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A case of multiple congenital anomalies including agenesis of the anterior cruciate ligament

Ön çapraz bağ agenezisini de içeren çoklu doğuştan anomalili bir olgu

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Ön çapraz bağın (ÖÇB) doğuştan agenezisi nadir rastlanan bir durumdur. Tanısı genellikle artroskopi sırasında fark edilerek konmaktadır. Bu yazıda, tanısı fizik muayene ve manyetik rezonans görüntüleme ile konan 15 yaşında bir erkek hasta sunuldu. Hastada doğuştan ÖÇB agenezisine ek olarak, femur boyunda kısalık, tibial interkondiler eminenste displazi, dizde valgus ve omurgada kompansatuvar skolyoz vardı. Hastanın diziyle ilgili hiçbir instabilite şikayetinin bulunmaması, pivot-shift testinin negatif olması ve çoklu doğuştan anomaliler bulunması nedeniyle ameliyat düşünülmedi.

Anahtar sözcükler: Ön çapraz bağ/anormallik/radyografi; femur; diz eklemi.

Congenital absence of the anterior cruciate ligament (ACL) is a very rare anomaly. It is usually diagnosed during arthroscopic intervention. We presented a 15-year old male patient whose diagnosis was based on findings of physical examination and magnetic resonance imaging. Congenital absence of the ACL was associated with femoral shortening, dysplasia of the tibial intercondylar eminence, valgus knee, and compensatory scoliosis. Since the patient had no instability symptoms, and due to the presence of a negative pivot-shift test and multiple congenital anomalies, surgical treatment was not considered.

Key words: Anterior cruciate ligament/abnormalities/radiography; femur; knee joint.

Absence of the anterior cruciate ligament (ACL) is a very rare congenital disease.[1-4] This condition may be associated with other embryologic abnormalities of the lower limb such as shortness of femur, hypoplasia of the intercondylar tibial eminence, of the intercondylar notch and discoid meniscus.[5-10]

We describe a case of absence of the ACL associated with other congenital anomalies in an adolescent without any knee instability.

Case report

On beş yaşında erkek hasta, başka bir merkezde A 15-year-old boy was referred to our clinic to have a reconstruction operation which was recommended by another clinic. He had complaints of pain on the anterior part of his left knee. His symptoms had started 3 weeks prior to his index evaluation after having a contact injury on his knee during playing basketball. He applied to an orthopedic surgeon initially with the complaints of mild knee pain that started after the trauma. Magnetic resonance imaging (MRI) views were ordered by the initial physician surgeon. MRI images were interpreted as an ACL rupture hence. The patient went through a comprehensive evaluation by the senior author. His history revealed that the patient was repeatedly seen by orthopedic surgeons because of leg length discrepancy since his infancy. Shortness of the left femur was diagnosed at the age

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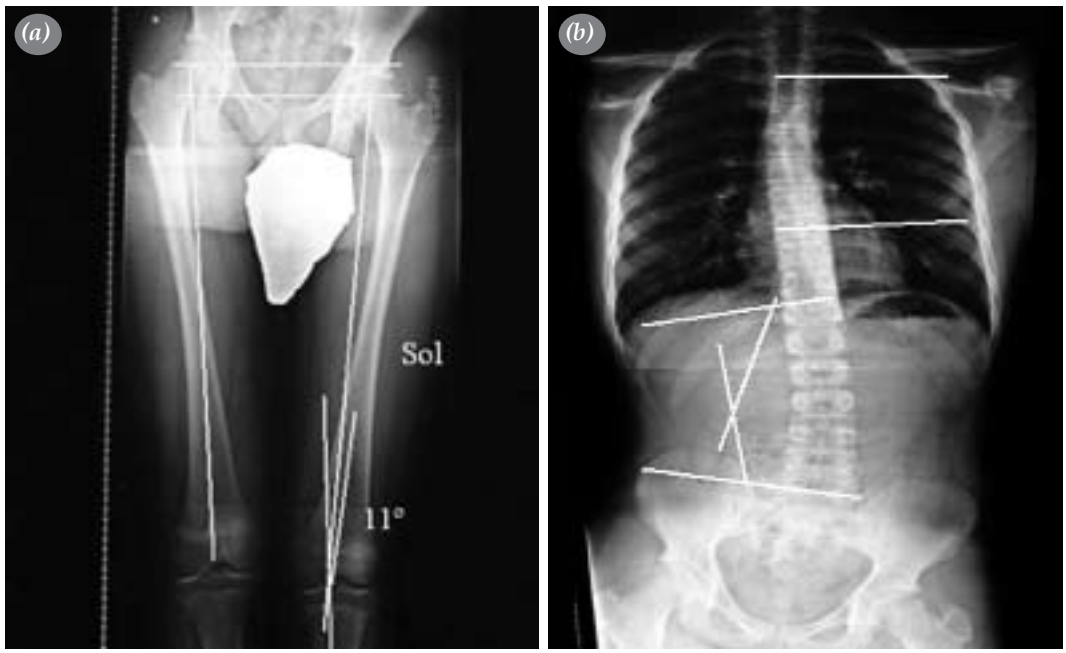

