Total Knee Arthroplasty in a Patient with Osteopetrosis: A Case Report and Review of the Current Literature

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Abstract

Introduction: Osteopetrosis can present distinct challenges in a patient undergoing total knee arthroplasty. The disease can manifest as generalized osteosclerosis and abnormal bony architecture. We report a case of total knee arthroplasty (TKA) in a patient with osteopetrosis.

Case Report: We report a 59 year old female with osteopetrosis who had failed non-operative management for knee arthritis. Patient specific instrumentation was utilized for the surgery to avoid instrumenting the femoral or tibial canals. Despite our best effort, our case was complicated by several intra-operative fractures. We outline our treatment and rationale, including recommendations on dealing with osteopetrosis if planning to perform TKA.

Conclusion: Osteopetrosis is a condition that may present to the orthopaedic surgeon. Planning for possible intra-operative difficulties given this condition is of vital importance in order to achieve a successful surgical outcome.

Keywords: Osteopetrosis; TKA; Patient Specific Instrumentation; Intra-Operative Fracture

Introduction

Osteopetrosis, also known as “Marble Bone Disease” was first described in the early 20th Century by German radiologist Dr. Albers-Schönberg after encountering a patient with a history of generalized osteosclerosis and multiple fractures [1]. Osteopetrosis is a genetically inherited condition with several possible modes of inheritance that all lead to impaired osteoclastic function and an inability to remodel bony matrix. Common features of osteopetrosis include pathological fractures, short stature, hematological and neurological abnormalities [2].

The most commonly encountered form of osteopetrosis by orthopaedic surgeons is the benign autosomal dominant type. This is due to long patient lifespans and the many complications of their disease process. While this type of osteopetrosis is still a relatively rare condition, surgeons should still be mindful of these patients due to their propensity to seek orthopaedic care and the unique set of challenges involved in surgical management of their disease complications [3]. Degenerative osteoarthritis is the most commonly encountered sequela of chronic osteopetrosis. These patients often exhaust conservative management options and total joint arthroplasty can present as a viable option for treatment of their osteoarthritis [4].

Previous case reports on patients with osteopetrosis undergoing TKA have described various technical challenges during surgery such as drilling the femoral canal, drilling pins for intramedullary cutting guides, performing the bony resections, cementing of prosthetic components, and sizing of the implant. Additionally, these previous reports have described precautions in order to avoid iatrogenic frac-
ture and instrumentation malfunctions, such as overheating of saw blades and breaking of drill bits [3-5]. In our case we tried to mitigate some of the complications described in the literature by assuring adequate instrumentation and the use of patient specific cutting guides. However, despite our preparations, we still encountered difficulties during the course of the operation.

Case Presentation

We report on a 59-year-old female with osteopetrosis who presented to our orthopaedic clinic with bilateral, left greater than right, knee pain. She reported that the left knee pain had been present for over six years and denied any history of trauma. At presentation, she described an aching, burning, and shooting pain localized to the lateral joint line of the left knee. She stated that the pain was worse with standing, sitting, and walking for long periods of time. She stated that the pain improved minimally with rest and elevation of the affected knee, and that all previous pharmacological efforts had only provided short-lived relief. The patient also underwent physical therapy, which failed to provide adequate pain relief or improvement in knee function.

Physical examination of the left knee demonstrated a valgus deformity of 20 degrees and range of motion of 5-110 degrees with crepitation. The valgus deformity was mildly correctable but there was no gross instability of the left knee, and there was no effusion. The left lower extremity was neurovascularly intact.

Preoperative radiographic examination of the left knee revealed diffusely sclerotic bone. The x-rays also showed tricompartmental osteoarthritis, worse in the lateral compartment (Figure 1).
After a thorough discussion of treatment options and risks, the patient elected to undergo a total knee arthroplasty. Based on our patient’s history of osteopetrosis, we had concerns regarding potential technical challenges of performing a total knee arthroplasty. After a review of the current literature, we decided to use patient specific instrumentation [4] and MRI based custom cutting blocks were ordered/obtained (MicroPort, Arlington, TN) (Figure 2 and 3).

**Figure 2:** Preoperative and estimated postoperative alignment measurements.

**Figure 3:** 3-D rendering of the femoral and tibial custom cutting guides. (Microport).
Operative report

After appropriate informed consent was obtained, the patient was taken to the operating room and underwent general endotracheal anesthesia. A Foley catheter was placed, and a nonsterile tourniquet was placed to the thigh. The left leg was then prepped and draped in a normal sterile fashion. A timeout was called confirming the patient, site, side, procedure, that she had received tranexamic acid and antibiotics within 30 minutes of incision, and that she had no known drug allergies.

