Treatment of Congenital Cruciate Ligament Absence in a Teenager Knee-Ding Stability

Gaurav Singh, BA[1]; Anna Bergquist, MD[2]; Rebecca Peebles, DO[2]; Justin Bartley, MD[3]

[1]University of Texas at Tyler School of Medicine, Tyler, Texas

[2]The University of Texas Health Science Center at Tyler, Tyler, Texas

[3]Azalea Orthopedics, Tyler, Texas

Corresponding Author Gaurav Singh, BA. University of Texas at Tyler School of Medicine, Tyler, TX 75708 (email: gsingh2@patriots.uttyler.edu).

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ABSTRACT

The congenital absence of the anterior cruciate ligament (ACL) and posterior cruciate ligament (PCL) is an extremely rare condition, with an estimated prevalence of 0.017 per 1,000 live births for ACL agenesis. The authors present the case of a 15-year-old adolescent girl with autosomal dominant multiple synostosis, who developed progressive bilateral knee pain and functional limitations. Bilateral knee x-rays revealed trochlear dysplasia and an absent tibial eminence, suggesting a congenital ligamentous abnormality. Magnetic resonance imaging confirmed the bilateral absence of both the ACL and PCL. Surgical reconstruction of both ligaments was performed, with the first procedure taking place in October 2024. Postoperatively, the patient followed a rehabilitation protocol in physical therapy. This case underscores the rarity of bilateral ACL and PCL agenesis and the importance of timely surgical intervention to alleviate symptoms and improve function.

Keywords: Anterior Cruciate Ligament; Congenital Abnormalities; Joint Instability; Posterior Cruciate Ligament

INTRODUCTION

Anterior cruciate ligament (ACL) and posterior cruciate ligament (PCL) agenesis are exceedingly rare congenital conditions characterized by the complete or partial absence of these essential stabilizing ligaments within the knee joint.[1] Congenital absence of the ACL, referred to as ACL agenesia, has an estimated prevalence of 0.017 per 1,000 live births.[2,3] The incidence of isolated PCL agenesis is poorly documented due to its rarity, but both congenital and traumatic PCL injuries occur at a rate of 1.8 per 100,000 individuals, often alongside ACL agenesis.[1,2,4]

Several genetic conditions have been linked with combined ACL and PCL agenesis, such as autosomal dominant multiple synostosis, Larsen Syndrome, Fibular Hemimelia, and Nail-Patella Syndrome.[5,6] Previous research has also indicated a genetically inherited autosomal dominant pattern that may account for agenesis of both the ACL and PCL, potentially linked to a copy number variation deletion in the CEP57L1 gene, which is expressed in ligament tissue.[7,8]

These ligaments, alongside the medial (MCL) and lateral (LCL) collateral ligaments, are integral to maintaining knee stability, proprioception, and proper biomechanics during dynamic activities.[8-10] Consequently, the congenital absence of the ACL can lead to substantial knee instability, joint malalignment, and an increased susceptibility to degenerative changes over time.[2,9,11] While some individuals can achieve high function despite PCL deficiency, biomechanical studies demonstrate that the PCL plays a critical role in knee stability, particularly in limiting posterior tibial translation and rotational laxity.[1,10] In cases of complete absence of both ligaments, the knee's ability to withstand rotational and anterior-posterior forces is substantially compromised, resulting in pronounced joint laxity, altered gait mechanics, and an increased risk of early osteoarthritic changes.[1,6,9,10] Although agenesis of these ligaments is typically an isolated defect, they may present with other congenital abnormalities affecting the knee, lower extremities, hips, and spine.[7] Diagnosis of ACL agenesis is typically confirmed via magnetic resonance imaging (MRI), which may also identify associated anomalies such as hypoplasia of the lateral femoral condyle, irregularities of the tibial intercondylar spines, absent or abnormal menisci, and patellar dislocation.[6,10-12]

CASE REPORT

A 15-year-old right-handed adolescent girl with a medical history of autosomal dominant multiple synostosis, anemia, anxiety, and migraines presented with bilateral knee pain. She reported a several-year history of discomfort that worsened with

increased physical activity, describing the pain as a dull, continuous ache rated 7 out of 10 in severity. Accompanying symptoms included a sense of knee instability on uneven terrain. Pain was aggravated by physical activity, knee flexion and extension, cold temperatures, and prolonged sitting or standing. The patient experienced significant functional impairments, including difficulties with squatting, kneeling, pivoting, running, jumping, and cutting, but denied any episodes of patellar dislocation, instability, or prior knee trauma.

