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Dentinogenesis imperfecta in a four year old child: A case report

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Abstract
Dentinogenesis imperfecta represents a group of hereditary conditions that are characterized by abnormal dentin formation. These conditions are genetically and clinically heterogenous and can affect only the teeth or can be associated with the condition osteogenesis imperfecta. It is an uncommon defect in the collagen formation that is transmitted as an autosomal dominant trait. This case report discusses the dental manifestations of DI in a 4-year-old. Rigorous home care instructions, including reinforcement of the oral hygiene practice were discussed with the parents.

Keywords: dentinogenesis imperfecta, osteogenesis imperfecta, clinical manifestations, medical and dental consideration

Introduction

Dentinogenesis imperfecta (DI) is an inheritable disorder of tooth development that occurs during the histodifferentiation stage. It is a genetic disorder of tooth development which affects an estimated 1 in 6,000 to 8,000 people [1]. 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 [2]. The term 'hereditary opalescent dentin' was first used by Skillen, Finn and Hodges to describe the brown translucent teeth that have an opalescent sheen and are lacking in pulp chambers [3]. Clinically, the appearance of the teeth with DI is characteristic. They show a high degree of amber like translucency and a variety of colors from yellow to blue-gray. The colors change according to whether the teeth are observed by transmitted light or reflected light. Affected teeth have broad crowns with constriction of cervical area resulting in tulip shape. The enamel easily fractures from the teeth and the crowns wear readily. In adults they may frequently wear down to the gingiva. 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. Radiographically the teeth appear solid, lacking pulp chambers and root canals. Radiographs may also reveal slight to marked attrition of the occlusal surface. The roots are usually short and slender. Early in development, the teeth may appear to have large pulp chambers, but these are quickly obliterated by the formation of dentin. Poorly developed teeth wear down rapidly, break easily and are prone to loss at a young age.

- **Type I:** Type of DI with similar dental abnormalities usually an autosomal dominant trait with variable expressivity but can be recessive if the associated osteogenesis imperfect (OI) is of recessive type [4].
- **Type II:** Occurs in people without other inherited disorders (i.e. OI). It is an autosomal dominant trait. A few families with type II have progressive hearing loss in addition to dental abnormalities.
- **Type III:** Type is rare; its predominant characteristic is bell-shaped crowns, especially in the permanent dentition. Unlike Types I and II, it involves teeth with shell-like appearance and multiple pulp exposures [5].

Mutations in the DSPP gene have been identified in people with type II and type III dentinogenesis imperfecta. Type I occurs as part of osteogenesis imperfecta. Clinical appearance is variable. However, the teeth usually involved and more severely affected are primary teeth in type I; whereas in type II both the dentitions are equally affected [6].

Case report

A 4-year-old girl reported to department of Oral Medicine, Diagnosis & Radiology, Institute of Dental Education & Advance Studies, Gwalior, Madhya Pradesh with a chief complaint of decayed teeth in upper and lower jaw region since 3 months. Patient medical history was non-contributory. The family history suggested that none of relatives suffered from a similar condition of discolored teeth. There was no abnormality noted on general and extra-oral examination.

Teeth present	55 54 53 52 51	6162 63 64 65
	85 84 83 82 81	31 72 73 74 75

Generalized brownish discoloration of teeth was noted. 51 52 61 62 72 81 82 83 85 were found to be discoloured. (Fig 1 & 2).

51 52 53 54 61 62 74 84 85 were grossly destructed. Pre shedding mobility was found to be associated with 81. Panoramic radiograph was taken (Fig 3). Clinical examination and radiographic examination were consistently with features of DI. There was a complete obliteration of pulp space with all deciduous teeth. 11 12 21 22 and all permanent second molars 16 26 36 46 showed widened pulp space. Cervical constriction with partial obliteration of pulp was noted with all 16 26 36 46. Patient was referred to department of pedodontics and preventive dentistry for management.

