

# Hereditary multiple exostoses: a case report

John R Pikula, BSc, DC, DACBR, FCCR(C), MSc\*

*Hereditary multiple exostoses (HME) is a dominantly inherited skeletal disorder which alters enchondral bone during growth and is characterized by exostoses of the juxta-epiphyseal regions. These exostoses are benign cartilaginous neoplasms that consist of a pedicle of normal bone covered with proliferating cartilage cells. Pathologic, clinical, and radiographic findings are discussed and a case of a nine-year-old male is reviewed.*

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**KEY WORDS:** hereditary multiple exostoses, painful joints, bony masses, chiropractic.

*La maladie exostosante héréditaire (HME) est un trouble squelettique dominant héréditaire, qui modifie l'os cartilagineux lors de sa croissance et se caractérise par des exostoses des régions juxta-épiphyseales. Ces exostoses sont des néoplasmes cartilagineux bénins constitués du pédicule d'un os normal recouvert de cellules cartilagineuses prolifératives. Nous discutons des résultats pathologiques, cliniques et radiographiques et étudions le cas d'un jeune mâle de neuf ans.*

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**MOTS CLÉS:** maladie exostosante héréditaire, articulations douloureuses, masses osseuses, chiropractie.

## Introduction

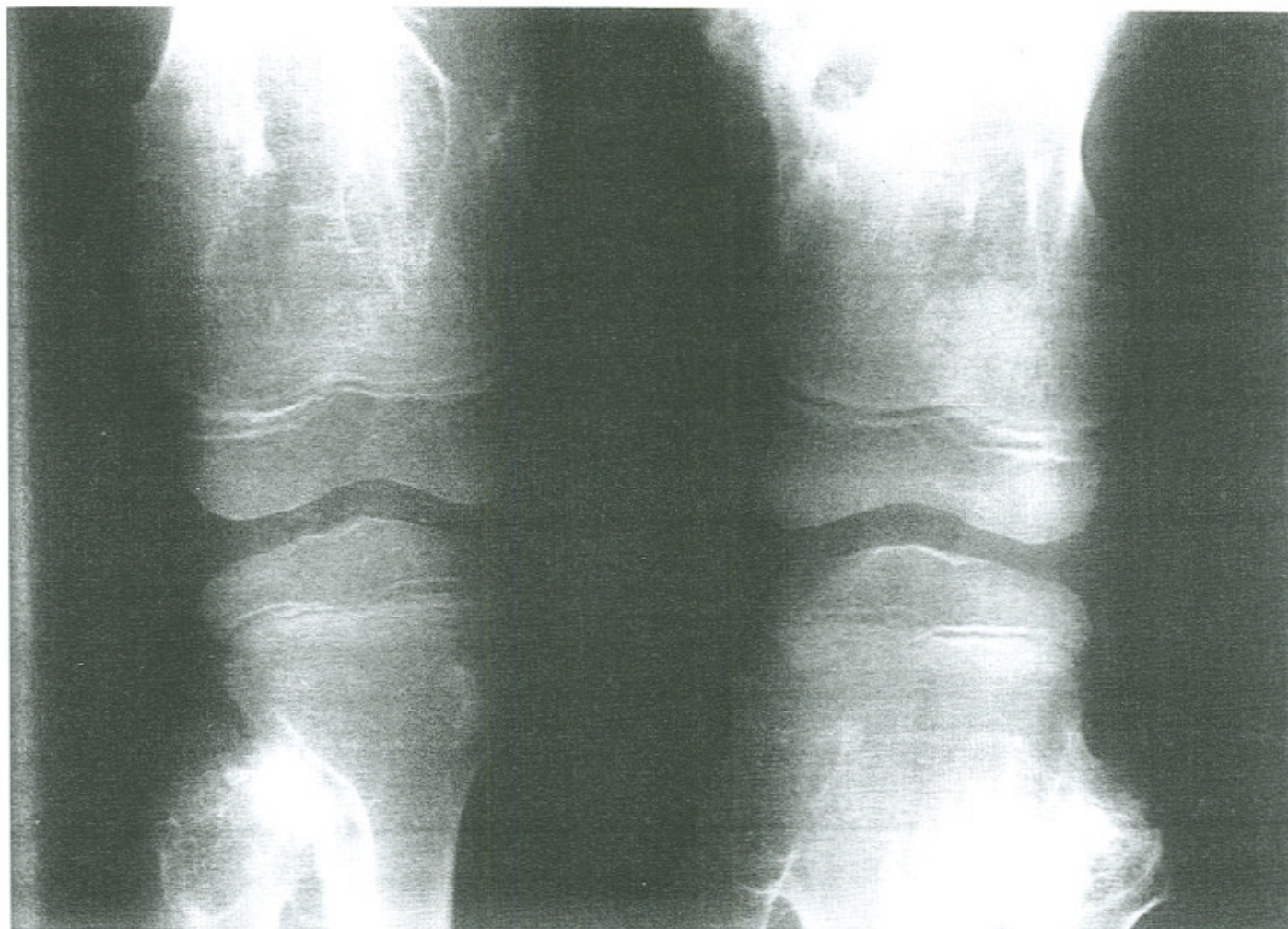
Extremity joint pain is a common complaint seen in chiropractic practice. Various etiologies can include a mechanical origin, trauma, infection, and pathology. Adults tend to delay seeking treatment for themselves hoping that the symptoms will subside with time. Concern for a potential serious disorder is why the parent brings the child to a clinician's attention. The following case demonstrates the clinical, pathologic and radiographic features of a rare hereditary disorder that is detected early in childhood. It is important to discuss this condition with the parents as to the potential future ramifications related to this disorder.

