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Oral and Skeletal Challenges in a Pediatric Case of Osteogenesis Imperfecta with Dentinogenesis Imperfecta

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ABSTRACT

Osteogenesis Imperfecta (OI) is a rare inherited connective tissue disorder primarily characterized by increased bone fragility and low bone mass. It results from mutations in genes encoding type I collagen, most commonly *COL1A1* and *COL1A2*. The clinical presentation is highly variable, ranging from mild forms with few fractures to perinatal lethality.

Case Presentation

We report the case of a six-year-old child who presented to the dental department with complaint of discolored teeth. Clinical examination revealed opalescent, discolored teeth with severe enamel attrition. Radiographic imaging showed bulbous crowns, cervical constriction, and obliterated pulp chambers—features consistent with dentinogenesis imperfecta. Further medical history and orthopedic examination revealed a background of recurrent long bone fractures and blue sclerae. A multidisciplinary assessment, including genetic testing, confirmed a diagnosis of Osteogenesis Imperfecta Type1V.

Conclusion

This case highlights the importance of early recognition and genetic diagnosis of Osteogenesis Imperfecta for appropriate management. Individualized, multidisciplinary care is essential to improve quality of life and reduce fracture risk in affected patients.

Keywords

Osteogenesis Imperfecta, bone fragility, COL1A1, Dentinogenesis imperfecta.

INTRODUCTION

Osteogenesis imperfecta (OI), also known as brittle bone disease, is a genetically diverse connective tissue disorder characterized by fragile bones and frequent fractures.[1,2] The incidence of osteogenesis imperfecta (OI) ranges from 6 to 20 cases per 100,000 newborns, while its prevalence is estimated at 4 to 10 cases per 100,000 individuals.[3] It is caused by mutations in the COL1A1 and COL1A2 genes, which encode the two chains of type I collagen. Type I OI is typically associated with a reduced amount of collagen, while types II, III, and IV involve both structural abnormalities and decreased production of collagen.[4] According to Sillence, OI is classified based on clinical, genetically, and radiographic features in four groups.[5] A decrease in type I collagen synthesis leads to type I OI, whereas types II, III, and IV result from either quantitative or qualitative defects in type I collagen. As a consequence of impaired collagen production, all tissues rich in type I collagen can be affected. This may lead to various clinical manifestations, including dentinogenesis imperfecta (DI), blue sclerae, hearing loss, growth retardation, and joint hypermobility.[2,6,7] Type I OI is the mildest form and follows an autosomal dominant inheritance pattern. Affected individuals typically exhibit varying degrees of bone fragility, blue sclerae, and hearing impairments. Type II OI, which may be inherited in either an autosomal dominant or recessive manner, is the most severe form, marked by extreme bone fragility, blue sclerae, and often results in perinatal death. Type III OI, usually autosomal recessive, presents with moderate severity and is characterized by significant bone fragility, normal sclerae, and severe long bone deformities. Type IV OI can be inherited dominantly or recessively and also shows moderate severity. Patients commonly present with bone fragility, normal sclerae, bowing of long bones, and frequent vertebral fractures. Types V through VII are more recent additions to the classification system.[8]

Dentinogenesis imperfecta (DI) has been reported in over 50% of individuals with osteogenesis imperfecta (OI). DI is a hereditary dentin disorder, most commonly inherited in an autosomal dominant pattern.[8] While DI Type I is considered the dental manifestation of defective collagen production and is closely associated with OI, Types II and III are linked to mutations in the dentin sialophosphoprotein (DSPP) gene.[9] Both primary and permanent teeth are affected in DI Type I, with primary teeth typically showing more severe involvement. Clinically, DI is characterized by tooth discoloration—appearing gray, opalescent, yellow, or brown—and enamel that fractures easily. Radiographically, affected teeth often display bulbous crowns due to pronounced cervical constriction, short roots, and defective dentin, although the enamel usually appears normal in thickness and density. Microscopically, all three types of DI show similar alterations in dentin structure,

including normal mantle dentin, irregular circumpulpal dentin with distorted dentinal tubules, and areas lacking tubules altogether.[2] Patients with osteogenesis imperfecta commonly exhibit midface hypoplasia, Class III malocclusion, unilateral or bilateral crossbites, ectopic eruption of the first or second permanent molars, and congenitally missing second premolars. Despite these anomalies, their dental development generally corresponds to their chronological age.[7]

Due to the extensive and often severe skeletal and dental abnormalities associated with osteogenesis imperfecta (OI), providing dental care to affected individuals presents significant challenges for both the patient and the clinician. Awareness of the potential complications that may arise during dental treatment is essential for effective management. Therefore, the objective of this study is to present a clinical case in order to describe the features of OI, along with its oral and dental manifestations, from clinical, radiographic, and histopathological perspectives.