The patient exhibited tenderness along the medial and lateral joint lines of both knees, a posterior sag sign, and Lachman and posterior drawer tests showing grade III instability. Other tests, including the patellofemoral grind, patellar apprehension, McMurray's, and varus and valgus stress tests, were negative. She walked with a heel-to-toe gait and showed neutral knee alignment, with no knee effusion. Leg strength was 5/5, sensory examination was normal, skin was intact, and both legs had good perfusion and capillary refill.

Bilateral knee x-rays were performed, which revealed the presence of trochlear dysplasia, absence of a femoral notch, and absent tibial eminences, suggesting complete agenesis of the ACL and PCL (Figures 1A-1C). An MRI of both knees confirmed the diagnosis, demonstrating the complete absence of bilateral ACL and PCL, in addition to macerated medial meniscus (Figure 2A).

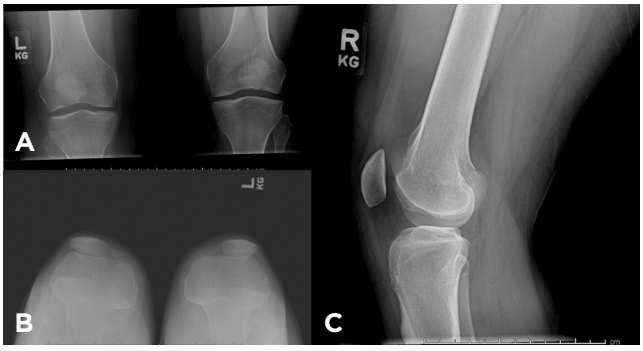

Figure 1. *A) Anteroposterior x-ray of bilateral knees; B) merchant x-ray of right knee; C) lateral x-ray of bilateral knees.*

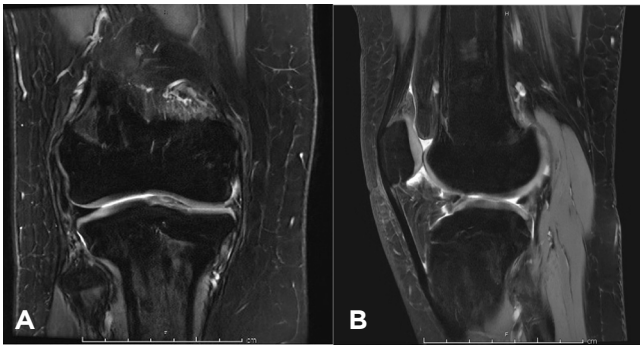

Figure 2. *A) T2-weighted Coronal MRI of right knee, demonstrating absence of ACL and PCL; B) T2-weighted Sagittal MRI of right knee, demonstrating absence of ACL and PCL.*

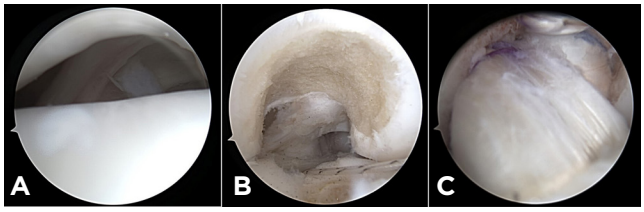

Figure 3. *A) Arthroscopic image demonstrating no intercondylar notch, no ACL, no PCL; B) arthroscopic image following notchplasty, meniscofemoral ligament visualized within the posterior notch; C) arthroscopic image demonstrating reconstructed ACL and PCL.*

The patient was diagnosed with congenital absence of the bilateral ACL and PCL. Due to the failure of conservative measures—including 12 physical therapy visits on two separate occasions over multiple years—and the severity of clinical symptoms and significant functional limitations, surgical reconstruction of the ACL and PCL was deemed necessary. The reconstruction procedure began with the right knee, as the first surgery was performed in October 2024 due to more pronounced symptoms and pain in that knee. The surgery consisted of arthroscopic exam notable for a macerated medial meniscus, preserved cartilage, complete absence of an intercondylar notch with hypoplastic tibial eminences, and complete absence of an ACL and PCL (Figure 2B). A notchplasty was performed to recreate an intercondylar notch, using the intact meniscofemoral ligament as a guide to determine the desired notch width (Figures 3A & 3B). ACL reconstruction was performed using an anterior tibialis allograft, and an all-inside PCL reconstruction was performed using a quadrupled hamstring allograft. The ACL and PCL were fixated on the femur and tibia with suspensory fixation (Figure 3C).