## Case report

A nine-year-old white male was brought by his mother to a chiropractic office with a chief complaint of joint pains. The mother pointed out numerous palpable osseous masses about his knees and ankles and one on his back. This young boy was very active and enjoyed playing soccer, however, when hit with the ball on these masses or falling on these masses, he experienced pain. Otherwise, these bony masses were painless.

A radiographic examination consisted of a skeletal survey of both knees, ankles, hands, pelvis, rib cage, and wrists. A single PA chest film demonstrated multiple bony abnormalities involving the first and third left ribs, both scapulae and the right humeral head. There were less obvious bony abnormalities involving the lower left ribs. A single AP view of the pelvis demonstrated multiple osteochondromas involving both femoral heads and acetabuli and the right iliac bone. Views of the knees,

\* Private practice, 189 Brant Avenue, Brantford, Ontario N3T 3J1. In partial fulfillment for requirements for the College of Chiropractic Sciences Field-practitioner program.  
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**Figure 1** Multiple exostoses (knees). Lack of tubulation of distal femur and proximal tibia. Note multiple osteochondromas directed away from the joint due to muscle pull.

ankles and wrists demonstrated many osteochondromatous lesions in these areas.

A diagnosis of hereditary multiple exostoses was made. No treatment was recommended at that time and the mother and son were counseled in regard to future potential complications and what action should be taken. It should be remembered that there is an increased incidence of malignant degeneration with HME.<sup>1</sup>

#### Discussion

Hereditary multiple exostosis (HME) is a hereditary disorder characterized by multiple osteochondromas. Other names for this disorder are external chondromatosis, mul-

tiple osteochondromatosis, and diaphyseal aclasis. These osteochondromas consist of multiple projections of bone (exostoses) or pedicle of normal bone covered with proliferating cartilage cells.<sup>2,3</sup> The lesions are most numerous in the metaphyses of long bones but may appear on diaphyses of long bones and on flat bones and vertebrae. These lesions may arise from any bone which was preformed in cartilage. The most common locations are the knees (Figure 1), ankles (Figure 2), shoulders, and wrists (Figure 3). They can be found in the pelvis as well and appear 'cauliflower-like' (Figure 4).

