

Congenital Absence of the Cruciate Ligaments

Derek B. Smith,¹ BScEng, MScEng MD '00, J. David Amirault,² MD, FRCS(C) and Lorne Leahey,³ MD, FRCS(C)

¹Department of Surgery, Dalhousie University

²Associate professor, Department of Surgery (Division of Orthopaedic Surgery), Dalhousie University and Chief of Surgery, New Halifax Infirmary, QEII Health Sciences Centre

³Associate professor, Department of Surgery (Division of Orthopaedic Surgery), Dalhousie University, Orthopaedic Surgery, IWK-Grace Health Centre.

Congenital absence of one or both cruciate ligaments is a rare event, and little literature exists regarding this abnormality. Patients with this condition often have associated anomalies. This study looked at 6 subjects with congenital abnormalities often associated with absence of the cruciate ligaments. The subjects were followed by a questionnaire and knee examination. This paper presents results of congenital absence of the cruciate ligaments on knee instability and activities of daily living.

INTRODUCTION

Congenital absence of one or both of the cruciate ligaments is an uncommon occurrence, and has been only sporadically reported in the literature. Most reports are in association with a congenitally short femur or other abnormality, such as congenital dislocation of the knee. A study by Giorgi (1956) looking at morphologic variations in the intercondylar eminence of the knee contained the first mention of congenital absence of the cruciates: one case showed total aplasia of the intercondylar eminence, which Giorgi attributed to a congenital lack of the cruciates. A study by Katz et al (1967) reported a relationship between congenital cruciate ligament anomalies and congenital dislocation of the knee. This study was the first to report confirmed congenital absence of the cruciate ligaments. Since then, it has been reported in association with a number of congenital abnormalities, [3-8]. Congenital absence of one or both cruciates is usually detected during examination for a separate abnormality, such as congenital dislocation of knee, [2], or preparation for leg-lengthening procedures, [3]. During clinical analysis for the presenting abnormality, ipsilateral instability of the knee is noted, with follow-up tests revealing cruciate ligament insufficiency. This study is intended to add to the existing base of reported cases of congenital absence of one or both cruciate ligaments.

METHODS

Using resources at the IWK Grace Health Centre Orthopaedics Clinic, a list of patients with possible congenital absence of the cruciate ligaments was compiled. These subjects all had associated abnormalities that are often linked to congenital absence of the cruciate ligaments, such as fibular hemimelia and proximal femoral focal deficiencies. A total of 9 patients were identified as having congenital abnormalities associated with congenital absence of the cruciate ligaments. Two of these patients lived a significant distance away and could not be obtained for examination purposes, and 1 patient could not be reached; this left 6 subjects with possible congenital absence of the cruciate ligaments. The subjects were relatively young, ranging in age from 7 to 24 years old. Each subject was asked a series of questions and had his or her knee examined. This was in an effort to clinically determine the presence or absence of the cruciate ligaments and, if absent, any effects this may have had on the subject. The questions included *The Knee Society* clinical rating system [10] as well as some questions regarding any impact the patient's knee might have had on the subject's life and/or activity levels, associated abnormalities and previous treatments. Each subject signed an informed consent form prior to any investigations.

Address correspondence to:

Derek Smith, Med IV

Box 80, Sir Charles Tupper Building, Dalhousie University

Halifax, Nova Scotia, Canada. dsmith@is2.dal.ca. (902)492-7153

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Table I - Associated Abnormalities As Identified in the Literature

- Congenital knee dislocation
- Congenital leg length discrepancy
- Hypoplastic or absent fibula
- Absent 5th ray of foot
- Dislocation of patella
- Femoral dysplasia
- Congenital talipes equinovarus
- Osteogenic scoliosis
- Bilateral dislocation of hip
- Incomplete sagittal septum
- Proximal femoral focal deficiencies
- Congenital thrombocytopenia
- Genu valgum
- Genu varum
- Absent radius syndrome
- Congenital absence of the menisci

RESULTS

The limited amount of literature regarding absence of the cruciate ligaments indicates the relative rarity of this condition. As well, there were only 9 patients who were suspected of congenital absence of one or both cruciate ligaments in a database of approximately 13000 patients spanning 3 years at the IWK Grace Health Centre Orthopaedics Clinic. From the literature, the most commonly associated abnormalities were congenital knee dislocation and congenital leg-length discrepancies, [2-8]. Table I contains specific conditions that have been reported in cases with congenital absence of the cruciate ligaments.

