Femoral trochlear dysplasia is an anatomic deformity that predisposes patients to patellar instability, including patellar subluxation and dislocation, and can lead to severe patellofemoral joint degeneration if left untreated. Femoral trochlear dysplasia leading to recurrent bilateral patellar dislocation has rarely been reported as having a familial association. Orthopedic surgeons who encounter patients presenting with chronic patellar instability without an underlying disease or syndrome should be aware of the presence of femoral trochlear dysplasia leading to recurrent bilateral patellar dislocation. Although femoral trochlear dysplasia remains uncommon, the presence of bilateral recurrent patellar dislocation in multiple members of the same family is highly suggestive of genetic inheritance.

This article describes 3 patients from 1 family who presented with femoral trochlear dysplasia leading to recurrent bilateral patellar dislocation. To our knowledge, this is the second article to describe a familial form of femoral trochlear dysplasia associated with recurrent bilateral patellar dislocation and is the first article in English. A lower threshold for screening and early intervention for symptomatic family members may be indicated to prevent the long-term effects of chronic patellar subluxation, dislocation, and patellofemoral arthritis.

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Drs Rebolledo, Nam, Cross, Green, and Sculco have no relevant financial relationships to disclose.

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doi: 10.3928/01477447-20120327-30

Figure: Preoperative anteroposterior (A) and Merchant (B) radiographs of 54-year-old woman with bilateral knee osteoarthritis and femoral trochlear dysplasia. Postoperative anteroposterior (C) and merchant (D) radiographs 1 year after bilateral total knee arthroplasty.
Patellar instability is common in patients with femoral trochlear dysplasia. Abnormalities in the bony geometry of the femur, such as a shallow trochlear groove or deficient lateral trochlear margin, can lead to chronic patellar instability and recurrent dislocation. Although several anatomic variants contribute to patellofemoral instability, femoral trochlear dysplasia exists in 85% of patients with recurrent patellar dislocation. Complications from chronic patellofemoral instability include recurrent dislocation, patellofemoral osteoarthritis, and loss of flexion from a permanently dislocated patella. In young, skeletally immature patients, early conservative measures and possible surgical management should be considered to prevent the progression of patellofemoral deformity and the development of patellofemoral osteoarthritis. Familial forms of recurrent patellar dislocation have been described in patients with normal trochlear anatomy. However, femoral trochlear dysplasia leading to recurrent bilateral patellar dislocation has rarely been reported as having a familial pattern or genetic component. This article describes 3 patients from 1 family who presented with femoral trochlear dysplasia leading to recurrent bilateral patellar dislocation.

CASE REPORT

Patient 1

A 54-year-old woman presented with a 20-year history of progressively worsening bilateral knee pain. The pain was severe in both knees but was worse in the right knee. The patient reported diffuse knee discomfort, anterior knee pain while ascending and descending stairs, and increased pain at night. The patient had been using a cane to ambulate for the past 9 years and had tried conservative measures, including nonsteroidal anti-inflammatory drugs (NSAIDs) and several physical therapy trials, which provided limited relief.

She reported a history of recurrent bilateral patellar dislocation that had started during early childhood, which was unrelated to previous trauma. She reported that her grandmother had similar symptoms, including recurrent bilateral patellar dislocation. Her medical history was significant for gout, which was limited to her right ankle and was being medically managed with allopurinol and colchicine. Pertinent surgical history included previous bilateral patellar realignment surgery >40 years previously in her home country of Laos; however, the surgical procedure details were unknown to the patient. Despite the previous surgery, the patient's symptoms of patellar instability had not resolved. She reported no alcohol or tobacco use.

On examination, the patient demonstrated a windswept deformity with her right knee in varus and left knee in valgus. Right knee active range of motion (ROM) was maximum flexion of 120° and full extension of 0°, with an appreciable mild effusion. Left knee ROM was maximum flexion of 120° and full extension of 0°. Bilaterally, the patient had mediolateral joint line tenderness and patellofemoral crepitus. Pain and positive patellar apprehension occurred in both knees during manipulation. Neurovascular examination was intact in both lower extremities.

Standing anteroposterior knee radiographs showed significant bilateral joint space narrowing. The right knee demonstrated joint space narrowing in the medial compartment with an anatomic valgus deformity of 3°. The left knee demonstrated osteoarthritic changes in the lateral compartment, with an anatomic varus deformity of 18° (Figure 1). Merchant view showed bilateral trochlear hypoplasia with a poorly visualized right patella and laterally subluxed left patella. The left femoral sulcus angle was 151°, whereas the right femoral sulcus angle was 150°. Using Tavernier Dejour classification, both knees were dysplasia type C. This classification was determined due to the presence of a shallow trochlear sulcus, supratrochlear spur, and asymmetry of the trochlear facets.

