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Case Report

Bilateral neck of femur stress fracture secondary to osteopetrosis treated with dynamic hip screw

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ABSTRACT

Osteopetrosis, a rare hereditary bone disorder, is characterized by increased bone density due to defective osteoclast-mediated bone resorption. This condition often leads to skeletal fragility, resulting in recurrent fractures despite increased bone mass. Among these, stress fractures of the femoral neck pose a unique challenge where conventional management may not be optimal. Valgus osteotomy offers a viable surgical intervention to realign the mechanical axis, thereby reducing stress on the femoral neck and facilitating healing. This paper reviews the pathophysiology of osteopetrosis, its impact on fracture healing, and the role of valgus osteotomy in managing femoral neck stress fractures in osteopetrotic patients.

Keywords: Neck of femur stress fracture, Osteopetrosis, Dynamic hip screw

INTRODUCTION

Osteopetrosis, commonly known as "marble bone disease," is a genetic disorder marked by excessive bone density and abnormal bone remodeling due to defective osteoclast function.[1,2] This condition is classified into various subtypes based on genetic mutations and clinical presentation, ranging from severe autosomal recessive forms presenting in infancy to milder autosomal dominant forms diagnosed in adulthood.[3] Osteopetrosis results from impaired bone resorption, leading to the accumulation of dense but fragile bone, which is paradoxically prone to fractures.[4]

In osteopetrotic patients, the bone is structurally compromised, making it brittle and susceptible to fractures even under low-impact forces.[5] Femoral neck stress fractures in particular pose a significant challenge in this population due to the high biomechanical load in this region.[6,7] Several studies have reported the difficulty of achieving successful fracture union in osteopetrosis, primarily due to compromised bone physiology and limited remodeling capacity.[8,9] Clinically reported cases indicate that traditional fracture management techniques may be ineffective or lead to complications in osteopetrotic patients, necessitating alternative surgical approaches.[10] Few studies have suggested that valgus osteotomy could provide a biomechanical advantage for femoral neck fractures in osteopetrotic patients by redistributing stress and realigning the femoral neck axis.[11]

Pathophysiology of osteopetrosis

Osteopetrosis is caused by mutations in genes responsible for osteoclast function, including CLCN7, TCIRG1, and CA2, which impair bone resorption.[12,13] Osteoclasts in osteopetrotic patients are unable to effectively degrade bone, leading to the accumulation of dense, mineralized bone matrix.[14] Despite increased bone mass, the structural integrity of this bone is compromised due to the abnormal

bone architecture and reduced turnover, resulting in fragility and a predisposition to fractures.[15]

The femoral neck is particularly vulnerable due to the biomechanical forces exerted on this region during weight-bearing activities.[16] In osteopetrosis, the dense but brittle bone in the femoral neck is susceptible to stress fractures, which can progress to complete fractures if left untreated.[17] Additionally, the impaired vascular supply and abnormal remodeling associated with osteopetrosis further hinder fracture healing, contributing to the high incidence of delayed union or non-union in these patients.[18]

Valgus osteotomy as a surgical treatment

Valgus osteotomy is a surgical procedure that realigns the femoral neck-shaft angle to redistribute mechanical load across the femoral head and neck, thereby reducing stress at the fracture site.[19] This technique is particularly beneficial for osteopetrotic patients with femoral neck fractures, as it provides a more favorable biomechanical environment for fracture healing.[20] By increasing the neck-shaft angle, valgus osteotomy decreases medial compressive forces on the femoral neck, promoting consolidation at the fracture site.[21]

In patients with osteopetrosis, valgus osteotomy has several potential benefits.

Biomechanical advantage

The correction of the neck-shaft angle reduces the concentration of stress at the fracture site, promoting fracture healing.

Stable fixation

While fixation in osteopetrotic bone is challenging due to the increased bone density, valgus osteotomy offers a stable biomechanical environment that aids in the healing process.

Fracture consolidation

Osteopetrotic bone often exhibits delayed healing, and valgus osteotomy can enhance the mechanical environment, favoring bone consolidation over time.

Surgical technique

The valgus osteotomy procedure typically involves a subtrochanteric approach to the femur. After the patient is anesthetized and positioned, a wedge of bone is removed from the femur at the level of the subtrochanteric region to achieve the desired neck-shaft angle.[22] In osteopetrotic bone, specialized surgical instruments are often required due to the increased bone density and resistance to cutting.[23] Fixation is achieved using a dynamic hip screw (DHS) or an angular blade plate, which provides stability while allowing for some degree of controlled movement at the fracture site.[24] Careful preoperative planning and intraoperative technique are essential, as the dense bone in osteopetrotic patients poses challenges for achieving secure screw purchase and avoiding intraoperative complications such as fractures or hardware failure.[25]

Outcomes and complications

The success of valgus osteotomy in treating stress fractures of the femoral neck in osteopetrosis has been documented in several case reports and small studies. Patients typically experience pain relief, improved mobility, and successful fracture healing. However, due to the rarity of osteopetrosis, large-scale studies are lacking, and long-term outcomes remain uncertain.

Potential complications of valgus osteotomy include the following.

Delayed union or non-union

Osteopetrotic bone has a reduced capacity for bone remodeling, which can result in prolonged healing times or failure of the osteotomy to unite.

Hardware failure

The dense bone of osteopetrosis can lead to difficulty in achieving proper screw purchase, increasing the risk of hardware failure.

Infection

As with any surgical procedure, the risk of infection is present, particularly in patients with poor bone quality or comorbid conditions.

