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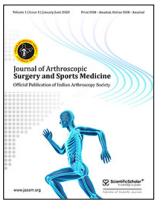

Case Report

Congenital absence of anterior and posterior cruciate ligaments – A rare case report and review of literature

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ABSTRACT

A rare occurrence, congenital absence of the cruciate ligaments has a prevalence of 0.017/1000 live births. A literature review on the PubMed database revealed only a few case reports of both coexisting anterior cruciate ligament and posterior cruciate ligament congenital absence. Most cases were found to be associated with other congenital abnormalities, making isolated congenital absence of the cruciate ligaments exceptionally rare. Liu *et al*. reported a total of nine cases to date, with our case being the second youngest and having no other associated knee pathologies. Our report presents a 4-year-old girl with unilateral congenital absence of a cruciate ligament, diagnosed through magnetic resonance imaging. It is crucial to be aware of this rare abnormality for proper management and to minimize morbidity.

Keywords: Prevalence, Live birth, Anterior cruciate ligament, Posterior cruciate ligament, Magnetic resonance imaging

INTRODUCTION

Cruciate ligaments play a crucial role in maintaining the stability of the knee joint. The congenital absence of these ligaments is a rare condition, with a global prevalence of 0.017/1000 live births.[1-6] This anomaly is often associated with various deformities, including hypoplasia of the lateral femoral condyle, congenital shortening of the femur, defects in the fibular head, abnormalities in the tibial intercondylar spines, abnormal or absent meniscus, and patellar dislocation.[4] The optimal management of this condition is still a topic of debate, with conservative and surgical treatments being considered.

Our case involves the second youngest patient with unilateral congenital absence of cruciate ligaments, who presented with walking difficulties and instability in the right knee. Due to the young age of the patient, physiotherapy was chosen as the preferred treatment modality. We present a unique case of a 4-year-old girl with a congenital absence of cruciate ligaments, leading to knee instability. Magnetic resonance imaging (MRI) imaging revealed the absence of both the anterior cruciate ligament (ACL) and posterior cruciate ligament (PCL). Imaging techniques, particularly MRI, play a crucial role in diagnosing this condition. It is essential for radiologists to be familiar with this anomaly to prevent long-term complications such as dislocations, hypertrophy of accessory knee ligaments, leg length inequalities, and early-onset osteoarthritis.

CASE REPORT

A 4-year-old girl presented to our department with a history of recurrent instability of the right knee. There was no history of trauma and past history was unremarkable. She had a normal birth and there were no associated genetic abnormalities. On clinical examination, there was no swelling of the right knee. Anterior and posterior drawer tests as well as the Lachman test were positive. McMurray's test and varus-valgus stress tests were negative. She had a full range of movements. Ipsilateral hip, ankle, and contralateral knee were normal.

Given the age and presentation, with the absence of trauma, a radiograph was not performed. MRI was performed to evaluate this further. Only proton density sagittal MR of the knee could be obtained on 1.5 Tesla MRI scanner. MRI revealed a complete absence of both ACL and PCL [Figure 1]. The tibial eminence was hypoplastic with absent tibial spines. The medial and lateral menisci, medial and lateral collateral ligaments, iliotibial band, posterolateral corner, and extensor

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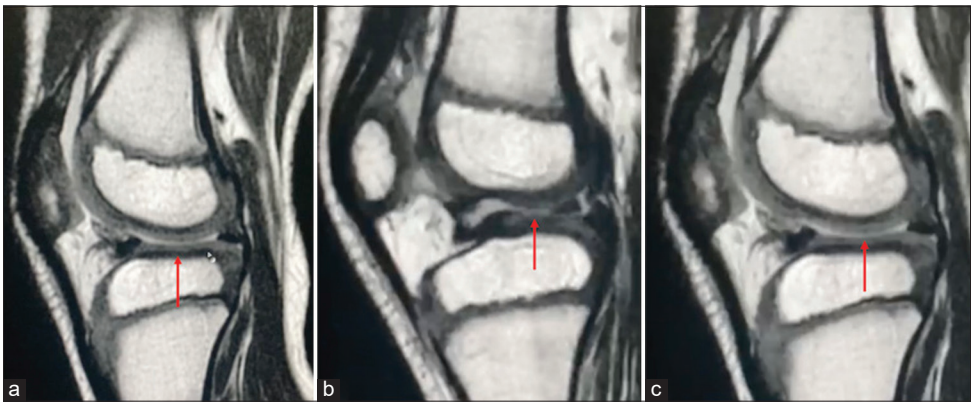

