

Knee Reconstruction in Syndromes With Congenital Absence of the Anterior Cruciate Ligament

Peter G. Gabos, MD, George El Rassi, MD, and Joshua Pahys, MD

Abstract: The authors review their experience with four patients with congenital deficiency of the anterior cruciate ligament (ACL) who underwent surgical treatment of symptomatic knee instability at a mean age of 15.8 years (range 14–17 years). Associated syndromes included fibular hemimelia, congenital short femur, and an unspecified skeletal dysplasia. All patients had undergone multiple previous realignment and leg lengthening procedures and were skeletally mature at the time of the reconstruction. All four patients underwent ACL reconstruction, and one patient underwent concomitant posterolateral corner reconstruction. One patient required an osteochondral autograft transplant procedure in addition to ACL reconstruction. Hypertrophy of the meniscofemoral ligament of Humphrey was a consistent anatomic finding at surgery. The patients were followed for a mean of 38 months (range 26–58 months) after the reconstruction. The mean preoperative Lysholm II score was 38 (range 28–56); the score had improved to a mean of 81 (range 78–93) at the latest follow-up. The authors conclude that reconstructive surgery is a viable option for restoration of knee stability and function in appropriately selected patients with congenital ACL deficiency.

Key Words: congenital anterior cruciate ligament deficiency, anterior cruciate ligament reconstruction

(*J Pediatr Orthop* 2005;25:210–214)

Congenital absence of the anterior cruciate ligament (ACL) has been reported as an isolated anomaly^{2,11,25} or as part of a syndrome complex.^{3,6,9,13,14,16,18,20–23,28,31,32} Studies involving congenital syndromes in which ACL deficiency is a common finding have focused mainly on correction of angular deformity, limb lengthening, or amputation and prosthetic fitting, with almost no mention of treatment of instability from the ACL deficiency.^{3,6,9,13,14,16,31} While several reports describe patients with no complaints of functional instability with daily activities,^{2,13,16,24,29,31} thus advocating nonoperative treatment, episodes of swelling and frequent giving way are also reported.^{11,24,31} Reconstruction of the ACL

in patients with congenital ACL deficiency has received only scant mention in the literature,^{16,18} with no reported follow-up.

Technological advancements in limb lengthening and a better understanding of limb alignment^{27,28} have allowed restoration of a more anatomically aligned limb in patients with congenital limb deficiencies. Not surprisingly, these advances have led to an increase in activity level and higher patient expectations for more normal function. We believe that ACL reconstruction is a viable and beneficial treatment option in the care of a symptomatic patient with congenital absence of the ACL, provided that some important principles are adhered to. The purpose of this study is to review our experience with ACL reconstruction in four patients with symptomatic congenital absence of the ACL.

PATIENTS AND METHODS

Four patients (three girls, one boy) underwent ACL reconstruction for symptomatic congenital deficiency of the ACL at our institution between 1998 and 2002. Associated conditions included an unspecified skeletal dysplasia in one patient (patient 1), isolated fibular hemimelia in one patient (patient 3), and fibular hemimelia with congenital short femur in two patients (patients 2 and 4). All patients had undergone multiple previous realignment osteotomies and limb lengthening procedures resulting in an anatomically aligned limb prior to ACL reconstruction. Anterior knee stability was assessed for each patient by anterior drawer, Lachman, and pivot shift testing. Posterior knee stability was assessed by posterior drawer and presence or absence of a posterior sag. Posterolateral rotatory instability of the knee was assessed by posterolateral drawer, reverse pivot shift, and external rotation recurvatum testing. Varus and valgus instability was tested at 0 and 30 degrees of knee flexion.^{5,7} Instability was graded according to the International Knee Documentation Committee (IKDC) knee form.¹ Subjective function was evaluated using the Lysholm II scoring system^{15,19} preoperatively and at the most recent follow-up evaluation.

