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Case report of proximal femoral replacement and total hip arthroplasty for bilateral hip nonunion in familial osteopetrosis

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Case Report

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Abstract

Background

Osteopetrosis is a rare skeletal dysplasia characterized by defective osteoclast-mediated bone resorption and impaired bone remodeling. Although the bone appears unusually dense radiographically, its abnormal microarchitecture results in increased fragility and a predisposition to fracture, delayed union, nonunion, and fixation failure. Orthopedic reconstruction is particularly challenging because of the dense cortical bone, narrowed or obliterated medullary canals, and impaired healing capacity.

Case Presentation

A 28-year-old woman with familial osteopetrosis, affecting four sisters, developed sequential complex injuries involving both hips. The right hip sustained a subtrochanteric femoral fracture that progressed to persistent nonunion despite multiple attempts at internal fixation, with progressive proximal femoral bone loss and compromised bone stock. A right proximal femoral replacement (PFR) was performed. The left hip subsequently sustained a femoral neck fracture that developed into an established nonunion with secondary degenerative changes. A cementless total hip arthroplasty (THA) was performed. At the latest follow-up, approximately 13 months after THA and 22 months after PFR, the patient was living normally and had returned to routine daily activities without reported postoperative complications. The Harris Hip Score improved from 29 to 88 following right PFR and from 34 to 92 following left THA. Visual analog scale pain scores improved from 8/10 preoperatively to 1/10 at final follow-up bilaterally.

Conclusions

This case demonstrates the need for individualized, pathology-specific reconstruction in patients with osteopetrosis. PFR provided a salvage solution for severe proximal femoral bone loss and recalcitrant subtrochanteric nonunion, whereas THA addressed femoral neck nonunion associated with secondary joint degeneration. Careful preoperative planning and modified surgical techniques can result in satisfactory functional outcomes despite the substantial technical challenges associated with osteopetrotic bone.

Introduction

Osteopetrosis comprises a heterogeneous group of rare skeletal disorders characterized by defective osteoclast function and impaired bone resorption. The resulting failure of normal bone remodeling produces abnormally dense bone with altered microarchitecture.[1–4] Despite its increased radiographic density, osteopetrotic bone is structurally abnormal and may be brittle and susceptible to fracture.[2–4]

Orthopedic management of fractures in patients with osteopetrosis is challenging. The cortex may be extremely dense, making drilling, broaching, and reaming technically difficult. The medullary canal may be markedly narrowed or completely obliterated, while impaired bone remodeling contributes to delayed union, nonunion, and failure of internal fixation.[5–9] Consequently, conventional fixation strategies may be unsuccessful, particularly in fractures occurring in mechanically demanding regions such as the subtrochanteric femur.

Total hip arthroplasty has been reported in patients with osteopetrosis for conditions including femoral neck fracture, nonunion, and secondary degenerative arthritis. However, preparation of the femoral canal and achievement of reliable implant fixation remain technically demanding because of the abnormal bone morphology.[11–17] In cases of severe proximal femoral bone loss in which conventional reconstruction is no longer feasible, proximal femoral replacement can provide a salvage option by replacing the structurally compromised proximal femur rather than relying on healing across a chronic nonunion.[18,19]

We present a case of familial osteopetrosis in a young woman who developed complex pathology in both hips requiring two different reconstructive strategies. The right hip underwent proximal femoral replacement for recalcitrant subtrochanteric nonunion with severe proximal femoral bone loss, while the contralateral hip subsequently underwent total hip arthroplasty for femoral neck nonunion with secondary degenerative changes.