Figure 1: (a-c) Sagittal proton density images showing the absence of both anterior cruciate ligament and posterior cruciate ligament (arrows) with (b) hypoplasia of tibia eminence and absence of tibial spines (arrow).

mechanisms were normal. The articular cartilages were also normal. She was managed with physiotherapy.

DISCUSSION

The coexistence of congenital absence of both the ACL and PCL is an uncommon occurrence. There have been only a few documented case reports on this condition. On the other hand, isolated congenital absence of the ACL is more frequently observed compared to the absence of the PCL. These conditions can be linked to knee dislocation.

The ACL originates from the anteromedial aspect of the intercondylar area of the tibial plateau and extends superiorly and posteriorly to insert on the posteromedial aspect of the lateral femoral condyle. Conversely, the PCL arises from the lateral surface of the medial femoral condyle and inserts onto the posterolateral part of the intercondylar surface of the tibial plateau. The absence of cruciate ligaments can be explained through embryological studies. The menisci, capsule, and ligaments begin to form between four and six weeks of gestation. By the eighth week, the fetal knee joint resembles that of an adult knee joint, with fully differentiated anterior and PCLs. The tibial spine, which emerges during the past two months of intrauterine life from the secondary superior epiphyseal nucleus of the tibia, becomes elevated around the age of 2 years. The intercondylar notch serves the purpose of housing the cruciate ligaments. Therefore, any congenital alteration of the cruciate ligaments is associated with morphological changes in the tibial spine.[7,8]

Manner *et al*. identified three distinct types of dysplasia affecting the cruciate ligaments.[2] Type I is characterized by aplastic or hypoplastic ACL alongside a normal PCL. Type II presents with an aplastic ACL and hypoplastic PCL. Type III is marked by aplastic ACL and PCL. Liu *et al.* described a genetically inherited autosomal dominant form of cruciate agenesis involving both ACL and PCL.[3]

The congenital absence of ACL is more prevalent than that of PCL, often presenting with additional abnormalities such as knee dislocation, lateral femoral condyle hypoplasia, femur shortening, fibular hemimelia, meniscus abnormalities, and patellar dislocation.[9]

While many individuals with congenital cruciate ligament absence may be asymptomatic due to compensatory mechanisms provided by surrounding muscles and ligaments, conservative treatment is typically recommended for such cases. This approach is favored due to the even distribution of forces achieved through compensatory joint surface adjustments in the absence of ligaments. However, some authors advocate for ligament reconstruction instead. Surgical intervention is reserved for cases where conservative therapy fails, with reconstructive surgery involving both ACL and PCL.[9,10] In the case of our 4-year-old patient, physiotherapy was the chosen management approach.

CONCLUSION

The present case study provides an overview of the MRI findings associated with aplasia of the ACL and PCL of Manner Type 3, without any additional anomalies. The presence of a hypoplastic tibial eminence and the absence of tibial spines strongly suggest a congenital origin. Congenital cruciate absence is an uncommon condition that is frequently misdiagnosed. Conservative treatment has traditionally been the preferred approach for the majority of patients, reserving ligament reconstruction for those experiencing symptomatic instability. Given the rarity of congenital cruciate absence, it is crucial to raise awareness of this condition to minimize patient morbidity.

Ethical approval

The Institutional Review Board approval is not required.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

Use of artificial intelligence (AI)-assisted technology for manuscript preparation

The authors confirm that there was no use of artificial intelligence (AI)-assisted technology for assisting in the writing or editing of the manuscript and no images were manipulated using AI.

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