# Femur fracture management in a newborn with osteogenesis imperfecta: a case report


## ABSTRACT

**Aims:** This article aims to report a case of osteogenesis imperfecta from an orthopedic perspective, highlighting a study on the orthopedic management of a femur fracture. **Presentation of Case:** A five-day-old Brazilian male neonate of South American ethnicity was diagnosed with type III osteogenesis imperfecta. Imaging tests identified common clinical manifestations of this disease. Bryant's traction was applied to immobilize the right femur, Pavlick's brace was used, and pamidronate disodium was prescribed. One month after birth, the patient was discharged from the Intensive Care Unit with excellent outcomes. **Discussion:** Osteogenesis imperfecta (OI) is a rare genetic disorder characterized by bone fragility and low bone mineral density, resulting in recurrent fractures. OI is classified into five types based on severity. Clinical manifestations include skeletal weakening, blue sclera, and deafness. Diagnosis during intrauterine life remains challenging, while fractures during pregnancy are atypical. Management necessitates a multidisciplinary approach, incorporating orthoses, physiotherapy, and pharmacological interventions, which improves bone density but poses risks with prolonged use. Fracture treatment emphasizes minimal immobilization, physiotherapy, and tailored neonatal interventions such as Bryant traction. Patient and caregiver education is integral to optimizing outcomes in OI care.
**Conclusion:** This is a case report of a newborn with osteogenesis imperfecta (OI) who received treatment for a femur fracture. OI is a rare disease that can have a significant impact on affected patients. Therefore, it is essential to document treatment options and outcomes to contribute to the global database on this rare pathology.



*Keywords: Osteogenesis imperfecta; Orthopedics; Femur fracture; Lacunal skull.*


## 1. INTRODUCTION

Osteogenesis imperfecta (OI) is a rare genetic disease of connective tissue that affects collagen production, resulting in a quantitative or qualitative abnormality of this structural protein in bones, skin, teeth, tendons, and sclera (Deguchi *et al*, 2021). In 1979, David Sillence formulated the current classification of osteogenesis imperfecta. However, in this systematization, alterations in type I collagen were not predominant, so the main characteristic of the disease is bone fragility, which may be accompanied by other nonskeletal signs that help in the diagnosis of OI, which is predominantly clinical (Sillence *et al*, 1979).

This unique case has not been widely reported, given that OI is a rare disease with a low epidemiological incidence that has evolved well thus far. Because of its low frequency, there

has been a lack of specific studies on the treatment of osteogenesis imperfecta.In addition to bone fragility, which is demonstrated by a greater incidence of fractures and a decrease in bone mineral density (BMD), specific individuals also have bluish sclerae, dentinogenesis imperfecta, joint hyperextensibility, bone deformities in the skull and long bones, and morbidities throughout life that impair mobility, the respiratory system, cardiac alterations, severe scoliosis and other conditions that determine the classification of OI into five types, each with particular clinical manifestations (Brazil, 2022).

According to the Department of Informatics of the Unified Health System (DATASUS), 74 individuals were diagnosed with an anomaly or congenital disability of the osteogenesis imperfecta type in Brazil between 2019 and 2021 (Brazil-DATASUS, 2023). Thus, case reports addressing the treatment of bone consequences in neonatal patients are not widespread due to the low incidence of OI. Treatment is multidisciplinary and involves physiotherapy to improve quality of life, drugs from the bisphosphonate class to reduce bone resorption by osteoclasts, and orthopedic surgical procedures when necessary (Etich *et al*, 2020).

As a rare genetic alteration, osteogenesis imperfecta represents a significant clinical challenge. This case report aims to describe a case of osteogenesis imperfecta from an orthopedic perspective, highlighting the orthopedic management of the femur fracture present in the patient. The case in question refers to a patient diagnosed with type III OI, which is characterized by severe bone fragility, multiple fractures, deformities in long bones, and a bluish sclera at birth. By disseminating information about the following case, we hope to contribute to advancements in clinical practice and the debate on the most appropriate and effective orthopedic approaches for patients with OI.

