Total Hip and Knee Replacement in a Patient with Arthrogryposis Multiplex Congenita

Kimberly A. Fisher, DO, MBA, and David A. Fisher, MD

Abstract

Arthrogryposis multiplex congenita (AMC) is a complex disorder that leads to joint stiffness and deformities in 2 or more joints in afflicted children. Late manifestations of this disorder can include secondary degeneration of the abnormal joints with arthritic symptoms of pain and loss of function. There are few reports in the orthopedic literature on the use of total joint arthroplasty to improve the pain and function in patients with arthrogryposis.

This case report presents one patient who underwent bilateral total hip and total knee arthroplasties for deteriorating function and pain in her hips and knees secondary to the congenital deformities created by arthrogryposis multiplex congenita. We discuss the intraoperative difficulties and techniques used to reconstruct her hips and knees, as well as the potential indications for joint arthroplasty in this challenging group of patients.

There are very few reports on the results of total joint arthroplasty in the treatment of arthrogryposis multiplex congenita (AMC) and concomitant arthritis of the hip or knee. We present a case of nongenetic arthrogryposis involving the hips and knees, and discuss the surgical reconstruction of hip and knee deformities in this patient. The patient provided written informed consent for print and electronic publication of this case report.

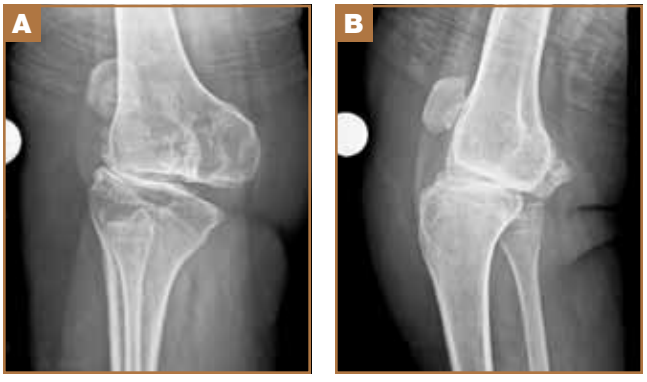
Case Report

A 38-year-old woman presented with bilateral knee pain with a history of congenital hip and knee dysplasia secondary to AMC. She underwent manipulations, splinting, physical therapy, and 11 surgical procedures as an infant and young child to improve her lower extremity joint function. She had also undergone a left total hip replacement 10 years previously for a painful and degenerated congenital hip deformity. Her upper extremity development was normal. At her initial presentation, pain had been present in both knees for over 5 years and was greatest in her left knee. She had intermittent swelling and buckling when walking. Pain occurred with weight bearing and at night. Venous insufficiency was present with ulceration of the left ankle, which had been present for 5 months. The patient was able to walk 1 to 2 blocks, used a rail when going up steps, had difficulty sitting and standing from a chair, and walked with a limp. She denied having had injections or previous intra-articular knee surgeries. Pharmacotherapy included occasional hydrocodone and diazepam. She was also undergoing physical therapy with limited improvement in pain or function. She denied using assistive devices to walk.

She had difficulty getting up from a chair, and walked with a waddling gait pattern. Upper extremities had full range of motion at the shoulders, elbows, wrists, and hands with symmetrical cervical spine rotation. Hips had decreased mobility with full extension and only 60° of flexion. Abduction of both hips was limited to 15° and rotation arcs were 30°. Hip motor strength was 4/5 on the right and 3/5 on the left in flexion and abduction. The right knee had 0° to 55° of flexion. The left knee had a 15° flexion contracture with 40° of flexion and a partially correctable 25° valgus deformity. Distally there was a 4 cm stasis ulcer on the medial side of the left ankle superior to the malleoli. Her pulses were intact, sensation was intact in both extremities, and 2+/4 reflexes were present in both lower extremities.

Radiographs of the knees demonstrated congenital abnor-

Figure 1. (A) Preoperative anteroposterior (AP) radiograph of the right knee. (B) Preoperative lateral radiograph of the right knee.



Authors' Disclosure Statement: Dr. DA Fisher wishes to report that he is a consultant with DePuy Orthopedics and has received royalties and research support from DePuy. The other author reports no actual or potential conflict of interest in relation to this article.

malities with flattening of the distal femoral condyles, large osteophytes, valgus deformity of the left knee with erosion of bone, hypoplastic trochlear grooves and patellae (**Figures 1A, 1B**). Radiographs of the pelvis and hips confirmed the presence of a left total hip with evidence of polyethylene wear and fibrous fixation of her cementless acetabular cup. The right hip had congenital dysplasia with secondary arthritic changes (**Figure 2**).