A standard medial parapatellar approach was performed. The patella was everted, and the knee was flexed. The custom distal femoral cutting guide was then placed, and the distal femoral cut was made (Figure 4). A sizing guide was then used to confirm the appropriate 4 in 1 cutting block. Upon insertion of the 4 in 1 cutting guide, it was found that the lateral condyle was split in a sagittal direction (Figure 5). We attempted cancellous screw fixation of the fracture fragment three times. However, with each attempt, the brittle fracture fragment became further comminuted (Figure 6). When it had become evident that the fracture fragments were not amenable to screw fixation, a decision was made to finish the distal femur cuts and secure the fracture fragments with cement externally at the time of cementing of implants. The 4 in 1 cutting block was reapplied and the remaining distal femoral cuts were made. Next a trial femur was placed and was noted to fit well. We then turned our attention to the tibia. The anterior cruciate ligament was transected followed by the resection of the medial and lateral menisci. The tibia was then subluxed anteriorly using a posterior cruciate ligament retractor. Next, the custom proximal tibial cutting guide was applied (Figure 7). This guide was then pinned in place and the tibial cut was made. A spacer block was inserted, and the knee was found to be well balanced in flexion and extension. The tibial plate was then sized, and a trial base plate was pinned in place. A reamer and keel punch were used to prepare the proximal tibia. This step was aided by the use of a drill and a saw to penetrate the extremely hard bone. A trial reduction with femoral and tibial components was successful and the knee was found to be well-balanced in extension and flexion. However, after removing the trial components, we found that the keel punch had fractured the anterior-medial aspect of the tibial plateau into a few small fragments (Figure 8). We again made the decision to address this defect during cementation of the tibial components. We then sized and cut the patella. It was prepared without difficulty and the trial patella button fit well.
Figure 6: Further comminution of the lateral femoral condyle after fixation attempts with cancellous screws.

Figure 7: Custom tibial cutting guide in place.

Next, the joint was irrigated, and the cement was mixed. A decision was made to use two different cement batches to allow for sufficient fixation of the fractures. Once the cement was ready, the tibial component was cemented in place while the anteromedial fracture fragments were manually kept in place. Excess cement was then used to hold the fragments in place externally. Once the first batch of cement had hardened, a second batch of cement was mixed, and the femoral component was cemented in place while the lateral condyle fracture was held in reduction manually. Excess cement was allowed to cover the lateral aspect of the lateral femoral condyle holding the fracture in place. The patella button was cemented without any problem also using the second batch of cement. Next a trial reduction was performed with a trial tibial insert and the knee was found to be stable and well balanced and the patella tracked well. Then a permanent tibial insert was placed, the knee was irrigated, and a standard closure was performed.

Post-operative imaging was collected (Figure 9).

After an uneventful recovery, the patient was discharged on post-operative day two and was scheduled for follow up in 2 weeks.

The patient was seen for follow up at 2 weeks, 4 weeks, 8 weeks, and 5 months, with no significant complications.

At 5 months post-operation, the patient reported that she was doing well. Patient was ambulating without any assistive device, and she stated that there was no pain or stiffness, even after very long walks. At the 5-month point, the patient had stopped receiving formal physical therapy and had transitioned to home therapy. Her physical exam revealed a well healed incision with no tenderness about the knee. The patient’s active range of motion was 5 degrees to 90 degrees of flexion. Postoperative imaging at 5 months showed no significant changes or loosening of hardware, no acute abnormalities or fractures, and heterotopic ossification at the posterior aspect of the joint (Figure 10).

**Discussion**

Osteopetrosis is an inherited disease that results in defective osteoclast function leading to brittle, sclerotic bone, and obliteration of the medullary canal. Osteopetrosis, while not a frequently encountered condition, may present to the orthopaedic surgeon due to complications from the disease process. A common sequela of osteopetrosis is osteoarthritis (OA), and the current literature suggests that...
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Figure 9: Immediate post-op x-rays.

Figure 10: X-rays at 5 months post op.

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surgical treatment of knee OA via TKA is a viable treatment option after failure of conservative measures. Orthopaedic surgeons should be aware of the difficulties in surgically managing these patients in order to adopt strategies to maximize the potential for good surgical outcomes [6].