Discussion

Dentin is the most abundant dental tissue and largely determines the size and shape of teeth. Dentin is formed by

odontoblast cells. The unique structure and composition of dentin allow it to function as the substructure for the rigid enamel tissue, thereby imparting teeth with the ability to flex and absorb tremendous functional loads without fracturing. Dentin contains about 60 percent mineral by weight and, unlike enamel, has a substantial organic component (20 percent). Dentin contains a complex organization of tubules that are approximately 1um in diameter, filled with fluid and/or the cellular processes of the odontoblasts and are thought to play a role in the neurosensory function of teeth. Additional dentin can be deposited along the tubule (sclerotic dentin) or the pulpal wall in a reparative or protective mode in response to environmental stimuli such as trauma, tooth wear or dental caries [7]. Dentin formation involves numerous genes that produce a complex extracellular matrix that is highly organized, is processed, sand that eventually mineralizes in a highly controlled fashion. Type I collagen (product of COL1A1 andCOL1A2 genes) is the most abundant dentin protein. This complex molecule has the structure of a heterotrimer and forms the foundation for several mineralized tissues including bone and dentin.

The collagen molecules interact with a variety of non-collagenous proteins to help initiate and regulate the mineralization process in these tissues. Interestingly, mutations in either type I collagen or proteins that interact with it can cause the DI dental phenotype. There are known to be hundreds of different mutations in these two genes that are associated with OI, a group of hereditary defects associated with bone fragility [8]. Interestingly only some of the collagen mutations result in the dental manifestations of DI. There are numerous non-collagenous proteins present in dentin, some of which interact with collagen to initiate and/or regulate mineralization. The most abundant non-collagenous protein, dentin sialophosphoprotein (product of DSPPgene) is a highly phosphorylated protein that attaches to the type 1 collagen fibril helping regulate mineralization at specific sites within the collagen. Mutations in either type 1 collagen or DSPP can alter this interaction resulting in abnormal mineralization and a DI dental phenotype [9]. The molecular defects in OI include numerous mutations in the pro-alpha chains of collagen type 1 that result in a phenotype characterized by increased bone fragility [50]. Although the dental phenotypes of DI types I and type II appear very similar, the latter disorder is not associated with any of the non-dental phenotypic features of osteogenesis imperfect and is not caused by a collagen 1 defect. DI type II and type III are autosomal dominant conditions that have been linked to chromosome 4q12-21, suggesting these may be allelic mutations of the DSPP gene. In all three DI types the teeth have a variable blue-gray to yellow brown discoloration that appears opalescent due to the defective, abnormally colored dentin shining through the translucent enamel. Due to the lack of support of the poorly mineralized underlying dentin, the enamel frequently fractures from the teeth leading to rapid wear and attrition of the teeth.

The severity of discoloration and enamel fracturing in all DI types is highly variable even within the same family. If left untreated it is not uncommon to see the entire DI affected dentition worn off to the gingiva. Radiographs show pulpal obliteration in DI types I and II due to rapid and excessive deposition of dentin. The pulp chambers are large in DI type III. These are usually known as "shell teeth" [10].

Dental pulp

The dental pulp is a specialized tissue comprised of a layer of odontoblasts, fibroblasts, blood vessels, nerves and a complex extracellular matrix. The pulp provides the reparative potential of teeth and neurosensory function. The dental pulp can increase production of dentin (reparative dentin) in an attempt to protect and wall off the vital pulp tissue from the injury or noxious stimuli. Prompt treatment of dental trauma and dental caries are critical steps towards maintaining a healthy vital pulp and allowing an injured or diseased tooth to retain a vital pulp. The pulp will continue to lay down small amounts of dentin throughout the life of a tooth as part of the normal pulp physiology. This process ultimately results in a smaller pulp chamber in people as they age and are part of the reason teeth continued to become more yellow in colour with age. It is especially critical to maintain a healthy dental pulp until the root is fully formed and its walls are of adequate thickness to maintain the tremendous forces transmitted from the crown during function. If the pulp becomes non-vital in a young tooth that lacks complete root formation, it is much more difficult to complete endodontic treatment successfully and the prognosis for retaining the tooth is diminished. In teeth with DI, the inside where the nerves and blood vessels are normally located may already be filled with dentine. This makes placing a post in the center of the root and/or root canal treatment difficult, if not impossible. Small reinforcing pins may be placed in the dentine away from the center of the root to help make the new crown of the tooth stronger [11].