The transmission is autosomal dominant.<sup>2</sup> Any affected person can transmit the disease. An unaffected male in a

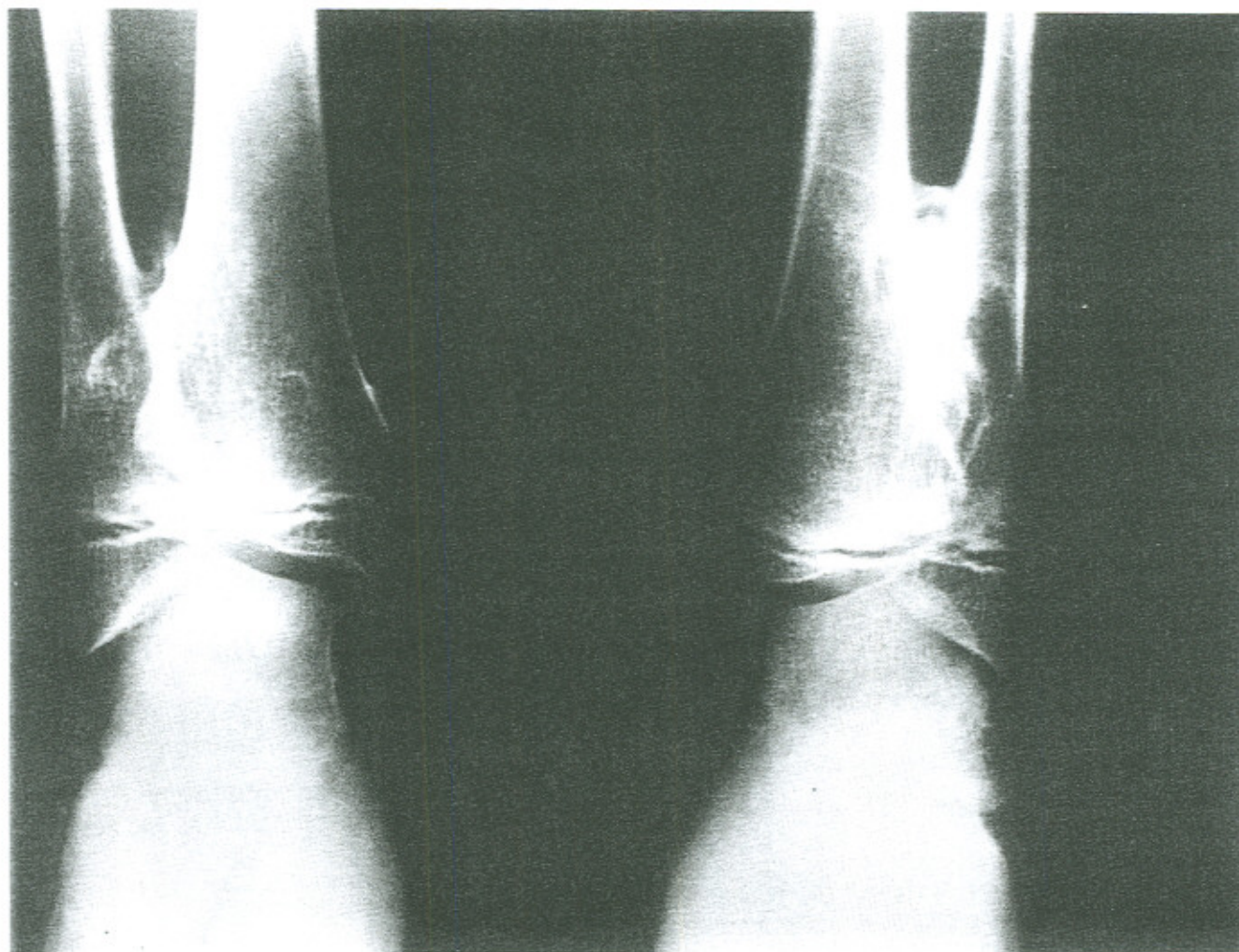


Figure 2 Osteochondroma (distal tibia), causing significant pressure erosion of distal fibula.