Abnormal laxity of the knee is noted upon clinical examination of a subject with congenital absence of one or both cruciate ligaments, [3-6]. Subjects with absent anterior cruciate ligaments show positive anterior drawer and Lachman tests, [3-5][9], medial lateral translational instability, [4][6][7][9], and possible habitual subluxation of the tibia in extension, [4][6]. In cases where the posterior cruciate liga-

ment is absent, subjects exhibit positive posterior drawer signs, [3][8][9]. Of the 6 subjects examined, four had posterior drawer tests of less than 5 mm deviation, and two had deviations of 5 - 10 mm. There were five subjects with anterior drawer tests of 5-10 mm deviation and one with a deviation of greater than 10 mm. Five subjects also had Lachman tests of 5-10 mm deviation and one had a deviation of less than 5 mm. Five of the six subjects had a positive pivot shift. All six subjects had normal range of motion for the knee in question, and 4 of the 6 had less than 5 degrees of mediolateral laxity (the other 2 had 6-9°).

In the majority of cases reported with congenital absence of the cruciate ligaments, subjects had no complaints of instability, and even subjects in whom the knee gives way more than once a week are active. Despite the clinically unstable knee, subjects are frequently involved in sports and other strenuous activities with no difficulties, unless precipitated by an associated abnormality, [3-6]. In agreement with this, none of the 6 subjects investigated claimed to have any problems with regards to knee stability, and 4 subjects were active in athletics, with no limitations. The remaining 2 subjects were active in sports, but found that other factors limited their participation (prosthetic devices and ankle deformity). None of the subjects felt that their knee affected their daily living, nor did any subject find any limitation in walking or stairs. Two subjects mentioned occasional discomfort, although neither would refer to it as pain. One subject mentioned that she thought her knee may have "given way" once, but that it was of no consequence for her. Another mentioned that her knee had locked, but that once again the subject did not feel that it had interfered with her life. A summary of the patients seen during this study is contained in Table II. It is interesting to note that although none of the subjects had intact knees according to objective evaluation, they all had complete function (both values are out of one hundred).

Radiographic findings can be quite useful in differentiating between congenital absence and traumatic effect. Radiographic changes in the subjects missing one or both cruciate ligaments can include: hypoplasia of the intercondylar tubercles (lateral for anterior cruciate; medial for posterior cruciate), [3][4][6]; hypoplasia of the femoral condyles (medial for anterior cruciate; lateral for posterior cruciate), [4-6]; tarsal coalition, [6]; and hypoplasia to aplasia of the intercondylar emi-

Table 2: Patient Characteristics

Gender	Age (yrs)	Cruciate(s)	Knee Society Score	Associated Abnormalities
M	13	ACL	92:100	fibular hemimelia, leg length discrepancy, absent 5 th ray of foot
F	21	ACL and PCL	82:100	fibular hemimelia, leg length discrepancy
F	24	ACL	90:100	proximal femoral focal deficiency, leg length discrepancy
F	9	ACL and PCL	80:100	fibular hemimelia, leg length discrepancy
M	6	ACL (R&L)	R - 75:100 L - 89:100	absent 5 th ray (upper limb), agenesis of lateral ray (R foot), L fibular hypoplasia, R fibular aplasia, R syndactyly of toes
M	7	ACL	92:100	absent 5 th ray of foot, club foot, fibular hemimelia, leg length discrepancy

