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Ipsilateral femoral neck and shaft fractures in a woman with osteogenesis imperfecta

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ABSTRACT

Osteogenesis Imperfecta (OI), also known as Ekman-Lobstein syndrome, is a hereditary disease of the connective tissue that primarily affects the bones, leading to bone fragility and increased susceptibility to fractures. The disease can also affect other connective tissues such as ligaments, teeth, and sclera.

Thanks to advances in genetic research, several genes involved in osteogenesis imperfecta have been identified, improving our understanding of the disease's pathogenesis. In addition, new therapeutic approaches such as gene therapy and pharmacological treatments have been developed to increase bone density and improve the quality of life of patients with OI.

Key Words

Femoral neck fracture, Osteogenesis Imperfecta, Rib fracture.

Clinical Image

The patient is a 21-year-old woman who was admitted to the emergency room due to a fall from her height, resulting in a closed pelvic and left thigh trauma. She has a history of osteogenesis with multiple fractures due to low-intensity traumas in 2003 and 2007. The radiological examination reveals a bilateral femoral diaphysis fracture, previously treated with double telescopic nail fixation, as well as extra-osseous manifestations of the disease, including blue sclera associated with capillary weakness, and signs of aortic and respiratory insufficiency with a history of recurrent pneumonia.

On clinical examination, the patient is stable hemodynamically and respiratory, but complains of pain and total functional impairment of the left lower limb, with a twisted attitude in external rotation and shortening of the limb. The radiological examination shows a left femoral neck fracture associated with an iterative fracture of the homolateral femoral diaphysis on the telescopic nail (Figure 1).

The patient was operated on two days after the trauma. At first, the telescopic nail was removed. Then Open Reduction with Internal Fixation (ORIF) using plate Osteosynthesis was performed with a screwing of the femoral neck (Figure 2). The patient received antibiotic prophylaxis for 48 hours and thromboprophylaxis for 4 weeks. Rehabilitation was initiated the day after surgery with partial weight bearing after 9 weeks of surgery. Full weight-bearing was authorized 3 months after surgery. With a follow-up of 6 months, we obtained a consolidation of the fracture without notable complications.

Discussion

Osteogenesis Imperfecta (OI) is a hereditary condition characterized by brittle bones that result in multiple fractures following minor traumas. This rare disease does not show any particular ethnic or racial predisposition and affects approximately 1 in 15,000 to 20,000 births[1-6]. It is caused by an anomaly in the production of type I collagen, which can be quantitative or qualitative, leading to a heterogeneous group of diseases ranging from lethal forms at birth to milder forms discovered in adulthood.

Clinically, Osteogenesis Imperfecta (OI) is characterized by bone fragility, increased susceptibility to fractures, short stature, and abnormalities in bone structure, dental problems, and vascular fragility[7]. Bone fragility leading to multiple fractures is the main manifestation of osteogenesis imperfecta, and it can lead to increased morbidity and mortality due to complications such as respiratory failure

secondary to rib fracture[8]. The extra-skeletal manifestations of osteogenesis imperfecta include a wide variety of symptoms and complications, including hearing problems, dental issues, cardiovascular abnormalities, kidney problems, and bleeding disorders. The frequency and severity of these manifestations can vary greatly depending on the individual and the type of OI.

It is possible to diagnose osteogenesis imperfecta before birth through amniocentesis or chorionic villus sampling, which allows for fetal DNA analysis[9]. However, due to the genetic heterogeneity of the disease and the possibility of de novo mutations, accurate prenatal diagnosis remains difficult. Genetic counselling is recommended for affected families to assess the risk of recurrence and discuss treatment options and prevention of associated complications[10,11]. The diagnosis of osteogenesis imperfecta is based on a combination of clinical, radiological, and genetic information, and the most commonly used diagnostic criteria are those proposed by Silence et al. in 1979, which classify the disease into four types based on clinical and radiological presentation. However, due to the significant genetic heterogeneity of the disease, molecular diagnosis through DNA analysis can also be used to confirm the diagnosis and determine the type of OI. Targeted genetic tests can also be performed to search for specific mutations in genes associated with the disease.

The primary objective of treating osteogenesis imperfecta is to enhance the patient's quality of life and prevent disease-related complications. Available therapeutic modalities consist of bisphosphonate medication, which helps to increase bone density and reduce fracture risk, bone replacement therapy, such as recombinant bone protein injection, which aids in strengthening bones and preventing fractures[12], orthopedic surgery to correct bone deformities and prevent fractures, as well as mobility aids, including wheelchairs and crutches, to improve mobility and diminish the risk of falls[11]. Nonetheless, managing osteogenesis imperfecta is a challenging task due to the complexity of the disease and the variations in symptoms and severity[13].

Conclusion

Osteogenesis imperfecta is a rare genetic disease that affects the production of collagen, an essential protein for the structure and strength of bones. The treatment of osteogenesis imperfecta is mainly symptomatic and aims to prevent fractures, improve quality of life, and maintain functional autonomy. It is important to note that osteogenesis imperfecta is a chronic disease that can have a significant impact on patients' quality of life. Regular follow-up and multidisciplinary management are necessary to ensure optimal care and improve the prognosis of the disease.

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Figures

Figure 1: : Image showing an iterative fracture of the femoral shaft on a telescopic nail associated with a homolateral fracture of the femoral neck

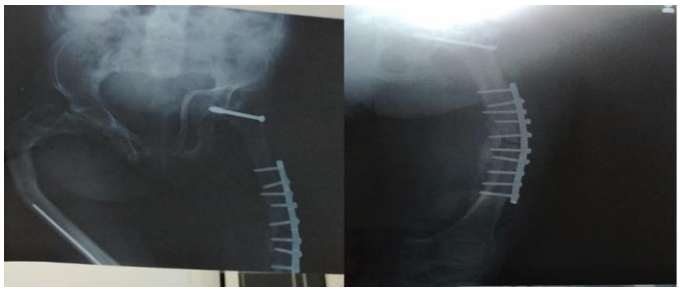

Figure 2: Postoperative radiography of the internal osteosynthesis.

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