Differential diagnosis

Includes conditions that may have similar clinical or radiographic features to DI such as OI and dentin dysplasia. The clinical symptoms may be mistaken for hypocalcified amelogenesis imperfecta, congenital erythropoietic porphyria, conditions leading to early tooth loss (Kostmann syndrome, cyclic neutropenia, Chediak-Hegashi syndrome, Langerhans cell histiocytosis, Papillon-Lefèvre syndrome, vitamin D-resistant rickets, as well as permanent teeth discoloration due to tetracyclines or dental fluorosis. It is important to investigate for a history of bone fractures with minimal trauma, hearing loss and to check for blue sclerae to rule out osteogenesis imperfecta [12].

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. Regional odontodysplasia is a localised anomaly restricted to a single tooth or a group of contiguous teeth while in DI all the teeth are involved [13].

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 DI 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 DI. 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 [14].
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 [15].
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 DI and also the crown has no opalescent appearance. Pulp obliteration is seen in DI which is absent in dental fluorosis [16].

Histologically
Histologically in DI 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.

Management and treatment
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. Protection of primary and then permanent teeth with preformed pediatric crowns, cast occlusal onlays on first permanent molars and eventually premolars, may help to minimize tooth wear and maintain the occlusal vertical dimension. Obliteration of pulp spaces in teeth that develop abscesses makes endodontic treatment extremely difficult if not impossible. Appropriate care makes it possible to achieve good esthetic appearance and functional performance. The replacement of teeth might be considered with dentures or implants. At first a patient is carefully examined in order to evaluate bone fragility and other symptoms. This is extremely important for young children or for patients with no family history of DI. Findings like frequent bone breaks can change later treatment tactics. Although DI cannot be cured but the natural teeth can be saved. Proper care also helps to avoid caries, abscesses, pain and ensure an optimized esthetic appearance. Although brushing, rinsing and other appropriate oral hygiene procedures are necessary, they do not make any difference to the colour of the teeth. Bleaching, which is the most popular procedure done to improve the look of the teeth, is not an effective option for DI either.
This is because the discolouration of the teeth is caused not by enamel but by alternation of the deeper lying dentin. Both brushing and bleaching work only on the enamel [17].

Sealants
As long as the enamel, the outer layer of the teeth, is intact sealants should be affective for the teeth. Sealants are transparent artificial plastic material teeth coverings that protect the chewing surfaces from decay due to germs and food particles. Adding sealants does not require drilling or damaging the tooth. If properly done, sealants can last up to 10 years.

Veneers
Usually bonding of veneers is preferred. Veneers are porcelain facings for the teeth. They come in various shapes, colours and sizes. They alter the teeth minimally but make a great improvement for the smile. Intra-coronal dental restorations (for example amalgams and composites) can also be done for patients without enamel loss [18].

Crowns and Bridges
In more severe cases, when enamel is severely damaged or worn off, full coverage crowns and bridges are preferred. Crowns are metal or ceramic caps that fully cover the teeth. They improve the appearance and strength of the teeth. Bridge is used when at least one artificial tooth is attached to one or more crowns.

Complete Dentures
Complete Dentures are used when there are no teeth remaining in one or both jaws. How well the denture fits depends on how much bone remains after the teeth are lost. There are no studies that compare bone loss under dentures in people with OI to people without OI. The bone loss that occurs when teeth are lost is a resorption of the bone, not a fracture process, so it is not known if bone loss would be more rapid in people with OI. Complete dentures in children and adolescents who are still growing will need to be adjusted and or remade on a routine basis to compensate for growth in the jaw [19].

Removable Partial Dentures
Removable Partial Dentures are used when some teeth remain in one or both jaws. A denture, typically made with a metal framework for strength and retention, is constructed to replace missing teeth.

Dental implants
Dental implants in some cases, if the jaw bone is not affected by a genetic disorder, dental implants can be considered. Sometimes only removable partial or complete dentures can be done.

