



Strength Beyond Fragility: A Case of Suspected Osteogenesis Imperfecta in a Child

Introduction

Osteogenesis Imperfecta (OI) is a rare inherited connective tissue disorder characterized by bone fragility and recurrent fractures. The condition results from abnormalities in type I collagen, which is essential for bone strength and connective tissue integrity.

OI occurs in approximately 1 in 20,000 births worldwide and can vary from mild forms to severe life-threatening disease. One of the most recognizable clinical signs is blue sclera, which results from thinning of the scleral collagen allowing the underlying choroid to become visible.

This case report describes a 7-year-old girl with recurrent fractures and blue sclera, highlighting the importance of early recognition of Osteogenesis Imperfecta.

Case Presentation

Patient Information

A 7-year-old girl was referred by an orthopedic surgeon on 14 January 2026 for evaluation of blue sclera and a history of repeated bone fractures.

The patient recently sustained a right forearm injury 10 days prior, reportedly caused by minor accidental trauma while playing with her mother.

Past Medical History

The child had a history of multiple fractures including:

- Clavicle fracture at **2 years of age**
- Suspected **tibia fracture at 3 years**
- Previous **left forearm fracture**

There were no associated injuries during these events.

Additional History

Important negative findings included:

- No fever or respiratory symptoms
- No evidence suggesting non-accidental injury (child abuse)
- No developmental delay
- No history of prolonged medication use

Family history was negative for similar conditions.

The parents were non-consanguineous, and the patient was the eldest of three siblings, with the other children having no similar medical history.

Physical Examination

On examination:

Parameter	Findings
Weight	18.7 kg (5th–10th percentile)
Height	119 cm (25th–50th percentile)
General Condition	Fair
Temperature	Normal

Systemic examination showed:

- Blue sclera present
- Teeth normal without discoloration
- Normal hearing
- Normal cardiovascular examination
- Normal respiratory examination
- Abdomen: no abnormalities detected

Musculoskeletal examination revealed:

- Right forearm immobilized with POP slab
- No limb deformities
- No spinal deformities
- No joint hypermobility

Investigations

X-ray Findings

Radiography of the right forearm showed:

- Distal ulnar fracture
- Evidence of callus formation

The fracture had been stabilized with a POP slab by the orthopedic surgeon on 12 January 2026.

Differential Diagnosis

Blue sclera can be associated with several conditions including:

1. Genetic connective tissue disorders

- Osteogenesis Imperfecta
- Ehlers-Danlos Syndrome
- Marfan Syndrome
- Incontinentia Pigmenti

2. Iron deficiency anemia

3. Drug-induced causes

Certain medications may cause scleral discoloration including:

- Steroids
- Amiodarone
- Phenytoin
- Phenothiazines

4. Idiopathic or physiological causes

Given the recurrent fractures and blue sclera, Osteogenesis Imperfecta was considered the most likely diagnosis.

Provisional Diagnosis

Closed right ulnar fracture with suspected underlying Osteogenesis Imperfecta

Osteogenesis Imperfecta Overview

Osteogenesis Imperfecta is a genetic disorder affecting collagen production.

Mutations in the COL1A1 and COL1A2 genes lead to abnormal type I collagen, resulting in fragile bones and connective tissue abnormalities.

Classic Clinical Features

Common manifestations include:

- Recurrent fractures after minimal trauma
- Blue sclera
- Joint laxity
- Hearing impairment

Other possible features include:

- Short stature
- Bone deformities
- Scoliosis
- Dentinogenesis imperfecta
- Wormian bones in skull X-ray

Diagnostic Evaluation

Diagnosis is based on clinical findings and supportive investigations such as:

- Genetic testing (confirmatory)
- X-ray showing osteoporosis or multiple fractures
- Bone density assessment using DEXA scan

- Blood tests:
 - Calcium
 - Phosphorus
 - Alkaline phosphatase
 - Parathyroid hormone
- Skin biopsy for collagen analysis

Prenatal diagnosis may be possible in high-risk pregnancies using:

- Ultrasound
- Chorionic villus sampling
- Amniocentesis

Management of Osteogenesis Imperfecta

Management is multidisciplinary and mainly supportive.

Conservative Treatment

- Physiotherapy
- Rehabilitation programs
- Casting and bracing
- Orthopedic monitoring

Medical Therapy

Bisphosphonates such as:

- Pamidronate
- Alendronate

These medications help:

- Increase bone density
- Reduce fracture frequency
- Improve bone pain

Additional treatment includes:

- Calcium supplementation
- Vitamin D supplementation

Surgical Management

Orthopedic surgery may be required for severe bone deformities.

The most common procedure is intramedullary rodding, which stabilizes long bones and prevents recurrent fractures.

Emerging Therapies

Recent research has explored new treatment options including:

- Bone marrow mesenchymal stem cells
- Denosumab (Anti-RANKL antibody)
- Sclerostin inhibitors
- Recombinant human parathyroid hormone
- Anti-TGF- β therapy
- Gene therapy approaches

These therapies remain under investigation but show promising potential.

Prognosis

The prognosis of Osteogenesis Imperfecta depends on the severity of the disease.

- Type II: usually fatal in infancy
- Type III: severe disability with shortened life expectancy
- Types I, IV, V: generally compatible with normal lifespan

Early diagnosis and proper management can significantly improve quality of life.

Genetic Counseling

OI can be inherited in autosomal dominant or autosomal recessive patterns.

- Autosomal dominant inheritance: 50% risk of transmission
- Autosomal recessive inheritance: 25% recurrence risk

Genetic counseling is essential for affected families.

Conclusion

This case highlights the importance of recognizing **recurrent fractures and blue sclera as key clinical indicators of Osteogenesis Imperfecta.**

Early identification allows timely management, appropriate counseling, and prevention of complications. Clinicians should consider OI when evaluating children presenting with fractures after minimal trauma.