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Management of femoral fractures in adult onset osteopetrosis: A multicentric retrospective study

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Abstract

Background: Osteopetrosis is a relatively rare disease with an incidence of approximately 1 case per 2,00,000 to 5,00,000 population. The clinical features of osteopetrosis includes pathological fractures, severe anemia due to defective erythropoiesis, bone marrow failure, coagulation defects, propensity for developing severe infections. Common fractures seen in these patients include hip and femoral fractures. Management of fractures in these patients is technically difficult and postoperative complications are common. We conducted a retrospective study of 20 patients with osteopetrosis who presented with femoral fractures and were treated in our institute.

Materials and Methods: This was a multicentric retrospective study of 20 patients with osteopetrosis who presented with femoral fractures. The patients were included in this study on the basis of predefined inclusion and exclusion criteria. The diagnosis was based upon imaging studies (X-Rays in all the patients and CT in selected cases). The operative procedure post-operative complications, functional outcome and complications during follow up were studied from case papers. The data was tabulated and analyzed using Minitab 17 version software.

Results: Out of 20 studied cases there were 16 (80%) males and 4 (20%) females with a M:F ratio of 1: 0.25. The most common affected age group was found to be between 30 – 35 years (35%). Majority of patient sustained fracture following motor vehicular accidents (65%) followed by fall from height (20%) and direct blow or assault (15%). 13 patients were found to be having co-morbidities including anemia, carpal tunnel syndrome and visual disturbances. Open reduction and internal fixation was done in all the cases. Technical difficulties seen during operative procedures included difficult because of obliterated canal, sclerosed medulla and dense swarf. Common complications in these patients included wound infection, hematoma formation and arthritis of hip joint (5%). Repeat surgeries were required in 3 (15%) patients and 1 (5%) patient developed non-union of the fracture ends.

Conclusion: Management of femoral fracture in patients with osteopetrosis is challenging for operating orthopedic surgeon. Common problems encountered during open reduction and internal fixation include overheating during drilling, smoke production and increased temperature of both bone and drill bits and production of dense bone swarf. Orthopedic surgeon should be aware of these technical difficulties encountered during operative interventions.

Keywords: Osteopetrosis, femoral fractures, open reduction and internal fixation, drilling

Introduction

Osteopetrosis is a relatively rare disease with an incidence of approximately 1 case per 2,00,000 to 5,00,000 population [1]. The basic pathology in osteopetrosis is failure of bone resorption by osteoclasts due to which there is defective bone modelling and remodeling. This defective bone remodeling leads to skeletal fragility despite an apparent increase in bone mass. The secondary consequences of such an increase in bone mass, along with bone fragility, is associated with secondary complications such as pathological fractures, defective hematopoiesis, dental defects, nerve entrapments, growth disturbances and renal tubular acidosis [2]. Frontal bossing may be seen in pediatric age group. Osteopetrosis is divided into infantile, intermediate and adult onset forms on the basis of age of onset and clinical features [3]. The clinical features of osteopetrosis includes pathological fractures, severe anemia due to defective erythropoiesis, bone marrow failure, coagulation defects, propensity for developing severe infections [4].

The infantile form osteoporosis usually causes growth retardation, infections and death usually occur by 1st decade of life due to severe anemia and congestive cardiac failure. Adult onset osteopetrosis may present with bony abnormalities such as marked sclerosis of skull and other bones. There is an abnormally high risk of fractures due to increased fragility of bones⁵. Other features of adult onset osteopetrosis includes cranial neuropathies due to nerve entrapments, carpal tunnel and tarsal tunnel syndrome, osteoarthritis and osteomyelitis especially of mandible [6]. Non-skeletal manifestation may include visual impairment due to retinal detachment, retinal degeneration and psychomotor retardation [7]. The diagnosis of osteopetrosis is generally made on the basis of imaging. The imaging features are distinct in infantile as well as adult form of osteopetrosis [8]. Autosomal dominant adult type of osteopetrosis usually presents with pronounced Osteosclerosis of cranial vault with clinical presentation as cranial nerve palsies (Type I) or thickening of endplate, also known as sandwich vertebra, and classical bone-within-bone" appearance in the pelvis (Type II) [9]. Irrespective of the type of osteopetrosis a generalized increased in the bone density is the hallmark of osteopetrosis. Though not diagnostic, Hypocalcemia, hyperparathyroidism and increased levels of acid phosphatase and Creatinine kinase isoform BB are the other biochemical abnormalities seen in these patients [10].