Literature Review

Reports of orthopedic complications in osteopetrosis are largely confined to isolated case reports and small single-institution case series, reflecting the rarity of the disease.[1–4] Across this literature, fracture healing is consistently described as impaired, and internal fixation is complicated by hardware failure, delayed union, and nonunion, particularly in mechanically loaded regions such as the subtrochanteric femur.[5–9]

A number of case reports and small series describe total hip arthroplasty performed for femoral neck fracture, nonunion, or secondary degenerative arthritis in osteopetrosis.[11–17] These reports consistently emphasize that femoral canal preparation is technically demanding because of the dense, poorly compliant bone, and describe a range of approaches to fixation, including cemented, cementless, and hybrid constructs, with authors generally recommending controlled, sequential reaming to reduce the risk of iatrogenic fracture or thermal injury during implantation.[11–17]

Proximal femoral replacement has been reported chiefly in non-oncologic settings such as periprosthetic fracture, failed arthroplasty, and severe proximal bone loss unrelated to osteopetrosis, where it functions as a salvage option when conventional fixation or revision arthroplasty is not feasible.[18,19] Reported outcomes are mixed rather than uniformly favorable: in one series of 48 patients with non-neoplastic proximal femoral bone loss, an excellent or good functional result was achieved in 22 of 43 hips, and ten patients required reoperation or revision for at least one complication.[19] Recognized risks include

dislocation, infection, and abductor dysfunction related to the extent of soft tissue disruption required for implantation; similar failure modes are described in megaprosthesis series for tumor reconstruction.[18–20]

Taken together, the available literature indicates that THA is an established option for osteopetrosis-related hip pathology, including bilateral THA,[14] and that PFR has been described mainly for non-neoplastic proximal femoral bone loss unrelated to osteopetrosis.[18,19] To the best of our knowledge, we did not identify a previous report of PFR for osteopetrosis-related subtrochanteric nonunion, or of PFR and THA being used in contralateral hips of the same patient with osteopetrosis. The present case illustrates how the choice between fixation, arthroplasty, and salvage reconstruction can differ even within a single patient, depending on the location, chronicity, and bone stock associated with each injury.

Methods

This case report was prepared in accordance with the CARE (CAse REport) reporting guidelines. Ethical approval was obtained from the Institutional Review Board (IRB) at Palestine Polytechnic University in Palestine (reference EA/2024/224). Khaled Surgical Hospital, where the patient was treated, additionally granted institutional permission for the preparation and publication of this case report in September 2026, through the treating surgeon. All methods were performed in accordance with the relevant guidelines and regulations, including the World Medical Association Declaration of Helsinki: Ethical Principles for Medical Research Involving Human Participants. Freely given, written informed consent to participate in this study and for publication of the clinical details and any accompanying images was obtained from the patient. Clinical data were collected retrospectively from the patient's medical records and de-identified to protect confidentiality. The literature review was based exclusively on previously published literature.

Case Presentation

A 28-year-old woman with known familial osteopetrosis presented with a history of significant bilateral hip pathology. Her family history was notable for four sisters who were similarly affected. A specific molecular subtype or genetic mutation was not available in the clinical records.

The initial major orthopedic problem involved the right hip. The patient sustained a subtrochanteric femoral fracture that subsequently developed into a persistent nonunion. Multiple attempts at internal fixation were undertaken; however, the fracture failed to unite. Continued mechanical stress and the impaired remodeling capacity associated with osteopetrosis resulted in progressive compromise of the proximal femoral bone stock. The persistent nonunion was associated with substantial pain and functional impairment.

Given the recalcitrant nature of the nonunion, the failure of previous fixation attempts, and the progressive loss of viable proximal femoral bone, further conventional fixation was considered unreliable. A decision was therefore made to proceed with proximal femoral replacement as a salvage reconstruction.

The patient underwent right proximal femoral replacement using a MUTARS® Proximal Femoral Replacement system. A direct lateral Hardinge approach was used. Intraoperatively, the femoral canal was found to be severely compromised and essentially obliterated. High-speed burrs were therefore used to carefully establish and prepare the femoral diaphyseal canal. Host bone was subsequently impacted to facilitate reconstruction and accommodate the cemented diaphyseal stem. The procedure provided immediate structural reconstruction of the deficient proximal femur while avoiding dependence on union across the chronic subtrochanteric nonunion.

