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**Conservative vs surgical management of congenital ACL aplasia--which is more effective in
improving pain and stability?**

By

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Abstract

There is currently no consensus on the treatment of congenital absence of the ACL. The rarity of congenital ACL aplasia precludes large randomized controlled trials which might determine what therapy is superior. This paper examines the limited medical literature available to compare surgical vs conservative therapies in patients with congenital ACL aplasia in order to produce a practical recommendation. Review of the literature shows inconsistent outcomes in terms of stability and pain after ACL construction and conservative management in patients with congenital ACL aplasia. Conservative measures may be sufficient to resolve symptoms of pain and instability but are not always enough. Surgical intervention can also be beneficial but is not without risk. The evidence from the literature suggests that the conservative treatment and patient education should be attempted first, and surgical treatment be considered on a case by case basis depending on the severity of symptoms and associated abnormalities.

Introduction

Congenital aplasia of the anterior cruciate ligament is a rare condition, with a prevalence of 1.7 per 100,000 live births.^{1, 2} The etiology of this condition is not well understood. A study done by Liu et al suggested bilateral ACL and PCL aplasia is associated with a copy number variation deletion impacting the exon sequences of CEP57L1, which is inherited by autosomal dominant transmission.¹ Isolated ACL aplasia has been reported but is often associated with other lower extremity abnormalities. Some abnormalities associated with ACL aplasia include: PCL hypoplasia or aplasia, tibial-fibular dysplasia, dislocation or hypoplasia of patella, femoral dysplasia, congenital talipes equinovarus, osteogenic scoliosis, dislocation of hip(s), discoid lateral meniscus, incomplete sagittal septum, femoral condyle hypoplasia, tibial spine hypoplasia, narrowing and deformation of intercondylar notch, tibial plateau hypoplasia, absent or hypoplastic fibula, congenital leg-length discrepancy, and hypertrophy of the meniscomfemoral ligaments.³⁻⁵ Some persons with ACL aplasia experience instability and pain, while others are found incidentally after an injury. An absent or non-functional ACL is particularly significant in those with femoral or tibial dysplasia because it increases the risk of subluxation or dislocation of the knee after leg-lengthening procedures.⁴ Another concern about congenital ACL aplasia is the predisposition to

osteoarthritis of the knee because of the potential for instability and increased translation of the tibia with daily activities. Conservative and surgical treatments have been utilized, but there is currently no consensus on the best treatment for congenital ACL aplasia or hypoplasia. The rarity of congenital absence of the ACL precludes large randomized controlled trials which might determine which therapy is superior. Nonetheless, examining the limited medical literature available comparing surgical vs conservative therapies in patients with congenital ACL aplasia may produce a practical recommendation. This paper will appraise the evidence on how effective these therapies are in terms of knee stability and pain at a minimum of 6 months follow up.

Discussion

The University of the Pacific library database was utilized to access several search sources including PubMed, Science Direct, Cochrane Library, and Trip. Search terms used included variations of congenital, anterior cruciate ligament, ACL, aplasia, hypoplasia, agenesis, absence, and ACL surgery risks, risks ACL revision surgery. Literature relevant to congenital ACL aplasia or hypoplasia were selected, along with articles discussing pre-op and post-op risks of ACL reconstruction in the setting of acute rupture. Of the articles selected, a full text copy of the article was obtained and critiqued for validity. Congenital absence of the ACL is a rare condition, therefore most studies that are published at this time are case reports or series. Currently there are no randomized controlled trials about congenital absence of the ACL. A table summarizing the articles selected about congenital ACL aplasia can be found in the “Supplemental Materials”.

Conservative Treatment

Conservative treatment for knee pain and instability of an ACL deficient knee consists of cryotherapy, passive range of motion, bracing, electrical neuromuscular stimulation, anti-inflammatory medication, and exercises for strengthening, balance, and proprioception.⁶ Conservative treatment has been successful in some patients with congenital absence of the ACL. A case discussed by Davanzo et al² discussed a 15-year-old female with congenital ACL aplasia with pain and instability who underwent conservative treatment and had subjective improvement in stability and gait at 2 years follow up. Sometimes