Periapical abscesses
Some patients with DI also suffer from multiple periapical abscesses. Because weak enamel and dentin wear off easily, the pulp - the center part of the tooth that has blood vessels,

nerves, connective tissue becomes exposed and very likely to become infected. Apical surgery might be required to maintain the abscessed teeth and to prevent the infection from spreading. With proper medical care people affected by dentinogenesis imperfecta can enjoy good quality of life and an appealing appearance [20].

Treating Malocclusions with Orthodontia or Orthognathic Surgery

A malocclusion is an abnormal relationship between maxillary and mandibular teeth, which creates problems with how the teeth come together. This may be due to the relationship of the upper and lower jaws to each other, the alignment of the teeth, or both. This type of problem includes crooked teeth, "underbite," "overbite" and "open bite." Treatment is usually provided by an orthodontist. The particular treatment plan depends on the specific problem with the bite and the teeth. If the malocclusion is caused by skeletal discrepancies, then orthognathic surgery may be required along with orthodontia. An orthodontist should examine each child with OI around the age of 7 years. At that time early orthodontic interventions in children who are developing a relatively small upper jaw compared to the lower jaw may help decrease the need for later orthognathic surgery.

Although there are only a few case reports and no published studies regarding orthodontia for people with OI, it seems to be safe to treat them if DI is not present. If DI is present, the orthodontist will have to decide if the enamel is strong enough for braces. Unfortunately, it is difficult to determine how strong the enamel is until it is tried. Conventional practice involves gluing brackets to the teeth for the braces and removing the brackets later. Plastic brackets can be used instead of metal brackets because they can be removed with a hand piece without disturbing the fragile enamel. If there is concern about the enamel cracking off and treatment is still desired, placing bands on all the teeth to hold the brackets may work. Although bands are considered an "old fashioned" method, the technique still works. It may be necessary to seek out an older orthodontist who learned to install braces before the current practice of gluing bands directly to teeth was discovered. The orthodontist will need to minimize forces on the teeth as well as movement of teeth over long distances. The wires which are attached to the bands should initiate slow and light movements.

Whenever possible, removable orthodontic appliances are preferred. Caps, or crowns, may also be effective in correcting rotations or mildly malpositioned teeth. In some children with OI the upper jaw, or maxilla, does not grow as much as the lower jaw, or mandible. Sometimes the way that both jaws grow makes it difficult, if not impossible, to bring the teeth together properly, even after orthodontic braces. If the malocclusion is due to a problem with the growth of one or both jaws, then a combination of orthodontic braces and orthognathic surgery may be used to align the teeth. Some period of orthodontic braces is also usually needed after the jaw surgery. There are a few published reports about these surgeries indicating good post-operative healing of the jaws. The same concerns that one would have with any surgery in people with OI, such as potential bleeding problems and reaction to general anesthesia, still apply.

Furthermore, the recent use of bisphosphonates to treat different bone disorders triggers many additional questions regarding maxillo facial surgeries. Bisphosphonates work by reducing the remodeling rate in the skeleton. In the short term, reduction of the remodeling rate produces bone with a greater density, although it is not clear if this results in greater strength. It is also not clear what impact this reduction in remodeling will have long term. Because the remodeling rates for bone surrounding teeth are typically higher than for other bones in the body, additional questions arise about the effect of bisphosphonates on the oral cavity. It is also not clear what effect bisphosphonates have on young children whose new teeth are erupting as they grow. Similarly, the effect of bisphosphonates on the necessary remodeling surrounding dental implants is not understood. Separate from the concern about BON is the likelihood that tooth movement from orthodontia will decrease if the patient is taking, or has within some period of time, been taking bisphosphonates. These include: pamidronate (Aredia®) and zoledronic acid (Zometa®) given by intravenous infusion, and alendronate (Fosamax®), risedronate (Actonel®), and ibandronate sodium (Boniva®) given in tablet (oral) form [21].

Treating Impacted Teeth

The dentist needs to consider if the impacted teeth should be left alone or extracted, or if an attempt should be made to move them into a functional position in the mouth. To move a tooth, a coordinated effort is needed between the oral surgeon and the orthodontist to surgically uncover the impacted tooth and glue an attachment onto the tooth so that light force from the braces can be used to bring the tooth into the proper position. The orthodontist may also use braces prior to surgery to be sure there is space to bring the impacted tooth into the proper position [22].