## 2. PRESENTATION OF CASE

The patient was a five-day-old Brazilian male neonate of South American ethnicity with an Apgar score of 2/7/7 and a gestational age of 40 weeks. After prolonged and laborious natural labor, he did not cry at birth, and neonatal resuscitation maneuvers were necessary. On physical examination after birth, a brain hematoma was observed on the left, which led to a request for a computed tomography (CT) scan of the skull and a full radiograph. The CT scan of the skull showed shallow orbits, craniofacial disproportion, with a predominance of the skull and signs of maxillary hypoplasia, and a skullcap with multiple bone defects, predominantly in the parietal and occipital regions, with a trabeculated appearance, indicating a lacunar skull, also known by the German term Luckenschadel (Figure 1). Radiographs (Figure 2) revealed a proximal fracture of the right femur, bone callus, shortening of the left femur, and scoliosis of the spine on the right.

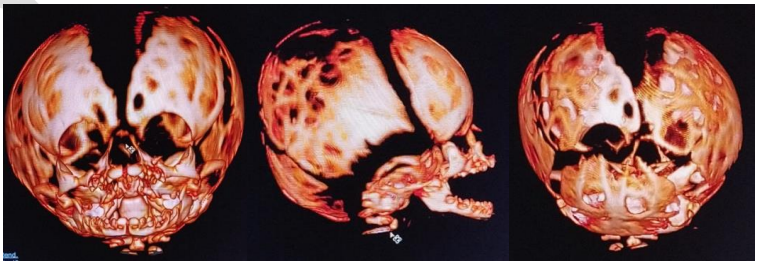

**Figure 1.** Computed tomography (Front, right side and back view) of the patient's skull, showing his trabeculated appearance.
**Source: Hospital records**

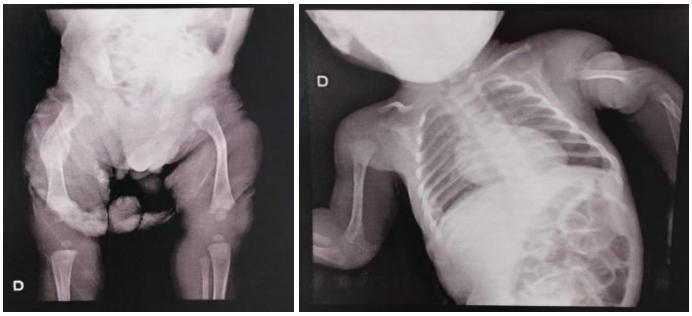

**Figure 2.** Radiograph of the chest and the pelvis, showing a proximal fracture of the right femur, bone callus and shortening of the left femur and scoliosis of the spine on the right.
**Source: Hospital records**

On physical examination and imaging, the orthopedic doctor noted severe bone fragility, multiple fractures, deformities in the long bones (varus thigh), a blue sclera at birth, and a triangular face with a frontal hump and short stature; all these findings are indicative and signs that confirm the diagnostic hypothesis for type III osteogenesis imperfecta. In addition, the orthopedic doctor immobilized the right femur with Bryant traction and concluded that the callus on the left femur was the result of an in-utero fracture. After a few days in the intensive care unit (ICU), the patient had a good recovery and continued treatment for the fracture. The orthopedic doctor advised the parents to buy and use the Pavlick harness, which was quickly implemented.

During the follow-up with the multidisciplinary team, it was observed that the patient was responsive to sound stimuli and physiotherapy. Before discharge, information was collected that the patient's father had delayed motor development.

The patient was released from the orthopedic ICU after using the Pavlick harness. As part of his treatment for Osteogenesis Imperfecta, he was prescribed pamidronate disodium, a drug belonging to the bisphosphonate group. In addition, he was referred to the Association of Parents and Friends of the Exceptional (APAE) for ongoing monitoring. The orthopedic service requested that he receive outpatient follow-up with pediatric orthopedics through the Unified Health System (SUS). Approximately one month after his birth, he was discharged from the hospital due to good progress in the treatment of his bone fractures, with normal serum calcium concentrations.

