Case study

Surgical management of subtrochanteric fracture with intramedullary nailing in osteopetrosis – A rare case report

Abstract

Background: Osteopetrosis, also called as "Osteosclerosis", "Marble bone disease" or "Albers-Schonberg disease, is an extremely rare inherited sclerotic bone disorder. The primary defect in osteopetrosis is due to mutation in CLCN-7 gene. Osteopetrosis is marked by increased bone density due to the defect in bone reabsorption by osteoclasts which leads to accumulation of bone with defective architecture, making them brittle and susceptible to fracture. **Case report:** We reported a 36 years old normotensive and non-diabetic female with type 2 adult type of osteopetrosis with subtrochanteric fracture of right femur and highlighted the surgical management with intramedullary interlocking nailing and technical difficulties encountered during the surgery. The classical features of osteopetrosis associated with this case and past history of left trochanteric fracture & its surgical management, iatrogenic fracture associated with surgical implant removal has been enlightened in this article to bring about the awareness among the readers. The patient has been explained about the natural history of disease and counselled for genetic screening to evaluate the mutant alleles. Due to lack of facilities, genetic testing could not be done. **Conclusion:** We recommend intramedullary interlocking nailing is the best surgical modality of choice for subtrochanteric fracture of femur in a case of osteopetrosis.

Keywords: Osteopetrosis; Sclerotic; Osteoclasts; Osteomyelitis; Intramedullary; AO cannulated screws

Introduction

Osteopetrosis, also called as "Osteosclerosis", "Marble bone disease" or "Albers-Schonberg disease, is an extremely rare inherited sclerotic bone disorder. The primary defect in osteopetrosis is due to mutation in CLCN-7 gene on chromosome 16q13.3[1]. Osteopetrosis is marked by increased bone density due to the defect in bone reabsorption by osteoclasts which leads to accumulation of bone with defective architecture, making them brittle and susceptible to fracture[2]. The primary manifestation of dysfunctional osteoclasts is either impaired acidification resulting in insufficient acid secretion and abnormal bone reabsorption or generation failure which involves the absence of RANKL on osteoblasts[3]. The resorption of calcified cartilage and primary trabecular weakens, thereby inhibiting secondary lamellar bone to replace the primary structure. Thus, bones fail to withstand the stress and reduplicative fracture exists in the course of osteopetrosis[4,5].

Here in this article, we have reported a rare case of type 2 adult type of osteopetrosis with subtrochanteric fracture of right femur and highlighted the surgical management and technical difficulties encountered during the surgery. The classical features of osteopetrosis associated with this case and past history of left trochanteric fracture & its surgical

management, iatrogenic fracture associated with surgical implant removal has been enlightened in this article to bring about the awareness among the readers.

Case report

A 36 years old normotensive and non-diabetic female patient came to Chigateri Government General Hospital attached to JJM Medical College with the chief complaints of pain over right hip and difficulty in walking following self fall at home from past 2 days. The pain was sudden in onset, non progressive, non radiating, aggravated on movements and partially relived by rest and medications.

The patient had given a past history of fracture around left hip 4 years back which was diagnosed as intertrochanteric fracture and operated with dynamic hip screw. After 1 year of surgical management, the patient was informed about non-union of fracture and the surgical implant removal was done.

While performing implant removal, stem of cortical screws were broken which were left inside the medullary canal of left femur. Then the patient's attenders were informed about the iatrogenic fracture of left neck of femur. After taking the consent, the iatrogenic fracture of left neck of femur were fixed with multiple AO cannulated screws (as shown in figure 1) and the patient was discharged. Within 2 months post op, the patient had good fracture union and started mobilization.

Figure 1 – Radiograph of left hip showing fracture neck of femur fixed with multiple AO cannulated screws and stems of cortical screw inside the medullary canal of left femur.

Then the patient had an episode of chronic osteomyelitis over left proximal femur(as shown in figure 2) after 6 months of second surgery for which the patient was treated conservatively with higher IV antibiotics for 6 weeks.