Postoperative radiographs demonstrated the reconstructed proximal femur and implant without a reported immediate radiographic complication (Fig. 1). The patient's postoperative recovery was satisfactory, with progressive improvement in pain and function.

The patient's second major hip problem subsequently developed on the left side. She sustained a left femoral neck fracture that progressed to an established nonunion. Over time, secondary degenerative changes developed in the hip joint, producing persistent pain and functional limitation. Unlike the right hip, the principal pathology on the left involved an intracapsular femoral neck nonunion associated with degenerative joint disease. Therefore, fixation of the femoral neck alone would not adequately address the associated articular pathology.

A decision was made to proceed with total hip arthroplasty. The patient underwent cementless left total hip arthroplasty through a posterior approach. The abnormal density of the osteopetrotic bone required careful and controlled preparation of the femur. Sequential power reaming was performed to prepare the femoral canal. A Corail® proximally porous-coated femoral stem was inserted using a press-fit technique. The acetabular reconstruction included a highly cross-linked polyethylene liner. Total operative time was 127 minutes, with an estimated blood loss of 450 mL. Radiographs after both reconstructions are shown in Fig. 2.

The combination of femoral neck nonunion and secondary degenerative changes was successfully addressed through arthroplasty, eliminating the need for biological union at the fractured femoral neck while restoring the hip joint.

Timeline

Dates are expressed relative to the right proximal femoral replacement (PFR). Exact dates of the earlier fixation procedures were not available.

Before PFR

Right subtrochanteric femoral fracture with multiple failed internal fixation attempts, progressing to recalcitrant nonunion with proximal femoral bone loss (dates unavailable).

Month 0

Right PFR (MUTARS®) for recalcitrant subtrochanteric nonunion. Preoperative Harris Hip Score (HHS) 29; visual analog scale (VAS) pain 8/10.

Interval between PFR and THA

Left femoral neck fracture progressing to nonunion with secondary degenerative change (exact dates unavailable).

Approximately month 9

Left cementless total hip arthroplasty (THA). Preoperative HHS 34; VAS pain 8/10.

Approximately month 22 (13 months after THA)

Final follow-up. HHS 88 (right) and 92 (left); VAS pain 1/10 bilaterally; no complications.

Follow-up and Outcome

The patient was followed clinically and radiographically following both reconstructions. At the most recent assessment, she was approximately 22 months after right proximal femoral replacement and 13 months after left total hip arthroplasty.

At final follow-up, the patient reported doing extremely well and had returned to normal daily activities. No postoperative infection, dislocation, periprosthetic fracture, or other reported clinical complication was identified. Follow-up radiographs demonstrated the bilateral reconstructions without reported radiographic complications (Fig. 2).

Following right proximal femoral replacement, the Harris Hip Score improved from 29 preoperatively to 88 at final follow-up. Her visual analog scale pain score improved from 8/10 to 1/10. She was able to walk more than 1 km without an assistive device, although a mild Trendelenburg gait remained. Following left total hip arthroplasty, the Harris Hip Score improved from 34 preoperatively to 92 at final follow-up, while the visual analog scale pain score improved from 8/10 to 1/10. At the final examination, left hip range of motion included approximately 110° of flexion, 30° of abduction, and 40° of external rotation.

Overall, both reconstructions resulted in substantial improvement in pain and function, allowing the patient to resume normal daily activities.

Discussion

Osteopetrosis presents a unique paradox in orthopedic surgery: the skeleton is radiographically dense but biologically and mechanically abnormal. Defective osteoclastic bone resorption leads to accumulation of poorly remodeled bone, resulting in increased density while compromising normal

structural organization.[1–4] This abnormal bone architecture contributes to fracture susceptibility and impaired healing.

