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Arthroplasty in patients with rare conditions

Total knee arthroplasty in osteogenesis imperfecta

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ABSTRACT

Osteogenesis imperfecta is a genetic disease resulting in abnormal collagen formation, with multiple clinical manifestations. Advancements in medical and surgical treatments have prolonged the life expectancy of these patients in recent decades. As a result, orthopedic surgeons are likely to be faced with the challenge of performing arthroplasty in these patients on a more frequent basis. Here, we describe a patient with osteogenesis imperfecta and subsequent severe osteoarthritis prompting primary total knee arthroplasty. This rare case presents an opportunity to explore special considerations unique to this patient population, including comorbid bone defects, the need for using extramedullary guides, careful alignment of prostheses to accommodate abnormalities in limb axes, and equipment utilization.

Introduction

Osteogenesis imperfecta (OI) is a metabolic bone disease caused by errors in collagen I synthesis due to mutations in COL1A1 or COL1A2 genes, resulting in abnormally fragile bone [1,2]. Current estimates suggest the prevalence of OI in the United States to be approximately 8 people in 100,000 [3]. There are currently sixteen recognized types of OI, each due to different genetic mutations and clinically manifesting with differing symptoms and degrees of severity [2,4]. Owing to the abnormal molecular characteristics of collagen, there is an increased risk of periarticular and diaphyseal fractures that can alter joint mechanics in OI patients, which translates to a propensity for both primary and posttraumatic arthritis [5-7]. Although OI type II is incompatible with life, other types may have a normal or near-normal life expectancy [8]. Advancements in fracture management, arthroplasty, and treatment for metabolic bone diseases will likely help to extend the lifespan for this group and may lead to more adults with posttraumatic and degenerative osteoarthritis, which may lead to a more frequent use of joint replacement in this population [6,9].

Very few cases of total knee arthroplasty (TKA) in adult patients with OI have been reported to date. To our knowledge, this is only the tenth published case. In this article, we describe the clinical course of an OI patient who elected to undergo TKA, review the available literature, and present technical considerations for managing this rare scenario.

Case history

A 69-year-old female with a history of OI presented to the clinic for evaluation (Fig. 1). Her chief complaints were left knee pain and knock-knee deformity with the knee caving in during ambulation. She also complained of a leg length discrepancy, for which she wore custom orthotics. She felt that her left knee discomfort had progressed to a degree warranting surgical intervention and thus sought consultation to proceed with TKA. Her surgical history included right hip resurfacing 40 years before this presentation and multiple surgeries for lower extremity fractures. At the time of presentation, she was 152-cm tall with a body mass index of 22.6. Although her OI subtype was not known to us, we assumed she has OI type I given her notably blue sclera. This patient was wearing orthotic shoes with a remarkable right foot elevation. The left lower extremity had 25 degrees of genu valgus of the left knee, with a significant valgus thrust on ambulation. Her deformity was passively correctable to a neutral coronal alignment, and she could range her knee from full extension to 130 degrees of flexion.

Radiographs obtained at this visit demonstrated surgical hardware in the right femur, which was grossly shorter than the left. The left knee radiographs revealed bone-on-bone degenerative changes of the lateral compartment with severe genu valgum. The tibia also showed medial bowing without remarkable deformity in the sagittal plane. Imaging also revealed gross osteopenia (Fig. 2).

After discussing the risks and benefits of the surgery, the patient elected to proceed with left TKA. Her severe valgus deformity made

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the knee very unstable, putting her at tremendous risks for falls, which would have catastrophic consequences for someone with similar systemic disease. The patient was agreeable to this plan, and informed consent was obtained for both surgery and the publication of her case as medical literature.

Preoperative templating revealed that standard implant sizes would suffice, and custom implants would not be necessary. The tibia deformity precluded the use of intramedullary guides for the tibia cut but was distal enough to allow the use of standard tibia reamers proximally. The femur was not significantly deformed in a way that would preclude the cautious use of intramedullary guides. There was minimal radiographic evidence of patellofemoral arthritis. The chronic genus valgus deformity led us to assume there would be ligamentous laxity, prompting us to have multiple levels of constraint available to make decisions based on operative findings. Given her poor bone quality, we also planned to use a small tibial stem extension to enhance fixation while avoiding the fracture risk from the use of a larger stem.