Figure 2 – Radiograph of left hip showing united fracture neck of femur fixed with multiple AO cannulated screws and stems of cortical screw inside the medullary canal of left femur with evidence of chronic osteomyelitis over left proximal femur

Then the patient has been counselled for multiple AO cannulated screw removal (as shown in figure 3). Following implant removal, the patient was pain free and functional range of movements over left hip was achieved.

Figure 3 – Radiograph of left hip with proximal femur showing the union of fracture of left neck of femur with no cortico-medullary differentiation over left proximal femur.

On examination, the patient was anemic without cyanosis and clubbing. Patient is unable to walk. Inspection of right hip showed swellingpresent over proximal one-third of

right thigh with no visible deformity, scars, sinus, dilated veins or visible pulsations. The patient had true shortening of 1 cm present over right lower limb. On palpation, local rise of temperature was present over proximal one-third of right thigh with crepitus over the fracture site. There were no transmission of movements at the right hip joint. Palpatory Bryant's triangle showed no proximal migration of right greater trochanter. Movements around right hip joint were painful and restricted. Measurements revealed 1 cm of shortening present over right thigh segment. The patient had no distal neurovascular deficit. Examination of right knee & ankle and left hip, knee & ankle were normal

The patient was subjected for further investigations

a) Hemogram
   - Hb – 11.3 gm/dL
   - Total count – 7310 cells/mm^3
   - RBC – 4.1 million/mm^3
   - Platelets – 2.01 lakh cells/mm^3
   - ESR – 8 mm/hour
   - CRP – 6 mg/L

b) Renal function tests – Urea 31 mg/dL and creatinine 1.6 mg/dL
c) Random blood glucose – 96 mg/dL
d) HIV and HbsAg – Non reactive
e) Serum calcium – 9.2 mg%
f) Serum phosphorus – 4.5 mg%
g) Serum alkaline phosphatase – 893 IU/L
h) Serum acid phosphatase –4.9 ng/mL
i) Serum Vitamin D3 – 30.3 ng/mL
j) Radiography of right hip with femur revealed subtrochanteric fracture (as shown in figure 4)

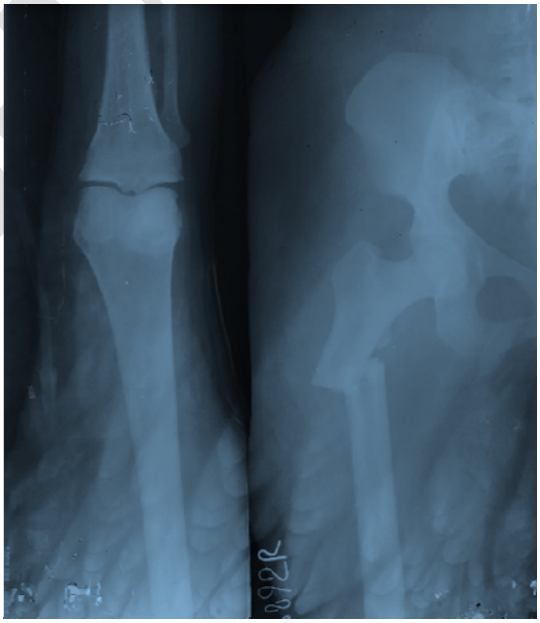

Figure 4 – Radiograph of right hip with femur showing subtrochanteric fracture of right femur

k) Skeletal survey shows diffuse sclerosis of all bones (as shown in figure 5, 6 & 7).

Figure 5 – Radiographs of bilateral wrist with bilateral hands showing sclerosis of bilateral radius & ulna, bilateral carpal & metacarpals.

Figure 6 – Radiographs of skull showing sclerosis of outer table of skull.

