Total Knee Arthroplasty in a Patient With Thrombocytopenia-Absent Radius Syndrome

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abstract

Thrombocytopenia-absent radius (TAR) syndrome is a rare genetic condition with a complex inheritance pattern. This syndrome is classically characterized by hypomegakaryocytic thrombocytopenia as well as bilateral absent radii, shortened ulna, and radially deviated 5-digit hands. During infancy, the predominant manifestations are hemorrhagic complications. Later in life, the bleeding disorder typically improves, but the musculoskeletal abnormalities become of greater concern because of the effects on quality of life. Although the classic musculoskeletal manifestations of TAR syndrome involve the upper extremity, multiple lower-extremity abnormalities have been described, especially dysplasia of the knee. Knee abnormalities include genu varum, varying degrees of laxity or stiffness, patellar abnormalities, concave distal femur, convex medial tibial plateau, and/or absence of the anterior cruciate ligament and posterior cruciate ligament. Several management strategies for lower-extremity abnormalities in TAR syndrome have been described, especially for pediatric patients. Management strategies have not halted the natural progression of knee disease in these patients, and the effect that these knee abnormalities have in adulthood is unclear. Management of knee abnormalities in adults with TAR syndrome is poorly described in the current literature. The authors report a 59-year-old patient with TAR syndrome and knee abnormalities who underwent successful total knee arthroplasty. The patient was followed to the 3-year postoperative visit. At various postoperative time points (7 weeks, 6 months, 1 year, and 3 years), Knee Society Scores and 12-Item Short Form Health Survey scores were recorded. Radiographs obtained at each clinical visit showed well-positioned, well-fixed components. The authors concluded that total knee arthroplasty may be a safe and effective surgical intervention for adults with TAR syndrome and associated knee osteoarthritis.
Thrombocytopenia-absent radius (TAR) syndrome is a rare genetic condition with a complex inheritance pattern that has been associated with 1q21.1 microdeletions. Although the association between thrombocytopenia and bilateral absence of the radii had been described in scattered case reports as early as 1929, it was first described as a distinct syndrome by Hall et al in 1969. It is classically characterized by hypomorphic karyocytic thrombocytopenia, bilateral absent radii, shortened ulna, and radially deviated 5-digit hands. During infancy, the predominant manifestations are hemorrhagic complications. Later, the bleeding disorder typically improves, but the musculoskeletal abnormalities become of greater concern because of the effects on quality of life. Although the classic musculoskeletal manifestations of TAR syndrome involve the upper extremity, multiple lower-extremity abnormalities have been described, specifically, dysplasia of the knee. These abnormalities were well documented by Schoenecker et al in 1984.

In a case series of 21 patients with TAR syndrome, 18 had deformities of the knee, including genu varum, varying degrees of laxity or stiffness, patellar abnormalities, concave distal femur, convex medial tibial plateau, and/or absence of the anterior cruciate ligament (ACL) and posterior cruciate ligament (PCL). In most cases described, abnormal laxity of the knee joints was found; however, rare cases of knee stiffness have been described. Similar anatomic abnormalities have been observed in subsequent case studies and case series. Various surgical and nonsurgical strategies for managing the manifestations of knee dysplasia in patients with TAR syndrome have been described. However, no treatment modalities have been shown to consistently slow the natural progression of the disease. In particular, among patients who chose surgical interventions, such as childhood osteotomy with or without combined soft tissue procedures to correct genu varum, most had a recurrence of varus deformity. This is believed to be related to the challenge of surgically correcting the intra-articular deficiency of the medial femoral condyle through osteotomy, leading to recurrence of the varus deformity. Many patients later required secondary tibial or femoral osteotomy. Importantly, this progression of varus instability in skeletally immature children was self-limited and tended to stabilize after skeletal maturity and into adulthood.

Because of the wide variety of anatomic deformities associated with TAR syndrome, no consensus has been reached about the ideal treatment for lower-extremity abnormalities. In a case series of 21 patients by Schoenecker et al, most patients elected surgical interventions, whereas in a case series by McLaurin et al that included 23 patients who were treated at a childhood amputee center, most patients elected nonsurgical interventions, such as bracing, casts, and adaptive devices, such as walkers and power wheelchairs. Regardless of the intervention, in both case series, knee abnormalities tended to worsen progressively until skeletal maturity was reached. Furthermore, most of the literature on TAR syndrome focused on pediatric patients, and follow-up rarely extended past childhood. No studies have examined knee pathology in adult patients with TAR syndrome. It is unknown whether the bony and soft tissue abnormalities typical of TAR syndrome predispose patients to osteoarthritis in late adulthood. Likewise, there have been no reported cases of total knee arthroplasty (TKA) in patients with TAR syndrome. The authors present the case, along with 3-year follow-up, of a 59-year-old woman with TAR syndrome who underwent successful right TKA at the authors’ institution.
History and Physical Examination