The patient underwent single-stage, bilateral total knee arthroplasty (TKA) under a spinal and epidural anesthetic. A medial parapatellar approach was used for both knees. During the procedure, the trochleas of both femurs were severely dysplastic, and both patellae were severely hypoplastic, with a thickness of approximately 17 mm. An Optetrak cemented posterior cruciate-substituting femoral prosthesis with a cemented tibial tray (Exactech, Gainesville, Florida) was used for both knees. Anterior referencing was used for femoral component sizing, and the transepicondylar axis was used for femoral component rotation. After
component implantation, both knees were passively flexed to 110°, which resulted in lateral patellar dislocation. An extensive release was performed through the lateral retinaculum using electrocauterization. This release extended from approximately 2 to 3 cm proximal to the patella, distally to the tibial component, and approximately 1 cm lateral to the patellar border. After performing the lateral release, patellar tracking was improved. Both patellae were maintained in the trochlea at maximum flexion. The wounds were irrigated and closed, and an intra-articular drain was placed. The patient tolerated the procedure well and was made weight bearing as tolerated with physical therapy on postoperative day 1. She was discharged on postoperative day 4.

At 6-week follow-up, the patient had decreased pain and improved ambulation. Both wounds had healed; however, residual swelling existed in both knees. The patient began to transition away from using a cane and was continuing with physical therapy. No patellar instability existed on examination. Active ROM maximum flexion was 95°, and extension was −5° in both knees. At 1-year follow-up, the patient had full extension and flexion to 110° bilaterally with no patellar instability or knee pain. Radiographs showed excellent prosthesis alignment and fixation.

**Patient 2**

A 6-year-old boy, the son of the patient 1, presented with episodes of frequent unintentional falls. Patient 1 described him as clumsy. The patient’s parents reported no history of trauma; however, instability was reported to precede many of the patient’s falls. The patient reported no pain during these episodes and no difficulty during physical play with his peers. The patient’s medical history included mild asthma, but he took no medications. The patient was born at full term with a normal spontaneous vaginal delivery, and the pregnancy was described as uncomplicated. He had met all developmental milestones.

On examination, the patient was well developed and active. On standing, the patient had a well-aligned trunk and a mild genu varum alignment with the knees locked in extension. Both patellae were shifted laterally, the left more so than the right. Examination demonstrated bilateral obligatory lateral patellar dislocation that occurred during knee flexion. Both knees had a maximum flexion of 150° and extension to 0° with active ROM. No varus or valgus laxity existed, and the tibial tubercles were not laterally displaced. Hip examination demonstrated normal ROM, and Thomas and Ely tests were negative bilaterally. The patient showed normal strength and had an intact neurovascular examination in the bilateral lower extremities. He was able to ambulate independently with appropriate step lengths and cadence; however, the stance phase was characterized by increased impact of the foot to the surface and locking of knees into extension.

Standing anteroposterior plain radiographs showed both patellar ossification centers inferolaterally positioned, consistent with patellar subluxation (Figure 2). However, both patellae were frankly dislocated on the skyline view. The trochlear grooves were dysplastic and shallow bilaterally, the right more so than the left. The growth plates were intact, and the joint spaces were normal.

The patient was treated with physical therapy focused on quadriceps strengthening for approximately 10 weeks. However, no improvement occurred in mechanical symptoms. The patient’s parents elected surgery to prevent further patellar dislocations and improve the likelihood for normal trochlear groove development.

Under tourniquet control, a lateral parapatellar approach was used in both knees. The rectus femoris tendon was lengthened in a V-Y fashion. A lateral release was performed and extended proximally to free the vastus lateralis, with a small tendon cuff from the rectus femoris. The vastus medialis was freed from the patellar retinaculum proximally and distally, leaving a cuff of retinaculum and vastus medialis. The pes anserine was inserted, and the semitendinosus tendon was freed proximally with a tendon stripper, leaving the distal end attached, as described by Galeazzi. The proximal tendon was then sutured to the inferomedial border of the patella with the vastus medialis cuff, holding the patella in a neutral position. The rectus femoris tendon cuff was sutured to the vastus medialis and rectus femoris with the knee placed in 90° of flexion.

![Figure 2: Preoperative anteroposterior (A) and Merchant (B) radiographs of a 6-year-old boy with femoral trochlear dysplasia and recurrent bilateral patellar dislocation. Postoperative anteroposterior (C) and merchant (D) radiographs 10 years after patellar realignment.](image-url)
The wound was irrigated and closed, and sterile dressings were applied. The patient lost 50 cc of blood, and no complications occurred. The patient was placed in bilateral Bledsoe braces locked in extension and was made weight bearing as tolerated on postoperative day 1. The patient tolerated the procedure and in-hospital rehabilitation well and was discharged on postoperative day 2.

