CASE REPORT

# Multiple Bone Fracture in Neonates with Suspected Osteogenesis Imperfecta: A Case Report and Review of the Management

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## ABSTRACT

Osteogenesis imperfecta (OI) is a rare hereditary skeletal dysplasia characterized by bone fragility, deformity, and short stature. In this case report, we provide an overview of the management of a suspected OI case in a peripheral hospital. A suspected case of OI in a 1-month-old neonate boy presents with typical clinical conditions including reduced mobility, bowed legs, shortened limbs, blue sclera, and pectus excavatum chest. The radiological finding showed a spiral fracture in the right and left femur. This patient was initially diagnosed type 1 OI with differential diagnoses of rickets and idiopathic juvenile osteoporosis. The initial management for this patient is to reduce the fracture angulation with skin traction, and then bilateral spica cast was performed after the appropriate angulation angle. Multidisciplinary management is recommended for these cases, including pediatric, orthopedic, and rehabilitation management.
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## INTRODUCTION

Osteogenesis imperfecta (OI), sometimes referred to as brittle bone disease, is a rare genetic skeletal dysplasia characterized by bone fragility, skeletal abnormalities, and low stature. Furthermore, OI impacts the functionality of several other connective tissues, in addition to its impact on bone structure [1]. These are mostly hereditary disorder that result from structural or quantitative abnormalities in the COL1A1 or COL1A2 genes, which are responsible for encoding the α1(I) and α2(I) chains of collagen type 1, respectively. The Sillence classification is the predominant categorization system for OI. The mildest form of OI is type 1, which is characterized by the presence of clearly blue sclera. Type 2 OI is characterized by severe symptoms, resulting in death either during the prenatal or postpartum period. Type 3 OI have the most profound impact, characterized by the presence of blue sclera and a high frequency of fractures since birth, often resulting in significant limb deformities. Type 4 OI have sclera with a normal shade and experience bone fragility of intermediate degree [2].

## CASE REPORT

A 1-month-old neonate boy came with his mother to the Outpatient Clinic of the Department of Orthopedic Surgery of a Hospital in East Java, Indonesia, with a chief complaint of frequent fussing. The history of falling was present in the last two days before visiting the hospital, his mother slipped together while carrying the patient, but the mode of injury could not be described by his mother. A few hours after the incident, the right and left femurs became swollen and slightly hard when pressed, he began to fuss and cry continuously. He also developed a blue sclera, which was normal at birth but turned blue after a few weeks.

He was born by spontaneous delivery with 3600 grams of birth weight, without any complications. Immediately after birth, he cried spontaneously and strongly with a 9-10 APGAR score. According to antenatal history, the mother did not regularly visit antenatal care, with a history of visiting the doctor three times during pregnancy. He was breastfed from immediately after birth until the time of coming to the hospital. He was the first child in his family and there was no previous history in his father and/or mother's family.

Anthropometric measurements were recorded, including 4000 grams of body weight and 49 cm of stature which were -1 SD and -2 SD respectively based on the WHO Child Growth Standards interpretation. He presented with reduced mobility, blue sclera (Figure 1 a), pectus excavatum chest (Figure 1 b), bowed legs, and shortened limbs. The radiological finding showed a spiral fracture in the right and left mid-diaphyseal femur. In addition, the lower leg showed a bone healing process indicating the possibility of a previous fracture (Figure 2).