An ambulatory 59-year-old woman with well-documented TAR syndrome presented in 2010 with progressive bilateral knee pain that affected quality of life and activities of daily living. Conservative management of knee pain, including the use of a cane for ambulation and intra-articular steroid injections, was unsuccessful. Pertinent surgical history included bilateral tibial osteotomy for correction of genu varum at 3 years old and left foot triple arthrodesis at 25 years old. On physical examination, the patient walked with a significant antalgic gait. There was obvious deformity of the bilateral upper extremities. Crepitus and discomfort in the knees occurred with flexion and extension. Range of motion was 5° to 110° bilaterally. No laxity was recorded in the medial-lateral or anterior-posterior plane.

Imaging

Five-view radiographs of both knees obtained at the initial visit showed bilateral complete loss of medial and lateral joint space, loss of patellofemoral joint space, osteophytes, sclerosis, and subchondral cyst formation (Figure 1). Bilaterally, the distal femur and proximal tibia were grossly abnormal in shape. The articular surface of the distal femur was concave and saddle-shaped, and the articular surface of the proximal tibia was characterized by a convex, dome-shaped contour and an underdeveloped intercondylar eminence (Figure 1A). Slight genu valgum, significant coxa valga, and apex-medial bowing deformity of both tibias were noted on long-standing films (Figure 1B). The right patella was subluxated laterally on sunrise view (Figure 1C). Noncontrast computed tomography (CT) scan of both knees was also performed for presurgical evaluation and planning. The CT scan was evaluated by the Custom Implant Department of DePuy Synthes (Warsaw, Indiana). The findings confirmed that standard implants could be inserted. A previous magnetic resonance imaging scan report was reviewed and showed bilateral absence of the ACL and PCL and probable absence of the medial menisci.

Surgical Procedure

Because of the significant anatomic abnormalities around the knee joint, custom components, custom cutting blocks, and computer-assisted alignment methods were considered. Preoperative radiographs and CT scan were evaluated by the Custom Implant Department at DePuy Synthes to determine whether to use custom vs standard components and cutting blocks and to assess the implant position. This custom evaluation led the primary surgeon (E.L.S.) to determine that computer-assisted alignment was not necessary for ideal alignment.

A standard medial parapatellar surgical approach was performed to expose the knee joint. On joint entry, marked dysplasia of the proximal tibia and distal femur was noted. Specifically, the articular surface of the tibia was dome-shaped, and the distal femur was concave, with bone loss centrally and retention of the lateral and medial epicondyles (Figure 2). The ACL, PCL, and menisci were absent. Because of the anatomic deformities, the tibia was prepared first. An extramedullary tibial alignment guide at 0° posterior slope was used for the tibial resection. The surgeon resected 10 mm of bone with an oscillating saw, which allowed clearance of the posterior portion of the femur. The distal femur, proximal tibia, and patella were osteotomized in the normal fashion and fitted with the appropriate components. Intramedullary guidance was used for the femur set for 5° valgus. The components inserted included a size 1.5 posterior-stabilized PFC Sigma femoral component (DePuy Synthes), a size 1.5 modular tibial base plate (DePuy Synthes), an 8-mm polyethylene insert (DePuy Synthes), and a 32-mm patellar component (DePuy Synthes). The knee was tested, and full extension was obtained with stability to varus and valgus stress testing. The wound was closed with 2 drains placed deep to the arthrotomy. Total tourniquet time was 89 minutes. The patient received 1.2 L crystalloid fluid intraoperatively and had minimal blood loss. No complications occurred intraoperatively. Preoperative hematocrit was 39.2%. Hematocrit was 37.3% in the immediate postoperative period, 29.4% on postoperative day 1, and 31.4% on discharge from the hospital. The patient required no transfusions preoperatively, intraoperatively, or postoperatively.
Hospital Course

The postoperative course was complicated by fever and an elevated white blood cell count on postoperative day 3. The cause was found to be *Clostridium difficile* infection. The patient was treated with a 14-day course of oral vancomycin. The rest of the hospital course was unremarkable, and the patient was discharged to a short-term nursing facility, with plans for routine postoperative follow-up.