Fracture fixation is particularly difficult because the dense cortical bone can make conventional drilling and implant preparation challenging. Narrow or obliterated medullary canals may further complicate intramedullary fixation and arthroplasty.[5–10] In addition, impaired remodeling can compromise the biological response required for fracture healing, contributing to delayed union and nonunion.[5,6]

The right hip in this patient illustrates the limitations of repeated internal fixation in the setting of severe osteopetrotic bone disease. The subtrochanteric region is exposed to substantial mechanical forces, and persistent nonunion in this location creates a challenging combination of poor biology and unfavorable biomechanics. Multiple fixation attempts had failed, while progressive proximal femoral bone loss reduced the amount of viable bone available for further reconstruction. Failure of fixation and secondary complications after proximal femoral fracture surgery have also been described by others in osteopetrosis.[21]

In this setting, proximal femoral replacement offered a fundamentally different solution. Rather than attempting another fixation construct and relying on union of the chronic fracture, PFR replaced the structurally deficient proximal femur with a prosthetic reconstruction.[18,19] PFR has been described as a salvage procedure for severe proximal femoral bone loss in non-neoplastic conditions and can provide immediate structural stability when conventional reconstruction is unlikely to succeed.[19] However, the procedure carries potential risks including instability, infection, abductor dysfunction, and periprosthetic fracture.[18–20] Despite these concerns, no such complications were reported in this patient during the available follow-up.

The left hip represented a different clinical problem. The femoral neck fracture progressed to nonunion and was accompanied by secondary degenerative changes of the hip joint. In this setting, fixation of the femoral neck would have addressed the fracture but not the established joint degeneration. Total hip arthroplasty therefore provided a means of simultaneously treating the nonunion and replacing the diseased joint surface.

Previous reports have demonstrated that THA can be successfully performed in patients with osteopetrosis, although femoral preparation and implant fixation can be technically demanding.[11–17] The surgical challenges encountered during the left THA were consistent with the known characteristics of osteopetrotic bone. The dense femoral bone required controlled sequential reaming, and careful preparation was necessary to establish a suitable canal for the femoral component. A cementless proximally porous-coated stem was ultimately implanted using a press-fit technique. The favorable clinical outcome suggests that, when adequate fixation can be achieved, cementless reconstruction may be feasible in selected patients with osteopetrosis.[11–17]

An important feature of this case is that the same underlying skeletal disorder produced two different reconstructive problems requiring different surgical solutions. The right hip had a chronic

subtrochanteric nonunion with severe proximal femoral bone loss after failed fixation, making PFR an appropriate salvage strategy. The left hip had femoral neck nonunion with secondary degenerative joint changes, for which THA was better suited. This distinction emphasizes that treatment decisions in osteopetrosis should be based not only on the underlying diagnosis but also on the location and biology of the fracture, the degree of bone loss, previous treatment failures, joint involvement, and the remaining bone available for reconstruction.

The familial nature of the patient's disease is also notable. Four sisters were similarly affected, supporting a familial form of osteopetrosis, although the specific genetic subtype was not available. Osteopetrosis encompasses multiple genetic disorders with variable clinical severity and inheritance patterns.[1,3,4] Identification of the underlying molecular subtype may be valuable in future cases because genotype can influence disease severity and associated systemic manifestations.

This report has several limitations. First, it represents a single patient and therefore cannot establish the superiority of either reconstructive strategy. Second, the precise genetic subtype of osteopetrosis was unavailable. Third, long-term implant survivorship cannot yet be determined because the latest follow-up was approximately 13 months after THA and 22 months after PFR. Finally, details of some of the patient's earlier internal fixation procedures were not available for comparison, and radiographic follow-up is limited to the images presented, so long-term osseointegration, implant migration, and periprosthetic bone changes could not be assessed. Longer follow-up will be necessary to assess the durability of both reconstructions.

Conclusions

Bilateral hip reconstruction in patients with familial osteopetrosis can be exceptionally challenging because of abnormal bone density, impaired remodeling, difficult canal preparation, and a high risk of delayed union or nonunion. In this patient, the right subtrochanteric fracture developed into a recalcitrant nonunion with substantial proximal femoral bone loss following multiple failed fixation attempts and was successfully managed with proximal femoral replacement. The subsequent left femoral neck nonunion, complicated by secondary degenerative changes, was treated with total hip arthroplasty.