## 3. DISCUSSION

Osteogenesis imperfecta (OI) is a rare primary bone fragility disorder with a prevalence of 1/15,000 live births. Most cases are inherited autosomal dominant through mutations in the COLIA1 and COLIA2 genes, responsible for coding the type I collagen subunit, an essential protein in the extracellular matrix (Botor *et al,* 2021; Jovanovic & Marini, 2024). The resulting phenotype is characterized, according to the updated silence classification, into five different types (I to V), depending on the severity of bone fragility, and its main manifestations are characterized by low bone mineral density (BMD) and increased bone fragility resulting in multiple fractures from minor trauma (Koumakis *et al*, 2022; Botor *et al,* 2021).

In OI, the occurrence of fractures during pregnancy and in the postpartum period is atypical, and the determinants that influence them are not well known. Genetic testing can be carried out during prenatal care in high-risk and low-risk pregnancies, but it is challenging to diagnose OI during intrauterine life (Steiner & Basel *et al,* 2021). Individuals with this condition have a bone matrix with a lower absorption capacity due to its low strength and

elasticity, which implies that the bone is more susceptible to microdamage and, concomitantly, to increased osteoclastic and osteoblastic activity and bone porosity (Nijhuis *et al*, 2022).

The clinical manifestations are divided into two groups: the first concerns signs related to the weakening of the skeleton, and the second deals with manifestations resulting from the disturbance of collagen tissue in other organs, such as the blue sclera and deafness (Ishida *et al*, 2017).

Fractures occurring in patients with osteogenesis imperfecta (OI) may, in some instances, initially be mistaken for signs of abuse. However, upon closer evaluation, the hypothesis of abuse is readily dismissed, as bone density observed in radiographic examination and associated bone deformities, in conjunction with the patient's physical characteristics, unequivocally indicate a case of osteogenesis imperfecta (Etich *et al*, 2020; Ishida *et al*, 2017).

The heterogeneous presentation of the disease is another point that makes it difficult to approach and treat this condition specifically since treatment involves a multidisciplinary team (general practitioner, orthopedist, geneticist, rehabilitation medicine specialist, pediatric dentist, otorhinolaryngologist, psychiatrist and psychologist) (Hidalgo Perea & Green, 2021). Some potential medical approaches for OI include orthoses to stabilize loose joints, physical activity, physiotherapy to maximize bone stability, improve mobility, prevent contractures, and improve muscle strengthening, mobility devices, if necessary, and pain management (Steiner & Basel, 2021).

About the pharmacological treatment of OI, pamidronate, a second-generation bisphosphonate that inhibits bone resorption and induces osteoclast apoptosis, has been widely used in children and adolescents with this disease. This is because bone renewal is generally not a problem in children. In addition, there is an improvement in bone mineral density and a decrease in the porosity of the bone structure, which can reduce the number of fractures (Nijhuis *et al*, 2022; Nolin & Friedman, 2019). Therefore, the prescription of disodium pamidronate by the medical team in the case described is justifiable and can reduce the harm that tends to occur in patients with OI.

However, no definitive evidence supports its use after adolescence, and its long-term effects on the growing skeleton are uncertain. In the United States, for example, pamidronate is available only for parenteral administration, aimed primarily at treating hypercalcemia in patients with malignant neoplasms and Paget's disease and preventing bone loss in patients with breast cancer. In this sense, the risk-benefit ratio must be rigorously assessed, as bisphosphonates have been associated with serious adverse effects such as stress fractures in the lateral cortex of the femoral body (Nijhuis *et al*, 2022; Ishida *et al*, 2017; Nolin & Friedman, 2019). It is, therefore, essential that the pharmacological options for the patient in this report are periodically reassessed so that the risk of long-term use of this bisphosphonate does not outweigh its benefit.

Most fractures are treated with immobilization for as little as possible since the average healing time in these cases is shorter due to more excellent bone remodeling. Physiotherapy treatment after the removal of casts and intramedullary reinforcement rods is indicated in cases of OI (Sinikumpu *et al*, 2015).

Another standard treatment for hip fractures in neonates is suspenders, which can be categorized into static and dynamic; the most common is the Pavlik brace due to its ease of handling, the possibility of movement by the newborn, and its cost-benefit ratio. Using the

Pavlik harness is recommended as early as possible, with a minimum use of 6 to 12 weeks, as this influences the successful management of the fracture (Yoshiyasu & Cunha,1999). However, prolonged use can lead to avascular necrosis and paralysis of the femoral nerve, as well as treatment failure due to a lack of correct adherence (Merchant *et al*, 2021).

