CASE REPORT

Teenage Female with Knee Pain and Instability

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INTRODUCTION

Congenital dislocation of the knee, resulting from an absence of the cruciate ligaments, is a condition affecting 0.017 per 1000 live births (1). Although very rare, it has drawn the attention of orthopaedic surgeons and radiologists because it is associated with other congenital anomalies. This paper presents abnormalities that are isolated to the knee and without evidence of associated syndrome. The absent anterior cruciate ligament (ACL) is associated with a hypoplastic posterior cruciate ligament (PCL), a shallow femoral notch, and hypoplastic tibial spines seen with radiographic and magnetic resonance imaging. The objective of this article is to review the clinical presentation and imaging findings associated with congenitally absent ACL.

THE CASE

A 16-year-old female presented to a pediatric orthopaedic surgeon with a history of right knee symptoms. Beginning at two years of age, her mother noticed atrophy of her right leg and abnormality in gait when she first began to walk, leading to recurrent falls. Her left leg was asymptomatic. At that time, a pediatric neurologist assessed her, and spine MRI and EMG studies were performed, showing no abnormalities. At age five, she began to function normally and she enrolled in dance and gymnastics. These activities appeared to help her with balance and strength.

At the time of evaluation, the patient was very active and played soccer. However, she described a five month history of poorly localized intermittent pain and tightness in the right knee after

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standing or walking long hours. Her pain was rated 4/10. She used an over-the-counter knee brace that provided some relief. She denied swelling, clicking, catching, or other symptoms of instability. There was no history of significant knee trauma or family history of similar problems.

Clinical examination revealed valgus alignment bilaterally, which was worse on the right side. The right leg was noted to be slightly smaller in girth than the left and there was no obvious leg length discrepancy. Gait demonstrated a valgus thrust and apparent instability in the sagittal plane with each step. She had full range of motion with no crepitations and no joint line or patellar tenderness. Positive Lachman, pivot shift, and anterior drawer tests were demonstrated with a negative McMurray's test. No other physical findings of interest were noted. The differential diagnosis for this patient was very limited and included traumatic or congenital absence of the right ACL.

AP radiograph demonstrated a shallow femoral intercondylar notch and hypoplastic tibial spines (Figure 1). Lateral view of patient in the right
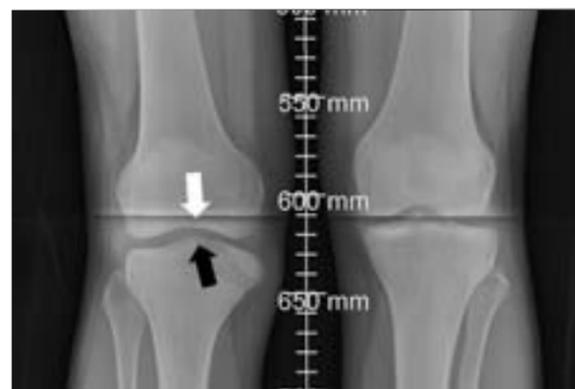

Figure 1: Standing AP radiograph from a full-length leg series shows valgus knees bilaterally, worse on the right. A shallow femoral intercondylar notch (white arrow) and hypoplastic tibial spines (black arrow) are visible in the right knee.

Figure 2: Lateral right knee radiograph with the patient positioned in right lateral decubitus reveals normal alignment without subluxation of the tibia on the femur. Tibial spines are hypoplastic (black arrow). Physes are closed.

Figure 3: Coronal proton density fat-suppressed MRI demonstrates a shallow femoral intercondylar notch (white arrow), absent intercondylar eminence (black arrow) and a downward slope of the medial tibial condyle. There is no evidence of the ACL

lateral decubitus position showed a hypoplastic intercondylar eminence in the right knee (See Figure 2). MRI demonstrated the shallow femoral intercondylar eminence and hypoplastic tibial spines. No ACL fibers were visible. There was complete hyaline cartilage covering the area where the tibial eminence appeared to be aplastic and complete cartilaginous coverage of the shallow femoral notch anteriorly. Sagittal view showed anterior subluxation of the tibia, the imaging equivalent of an anterior drawer test. This is seen on the MRI with the patient in a supine position, because this position allows the weight of the upper leg to posteriorly subluxate the tibia. Associated hyperbuckling of the hypoplastic posterior cruciate ligament is also identified (See Figures 3 and 4).

Although examination showed significant instability of the knee and imaging studies were abnormal, the patient said she was able to perform and function at a reasonable level. She was advised to follow up with her orthopaedic surgeon annually for further imaging studies and to evaluate any progression of symptoms, at which time large ligament reconstruction may be considered.

