Matching Osteochondritis Dissecans Lesions in Identical Twin Brothers

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abstract

Osteochondritis dissecans is a disorder of unknown etiology that can result in fragmentation of osteochondral surfaces, most commonly of the knee, shoulder, elbow, and ankle. This may lead to sequelae of pain and an inability to participate in desired activities. Multiple theories exist as to the true cause of the disorder, but none have been fully proven. One such proposed etiology is genetic causation.

Familial cases of osteochondritis dissecans are rare, yet these cases offer support to growing evidence that may support a genetic link. This article describes osteochondritis dissecans lesions of the femoral trochlea in monozygotic (identical) twins. Both twins presented with similar symptoms 1 year apart. Neither twin had any clear inciting trauma. Magnetic resonance imaging revealed osteochondral lesions in similar positions of the lateral trochlear of the same knee in both brothers. Osteochondral autograft transfer and tibial tubercle anteromedialization were performed on both patients. An identical postoperative protocol was followed, and recovery with full return to sport was comparable for the brothers. To the authors’ knowledge, only 1 other case report exists of osteochondritis dissecans lesions in monozygotic twins.

Although debate continues regarding the true etiology of this disorder, cases of identical twins presenting with a similar disease process are highly suggestive of a genetic component and may lead to early identification and treatment of these lesions. Continued research in the area of osteochondritis dissecans and its genetic basis is needed to completely understand this disorder.

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Osteochondritis dissecans, an idiopathic condition involving the articular surface of a joint, can affect any number of joints within the body. First described in the late 19th century by Sir James Paget,1 osteochondritis dissecans earned its name from Franz König,2 who felt it was attributable to dissecting inflammation of the joint cartilage. During the past 120 years, many theories have been proposed for the etiology of osteochondritis dissecans; however, the precise etiology of this condition is yet to be determined.

Familial cases of osteochondritis dissecans are rare, yet evidence exists to support a genetic link. Lee et al3 describes bilateral femoral head osteochondritis dissecans lesions involving a father, his son, and his nephew. Mei-Dan et al4 and Kenniston et al5 presented cases of osteochondritis dissecans lesions in dizygotic, or fraternal, and monozygotic, or identical, twins, respectively. Most relevant report to the current articles is the case by Onoda et al,6 which describes identical twins with osteochondritis dissecans lesions, both of which achieved successful union after surgical intervention for unstable osteochondritis dissecans lesions. These reports give support to theories of a genetic predisposition, which has yet to be completely understood or identified, but coincide with the findings of our case reports.

This article describes osteochondritis dissecans lesions of the femoral trochlea in identical twins. The brothers presented with similar clinical symptoms within 1 year of each other, and neither brother had a traumatic event preceding the onset of his knee pain. Both were active in high school athletics. The brothers underwent the same surgical procedure and rehabilitation and were able to return to competitive sport. Identical twins with osteochondritis dissecans lesions have been described previously5,6; however, the current cases are interesting and unique because the osteochondritis dissecans lesions occurred in the same location of the left knee and became symptomatic at approximately the same time.

**CASE REPORT**

**Patient 1**

In December 2010, a 15-year-old boy presented to his pediatrician with diffuse left knee pain and intermittent swelling that had been present for at least 1 year. No mechanical symptoms were present, and he reported no inciting injury. After failing to improve with conservative treatment of nonsteroidal anti-inflammatory drugs and rest from athletic activity, he was referred to an orthopedist. Orthopedic examination demonstrated full range of motion, no mechanical symptoms, mild joint effusion, and pain localized anteriorly near the patellofemoral joint that worsened with active extension.

Magnetic resonance imaging revealed an osteochondral lesion of the lateral trochlea. The lesion, within the concav-
ity of the trochlea, measured $16 \times 20$ mm with a depth of 6 mm with fluid signal beneath the fragment, indicating instability (Figure 1). Loose body removal, lateral release, osteochondral autograft transfer, and tibial tubercle anteromedialization were performed (Figures 2A, B).

Two months postoperatively, the patient exhibited mechanical symptoms in the operative knee. Magnetic resonance imaging indicated intra-articular loose bodies present. During arthroscopic removal of the loose bodies, an inspection of the previous osteochondral autograft transfer showed stable and smooth incorporation of the transplanted osteochondral plugs (Figure 2C). Postoperative rehabilitation was followed, and at 6 months he was cleared to return to competitive sports. The patient has had no further issues to date.

**Patient 2**

Approximately 1 year after his identical twin brother’s initial visit, the patient presented with a 4-month history of diffuse knee pain that worsened with activity. Examination demonstrated full active range of motion, positive patellar grind test, and no effusion.

Magnetic resonance imaging revealed a $22 \times 10$-mm osteochondral lesion with a depth of 8 mm on the left lateral trochlea (Figure 3). Fluid behind the fragment indicated an unstable lesion. Loose body removal, osteochondral autograft transfer, tibial tubercle transfer, and lateral release were performed (Figure 4). The patient followed a postoperative protocol similar to that of his brother A. He progressed well, with full return to competitive sports at 6 months postoperatively.

**DISCUSSION**

Osteochondritis dissecans is a condition of unknown etiology. It most commonly affects the knee and is a source of pain in adults and juveniles. In their cumulative review of knowledge on this disorder, Edmonds and Polousky recognized osteochondritis dissecans as a lesion affecting subchondral bone with varying degrees of osseous resorption and collapse. The overlying articular surface appears to undergo softening, which then progresses to separation of the articular piece and subsequently results in osteochondral loose bodies. Despite various suggested etiologies, such as trauma (repetitive microtrauma vs acute macrotrauma), inflammation, constitutional factors including abnormal collagen formation, and vascular abnormalities, no clear cause exists for this disorder.

The appearance of osteochondritis dissecans lesions in identical twin brothers without an inciting event leads to a discussion regarding a potential genetic basis for this disorder. Both brothers were competitive high school athletes, which could provide some support to the hypothesis of repetitive trauma injury that may be more
prominent in athletes. The European Pediatric Orthopaedic Society reported that 55% of patients with osteochondritis dissecans lesions were regularly involved in sports or performing strenuous athletic activity.10

Even with these findings potentially supporting a repetitive microtrauma etiology, it seems more than coincidence that identical twins would develop similar lesions at the same location within their knee around the same time in life. This is reasonable evidence to suggest that the etiology of osteochondritis dissecans has a genetic component. Stattin et al11 reported a missense mutation resulting in abnormal C-type lectin, which normally mediates interactions of proteins and the cartilage extracellular matrix. Five generations of a family were studied, and an autosomal-dominant inheritance pattern for osteochondritis dissecans was revealed.11 Other studies have also reported an autosomal-dominant familial inheritance pattern for osteochondritis dissecans.12,13 The current case report provides further evidence to support a genetic basis for osteochondritis dissecans.

CONCLUSION

This article described identical twins, both highly active in competitive sports, with essentially identical trochlear lesions. Despite this case report and others denoting a familial incidence, it seems likely that this disorder may be multifactorial. Identical twins with similar histories, lesion locations, and timing are highly suggestive of a genetic component. However, the true etiology of this disorder remains controversial. Contributing variables, such as athletic endeavors with presumed repetitive stress and microtrauma, need to be considered. Although this case report does not identify the true etiology of osteochondritis dissecans, it lends substantive support to a genetic link. Clinicians should be aware of a potential genetic predilection for osteochondritis dissecans, in addition to other potential contributory factors, when treating early joint pain in siblings of osteochondritis dissecans patients so that they can facilitate prompt examination and timely diagnosis, which may potentially alter treatment.

REFERENCES