In 1989, Casden., et al. reported on a case of a 50-year-old female with longstanding history of osteopetrosis undergoing TKA. The case report described bone that was extremely dense and difficult to osteotomize. Additionally, the saw blade had to be frequently cooled, cutting jigs were difficult to place, and creating the tibial peg trough presented a challenging problem [7]. Despite how long ago this first reported case was, similar difficulties were seen in all the other cases.

In 2005, Strickland., et al. reported two cases of osteopetrotic patients receiving total knee arthroplasty. Extramedullary alignment was used for both patients and long operative times were also reported. They also reported difficulty with making bone cuts, and these cases were the first to describe using a burr instead of a broach for preparation of the tibia [8].

The next reported case of total knee arthroplasty in an osteopetrotic patient was reported in 2012 by Mayer., et al. They were the first to report using patient specific instrumentation (PSI). They reported difficult bone preparation using multiple drill bits, saw blades, and batteries. Complications reported in this case included fracture of one of the femoral condyles when making initial bone cuts [4].

Van Hove., et al. reported a case in 2014 using extramedullary alignment and mentioned difficulties with making bone cuts along with the need for additional saw blades. A complication in this case included a fissure fracture distal to the tibial cone [9].

Xie., et al. reported a case in 2015 using extramedullary alignment. They reported intraoperative difficulties consistent with previous case reports, including the use of multiple saw blades due to difficulty in resecting bone. Similarly, to Mayer., et al. they reported an iatrogenic fracture of the medial condyle that occurred during preparation of the femur.

Most recently, Soberon., et al. reported a case in 2019 that also involved the use of multiple saw blades, drill bits, and batteries due to difficulty with bone preparation. They reported a long operative time and multiple intraoperative fractures including a fracture of the medial tibial plateau and the medial femoral condyle [10].

We reported on a case involving a 58-year-old female with a previous diagnosis of osteopetrosis undergoing TKA using patient specific instrumentation (MicroPort, Arlington, TN). In preparation for this case, which included a review of the literature, it was decided that patient specific instrumentation, involving minimal drilling into the bone, would be most advantageous due to its sclerotic nature. Additionally, adequate instrumentation was made available for the case, including extra saw blades, drill bits, and batteries, along with fracture trays, appropriate staffing, and scheduling of the OR suite to reflect anticipation of a long operative time. The measures were undertaken to mitigate potential difficulties in cases previously reported in the literature.

Despite these anticipatory measures, the case was still complicated by a long operative time, difficulty with bone preparation, and multiple intraoperative fractures. Nonetheless, at five months post procedure, the patient appears to be doing well. Intraoperative fractures appear to be healing appropriately on imaging and implants are stable without evidence of loosening. The patient reported a decrease in pain and stiffness with ambulation and appears to be satisfied with the results.

Conclusion

Performing a TKA on a patient with osteopetrosis can be fraught with difficulty. However, in this case, the benefits to the patient outweighed the risks. The decision to proceed with TKA in these patients should be one of last resort and only be made with careful consideration of the risks involved. It is essential to manage patient expectations and provide thorough patient education regarding expected outcomes of this procedure.

Based on our experience, as well as previous cases reported in the literature, we would recommend the following when performing a total knee arthroplasty on a patient with osteopetrosis.

It is important that the patient be well informed of the increased risk of complications as a result of their diagnosis of osteopetrosis, and they should also have their expectations regarding postoperative outcomes carefully managed. It is important to undertake good pre-operative planning, and we would recommend the use of patient-specific, extramedullary cutting guides obtained prior to the operation, as it would have been exceedingly difficult to prepare the medullary canal in our patient. We would also recommend that the surgeon be cognizant of longer than normal case lengths and ensure adequate OR staffing. Bone preparation was quite difficult in our case, and we would recommend adequate instrumentation be present including the availability of extra saw blades, batteries, drill bits and burrs. In addition, intraoperative fractures seem to be a frequent complication reported in the literature, and our case was no exception. We recommend having readily available fracture trays to possibly deal with any such occurrences. Finally, we would recommend more vigilant follow up of this patient population including shorter follow up intervals, frequent postoperative radiographs, and continuing management of patient expectations.

Although we have created a list of recommendations based off our experience as well as previously reported cases in the literature, it is important to recognize the relative paucity of case reports that exist on performing TKA in patients with osteopetrosis. Therefore, we feel it is important to continually re-examine how and when we perform total knee arthroplasty in patients with osteopetrosis based on continual expansion of our collective knowledge on the subject.

Conflicts of Interest
No authors report conflicts of interest in regard to this case report.

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Bibliography
