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Case Report

A Case Report on Osteogenesis Imperfecta

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Abstract: ***Background*:** Osteogenesis imperfecta (OI) is a rare heritable connective tissue disorder which is characterized by fragile bones and heterogeneous clinical presentation. There is mutation in genes of collagen Type I-COL1A1 and COL1A2 genes. ***Case Presentation*:** We report the case of a 16 year old male with a history of recurrent fractures since early infancy following the trivial trauma. The patient sustained multiple fractures involving long bones, associated with progressive deformities of the lower extremities. Notably, no etiological workup had been performed during childhood. There was no significant family history. On admission, physical examination revealed blue sclerae, dentinogenesis imperfecta, short stature, arachnodactyly, scoliosis and bowing deformity of bilateral lower limbs. Radiographic evaluation showed sequelae of previous fractures associated with bone deformities and cortical thinning. Based on the clinical presentation and radiological evaluation, a diagnosis of osteogenesis imperfecta was established. ***Conclusion*:** This case underscores the critical importance of early recognition of suggestive clinical features in children presenting with recurrent fractures. Improved clinician awareness may help reduce diagnostic delays, prevent complications, and enable timely genetic counselling and appropriate multidisciplinary management.

Keywords: Congenital Connective Tissue Disorder, Genetic Disorder, Osteogenesis Imperfecta.

INTRODUCTION

Osteogenesis imperfecta (OI) is a rare genetic connective tissue disorder resulting from abnormalities in type I collagen synthesis, most frequently caused by mutations in the COL1A1 and COL1A2 genes [1]. The disorder is marked by reduced bone mass and increased skeletal fragility, leading to recurrent fractures that may occur with minimal or no trauma.

The clinical presentation of OI varies widely, ranging from mild forms with limited skeletal involvement to severe variants associated with pronounced deformities and increased perinatal mortality. Common clinical features include blue sclera, dentinogenesis imperfecta, short stature, ligamentous laxity, and progressive bone deformities. In addition to skeletal manifestations, extra skeletal features such as hearing impairment and cardiovascular involvement, including valvular dysfunction and aortic root dilation, have also been documented [2-4].

In moderate forms, diagnosis is often delayed, particularly when characteristic clinical features go unrecognized. Early identification and a coordinated multidisciplinary approach are crucial in minimizing complications and improving functional outcomes. However, in resource-limited settings, the diagnosis may rely primarily on clinical and radiological findings in the absence of molecular confirmation.

We report the case of a 16 year old male with a clinical presentation highly suggestive of osteogenesis imperfecta, in whom the diagnosis was established late following recurrent low-energy fractures beginning in infancy.

CASE PRESENTATION

A 16-year-old male presented to Guru Gobind Singh Medical College and Hospital, Faridkot, with complaints of recurrent fractures since early infancy following trivial trauma and pain in the bilateral lower limbs for 1 day.

The patient was born at term via spontaneous vaginal delivery with no history of perinatal asphyxia or fetal distress. Birth weight and length were reportedly normal. Developmental milestones were delayed, particularly gross motor milestones, as the patient was unable to sit and walk independently within the expected age range. However, cognitive development was appropriate for age.

There was no family history of similar illness, and the parents were non-consanguineous and apparently healthy. The mother had a history of three intrauterine fetal deaths, the causes of which were not investigated. There was no history of long-term medication use, including corticosteroids or other drugs known to affect bone metabolism.

On physical examination, the patient was conscious and oriented to time, place and person. Classical stigmata of osteogenesis imperfecta were present, including blue sclera (Fig. 1) and dentinogenesis imperfecta, with multiple missing, discolored, and malformed teeth. A high-arched palate was also noted (Fig. 2).

