Congenital absence of the anterior cruciate ligament: eight cases in the same family

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ABSTRACT

Purpose of the study
The purpose of this study was to present a descriptive analysis of eight cases of congenital agenesia of the anterior cruciate ligament (ACL). Congenital absence of the ACL is rare. The few series reported in the literature have been limited to sporadic cases, all associated with diverse malformations. We report a series of eight cases of knees in five patients presenting congenital knee laxity. These patients were all members of the same family, raising the question of an inherited condition. We describe the characteristic features and natural history of ACL agenesia compared with post-traumatic loss of the ACL.

Material
This descriptive study included five patients (eight knees) with agenesia of the ACL. There were four men and one woman, with a mean age of 46 years. All five patients had a common ancestor.

Results
The main symptom was medial femorotibial and patellofemoral pain, present in all patients. Physical examination revealed major anterior laxity (positive pivot test, Trillat Lachman test) involving the posterior capsule and ligament structures and an abnormal knob on the anterior tibial tubercle. Plain x-rays demonstrated an abnormal aspect of the tibial spines, suggestive of ACL agenesia. The spines were flat in three knees, smooth in three, and dome-shaped in two. On the lateral view, the femoral condyles presented an abnormal rounded posterior curvature in all of the affected knees. The tibial slope was increased in all knees, 20.8° on average. MRI and arthroscopy confirmed the diagnosis in three knees.

Discussion
Examining our cases and the data in the literature led to the following remarks. The family pedigree in our patients suggested autosomal dominant inheritance. Episodes of serious laxity in subjects with congenital absence of the ACL are rare in subjects with congenital absence of the ACL compared with post-trauma patients, probably because of adaptation since infancy. Hypoplasia of the tibial spine and the lateral femoral condyle are characteristic consequences of ACL agenesia. In comparison with post-trauma cases, the natural history of ACL agenesia is characterized by better functional tolerance and inevitable progression to osteoarthritis due to the permanent anterior laxity. Degenerative disease may develop late and more slowly than after traumatic injury of the ACL. Unlike common degenerative arthritis, the lateral femorotibial and patellofemoral compartments are preserved longer.

Conclusion
Though rare, congenital absence of the ACL should be evoked as a possibility in patients with chronic anterior laxity without trauma. The radiological aspect is highly suggestive of the diagnosis which can be confirmed by MRI or arthroscopy. Study of the present series enabled a description of the natural history of ACL agenesia, which is different from that of traumatic ACL tears because of the lack of secondary meniscal lesions and the later progression of osteoarthritic degeneration.

Key words: Knee, anterior cruciate ligament, aplasia.

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INTRODUCTION

The congenital absence of the anterior cruciate ligament (ACL) is rare [Thomas et al. (1)]. Since Katz’s first publication in 1967 describing a congenital luxation of the knee [Thomas et al. (1)], exceptional cases have been reported associated with a variety of malformations: short femur, coxa vara, malformation of the leg and foot skeletal structure, discoid meniscus, and absence of medial meniscus [Thomas et al. (1), Dejour et al. (2), Carlioz (3)].

The few series reported in the literature are case studies, rarely more [Thomas et al. (1), Dejour et al. (2), Kaelin et al. (4), Noble (5)]. We report a series including eight knees with congenital laxity in five patients. The series is exceptional because all patients come from the same family, thus raising the question of the genetic origin of this condition and making this study particularly interesting.

The purpose of this study was to describe and analyze the results of this series, to determine patient characteristics, and finally to note the natural progression of ACL agenesis compared to the progression of post-traumatic ACL tear.

MATERIAL AND METHODS

This study investigated five patients, eight knees, presenting congenital absence of the ACL. Three patients had bilateral ACL agenesis. The study followed a descriptive method consisting of a case report: starting with an index case, other cases were discovered through screening.

This was a familial condition discovered by chance in 1996 in a 47-year-old patient who consulted for knee pain. Clinical and radiological examination showed bilateral gonarthrosis. The presence of a dampened brake on the Trillat-Lachman test, a positive pivot test, and a very high, direct anterior drawer sign, with no traumatic context in these arthrotic knees attracted our attention, suggesting a congenital origin for this laxity. We therefore launched a family screening survey. The ascendants, collaterals, and descendants of this patient (index case) were called in (Fig. 1), nine of whom were examined: three descendants (III4, III8, III10), four collaterals (III17), and two ascendants (II9, III8, III10, IV5).