Pathological fractures are one of the hallmarks feature of the affected patients. Defective modelling and remodeling is responsible for increasing fragility of bones predisposing them for pathological fractures [11]. Relatively low-energy or trivial fall may cause fractures in these patients. Many of the patients with osteopetrosis remain asymptomatic till they present with pathological fracture. The hip and femur fractures are common pathological fractures seen in these patients [12]. The management of these fractures are challenging because of defective bone remodeling and increased bone density [13]. Moreover, fixation of an Osteopetrotic femoral fracture is difficult and is open to complications. Delayed union, malunion, non-union and Varus deformities of these fractures are some of the common complications seen in these patients. Even in otherwise uneventful open reduction and internal fixation cases these patients are predisposed for development of infections which may further complicate the management [14].

We conducted this retrospective study of 20 patients with osteopetrosis who presented with femoral fracture and were treated in our institute.

Materials and Methods

This was a multicentric retrospective study of patients who presented with femoral fracture who were either known cases of osteopetrosis or were diagnosis to be having osteopetrosis for the first time after they were admitted for fracture of femur. A total of 20 patients who had been admitted for femoral fractures were diagnosed or were known to be having osteopetrosis. The cases were included in the study on the basis of a predefined inclusion criteria. Cases having any of the exclusion criteria were excluded from the study. A detailed history was taken in all the patients specially to know similar complaints in any other family members, history of past fractures and other significant illness in past. History of blood transfusion was asked. Previous investigation, if available, were reviewed.

Operative Procedure

Under general anesthesia, the patient was placed into the right

or left decubitus position depending upon the side of fracture and the affected extremity was prepared. Patients were operated under general anesthesia. The fracture line was opened with a longitudinal incision from the lateral side. After the fracture ends were reached the medulla was drilled. In almost all cases there was sclerotic medulla and it was difficult to drill the bone with routine drills. In many of the cases the medullary canal was obliterated. For drilling purposes, A 2.5 mm high-speed steel (HSS) drill bit was used with saline cooling. Low speed and high torque setting were used. During drilling a dense bone swarf was frequently encountered that required frequent removing of drill and saline irrigation to clear the accumulated swarf. Drilling was found to be technically very difficult because of obliterated canal, sclerosed medulla and dense swarf. The fracture was then reduced and fixation was made with screws. Bone specimens which were obtained by drilling and the intraoperative biopsy were sent for pathological examination. The histopathological examination confirmed the diagnosis of osteopetrosis. Fluoroscopy checking was applied and the operation was concluded. On the radiograph taken on postoperative Day 2. If the screws were found to be in place and proper reduction was confirmed then the patients were referred for the rehabilitation program on the 4-5th postoperative day and sutures were removed at 15 days. Patients were mobilized without loading on the healing fracture, using a crutch at two weeks and with partial loading at six weeks.



Fig 1: An anteroposterior view of the right hip at the time of initial presentation shows femoral fracture and the Osteosclerotic appearance of the Osteopetrotic bone (Left). Postoperative radiograph after operation with proximal femoral anatomic plate and screws (Right).

The follow up records of at least 1 year was reviewed in all the patients. The data was collected from the case papers of the patients. Statistical analysis was done with Minitab version 17 running on windows 10. Microsoft word was used for manuscript preparation while excel was used for creation of figures and graphs.

Inclusion Criteria

1. All patients who were admitted with femoral fractures and were either know case of osteopetrosis or were diagnosed to be having osteopetrosis after admission to our institute.

- Age of the patients should be more than 18 years.
- The case papers of all the patients included in this study had complete record of treatment as well as of follow up visits for at least 1 year after the discharge of patients from our institute.