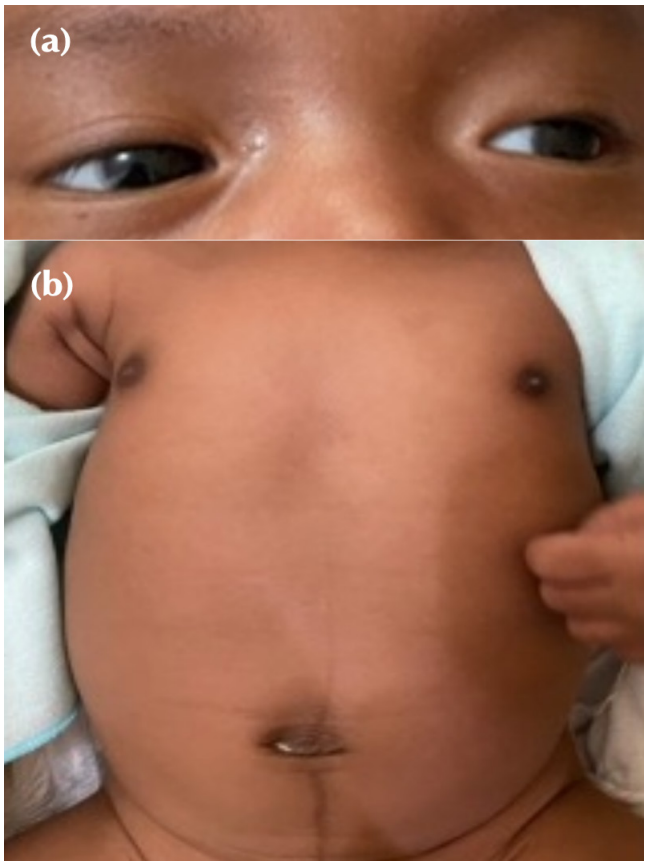


**Figure 1: (a) Blue sclera . (b) Pectus excavatum**

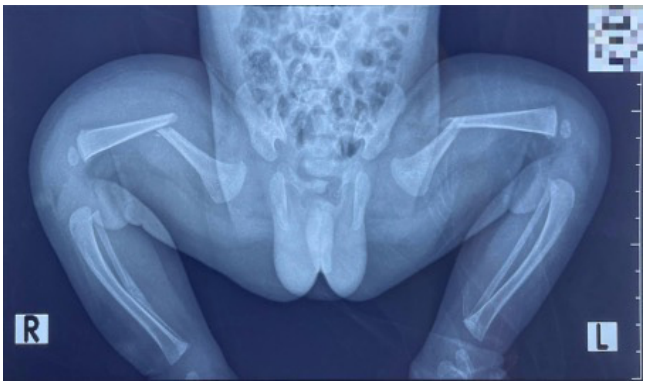


**Figure 2: Radiological findings showed multiple fractures in the right and left femur with spiral fracture type.**

The generally preferred investigation for OI diagnosing, including genetic DNA tests (COL1 A1 and A2), was not performed due to limited facilities in this hospital. For information, this hospital is a type B hospital (non-tertiary hospital) in one of the regions in East Java Province, which requires referral to a type A hospital (tertiary hospital) for certain diseases located in the capital city of East Java, Indonesia. Based on the clinical and radiological finding, the patient was diagnosed with suspected type 1 OI with differential diagnosis in this case, including rickets and idiopathic juvenile osteoporosis.

This patient planned symptomatic treatment including 0.5 kg skin traction or spica cast for seven days and then repeated radiological monitoring was taken to evaluate the angulation and shortening (Figure 3). A bilateral spica cast was performed (Figure 4).