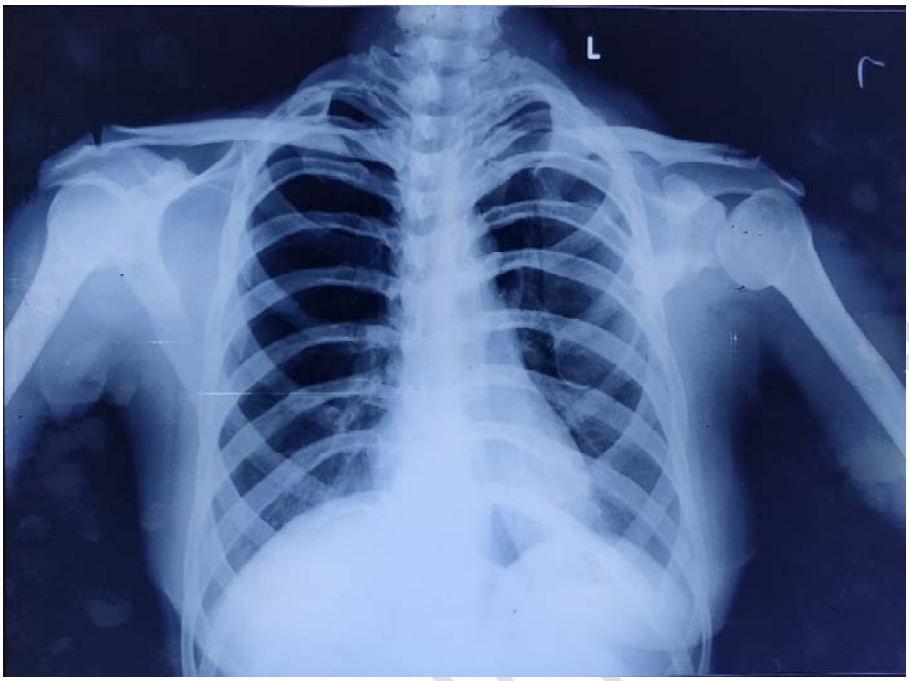

Figure 7 – Radiograph of chest showing diffuse sclerosis of all ribs and clavicle.

l) CT scan of pelvis with both hips

- Right hip showed displaced subtrochanteric fracture of right femur secondary to osteopetrosis as right femur showed diffuse sclerosis (as shown in figure 8, 10a& 10b).
- Left hip showed multiple areas of sclerosis with irregular bony defect and bony outgrowths involving head, neck and proximal shaft of left femur with metallic screws in situ and with the evidence of chronic osteomyelitis (as shown in figure 9, 10a & 10b).

Figure 8 – CT 3D construct showing subtrochanteric fracture of right femur

Figure 9 – CT 3D construct showing chronic osteomyelitis of left proximal femur

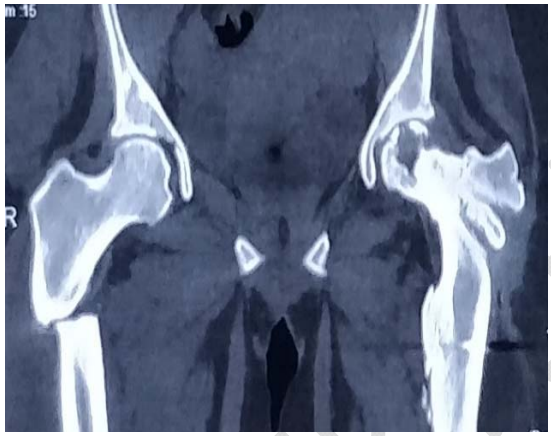

Figure 10a

Figure 10b

After obtaining IEC and the informed and written consent from the patient and her attenders, the patient was subjected for surgical management for subtrochanteric fracture of right femur. Under spinal anaesthesia, the patient was operated in left lateral position. After opening the fracture site, we noticed the closed medullary canal at proximal and distal end of fracture site (as shown in figure 11a and 11b). With vigorous reaming, the medullary canal was opened over both proximal and distal fracture fragments. We noticed an iatrogenic fracture at the end of proximal fracture fragment. Then intramedullary nailing was performed and cerclage wiring was done at the iatrogenic fracture site. The post operative period was uneventful. Then the patient got discharged after 15 days.

