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Abstract

Femoral shaft fractures in patients with osteogenesis imperfecta (OI) and paraparesis due to meningomyelocele are rare and complex, requiring careful surgical and rehabilitative management. We report a 12-year-old male with OI, paraparesis, and multiple skeletal deformities who presented with a spontaneous femoral shaft fracture. Clinical and radiological evaluation revealed generalized osteopenia, multiple healed fractures, Wormian bones, and dentinogenesis imperfecta, confirming OI. The fracture was managed by open reduction and stabilization with double Titanium Elastic Nailing System (TENS) in a retrograde manner. Postoperative rehabilitation included early passive knee motion and bisphosphonate therapy. The outcome demonstrated satisfactory alignment, stability, and functional preservation. This case underscores the importance of individualized surgical intervention, multidisciplinary care, and the role of intramedullary fixation in achieving favorable outcomes in children with OI complicated by neurological comorbidities.

Keywords: Osteogenesis imperfecta, femoral shaft fracture, paraparesis, meningomyelocele, titanium elastic nailing

Introduction

Shaft of femur fractures in patients with *osteogenesis imperfecta* (OI) and *paraparesis* due to *meningomyelocele* represent a rare and complex orthopaedic challenge. These patients have inherently brittle bones from OI and compromised lower limb function and sensation from neural tube defects. Fractures may occur spontaneously or go unnoticed due to reduced pain perception. Deformities, poor bone stock, and altered biomechanics complicate both diagnosis and management. Treatment strategies must balance fracture stabilization with the need to preserve mobility and minimize complications like pressure sores and malunion, often requiring individualized surgical and rehabilitative approaches.

Case report

A 12-year-old male born of a non-consanguineous marriage presented with acute pain and swelling in the thigh without any history of obvious trauma. The patient is a known case of paraparesis secondary to meningomyelocele, for which he underwent surgery during infancy. He has been non-ambulatory since birth. The absence of trauma history, along with neurologic impairment, raises suspicion for a pathological fracture or underlying bone fragility, necessitating further evaluation.

On clinical examination, the patient exhibited dental malocclusion, blue sclerae, and delayed attainment of motor milestones. Notable musculoskeletal features included thoracolumbar scoliosis, anterolateral bowing of the long bones, and a broad, barrel-shaped chest. A surgical scar was present over the lumbosacral region, consistent with prior meningomyelocele repair. Craniofacial anomalies included an overhanging occiput and triangular facies. Additionally, the patient demonstrated generalized hypotonia, thin, translucent skin, and features suggestive of an underlying connective tissue disorder.

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Fig 1: Clinical pictures.

On skeletal survey, Transverse # shaft of femur left, Multiple fractures at other regions in various stages of healing, resulting in deformed bones, Generalised Osteopenia, Thin cortices, Absence of funneling of diaphyses of long bones, Popcorn calcifications at metaphyses and epiphyses, Wormian bones, Scoliosis, Dental malocclusion, dentogenesis imperfecta, Thinned calvarial bones, Elongated pedicles of vertebrae were found.

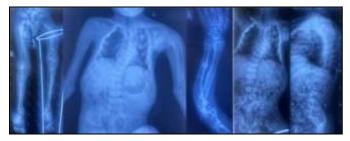


Fig 2: Skeletal survey.

Magnetic Resonance Imaging (MRI) of the brain demonstrated moderate hydrocephalus. Ultrasound of the whole abdomen and kidneys, ureters, and bladder (USG KUB), as well as Brainstem Evoked Response Audiometry (BERA), Visual Evoked Potentials (VEP), and fundoscopic examination, were all within normal limits. 2D Echocardiography showed situs solitus with levocardia. Bone Mineral Density (BMD) was significantly reduced, with a score of -4.2, indicative of osteoporosis. The diagnosis of osteogenesis imperfecta was established based on clinicoradiological grounds, along with a femur shaft fracture and paraparesis.

The patient underwent surgical management. Closed reduction of the femoral shaft fracture was attempted but failed; open reduction was done, followed by the insertion of a double intramedullary Titanium Elastic Nailing System (TENS), both in a retrograde manner for stabilization. Intraoperatively, hypertrophic callus was found. Postoperatively, the patient was initiated on regular passive knee range of motion exercises to maintain joint mobility and prevent stiffness using hinge knee brace, in accordance with standard rehabilitation protocols for non-ambulatory individuals with underlying bone fragility. Intravenous bisphosphonate is given after adequate prehydration and subsequent serum calcium monitoring.

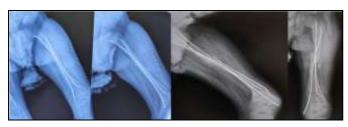


Fig 3: immediate post op and 7 weeks post op radiographs.

Discussion

Osteogenesis imperfecta (OI) is a heterogeneous group of

inherited disorders characterized by bone fragility, frequent fractures, and connective tissue manifestations. The presence of multiple fractures in various stages of healing, Wormian bones, and reduced bone mineral density in this case strongly supported the clinical diagnosis of OI, as per the criteria outlined by Sillence *et al.* [1]. The co-existence of meningomyelocele adds to the complexity, posing challenges in both diagnosis and postoperative care due to paraparesis and altered biomechanics.

Femoral shaft fractures in children with OI are often the result of minimal or no trauma and are best managed by individualized surgical intervention to provide stability and minimize complications. The use of intramedullary fixation has shown favorable outcomes in such cases, as noted by Gamble *et al.* [2], who reported reduced fracture recurrence and improved functional outcomes using flexible intramedullary nailing.

In this case, closed reduction followed by double Titanium Elastic Nailing System (TENS) insertion provided satisfactory alignment and stability. TENS is considered an effective option for long bone fractures in pediatric patients with OI due to its minimally invasive nature, dynamic fixation, and ability to accommodate growing bones [3]. Postoperative rehabilitation with passive knee range of motion exercises was initiated early to prevent joint stiffness, consistent with guidelines from Choi *et al.* [4].

While conservative approaches like bracing bisphosphonate therapy may reduce fracture risk in mild OI, surgical stabilization remains the cornerstone in moderate to severe cases. In a study by Esposito et al. [5] intramedullary rodding significantly improved mobility and reduced refracture rates in children with moderate-to-severe OI. Telescopic intramedullary rods are the gold standard in osteogenesis imperfecta as they grow with the bone, reduce revision surgeries, and have fewer complications like refractures and deformities. They offer longer implant survival and better biomechanical support compared to plates or nails. Newer designs like Fassier-Duval rods allow minimally invasive insertion, reducing tissue trauma.

In-situ TENS offers effective internal splintage for managing new fractures in osteogenesis imperfecta, minimizing the need for multiple surgical interventions⁶. Its design allows insertion without the use of fluoroscopy, reducing radiation exposure. The procedure is minimally invasive, technically simple, and can be performed even in resource-limited settings, making it a practical option for widespread use.

This case highlights the importance of a multidisciplinary approach tailored to the patient's functional status and skeletal pathology. With timely surgical intervention and structured rehabilitation, favorable outcomes can be achieved even in complex cases of OI with neurological comorbidities.

Conflict of Interest

Not available

Financial Support

Not available

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