Fig. 1: Blue sclera observed in the patient

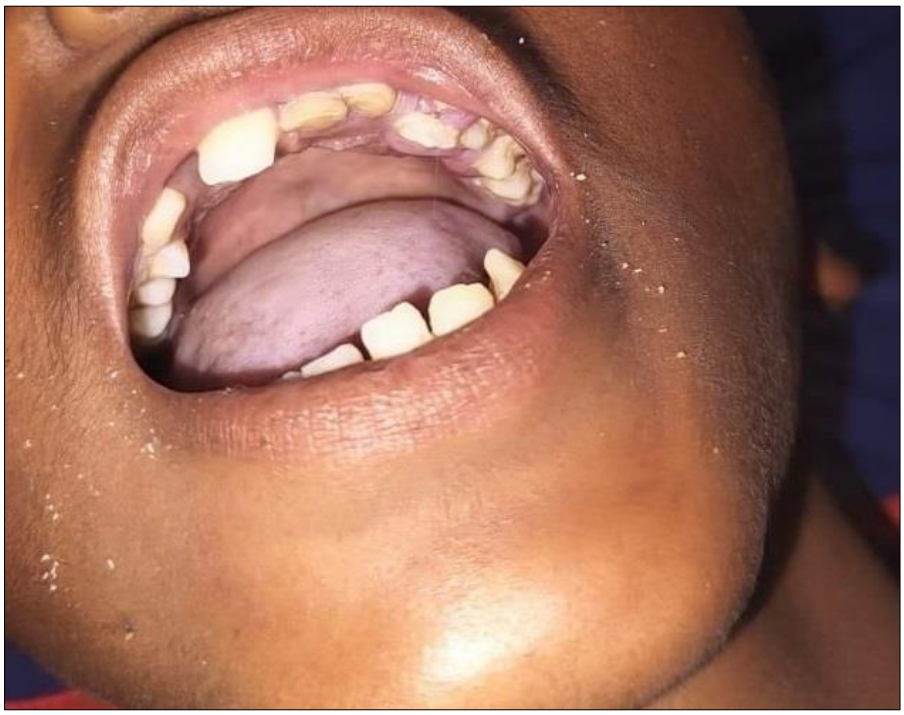

Fig. 2: Dentinogenesis Imperfecta with malformed teeth

There was marked ligamentous laxity with increased joint mobility. The patient had scoliosis, long and slender extremities (Fig. 3) with bowing deformities of both lower limbs (Fig. 4). Cardiovascular examination was unremarkable with no audible murmurs, and respiratory examination showed no abnormality. Audiometric assessment did not reveal hearing impairment, and neurological examination was within normal limits.

