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Dentinogenesis Imperfecta Associated with Osteogenesis Imperfecta in Children: phenotypic features and Dental Management

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ABSTRACT

Dentinogenesis Imperfecta (DI) is a hereditary dental condition commonly associated with Osteogenesis Imperfecta (OI), a genetic connective tissue disorder characterized by bone fragility and skeletal deformities. This article presents a case report of a child diagnosed with OI Type I and concurrent DI, highlighting the clinical, radiographic, and histological features that support the diagnosis. The patient exhibited classical signs of DI, including brown-grey opalescent teeth, generalized attrition, crossbite, and significant enamel chipping. Imaging revealed bulbous crowns, large pulp chambers, and a hypoplastic maxilla consistent with DI Type I. A comprehensive dental management plan involving preventive care, restorative treatment, and orthodontic planning was implemented to preserve oral function and aesthetics. This case reinforces the importance of recognizing dental anomalies as phenotypic indicators of systemic conditions and underscores the need for an interdisciplinary approach in managing children with OI and associated orodental manifestations.

1. INTRODUCTION

Osteogenesis Imperfecta (OI) is a rare genetic disorder characterized by increased bone fragility, skeletal deformities, and connective tissue anomalies. [1] It is primarily caused by mutations in the COL1A1 and COL1A2 genes, which encode type I collagen, a key structural component of bone and dentin. [2] Dentinogenesis Imperfecta (DI), seen in approximately 50% of OI cases, manifests as discolored, weak, and rapidly wearing teeth. This condition not only affects aesthetics and function but also plays a diagnostic role, particularly when skeletal findings are subtle.

Casereport:

A male child in middle childhood reported with discoloured and fractured teeth. His medical history revealed frequent fractures with a history of left femur fracture due to a fall from a bed, intramedullary rod insertion before 2 months, and a history of right radius fracture at the toddler age. The child was diagnosed with osteogenesis imperfecta type I and referred for dental management by his physician to evaluate orofacial aberrations related to osteogenesis imperfecta. This was the child's first dental visit. The parents revealed a personal history of second-degree consanguineous marriage with a history of opalescent teeth in the mother with no history of fractures. On Clinical examination, the child had abnormal physical development, short stature, demonstrated limited mobility, and abnormal gait. The extraoral examination revealed a mild blue-tinted hue in the sclera of the eyes (fig 1A). The patient's face appeared symmetrical with hypoplasia of the maxilla,

causing midface deficiency and reduced lower facial height. His facial profile appeared to be concave (fig1 B). Intraoral examination revealed brownish-grey opalescence of the upper and lower teeth with severe attrition in the mandibular anterior teeth. The left mandibular central incisor showed severe attrition and chipping of enamel(fig1C). Generalized crossbite was also observed (fig1D). An orthopantomogram (OPG) was advised for investigation, revealing tulip-shaped teeth with a bulbous coronal portion and prominent cervical constriction. The teeth appear shell-like with thin dentin, normal enamel deposition, absent pulp horns, and a large pulpal space. A rudimentary premolar is present in the left upper maxillary posterior region. Bilateral flattening of the condyle and shallow sigmoid notch were evident in the OPG (fig 2). The lateral cephalogram revealed lordosis of the neck spine, a hypoplastic maxilla, and a prognathic mandible, leading to skeletal class III (fig 3). Correlating the history, radiographic features, and histological features, Correlating the history, radiographic features, and histological features, a final diagnosis of dentinogenesis imperfecta associated with osteogenesis imperfecta was made.. Extensive treatment that focused on prevention and restoration was planned,which included fluoride application and pit and fissure sealants in posterior teeth and full coronal restoration of the lower anterior teeth to prevent further attrition.Staged orthodontic correction of the malocclusion as also advised. Anticipatory guidance was advised to prevent further trauma and occlusal deformation.