CASE PRESENTATION

A six-year-old female child was referred to the Department of Pediatric and Preventive Dentistry, University College of Medical Sciences, New Delhi with a chief complaint of discoloration in teeth. Clinical examination revealed bluish sclera [Figure 1], while a review of medical records indicated a history of multiple fractures since the age of one year, involving the left wrist, right wrist, right ankle, right humerus, and right elbow, each of which had healed within approximately one month [Figure 2]. No history of hyperextensibility of joints, head shape and size were normal, no growth developmental delay were noted. The patient's family history showed that her parents (mother, 27 and father 35 years old) were not related (non-consanguineous marriage). The patient's father is a known case of osteogenesis imperfecta with clinically positive bluish sclera[Figure 3]. Based on the patient's medical and radiographic results, the diagnosis of osteogenesis imperfecta Type 1V was confirmed, and the child is under medication (Syrup calcium and vitamin D3).

Intraoral examinations showed yellow/brown discoloration in all primary teeth, with severe attrition on the lingual and vertical cracks on the labial side of the upper and lower anterior teeth. Moreover, enamel disintegration was evident on the buccal surface of the canines [Figure 1]. Radiographic imaging showed bulbous crowns, cervical constriction, and obliterated pulp chambers—features consistent with dentinogenesis imperfecta (Type 1) [Figure 2]. Later, patient developed pain and sensitivity in her left upper first primary molar. Extraction of maxillary primary molar was done under local anesthesia with proper precautions. Preventive dental procedures, diet counselling and oral hygiene instructions were explained to the parent and the child to avoid further dental complications.

DISCUSSION

Osteogenesis imperfecta is a heritable connective tissue disorder primarily caused by mutations in the COL1A1 and COL1A2 genes, which encode the alpha chains of type I collagen-the predominant structural protein in bone, dentin, sclera, and ligaments.[10] Clinically, osteogenesis imperfecta is characterized by increased bone fragility, frequent fractures, skeletal deformities, blue sclerae, hearing loss, and growth deficiency.[11]

Fractures in patients with osteogenesis imperfecta are caused by a decreased strength of bone due to a decreased quality and quantity of bone matrix and architecture. The bone matrix has reduced strength and elasticity, resulting in lower energy absorption and increased susceptibility to micro-damage. This damage stimulates higher osteoclast and osteoblast activity, raising osteocyte density and bone porosity due to enlarged lacunae and increased vascularity. The combined effects of increased porosity and weakened bone matrix accelerate crack propagation, especially under repetitive stress. A rise in bone pore percentage from 4% to 20% leads to a threefold decrease in bone deformation before fracture, highlighting the bone's increased brittleness in osteogenesis imperfecta. In our case, the patient had sustained multiple fractures beginning at the age of one, involving the left wrist, right wrist, right ankle, right humerus, and right elbow. Notably, each fracture demonstrated a consistent healing period of approximately one month. Fracture management in osteogenesis imperfecta requires individualized care, with prompt emergency treatment and options ranging from observation to immobilization or surgical intervention. Patient and family education is essential, and flexible casting materials are preferred to minimize secondary fractures.[12,13]

Based on the medical history provided by the patient, the fractures were managed with immobilization using flexible plaster of Paris and splints to minimize stress risers, alongside analgesics and gentle handling for effective pain control. Physical therapy was provided to promote safe mobility and prevent joint contractures, while caregivers were trained in emergency fracture response and temporary splint application to improve outcomes and reduce treatment delays.

Dental abnormalities in individuals with osteogenesis imperfecta vary in severity, ranging from mild morphological changes to significant structural defects in dentin. A common dental manifestation in osteogenesis imperfecta patients is dentinogenesis imperfecta type I**,** which is uniquely associated with osteogenesis imperfecta especially in types I, III, and IV. Dentinogenesis imperfecta is a genetic disorder affecting dentin development and is classified into three types (I, II, III), which are clinically and radiographically similar but differ genetically. While dentinogenesis imperfecta types II and III are inherited in an autosomal dominant pattern and linked to mutations in the *dentin sialophosphoprotein* (DSPP**)** gene, they occur independently of systemic conditions like osteogenesis imperfecta. In contrast, dentinogenesis imperfecta type I is the only form associated with osteogenesis imperfecta and reflects the broader connective tissue defects characteristic of the disorder.[14]

According to Okawa R et al, dental abnormalities are prevalent in osteogenesis imperfecta, with dentinogenesis imperfecta prevalence ranging from approximately 20 to 48%. Those with a more severe skeletal phenotype (osteogenesis imperfecta type III/IV) exhibited more dental abnormalities than those with a milder skeletal phenotype (type I).[15] The pathophysiology of dentinogenesis imperfecta in osteogenesis imperfecta stems from the same underlying collagen defect affecting bone, leading to defective dentin matrix formation, inadequate mineralization, and disruption of the dentin-pulp complex. Consequently, teeth in affected individuals

demonstrate a range of structural and aesthetic anomalies.[16] Clinically, all three types of dentinogenesis imperfecta share common features in both primary and permanent teeth, including a translucent, opalescent appearance with discoloration ranging from yellow-brown to gray.

