Identical Osteochondritis Dissecans Lesions of the Knee in Sets of Monozygotic Twins

Itai Gans, MD; Eric J. Sarkissian, MD; Struan F.A. Grant, PhD; Theodore J. Ganley, MD

abstract

Osteochondritis dissecans (OCD) is a focal, idiopathic alteration of subchondral bone structure with the risk for secondary damage to adjacent articular cartilage and the development of premature osteoarthritis. The exact etiology of OCD is unknown, although repetitive microtrauma and vascular insufficiency have been previously described. A genetic predisposition has been suggested, but the existing evidence is sparse. There are multiple case reports of twins and siblings with OCD and a few large family series in the literature, promoting the theory that OCD may have a genetic component to its etiology.

This article describes 2 sets of monozygotic twins presenting concurrently with OCD of their dominant knees, offering further support for a genetic component to the etiology of OCD. Interestingly, in both sets of twins, 1 was left-handed and 1 was right-handed. Both sets of twins had simultaneous presentations and clinical courses, lending support to a genetic element to OCD. The development of the OCD lesion in the dominant knee of each patient suggests an environmental influence, perhaps due to repetitive microtrauma and overuse.

Recently, a genome-wide linkage study identified a prime candidate locus for OCD. However, despite the suggested association, genetic and developmental factors in the development of OCD remain relatively unstudied. The authors believe monozygotic twins provide an excellent clinical opportunity for future examination of the role of familial inheritance in the etiology of OCD.

Figure: Anteroposterior radiograph (A) and sagittal magnetic resonance image (B) of the osteochondritis dissecans lesion in case 2, twin A.
Osteochondritis dissecans (OCD) is a focal, idiopathic alteration of subchondral bone structure with the risk for secondary damage to adjacent articular cartilage and the development of premature osteoarthritis. These changes most commonly occur at the medial femoral condyle and can manifest as early articular cartilage separation, partial detachment of an articular lesion, and osteochondral separation with loose bodies. The etiology of OCD remains unclear, and no theory regarding the cause of OCD is universally accepted. Repetitive microtrauma, secondary effects associated with vascular insufficiency, avascular necrosis, inherited factors, and genetic predisposition have been proposed as causes of OCD. Despite the suggested association, genetic and developmental factors remain relatively unstudied, and few cases of monozygotic twins with OCD of the knee have been reported.

This article describes 2 sets of monozygotic twins, each with 1 left-handed and 1 right-handed sibling, with dominant-knee OCD of the weight-bearing surface of the femoral condyles.

**Case Reports**

**Case 1**

Seventeen-year-old monozygotic twin brothers concurrently presented to the authors’ clinic for evaluation of chronic knee pain. Twin A, a right-hand–dominant football player, reported activity-induced right knee pain and a positive history of intermittent mechanical symptoms. Twin B, a left-hand–dominant lacrosse player, reported left knee discomfort. Neither twin described a traumatic event prior to symptom onset. On physical examination, both twins had positive lateral joint line and lateral femoral condyle tenderness to palpation. Both twins had stable knees with full range of motion, no knee effusions, and no signs of pathological laxity.

Plain radiographs of the affected knees raised suspicion for lateral femoral condyle OCD. Magnetic resonance imaging (MRI) of twin A’s right knee demonstrated a large OCD lesion at the weight-bearing portion of the lateral femoral condyle with underlying subchondral cystic changes and a partial discoid lateral meniscus. Twin B’s MRI revealed the same findings in the left knee. Based on the low likelihood of healing with conservative measures in both cases, the surgeon and family agreed to proceed with surgery.

Arthroscopy of twin A’s right knee revealed a discoid lateral meniscus with intrasubstance delamination and tearing and a large, friable OCD lesion exceeding 2.5×2.5 cm with fractured cartilage fragments. Twin A underwent a partial meniscectomy, loose body removal, and autologous chondrocyte implantation to address the unsalvageable OCD lesion.

Arthroscopic evaluation of twin B’s left knee revealed a large (3×3 cm) but intact OCD lesion as well as a partial discoid lateral meniscus. A partial lateral meniscectomy and drilling of the OCD lesion were performed. Both twins progressed without complication at a minimum 6-month follow-up.

**Case 2**

Twin A, a 14-year-old, left-hand–dominant, multisport male athlete, presented to the authors’ clinic for evaluation of chronic left knee pain. Findings on physical examination were within normal limits, with full range of motion, no tenderness about the knee, and no effusion. Knee radiographs and MRI indicated a stable, intact OCD lesion at the lateral aspect of the medial femoral condyle (Figure 1). A 6-month trial of nonoperative therapy consisting of activity modification, bracing, and physical therapy did not improve the patient’s symptoms. Arthroscopic transarticular drilling of the OCD lesion was performed. At 14-month follow-up, knee radiographs revealed no osseous abnormalities, and the patient had resumed all activities.

Twin B, a right-hand–dominant male athlete, presented at age 16 years for evaluation of chronic activity-induced right knee pain. Physical examination revealed normal range of motion and no tenderness or effusion. Knee radiographs and MRI revealed a 2.9×2.9-cm OCD lesion at the lateral aspect of the medial femoral condyle with subchondral cystic changes and irregular overlying cartilage (Figure 2). Arthroscopic transarticular drilling and bone grafting of the OCD lesion were performed. At 6-month follow-up, knee radiographs revealed no osseous abnormalities, and the patient had resumed all activities.

**Discussion**

Osteochondritis dissecans can be incapacitating, potentially leading to premature arthritis and considerable pain. The exact etiology of OCD is unknown, but theories include repetitive microtrauma, vascular insufficiency, avascular necrosis, inflammation, and genetic predisposition.

Previously, 3 cases of monozygotic twins with OCD lesions of the knee were reported.
Of particular interest, both sets and endocrine disorders. Lee et al identified a mutation in all with a seemingly conducted a study of OCD have been described by many others. Lee et al reported 10 cases of asymptomatic men, Nielsen noted OCD in 4.1% of those studied. In addition, 14.6% of the male relatives of affected men showed unmistakable radiographic evidence of OCD, providing strong evidence for a genetic component to OCD. Reports of familial OCD have been drawn on many others. In 1977, Petrie reported that genetic causation could not be determined in a 3-generation family, concluding a likely genetic influence. In addition, many reports on 3- to 5-generation familial cases of OCD exist, all with a seemingly autosomal-dominant mode of inheritance with fairly high penetrance. Furthermore, OCD has been reported to have a high association with genetic syndromes, including Stickler’s syndrome, familial short stature, and endocrine disorders.

More recently, genetic linkage assays have demonstrated a few polymorphisms associated with the development of OCD. Jackson et al identified a mutation in COL9A2, located in an exon splice site, which was found to be associated with OCD lesions in 2 unrelated families. In 2010, Stattin et al conducted a genome-wide linkage study and identified aggrecan (ACAN) as a prime candidate locus for OCD.

The current literature regarding the genetic nature of OCD is mostly low evidence and still lacking. Future higher-quality studies must be conducted to determine the extent of a genetic relationship in the etiology of OCD. The current article described 2 sets of monozygotic twins developing OCD lesions of the knee. Both sets of twins presented with similar complaints and were found to have similar OCD lesions on the weight-bearing surface of their dominant knees. The authors believe that monozygotic twins offer an excellent opportunity to examine the possibility of a genetic predisposition for OCD.

**REFERENCES**