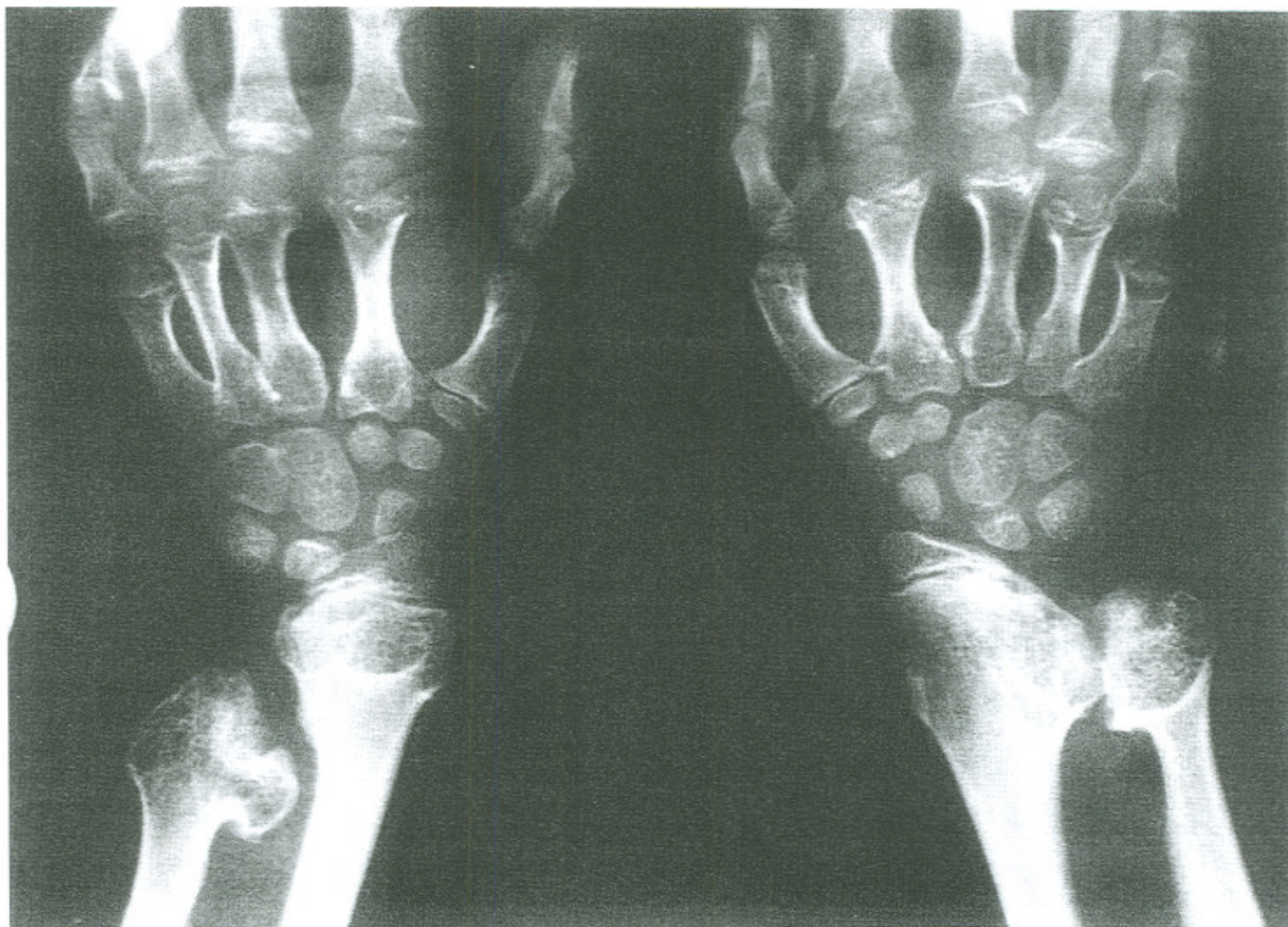
family with HME does not transmit the disease but an unaffected female may do so. Close to 50% of the children in families in which one parent has HME will manifest the condition. Both sexes are affected almost equally with males being more severe.<sup>4,5,6</sup>