Postoperatively, the patient was advised to use crutches with 50.0% weight bearing and to gradually increase activities while wearing a knee brace. She began physical therapy afterward, with follow-up appointments at two weeks, six weeks, and three months. At her six-week appointment, the patient was walking with minimal assistance, reported major improvements in stability and knee pain, exhibited a range of motion from 0° to 80°, and showed stable results on the Lachman and posterior drawer tests.

DISCUSSION

Although isolated congenital absence of the ACL is more common, PCL absence typically occurs alongside ACL agenesis, underscoring the rarity of this condition.[2] Most cases of congenital absence of the cruciate ligaments also involve other knee abnormalities, such as hypoplasia of the lateral femoral condyle, tibial intercondylar spines, trochlear dysplasia, and either abnormal or absent menisci.[13-15] Additionally, these defects increase mechanical stress on the knee ligaments and are commonly found in skeletally-

immature patients, which raises the likelihood of ligamentous abnormalities.[1,16-18]

Treatment for this condition ranges from conservative treatment to surgical intervention.[14,19,20] Based on the literature review, there are only a few dozen reports of ACL agenesis worldwide, limiting research regarding the efficacy of surgical treatment of congenital absence of the ACL and PCL.[15,16,21] The few case reports found in the literature address the treatment of ACL agenesis, but not PCL.[1] The congenital absence of the cruciate ligaments is usually asymptomatic and is often managed conservatively, as the joint surface may adapt to compensate for the missing ligaments, allowing for an even distribution of forces.[22,23] However, after failure of conservative measures, including extensive physical therapy, surgical intervention is recommended to reconstruct ligaments and restore knee stability.[2,6] Despite treatment, most of these patients will likely progress to knee replacement later in life, secondary to resulting knee arthritis.[6,24] Manner et al[16] created a three-tier classification system for cruciate ligament dysplasia, which aids in diagnosing patients with cruciate agenesis.[2,3,13,15] In this case, the patient presents with a Type III classification. However, the classification system does not offer specific treatment guidelines for this level of dysplasia, highlighting the need for further research.[15]

CONCLUSION

This case of congenital ACL and PCL absence in an adolescent girl highlights the need for early diagnosis and intervention of rare ligament abnormalities. Although this condition affects knee stability, timely surgical reconstruction may provide better outcomes. Clinicians should consider ligament agenesis in cases of unexplained knee instability, particularly with congenital anomalies. Further research is needed to examine genetic factors and improve treatment and long-term outcomes.