On the day of surgery, a medial parapatellar arthrotomy was created to expose the knee joint. Upon entering the knee, the operative team identified significant cartilage wear. The anterior cruciate ligament was completely absent, and the posterior cruciate ligament was significantly attenuated. There was severe wear of the posterior lateral aspect of the tibia with complete eburnation of the lateral femoral condyle. The remnants of the cruciate ligaments were debrided, and a retrograde intramedullary rod was placed in the femur. A flexible rod was used to minimize the risk of iatrogenic fracture during this step. After making the distal femur and chamfer cuts, an osteopenic change was identified in the medial femoral condyle, juxtaposed with dense, sclerotic bone in the lateral condyle. Rotation of the femur was determined with reference to Whiteside's line given her significant femoral dysplasia.

Attention was then turned to the tibia, and an extramedullary guide referenced to the second ray to guide axial alignment was used, with 3 degrees of posterior tibial slope factored into the cut. Given the patient's osteopenia, a short stem extension was selected to augment fixation in the distal metaphysis. The shaft was reamed proximally without complications. After final preparations of the joint and placement of trial components, the knee was extended and the patella everted and was denervated circumferentially. Patellar osteophytes were removed, and the edges were smoothed. There was minimal cartilaginous wear on the patella, and the operative team opted to retain the native patella to reduce the potential risk of a postoperative fracture.

At this point, the trial components were replaced with the final prosthesis. The knee was put through a range of motion of 0–130 degrees, demonstrating smooth patellar tracking. Although the native MCL was retained, it has been stretched out over time due to her valgus deformity, prompting the selection of a constrained posterior stabilized implant to create additional intrinsic stability within the construct. A fixed hinge was not necessary. After placement of the constrained posterior stabilized implant, there was no further valgus laxity. No soft tissue releases were necessary to balance the flexion or extension gaps (Fig. 3).

At the patient's 6-week follow-up appointment, she reported complete resolution of her left knee pain and ambulated independently. Follow-up AP (Fig. 4a), lateral (Fig. 4b) radiographs were obtained. She had a nonpainful range of motion from 0 to 130 degrees of flexion, and the valgus angulation and thrust noted during preoperative examination had been completely eliminated. At that time, a new pair of custom shoes was then measured and obtained, to accommodate her newly neutral left limb alignment, further improving her capability for gait and balance, and provide support for her ankle joints. At 4 months, an updated standing long leg film was obtained with her new shoes in place to assess limb length and alignment (Fig. 4c). At that time, she was walking progressively and bearing weight with the help of a cane. She had no pain, full range of motion, and was performing all desired activities of daily living.

Discussion

In this article, we present a patient with the diagnosis of OI who underwent TKA. Such a case requires several special considerations. If possible, the patient's operative anesthesia should be epidural or regional block, thereby avoiding general anesthesia. This will

Figure 1. Preoperative clinical photographs demonstrating severe valgus deformity.