The chief complaint is the discovery of single or multiple hard, painless masses near joints. The number of osteochondromas varies from a few to hundreds, with an average of 10.<sup>7</sup> These lesions form and enlarge only during the growth period. They are most commonly discovered between the ages of 2 and 10 years. The distribution is usually bilateral and may be symmetrical. As the exostoses grow there may be symptoms associated with pressure on

adjacent structures such as tendons,<sup>8</sup> spinal cord,<sup>9,10,11,12</sup> vessels,<sup>2</sup> restriction of joint function,<sup>2,14</sup> or bursa formation.<sup>2</sup>

Deformities may occur. The forearm is involved in approximately 30% of patients giving the characteristic "bayonet hand" deformity as a result of retardation of growth.<sup>15</sup> This is characterized by shortening of the ulna which does not extend far enough to take part in the formation of the wrist, outward bowing of the radius, and a subluxation of the radioulnar joint outward and posteriorly.<sup>7,16,17,18</sup>

The hand when involved rarely will show hand deformity. The principle area of involvement appears to be around



**Figure 3** Multiple exostoses. Note shortening of the metacarpals and of the ulna and curvature of the radius bilaterally. The phalanges are shortened as well in this case.

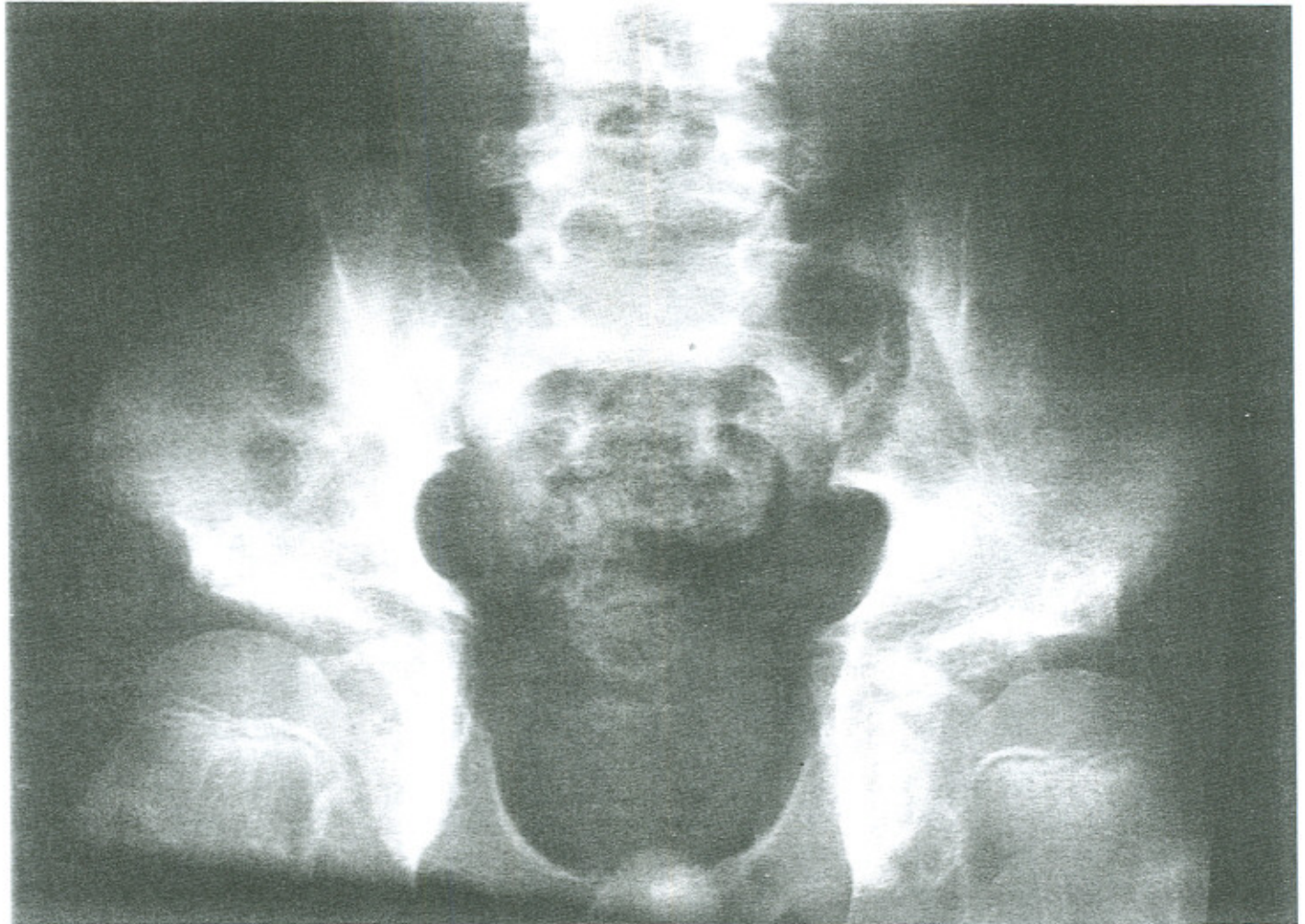
the MCP joint but the PIP joint is the most common area of deformity.<sup>19</sup> Brachydactyly can be seen in patients with HME when no exostoses are present, however, the presence of an exostosis results in even more shortening. The location and size of the exostosis has no relationship to increased bone shortening.<sup>20</sup>