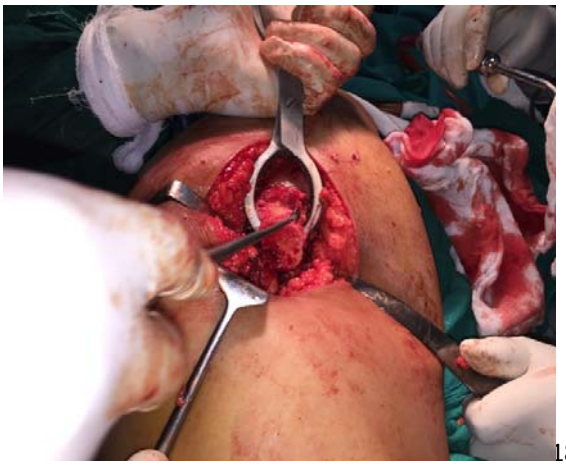

Figure 11a

Figure 11b

During first month follow up, a minimal callus was noticed at the fracture site (as shown in figure 12). At the end of 6 months follow up, a good fracture union was appreciated (as shown in figure 13). The patient had full and pain free range of movements over right hip at the end of 6 months (as shown in figure 14a, 14b, 14c, 14d and 14e).

Figure 12 – Radiograph of right femur with intramedullary nailing at the end of 1st month

Figure 13 – Radiograph of right femur with intramedullary nailing at the end of 6th month

Figure 14a – Flexion

Figure 14b – Extension

Figure 14c – Adduction

Figure 14d – Abduction

Figure 14e – Sitting cross legged

Discussion

In 1904, a German radiologist Albers-Schonberg coined the term Osteopetrosis. Osteopetrosis is also called as “Osteosclerosis”, “Marble bone disease” or “Albers-Schonberg disease”. It is an extremely rare inherited disorder and the most common hereditary cause of sclerotic bone disease. The primary defect in osteopetrosis is due to mutation in CLCN-7 gene on chromosome 16q13.3[1,5]. Osteopetrosis is marked by increased bone density due to the defect in bone reabsorption by osteoclasts which leads to accumulation of bone with defective architecture, making them brittle and susceptible to fracture[2].

In cytopathology of osteopetrosis, the primary manifestation of dysfunctional osteoclasts is either impaired acidification which is due to mutant gene ClCN7 or TCIRG1, which compromise chloride channel and proton pump, subsequently resulting in insufficient acid secretion and abnormal bone reabsorption or generation failure which involves the absence of RANKL on osteoblasts[2,3]. The resorption of calcified cartilage and primary trabecular weakens, thereby inhibiting secondary lamellar bone to replace the primary structure. Thus, bones fail to withstand the stress and reduplicative fracture exists in the course of osteopetrosis.

Forms of osteopetrosis[3,4]

Form of osteopetrosis	Significance	Clinical features
Autosomal Recessive; Malignant Infantile Type	Most apparent at birth but if left untreated, can lead to death in first decade of life	Macrocephaly, hydrocephalus, retinal atrophy, hypertelorism, exophthalmos, strabismus, nystagmus, blindness, sensorineural hearing loss, delayed psychomotor development, osteosclerosis, osteomyelitis, cranial hyperostosis, extramedullary hematopoiesis.
Autosomal Dominant; Adult Type	Milder form of osteopetrosis diagnosed in late childhood or adulthood	Osteosclerosis, fractures after trivial trauma, osteomyelitis and cranial hyperostosis, hepatosplenomegaly, anemia, extramedullary hematopoiesis.
Intermediate	Found in children and can be inherited as a autosomal recessive or autosomal dominant trait with variable severity	Abnormal hardening bones, fractures, mandibular osteomyelitis, genu valgum, cranial hyperostosis, optic atrophy, blindness, mandibular prognathism, dental anomalies, dental caries, facial paralysis, anemia, pancytopenia, extramedullary hematopoiesis.
X-linked Recessive	Extremely rare but severe	Associated with immunodeficiency and ectodermal dysplasia

Adult type Albers – Schonberg disease

Albers – Schonberg disease, also called as marble bone disease or osteopetrosis tarda, is an adult autosomal dominant type 2 osteopetrosis, caused by severe impairment of osteoclast-mediated bone resorption due to mutation in CLCN-7 gene on chromosome 16q13.3[5]. the incidence of autosomal dominant osteopetrosis is 1 in 20,000 births. It is

diagnosed incidentally based on presence of pathological fracture, which usually involves proximal femur and hip.The primary defect in osteopetrosis is loss of osteoclastic bone resorption with preservation of osteoblastic bone formation with the persistent primary spongiosa[6]. Albers-Schonberg disease is characterized by the increased bone density, diffuse and focal sclerosis of varying severity with thickening of bone.