Figure 1.(a) AP view of lower extremity. Shortness of femur, dysplasia of intercondylar tibial eminence and 11 o of valgus can be seen on the left leg and left knee. **(b)** AP view of vertebral column. Compensatory vertebral scoliosis can be seen, especially in lumbar region.

of 2 and followed by an orthopedic surgeon until the age of 10. It was concluded by the responsible physician that it was an idiopathic situation and simple shoe lift was recommended. Previous x-rays dating back to 4 years of age revealed shortness of femur on the left leg. Current x-rays showed dysplasia of the intercondylar tibial eminence, 11o valgus on the left knee and compensatory vertebral scoliosis (thorocal Cobb 5 o, lumbar Cobb 22 o) in addition to 3 cm shortness of femur (Figure 1). At the time of examination the patient had a pain level of 2 on a scale ranging from 0 – 10. His pain level had been decreasing since the time of injury. On inspection, left knee had 10o valgus. Q angle was in its normal range. There was a leg length discrepancy of 3 centimeters disfavoring the left side when measured from the anterosuperior iliac spine to the medial malleolus. On physical examination, there was no effusion or tenderness on his left knee with full range of motion. Ligament tests involving varus-valgus stress, anterior-posterior drawer and pivot-shift tests were all negative. Medial and lateral Mc-Murray tests were negative as well. On his MRI, absence of ACL and thinness of the posterior cruciate ligament (PCL) were clearly seen (Figure 2).

It was decided to follow the patient with first line analgesic consisting of paracetamol in addition to basic knee musculature strengthening exercises. Since the patient did not complain of about any instability problem, it was decided that there was no need to carry out an ACL reconstruction operation. Bearing in mind that the patient did not have any pain, locking, swelling or other kind of problem that would hint a mechanical irritation, diagnostic arthroscopy was not performed either. His decreasing symptoms in follow-up examinations were correlated with the absence of any mechanical irritation. In the sixth

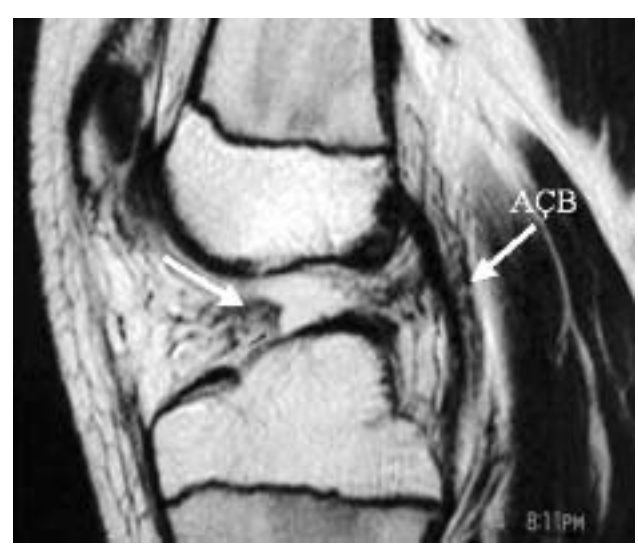

Figure 2. On sagittal plane MRI of the left knee, absence of anterior cruciate ligament and a posterior cruciate ligament which is thinner than normal is visible.

month of follow-up, his last examination was done and there was no pathological finding.