Marriage between blood relations.

I  
II  
III  
IV  

Tree of the family.

Healthy man (examined clinically).

Healthy woman (examined clinically).

Woman with condition examined clinically (IV3).

Deceased man.

Deceased woman.

Infertile couple.

Index case.

Deceased woman with condition, diagnosis based on questioning other family members (II2).

Deceased man with condition, diagnosis based on questioning other family members (II).

Man with condition examined clinically (II9, III8, III10, IV5).

Woman with condition examined clinically (IV3).

Healthy man (examined clinically).

Healthy woman (questioned).

All our patients had a meticulous examination completed paraclinical examinations. Plain frontal and lateral, weight-bearing x-rays of the knees were taken systematically. Dynamic lateral x-rays at 90° of flexion were taken in a single patient (passive anterior drawer). An MRI was done in two patients and arthroscopies of two knees were taken in a single session.

The surgical indication was only proposed for patients with degenerative complications: one total knee prosthesis (TKP) for the oldest patient and tibial valgization osteotomy for the other two.

RESULTS

The patients’ mean age was 46 years (range, 20-69 years). There were four men and one woman.

Of the five patients, three had congenital bilateral absence of the ACL and two had unilateral absence. All patients had a common ancestor.

Clincial signs

The mean age of symptom appearance was 31 years (range, 16-47 years). The main symptom was medial femorotibial and patellofemoral pain, present in all patients. It
was mechanical and worsened when climbing and descending stairs. Although always present, knee buckling was secondary. The knees did not lock in any of the patients. According to the Arpège index [Dejour (6)], four patients were classified as active and one as sedentary.

The clinical examination found both cardinal signs of anterior laxity of the knee (positive pivot test and a positive Trillat-Lachman test) and a frank, direct anterior drawer in all patients. In six cases, frontal laxity was found. No posterior drawer was observed. All patients presented an abnormal knob on the anterior tibial tuberosity related to anterior translation of the tibia. Meticulous general examination to look for associated malformations found only bilateral flat feet in three patients. However, all patients had congenital ligament hyperlaxity.

**Complementary examinations**

**Radiological tests**

The frontal x-ray showed an abnormal aspect of the tibial spines, suggestive of ACL agenesis. The tibial spines were flat in three cases, smooth in three patients, and dome-shaped in two cases (Fig. 2). The tibial notch was abnormally narrow in all cases. There was also external separation in six out of eight knees. On the lateral x-ray, the femoral condyles presented an abnormal rounded posterior curvature in all cases (Fig. 3). Tibial slope increased in all cases a mean of 20.6°, ranging from 20° to 22° (Fig. 4). The anterior tibial translation, measured on weight-bearing images, was a mean of 8 mm, with a minimum of 5 mm and a maximum of 12 mm. This translation worsened with age and with the degree of tibial slope. According to the Ahlback classification (7), only one patient had stage IV bilateral medial femorotibial degenerative arthritis on one side and stage III on the other. This was in the oldest patient (69 years). Four other knees had beginning osteoarthrosis.

Magnetic resonance imaging (MRI) was done in two patients (three knees), 20–69 years of age. In the first patient, it showed a very hypoplastic aspect of the ACL and a normal aspect of the PCL the length of its course (Fig. 5). In the second patient, the MRI showed bilateral agenesis of the ACL, a normal aspect of the PCL (Fig. 6), bilateral meniscus involvement (the medial meniscus was luxated externally), and a substantial bilateral degenerative reorganization.

**Arthroscopy**

Exploratory arthroscopy was done in 1996 in the index case patient for both knees, concluding in a bilateral absence of the ACL, a crack in the posterior segment of the meniscus of the left knee, and both PCLs normal.