Adult Autosomal dominant osteopetrosis has 2 distinct phenotypic variants[7]. There is no significant difference in radiographic findings of long bones of appendicular skeleton in both thesetypes. Type II shows rugger jersy spine. Serum levels of alkaline phosphatase are reduced in type I and increased in type II. The most common locations for fractures are inferior neck of the femur, the proximal third of the femoral shaftand the proximal tibia. Bone is grossly grayish white on cut section, as hard as marble, brittleness of chalk with obliterated medulary cavity. Histologically, mature osteopetrotic fracture callus contains no haversian organization, with paucity of osteoclasts and normal or increased number of osteoblasts may be normal or increased. Radiographically, the bones have a dense, chalk-like appearance, sandwich or rugger jersey spine appearance,long bone shows marble-like appearance and erlenmeyer's flask shape at their ends[7,8].

In literature, both operative and non-operative modalities are available for treatment of pathological fracture in osteopetrosis[8].Surgery for osteopetrotic fractures is associated with considerable difficulties and complications. Technical difficulties include bending of drill bits or screws during surgery due to hard-fragile sclerotic bones and narrow medullary canal[9]. Many studies report complications like non union, mal union and coxa vara. Osteosynthesis has been the primary method for the surgical treatment of femoral osteopetrotic fractures[10,11,12].

There are case reports in which various implants e.g. locking plates, cannulated screws, dynamic condylar screw (DCS), dynamic hip screws (DHS), and intramedullary nailing (IMN) were used during surgery[13, 14, 15]. In a study including 42 patients with osteopetrosis, Benichou et al reported a fracture rate of 78% and most common localization was femur[2].Kleinberg described the treatment of a peri-trochanteric fracture with a plate, screw and cortical strut allograft. The plate broke and the fracture site became angulated but the fracture united[3].

Kulkarni et al reported a 22-years male case of ADO type II with the left femoral shaft fracture and a 47-years male case of ADO type II with the rightsub-trochanteric fracture. Both patients who underwent open reduction internal fixation under combined spinal + epidural anesthesia were successfully treated[16].

Kumar et al reported a 45-year-old male patient with osteopetrosis in whom the left femoral sub-trochanteric fracture was surgically treated. Internal fixation was performed with a DHS instead of intramedullary nailing due to the presence of a narrow femoral canal[17].

In our case report, a 36 years old short statured female diagnosed with right subtrochanteric fracture of femur. With the help of skeletal survey and other laboratory tests, this case was proved as osteopetrosis. After pre-operative planning and workup, under spinal anesthesia in left lateral position, the standard lateral straight incision was performed in the proximal one-third of right thigh and fracture site was opened. During intra-operative period, closed ends of medullary canal was observed at proximal and distal fracture ends which made reaming very difficult. While reaming hard, we observed iatrogenic fracture at the proximal

fracture end. Then osteosynthesis with internal fixation was performed using intramedullary interlocking femur nail and cerclage wiring at the proximal fracture fragment. Intra-operative successful reduction and fixation was confirmed under C-arm. Patient had no post operative complications and partial weight bearing was started on 2nd post operative day and staple removal was done of 12th post operative day and patient was discharged. Patient was followed up in our hospital and follow up period was uneventful.

In this case, we encountered short stature, non union of trochanteric fracture of left femur, failure of implant removal, iatrogenic fracture neck of left femur, osteomyelitis of left proximal femur, subtrochanteric fracture of right femur after trivial fall, completely closed medullary canal at both proximal and distal fracture fragments and iatrogenic fracture at proximal fracture fragment due to vigorous reaming. With all these findings, this case fit into type II adult type of osteopetrosis.

The patient has been explained about the natural history of disease and counselled for further genetic testing. Due to lack of facilities, genetic tests could not be done.