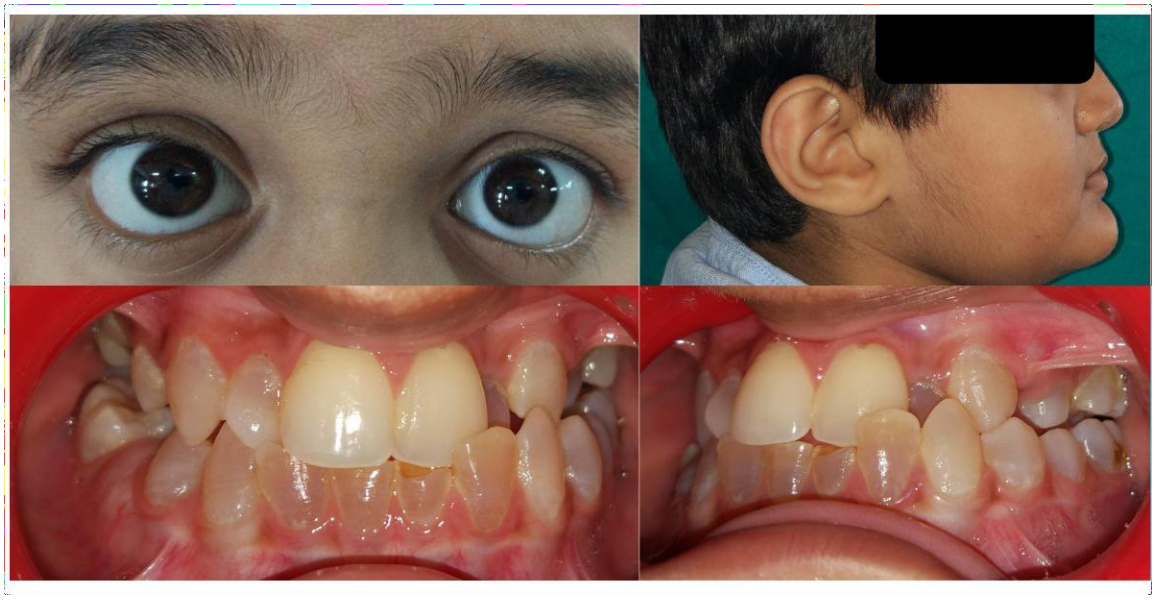

Figure 1 A:Bluish hue of the sclera.B:Lateral view of face depicting midface hypoplasia, prognathic mandible, reduced lower facial height.C:Teeth exhibiting brownish grey hue, with attrition in anterior teeth.D:bulbous tooth with generalised cross bite present.

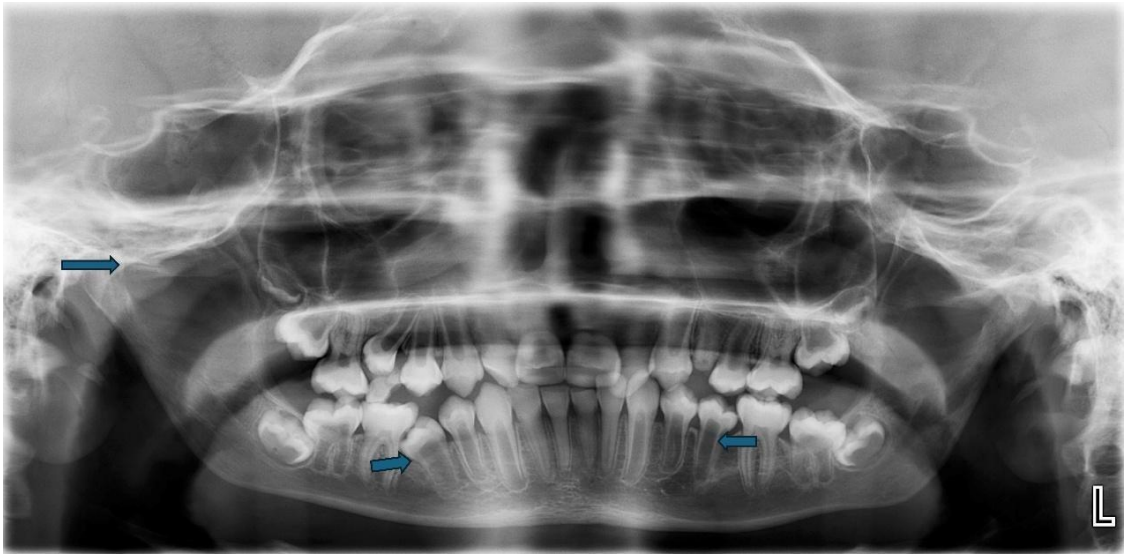

Figure 2:OPG reveals tulip like teeth with constriction in the CEJ, reduced dentin deposition,large pulp space, no pulp horns,wide open apex, agenesis of left maxillary first premolar, flat condyle head and shallow sigmoid notch.

