerous in a given volume of dentin than in normal teeth.<sup>16</sup>

### Molecular Genetic Diagnosis

Diagnosis is based on history, clinical examination and radiographic features<sup>11</sup>. However, molecular genetics diagnosis may prove to be a useful adjunct to clinical analysis, particularly where the precise diagnosis is in doubt as the genetic mutations underlying these conditions are delineated<sup>17</sup>.

### Management

Treatment of DI has objectives of maintaining dental health and preserve vitality, form and size of the dentition, providing patient with an esthetic appearance at an early age in order to prevent psychological problems, providing patient with a functional dentition, preventing loss of vertical dimension; to avoid interfering with the eruption of the remaining permanent teeth and to allow normal growth of the facial bones and temporomandibular joint. Treatment of

mixed and permanent dentition is challenging and frequently demands a multidisciplinary approach. In the permanent dentition, full-coverage restorations are preferable, particularly when enamel chipping begins. In cases involving extensive attrition, over dentures are an option.<sup>18,19</sup>

### CONCLUSION

This case report presented a rare case of dentinogenesis imperfecta associated with osteogenesis imperfecta type I and illustrated that early and appropriate dental care can lead to improved control of oral disease, function and esthetics in DI.

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