Majorana et al. reported that yellow-brown discoloration is more commonly observed than gray discoloration in deciduous teeth affected by dentinogenesis imperfecta.[17]

Despite structurally normal enamel, it fractures easily due to inadequate support from abnormal dentin and the absence of the typical scalloped dentinoenamel junction. Tooth crowns show a tulip or bell shape due to marked cervical constriction, and roots are typically short and blunted. Caries susceptibility is not increased and may even be reduced due to rapid tooth wear and lack of contact points. Radiographically, types I and II show identical findings: short roots, bell-shaped crowns, and pulpal obliteration due to continued deposition of abnormal dentin. In contrast, type III features thin dentin and markedly enlarged pulp chambers and root canals, resembling "shell teeth." Microscopically, affected dentin has fewer but larger and irregular tubules, and the pulp is progressively replaced by irregular dentin. The enamel is normal in structure, but the dentinoenamel junction is smooth, lacking the typical scalloping. Treatment focuses on minimizing wear and enhancing the esthetic appearance of affected teeth.[17]

In this case, intraoral examination demonstrated a generalized yellow-brown discoloration affecting the primary dentition, indicative of underlying dentin abnormalities. The teeth showed severe lingual attrition, reflecting extensive enamel wear likely caused by the compromised structural integrity of the dentin. Additionally, vertical fissures were observed on the labial surfaces of the anterior teeth, suggesting enamel fragility and microfractures. Buccal enamel breakdown was particularly evident on the canines, further highlighting the enamel's susceptibility to chipping and erosion. These clinical features collectively align with a diagnosis of dentinogenesis imperfecta, characterized by defective dentin formation that leads to enamel chipping, abnormal tooth coloration, and rapid dental wear.

The co-manifestation of osteogenesis imperfecta and dentinogenesis imperfecta necessitates a multidisciplinary approach for comprehensive patient management. Early diagnosis through clinical and radiographic evaluation, coupled with genetic testing, enables prompt intervention. Dental care should focus on preserving tooth structure, managing occlusal function, and addressing esthetic concerns. Treatment modalities may include stainless steel crowns in pediatric patients and composite resin restorations, or full-coverage crowns. Preventive strategies such as fluoride application and meticulous oral hygiene are also vital to mitigate secondary complications like caries and periodontal disease.[18]

Moreover, the psychosocial impact of visible dental abnormalities should not be underestimated, especially in young patients. Integrating psychological support and counselling into the management plan can significantly enhance patient quality of life. Advances in gene therapy and biomaterials hold promise for future therapeutic strategies that may address both skeletal and dental components of osteogenesis imperfecta.[19]

In **conclusion,** the presence of dentinogenesis imperfecta in individuals with osteogenesis imperfecta reflects the systemic involvement of connective tissue abnormalities and underscores the need for integrated medical and dental care**.** Continued research into the molecular mechanisms and clinical management of osteogenesis imperfecta and dentinogenesis imperfecta will contribute to improved outcomes and quality of life for affected individuals.

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FIGURE 1:

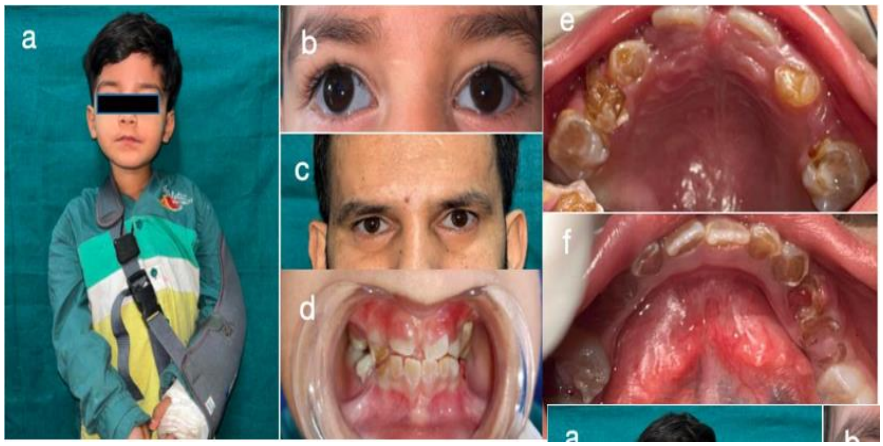
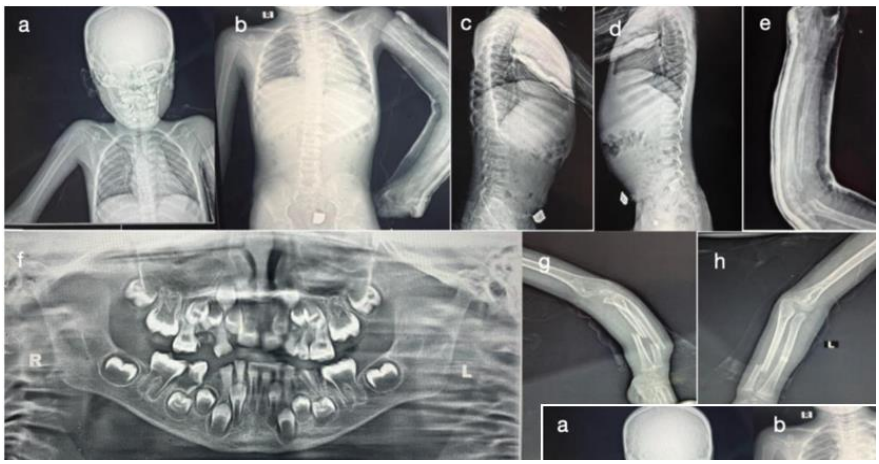


FIGURE 2:



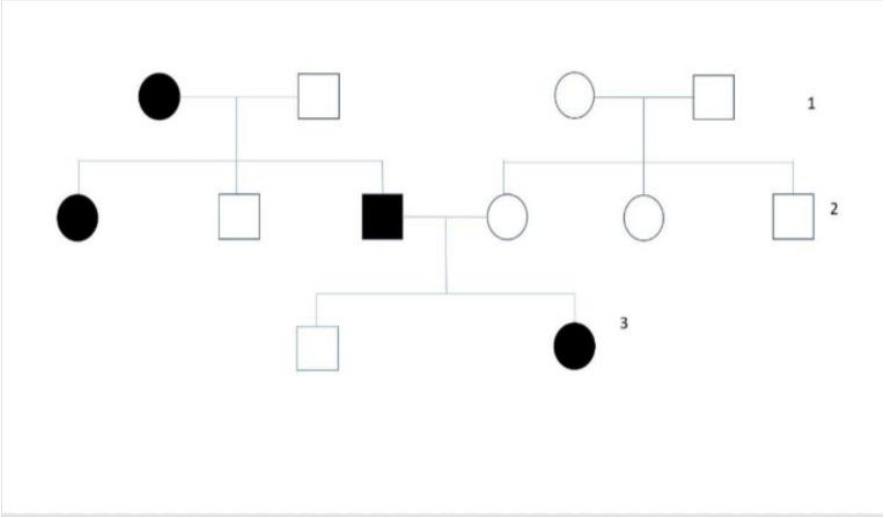


FIGURE LEGENDS:

FIGURE 1:

1a. Frontal profile image captured at initial presentation, revealing an active fracture involving the left forearm.

1b. Bluish hue of the scleral tissue observed upon clinical examination

1c. A positive family history of osteogenesis imperfecta was noted, with the patient's father being affected. The patient exhibited characteristic blue sclerae on clinical evaluation.

1d,e,f. Yellow-brown discoloration was observed across the entire primary dentition, accompanied by severe lingual attrition and vertical fissures along the labial surfaces of the upper and lower anterior teeth.

FIGURE 2:

a,b. Anteroposterior (AP) chest radiograph demonstrating a fracture involving the right shoulder joint

c,d. Thoracic spine radiograph (AP view) showed normal spinal alignment, with no signs of kyphosis or scoliosis.

e. Lateral view radiograph of the elbow demonstrates a fracture involving the right elbow joint

f. Panoramic radiographic evaluation (orthopantomogram) demonstrates bulbous tooth crowns, pronounced cervical constriction, and pulp chamber obliteration, findings consistent with dentinogenesis imperfecta.

g,h. Anteroposterior (AP) view of the elbow joint reveals a fracture involving the adjacent long bones.

FIGURE 3:

Pedigree chart illustrating the familial occurrence of osteogenesis imperfecta in 3 generations.