ACL aplasia is found incidentally because of an unrelated injury to the knee and does not cause instability. One case involved a 15-year-old male with ACL aplasia, femoral shortening, dysplasia of the tibial intercondylar eminence, valgus knee, and compensatory scoliosis experienced knee pain without instability after an injury. He was treated conservatively and improved without pathological findings on physical exam at 6 months follow up.⁷ A 20-year-old female with a history of leg length discrepancy was found to have ACL aplasia incidentally when she was being treated for a medial meniscal tear. She underwent a partial medial menisectomy without ACL construction because she did not experience any instability. At more than 2 years follow up, she was without pain or instability and exercising without issue.⁸ It is suggested that hypertrophy of the menisofemoral ligaments provide stability to ACL deficient knees.^{4,8} The potential for stable, asymptomatic knees with congenital ACL aplasia suggests that the condition may be more prevalent than originally thought. In view of the favorable outcomes in these studies and the possible stabilization of the knee with hypertrophied ligaments, surgical construction for ACL aplasia may not always be necessary. Furthermore, conservative treatment for ACL aplasia is similar to the same treatment for acute ACL rupture, which can be sufficient. However, it should be noted that 39% of those who underwent non-operative treatment of ACL rupture had either ACL reconstruction for continued knee instability or meniscus repair within 2 years of their ACL rupture, and 51% did within 5 years.^{6,9}

Surgical Treatment

Patients with pain and instability associated with congenital ACL aplasia can consider surgical intervention. Ceruli et al described a 17-year-old male with bilateral ACL agenesis with associated pain and abnormal gait who attempted 6 months of conservative treatment without improvement. He went on to have ACL construction, resulting in improved stability and elimination of knee pain 8 months post-op.¹⁰ A study done by Sonn et al reported 15-year-old monozygotic twins with isolated congenital ACL aplasia with associated knee pain and instability who underwent ACL construction and return to high-level athletics without symptoms at 32 months post-op.¹¹ Chala et al reported a 17-year-old male and a 30-year-old female with congenital ACL aplasia and meniscal tear with associated pain and instability. They both underwent ACL construction and menisectomy/repair and were doing well at 1 year follow up.¹² A retrospective study

by Sachleben et al discussed 13 patients age 3 to 22 with instability who failed conservative treatment for congenital ACL aplasia and underwent ACL construction surgery. None of the patients needed revision surgery due to graft tear at a minimum of 1 year follow up. However, one patient with congenital absence of multiple knee ligaments needed revision surgery, with concurrent first-time PCL reconstruction due to persistent instability.¹³

Successful surgical correction of congenital ACL aplasia has been reported but is not without risks. A study by Davanzo et al included a 6-year-old male with pain and instability associated with congenital ACL aplasia who underwent ACL construction. At 40 months post-op, he was not impaired in daily activities but continued to have bilateral laxity on PE and frequent giving way, with swelling and tenderness. At 5 years post-op, he had limited daily ambulation but continued recreational sports with clear limitation and aches.² One in 9 patients who undergo ACL reconstruction will experience re-rupture or clinical failure at long term follow up.¹⁴ While similar to an ACL reconstruction, it is controversial to what extent a procedure can restore normal kinematics of a joint that is congenitally malformed.¹⁵ Construction of an ACL in someone with congenital ACL aplasia often requires additional steps due to different knee anatomy. Additional procedures done at the time of ACL construction may include notchplasty and/or multiligament reconstruction of associated ligament deficiencies.^{10, 12, 13, 15} Since this disorder affects children, another potential complication for surgery is the proximity and risk of damaging growth plates. The Clocheville technique, which does not cross growth cartilage, was developed to decrease the risk of epiphysiodesis.¹⁶ Narrowing of intercondylar notch, which is commonly associated with ACL aplasia, particularly when combined with absence of the PCL, may limit field of view & space and increase the technical difficulty and likelihood of failure of the procedure.¹⁵ It also should be noted that in patients with acute ACL tears, no difference was found in patient-reported knee scores at 2 and 5 years between conservative versus surgical treatment.^{6, 9}

Osteoarthritis Considerations

Another concern about congenital ACL aplasia is the potential risk for developing premature osteoarthritis of the knee. Arner et al developed a cadaveric model that suggests that an ACL deficient knee

has greater tibial and meniscal translation, which may lead to early degeneration.¹⁷ This finding suggests that constructing an ACL, would decrease tibial and meniscal translation and decrease the risk of developing osteoarthritis. However, it does not take into account those with congenital ACL aplasia who do not have instability. Those individuals without instability are likely to have hypertrophy of the menisofemoral ligament, which can provide stability.^{4, 8} Moreover, surgical reconstruction of a ruptured ACL has not been shown to reduce the risk of future osteoarthritis back to baseline.¹⁸ Meniscal injury, commonly associated with ACL rupture, appears to be the largest risk factor for developing osteoarthritis.¹⁸ This finding suggests that knees without an ACL that have not sustained an injury damaging the meniscus, may not be at an increased risk for osteoarthritis. This idea is supported by a study done by Johansson et al which examined the knees of 6 active patients with congenital ACL aplasia. None of the patients had signs of osteoarthritis, including one followed to age 60.¹⁹

Conclusion

ACL aplasia is a rare congenital condition that may be associated with knee pain and instability and various lower extremity abnormalities. However, it can be stable and asymptomatic. Currently there is no consensus on best treatment for this condition. Literature is limited to case reports and studies with small sample sizes. Further research investigating the long-term effects of congenital ACL aplasia with and without surgical intervention is needed. Treatment decisions can be particularly complicated when there are other lower extremity abnormalities associated with ACL aplasia. The data suggest that conservative measures may be sufficient to resolve symptoms of pain and instability but are not always enough. Surgical intervention can be beneficial but is not without risk. The evidence from the literature suggests that the conservative treatment should be attempted first, and the surgical treatment be on a case by case basis depending on the severity of symptoms and associated abnormalities.

