

Osteopetrosis– An Experience of Open Reduction and Internal Fixation: A Case Report

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Abstract

We describe a patient with osteopetrosis who sustained multiple fractures including, right neck of femur fracture left subtrochanteric fracture and left forearm both bone fracture. All fractures were internally fixed. Our patient showed fracture union and successful return to normal activities of daily living. Operative treatment in osteopetrosis fractures are extremely difficult along with associated complications including infection and non union. Our experience shows internal fixation of peritrochanteric fracture with good outcome.

Keywords: Osteopetrosis, Internal fixation, Neck of femur fracture, Subtrochanteric fracture, Both bone fracture forearm.

Introduction

Osteopetrosis is a rare hereditary disease characterized by defective osteoclastic function resulting in high bone density [1]. In 1904, Albers-Schoenberg gave a description of hypersclerotic radiographic appearance of the bone in osteopetrosis. There are three clinical patterns based on the age of onset and inheritance pattern:-

1. Autosomal recessive malignant or infantile,
2. Autosomal recessive intermediate and
3. Autosomal dominant adult-onset type, the autosomal dominant type being the common among others.

In 50–60% of the autosomal recessive malignant osteopetrosis, patients have mutation in the TCIR1 gene on chromosome 11, which codes for the H⁺-adenosine triphosphatase proton-pump subunit of the osteoclast [2,3], whereas in autosomal- dominant osteopetrosis, patient mutation occurs in the chloride channel of the osteoclast, specifically in the CLCN7 gene on chromosome 16p13.3 [4]. Although bone contains increased number of osteoclasts, they lack the inherent

ability to activate macrophages and monocytes, as there is the absence of ruffled border and clear zone in the osteoclast, also fail to respond to PTH [5].

Even though sclerotic bones are very fragile and prone to fractures. Osteopetrosis is characterized by minor trauma-related fractures and typical radiographic findings like such as “bone within a bone” appearance and failure in bone tabulation [6]. With an increased risk of non-union and infection, internal fixation of fractures are is complicated in cases of osteopetrosis [1].

We report a case of osteopetrosis with bilateral peritrochanteric fracture and both bone fractures of forearm, all were internally fixed and the outcome of it.

Case Report

A 31-year-old lady female who is a known case of osteopetrosis, diagnosed first when she presented with a fracture of the right hip on 2015. She has a history of multiple fractures. She was presented with subtrochanteric fracture left femur and both bone fractures of the left forearm following fall. The patient has undergone dynamic hip screw fixation for fracture neck of femur right side and implant exit was done 2 years later when she developed pain over the right hip, she was advised partial weight-bearing and mobilisation following implant removal. Two months following



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Figure 1: The subtrochanteric fracture fixed with locking proximal plate, note the “pelvis in pelvis appearance.” DHS fixation was done for the right neck of femur fracture for this patient and removed 2 years later.

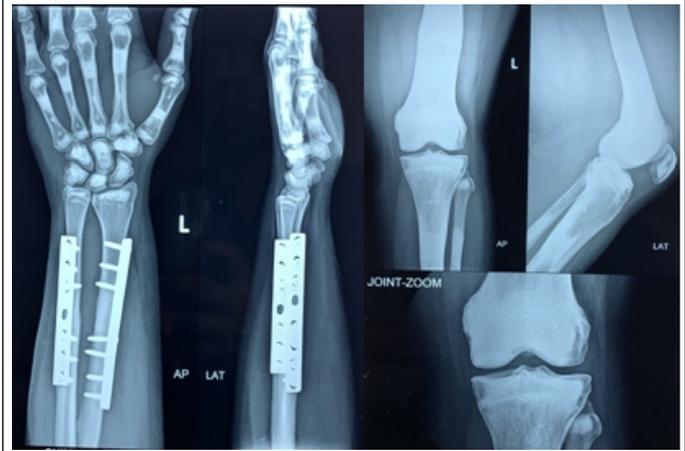


Figure 2: (a) Image of both bone fractures fixed with DCP shows union at the fracture site. (b) “Bone in bone” appearance of the knee joint.

implant exit, she had a history of fall and sustained subtrochanteric fracture of the left femur and both bone fractures of the left forearm in 2017. Subtrochanteric fracture was fixed with anatomical locking proximal femoral plate (fig. 1) and 3.5 DCP for both bone fractures. The patient was on further follow-up and nearly after one year of procedure, radiological union of the fracture was evident. Two years following the fixation, she has presented to us with pain over the left hip, on further evaluation found to have clinical and radiological union of the fracture, no signs of infection, or implant loosening. Pain was persisting with conservative management and metal exit done. She has relieved of pain and started walking. Following metal exit, the patient was advised non-weight-bearing for 4 weeks and was advised to start weight-bearing with support later on.