**DISCUSSION**

This case of ACL agenesis was discovered at a young age (Table 1), which can be explained by the presence of associated malformations [Thomas et al. (1), Carlioz (3), Kaelin et al. (4)]. In our series, there were no malformations associated with the ACL agenesis. Moreover, this condition was well tolerated by the majority of the patients, which could explain why it was discovered relatively late.

All of the studies published in the area reported sporadic cases. Our series differs because it involves a family series. All the patients had a common ancestor, which brings us to suggest a possible genetic component. Studying the family’s genealogical tree shows that (Fig. 1):

This is a dominant disease because one of the affected child’s parents always has the disease and the pathology is transmitted directly from parent to child without skipping a generation.

This disease is autosomal (affecting both sexes).

Its expressivity is variable, with two varied phenotypes: ACL agenesis and hyperplasia of the ACL (demonstrated on MRI).
From a practical point of view, the karyotype has no indication in cases of dominant autosomal disease [Feingold et al. (8)]. Given the familial character of our series and the absence of a genetic study on this disease in the series studied, a karyotype done on a blood sample was indicated to look for a balanced chromosomal arrangement that could help locate a gene responsible for this disease at the breakpoint (the investigation is in progress).

**Clinical study**

For our patients, the knee giving way was not a major complaint. This could be explained by the relatively advanced age of most of our patients. Dejour et al. (2)
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explain this phenomenon by the presence of degenerative arthritis or beginning degenerative arthritis: patient’s complaints center more around pain than apprehension and instability. We attribute the rareness of knee buckling in agenesis of the ACL to an adaptation lasting since childhood. In our series, patellofemoral pain was present in all cases. Dejour et al. (2) and Carlioz (3) made the same observations, explained by the circular shape of the femoral condyles. In a normal knee, the femoral condyles are covered with an articular surface occupying their anterior, inferior, and some of the posterior facet. The lateral condyles draw a spiral curve whose radius decreases from front to back [Kapandji (9), Frain et al. (10)]. The round shape of the condyles entails a second, rolling component, rather than a combination of rolling and sliding [Kapandji (9)], which leads to an increase in the pressure of the patella against the trochlea, which may be the cause of the patellofemoral pain.

No studies report knee locking. In the series published in the literature, ACL agenesis is always associated with agenesis or a ring aspect of the medial meniscus. [Thomas et al. (1), Dejour et al. (2), Nobel (5), Tolo (11), Curtis et al. (12)], which could explain the absence of knee locking.

The association of ACL agenesis and an absence of medial meniscus is related to their common embryological origin [Clark et al. (13)]. These two structures differentiate between the 4th and 9th weeks of gestation from a single mesenchymal tissue that then differentiates into collagen fiber and fibroblasts at the end of the 3rd month of gestation. At this time, the knee joint has been clearly individualized. We observed no congenital abnormalities in the cases explored by arthroscopy or MRI.

Front laxity was found in six knees and can be explained by a false ligament distension secondary to degenerative lesions corresponding to wear laxity, whereas the literature reports this laxity to be secondary to a congenital condition of the collateral ligaments.

The Trillat-Lachmann test and the pivot test, two major signs of anterior laxity of the knee, were frankly positive. Similarly, the direct front drawer was positive, a sign of ACL and posterior capsule and ligament structure involvement [Imbert et al. (14)]. In this series, no direct posterior drawer was found. This is evidence of a healthy PCL. An association of ACL and PCL absence was found by Thomas et al. (1), Carlioz (3), Kaelin et al. (4), Johanson et al. (15). As for Dejour et al. (2) and Tolo (11), absence of the ACL was concurrent with normal PCL.

Conventional x-rays

Hypoplasia of the tibia spines was noted in all cases. Thomas et al. (1), Dejour et al. (2), Carlioz (3), Kaelin et al. (4), and Johanson et al. (15) all reported this anomaly. This hypoplasia could be partly explained by the hypothesis put forward by Giorgi [in Dejour et al. (2)], which suggests that the development of the tibial spines stops if there is no traction from the ACL. The rounded shape of the lateral femoral condyle was found in all cases. This observation was also reported by Thomas et al. (1), Dejour et al. (2), Carlioz (3), and Kaelin et al. (4), and may in part be explained by the Delpech law [in Dejour et al. (2)]: during anterior subluxation and hyperextension, stresses are at a maximum level at the back of the condyles and the tibial articular surface disturbs of the bone’s development.