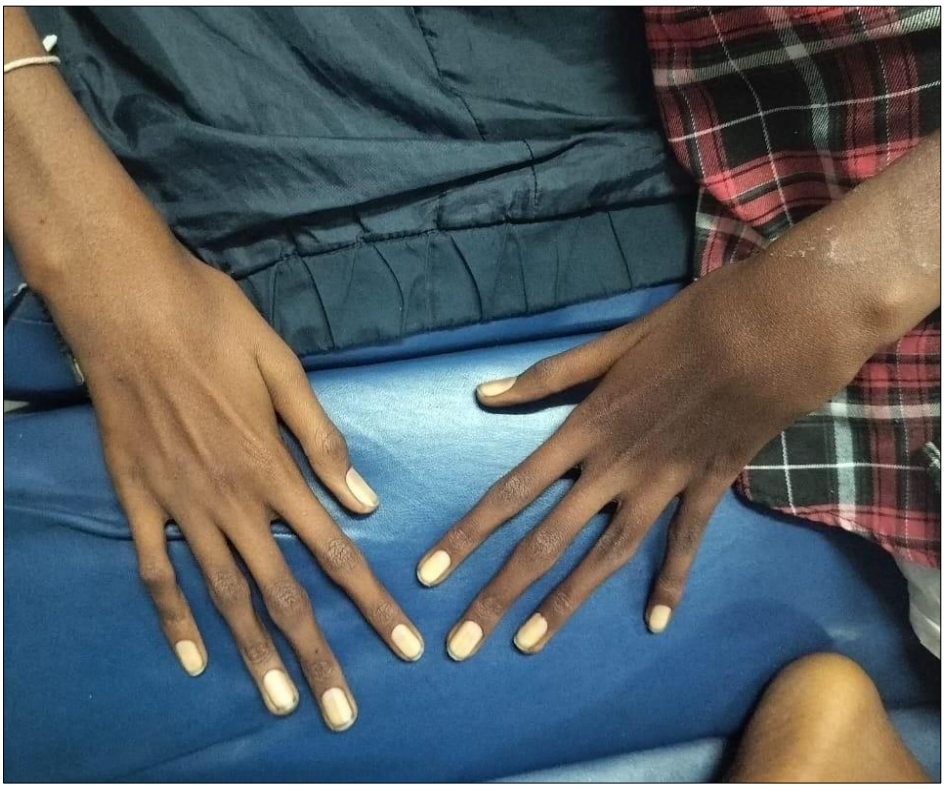

Fig. 3: Arachnodactyly with long, slender fingers

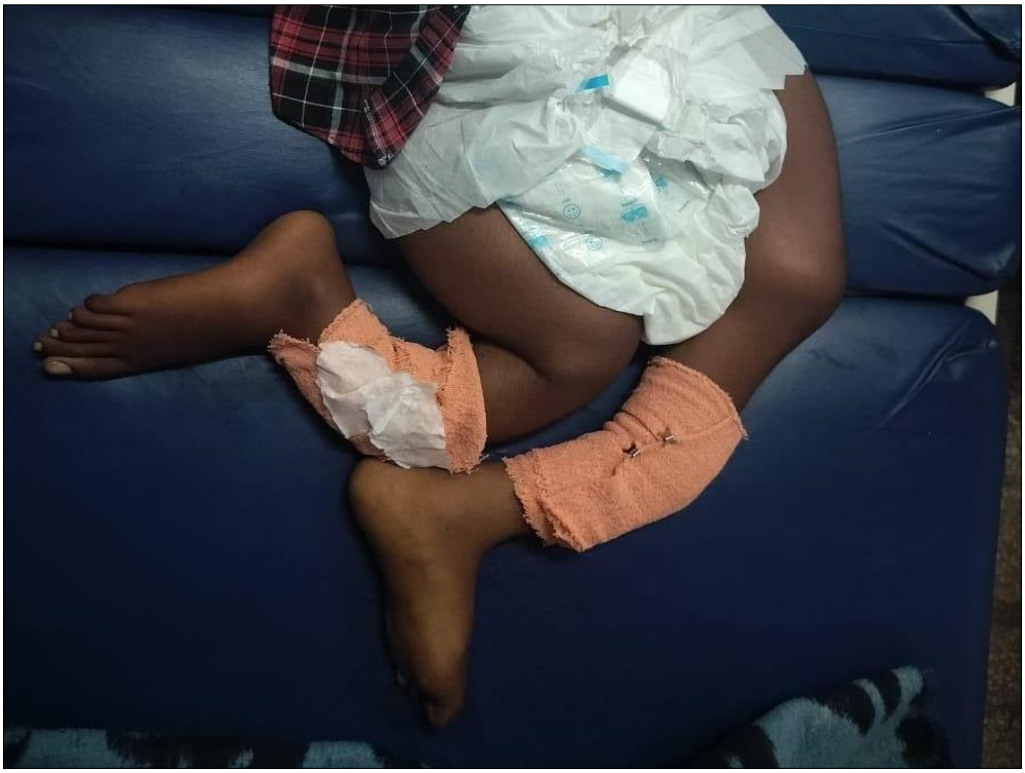

Fig. 4: Bilateral bowing deformity of the lower limbs

Radiographic evaluation showed sequelae of previous fractures associated with bone deformities and cortical thinning (Fig 5).

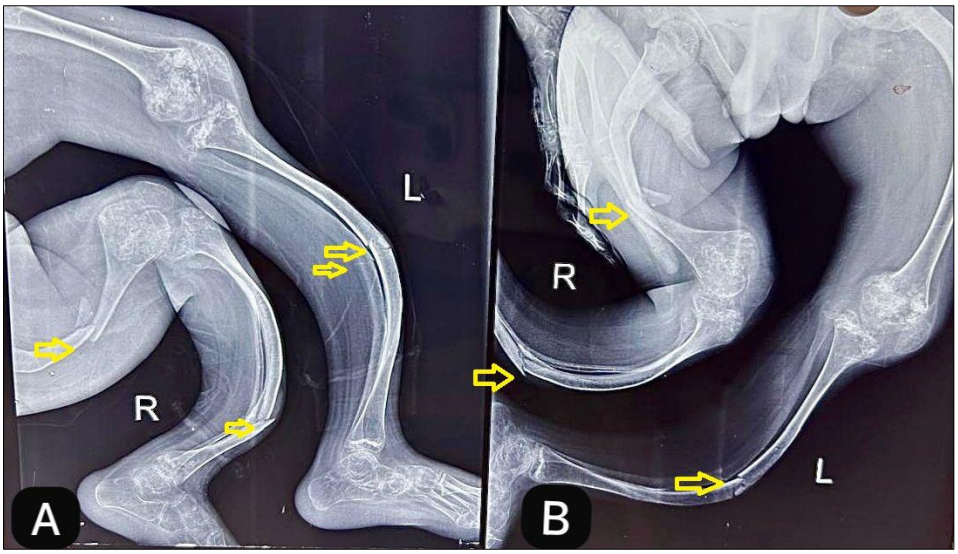

Fig. 5: Plain radiographs of bilateral lower limbs demonstrating severe skeletal deformities
(A) Lateral view of bilateral lower limbs showing diffuse osteopenia, cortical thinning, multiple healed fractures.
(B) Anteroposterior view of bilateral lower limbs showing diffuse osteopenia, cortical thinning, multiple healed fractures consistent with osteogenesis imperfecta. L: Left, R: Right

A comprehensive laboratory workup was performed which showed serum calcium of 83.9 mg/L with albumin at 42.7 g/L, phosphate at 34.4 mg/L, 25-hydroxyvitamin D at 24 ng/mL. Alkaline phosphatase was 120 IU/L. Renal function was preserved, with a creatinine level of 6.32mg/L (Table 1).