The knees are most severely affected (Figure 1). Exostoses about the knee joint can cause shortening of the fibula with curving of the tibia. If one leg is more involved then shortening of the lower extremity can result in a compensatory spinal scoliosis.<sup>21</sup>

Large osteochondromas in the distal tibia or fibula may cause a pressure erosion or deformity of the adjacent bone

(Figure 2). The base of the skull may be involved but the calvarium and mandible are spared. The scapula usually has multiple protrusions at the medial border and inferior angle due to the presence of epiphyses at these areas. Subscapular osteochondromata can cause winged scapula as well.<sup>22</sup> Pelvic lesions may be noted at the iliac crest and at the ischiopubic synchondroses (Figure 4). The ribs are usually involved at the costochondral junctions.

Each exostosis consists of a cortex of compact bone with a medulla of cancellous bone. These are both continuous with that of the host bone. The exostosis is covered with a cap of hyaline cartilage. It should be noted that there are two types of exostoses. Pedunculated exostoses have a



**Figure 4** Osteochondromas at the ischiopubic synchondroses are noted. Also note the metaphyseal widening of the femoral necks near the growth plates. Both acetabuli demonstrate osteochondromas.

narrow base which have a rounded cap which may be expansile or 'cauliflower-like.' Sessile exostoses have a broad-base attachment which extends outward away from the nearest joint and can form a small, hook-like projection.<sup>4</sup> Malignant degeneration occurs in 5%–25% of cases into chondrosarcoma.<sup>1,23</sup> The most common sites are the pelvis and shoulder girdle.<sup>24</sup> Malignant degeneration should be considered when a known HME patient complains of pain or growth of an osteochondroma.

Ultrasonography enables accurate assessment of the cartilage cap of exostoses. The detection rate and measurement accuracy of ultrasound is higher than with computed tomography and comparable to magnetic resonance

imaging. Ultrasonography appears to be a good procedure for evaluating the cartilage cap, which is usually thin for a benign exostosis and thick for a malignancy.<sup>25</sup>

Treatment of HME is surgical, usually for cosmetic rather than symptomatic reasons. Recurrence of the tumor following adequate excision is highly suggestive of an aggressive lesion. Another reason for surgery is for the removal of the osteochondroma in the event of sarcomatous degeneration.<sup>7</sup>

#### **Summary**

Hereditary multiple exostosis is an inherited disorder characterized by multiple osteochondromas. Clinical manifes-

tations are usually absent before age two but discovered before age 10. The chief complaint is the discovery of single or multiple hard painless masses near joints. The distribution is usually bilateral and may be symmetrical leading to dwarfing and deformities. Malignant degeneration into chondrosarcoma occurs in 5–25%. It is therefore important to monitor all cases of HME especially if the patient complains of pain or growth of an osteochondroma.

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