Exclusion Criteria

- Patients who have undergone failed attempts at open reduction and internal fixation prior to being referred to our institute.
- Patients in whom treatment record was not complete or missing.
- Follow up record for at least 1 year was not present.
- Age less than 18 years.

Results

This was a retrospective study of patients of osteopetrosis admitted with femoral fracture. Hospital records of patients were studied for demographic details. Out of 20 patients who met the inclusion criteria of the study there were 16 (80%) males and 4 (20%) females with a M:F ratio of 1: 0.25.

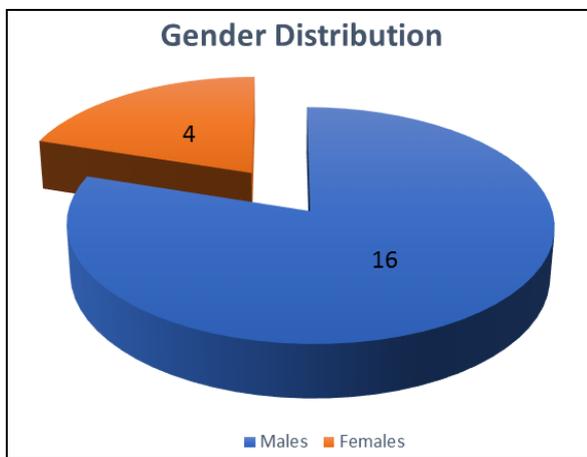


Fig 2: Gender Distribution of the studied cases

The analysis of the age group of the patients showed that the most common affected age group was found to be between 30 – 35 years (35%). Pathological fractures of femur were seen in elderly but those cases were excluded from the study.

Table 1: Age distribution of the studied cases

Age	N	%
≤/ < 20 years	1	5.00%
21-25 years	3	15.00%
26-30 years	4	20.00%
31-35 years	7	35.00%
36-40 years	2	10.00%
> 40 years	3	15.00%
Total	20	100%

Majority of patient sustained fracture following motor vehicular accidents (65%) followed by fall from height (20%) and direct blow or assault (15%).

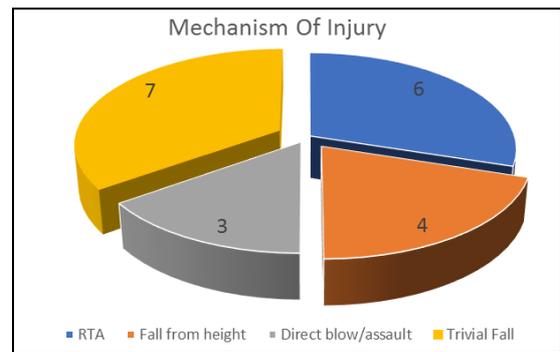


Fig 3: Mechanism of Injury in the studied cases

In 9 (45%) patients fracture of femur was on right side while in 7 (35%) patients was fracture was on seen on left side. Bilateral Femoral Fractures were seen in 4 (20%) patients.

Table 2: Affected side in the studied cases

Affected Side	N	%
Right Femoral Fracture	9	45.00%
Left Femoral Fracture	7	35.00%
Bilateral Femoral Fractures	4	20.00%
Total	20	100%

The analysis of the cases on the basis of associated co-morbidities showed that out of 20 patients 7 were not having any co-morbidities. Anemia was present in 5 patients while carpal tunnel syndrome was present in 3 patients. Other co-morbidities present were osteomyelitis (10%), visual disturbance (10%) and facial nerve palsy (5%).

