Dentinogenesis imperfecta: case report and review of literature

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Abstract

Dentinogenesis imperfecta (DI) is one of the most common hereditary disorders of dentin formation. It follows an autosomal dominant pattern of transmission, affecting both the formation and mineralization of dentin. Either or both primary and permanent dentition is affected by it. It is characterized by the presence of opalescent dentin, resulting in a dusky blue to brownish discoloration of the teeth. Here we present a case of DI in a 20 year old female with remarkable clinical, radiological and histological presentation.

Keywords: Autosomal; Dentin; Odontoblasts; Opalescent.

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Introduction

The structural and numerical integrity of human dentition depends on various external as well as internal influences which attributes to development of several clinical disorders. Dentinogenesis imperfecta (DI) is one such autosomal dominant disorder that affects both the primary and permanent dentitions.[1] It is a localized mesodermal dysplasia consisting of opalescent teeth composed of irregularly formed and undermineralized dentin obliterating the pulp cambers and canals.[2] It is also known as 'Hereditary opalescent dentin' and 'Capdepont's teeth' affecting 1 in 8000 people.[3]

It was probably first recognized by Barret in 1882. The first published report describing the disorder as an enamel defect was by Talbot as quoted by Witkop (1971). The term "hereditary opalescent dentin" was first used by Finn (1933), Skillen (1937) and Hodges (1938) to describe the brown translucent teeth, which are opalescent lacking pulp chambers.[4] Here we present a case of DI in a 20 year old female.

Case Report

A 20 year old female reported with the complaint of pain in left lower back teeth region past 1 week which was dull and diffuse in nature. She also complained of poor aesthetics due to rapid wearing off the teeth surfaces. She gave history of similar coloured milk teeth which exfoliated and the permanent also resulted in same. Patient's medical history was non-contributory. Family history revealed that the patient's maternal grand-father and mother had similar features.

Oral examination revealed generalized opalescent teeth with yellowish brown discoloration. Generalized chipping of enamel was present. Fractured cusps were present in multiple teeth. Root stumps were present in relation to 36 and 37. Fractured crown portion with pulpal exposure was present in 38 which was non tender on vertical percussion.[Fig. 1 and 2]

Orthopantomograph (OPG) and intraoral periapical radiograph of 36, 37 and 38 revealed short roots, missing pulp chambers, obliteration of canals, with well-defined large periapical radiolucency in relation to fused 38 surrounded by a thin sclerotic border.[Fig. 3]

Based on the history, clinical and radiological investigations, a provisional diagnosis of Dentinogenesis imperfecta type II and periapical cyst in relation to 38 was given. The patient was sent for extraction of left lower third molar and tooth sectioning was done for histopathological examination.[Fig. 4]

Ground section of the specimen showed the presence of a thin layer of normal appearing enamel rods, lamellae and tufts. Presence of a disturbed dentinal matrix was seen with irregular dentinal tubules and degenerated, uncalcified matrix. Few areas showed presence of cellular inclusions indicating tall columnar cells (odontoblast like cells).[Fig. 5] All these features were suggestive of uncalcified dentinal matrix with normal appearing enamel confirming the diagnosis of Dentinogenesis Imperfecta.

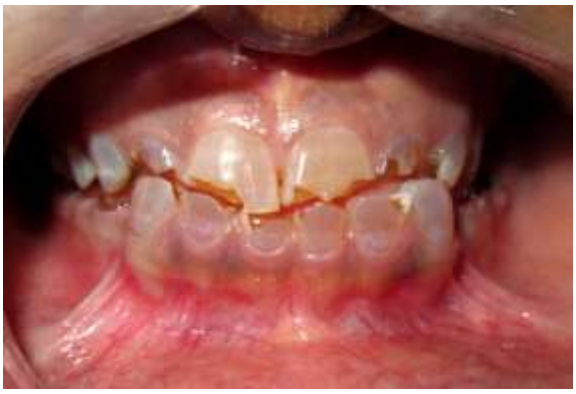

Fig. 1: Generalized opalescent teeth with yellowish brown discoloration

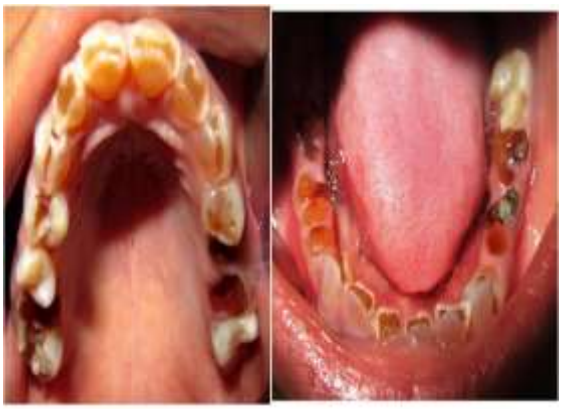

Fig. 2: Generalized chipping of enamel and fractured cusps in multiple teeth in maxillary and mandibular arch

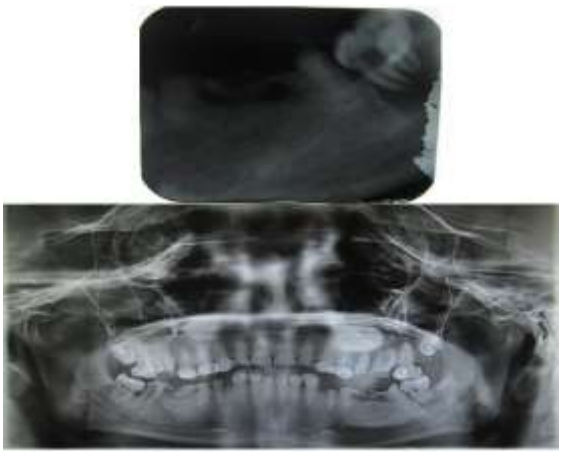

Fig. 3: OPG and intraoral periapical radiograph of 36, 37 and 38 showing short roots, missing pulp chambers, obliteration of canals, and periapical cyst in fused 38