REFERENCES

1. Waghe VR, Tikhile P, Patil DS. The integral role of physiotherapy in combined complete anterior cruciate ligament and posterior cruciate ligament arthroscopic reconstruction: a case report. Cureus. 2024;16(4):e57556. doi: 10.7759/cureus.57556.
2. Gavvala SN, Giliyaru S, Ariyaratne S, et al. Congenital absence of anterior and posterior cruciate ligaments – A rare case report and review of literature. J Arthrosc Surg Sports Med. 2024;5(1):38-40. doi:10.25259/JASSM_3_2024.
3. García N, Debandi A, Delgado G, et al. Isolated posterior cruciate ligament aplasia: a case report. Skeletal Radiol. 2019;48(9):1439-1442. doi: 10.1007/s00256-019-3161-3.
4. LaPrade CM, Civitarese DM, Rasmussen MT, et al. Emerging updates on the posterior cruciate ligament: a review of the current literature. Am J Sports Med. 2015;43(12):3077-3092. doi: 10.1177/0363546515572770.
5. Berruto M, Gala L, Usellini E, et al. Congenital absence of the cruciate ligaments. Knee Surg Sports Traumatol Arthrosc. 2012;20(8):1622-1625. doi: 10.1007/s00167-011-1816-2.
6. Sweeney E, Hoover-Fong JE, McIntosh I. Nail-patella syndrome. In: Adam MP, Feldman J, Mirzaa GM, Pagon RA, Wallace SE, Amemiya A, eds. GeneReviews®. University of Washington, Seattle; 1993. Accessed March 24, 2025. http://www.ncbi.nlm.nih.gov/books/NBK1132/.
7. Benassi AB, Guerreiro JPF, de Oliveira Queiroz A, et al. Anterior and posterior cruciate ligament agenesis. J Surg Case Rep. 2018;2018(8):rjy216. doi: 10.1093/jscr/rjy216.
8. Liu Y, Li Y, March ME, et al. Copy number variation in CEP57L1 predisposes to congenital absence of bilateral ACL and PCL ligaments. Hum Genomics. 2015;9:31. doi: 10.1186/s40246-015-0053-z. Erratum in: Hum Genomics. 2016;10:5. doi: 10.1186/s40246-016-0060-8.
9. Raj MA, Mabrouk A, Varacallo M. Posterior cruciate ligament knee injuries. In: StatPearls. StatPearls Publishing; 2024. Accessed December 30, 2024. http://www.ncbi.nlm.nih.gov/books/NBK430726/.
10. Goyal K, Tashman S, Wang JH, et al. In vivo analysis of the isolated posterior cruciate ligament-deficient knee during functional activities. Am J Sports Med. 2012;40(4):777-785. doi: 10.1177/0363546511435783.
11. Kim KK, Kim TH, Kim DY, et al. Bilateral congenital absence of the anterior cruciate ligament associated with bilateral knee and hip osteoarthritis: case report. Int J Surg Case Rep. 2020;72:313-317. doi: 10.1016/j.ijscr.2020.05.099.
12. Arteaga G, Ramos P, Vargas M, et al. Anterior cruciate ligament agenesia associated with hypermobile lateral meniscus, bilateral prominent L5 transverse process, and ipsilateral femoral shortening: a case report. J Orthop Rep. 2023;2(4):100216. doi:10.1016/j.jorep.2023.100216.
13. Ossola C, Duvia M, Carlizzi CN, et al. MR-arthrography: anatomic variant from link between lateral meniscus and anterior and posterior cruciate ligaments. a case report and review of the literature. Radiol Case Rep. 2021;16(10):3012-3015. doi: 10.1016/j.radcr.2021.07.012.
14. Walker JL, Milbrandt TA, Iwinski HJ, et al. Classification of cruciate ligament dysplasia and the severity of congenital fibular deficiency. J Pediatr Orthop. 2019;39(3):136-140. doi: 10.1097/BPO.0000000000000910.
15. Gabos PG, El Rassi G, Pahys J. Knee reconstruction in syndromes with congenital absence of the anterior cruciate ligament. J Pediatr Orthop. 2005;25(2):210-214. doi: 10.1097/01.bpo.0000153874.74819.29.

16. Manner HM, Radler C, Ganger R, et al. Dysplasia of the cruciate ligaments: radiographic assessment and classification. J Bone Joint Surg Am. 2006;88(1):130-137. doi: 10.2106/JBJS.E.00146.
17. Chahla J, Pascual-Garrido C, Rodeo SA. Ligament reconstruction in congenital absence of the anterior cruciate ligament: a report of two cases. HSS J. 2015;11(2):177-181. doi: 10.1007/s11420-015-9448-6.
18. Murali J, Monchik K, Fadale P. Congenital absence of the anterior cruciate ligament. Am J Orthop (Belle Mead NJ). 2015;44(8):E283-E285.
19. Kaplan D, Jazrawi T, Capo J, et al. Multiple ligament reconstruction for absent cruciate and a congenital short femur case report and review of the literature. Bull Hosp Jt Dis (2013). 2017;75(2):143-147.
20. Reeves B, Roper B, Salton R, et al. Outcomes of simultaneous lengthening and ACL reconstruction in fibular hemimelia: a retrospective case series. Iowa Orthop J. 2024;44(1):93-98.
21. Frikha R, Dahmene J, Ben Hamida R, et al. Absence congénitale du ligament croisé antérieur. A propos de 8 cas [Congenital absence of the anterior cruciate ligament: eight cases in the same family]. Rev Chir Orthop Reparatrice Appar Mot. 2005;91(7):642-648. French. doi: 10.1016/s0035-1040(05)84468-x.
22. Davanzo D, Fornaciari P, Barbier G, et al. Review and long-term outcomes of cruciate ligament reconstruction versus conservative treatment in siblings with congenital anterior cruciate ligament aplasia. Case Rep Orthop. 2017;2017:1636578. doi: 10.1155/2017/1636578.
23. Kambhampati SBS, Vishwanathan K, Vasudeva N, et al. Management of anterior cruciate ligament aplasia for the arthroscopy surgeons: a systematic review. Indian J Orthop. 2022;56(7):1123-1138. doi: 10.1007/s43465-022-00651-2.
24. Balke M, Mueller-Huebenthal J, Shafizadeh S, et al. Unilateral aplasia of both cruciate ligaments. J Orthop Surg Res. 2010;5:11. doi: 10.1186/1749-799X-5-11.