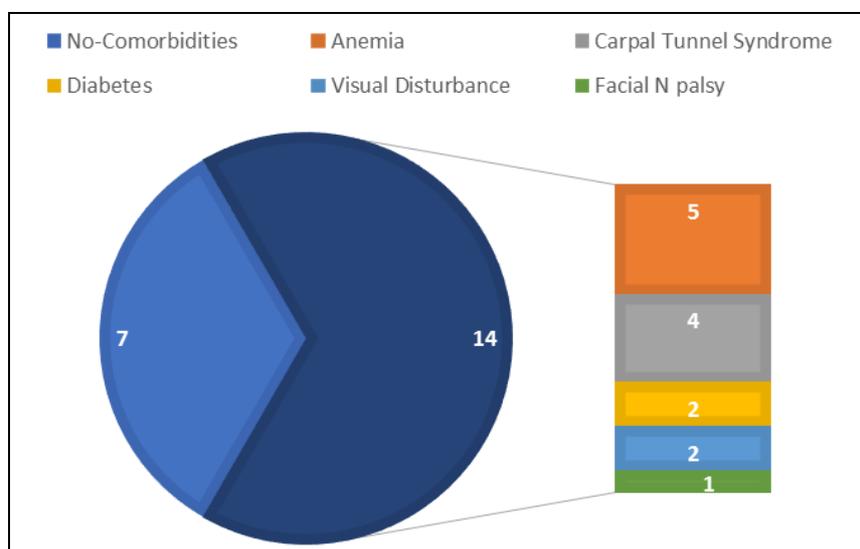


Fig 4: Presence of co-morbidities in affected cases

The classification of fractures on the basis of imaging showed that out of 20 studied cases 7 patients had simple fractures of proximal femur whereas 8 patients has wedge fractures. Remaining 5 patients had complex fractures of distal femur.

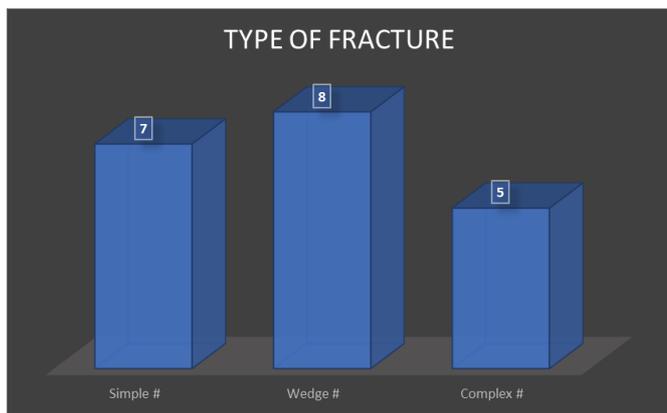


Fig 5: Type of fracture in the studied cases

All patients were treated by open reduction and internal fixation as per the institutional protocol. Patients were operated under general anesthesia. The fracture line was opened with a longitudinal incision from the lateral side. After the fracture ends were reached the medulla was drilled. In almost all cases there was sclerotic medulla and it was difficult to drill the bone with routine drills. Post operatively referred for the rehabilitation program on the 4-5th postoperative day and sutures were removed at 15 days. Post operatively during follow up X-Rays were done to know the radiological evidence of union.



Fig 6: Radiological evidence of union at 1 month and 3 months follow up

Analysis of the complications during follow up showed that there were 5 cases with postoperative complications. These complications included wound infection in 2 (10%) patients. Other complications included hematoma formation (5%) and arthritis of hip joint (5%). Repeat surgeries were required in 3 (15%) patients and 1 (5%) patient developed non-union of the fracture ends for which further therapeutic measures were taken.

Table 3: Complications in the studied cases.

Complications	No of Patients	Percentage
Wound Infection	2	10%
Hematoma Formation	1	5%
Arthritis of Hip Joint	1	5%
Repeat Surgery	3	15%
Non-Union	1	5%
Total	8	40%

The functional outcome of the patients was assessed from the case papers. It was found that out of 20 patients 12 patients (60%) had excellent outcome while Good and fair outcome was seen in 4 (20%) and 3 (15%) patients respectively. Only 1 patient had poor functional outcome.