Discussion

Unilaterally or bilaterally congenital absence of the ACL is not an infrequent a rare condition.[1-5] This situation may be associated with discoid meniscus[2], hypoplasia of intercondylar notch, hypoplasia of intercondylar tibial eminence, shortness of femur, hypertrophy of the meniscofemoral ligament of Humphrey, and fibular hemimelia.[11]

When we look at the embryology of the knee, the connective tissue and cartilage of the knee develops from the interzonal mesenchyme. Ratajczak[12] has shown that the cruciate ligaments first appear in stage 19 (nearly 39th postovulatory days). The primordial cells, lie in the middle part of the interzone which faces the intercondylar fossa of the femur where the cells are more loosely arranged in obliquely passing strands, are forming the cruciate ligaments. During stage 22, intercondylar tibial eminence formation occurs, and in stage 23 all intraarticular structures are formed and the shafts of the tibia and femur begin to ossify.

In our case, the congenital origin of the condition must be strongly suspected if not proved because of its association with other previously reported joint anomalies, such as shortness of femur, dysplasia of the intercondylar tibial eminence and thinness of the PCL.[7-10] ACL is the primary restraint for anterior translation loads affecting the knee. Rupture of the ACL results in an unstable knee. Patients with a rupture of the ACL may not always feel the instability symptoms depending on the velocity and the magnitude of the loads. Low velocity and low magnitude loads may easily be absorbed by the muscle based reflex mechanism, however higher magnitude loads will not be able to resisted by an ACL ruptured knee. Irrelevant of the presence of instability symptoms, patients with rupture of the ACL will always have a positive pivot shift sign.

In spite of an absent ACL the patient did not have a positive lachman, a positive anterior drawer or a positive pivot shift sign. Absence of the instability related physical findings may be explained by the angulatory deformation of the knee. Valgus osteotomy alone or in addition to ACL reconstruction is a well accepted procedure for complex anterior instability cases. Valgus knees do not experience instability episodes.[14]

Other conditions such as other intraarticular structures may mask the expected pivot shift test. ACL compensating hypertrophied PCL is an example.[13] But we didn't see such any structure on MRI. Besides, compensatory scoliosis is unique for this case.

Since there is no standardized therapeutic approach in the case of congenital absence of the ACL, some of the studies propose surgical treatment with ACL reconstruction alone or associated with other extraarticular procedures to correct associated anomalies. Others suggest only conservative treatment with good results.[4] Kaelin et al.[5] reported 6 cases, 4 of which were treated conservatively and remaining surgically (a simple meniscectomy and a ligament reconstruction). The authors reported good results in the patient treated by meniscectomy alone and improved stability at clinical examination of the patient treated by ligament reconstruction.

Previous studies do not clearly mention the situation of the pivot shift of the patients reported. However, presence of functional ACL deficiency is primarily necessary to determine the therapeutic strategies. We have decided that the patient's clinical presentation has not warranted any kind of surgery due to 2 main reasons. First, the patient did not seek medical attention because of instability symptoms; secondly the patient did not have positive pivot shift sign. Since the ligament reconstructions are done only in the case of symptomatic and unstable knee, ACL reconstruction surgery was not considered.

This patient has shown us that a knee with a congenital absence of ACL may not have any instability symptom or finding. Although loss of continuity of the ACL due to traumatic reasons may lead to ACL reconstruction, congenital absence of the ACL may not require ACL reconstruction as in this case.

Johansson and Aparisi[1] also mentioned that risk of early osteoarthritis in congenitally absence of ACL is very low because none of their five younger patients had any arthroscopic or radiographic signs of osteoarthritis, also in the sixth patient whose instability had been present for sixty years without causing any significant osteoarthritis.

As a result, while absence of the ACL can sometimes be together with a severe articular instability and

frequent giving way episodes, can also have a negative pivot-shift test. While these are the possibilities, the most important point is to find the right theraphy among the therapeutic index in between surgical ACL reconstruction and only a yearly follow up according to the patient's age, type and level of activity, quality and amount of symptoms and the physical examination.

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