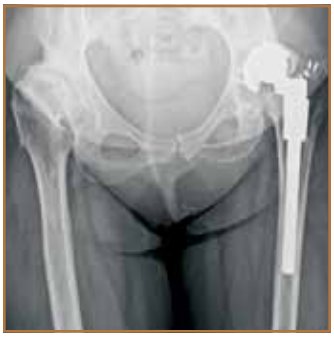


Figure 2. AP pelvis radiograph prior to right total hip arthroplasty.

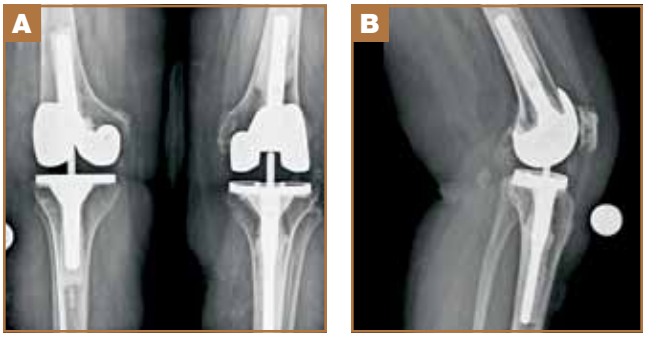


Figure 3. (A) Postoperative AP radiograph of bilateral total knees. (B) Postoperative lateral radiograph of left total knee arthroplasty.

Her options for management were discussed and we made the recommendation for a left total knee arthroplasty to correct alignment, relieve pain, and improve mobility. Less certain benefits included a possible improvement in range of motion and improved circulation with relieved venous stasis.

The patient underwent a left total knee arthroplasty. Because of the deformities and limited motion, the surgical procedure included a quadriceps snip to enhance exposure, followed by excision of the cruciate ligaments, downsizing of the femoral component to allow greater knee flexion, and a constrained Total Condylar 3 implant (Depuy Orthopedics, Warsaw, Indiana). Due to uncertainty about joint line placement and ligament releases and balancing of the deformed knee, we decided to use a more constrained articulating knee system. In addition, stem extensions were added to the femoral and tibial implants to aid in fixation and stress transfer. Our team used a continuous passive motion device postoperatively to assist range of motion. Postoperative range of motion improved from 0° to 90° of flexion and has been maintained over the following 8 years. The patient's pain level was greatly reduced, the venous stasis ulceration resolved, and her walking and transfer abilities were greatly enhanced.

She returned 2 years later to undergo a right total hip replacement for increasing pain and loss of function. The right hip replacement was notable for the congenital deformities present, including excessive femoral anteversion and acetabular dysplasia. The implants used included a modular femoral stem and a modular acetabular cup with a metal-on-metal insert bearing (S-ROM stem, Pinnacle cup, Ultimet liner, Depuy Orthopedics) (**Figures 3A, 3B**). While the right hip pain was resolved and her function improved, her postoperative range of motion remained limited with 0° to 60° of flexion, 40° of abduction, 25° of adduction, 30° of external rotation, and 20° of internal rotation.

She developed pain in the left hip secondary to polyethylene wear and acetabular cup loosening the following year and underwent a left acetabular cup revision. Postoperative left hip motion was 0° to 80° of flexion, 40° of abduction, 25° adduction, 40° external rotation, and 30° of internal rotation (**Figure 4**).

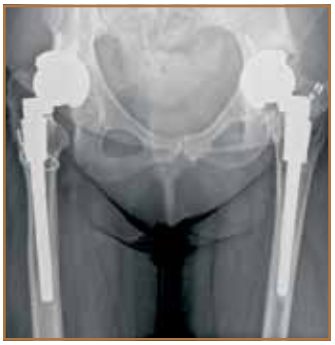


Figure 4. AP pelvis radiograph after right total hip and left acetabular cup revision.

Her most recent intervention was a right total knee arthroplasty performed 4 years after the previous operation. Her preoperative motion in that knee was 0° to 55° of flexion and her symptoms included increasing pain, decreasing walking ability, inability to climb steps, and frequent buckling of the knee. She again received a Total Condylar 3 knee prosthesis with similar intraoperative management as the left knee. At her 6-month visit, the patient could walk over 10 blocks without assistance, climb stairs with a rail, and had an active range of motion of 0° to 90° of flexion in both knees with good ligament stability.

After bilateral hip and knee arthroplasties, she is quite pleased with the functional improvement and relief of pain that she suffered prior to the reconstructions. She states her current status is "the most functional she has ever felt in her life," and she is able to complete all activities of daily living.