Additionally, another alternative for treating hip fractures is Spica plaster casting, which can be considered in appropriate cases, particularly when other treatments, such as the Pavlik harness, have proven unsuccessful (Bitar *et al,* 2016).

Patients with osteogenesis imperfecta (OI) are at an increased risk of fractures and deformities. Consequently, surgical treatment may be indicated, particularly through the use of telescoping intramedullary nailing, such as the Fassier-Duval system, to correct long bones. This approach can improve quality of life and stability while also reducing the frequency of severe fractures. (Hidalgo Perea & Green, 2021)

In the clinical scenario described, the application of traction, specifically Bryant traction, was primarily indicated due to its established efficacy in treating femur fractures in newborns. This method is recognized as a standard treatment because it aids in correcting femur deformities (Kim & Herring, 2021). Notably, in osteogenesis imperfecta (O.I.) cases, children may experience multiple femur fractures, potentially resulting in deformities (Koumakis *et al*, 2022; Bayram *et al*, 2018). Furthermore, the patient in question had a low birth weight and height and was in poor overall condition, discouraging the use of casting as a therapeutic modality. Additionally, using the Pavlik harness was not deemed feasible due to the small size of the child, which would complicate its application, as well as the challenging management of a patient hospitalized in the intensive care unit (ICU) and compromised general condition.

It is also worth noting that, regardless of the type of fracture and the therapy adopted, it is crucial to educate the patient, when possible, and family members about logistics, emergencies, transportation, and pain management (Steiner & Basel, 2021; Nijhuis *et al*, 2022).

## 4. CONCLUSION

This is a case report on type III OI in a newborn who presented classic clinical findings of this disease. For medical management, the first action was to immobilize the right femur with Bryant traction, which had a good effect, followed by adherence to the Pavlick brace and prescription of disodium pamidronate as part of his treatment for osteogenesis imperfecta. There are several potential therapies for this condition, ranging from the use of harnesses to surgical intervention. However, due to the lack of sufficient data in the literature on this subject, a significant gap remains regarding the specific aspects of treatment for this pathology. Therefore, it is essential to document treatment options and outcomes to contribute to the global database on this rare pathology.

## CONSENT

All authors declare that written informed consent was obtained from the patient for publication of this case report and accompanying images.

## ETHICAL APPROVAL

This case report was developed in a referral hospital through an interview with the orthopedic doctor in charge and analysis of the medical records. After talking to the orthopedic doctor in charge, we were told that this was a case of osteogenesis imperfecta type III in a newborn that occurred at a referral hospital. The case was carefully analyzed, and data were collected following the ethical standards established by the guidelines of Resolution No. 580 of 2018. Data collection was based on the CARE (Case Report) guidelines, and the following data were assessed: medical records, the report of the doctor in charge, and the results of computed tomography (CT) scans and radiographs.

The study was reviewed and approved by the Research Ethics Committee under the registration number CAAE: 70459223.4.0000.5076.
All authors hereby declare that all experiments have been examined and approved by the appropriate ethics committee and have therefore been performed in accordance with ethical standards.

## DISCLAIMER (USE OF ARTIFICIAL INTELLIGENCE)

The authors acknowledge the use of GPT-4 in the revision and editing of this manuscript. Its sole purpose was to enhance the grammatical quality of the translated text, originally written in Brazilian Portuguese, and to align it with academic writing standards. The AI's role was strictly limited to improving linguistic accuracy without adding or altering the content. The following outlines its involvement:

1. The manuscript was initially written in Brazilian Portuguese and subsequently translated into English. AI was employed to refine the translation, ensuring it adhered to academic English conventions.
2. Specific instructions were provided to guide the AI in correcting grammatical errors and ensuring the text maintained a high standard of academic language.

### DEFINITIONS, ACRONYMS, ABBREVIATIONS

**OI:** Osteogenesis imperfecta

**ICU:** Intensive Care Unit

**SUS:** Brazilian Unified Health System

**APAE:** Brazilian Association of Parents and Friends of the Exceptional

**BMD:** Bone mineral density

**DATASUS:** Department of Informatics of the Unified Health System

**CT:** Computed Tomography

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