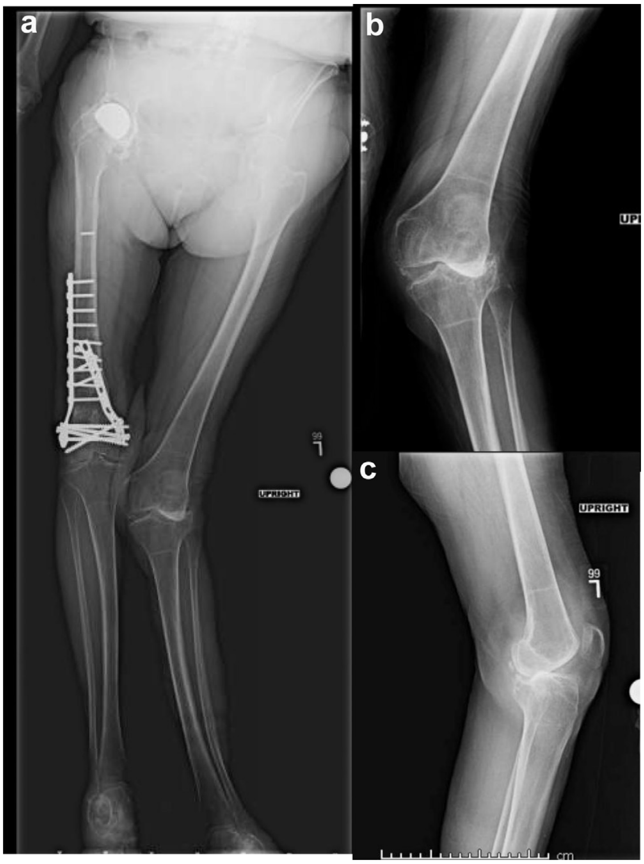

Figure 2. Preoperative bone length study of the bilateral lower extremities (a), anteroposterior radiograph of the left knee (b), and lateral radiograph of the left knee (c).

decrease manipulation of the cervical spine and risk for iatrogenic cervical fracture. If general anesthesia cannot be avoided, the surgeon should consider cervical spine imaging to evaluate for subclinical fracture or instability. Papagelopoulos et al. were among the first to publish considerations for arthroplasty in patients with OI. They offered several recommendations to minimize the risk of iatrogenic fracture while preparing for the operation, including ruling out subclinical cervical spine fractures or instability in

Figure 3. Immediate postoperative (a) anteroposterior and (b) lateral radiographs.

patients undergoing general anesthesia. [6] They also suggest using a well-padded operating table to minimize the potential of other iatrogenic fractures during positioning or trialing components [6]

A thorough evaluation of a patient's mechanical axis and angular deformities is especially useful in this population. These patients may have a history of extremity fractures that distort the limb mechanical axis more severely than that of the general population, and full-length standing Radiograph will help guide planning to restore a normal mechanical axis. As many as 46%–86% of patients have anterolateral bowing of the femur, and 27%–86% will have anterior bowing of the tibia [6]. Nishimura et al. published a case report of a total knee replacement in a patient with OI in 2008. In their patient, a tibial shaft deformity hindered the use of intramedullary guide rod, and thus, an extramedullary device was used. Despite the tibial deformity, the mechanical axis of the extremity still transected the middle of the knee in the coronal plane, negating the need for excessive medial or lateral soft tissue release. Nevertheless, Nishimura stated that extra-articular deformities may prevent the use of traditional intramedullary guides and should be recognized preoperatively [7]. In addition, the use of standard intramedullary guides in the relatively narrow canal may predispose to iatrogenic fractures even in the absence of significant long bone angulation. It is important to note that the presence of femoral rotational deformities may render a posterior condylar or epicondylar referencing guide inaccurate. In those cases, the femoral cut should be made in a fashion perpendicular to the anteroposterior axis of the femur to adequately restore knee kinematics. Alternatively, the surgeon may elect to perform the tibia cut first, followed by a gap-balancing technique to aide in accurate femoral component alignment.

In addition, such deformities must be taken into account when performing bone resections. Significant deformities may be corrected either extraarticularly or intraarticularly. Wang's retrospective case series demonstrated that the mechanical axis of the knee can be improved by TKA using juxtaarticular bone resection and soft tissue balancing in patients with extraarticular deformities [10]. However, the patients in this series had extraarticular deformities as a result of fracture malunions, and the axis distortions likely lacked the complexity of patients with OI, whose deformities often result as a combination of bowing, numerous traumatic fractures, and the accumulative result of multiple subclinical fractures. Wolff et al. also discuss the effect of extraarticular and valgus deformity on TKAs and present several key points that should be taken into consideration when planning a surgery for OI [11]. For example, the closer the deformity is to the joint, the greater the impact on knee mechanical axis. In addition, an intraarticular correction of varus malalignment produces lateral instability, which is better tolerated than medial instability in most patients. Finally, tibial deformities are easier to correct with intraarticular resection than femoral deformities, as compensatory femoral resections only affect the extension gap [11]. For patients with especially small bone structure or large deformities, computer-aided navigation or custom cutting blocks may be necessary to restore an acceptable axis [12]. Although careful intraarticular wedge resections can compensate for some extra-articular deformities, extraarticular osteotomies may be necessary for some patients. Wagner et al. presented two patients with three primary knee arthroplasties in which cases extraarticular deformities resulted in a distorted mechanical axis. They treated these patients with femoral and tibial osteotomies performed at the time of primary joint replacement. Although both the patients had satisfactory long-term outcomes, they both experienced complications at one or more osteotomy sites requiring revision fixation [13].