Discussion

Arthrogryposis or arthrogryposis multiplex congenita are descriptive terms used to define the physical finding of 2 or more joint contractures present at birth.[1] It affects a heterogenous group of individuals and actually consists of several hundred different disorders. Overall prevalence of arthrogryposis is 1 in 3,000 live births.[2-4] With limited movement of the fetal extremities in utero, fibrosis and ankylosis develop at multiple joints. Despite treatment in early childhood, patients may develop arthritic manifestations in their contracted and stiffened joints in adulthood. AMC is a physical limitation described within a broad range of complex disorders, including those with neurologic involvement and those without.[5] These groups can be further categorized into genetic versus nongenetic, based on syndromal findings, or extremity involvement.[6] Neu-

rogenic arthrogryposis is due to an abnormality in the central or peripheral nervous system and includes, but is not limited to, disproportions of muscle type as in Pierre Robin or Möbius syndromes, spinal muscular atrophy, dysgenesis of the central nervous system (associated with trisomy 18) and Zellweger syndrome, and dysgenesis of the anterior horn.[7] Individuals with arthrogryposis and a normal neurologic examination are affected secondary to fetal crowding, connective tissue disorders, distal arthrogryposes, or amyoplasia. Amyoplasia is the most common form and is characterized by a typical symmetric positioning of limbs.[8,9] Myopathic disorders include central core disease caused by a lack of oxidative activity within muscle fibers, nemaline myopathy diagnosed by the presence of nemaline rods within muscle fibers, congenital muscular dystrophy, and mitochondrial cytopathy. Our patient is an example of nongenetic, non-neurologic arthrogryposis likely secondary to mechanical restrictions.

The diagnosis of arthrogryposis is largely based on history and physical examination.[10] Limited joint movement on physical examination and nonspecific signs of muscle atrophy are keys to diagnosis. Deformities depend on etiology and are usually symmetric. Distal joints are affected more frequently than proximal joints. Joint dislocation is common. Lab tests can confirm genetic variations and connective tissue disorders. Skin biopsy can be done for fibroblast chromosome analysis. Genetic links exist for several forms of arthrogryposis, and muscle biopsy or electromyography can help detect muscular versus neuropathic involvement. Risk factors should be determined and can be used for preventive and therapeutic measures.

AMC is correlated with significant morbidity due to functional limitations and pain. Early treatment is centered around improving range of motion and functional positioning.[11,12] Hips and knees are often involved with dysplastic changes.[13-17] There is a high incidence of arthritis in adults secondary to joint dysfunction. Treatment options should be directed towards improving function and pain relief. Treatment options range from conservative therapies to surgical intervention and the treatment is varied depending on the breadth of system involvement. Treatment goals include lower-limb alignment and stability for ambulation and upper-limb function for self-care.

Patients who walk regularly and who develop secondary arthritis are at risk of losing their mobility. Consider medication, bracing, and assistive devices before surgical intervention. Total joint arthroplasty has not been widely promoted for AMC. Södergard[18] reported on 52 patients with AMC with hip involvement. Total hip replacements were performed in 3 hips with reportedly good results. Cameron[19] published a case report in 1998 on a 34-year-old man who underwent 2 total hip and 1 total knee replacements. Despite good motion obtained at the time of surgery, the joints returned to the preoperative state within 2 years and the author concluded that total joint arthroplasty offered little benefit for such patients. The only other report was by Leonard and Nicholson[20] in 2010 and involved a patient with AMC and an above knee amputation that underwent an ispilateral total hip replacement with good results.

From our experience, recommendations regarding total hip and knee arthroplasty in patients with AMC include the following: Patients should be skeletally mature community ambulators with intact neuromuscular function in the opposing muscle groups around the affected joint. Degenerative arthropathy must be demonstrated on plain radiographs. Pain and demonstrable loss of function should be evident. Upper extremities should allow transfer and ambulation with assistive devices to allow postoperative rehabilitation. Intact cognitive function and a high level of self-motivation are required.

There is a high incidence of arthritis in adults secondary to joint dysfunction. Treatment options should be directed towards improving function and pain relief.

Conclusion

Our patient is an example of a nongenetic, non-neurogenic form of AMC with congenital contractures that were limited to the lower extremities. She suffered from decreased functional activity and pain. Following bilateral total hip and knee arthroplasty, she enjoyed improved function and pain relief, while maintaining independence in mobility. This case study provides evidence of a successful outcome of total hip and knee arthroplasty in a skeletally mature patient with non-neurogenic arthrogryposis and secondary arthropathy.

Dr. Fisher (KA) is Resident, Ohio State University, Columbus, Ohio. Dr. Fisher (DA) is Assistant Clinical Professor of Orthopaedic Surgery, Indiana University School of Medicine, and Director, Total Joint Center, Indiana Orthopaedic Hospital, Indianapolis, Indiana.

Address Correspondence to: David A. Fisher, MD, 8450 Northwest Blvd, Indianapolis, IN 46278 (tel, 317-802-2852; fax, 317-802-2898; e-mail, dafishermd@aol.com).

Am J Orthop. 2014;43(4):E79-E82.

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This paper will be judged for the Resident Writer's Award.