Conclusion

The patients with the DI condition present with different degrees of severity. A comprehensive interdisciplinary treatment planning is required to rehabilitate these patients. This present case have been reported as an attempt on our part to create awareness among the general dentists, so that they can make the common masses aware of these genetic oral diseases.

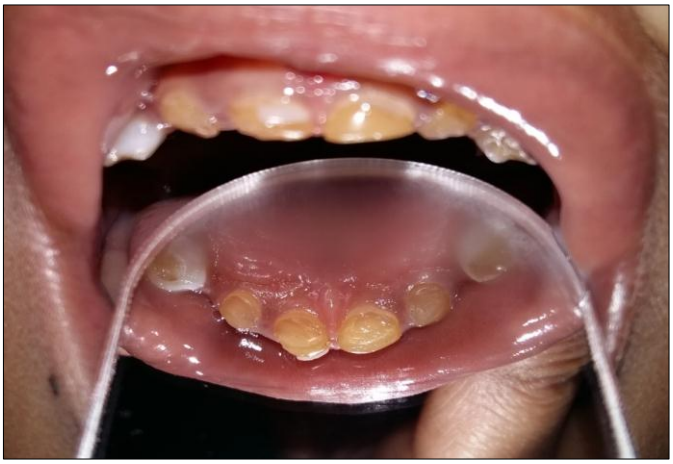

Fig 1: Clinical picture showing dentinogenesis imperfecta involving maxillary teeth

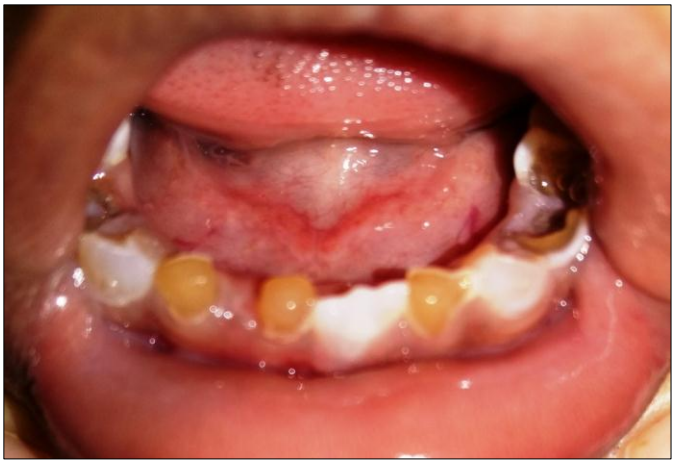

Fig 2: Clinical picture showing dentinogenesis imperfecta involving mandibular teeth

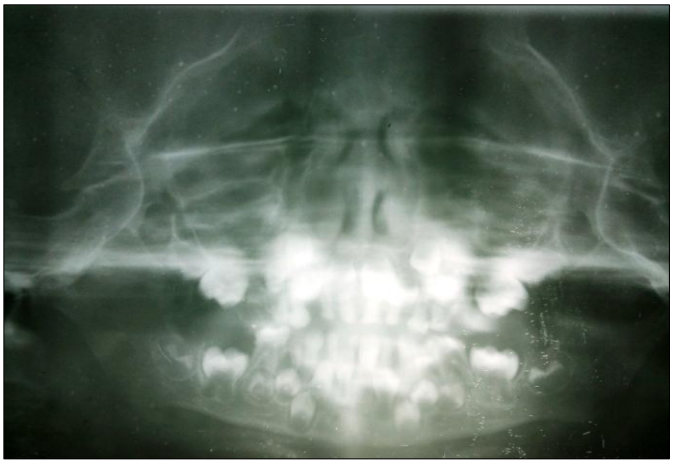

Fig 3: Panoramic radiograph revealing short and slender roots, constricted neck, prominent in posterior teeth, increased contrast between crown and root, and generalized obliteration of pulp canal. Radiographic features were characteristic and confirmed the clinical diagnosis of dentinogenesis imperfecta.

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