Another consideration is the sizing of the prosthetic components for this population. Patients with OI frequently experience growth retardation, and standard implants may be oversized in these patients. Therefore, the surgeon must perform preoperative digital

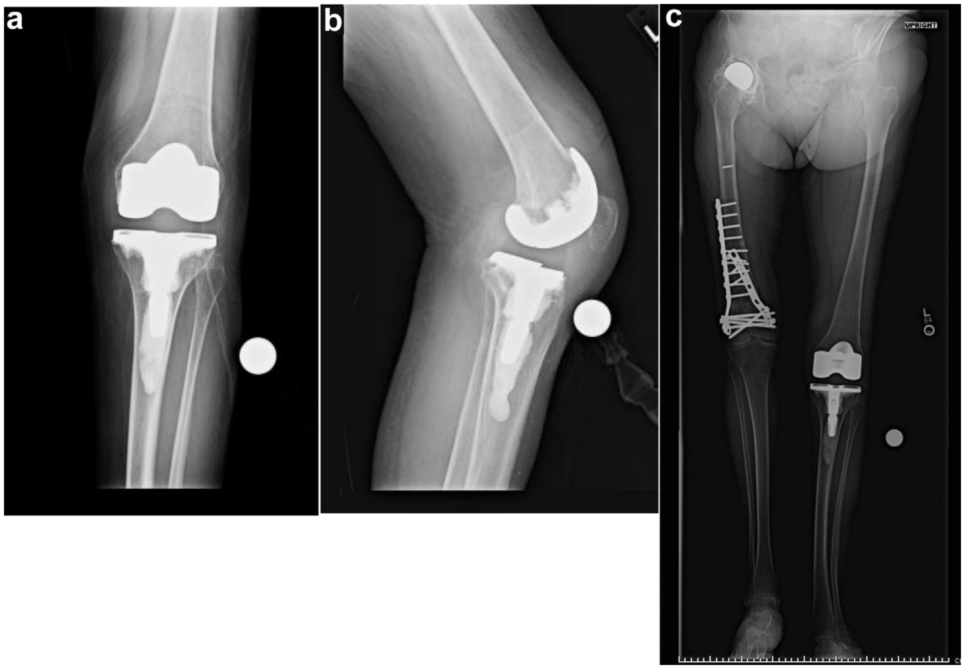

Figure 4. Six-week postoperative anteroposterior (a) and lateral radiographs (b); 4-month postoperative weight-bearing bilateral lower extremity radiograph (c).

templating to ensure that an appropriate range of implant sizes are available intraoperatively to allow for appropriate component selection once bone cuts are performed [5]. Again, custom implants and cutting guides based on preoperative templating may be of benefit for patients with an especially small skeletal structure.

In patients with severe coronal deformities, the lateral or medial soft tissues are often lax on the convexity of the deformity. Patients with OI are likely to have chronic coronal deformities. The long-lasting excessive stretch on collateral ligaments may cause these structures to lose most of their elasticity. Such soft tissue imbalance should be recognized and addressed at the time of surgery. Although mild deformities may be stabilized by addressing soft tissues alone, severe deformities of greater than 20 degrees, such as in this patient, should prompt consideration for the use of some degree of implant constraint [14].