Preoperative radiographs included standing anteroposterior (AP), lateral and femoral notch views of the knee, as well as a standing AP radiograph of both lower extremities on a 90-cm trifold film with the patellae oriented directly forward.^{27,28} The morphology of the tibial spines, presence of condylar hypoplasia, and patellar position using the modified Insall-Savati ratio⁸ were recorded. Mechanical axis deviation, lateral distal femoral angle, medial proximal tibial angle, and knee joint line congruence angle were measured.^{27,28} Magnetic resonance imaging (MRI) of the knee

Study conducted at Alfred I. duPont Hospital for Children, Wilmington, Delaware

From Alfred I. duPont Hospital for Children, Wilmington, Delaware.

None of the authors received financial support for this study.

Reprints: Peter G. Gabos, MD, Department of Orthopaedic Surgery, Alfred I. duPont Hospital for Children, 1600 Rockland Road, P.O. Box 269, Wilmington, DE 19803 (e-mail: pgabos@nemours.org).

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was performed for each patient to verify presence or absence of the major knee ligaments and to detect any other intra-articular abnormalities.

RESULTS

The study included four knees (three right knees, one left knee) in four patients, with an average age at the time of ACL reconstruction of 15.8 years (range 14–17 years). All patients had undergone multiple previous operative procedures to correct malalignment and limb length inequality prior to ACL reconstruction, including femoral and/or tibial lengthenings, osteotomies, and contralateral distal femoral and proximal tibial physeodeses. The patients required an average of 3.7 procedures (range 2–6 procedures) prior to the knee reconstruction. All of the previous procedures were performed at the senior author's (P.G.G.) institution. At the time of the knee reconstruction, all four patients were at or near skeletal maturity.

Patient 1 presented with an undefined skeletal dysplasia characterized by bilateral genu valgum, short stature, and bilateral congenital absence of the ACL. Bilateral distal femoral varus osteotomies were performed at age 7 years, followed by a right distal femoral valgus osteotomy to correct varus overcorrection at age 11 years. Patient 2 presented with right congenital short femur and fibular hemimelia. Left distal femoral and proximal tibial physeodesis was performed at age 11 years, followed by right distal femoral varus osteotomy and femoral lengthening at age 12 years. Patient 3 presented with left fibular hemimelia and absence of the fifth ray of the foot. The patient underwent four tibial varus osteotomies for recurrent angular deformity between ages 4 and 8 years, with the last procedure including soft tissue realignment of the patella for lateral patella dislocation. He underwent two tibial lengthening procedures, at age 13 and age 14 years. Patient 4 presented with right congenital short femur and fibular hemimelia. The patient underwent a proximal femoral valgus osteotomy at age 7 years, a femoral and tibial lengthening at age 9 years, and another femoral lengthening at age 15 years.

All four patients reported knee instability with walking despite use of a custom ACL brace and multiple courses of physical therapy directed at muscular strengthening of the lower limb. One patient (patient 3) was a member of his high school basketball team and could not continue to play due to symptomatic knee instability and locking episodes. On examination, all four patients showed marked anterior knee instability, with a grade 3+ (>10 mm) Lachman and anterior drawer and grossly positive pivot shift test. One patient (patient 2) also showed a grade 1+ (5 mm) posterior drawer and presence of posterolateral rotatory instability. The mean preoperative Lysholm II score was 38 (range 28–56).

At the time of the ACL reconstruction, the mean mechanical axis deviation was 12 mm (range 7–15 mm) lateral to the center of the knee joint. The mean lateral distal femoral angle was 89.75 degrees (range 89–92 degrees). The mean medial proximal tibial angle was 92.5 degrees (range 91–94 degrees). The mean joint line congruence angle was 3 degrees of valgus (range 0–4 degrees of valgus).

Radiographic studies of the knee showed hypoplasia of the lateral femoral condyle, a narrow intercondylar notch, and hypoplasia of the tibial spines in all four patients (Fig. 1). One patient (patient 3) had a large osteochondral defect of the weight-bearing surface of the medial femoral condyle, and MRI showed incarceration of the medial meniscus between the femoral defect and the posterior rim of the tibial plateau (Fig. 2).