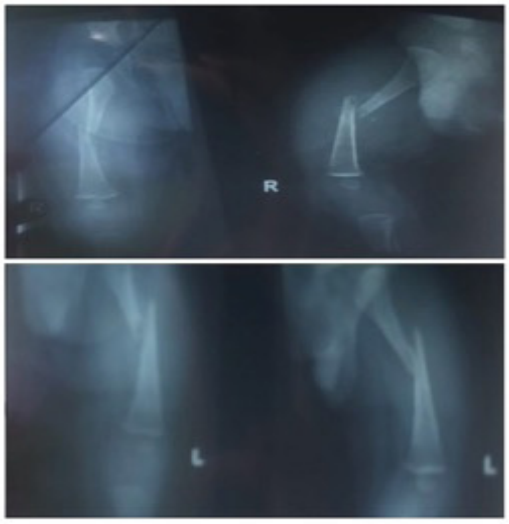


**Figure 3: Radiological evaluation on seventh days during spica cast installation**

**Figure 4: Bilateral spica cast.**

## DISCUSSION

The clinical diagnosis is typically established by evaluating skeletal manifestations, as shown in our patient. The OI is a prevalent factor in the development of bone fragility in youngsters. In addition to skeletal symptoms, this genetic disorder can also manifest with non-skeletal symptoms such as difficulty breathing, problems in the cardiovascular system, improper formation of tooth dentin, a bluish tint in the whites of the eyes, hearing impairment, and fragility of blood vessels. Genetic testing can provide definitive confirmation of the diagnosis, but molecular irregularities in collagen type 1 (COL1A1 and COL1A2) are only detected in 85% of cases. The occurrence of this disorder during the prenatal or postnatal period, as shown in our patient, is associated with severe manifestations and a significant risk of death. Antenatal ultrasound scans can detect these types by the identification of bone abnormalities, fractures, and bone shortening [3].

Providing a diagnosis in a non-tertiary hospital or a hospital with limited resources is a challenge. We only optimizing the existing facilities and explore a comprehensive history taking. From the history taking, physical examinations, and radiological finding this patient was diagnose as type 1 OI. Rickets and idiopathic juvenile osteoporosis are considered as another appropriate differential diagnosis due to the similar clinical symptoms. The most common type of fracture in OI is the transverse type, but it is possible that other types of fractures can also occur in certain conditions and cases. As in the case report by Matsukura and Takashima (2020) who reported a case of OI with a spiral fracture occurred in a boy due to a fall, this is similar to this case [4]. Type I collagen is the most abundant protein of bone and is also present in ligaments, tendons, dentin, sclera, and skin. Patients with OI have less or poorer quality (or both) type-I collagen than unaffected people, causing their bones to deform or fracture [3].

A multidisciplinary approach to this condition is indeed needed from pediatricians, orthopedics, and rehabilitation management. From the pediatrician's approach, pediatric care must consider how many calories and nutrients are needed to accelerate bone remodeling. Pharmacological therapies considered for OI are bisphosphonates [5], to reduce pain, increase bone mass density, and improve fracture healing [2].

From the orthopedic approach, specifically to improve the condition of the bones either by surgical treatment or other orthopedic therapy approaches. Follow-up requires orthopedic or surgical treatment of fractures and bone abnormalities [2]. While under anesthesia, bilateral spica casts or circular casts are surgically placed. Limit angulation to 15°, shortening to 2 cm, and central long bone fracture for spica cast installation. A spica cast immobilizes and preserves a body component till healing [1] . This patient had two closed fractures on the right and left femurs with a shortening of less than 3 cm but an angulation of more than 15°, so skin traction was placed for seven days before an evaluation showed that the angulation had decreased enough for bilateral spica cast.

From the physical medicine and rehabilitation approach, restoring normal function by minimizing disability is very necessary in this case. Rehabilitation plays an important role alongside surgical management and pharmacotherapy to maximizing mobility, function, and activities. Physical therapy includes aerobic exercise, leisure activities, and muscular building is essential for maximizing their function. The exercise program depends on age, function level, severity of OI, demands, and preferences [2].

Limitations of this case report are, first, we could not evaluate this case longer because the patient was lost to follow up. Second, specific examinations to confirm the diagnosis of OI could not be performed due to limited facilities in peripheral hospitals.

## CONCLUSION

Osteogenesis Imperfecta is a rare genetic disorder characterized by the presence of abnormally fragile bones. The patient was diagnosis with suspected to type I OI according to clinical and radiological findings. A comprehensive approach is necessary, involving the use of pharmacology, surgery, and rehabilitation management. The role of rehabilitation management is crucial in both the short- and long-term management of OI. Its objective is to optimize physiological function, enhance the patient's ability to perform everyday tasks independently, and enhance the patient's overall quality of life.

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