OI patients are at an inherently increased risk for fractures. The comparatively lower thickness of the patella combined with osteopenia may place the OI patient at an increased risk of patellar fracture if resurfacing is used. The surgeons involved in the aforementioned case noted minimal degenerative changes to the patellar facets and thus opted not to resurface the patella. The same decision and rationale was presented by Nishimura et al. [7]. When there are minimal arthritic changes to the patella, resurfacing should be avoided to minimize the risk of patellar fracture in the postoperative period. The rate of patellar fracture in all patients after TKA is approximately 0.7% [15]. To our knowledge, no data exist on the incidence of patellar fractures in patients with OI. However, one can infer that this disease process likely increases the risk of this catastrophic complication after patellar resurfacing. In addition, the use of activity restrictions and walking aides for a prolonged period of time may be appropriate for this population. The risk of fall or periprosthetic fracture in patients with osteopenic OI bone is intrinsic, and any such injury would likely be catastrophic in this population. In the present case, the patient did experience a type C periprosthetic fracture of her distal femur after her right hip resurfacing procedure, so an extremely conservative postoperative course was encouraged to help maximize her safety and gradually increase her activity levels over the 6 months after the procedure.

Current controversies and future considerations

Given that there is so little literature pertaining to arthroplasty in OI, many controversies exist. At this time, there is insufficient literature to determine if it is advantageous to address extra-articular deformities before or after arthroplasty, at the expense of the costs and risks associated with multiple procedures in the operating room. Additional data should be obtained to determine the risks and benefits of extraarticular osteotomies in conjunction with primary arthroplasty vs staged procedures. It would also be helpful to have more data to direct postoperative care. For our patient, the history of periprosthetic fracture during the recovery period of a prior procedure provoked extreme caution in returning to baseline ambulation. Further research on postoperative care in the OI population would help assist surgeons in making a decision toward weight bearing and other precautions at the expenses of temporarily decreased ambulation.

Although our patient was a candidate for standard-size implants and cutting guides, this may not be true for all OI patients. There is ongoing research exploring the use of custom cutting blocks and navigation assistance. The role of these tools has not yet been clearly delineated, but they may potentially be useful for OI patients with severe extraarticular deformities or abnormally small extremities [16,17]. Stem extensions may be helpful to restore function in patients with significant joint destruction or prevent loosening in patients with a significant angular deformity [18,19]. However, further data to help analyze the costs and benefits of extended stems are still needed.

Summary

Advances in medical and operative care for patients affected by OI will undoubtedly lead to improved survival later into adulthood, and an increased incidence of joint replacement is likely to be seen in these patients. Although there are few published examples of arthroplasty in the OI population, this disorder warrants additional precautions and planning in the perioperative periods. One should take special care to protect the patient from iatrogenic injuries by carefully preparing the operating table, monitoring for cervical spine injuries if using general anesthesia, and hypervigilance regarding extraarticular deformities. In addition, the physician should avoid patellar resurfacing when patella changes are felt to have minimal attribution to the patient's symptoms. The surgeon should be prepared to template-use smaller components as needed, and he or she may need to consider extramedullary guides to determine anatomic and mechanical axes. As reported in this article, chronic abnormalities in the coronal plane may result in ligamentous laxity, which is best treated with the option for partially or fully constrained prosthetic components.

Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.artd.2018.09.006.

KEY POINTS

- Use extreme caution when templating and preparing the patient for surgery. Extraarticular deformities may preclude the use of intramedullary guides or other devices a surgeon may otherwise be accustomed to use. Custom implants may be required in certain patients.
- Extraarticular deformities should also prompt the operating surgeon to consider alterations in the patient's mechanical and anatomic axes, which may require bone resections and prosthetic placement, which differ from total knee arthroplasties for primary osteoarthritis. Certain patients may require osteotomies to improve alignment.
- Patients with osteogenesis imperfecta are likely at a higher risk to experience osseous (iatrogenic intraoperative fracture, postoperative periprosthetic fracture, and so forth) complications after the surgery, and the care team should be hypervigilant in monitoring for such events.

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