Conclusion

We suggest that surgery is an effective treatment modality in patients with osteopetrotic fractures, although technical difficulties may be experienced and fracture healing is slower than normal. Technical challenges and complications may occur during surgery; however, we believe that osteopetroticsubtrochanteric femur fractures can be successfully treated with load sharing device such as intramedullary interlocking nailing without using any graft, which promotes fracture healing during primary surgery. Orthopaedics surgeons should be aware of intraoperative technical difficulties and possible postoperative complications during the follow-up period. Investigation would be beneficial for the diagnosis of osteopetrosis such the patient with fractures who has minor trauma history and increased bone density in radiography.

Ethical Approval:

As per international standard or university standard ethical approval has been collected and preserved by the authors.

Funding sources: Nil
Conflicts of interest: Nil
Acknowledgements: Nil

References

1. Stark Z, Savarirayan R. “Osteopetrosis,” Orphanet Journal of Rare Diseases, 2009; 4(1):5.
2. Bénichou OD, Laredo JD, de Vernejoul MC. Type II autosomal dominant osteopetrosis (Albers-Schonberg disease): clinical and radiological manifestations in 42 patients. Bone. 2000;26(1):87–93

3. Kleinberg S. Osteopetrosis. Am J Surg. 1954;87:50–62.
4. Gupta RK. Long bone fractures in osteopetrosis: awareness of primary pathology and appropriate preoperativeplanning necessary to avoid pitfalls in fixation. Injury Extra. 2005;36(3):37–41.
5. Aslan A, Baykal YB, Uysal E, et al. Surgical treatment of osteopetrosis-related femoral fractures: two casereports and Literature Review. Case Rep Orthop. 2014;10.1155:2014.
6. Gupta R, Gupta N. Femoral fractures in osteopetrosis: case reports. J Trauma. 2001;51(5):997–999.
7. Kumbaraci M, Karapinar L, Incesu M, Kaya A. Treatment of bilateral simultaneous subtrochanteric femurfractures with proximal femoral nail antirotation (PFNA) in a patient with osteopetrosis: case report andreview of the literature. J Orthop Sci. 2013;18(3):486–489.
8. Cadosch D, Gautschi OP, Brockamp T, Zellweger R. Osteopetrosis – a challenge for the orthopaedicsurgeon. S Afr J Surg. 2009;47(4):131–133.
9. Breck LW, Cornell RC, Emmett JE. Intramedullary fixation of fractures of the femur in case ofosteopetrosis. J Bone Joint Surg Am. 1957;39(6):1389–1394.
10. Sen RK, Gopinathan NR, Kumar R, Saini UC. Simple reproducible technique in treatment for osteopetroticfractures. Musculoskelet Surg. 2013;97(2):117–121.
11. Chabra A, Westerlund LE, Kline AJ, McLaughlin R. Management of proximal femoral shaft fractures inosteopetrosis: a case series using internal fixation. Orthopedics. 2005;28(6):587–98
12. Bhargava A, Vagela M, Lennox CM. Challenges in the management of fractures in osteopetrosis. Review of literature and technical tips learned from long-term management of seven patients. Injury. 2009;40(11):1167–1171.
13. Ahmet A, Yakup BB, Emin US. Surgical treatment of osteopetrosis-related femoral fractures: two case reports and literature review. Case Rep Orthop2014;2014:891–963.
14. Napoleon Burt, Gary Haynes R, Melinda Bailey K. Patients with malignant osteopetrosis are at high risk of anaesthetic morbidity and mortality Anesthesia&Aalgesia. 1999; 88(6):1292.
15. Birmingham P, Mchale KA. “Case reports: treatment of subtrochanteric and ipsilateral femoral neck fractures in an adult with osteopetrosis,” Clinical Orthopaedics and Related Research, 2008; 466(8):2002–2008.
16. J. V. Kulkarni, R. Bengali, S. Jewalikar, and A. Joshi, “Osteopetrosis— a challenge in rare situation. Case report,” Journal of Evolution of Medical and Dental Sciences, 2012;1(4):532–537.
17. D. Kumar, V. K. Jain, H. Lal, R. K. Arya, and S. Sinha, “Metachronous bilateral subtrochanteric fracture of femur in an osteopetrotic bone: a case report with technical note,” Journal of Clinical Orthopaedics & Trauma, 2012;3(2):103–106.