Case Report

Total Knee Arthroplasty in Osteopetrosis Using Patient-Specific Instrumentation

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Abstract: Osteopetrosis is an uncommon endocrine disease characterized by defective osteoclast resorption of bones. This causes a hard, sclerotic, and brittle bone throughout the skeleton. Fractures and unforgiving subchondral bone are common in this condition, both of which can lead to osteoarthritis. Total knee arthroplasty is often the treatment of choice but presents challenges due to the hard and sclerotic bone present throughout the metaphysis and diaphysis of the femur and the tibia. We present a case of knee osteoarthritis in a patient with osteopetrosis who underwent total knee arthroplasty using patient-specific instrumentation. This technique eliminates intramedullary alignment and minimizes drilling, reaming, and saw passes, making it attractive in the setting of diseases such as osteopetrosis to decrease operative time and potential complications. **Keywords:** total knee arthroplasty, osteopetrosis, patient-specific instrumentation.

Osteopetrosis, also known as marble bone disease and Albers-Schonberg disease, is an uncommon, heritable skeletal condition first described in 1904 by Dr Albers-Schönberg, a German radiologist [1,2]. In most forms, it is characterized by defective osteoclasts, which are unable to form the acidic, ruffled border necessary to resorb calcified cartilage during childhood and bone during adulthood. This results in an inhibition of bone turnover and remodeling, leading to bones made of disorganized calcified cartilage with immature, thickened trabeculae and cortices. Despite its thickened and sclerotic nature, it is substantially weaker and more brittle than a normal bone. Disruption of bone metabolism results in clinical manifestations including multiple recurrent fractures, osteoarthritis, osteomyelitis, and obliteration of the medullary cavity [1,2]. The 3 main types of osteopetrosis are classified as infantile malignant, intermediate, and benign. The malignant autosomal recessive form of this disease results in death in the first decade of life. The intermediate autosomal recessive form carries a life expectancy into adulthood and has the highest incidence of osteomyelitis of the jaw. The benign autosomal dominant form is the most common and typically carries a full life expectancy, however, with increased propensity for fractures and other musculoskeletal problems. Radiographic traits of osteopetrosis include increased bone density, osteosclerosis, metaphyseal widening, endobone formation, and skull-base and vertebral endplate thickening [3].

Degenerative osteoarthritis is a commonly encountered complication of long-standing osteopetrosis [4]. Total knee arthroplasty (TKA) remains a good option for these patients, but there are unique challenges even during what would otherwise be a routine procedure due to replacement of cancellous bone with hard, sclerotic trabeculae and cortical bone and, therefore, obliteration of the cancellous bone of the medullary canal and the metaphyseal regions. These challenges consist of reaming of the femoral canal, drilling for multiple pins to secure jigs, and challenging saw resections of the distal femur and proximal tibia. These obstacles can lead to malpositioned components and inadequate surfaces for cement interdigitation. To circumvent these obstacles, surgeons have used external alignment guides, customized implant components with smaller stems, and computer-assisted navigation techniques and have reported successful outcomes in short- and medium-term follow-up in several case reports

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[5-7]. In each of these techniques, the intraoperative goal was to minimize the amount of drilling, reaming, and saw passes while still achieving appropriate alignment and ingrowth surfaces for the components. We report the use of patient-specific instrumentation (PSI; Zimmer, Warsaw, Ind) for TKA in an osteopetrosis patient as another alternative to minimize these difficult and time-consuming steps. PSI uses a preoperative long-axis magnetic resonance imaging (MRI), which is used to manufacture unique cutting jigs specific to a patient's femoral and tibial anatomy through the use of rapid prototyping technology (Fig. 1A-C). This allows the surgeon to use one set of jigs that are predesigned to contour to the patient's distal femur and proximal tibia to perform the appropriate femoral and tibial resections. This eliminates the need to ream the intramedullary canal for an alignment guide, minimizes the number of femoral and tibial pins necessary for securing cutting jigs, and reduces the number of saw blade passes because the guide is fabricated for a predetermined resection. Short-term results from a feasibility trial and case series using a similar approach for routine TKA suggest that the procedure achieves the same level of precision in component placement as the conventional TKA technique [8].