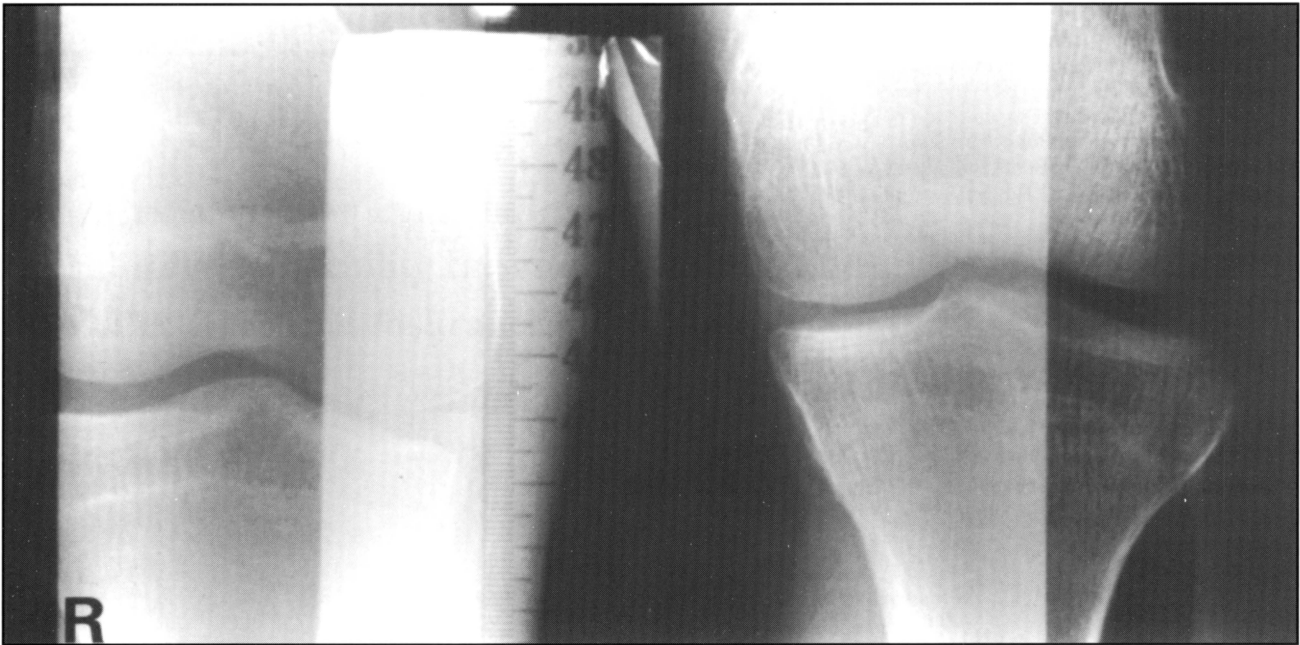


Figure 1 - Radiograph of subject with suspected absence of ACL and PCL.

nence and corresponding adjustment in the shape of the intercondylar fossa, [3][4][6][8]. Hypoplasia of the lateral femoral condyles has also been accompanied by a valgus deformity, [6]. As well, there have been reports of patellar hypoplasia and absent or hypoplastic tibia being associated with absence of the anterior cruciate ligament [3][4]. Note that while any of the above radiographic changes may occur, they may also accompany each other or not be present at all. Figure 1 presents a sample knee radiograph from this study. In the image, radiographic changes in the affected (right) knee are evident. Note in particular the hypoplasia of the intercondylar eminence and intercondylar groove.

DISCUSSION

The long-term effects on knees of patients with congenital absence of the cruciate ligaments are not completely known [3-5]. The largest concern is whether or not patients will develop the degenerative changes seen in patients who have suffered acute cruciate rupture [3-5]. This is particularly intriguing in that a study by Johansson and Aparisi (1983) contained data on a sixty year old patient with congenital absence of the anterior cruciate ligament who exhibited no significant osteoarthritis. As well, an autopsy performed on an 81 year old male with congenital absence of the cruciate ligaments and a ring meniscus showed no arthritic changes, significant at age 80 [3][5][8]. It has been postulated that the meniscus rather than the anterior cruciate ligament is a better prognosticator of degenerative joint disease, [3][5], and that ring meniscus may be a developmental change attempting to provide more stability [5]. As well, the authors of a study in which 5 patients complained of their knee giving way more than once a week, [4], have suggested that this might lead to the development of degenerative changes similar to those seen in adults who have suffered acute ligamentous disruptions;

however, this was only an assumption at the time of their study. Further long term studies are needed to evaluate this area. The current study cannot comment on long term effects because of the young age of the subjects.

Congenital absence of the cruciate ligaments is a relatively rare condition usually associated with other abnormalities. The majority of subjects suffer no ill effects in the short term, and are often active, with no difficulties or restrictions arising from the absence of the cruciate ligament(s). Patients don't have the same demand on the knee because of associated abnormalities, but they do function at a high level. Although these patients are not elite athletes, many engage in competitive sport as associated abnormalities permit. For example, one young male plays in the area hockey league, and the females in their twenties engage in recreational soccer and volleyball. These characteristics are found in both the literature and the results of this study. Finally, due to the dearth of information currently available, further work in determining the long term effects of congenital absence of the cruciate ligaments is indicated.

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