Discussion

Osteopetrotic patients are more prone to fractures, especially proximal femur fractures. The fracture healing mechanism is not different from normal bones here, but the time of union is usually longer than normal. The callus of mature osteopetrotic fractures has no Haversian organization and a scarcity of osteoclasts [7]. In osteopetrosis, there is a qualitative defect in bone metabolism characterised by defective osteoclastic bone resorption, leading to dense disorganized bone. The importance of RANK-L and RANK receptors in

osteoblasts and osteoclasts, respectively, are described in (Fig. 3). The normal physiology of RANK receptors in bone metabolism can be simplified like such as, PTH stimulating osteoblasts to produce the RANK-L which binds to specialized receptors in osteoclasts precursors and stimulates into the formation of active osteoclasts. The available literature are is more in the form of case reports, as for operative or non-operative management of fractures associated with osteopetrosis. Case report as early as 1923 by Alexander has shown pseudoarthrosis developed in a peritrochanteric fracture treated in a plaster cast. Likewise, many case reports of non-operative methods have shown to have union in 10–24 weeks duration [8].

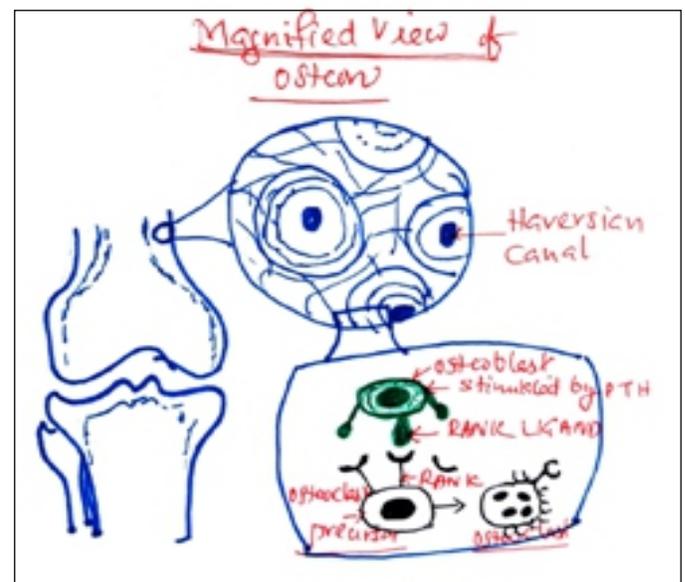


Figure 3: Pictorial representation of the molecular anatomy of bone. Animal studies where the absence of RANK-L gene resulted in osteopetrosis like condition. PTH:Parathyroid Hormone.

Internal fixations like such as interlocking nail, DHS, and anatomical locking plates are few of the options available for these types of fractures. The challenge to opt for internal fixation of osteopetrosis fractures are due to high density of the bone. Intra-medullary devices are used in proximal femur fractures associated with osteopetrosis, but it is more technically demanding and has a higher incidence of difficulty in opening intramedullary canal and drill breakage [9]. Other case reports by Aslan et al. used proximal femur plates, in which they have no complications and obtained union [10], which supports our case report. Infection and non-union are the main pitfalls of osteopetrotic fractures.

In our study, the patient had a previous neck of femur fracture of the right which was managed with dynamic hip screw and shown to have good results and functional outcome. Later on, she presented to us with subtrochanteric fracture of the left femur and both bone fractures of the left forearm, osteosynthesis with

proximal femoral locking plate was done for the subtrochanteric fracture and 3.5 dynamic compression plate for the forearm fracture. Fracture union was achieved in 12-months follow-up and with very good functional outcome. Animal studies in mice have shown that calcitriol and parathyroid hormone increased the function of osteoclasts in osteopetrosis. Clinical evidence also shown that high dose of calcitriol reduced the symptoms of osteopetrosis [11]. Our patient refused any type of medical augmentation of bone strength. Although internal fixation is quite technically demanding and with increased risk of nonunion and infection, we suggest internal fixation for peritrochanteric fractures associated with osteopetrosis.

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