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Dentinogenesis Imperfecta Type I: From Opalescent Teeth to Obliterated Pulp Chambers—A Clinicoradiographic Insight

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Abstract

Dentinogenesis imperfecta (DI) is an autosomal dominant inherited disorder that occurs during the development of teeth at the time of histodifferentiation. DI causes structural defects in dentin formation, affecting both deciduous and permanent dentition.The incidence of DI is approximately 1 in 8,000. DI Type 1 is a disease that not only affects the dentition but also has a major impact on the routine activities of the patient due to its widespread involvement of bone, leading to long bone deformities (such as bowing of legs), early deafness, and a pathognomonic manifestation in the eye known as "blue sclera." Wormian bones seen on a lateral skull radiograph serve as a constant and confirmatory radiological finding of this "brittle bone disease." This paper highlights the correct diagnosis of DI Type 1 through comprehensive history taking, clinical and radiographic examination, and unstained ground sectioning (histopathology) of the involved teeth, followed by effective management and comprehensive dental care.

Keywords

Dentinogenesis imperfecta; Osteogenesis imperfecta ; Wormian bones; Developmental disorder.

Introduction

Human dentition displays extensive variation in tooth length, shape, number, and dental tissue structure.[1] Dentinogenesis imperfecta is an autosomal dominant developmental disorder. It is

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primarily linked to a mutation in the dentin sialophosphoprotein (DSPP) gene in isolated types, resulting in two of the three allelic DI types: DI Type II and DI Type III.[2] DSPP is expressed predominantly in odontoblasts, but is also found in salivary glands, kidneys, and lungs.[3]

DI was first defined in the late nineteenth century and can affect both dentitions as a localized mesodermal dysplasia.[4] When associated with Osteogenesis Imperfecta (OI)—commonly known as "brittle bone disease, brittle bone dysplasia, Fragilitas ossium, Vrolik disorder, or Glass baby sickness"—it is classified as DI Type 1.[5] OI affects connective tissue and is characterized by easily fracturing bones. While OI is classified into multiple types, the core collagen structure involves mutations in the COL1A1 and COL1A2 genes, which are also reflected in the manifestation of DI Type 1.[6] Enamel abnormalities and associated dental structural degradation frequently serve as critical diagnostic indicators for the underlying systemic syndrome.

Case Report

A 26-year-old male patient presented with a chief complaint requiring the replacement of missing teeth in the lower arch. History taking revealed a smoking habit of 9–12 cigarettes/day for 10–12 years. The patient reported a history of severe deformities in his lower extremities since childhood. Additionally, his brother was reported to have suffered from progressive hearing loss over the last few years. The patient stated that his teeth had been brownish since childhood and tended to fracture and chip off easily during daily activities. Consequently, he had undergone multiple teeth extractions at a private clinic. No other family members (siblings or parents) presented with a similar dental or skeletal complaint.

On physical examination, the patient was short-statured, presented with grey-to-blue sclera, and had visible lower limb deformities. Precise height and weight could not be measured standardly as the patient was unable to walk or stand independently**.** Intra-oral examination revealed marked generalized discoloration of the remaining dentition with extensive chipping of the enamel, imparting an opalescent amber/grey appearance. **(Figure 1)**



Figure 1: showing tooth discoloration with extensive enamel chipping and an opalescent amber-grey appearance.

All present teeth exhibited a heavily reduced occlusal height indicative of severe attrition. The following teeth were clinically missing: 22, 23, 24, 31, 32, 33, 34, 35, 36, 37, and 41–47. Following clinical evaluation, comprehensive radiological and histopathological investigations were ordered:

- Panoramic Radiograph: Revealed total obliteration of the pulp chambers and root canals in all remaining teeth. **(Figure 2)**



Figure 2: showing Total pulp chamber and root canal obliteration in all remaining teeth

- Full-Mouth Intra-Oral Periapical Radiographs: Confirmed generalized total pulp chamber obliteration across the surviving dentition**. (Figure 3)**

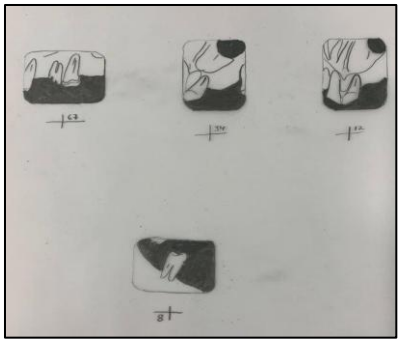


Figure 3: shows pulp chamber obliteration across all surviving teeth

- Lateral Skull Radiograph: Revealed the presence of multiple Wormian bones within the cranial sutures**.(Figure 4)**



Figure 4: Shows Multiple Wormian bones present within the cranial sutures.