All four patients underwent arthroscopic evaluation of the knee joint at the time of the reconstruction. Complete absence of the ACL and marked narrowing of the femoral notch was a consistent finding. The meniscofemoral ligament of Humphrey was noted to be markedly hypertrophied in every patient (Fig. 3). One patient (patient 1) showed fraying of the medial and lateral meniscus, but no patient had a complete meniscal tear. Outerbridge²⁶ grade II changes were noted about the medial and lateral compartments in every patient.

All four patients underwent ACL reconstruction using tendon allografts and interference screw fixation. Two patients (patients 1 and 4) underwent isolated arthroscopically assisted, single-incision ACL reconstruction. Two patients (patient 2

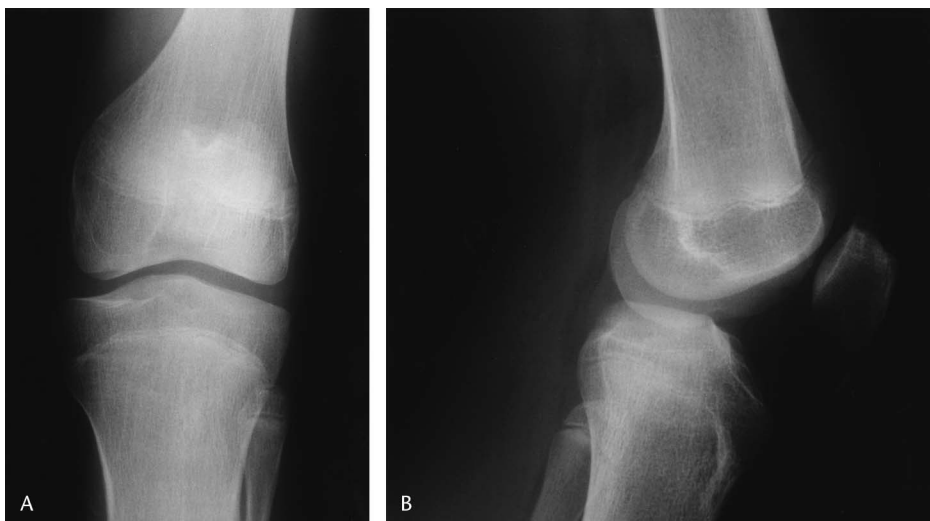


FIGURE 1. Patient 4. Patient with fibular hemimelia and congenital short femur 1 week prior to single-incision, arthroscopically assisted ACL reconstruction. A, AP radiograph of the knee shows hypoplasia of the lateral femoral condyle, a narrow intercondylar notch, a short fibula, and hypoplasia of the tibial spines. B, Lateral radiograph of the knee shows patella baja and hypoplastic tibial spines.

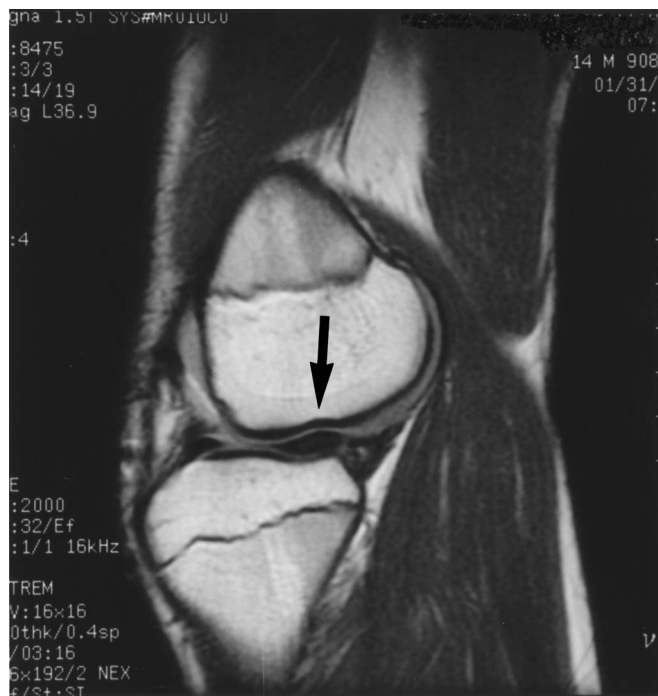


FIGURE 2. Patient 3. Sagittal MRI of the knee shows anterior displacement of the tibia and incarceration of the posterior horn of the medial meniscus within a large osteochondral defect (arrow).