Tab 1:

Parameters	Patient Result	Reference Range
Serum Calcium	83.9 mg/L	85–105 mg/L
Albumin	42.7 g/L	32-45 g/L
Phosphate	34.4 mg/L	25-45 mg/L
25-hydroxyvitamin D	24 ng/mL	30-100 ng/mL
Alkaline phosphatase (ALP)	120 IU/L	40-150 UI/L
Urea	0.24 g/L	0.19-0.45 g/L
Creatinine	6.32 mg/L	7-12 mg/L

Based on the clinical presentation and characteristic radiological findings, a diagnosis of osteogenesis imperfecta was established, although no genetic testing was performed. The patient was managed conservatively with calcium (1,000 mg/day) and vitamin D (800 IU/day) supplementation, and physiotherapy.

DISCUSSION

Osteogenesis imperfecta (OI) is a rare genetic connective tissue disorder caused by abnormalities in type I collagen, an essential structural component of the bone extracellular matrix [1]. Approximately 85-90% of cases demonstrate autosomal dominant inheritance, most often linked to COL1A1 and COL1A2 mutations, which encode the alpha chains of type I collagen [2]. These defects result in decreased bone mass and increased skeletal fragility, predisposing affected individuals to recurrent fractures over time, often accompanied by progressive deformities and joint instability.

The clinical spectrum of OI is broad and may include features such as blue sclera, dentinogenesis imperfecta, short stature, progressive hearing loss, and cardiovascular involvement, including valvular abnormalities and aortic root dilation [3, 4]. Pulmonary complications are primarily related to thoracic skeletal deformities such as scoliosis and rib cage abnormalities, resulting in restrictive lung disease and reduced pulmonary function [5]. Our patient presented with blue sclerae, dentinogenesis imperfecta, short stature, arachnodactyly, scoliosis and bowing deformity of bilateral lower limbs.

Disease severity ranges from mild forms with minimal symptoms to severe variants presenting in infancy with multiple fractures, skeletal deformities, and high perinatal mortality [6]. Our patient experienced his first fracture in early infancy following the trivial trauma, with no significant birth history.

The classification of OI was initially proposed by Sillence and Danks, who described four major types based on clinical and genetic characteristics. Subsequent advances in genetic understanding have led to the identification of additional subtypes (types V-XI) with type V showing autosomal dominant inheritance and types VI-XI demonstrating predominantly autosomal recessive transmission, highlighting the heterogeneity of the disorder [6, 7]. The clinical phenotype was most consistent with a severe deforming form of OI, likely corresponding to Sillence type III.

The estimated incidence of OI is approximately 1 in 15,000-20,000 live births, with a prevalence of about 10 per 100,000 individuals, affecting both genders equally without racial predilection [8, 9].

Diagnosis is largely based on clinical and radiological findings. In selected cases, confirmation may be achieved through collagen analysis or molecular genetic testing. In our case, the diagnosis was established on the basis of clinical and radiological features. Genetic testing was not available in our setting due to financial constraints. Management requires a multidisciplinary approach, including orthopedic care, physiotherapy, rehabilitation, and pharmacological therapy. Bisphosphonates have demonstrated beneficial effects in improving bone mineral density and reducing fracture frequency [10].

Recent literature has further expanded the understanding of osteogenesis imperfecta, emphasizing its genetic diversity and evolving classification beyond the traditional Sillence system [11-12]. Despite these advancements, clinical diagnosis continues to play a central role, particularly in resource-limited settings where advanced diagnostic tools may not be readily available.

In patients presenting with recurrent fractures, several differential diagnoses must be considered. Conditions such as rickets, osteopetrosis, hypophosphatasia, fibrous dysplasia, and non-accidental injury can clinically resemble osteogenesis imperfecta. Rickets is associated with impaired mineralization and characteristic biochemical abnormalities, whereas osteopetrosis is characterized by increased bone density and may be associated with bone marrow failure [13]. Hypophosphatasia presents with defective mineralization and low serum alkaline phosphatase levels [14]. Fibrous dysplasia typically manifests as localized bone lesions rather than generalized skeletal fragility [15]. Non-accidental injury should also be carefully excluded, particularly in pediatric populations, by correlating clinical findings with history and social context [16, 17].

Limitations

A major limitation of this case is the absence of genetic testing, which prevents molecular confirmation of the diagnosis. Thus, the diagnosis remains clinically based and cannot be considered definitive. Nevertheless, the combination of characteristic clinical features and typical radiological findings strongly supports a diagnosis of osteogenesis imperfecta.

CONCLUSIONS

Osteogenesis imperfecta is a clinically diverse connective tissue disorder characterized by increased bone fragility and variable systemic involvement. While clinical and radiological findings may strongly suggest the diagnosis, genetic testing provides molecular confirmation and aids classification when possible. Early identification is crucial for timely management and prevention of long-term complications. A multidisciplinary management strategy, including orthopedic care, nutritional support, and rehabilitation, remains essential.

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