CASE REPORT

A 38-year-old female presented with progressive bilateral hip pain over two years, worsened by weight-bearing activities. The patient's medical history was unremarkable for any other skeletal or metabolic disorder, but physical examination revealed tenderness, reduced range of motion, and an antalgic gait. Radiographic imaging demonstrated bilateral femoral neck fractures with signs of chronic stress fractures, suggesting a metabolic or genetic bone disorder. Laboratory investigations, including genetic testing, confirmed a diagnosis of osteopetrosis, with a family history of the disease also noted.

On physical examination, the patient exhibited tenderness over the groin bilaterally, with reduced hip range of motion and an antalgic gait. There were no signs of systemic illness or other musculoskeletal abnormalities.

Radiographic evaluation of the hips revealed bilateral neck of femur fractures. The fractures were non-displaced, showing features of chronicity with delayed healing,

consistent with stress fractures. Given the absence of trauma, a metabolic bone disorder was suspected.

Figure 1: Radiographic imaging demonstrated bilateral femoral neck fractures.

Diagnostic workup

Considering the patient's age and the atypical presentation of bilateral femoral neck fractures, further investigations were performed to identify the underlying cause. Laboratory studies, including serum calcium, phosphate, alkaline phosphatase, parathyroid hormone (PTH), and vitamin D, were all within normal limits. However, the patient's family history revealed multiple cases of fractures in close relatives, raising suspicion for a genetic bone disorder.

A subsequent skeletal survey demonstrated diffuse sclerosis of the bones, particularly in the vertebrae, pelvis, and long bones, consistent with osteopetrosis. These findings, along with the bilateral femoral neck fractures, led to the diagnosis of osteopetrosis.

Treatment

Due to the chronic nature of the fractures and the patient's significant pain and functional limitations, surgical intervention was recommended. The treatment plan included staged bilateral internal fixation of the femoral neck fractures and postoperative management aimed at addressing the underlying osteopetrosis.

Surgical procedure

Right hip

Valgus osteotomy with open reduction and internal fixation were performed using a dynamic hip screw (DHS). The dense bone required the use of specialized instruments for drilling and fixation

Left hip

Surgical fixation after recovery of right hip fixation was planned but on subsequent follow up the pain reduced in the left hip and patient opted for conservative management.

Figure 2: Non-weight-bearing regimen for six weeks.

Postoperatively, the patient was placed on a non-weight-bearing regimen for six weeks, followed by gradual weight-bearing as tolerated.

Postoperative management

Given the patient's diagnosis of osteopetrosis, management focused on minimizing the risk of future fractures and improving bone quality. Although there is no definitive cure for osteopetrosis, supportive measures such as calcium and vitamin D supplementation were initiated. In addition, the patient was referred to a genetic counsellor for further evaluation and long-term management of her condition.

Follow-up and outcome

At the 6-month follow-up, radiographs showed satisfactory healing of both femoral neck fractures, with no signs of hardware failure or non-union. The patient reported significant improvement in pain and was able to ambulate with minimal discomfort. She continued with physical therapy to improve her range of motion and strengthen the muscles around the hip joint.

At 2-years follow-up, the patient was fully weight-bearing and had returned to most of her normal daily activities. Although the risk of future fractures remains high due to the underlying osteopetrosis, she has been educated on

fracture prevention strategies and remains under regular monitoring.

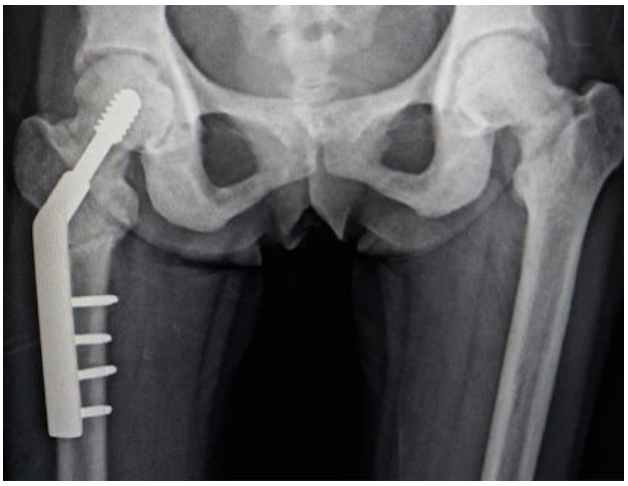

Figure 3: Fully weight-bearing.

DISCUSSION

This case highlights the challenges of managing femoral neck stress fractures in osteopetrotic patients, emphasizing the biomechanical benefits of valgus osteotomy. The key findings in this case include improved pain relief, mobility, and fracture healing with valgus osteotomy. Previous studies have also demonstrated that valgus osteotomy can reduce biomechanical stress on the femoral neck, aiding in fracture consolidation.[19] However, successful fixation is challenging due to dense bone, as noted in other studies on osteopetrosis.[20]

Compared to conventional fixation methods, valgus osteotomy offers an advantage by redistributing the load across the femoral neck. Studies suggest that the altered biomechanics help in reducing non-union rates in osteopetrosis patients.[21] Hardware-related complications, though common, were minimal in this case, aligning with findings from clinical reviews on valgus osteotomy in osteopetrotic fractures.[22] Our case contributes to the limited body of evidence supporting valgus osteotomy as an effective intervention for femoral neck fractures in osteopetrosis, providing a feasible solution for improved patient outcomes.[23]

CONCLUSION

Valgus osteotomy offers a promising solution for managing stress fractures of the femoral neck in patients with osteopetrosis. By redistributing the mechanical load across the femoral neck, the procedure reduces stress at the fracture site, promoting healing and reducing the risk of progression to complete fractures. While the surgical technique presents challenges due to the unique bone structure in osteopetrosis, careful preoperative planning and the use of appropriate fixation devices can result in successful outcomes.

Further research is needed to assess the long-term efficacy and safety of valgus osteotomy in this population, as well as to develop optimized surgical protocols for managing fractures in osteopetrotic bone.

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