At approximately 13 months following left THA and 22 months following right PFR, the patient demonstrated substantial improvements in pain and hip function and had returned to normal daily activities without reported postoperative complications. This case highlights the importance of individualized, pathology-specific reconstruction in osteopetrosis and demonstrates that successful bilateral hip reconstruction can be achieved despite the substantial technical challenges posed by this rare skeletal disorder.

Declarations

Ethics approval and consent to participate:

This study was conducted in accordance with the ethical principles outlined in the Declaration of Helsinki. Ethical approval was obtained from the Institutional Review Board (IRB) at Palestine Polytechnic University in Palestine (reference EA/2024/224). Khaled Surgical Hospital, where the patient was treated, additionally granted institutional permission for the preparation and publication of this case report in September 2026, through the treating surgeon. Freely given, written informed consent to participate in this study was obtained from the patient. Clinical data were collected retrospectively from the patient's medical records and de-identified to protect confidentiality.

Consent for publication:

Written informed consent for publication of this case report, including the clinical details and any accompanying images, was obtained from the patient. A copy of the written consent is available for review by the Editor-in-Chief of this journal.

Competing interests:

The authors declare no competing interests.

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Author Contribution

Author Contributions StatementM.A. contributed to the conceptualization of the study, literature review, data collection and interpretation, and drafting and revision of the manuscript. D.M.A.H. contributed to the literature review, data collection, and manuscript drafting. Q.M.A. contributed to the clinical interpretation of the case, literature review, and critical revision of the manuscript. H.A. contributed to the literature review and critical revision of the manuscript. S.A.H. contributed to data interpretation and manuscript revision. All authors reviewed and approved the final manuscript and agree to be accountable for all aspects of the work.

Data Availability

All data supporting the findings of this case report are included within the article, including the clinical details, outcome scores, and radiographic images (Figs. 1 and 2). The patient's underlying medical

records are not publicly available because of patient confidentiality and consent restrictions. De-identified information beyond what is reported in the article is available from the corresponding author on reasonable request, subject to the patient's consent and institutional approval.

References

1. Tolar J, Teitelbaum SL, Orchard PJ. Osteopetrosis. N Engl J Med. 2004;351(27):2839-2849. DOI: https://doi.org/10.1056/NEJMra040952
2. Shapiro F. Osteopetrosis. Current clinical considerations. Clin Orthop Relat Res. 1993;(294):34-44. PubMed: https://pubmed.ncbi.nlm.nih.gov/8358940/
3. Del Fattore A, Cappariello A, Teti A. Genetics, pathogenesis and complications of osteopetrosis. Bone. 2008;42(1):19-29. DOI: https://doi.org/10.1016/j.bone.2007.08.029
4. Stark Z, Savarirayan R. Osteopetrosis. Orphanet J Rare Dis. 2009;4:5. DOI: https://doi.org/10.1186/1750-1172-4-5
5. de Palma L, Tulli A, Maccauro G, Sabetta SP, del Torto M. Fracture callus in osteopetrosis. Clin Orthop Relat Res. 1994;(308):85-89. DOI: https://doi.org/10.1097/00003086-199411000-00014
6. Landa J, Margolis N, Di Cesare P. Orthopaedic management of the patient with osteopetrosis. J Am Acad Orthop Surg. 2007;15(11):654-662. DOI: https://doi.org/10.5435/00124635-200711000-00004
7. Gupta R, Gupta N. Femoral fractures in osteopetrosis: case reports. J Trauma. 2001;51(5):997-999. DOI: https://doi.org/10.1097/00005373-200111000-00027
8. Bhargava A, Vagela M, Lennox CME. 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. DOI: https://doi.org/10.1016/j.injury.2009.02.009
9. Armstrong DG, Newfield JT, Gillespie R. Orthopedic management of osteopetrosis: results of a survey and review of the literature. J Pediatr Orthop. 1999;19(1):122-132. DOI: https://doi.org/10.1097/00004694-199901000-00026
10. Spinnato P, Pedrini E, Petrera MR, Zarantonello P, Trisolino G, Sangiorgi L, Carpenzano M, Crombe A, Tetta C. Spectrum of skeletal imaging features in osteopetrosis: inheritance pattern and radiological associations. Genes (Basel). 2022;13(11):1965. DOI: https://doi.org/10.3390/genes13111965
11. Matsuno T, Katayama N. Osteopetrosis and total hip arthroplasty. Report of two cases. Int Orthop. 1997;21(6):409-411. DOI: https://doi.org/10.1007/s002640050196
12. Cameron HU, Dewar FP. Degenerative osteoarthritis associated with osteopetrosis. Clin Orthop Relat Res. 1977;(127):148-149. DOI: https://doi.org/10.1097/00003086-197709000-00019
13. Egawa H, Nakano S, Hamada D, Sato R, Yasui N. Total hip arthroplasty in osteopetrosis using computer-assisted fluoroscopic navigation. J Arthroplasty. 2005;20(8):1074-1077. DOI: https://doi.org/10.1016/j.arth.2005.03.037
14. Jones DPG, Hodgson BF, Hung NA. Bilateral, uncemented total hip arthroplasty in osteopetrosis. J Bone Joint Surg Br. 2004;86(2):276-278. DOI: https://doi.org/10.1302/0301-620X.86B2.14354