Case Report

We report on a 58-year-old man with osteopetrosis who presented to the orthopaedic clinic at our institution with bilateral, right greater-than-left, knee pain. His pain and effusions had been persistent for several years, beginning during a period of increased physical activity. He had previously been evaluated by an outside orthopedist who initially suspected meniscal pathology and, after an MRI, recommended right knee arthroscopy. At the time of surgery, degenerative meniscal tears, as well as extensive lateral compartment chondral loss, was noted. He was subsequently managed with aspirations for recurrent effusions and steroid injections, both of which provided short-lived relief. He was referred to our institution for consideration of TKA, given his diagnosis of osteopetrosis. At the time of our initial evaluation, he was noted to have a partially correctable clinical valgus deformity and a 7° flexion contracture with active flexion to 120°. He had no neurovascular compromise in either lower extremity. On plain film imaging, approximately 10° of overall valgus deformity with mild osteophyte formation and mild lateral joint space narrowing was observed (Fig. 2A). Because of his localizing symptoms and history of failed conservative management in the face of known full-thickness chondral lesions by arthroscopy, a TKA was recommended. His history of osteopetrosis caused concern for challenges with intramedullary guide rod placement for positioning of femoral component cutting jigs, placement of the tibial component keel, drilling for pins, and the possibility of multiple resections for soft tissue balancing. It was felt that PSI was a good option in this case. After the risks and benefits of the procedure were explained to the patient, he was consented for TKA using PSI. He underwent a routine MRI for preoperative fabrication of custom-cutting blocks (Figs. 1B, C) and was taken to the operating room where a right TKA was performed using a standard medial parapatellar approach. Zimmer Gender Solutions NexGen High-Flex Knee Implants and posterior cruciate ligament–sacrificing NexGen High-Flex Knee Implants were used. The implanted components consisted of a size 4 gender-specific male femur, size 3 tibia, and 9-mm ultracongruent high-molecular-weight highly cross-linked tibial polyethylene insert. Despite the use of PSI, there was difficulty in drilling even for the limited numbers of pins

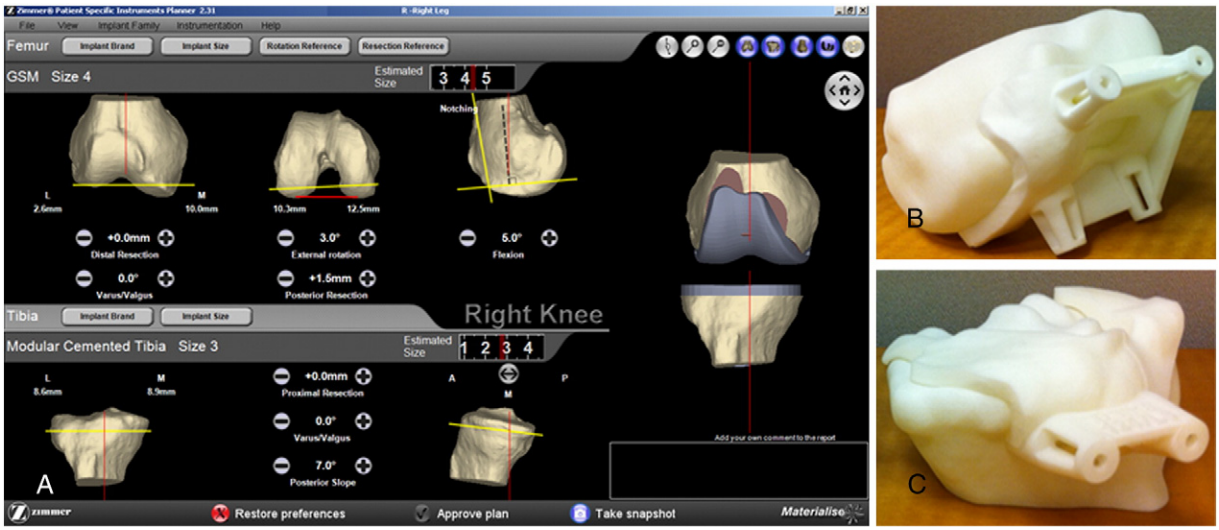

Fig. 1. (A) Screenshot of the PSI planning software for cutting jig design. Completed patient-specific cutting jigs fabricated for our patient's distal femur (B) and (C) tibial plateau.