Absence of the ACL leads to abnormal anterior tibial translation caused by shear force [Dejour et al. (16)]. This translation is increased when both ACL and the medial meniscus are absent [Dejour et al. (2), Curtis et al. (2), Johanson et al. (5)]. In the series studied here, an 8-mm anterior tibial translation with a minimum of 5 mm and a maximum of 12 mm was observed. This laxity was less than that found in the literature. Dejour et al. [2] report 30-mm anterior tibial translation in one case of congenital bilateral ACL absence. Tibial translation was moderate in our series, perhaps because of the presence of the medial meniscus in our patients, whereas it was absent in the other series.
We observed an increased tibial slope in all cases (mean, 20.6°) and the anterior tibial translation was even greater when the tibial slope value was high. The same observations were reported by Dejour et al. (2).

Therapeutic measures

There is no indication for surgical reconstruction of the ACL in cases of agenesis, for several reasons: congenital absence of the ACL is often well tolerated [Carlioz (3), Johanson et al. (15)]; reconstructing the anterior cruciate ligament in cases of substantial laxity of the knee can involve difficult technical problems, with a high level of failure or poor results [Carlioz (3)]; the shape of the articular surfaces is abnormal and it does not seem logical in these conditions to attempt to establish normal kinematics of the knee [Dejour et al. (2), Carlioz (3)]. In our opinion, surgery was only proposed for patients with degenerative complications (total knee prosthesis, valgization tibial osteotomy).

Comparative study of the progression of post-traumatic and congenital laxity

Traumatic ACL lesions are frequent and well known, as is their potential for further joint disorders [Imbert et al. (14), Aubriot et al. (17), Segal et al. (18) Dejour et al. (19)]. The period of functional tolerance is highly variable, with a natural history ranging from 10 to 50 years, with a mean period of 35 years in patients with a low level of activity and little axial deviation [Dejour et al. (19)]. In cases of meniscectomy, this period of tolerance decreases to a mean of 25 years [Dejour et al. (19)].

Absence of ACL is rare. The few series reported in the literature and the low number of cases does not allow the natural progression of this condition to be studied. Nevertheless, our series is particular in that five patients had a wide range of ages (16, 26, 47, 56, 69 years) and some knees were explored several times at 4-year intervals.

To assess the natural progression of ACL agenesis, our patients, all members of the same family with relatively similar levels of physical activity, we simulated a single “fictitious patient,” using data from the youngest member of the family (16 years old) through to the oldest member of the family (69 years old). This study included a comparative and progressive analysis, both clinical and radiological, based on frontal and lateral x-rays of the knees en charge. Our purpose was to identify the natural progression of this pathology and its specificities.

Although short, the chronological study of our series, allowed us to make the following observations:

• The functional tolerance is good at least for 26 years. By 26 years of progression, no meniscus lesion has complicated the clinical picture, which is exceptional in cases of traumatic tear of the ACL.
• The first signs of arthritic degeneration appear between 26 and 47 years of progression.
• The appearance of osteoarthritis with congenital anterior laxity is unavoidable. This osteoarthritis particularly involves the medial compartment. The lateral femorotibial and patellofemoral compartments remain healthy for a very long time. The condition evolves more slowly that osteoarthritis after traumatic ACL tear. This can be explained by mechanisms of adaptation.

• There is frontal laxity expressed by external separation with no lateral subluxation

CONCLUSION

Even though congenital absence of the anterior cruciate ligament is rare, it must be raised when there is a clinical picture of chronic anterior laxity but no traumaticism. The radiological aspect helps diagnosis, which is confirmed by MRI or arthroscopy. The familial aspect of our series should encourage a genetic investigation when agenesis of the ACL is found. The study of this series allowed us to observe that the natural progression of ACL absence may be different than that of traumatic rupture of the ACL when there are no secondary meniscus lesions and a later appearance of degenerative lesions. The reason for this difference may be progressive morphological adaptation of the knee to stresses altered by the absence of the ACL.

References