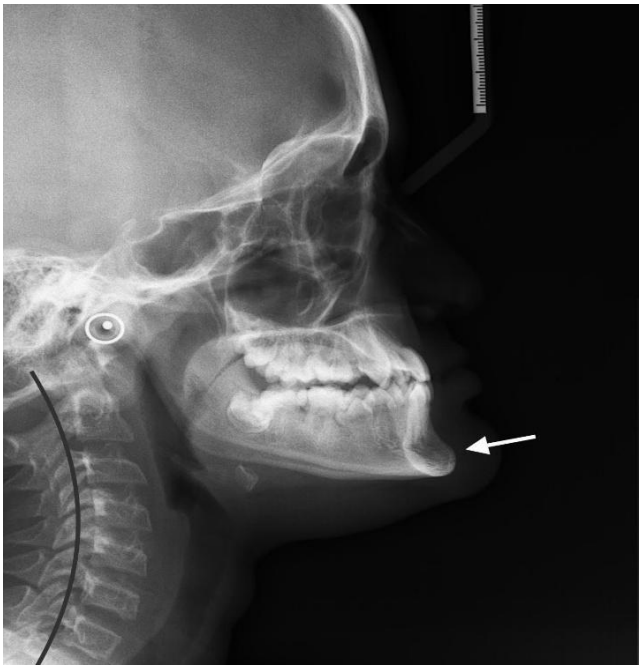

Figure 3:Lateral cephalogram showing lordosis of neck spine,mandibular prognathism.

2. DISCUSSION

Dentinogenesis Imperfecta (DI) in the context of Osteogenesis Imperfecta (OI) represents a significant clinical challenge due to its complex pathophysiology, aesthetic implications, and functional impairments. In children, DI typically becomes apparent with the eruption of the primary teeth and may persist or worsen with the eruption of permanent teeth. The teeth often appear discolored—ranging from yellowish-brown to bluish-grey—due to the translucency of the defective dentin beneath the enamel. These teeth are prone to rapid wear, attrition, and enamel chipping, particularly in areas subjected to occlusal forces.

The structural defect in dentin results from mutations in the genes encoding type I collagen, leading to reduced dentinogenesis and poor mineralization. As a consequence, teeth exhibit abnormal morphology such as bulbous crowns with cervical constriction, short or underdeveloped roots, and pulpal obliteration. These features are clearly visible on radiographs and are critical for confirming the diagnosis. [3] Histologically, the dentin appears dysplastic with an irregular tubular pattern, amorphous intertubular areas, and inclusions of cellular debris. [4] Additional studies have further characterized the dental phenotype in OI-related DI. Major et al. reported a high prevalence of DI among children with types III and IV OI and emphasized the correlation between DI severity and the degree of skeletal involvement. [5] Malmgren and Norgren highlighted the impact of DI on oral health-related quality of life and functional limitations such as chewing difficulty and hypersensitivity.[6] Moreover, studies by Renders et al. and Tolar et al. have investigated the effects of bisphosphonate therapy on dental and skeletal outcomes in OI, with some noting a possible reduction in fracture incidence but no significant impact on the expression of DI. [7][8]

One of the key challenges in managing children with DI and OI is balancing aesthetics with function while minimizing trauma to both the teeth and underlying bone. The fragility of bones in OI limits the extent of orthodontic or surgical interventions, requiring clinicians to take a conservative yet proactive approach. Moreover, as OI is a systemic condition, a multidisciplinary team involving pediatricians, geneticists, and orthopedic specialists is essential for comprehensive care. [9] In addition to dental complications, these children may experience delayed eruption, ectopic positioning of teeth, and increased incidence of dental anomalies such as congenitally missing teeth or taurodontism. Malocclusion, especially Class III due to maxillary hypoplasia and mandibular prognathism, is frequently observed and often necessitates long-term orthodontic planning. [10] Early diagnosis of DI, especially when associated with subtle skeletal signs of OI, can aid in the timely referral for genetic counseling and systemic evaluation. Furthermore, the psychological impact of dental disfigurement should not be underestimated. Dental rehabilitation plays a crucial role in improving the child's self-esteem and quality of life.Ultimately, long-term dental care for these patients involves regular monitoring, preventive strategies, and phased restorative or prosthodontic treatment to preserve tooth structure, restore function, and enhance aesthetics. Preventive measures such as regular dental check-ups, topical fluoride applications and other remineralising agents, dietary modifications, and the use of fissure sealants are crucial to limit caries and mechanical tooth damage. Restorative strategies include the use of full coronal restorations like stainless steel or composite crowns to protect teeth from attrition and enhance aesthetics. Glass ionomer cements and resin-modified materials are often preferred due to their better bonding properties

with defective dentin. Orthodontic correction of malocclusion, particularly Class III due to skeletal discrepancies, should be pursued cautiously, considering the underlying bone fragility. In severe cases, growth modification or orthognathic interventions may be reserved for post-pubertal stages. Prosthodontic management with space maintainers or partial dentures becomes necessary when there is premature tooth loss, while implant therapy may be considered in adulthood under specific conditions. Anticipatory guidance and caregiver education about trauma avoidance, oral hygiene maintenance, and long-term care expectations are vital components of successful management.

3. CONCLUSION

Dentinogenesis Imperfecta in children with Osteogenesis Imperfecta poses significant diagnostic and therapeutic challenges. Early identification, preventive strategies, and coordinated multidisciplinary care are vital to preserving oral health, function, and quality of life.

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