and 3) underwent open ACL reconstruction through a pre-existing midline arthrotomy scar. In patient 2, the ACL was reconstructed using bone–patella tendon–bone allograft, followed by open posterolateral corner reconstruction using a double-limbed bone–Achilles tendon allograft to recreate the popliteus and popliteofibular ligament and an inferior slip of

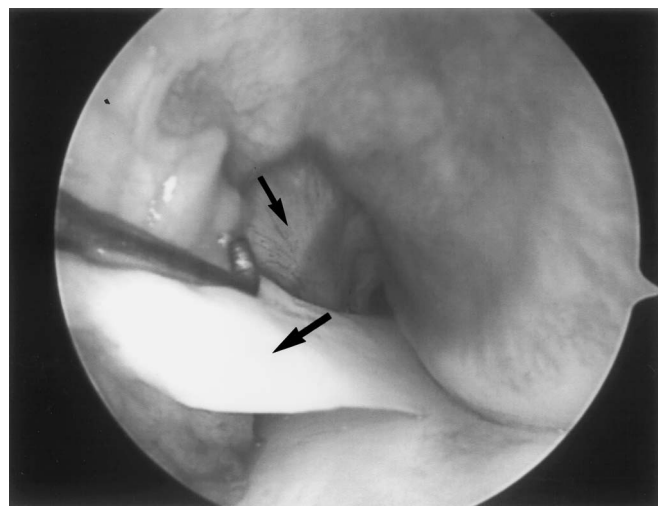


FIGURE 3. Patient 1. Arthroscopic image of the knee showing absence of the ACL within a narrow intercondylar notch. A hypertrophied meniscomfemoral ligament of Humphrey (large arrow) is seen anterior to the posterior cruciate ligament (small arrow).

autogenous bicep tendon to recreate the lateral collateral ligament.³³ In patient 3, an approximately 1.5-cm-diameter, 5-mm-deep osteochondral defect on the weight-bearing surface of the medial femoral condyle was grafted using a 1-cm osteochondral autograft prior to securing the ACL graft because it was noted that the posterior horn of the medial meniscus and the posterior portion of the tibial plateau tended to sublux into the defect when the knee was brought into full extension.

All patients began a structured postoperative rehabilitation protocol in a hinged knee brace immediately after surgery. Toe-touch weight bearing was permitted with the brace locked in full extension for 6 weeks. Crutches were discontinued at 6 weeks postoperatively, with the brace unlocked for controlled weight bearing. The brace was discontinued when a normal gait pattern was achieved, at a mean of 8 weeks postoperatively (range 7–10 weeks). Progressive strengthening was continued for an additional 6 months after surgery.

Patients were followed for an average of 31 months (range 26–58 months) after knee reconstruction. Examination of knee range of motion revealed no deficits of knee flexion. One patient (patient 4) had an extension deficit of 10 degrees compared with the contralateral knee. The Lachman and anterior drawer tests were negative in one patient (patient 1) and grade 1+ (3–5 mm) in the remaining three patients, and this did not deteriorate after surgery. The pivot shift test was negative in all four patients. Instrumented measurements of ligament laxity using a KT-1000 arthrometer⁴ (MEDmetric, San Diego, CA) were available for three patients postoperatively and revealed a mean side-to-side difference of 3 ± 1.1 mm (range 2–4 mm) at the 134-N force, confirming reasonable restoration of stability. The mean postoperative Lysholm II score was 81 (range 78–93). One patient (patient 3) returned to high school basketball at 6 months after surgery and remained asymptomatic at a follow-up of 26 months after knee reconstruction.

