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Case report

Hereditary bilateral genu recurvatum: Case report of a family

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ABSTRACT

Background: Genu recurvatum is a rare condition in children and adolescents. The origin can be osseous, ligamentous and mixed.
Methods: We describe for the first time a family inclusive two brothers and their mother with hereditary bilateral genu recurvatum of unknown etiology. The possible underlying pathology and treatment are discussed.
Results: The underlying pathology of the early closure of the apophysis of the tibial tuberosity remained unclear. The mother was never treated, one of the brothers received a tibial osteotomy as a young adult. Both patients developed end-stage osteoarthritis of both knees which was successfully treated by a bilateral computer-navigated TKA.
At seven to eight years after implantation in the older brother (left and right knee, respectively) and three to four years after implantation in the younger brother (right and left knee, respectively), the Visual Analogue Scale (VAS) pain score on a 0 to 100 scale was 0 of both brothers, the EQ-5D health status was 80 (scale of 0 to 100), the satisfaction was 10 (scale of 10) of both brothers.
The Knee Injury and Osteoarthritis Outcome Score (KOOS) function of the younger brother of 75 (scale 0 to 100) and of the older brother 100. The KOOS sport of the younger brother was 10 (scale 0 to 100), whereas that of the older brother 85.
Conclusions: Hereditary bilateral genu recurvatum with end-stage osteoarthritis can be successfully treated with computer-navigated TKAs; however, impingement of the patella on the proximal tibia and the position of the tibial keel are of concern.

1. Introduction

Genu recurvatum is a rare condition in children and adolescents. The origin can be osseous, ligamentous, and mixed [1]. Traumatic premature closure is the most common reason, even though the incidence of an injury of the proximal tibial physis is very low [2]. Further, multiple causes of genu recurvatum have been reported, including: fibrocartilaginous dysplasia [3], prolonged immobilisation with excessive pressure on the tibial tubercle [4], operation [1,4], skeletal trauma [3,4,5], slipped capital femoral epiphysis [6], spontaneous closure of the physis [7], Blount's disease [8], Osgood–Schlatter disease [9–11], avulsion of the tibial tubercle [12], tibial tubercle transfer [13], skeletal traction [14,15], haemophilia [16], infection [1], spondyloepiphyseal dysplasia [17], and neuromuscular disorders [18].

Genu recurvatum is a debilitating deformity of the knee, with clinical symptoms including pain, instability, weakness, leg-length discrepancies, and loss of range of movement (ROM) [1]. The present case report describes two cases in one family with

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hereditary, bilateral genu recurvatum, which resulted in end-stage osteoarthritis. An outpatient consultation with the geneticist did not reveal any known syndrome. The chosen treatment for both patients was computer-navigated total knee arthroplasty of both knees.

2. Materials and methods

2.1. Presentation of the cases

2.1.1. Medical history

A male patient (younger brother) at the age of 56 years was presented to the Department of Orthopaedics, Maastricht University Medical Centre (MUMC+) with debilitating genu recurvatum of both knees. He had been treated conservatively for several years, and neither knee had been operated on. No reason (e.g. trauma, infection or sports activities) was found in his medical history that could explain the bilateral genu recurvatum. He had an older brother who had the same deformity of both knees, and who had received a closing-wedge correction osteotomy with staple fixation at the age of 19 years. He had also received a computer-navigated total knee arthroplasty of both knees at the age of 54 years. Both brothers also had destructive osteoarthritis of both wrists, which had resulted in scapholunate advanced collapse (SLAC wrist) (Figure 1). The older brother had known spondylarthritis of multiple segments. By thorough physical examination, no other musculoskeletal disorders were found. An outpatient consultation with the geneticist did not reveal any known syndrome.

Their mother had the same knee deformity, but she was never treated. Her sons confirmed the description of the mother's malformation. However, because the mother had passed away years before the youngest brother was treated, her medical history records could not confirm the diagnosis: only examination of family pictures confirmed the malformation. Neither their two other brothers nor the children of the affected brothers had a bilateral genu recurvatum nor did they have osteoarthritis (OA) of other joints (Figure 2).

2.1.2. Physical examination

Physical examination of the younger brother showed recurvatum of both knees with valgus mechanical leg axis, which is a known combination with recurvatum deformity [19]. There was no leg-length discrepancy. The mechanical long leg axis on the left side was seven degrees valgus, with 20° recurvatum; knee flexion was 120°. The mechanical long leg axis on the right side was six degrees valgus, with 25° recurvatum; knee flexion was 120°. There was some effusion of the knee joints without weakness of the quadriceps muscle (Grade 5/5) [20]. The laxity of the collateral ligaments of both knees was five to 10 mm (Grade 2) [21,22], and the anterior cruciate ligament (ACL), posterior cruciate ligament (PCL) [23], and posterolateral corner (PLC) were stable. Both knees had a patella baja, despite a very long patellar tendon; the patellar tracking was normal and there was no positive J sign of the patella.

Clinically, there was a clear malformation of the tibial tuberosity, which was more clearly seen on radiograph (Figures 3 and 4). Both hips had a symmetrical ROM without symptoms. Standard plain radiographs showed an anterior slope of the tibial plateaus (16° on the left side; 19° on the right side). Despite the very long patellar tendon, the patella was positioned too distally, but central (patella alta). There were no crossing signs, indicating a normal depth of the trochlear groove. The degree of osteoarthritis was

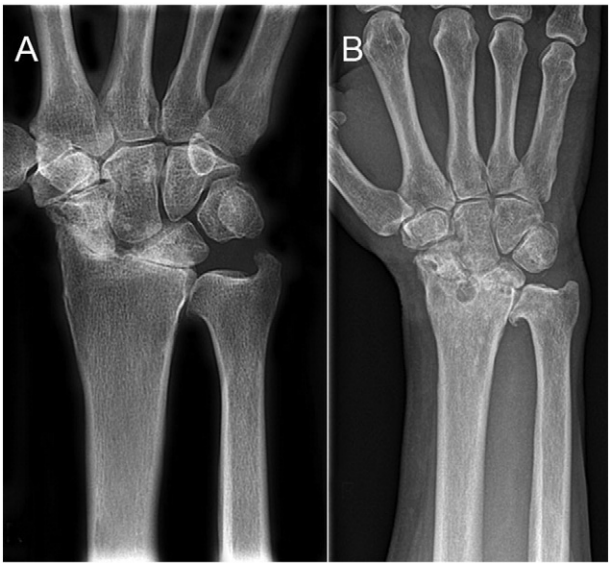

Figure 1. Standard antero-posterior (AP) radiographs of the right wrist of the younger brother (A) and the right wrist of the older brother (B). Note the diastasis between the scaphoid and lunate, the migration of the capitate into the space created by the scapholunate dissociation, and the sclerosis and joint space narrowing between the lunate and capitate in the radio-carpal joint. Similar radiological findings can be found on the left wrists of both brothers.

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