At 7-week follow-up, both patellae were centrally located on knee extension. Knee ROM in both knees was approximately 45° with normal patellar tracking. At 18 weeks postoperatively, the patient had full, active bilateral knee ROM and centrally located patellae that were stable to general stress. At 10-year follow-up, the patient returned with recurrent symptoms of right patellar dislocation that restricted him from sports. The left knee remained asymptomatic. Anteroposterior and Merchant radiographs showed that both patellar components were centrally located. Lateral radiographs showed bilateral patella baja and right patellar deformity (Figure 3).

Patient 3

A 9-year-old girl, the daughter of patient 1, presented with bilateral knee instability and frequent falls. The patient reported no history of trauma and no pain or swelling associated with knee instability. Previously, the patient had presented to her primary care physician at age 6 with an acute right patellar dislocation. Her right patella had dislocated during flexion. However, the patient had normal left knee stability at that time. Her medical history included growth hormone deficiency treated with growth hormone replacement for the past 3 years. The patient was born at full term with a normal spontaneous vaginal delivery, and the pregnancy was described as uncomplicated. The patient had met all developmental milestones.

On examination, the patient was well developed with a short stature. Knee examination showed a full, bilateral active ROM of 140° of flexion and 0° of extension with obligatory bilateral patellar dislocation on extreme flexion. Both patellae were centrally located during knee extension. A bilateral positive patellar apprehension sign existed. Normal strength existed in both lower extremities, and neurovascular examination was intact. The patient had equal leg lengths and full ROM in both hips.

Standing anteroposterior plain radiographs showed normal patellar ossification centers and intact joint spaces (Figure 4). Merchant radiographs showed bilateral lateral patellar dislocation, and both patellae displayed flat articular surfaces. The trochlear grooves were markedly flattened and shallow bilaterally.

A lateral parapatellar approach was used bilaterally under tourniquet control. The rectus femoris tendon was lengthened in a V-Y fashion. A lateral release was performed and extended proximally to free the vastus lateralis. The pes anserine was inserted, and the semitendinosus tendon was freed proximally with a tendon stripper, leaving the distal end attached. Using a 4-mm drill, a bony tunnel was created by drilling medially to anterolaterally. The proximal tendon was secured to the patella. The vastus lateralis was medially imbricated on the quadriceps tendon. The wound was irrigated and closed, and sterile dressings were applied to the closed wound. The patient lost 75 cc of blood, and no complications occurred. The patient was placed in bilateral Bledsoe braces locked in extension and was made weight bearing as tolerated on postoperative day 1. The procedure and in-hospital rehabilitation were tolerated well, and the patient was discharged on postoperative day 4.

At 4-year follow-up, radiographs showed anterior centrally located patellae and asymptomatic patella baja. The patient had full bilateral active knee ROM with good patellar tracking. She remained asymptomatic and reported no restrictions in activities of daily living.

DISCUSSION

Patellofemoral joint stability is maintained by soft tissue structures, the quadriceps musculature, retinacular ligaments, and the bony geometry of the trochlear groove. Femoral trochlear dysplasia is characterized by a shallow to absent femoral trochlea, a hypoplastic lateral femoral condyle, and a hypoplastic patella.8,9 Femoral trochlear dysplasia can lead to patellar instability, starting at approximately 20° of knee flexion when the patella begins to engage the trochlear groove.10 Dejour et al10 reported that the diagnosis of femoral trochlear dysplasia is made by a true lateral knee radiograph showing the crossing sign, which indicates a shallow trochlear sulcus. The presence of femoral trochlear dysplasia contributes to patellar instability and subsequent recurrent patellar dislocation in severe cases. However, femoral trochlear dysplasia is rare, occurring in 0.7% to 2% of patients with knee pain.11,12

Although femoral trochlear dysplasia is uncommon, bilateral recurrent patellar dislocation in multiple members of the same family suggests a genetic origin. Congenital forms indicate patellar dislocation at birth and are typically fixed and irreducible.13 This should be differentiated from developmental forms that occur after birth and obligatory dislocations that refer to dislocation during knee flexion.
ion. Orthopedic surgeons who encounter patients presenting with chronic patellar instability with no underlying disease or syndrome should be aware of the presence of femoral trochlear dysplasia leading to recurrent bilateral patellar dislocation. Immediate screening of family members who are symptomatic may allow earlier intervention to prevent long-term complications of recurrent patellar dislocation.