DISCUSSION

Congenital ACL absence is uncommon and has been reported as an isolated anomaly^{2,11,25} or as part of a syndrome complex. These syndromes include congenital short femur,^{13,14,16} proximal femoral focal deficiency,³¹ fibular hemimelia,^{3,13,31} congenital knee dislocation,^{6,18} Larsen syndrome,²⁰ and absence of the radius.^{9,30,32} Associated intra-articular anomalies in the knee may include absent or hypoplastic menisci, ring meniscus, and osteochondritis dissecans.^{2,16,24,25,32} Complications of congenital absence of the ACL include meniscal tears,^{2,16,18} clinical knee instability,^{2,11,13,16,31,32} and a tendency for knee subluxation or dislocation during femoral lengthening for congenital short femur or proximal femoral focal deficiency^{16,17,31} or in tibial lengthening for fibula hypoplasia.³

Studies involving these syndromes have had as their main focus correction of malalignment and limb length discrepancy, with scant mention of treatment of instability from the ACL deficiency.^{3,6,9,13,14,16,31} The majority of reports describe patients as having no complaints of knee instability

with daily activities,^{2,13,16,24,29,31} however, episodes of swelling and frequent giving way are also reported.^{11,24,31} Other studies indicate only a “modest loss of function,”¹¹ with patients “adapting to their knee instability,”¹⁶ thus postulating that ACL reconstruction would be unwarranted. These studies are unified by a lack of standardized tests of knee function, description of activity level, and patient follow-up.

We know of only two reports in the orthopaedic literature describing ACL reconstruction in patients with congenital ACL deficiency. Katz et al¹⁸ reported five cases of congenital anterior dislocation of the knee in three children in which the ACL was reconstructed as part of a procedure to maintain reduction of the knee. The authors reported that one knee dislocated postoperatively. No further follow-up information was provided in that study, other than stating that “the other knees are still being protected in a brace.” Kaelin et al¹⁶ described an extra-articular reconstruction of the ACL in one of six patients with congenital limb length deficiency and absent cruciates performed at the time of a meniscectomy, without follow-up.

Improvements in limb lengthening technology and a better understanding of limb alignment^{27,28} have allowed patients with congenital limb deficiencies to achieve a more anatomically aligned limb, as well as equalization of limb lengths and restoration of a plantigrade foot. As the outcomes of these procedures improve, we have noted a concomitant increase in the activity level and expectations of the patient for less restricted function. All of our patients had prolonged attempts at nonoperative treatment, including use of custom ACL braces and physical therapy, but remained symptomatic from their ACL deficiency. Principles adhered to in patients who were considered for ACL reconstruction included restoration of the mechanical axis^{27,28} of the limb, skeletal maturity, reconstruction of all components of knee instability and associated intra-articular pathology, and reinforcement of realistic postoperative expectations and surgical goals.

Interestingly, three of the four patients in our study were found to have a grossly hypertrophied meniscomfemoral ligament of Humphry. There is little information in the literature concerning the exact function of this ligament.^{10,12} The findings of a hypertrophied meniscomfemoral ligament of Humphry in conjunction with congenital absence of the ACL in the present study has not been previously reported, to our knowledge, and may suggest a role of the meniscomfemoral ligament of Humphry in reducing anterior instability.

Ultimately, the long-term outcome of these knees is unknown, and the presence of fairly extensive cartilage changes noted in each patient in this study suggests potentially severe degenerative arthritis over time. Whether this degree of degeneration is related to multiple previous episodes of giving way or is a function of the congenital deficiencies present and/or previous multiple surgical procedures is unclear. Currently, we know of no long-term natural history study of knee function in patients with congenital absence of the ACL who have undergone realignment and limb lengthening with or without ACL reconstruction. It is possible that achieving stability with a transepiphyseal reconstruction of the ACL performed at an earlier age, prior to skeletal maturity, may have led to fewer degenerative changes within the knee. We

felt that anatomic limb alignment was critical for the reconstruction, and this could not be achieved until at or near skeletal maturity in the patients reported here. Also, the potentially deleterious affect of a recurrence of angular deformity requiring osteotomy or of a subsequent limb length discrepancy requiring additional surgical lengthening on a knee reconstructed prior to skeletal maturity would make us hesitant to perform the ACL reconstruction at an earlier age.

The current study shows the efficacy of reconstructive surgery in restoring knee stability and function for appropriately selected patients with congenital deficiency of the ACL.

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