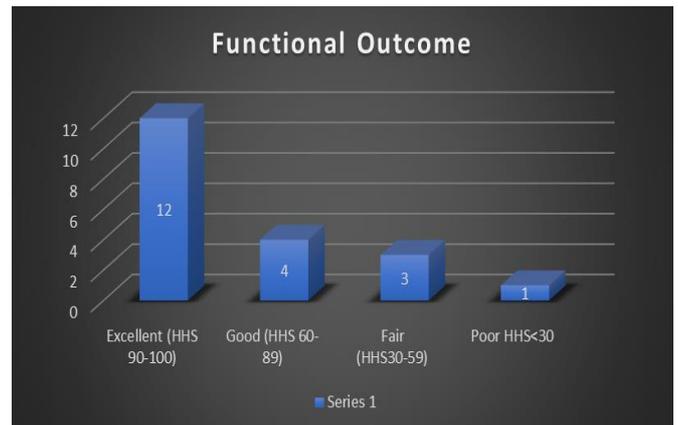


Fig 7: Functional Outcome in studied cases.

Discussion

This was a retrospective study of patients of femoral fracture with osteopetrosis. The study comprised of 20 patients who were admitted with femoral fracture and either were known cases of osteopetrosis or were diagnosed to be having it during the course of imaging. Out of the 20 cases there were 16 (80%) males and 4 females (20%) with M:F ratio of 1:0.25. Adult onset osteopetrosis is autosomal dominant hence males and females are expected to be affected equally. Many case series and case reports have reported approximately equal incidence of osteopetrosis in males as well as females. Though the incidence of osteopetrosis may be equal in males as well as females there are studies which showed that in males fractures may be more commonly seen than females. Sen *et al.* conducted a prospective study of patients with osteopetrosis in which the patients in the age group of 10-50 years with a mean age of 26 years were studied. Five cases were included in the study: four patients had subtrochanteric fractures, and one had segmental fracture of the humerus. Open reduction and internal fixation was done in all the fractures using metal-cutting drill bit. Out of these 5 patients there were 4 males and 1 female with a M:F ratio of 1:0.25. This was similar to male preponderance seen in our study [15].

The most common age group affected was found to be 31-35 years followed by in our study followed by 26-30 years. Adult onset osteopetrosis is frequently symptomatic with majority of the patients experiencing clinical manifestations like pathological fractures, visual disturbances and entrapment neuropathies that gradually increases as the age advance. All these complications usually increase after 20 years of age. The fractures in the patients with osteopetrosis are usually caused following trivial trauma such as a fall within home or following minor accidents which is not expected to cause fractures in otherwise healthy individuals. There are many authors including Gandhi GS *et al.* [16] and Rathod A K *et al.* [17] who have reported patients of osteopetrosis who presented with fractures after trivial trauma.

The common comorbidities seen in our patients included Anemia and carpal tunnel syndrome. Other co-morbidities present were osteomyelitis (10%), visual disturbance (10%) and facial nerve palsy (5%). The cause of anemia in osteopetrosis is usually hyperproliferation of haemopoietic

tissue or increased destruction of red cells or a combination of both these factors [18]. Anemia in adult onset osteopetrosis is difficult to diagnose as most of these patients usually remain asymptomatic and the anemia is usually detected when these patients are investigated for other purposes (preoperative evaluation in cases of fractures) [19].

The management of fractures in osteopetrosis is challenging as it is difficult to achieve proper fixation in these patients. The technical difficulties in drilling in these patients are well known to orthopedic surgeons. There are many instances of breaking of drills during drilling of bones in patients with osteopetrosis. Moreover, screw fixation is also a challenging task due to the strength required. The technical difficulties while drilling bones in patients with osteopetrosis include overheating during drilling, smoke production and increased temperature of both bone and drill bits and production of dense bone swarf. Low speed drilling, frequent changing of drill bits and continuous saline irrigations are some of the measures which can be taken to overcome these difficulties while doing drilling in these patients [20].

The outcome of management of femoral fractures in patients with osteopetrosis is variable and complications such as nonunion or varus malunion are common. In some cases, revision surgeries may also be required. Orthopedic surgeons should be aware of possible challenges during treatment of such patients.

Conclusion

The patients with osteopetrosis are prone for fractures following trivial trauma. Management of these fractures is complicated by factors such as overheating during drilling, smoke production and increased temperature of both bone and drill bits and production of dense bone swarf. Moreover, fractures in these patients may be complicated by malunion and nonunion. It is essential for operating orthopedic surgeon to be aware of intraoperative technical difficulties and postoperative complications seen in these patients.