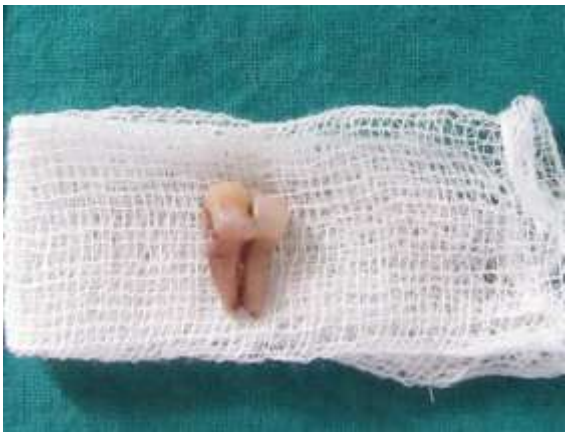

Fig. 4: Specimen of extracted 38

Fig. 5: Ground section showing presence of thin layer of normal appearing enamel rods, lamellae and tufts (10X) and disturbed dentinal matrix with irregular dentinal tubules and degenerated, uncalcified matrix (20X)

Discussion

DI was first classified by Shields in 1973 into three types: a) Type I with Osteogenesis imperfect (OI), b) Type II not associated with OI; also known as hereditary opalescent dentin and c) Type III-DI of the "Brandywine type" which was found in the Brandywine triracial isolate in Southern Maryland.[4] Multiple pulp exposures and "shell teeth" (due to large pulp chambers and thin dentinal walls) are two characteristic features used to distinguish DI type III from type II.

Extensive research over the years has proven that DI and OI are two separate and discrete entities, unrelated to each other. Therefore, a revised classification is proposed where DI is classified as 1 and 2. Both types are not associated with OI. DI1 correspondes to DI type II and DI2 to DI type III of Shields classification, respectively. There is no substitute for DI type I in this revised classification.[5]

Dentin has two proteins in its composition: dentinphosphoprotein (DSPP) and dentin sialoprotein (DSP). DSPP is expressed in a number of tissues including bone, kidney, salivary glands and lungs but its expression in dentin is hundred times higher than in

other tissues Disturbances in the secretion of these proteins, their accurate shape and arrangement of dentinal matrix hydroxyapatite crystals, clinically depicts DI. The genes responsible for producing both DSPP and DSP are located at 4q12-21. Type I and Type III DGI appear to result from mutations in the gene encoding DSPP suggesting that these conditions are allelic.[5,6]

Clinically, the appearance of the teeth with DI is characteristic showing a high degree of amber like translucency and color ranging from yellow to bluish grey when observed under transmitted or reflected light. Affected teeth have broad crowns with constriction of the cervical area which gives the teeth a tulip shape. The enamel easily fractures from the teeth and the crowns wear readily due to the poor support provided by the abnormal dentin and possibly in part to the absence of the scalloping normally seen between dentin and enamel.[4] Dental tissues in DI will have low hardness, elasticity and stiffness leading to a phenomenon of micro movement resulting in failure of restorations.[5] In adults, they may frequently wear down to the gingival and the exposed dentin becomes stained. The color of the abraded teeth may change to dark brown or even black. Some patients demonstrate an anterior open bite.[4,6] All these clinical findings were in accordance with present case.

Radiographically, the teeth appear solid, lacking pulp chambers and root canals, slight to marked attrition of the occlusal surface, short and slender roots and constriction of the cervical portion of the tooth giving the crown a bulbous appearance. Types I and II DI usually show partial or complete obliteration of the pulp chambers which was evident in the aforementioned case. Early in development, the teeth may appear to have large pulp chambers, but these are quickly obliterated by the formation of dentin. Ultimately the root canals may be absent or threadlike.[6,7]

Syndromes associated with dentinogenesis imperfecta are OI, Ehlers Danlos syndrome, Goldblatt syndrome, Schimke immunoosseousdysplasia, Brachio-skeletogenital syndrome.[8] DI should be differentiated both clinically and radiographically from Amelogenesis imperfecta, Regional odontodysplasia, Dentin dysplasia (DD), Tetracycline staining, Irradiation to jaws or chemotherapy during root development, Congenital erythropoietic porphyria and Dental Fluorosis.[5]

The entire dentition is at risk because of numerous problems. The root canals become thread-like and may develop micro exposures, resulting in periapical inflammatory lesion which was obvious in our case (38 affected with periapical cyst). Inspite of the risk of enamel loss and significant attrition, the teeth are not good candidates for full crowns because of cervical fracture. The success of full coverage is best in teeth with crowns and roots that exhibit close to a normal shape and size. Overlay dentures placed on teeth that are covered with fluoride releasing glass ionomer cement have been used with success in some cases.[3]

Conclusion

DI causes aesthetic as well as functional problems affecting the patient's mental and physical health. As an oral physician, it is a challenge to diagnose such disorders at the earliest so that good aesthetics and function can be restored thereby minimising nutritional deficits and psychosocial distress.

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