Clinic Visits

At the 7-week postoperative visit, right knee radiographs showed a well-positioned and stable implant (Figure 3). Subsequent follow-up radiographs were taken at each clinical visit and continued to show a well-positioned, well-aligned, and stable implant, as evidenced by radiographs from the 6-month (Figure 4) and 3-year clinic visits (Figure 5). Additionally, at each clinic visit, Knee Society Score (KSS) and 12-Item Short Form Health Survey (SF-12) forms were completed.\(^9,10\) As evidenced by both KSS and SF-12 data, the patient continued to improve over the first year postoperatively. By 6 months postoperatively, the patient had improved from moderate occasional pain to mild occasional pain with walking and climbing stairs. She had progressed to needing a cane only when outdoors, and the KSS and the physical component score of the SF-12 improved. By 1 year postoperatively, the patient no longer used a cane to walk, could walk unlimited distances, and could transfer without assistance. Further, the KSS and the physical component score of the SF-12 continued to improve (Figure 6). Range of motion improved from baseline throughout the first year postoperatively. Final range of motion was 0° to 120°. At the 3-year follow-up visit, the patient had new-onset right hip pain that affected the ability to walk distances. The patient had mild dysplasia and degenerative joint disease of the hips bilaterally, as seen on dedicated right hip radiographs (Figure 7). However, clinically, the patient reported no pain in the groin or buttock, but noted isolated
pain overlying the right trochanteric region. The differential diagnosis included trochanteric bursitis, abductor tendinosis, and atypical pain from the degenerative changes of femoroacetabular articulation. Because the pain was primarily located over the trochanteric bursa, the patient elected to try steroid injection for presumed right hip trochanteric bursitis. The patient reported 100% relief of pain with this injection, and so the pain appeared to be primarily related to presumed right hip trochanteric bursitis. There are currently no plans for surgical intervention on either hip. The knee score component of the KSS continued to improve; however, the functional score declined secondary to decreased walking ability because of hip pain (Figure 6A). When an SF-12 score was obtained at year 3 postoperatively, the physical component score decreased and the mental component score increased slightly (Figure 6B).

**DISCUSSION**

The estimated prevalence of lower-extremity abnormalities in patients with TAR syndrome is 40% to 80%, based on previously published literature. The knee is the most commonly affected area. Cited abnormalities include genu varum, medial tibial torsion, abnormal laxity or stiffness of the joint, concave distal femur, convex medial tibial plateau, and hypoplastic or absent ACL, PCL, medial meniscus, and patella. The current patient showed many of these abnormalities, including a grossly concave distal femur and convex proximal tibia as well as absent ACL, PCL, and medial meniscus. The patient also had shallow intercondylar grooves on sunrise view radiographs, with subluxation of the patella on the right. Slight genu valgum and significant coxa valga were noted on radiographs. The patient reported a history of genu varum as a child that was corrected with bilateral tibial osteotomies at 3 years of age. Apex-medial tibial bowing was noted bilaterally, as evidenced by long-standing radiographs. Tibial bowing in the absence of surgical intervention does not appear in any of the literature on TAR syndrome. Thus, tibial bowing appears more likely to
be related to the bilateral childhood tibial osteotomies than to be a natural sequela of the disease process. Additionally, the patient showed evidence of severe degenerative joint disease, with bilateral complete loss of the joint space in all 3 compartments as well as osteophytosis, sclerosis, and subchondral cysts. Although no studies have examined the incidence of osteoarthritis in adults with TAR syndrome, it is reasonable to hypothesize that this patient’s anatomic abnormalities likely predisposed her to degenerative joint disease.

This patient met the indications for TKA, having severe degenerative joint disease that affected quality of life and activities of daily living and having undergone unsuccessful conservative management. The surgery was successful, with no intraoperative complications. Knee function over the first year postoperatively improved, as measured by both components of the KSS. At the 3-year follow-up visit, although the knee score component of the KSS improved, the functional score component was significantly lower. Concurrent hip pain, and thus the inability to walk long distances, was likely a confounding factor. Although the physical component score of the self-completed SF-12 survey also decreased significantly over the first year postoperatively, the patient appears to be doing remarkably well, with well-aligned stable components on follow-up radiographs and overall improvement in symptoms. Because of the successful outcome with right TKA and the similar anatomic abnormalities in the left knee, it would be reasonable to consider left TKA if the patient has symptomatic osteoarthritis in the left knee affecting activities of daily living and if conservative management is unsuccessful.

**CONCLUSION**

The authors reported the case of a 59-year-old woman with TAR syndrome who underwent successful TKA. She had a number of anatomic abnormalities of the knee that were evident both radiographically and intraoperatively. As measured by validated surveys, right knee function improved over the first year postoperatively. At year 3, mixed results on surveys were found, likely because of concurrent right hip trochanteric bursitis. Radiographs obtained at each clinical visit showed well-positioned and well-fixed components. The authors concluded that TKA may be a safe and effective surgical intervention for adults with TAR syndrome and associated knee osteoarthritis.

**REFERENCES**