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Supplementary Materials

Summary of patients and outcomes of congenital ACL aplasia treatment

Ref	Age/Gender/S/Sx	Associated Abnormalities	Intervention	Outcome
2	15 y/o F (sibling). Intermittent pain, instability. (+) Anterior Drawer, Lachman. (-) Pivot Shift.	None.	Conservative tx.	Improvement in stability & gait at 2 yr f/u.
2	6 y/o M (sibling). Pain, instability after a sprain. (+) Anterior Drawer, Lachman, Pivot Shift.	Hypoplasia of tibial spine & intercondylar eminence, hypoplasia and cartilage coverage of femoral intercondylar notch, hypoplasia of PCL, atypical meniscus, lateral femoral condyle grade II & III Outerbridge lesions.	Bilateral ACL reconstruction, epiphyseal femoral fixation and transepiphyseal tibia fixation, MacIntosh lateral ligamentoplasty	5 yrs post-op limited daily ambulation but continued sports w/ clear limitation & aches.
3	10 pts, 9F 1M (12 knees) age 2-16 y/o w/ congenital absence of ACL & 1+ associated abnl. ½ asymptomatic, others w/giving way of knee >1x/wk. All but 1 active. All (+) Anterior Drawer, Lachman.	Tibial-fibular dysplasia (6), Dislocation of patella (5), Femoral dysplasia (3), Congenital talipes equinovarus (2), Osteogenic scoliosis (2), Dislocation of hip (bilateral) (1), Discoid lateral meniscus (1), Incomplete sagittal septum (1). Lateral femoral condyle hypoplasia (6), Patellar hypoplasia (5 dislocated, 1 subluxed), lateral tibial spine hypoplasia (4), intercondylar notch (v shape 1, narrow 2), medial tibial plateau hypoplasia (2), fibula (1 hypoplasia, 1 absent).	Radiographic eval, surgical records. <u>Previous procedures</u> : Soft-tissue release & repositioning of patella (4), Syme's amputation (2), Supracondylar femoral osteotomy (1), Adductor tenotomy (1), Posteromedial release for talipes equinovarus (2), Femoral lengthening (1). None to correct laxity from absent ACL.	Congenital absence of ACL more common than generally suspected, associated w/ other developmental abnormalities of bone & soft tissue in lower limb.
4	15 y/o F. Medial knee pain x 4 mo w/o trauma. (+) anterior/posterior drawer, Lachman, Pivot Shift, Reverse Pivot Shift, Dial Test at 30° & 90° flexion, ↑ external rotation.	Hypoplasia of tibial spines, narrowing & deformation of intercondylar notch, femoral trochlea flattened, patella w/ medial hypoplasia, posterior menisco-femoral ligament of Wrisberg, hypoplastic PCL.	Arthroscopic surgical tx of meniscal lesion.	Posterior menisco-femoral ligament of Wrisberg gives insight on knee development. No f/u noted.
5	6 pts (2M 4 F) age 9-22. No subjective instability, but all (+) Anterior Drawer. (+) Posterior Drawer and Jerk Test in 5. (+) Lachman in 3. 2 locked knee following sporting injury. 2 retropatellar pain.	All w/congenital leg-length discrepancy. 5 w/valgus deformity, hypoplasia of lateral femoral condyle, hypoplasia/ aplasia of intercondylar fossa & flattening of intercondylar eminence. Patella small & lateral (3), low (2). ACL completely absent (4), underdeveloped & not functional (2). Bucket handle tears of medial meniscus (2).	All had knee arthroscopy. 2 w/ bucket handle tear had meniscectomy, 1 also underwent extraarticular ligamentoplasty. 3 previously had leg-lengthening, 1 awaiting, 2 not needed.	Post-op ligamentoplasty knee more stable on PE, but no Δ subjective assessment. Other had no later complaints of knee instability or pain.
7	15 y/o M. Mild, improving pain after injury, no instability. (-) Lachmen, Anterior Drawer.	Leg length discrepancy, dysplasia of tibial intercondylar eminence, valgus knee, compensatory scoliosis.	Conservative tx.	6 mo f/u, no pain or instability.
8	20 y/o F. Knee pain after injury. (+) McMurray, Lachman.	Medial meniscal tear, narrow intercondylar notch, hypoplasia of tibial eminences & lateral femoral condyle, hx leg-length discrepancy, hypertrophy of menisiofemoral ligament of Humphrey.	Partial medial meniscectomy. Hx proximal tibial percutaneous epiphysiodesis.	>2 yrs post-op, no pain/ instability, exercising regularly w/o difficulty.
10	17 y/o M. Alteration in gait pattern, pain, instability. (+) Anterior/Posterior Drawer, Pivot Shift, Reverse Pivot	Bilateral agenesis of both cruciate ligaments, hypoplastic and cartilage covered tibial eminence and femoral notch, hypoplastic lateral femoral condyle, narrow intercondylar	Meniscectomy. 6 mo conservative, no improvements knee stability/ pain. Surgery of intercondylar notch & both	6 mo conservative, no improvement. 6 mo rehab after surgery of L knee, no pain. 8 mo