This article describes 3 patients in the same family with recurrent bilateral patellar dislocation associated with femoral trochlear dysplasia. Although previous reports have described a familial pattern of recurrent patellar dislocation, a familial association of femoral trochlear dysplasia leading to recurrent bilateral patellar dislocation has been described in the literature once. Patellar instability is linked to underlying medical syndromes and includes patients with excessive joint laxity. However, femoral trochlear dysplasia is not common as a genetic trait in many of the syndromes leading to patellar instability. Limited information exists regarding a heritable basis for recurrent bilateral patellar dislocation, and further studies are needed to identify specific genetic traits leading to this condition.

Rouvillain et al described 4 cases of bilateral recurrent patellar dislocation associated with femoral trochlear dysplasia in the same family. The presence of femoral trochlear dysplasia leading to bilateral recurrent patellar dislocation has also been reported as a characteristic of Kubuki syndrome, a rare disorder associated with severe mental retardation. Borochowitz et al reported 10 family members in 5 generations with recurrent bilateral patellar dislocations. However, the affected individuals in the current article had no evidence of excessive joint laxity and no bony abnormalities (including trochlear dysplasia) around the knee on radiographs. An autosomal-dominant mode of inheritance with male-to-male transmission was proposed in this series. Similarly, previous reports have suggested an autosomal-dominant mode of inheritance in familial forms of recurrent patellar dislocation.

Each patient in the current article had obligatory patellar dislocations not associated with previous trauma. Although the full family genealogy was not available, it is suggested that an autosomal dominant inheritance existed. The middle child in this family, not described in the current article, had no evidence of patellar disease or subluxation, also suggesting a heritable basis for the development of recurrent patellar dislocation in this family. To our knowledge, no history existed of patellar dislocation in the father’s family, which suggests transmission by the mother to both children. A limitation of this study is that a full family pedigree was not available to better identify transmission. The mother in this report was from Laos, and limited family contact made it difficult to better appreciate the mode of inheritance.

Patient 1 was a 54-year-old woman with a history of bilateral patellar instability since age 14. Complications of recurrent patellar dislocation with femoral trochlear dysplasia led to severe patellofemoral arthritis and degenerative tibiofemoral joint arthritis. Despite a trial of conservative measures, definitive surgical management included bilateral TKA. The use of TKA for the management of patellofemoral dysplasia with congenital dislocation of the patella and degenerative arthritis has been successful. Surgical considerations of TKA in this population include use of the transepicondylar axis for obtaining proper external rotation of the femoral component instead of the anteroposterior axis, which may result in excessive external rotation due to trochlear dysplasia. In addition, a lateral parapatellar approach could be considered when coupled with the release of the lateral retinaculum to decrease the risk of patellar avascular necrosis.

The 2 children in this report were surgically treated with semitendinosus tenodesis using a modified Galeazzi technique and proximal realignment for recurrent
bilateral patellar dislocation associated with femoral trochlear dysplasia. Various reports have described the successful use of the Galeazzi technique for recurrent patellar dislocation. 19, 20 Baker et al 19 reported a series of 53 knees treated with the Galeazzi technique and described good to excellent results in up to 88% of children, with a recurrence rate <5%. The clinical outcomes of all 3 patients showed successful patellar reduction and improvement in symptoms postoperatively. Surgical management in younger patients with femoral trochlear dysplasia leading to chronic patellar instability is recommended to preserve more natural patellofemoral joint congruity and the formation of an adequate trochlea. 9 Children can remain largely asymptomatic, but due to the unfavorable natural history of recurrent patellar dislocation, early corrective surgical management may prevent subsequent complications as they reach adulthood. Although several surgical options are available for patellar stabilization, soft tissue reconstructions are preferred in this population. Surgical options should preclude the use of osteotomies around the knee to correct patellar alignment due to the presence of open physes in skeletally immature patients.

Initial assessment of patients with patellar instability should include a comprehensive medical history and physical examination, including the patient’s age, weight, medical history, history of instability, and family history focused on symptoms or risk factors for patellar instability. Physical examination of patients with patellar instability should focus on balance and gait disturbances and clinical signs of potential collagen disorders, such as joint laxity, skin hyperextensibility, blue sclera, and other skeletal abnormalities. No genetic testing is necessary unless indicated by physical examination.

**CONCLUSION**

Few reports of familial forms of femoral trochlear dysplasia leading to recurrent bilateral patellar dislocation have been reported in the literature. This article described the possible genetic association of femoral trochlear dysplasia leading to recurrent bilateral patellar dislocation, as evidenced by 3 cases in 1 family. Recognition of familial forms and early intervention may halt the progression of patellofemoral instability and prevent long-term complications in these patients.

**REFERENCES**