DISCUSSION

The first suspected case of congenital absence of the ACL was reported in 1956 (2) and was later confirmed in 1967 by surgical exploration in

patients with congenital dislocation of the knee (3). Since then, it has been described in several cases in association with dysplasia of other structures in the knee including the menisci (4,5), tibial spines (2,6), intercondylar notch (7), and the PCL (8). In addition, congenital absence of the ACL may coexist with other congenital anomalies as part of a syndrome complex (9-11). It is less commonly seen as an isolated abnormality (12).

Thomas et al. identified tibial and fibular dysplasia as well as dislocation of the patella as the most common radiographic findings associated with congenital absence of the ACL (8). Others have frequently found absence of the cruciate ligaments in those with congenital femoral deficiency and post-axial hypoplasia (7,13,14). Associated conditions beyond the lower extremities include thrombocytopenia-absent radius syndrome (11) and arthrogryposis (10).

Abnormalities of structures exisat within the knee joint itself. Manner et al. evaluated a series of 34 knees in 31 patients with congenital cruciate ligament abnormalities on magnetic resonance imaging (7). The author defined three patterns of cruciate ligament dysplasia: hypoplasia or absence of the ACL with normal PCL (type 1 in 56%), aplasia of the ACL with hypoplastic PCL (type 2 in 21%), and total absence of both ACL and PCL (Type 3 in 24%) (7). Aberrations of the menisci have also been de-

Figure 4: Sagittal proton density fat-suppressed MRI reveals anterior subluxation of the tibia on the femur, the radiological equivalent of the anterior drawer test. The posterior cruciate ligament is hypoplastic and buckled (black arrows). A single cartilaginous surface covers the majority of the tibia (white arrows).

scribed in some cases as being ring-shaped (4,12) or discoid (5,7). Moreover, osteochondritis dessicans has been identified in conjunction with absent ACL (7,15).

Embryological studies of the knee joint indicate that at approximately seven weeks post ovulation, the femoral and tibial condyles are well defined and the interzone contains a three-layered blastema (16). The menisci, capsule, and cruciate ligaments all arise from this blastema, perhaps explaining why abnormalities in these structures commonly co-exist (3).

Often times, it can be difficult for radiologists to distinguish between traumatic and congenital causes for an absent ACL, however; there may be several clues that may suggest one or the other. For children younger than fourteen years of age, injuries to the ACL are less commonly seen since the physeal plates are not yet fused and traction forces are more likely to cause epiphyseal separation, long bone fractures, or avulsions of the tibial eminence rather than a disruption of the ligament (17). History of trauma to the knee suggests a traumatic cause while the radiographic finding of a hypoplastic intercondylar eminence may support a congenital cause for ACL deficiency (18).

Manner et al. performed the largest known study examining radiographic changes in knees with arthroscopically proven aplasia of the cruciate ligaments (7). Based on tunnel view radiography, the intercondylar notch was found to be more narrow and shallow in deficient ACL knees when compared to unaffected knees (7). It is thought that the purpose of the femoral intercondylar notch is to house the cruciate ligaments and the tension created by their insertion can cause secondary development of the tibial spines (2).

In those with absent ACL and normal or hypoplastic PCL, the lateral tibial spine was found to be aplastic and the medial spine was minimally affected (7). Since the medial tibial spine is the location for ACL insertion, Manner et al. hypothesized that the lateral tibial spine is affected rather than the medial spine. He speculated this because the lateral aspect of the femoral notch was also found to be hypoplastic in those with absent ACL and this resulted in molding and underdevelopment of the lateral tibial spine (7). In those with complete absence of both the ACL and PCL, he found both tibial spines were aplastic and the tibial eminence was flattened (7). With these type 3 deficiencies, the distal femoral epiphysis appeared concave and the proximal tibial epiphysis was convex, giving a "ball-and socket"- type knee joint (7). In the same study, magnetic resonance imaging showed hyaline cartilage covering the area of the femoral notch when both cruciate ligaments were absent (7). Authors continue to debate whether the changes in the femoral intercondylar notch and the tibial spines are congenital or simply a secondary response to the aplastic cruciate ligaments.

Reconstruction of the ACL has been shown to be a viable option for symptomatic patients with absence of the ACL (19). Nevertheless, there are indeed patients who remain asymptomatic and continue to be observed. Long-term outcome of those with knee instability caused by congenital absence of the cruciate ligaments is very good and many do not develop longer-term degenerative changes (6).

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