	Shift, Posterior Sag, Dial Test at 30 and 90°.	notch, patella alta, flap lesion of posterior horn of medial meniscus.	cruciates 1 knee at a time, R knee 2 yrs after L.	post-op R knee ↑ stability, no pain.
11	15 y/o M, monozygotic twin. R knee pain & instability. (+) Lachman.	None.	Arthroscopically assisted ACL construction.	32 mo post-op, in sports. No instability/pain. (-) Lachman.
11	15 y/o M, monozygotic twin. R knee pain & instability after injury. (+) Lachman, Pivot Shift.	Bucket handle tear of posterior horn of medial meniscus.	Partial meniscectomy, continued instability. ACL construction.	32 mo post-op, in sports. No instability or pain. (-) Lachman.
12	17 y/o M. Pain, instability, (+) Lachman, Anterior Drawer, Pivot Shift.	Hx leg-length discrepancy. Procurvatum deformity of distal femur, patella baja, stenotic intercondylar notch, dysplastic femoral trochlea, hypoplastic tibial eminences, popliteus tendon atresia, discoid lateral meniscus, tear of medial meniscus.	ACL reconstruction, partial meniscectomy, notchplasty. Hx leg-lengthening procedure.	1 yr f/u in rehab doing well. (-) Anterior Drawer, Lachman, Pivot Shift.
12	30 y/o F. Pain, instability, (+) Lachman, Pivot Shift. (+) Patellar Apprehension.	Absence of intercondylar notch, hypoplastic tibial eminences, dysplastic PCL, bucket handle tear of medial meniscus.	ACL reconstruction, medial meniscus repair, medial patellofemoral ligament reconstruction, notchplasty	1 yr f/u, no pain/instability, in sports. (-) Lachman, Pivot Shift, Patellar Apprehension.
13	13 pts (14 knees) age 3-22. All sx instability & failed nonoperative tx. All (+) Lachman, Pivot Shift.	9/13 w/ associated syndrome. Including congenital short femur, fibular hemimelia, osseofibrous dysplasia, Larsens syndrome.	ACL reconstruction in all, multiligament reconstruction of associated ligamentous deficiency (7). 1 with congenital absence of multiple knee ligaments needed revision ACL reconstruction surgery, w/ concurrent 1 st -time PCL reconstruction (instability)	F/u 1-6.6 yrs. Post-op improved clinical stability in all. None needed revision surgery due to graft tear at min 1 yr f/u.
15	21 y/o F. Instability, (+) Lachman, Anterior Drawer, Pivot Shift.	Leg-length discrepancy, hypoplasia of left lateral tibial spine, decreased posterior tibial slope, lateral femoral intercondylar notch hypoplastic & covered w/ cartilage. Incomplete longitudinal tear at posterior horn of medial meniscus, left in situ, did not appear pathological.	Notchplasty, ACL reconstruction.	6 mo f/u no instability.
16	5 y/o M. Instability, (+) Lachman, Anterior Drawer.	Leg-length discrepancy, agenesis of tibial spine, hypoplasia of medial meniscus.	ACL reconstruction, Clocheville technique.	5 yr f/u, in sports w/o pain/instability. 10° limit in knee flexion.
19	6 pts (3F 3M), 1 age 60, instability, active lifestyles. All with (+) Anterior Drawer. 3 with (+) Posterior Drawer.	All with hx leg-length discrepancy. None w/ signs of OA (1 with erosion of condylar cartilage after posterior subluxation). All with hypoplasia of lateral tubercle. Hypoplasia of medial tubercle (3), absent intercondylar notch (3), short intercondylar notch (3), hypoplastic fibula (2), absent fibula (1), short tibia (2), absent 5 th ray of foot (1).	All with hx leg-lengthening procedure, 1 with posterior subluxation complication. Radiographic and arthroscopic evaluation.	None had arthroscopic or important radiographic signs of osteoarthritis.

*Data excerpted from references cited herein. Author: Alexa Mull, 04/01/19