15. Ashby ME. Total hip arthroplasty in osteopetrosis: a report of two cases. Clin Orthop Relat Res. 1992;(276):214-221. PubMed: https://pubmed.ncbi.nlm.nih.gov/1537156/
16. Siljander MP, Trousdale RT, Perry KI, Mabry TM, Berry DJ, Abdel MP. Total hip arthroplasty in patients with osteopetrosis. J Arthroplasty. 2021. DOI: https://doi.org/10.1016/j.arth.2020.10.018
17. Xie L, Ding F, Jiao J, Kan W, Wang J. Total hip and knee arthroplasty in a patient with osteopetrosis: a case report and review of the literature. BMC Musculoskelet Disord. 2015;16:259. DOI: https://doi.org/10.1186/s12891-015-0716-x
18. Klein GR. Proximal femoral replacement for the treatment of periprosthetic fractures. J Bone Joint Surg Am. 2005;87(8):1777-1781. DOI: https://doi.org/10.2106/JBJS.D.02420
19. Parvizi J, Tarity TD, Slenker N, Wade F, Trappler R, Hozack WJ, Sim FH. Proximal femoral replacement in patients with non-neoplastic conditions. J Bone Joint Surg Am. 2007;89(5):1036-1043. DOI: https://doi.org/10.2106/JBJS.F.00241
20. Henderson ER, Groundland JS, Pala E, Dennis JA, Wooten R, Cheong D, Windhager R, Kotz RI, Mercuri M, Funovics PT, Hornicek FJ, Temple HT, Ruggieri P, Letson GD. Failure mode classification for tumor endoprostheses: retrospective review of five institutions and a literature review. J Bone Joint Surg Am. 2011;93(5):418-429. DOI: https://doi.org/10.2106/JBJS.J.00834
21. Seyfettinoglu F, Tuhanioglu U, Ogur HU, Cicek H. Proximal femoral fracture surgery in a patient with osteopetrosis tarda: complications and treatment strategy. Int Med Case Rep J. 2016;9:347-351. DOI: https://doi.org/10.2147/IMCRJ.S114710

Figures



Figure 1

Anteroposterior pelvic radiograph after right proximal femoral replacement and before left total hip arthroplasty, showing the right proximal femoral replacement with a cemented diaphyseal stem and acetabular screw fixation, and the native left hip.



Figure 2

Follow-up anteroposterior pelvic radiograph showing the right proximal femoral replacement and the left cementless total hip arthroplasty with a proximally porous-coated femoral stem, with acetabular screw fixation bilaterally. Cerclage wires are visible around the left femoral shaft.