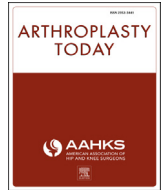




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Arthroplasty in patients with rare conditions

## Total knee arthroplasty in patients with multiple hereditary exostoses

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## ABSTRACT

We present a case report of a patient with severe valgus deformity of the right knee due to multiple hereditary exostoses (MHEs) treated with total knee arthroplasty (TKA). The surgical management of MHE affecting the knee encompasses exostoses resection, joint deformity rectification, and limb-length discrepancy alignment. On rare occasions, distraction osteogenesis and TKA have been used to correct valgus deformities of the knee. TKA in MHE patients with knee involvement has only been described in 6 cases. Several considerations, such as extensive knowledge of frequently occurring skeletal aberrations, are required to successfully correct the deformities associated with MHE via TKA. This report describes a case of severe valgus knee deformity with a rotational component in MHE managed with TKA, the surgical technique, and future recommendations.

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## Introduction

Multiple hereditary exostosis (MHE) is an autosomal dominant condition caused by a point mutation in the family of exostosin genes [1]. Mutations in the exostosin-1 gene are associated with more severe clinical manifestations along with an increased risk of malignant degeneration [2]. The overall prevalence of MHE is currently unknown due to many unreported asymptomatic cases. However, the approximated prevalence in Caucasian populations (the most widely studied population) spans from 0.9 to 2 individuals per 100,000 [3]. MHE is characterized by multiple pedunculated or sessile cartilage-capped bony outgrowths that may lead to joint deformities, restricted range of motion (ROM), and early-onset osteoarthritis [4]. Other associated findings are limb-length discrepancies, pectoral/pelvic asymmetry, and short stature, with limb-length discrepancies

(10%–50%) and short stature (37%–44%) being the most prevalent [5,6]. The clinical manifestations of MHE most commonly affect the long bones but may also affect the iliac crests, ribs, and scapulae [6]. The most common location of exostoses in MHE is the knee with prevalence of distal femur and proximal tibia involvement ranging from 70% to 98%. The fibula is less commonly involved with prevalence ranging from 30% to 97% [6]. Complications associated with MHE include: aneurysms/pseudoaneurysms, arterial/venous thrombosis, neurovascular compromise, and malignant degeneration [7].

It is a known fact that the knee is the most frequently affected joint, yet little is known about the surgical management of MHE with severe valgus deformities of the knee. Surgical management of MHE affecting the knee encompasses exostoses resection, joint deformity rectification, and limb-length discrepancy alignment [8]. On rare occasions, distraction osteogenesis and total knee arthroplasty (TKA) have been used to correct valgus deformities of the knee [1,8,9]. To our knowledge, only 6 reported cases of valgus deformity due to MHE have been managed via TKA [8,9]. We present the first case of MHE with severe valgus deformity, in excess of 45°, corrected by TKA using metaphyseal sleeves and describe our surgical technique.

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## Case history

### Preoperative evaluation

A 67-year-old Hispanic man, former smoker and construction worker presented to the clinic due to progressive bilateral knee pain and severe valgus angulation of the right knee. The patient stated that he began to notice aberrant growths on his right knee at age 12. However, the patient failed to seek medical advice due to lack of symptoms. Approximately 8 years before presentation, the patient began using a cane due to progressive bilateral knee pain and difficulty in walking. Family history revealed a daughter, brother, and 2 nephews who suffer from aberrant growths on the knees.

Upon physical examination, inspection of the right lower extremity revealed external rotation and severe valgus deformity of the knee, valgus angulation of the ankle, and no protruding or visible masses (Fig. 1). Palpation of the right knee elicited pain in the lateral compartment. Patellar evaluation failed to reveal hypermobility, hypomobility, or tilting. Evaluation of the peripheral vasculature resulted in findings within normal limits. Right knee ROM was 0°–120°. On valgus/varus stress test, the valgus deformity of the right knee was fixed and laxity of the medial collateral ligament (MCL) was identified. Evaluation of bilateral lower extremity motor function using the Oxford muscle grading scale was 3/5 for dorsiflexion of the right ankle. All other lower extremity movements were 5/5. Preoperative Knee Society Knee Score was 14 and Function Score was 55, for a total Knee Society Score of 69. Preoperative Western Ontario McMaster Score was 61.4.

Radiographic imaging using anteroposterior and lateral views showed 2 radiopaque sessile growths affecting the proximal medial and lateral portions of the right tibia and proximal fibula (Figs. 2 and 3). Using the anteroposterior radiograph, the valgus angulation of the right knee was determined by using the anatomical axis of the femur and tibia. Given the patient's clinical presentation, family history, and radiological findings, a clinical diagnosis of MHE was made and TKA was recommended. The patient was informed of the alternative treatment options, along with the risks and benefits



**Figure 1.** Preoperative standing clinical image.



**Figure 2.** Preoperative standing radiograph with anteroposterior view of the right knee exhibiting the severe valgus alignment and rotational component of the deformity.

associated with each alternative, and the patient consented to proceed with the TKA.

### Surgical technique

The surgery was performed by the senior author with the use of a tourniquet. Using standard instrumentation, the angle of bone



**Figure 3.** Preoperative standing radiograph with lateral view of the right knee elucidating the rotational component of the deformity.

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