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Clinical Image

Osteopetrosis Revealed by Pseudarthrosis of a Neglected Subtrochanteric Fracture in an Adult: Case Report

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Introduction

First described in 1904, Osteopetrosis (OP) is a rare genetic disorder [1] affecting osteoclast activity resulting in decreased bone resorption and bone densification [2]. There are three types of osteopetrosis depending on the world of transmission: autosomal dominant OP, autosomal recessive OP, and intermediate autosomal OP [3]. Indeed, osteopetrosis is characterized by a hard but fragile bone with a narrow or absent medullary canal [4]. Consequently, the bone is so dense and fragile that it is subject to an increased risk of fractures and complications during surgery, which is why patients with OP are treated conservatively [5]. Furthermore, due to poor blood supply, patients with osteopetrosis have a high risk of infection and osteomyelitis, in addition to the risk of pseudarthrosis [6].

Clinical Image

A 23-year-old patient consulted for chronic intermittent pain with limited mobility of the left hip and shortening of the left lower limb. On history, the patient reported a fall on the pelvis 2 years earlier, which caused acute pain in the left hip that later, became chronic and intermittent. The patient had not initially consulted. There was no history of anemia, fractures or recurrent infection. However, the patient reported a family history of bone disease without genetic evidence. The physical examination found a left lower limb shortened by 2cm; the mobility of the left hip was painless but limited, in particular abduction which was limited to 10°. We did not notice any pain on palpation or mobilization. Walking was possible but with a limp secondary to the shortening of the limb. The systemic examination did not reveal any other abnormalities. Biologically, there was no anemia, phosphocalcic abnormality, or alteration of renal function. Radiographically, there was a displaced left subtrochanteric fracture with an absent medullary canal and diffuse densification of the bone. The patient refused to undergo genetic testing. The diagnosis of late osteopetrosis with pseudarthrosis of a neglected subtrochanteric fracture was made. The patient underwent surgical management with open reduction and locked plate osteosynthesis.

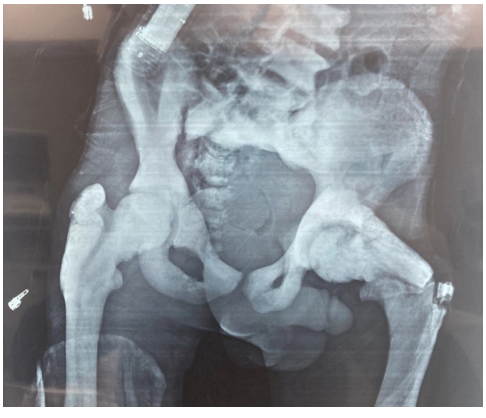

Figure 1:

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Discussion

Osteopetrosis is a rare hereditary disease that affects the process of bone remodeling by affecting the function of osteoclasts, resulting in decreased bone resorption and increased mineral density leading to hard bones and loss of the medullary canal [7-10]. Many patients with osteopetrosis are asymptomatic and diagnosed incidentally. This disease can also be revealed after a fracture [11]. In autosomal dominant OP, a fracture can occur in any bone but with a preponderance of the hip and trochanter region [12-16]. Fractures are often of simple transverse character following minor trauma [17].

The management of fractures in patients with osteopetrosis has been discussed in small case series [14]. The current literature suggests conservative management of some long bone fractures [18]. However, intertrochanteric and subtrochanteric fractures are at risk of pseudarthrosis and coxa vara [14,18,19]. These fractures should be treated surgically [18]. Several implants have been used in the reported case series, such as locked anatomic plates [11]. Surgical intervention requires wide availability of material. Moreover, during the osteosynthesis, a recurrent change of drill bits and cooling of the bone during drilling is necessary to avoid thermal bone necrosis [15]. Bhargava et al [15] report the occurrence of a subcapital fracture during tapping for dynamic hip screw fixation. In the case of pseudarthrosis, the absence of cancellous bone during OP limits its access to an autologous graft, which constitutes a particular difficulty.

Author Statements

Ethics Approval and Consent to Participate

Ethical approval was not sought. Written consent was obtained from the patients.

Availability of Data and Materials

The datadets used and analysed during the study are available from the corresponding author.

Declaration of Conflicting Interest

The authors declare that there is no conflict of interest.

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Authors Contributions

YB has conceived and developed the technique. YB has collected the data. All authors have read and approved the final manuscript.

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