- Histopathological Ground Section (Unstained): Ground section with respect to tooth 26 demonstrated highly irregular, disordered dentinal tubules and a completely obliterated pulp chamber space. **(Figure 5)**

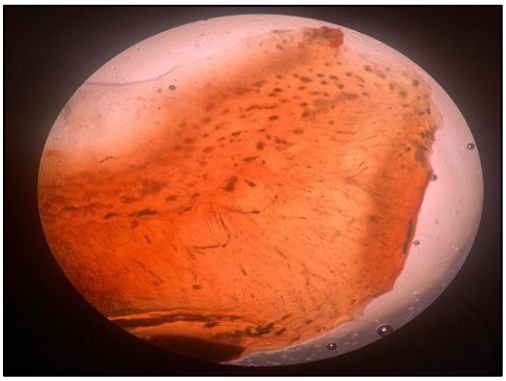


Figure 5: Reveals Irregular dentinal tubules and complete pulp chamber obliteration in tooth 26.

Based on a thorough correlation of family history, physical characteristics, clinical findings, pathognomonic radiographic features, and ground-section histopathology, a definitive diagnosis of Dentinogenesis Imperfecta Type 1 (associated with Osteogenesis Imperfecta) was established. As part of the comprehensive management plan, the remaining severely attrited and non-rehabilitable teeth (16, 15, and 26) were extracted. A comprehensive dental care approach using an overdenture was planned to restore esthetics, prevent loss of vertical dimension, and provide functional dentition**.(Figure 6)**

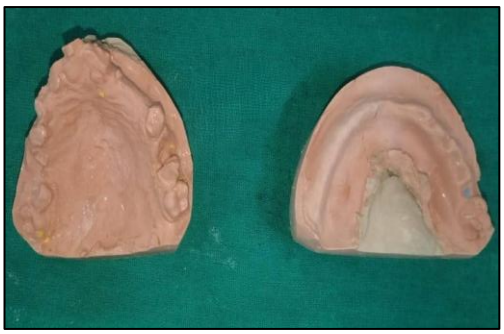


Figure 6: Overdenture planned to restore esthetics, preserve vertical dimension, and restore function

Discussion

Dentinogenesis imperfecta affects both primary and permanent dentitions through hereditary mesodermal dysplasia [7]. Dentin defect inheritance patterns can be autosomal dominant,

autosomal recessive, or X-linked. Syndromes exhibiting dentin defects include Osteogenesis Imperfecta (OI), Ehlers-Danlos syndrome, tumoral calcinosis, and hypophosphatemic rickets. Historically, DI was described first by Barrett in 1882 [8,9]. Statistical data indicates that approximately 50% of individuals with OI present with DI Type 1. This malformation occurs due to mutations in the COL1A1 and COL1A2 genes, which code for the pro-alpha 1 and pro-alpha 2 polypeptide chains of type 1 collagen. Mutations in these genes account for 80–90% of OI cases, primarily impacting bones and dentin. Conversely, isolated mutations in the DSPP gene located on chromosome 4q22.1 cause DGI Type II and Type III.[10]

Patients with Dentinogenesis Imperfecta Type 1 exhibit characteristic features categorized as follows:

- Extraoral Features: Bone fragility leading to frequent fractures, bowing of long bones (tibia and femur), growth deficiency resulting in short stature, blue/grey sclera that may become more pronounced with age, and early hearing loss caused by irregularities in the development of the labyrinth and temporal bone [5].
- Intraoral Features: Tooth color variations from opalescent grey/brown to yellow, tendency for Class III malocclusion, crossbites, ectopic eruption of permanent molars, and missing teeth (e.g., second premolars). Enamel cracks are common due to hypoplastic or hypocalcified defects; once the underlying soft dentin is exposed, it undergoes rapid, intense attrition. Periapical abscess formation and increased tooth mobility are also frequently appreciated [8].
- Radiographic Features: Teeth typically demonstrate short, blunted roots and bulbous crowns with normal enamel thickness but altered radiodensity, alongside total pulp chamber obliteration. Lateral skull films show pathognomonic "Wormian bones" within the cranial sutures [11]
- Histopathological Features: Characterized by highly irregular dentinal tubules that are larger in diameter but reduced in number, embedded within a large, uncalcified matrix [12].

The primary clinical objectives of comprehensive dental management in these patients are: to preserve dental health and maintain the strength, form, and duration of the dentition; to provide an esthetic appearance at an early age to prevent psychological and social issues; to offer a

functional dentition; to prevent the loss of vertical dimension; and to avoid interfering with the natural eruption of permanent teeth, thereby allowing normal growth of the facial bones and the temporomandibular joint. Differential diagnoses and their distinguishing clinical, radiographic, and/or histopathological features are summarized in **Table 1.**

S. No.	Condition / Differential Diagnosis	Differentiating Feature(s)
1	Amelogenesis imperfecta	Enamel is less radiodense than dentin.
2	Congenital erythropoietic porphyria	Yellow to green, brown, and grey to black discoloration localized primarily on the neck and coronal third of the teeth; associated with enamel hypoplasia.
3	Dentin dysplasia	"Thistle tube" shaped pulp chambers in single-rooted teeth.
4	Hypophosphatemia	Presence of aplasia or marked hypoplasia of cementum.
5	Regional odontodysplasia	Localized manifestation associated strictly with a single tooth or localized group of teeth ("ghost teeth").
6	Vitamin D-dependent rickets	Characterized by a distinct loss of the lamina dura.

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