References

1. Del Fattore A, Cappariello A, Teti A, Genetics, pathogenesis and complications of osteopetrosis. *Bone*. 2008; 42(1):19-29.
2. Stark Z, Savarirayan R. Osteopetrosis. *Orphanet Journal of Rare Diseases*. 2009; 4:5.
3. Sobacchi C, Schulz A, Coxon FP, Villa A, Helfrich MH. Osteopetrosis: genetics, treatment and new insights into osteoclast function. *Nat Rev Endocrinol*. 2013; 9(9):522-36.
4. Bedi RS, Goel P, Pasricha N, Sachin, Goel A. Osteopetrosis-A rare entity with osteomyelitis. *Annals of Maxillofacial Surgery*. 2011; 1(2):155-159.
5. Hamdan A-LH, Nabulsi MM, Farhat FT, Haidar RK, Fuleihan NS. When bone becomes marble: Head and neck manifestations of osteopetrosis. *Paediatrics & Child Health*. 2006; 11(1):37-40.
6. García CM, García MAP, García RG, Gil FM. Osteomyelitis of the Mandible in a Patient with Osteopetrosis. Case Report and Review of the Literature. *Journal of Maxillofacial & Oral Surgery*. 2013; 12(1):94-99.
7. Wangai AM, Waa S, Wangai M, Amayo E, Olunya O. Late-onset Visual Loss in Osteopetrosis. *Sultan Qaboos University Medical Journal*. 2011; 11(3):407-411.
8. Sit C, Agrawal K, Fogelman I, Gnanasegaran G. Osteopetrosis: Radiological & Radionuclide Imaging. *Indian Journal of Nuclear Medicine : IJNM : The Official Journal of the Society of Nuclear Medicine, India*. 2015; 30(1):55-58.
9. Shen FH, Samartzis D, Gaskin CM. Osteopetrosis: "sandwich vertebrae". *Am J Orthop (Belle Mead NJ)*. 2008; 37(3):165-6.
10. Kocher MS, Kasser JR. Osteopetrosis. *Am J Orthop (Belle Mead NJ)*. 2003; 32(5):222-8.
11. Alokaily F. Pathological fracture. *Saudi Medical Journal*. 2015; 36(1):124-125.
12. Aslan A, Baykal YB, Uysal E. Surgical Treatment of Osteopetrosis-Related Femoral Fractures: Two Case Reports and Literature Review. *Case Reports in Orthopedics*. 2014; 2014:891963.
13. Bénichou OD, Bénichou B, de Vernejoul MC. Osteopetrosis as a model for studying bone resorption. *Rev Rhum Engl Ed*. 1998; 65(12):778-87.
14. Arumugam E, Harinathbabu M, Thillaigovindan R, Prabhu G. Marble Bone Disease: A Rare Bone Disorder. Muacevic A, Adler JR, eds. *Cureus*. 2015; 7(10):e339.
15. Sen RK, Gopinathan NR, Kumar R, Saini UC. Simple reproducible technique in treatment for osteopetrotic fractures. *Musculoskeletal Surg*. 2013; 97(2):117-21.
16. Gandhi GS, Vijayanarasimhan V, John L, Kailash S, Balaji ES. Fracture Management in Pyknodysostosis - A Rare Case Report. *Journal of Orthopaedic Case Reports*. 2017; 7(3):54-58.
17. Rathod AK, Dhake RP, Borde MD. Traumatic multiple cervical spine injuries in a patient with osteopetrosis and its management. *Eur Spine J*. 2017; 26(Suppl 1):229-235.
18. Sreehari S, Naik DR, Eapen M. Osteopetrosis: a rare cause of anemia. *Hematology Reports*. 2011; 3(1):e1.
19. Peer M, O' Donoghue K. Osteopetrosis in pregnancy: a rare case report. *Obstetric Medicine*. 2012; 5(1):27-29.
20. Rafiq I, Kapoor A, Burton DJ, Haines JF. A new modality of treatment for non-united fracture of the humerus in a patient with osteopetrosis: a case report. *Journal of Medical Case Reports*. 2009; 3:15.