A Case of Multiple Hereditary Osteochondromatosis

Multiple hereditary osteochondromatosis (also known as multiple hereditary exostosis) is an uncommon autosomal dominant condition characterized by multiple osteochondromas (bony excretion covered by cartilage) (1-3). We report an 11-year-old male with history of multiple hereditary osteochondromatosis who presented to our institution for follow up of known osteochondromas. He had mild pain in the right knee. On physical examination he had normal range of motion, normal pulses, and no neurological deficits. These findings have remained stable over time. Anteroposterior and lateral radiographs of the right knee demonstrated multiple osteochondromas throughout the distal femoral, proximal tibial, and fibular metaphysis without evidence of destruction of the underlying osseous cortex or widening of the distal femoral metaphysis (Figures 1 and 2); anteroposterior and lateral views of right knee respectively. Multiple hereditary osteochondromatosis is usually apparent during childhood. It is more common in whites, and affects equally males and females. Knees are usually involved. This condition presents as painless bony lumps in the tibia and scapula. However, pain may occur as the result of the mass effect of osteochondromas in the adjacent soft tissues and nervous structures. The imaging modality of choice for the diagnosis is plain radiography given that the radiographic appearance of osteochondromas is characteristic. Other imaging modalities such as computed tomography, magnetic resonance imaging (MRI), ultrasound, and nuclear medicine have been used for further evaluation of these lesions and related complications. MRI is particularly useful for the evaluation of the hyaline cartilage cap, which is the site of origin of the malignant transformation (reported rate up to 20%). Possible complications related to osteochondromas include bone deformities, fractures, vascular and neurologic injuries, bursa formation, and malignant degeneration. Symptomatic or deforming osteochondromas may be amenable for surgical correction, depending on the severity of the case.

References