Rare Presentation of Hereditary Multiple Exostoses
A Case Report

Hereditary multiple exostoses is a relatively uncommon disorder of endochondral bone characterized by the presence of multiple, cartilaginous-capped exostoses arising from the metaphyses. A rare presentation of hereditary multiple exostoses in the calcaneus of a 35-year-old man is reviewed and discussed. A brief review of the literature is provided, as well as a discussion of the patient’s family history. (J Am Podiatr Med Assoc 88(3): 135-139, 1998)

OSTEOCHONDROMAS ARE THE MOST COMMON BENIGN LESIONS FOUND IN BONE.1-3 Also referred to as exostoses, they are characterized by a bony stalk and a cartilaginous cap. They are generally found in endochondral bones.4 These lesions rarely develop from bones of the foot; they are more likely to occur in long tubular bones such as the tibia and the femur.5 If an individual has several osteochondromas, he or she may be diagnosed as having hereditary multiple exostoses.3, 6 This condition is defined as a disorder of endochondral bone characterized by the presence of multiple, cartilaginous-capped exostoses arising from the metaphyses. The disorder is also known as osteochondromata, hereditary multiple osteochondromas, hereditary cartilaginous exostoses, hereditary deforming dyschondroplasia, and diaphyseal aclasia.3,6,7

Hereditary multiple exostoses is an autosomal dominant trait that has a penetrance of 93% to 100%,5, 8 making it highly probable that it will be inherited.

The less-than-complete penetrance is due to the variability in expression of the disorder in females. This entity is thus observed more frequently in males than in females, with males accounting for 59% of cases and females for 41%. This disparity in expression remains unexplained. Several studies have found that 10% of patients have no family history of the disorder. It was concluded that the apparent "skipping of generations" was accounted for by asymptomatic lesions that are easily overlooked.5 Hereditary multiple exostoses is relatively uncommon. In Europe, it is reported to have an incidence of 0.9 to 1.4 in 100,000.9 A study of the population of Washington State found an incidence of 1 in 50,000.8

Case Report

A 35-year-old man presented to his podiatric physician with a chief concern of pain in the left heel of 3 weeks’ duration; a noticeable lump had been present for several months. The heel pain was isolated to the area around the bump and was of a dull, aching nature. It was present only during weightbearing. The patient reported that the pain began when he jumped from a loading dock while working. He denied any previous treatment for this ailment.

The patient’s medical history was notable for a motor-vehicle accident occurring during adolescence that caused mild brain injury and required multiple

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reconstructive surgeries of the left lower extremity. The patient also had a surgical history of removal of three osteochondromas from the tibia, femur, and humerus as a child. He was subsequently given the diagnosis of hereditary multiple exostoses. He was an otherwise healthy man who had no known allergies and was taking no medications. He was employed as a deliveryman and lived with his mother. His family’s medical history was significant for coronary artery disease and hereditary multiple exostoses, both on the paternal side. His father had died of a myocardial infarction. His mother was healthy with no significant medical history. His mother claimed to have no family history of hereditary multiple exostoses and reported that her late husband had no previous family history of hereditary multiple exostoses. The patient had three living siblings, two sisters and a brother. One sister and the brother are afflicted with hereditary multiple exostoses. A second brother, who died following a motor-vehicle accident, had been diagnosed with hereditary multiple exostoses as well. Thus four of the five offspring in the family had hereditary multiple exostoses. The sister who was not afflicted had a son and a daughter with the disorder (Fig. 1).

Physical examination revealed that the patient was 5 feet 3 inches tall and weighed 140 pounds. His left lower limb was approximately 2 inches shorter than the right limb. He had shortened forearms bilaterally with marked ulnar bowing. Scars from previous surgical interventions were present on the left upper arm and proximal leg as well as on the right lower thigh and upper leg. Focal pedal examination revealed sensate and well-perfused feet bilaterally. The left foot had a markedly elongated second digit and a shortened, floating fourth digit (Fig. 2). He also had a moderate, asymptomatic hallux abductus deformity of the left foot. He had a hyperkeratotic lesion that was well circumscribed above a firm, non-fluctuant mass on the posterolateral side of the left foot (Fig. 3). Application of direct pressure to the protuberance elicited moderate pain.

The patient underwent radiographic examination, which revealed a homogenous bony lesion contained by an expanded cortex. The base of the lesion was contiguous with the cancellous bone of the calcaneus (Fig. 4). On the basis of the history and the clinical and radiographic appearance, the authors strongly suspected osteochondroma.

Treatment, including nonsurgical and surgical options, was discussed with the patient. Nonsurgical treatment would consist of accommodative orthoses or custom-molded shoes. Given the nature of his employment and his activity level, it was mutually agreed that the surgical option of excision of the lesion would be most beneficial.

![Figure 1](image1.png)

**Figure 1.** Nomogram of the patient’s family. The squares indicate male members, and the circles, female members. Those shapes representing individuals with a positive diagnosis of hereditary multiple exostoses are shaded gray. The patient is indicated by the square nearest to the arrow. The patient’s brother was afflicted with the disorder, but is now deceased, as indicated by the black box.

![Figure 2](image2.png)

**Figure 2.** Clinical photograph of the patient’s left foot. Note the growth disparity among the long bones of the foot. Clinically significant shortening of the fourth ray with a floating digit is evident.
The surgery was performed in an outpatient surgery center with monitored anesthesia care and a local ankle block to the left leg. The limb was prepared and draped in the usual aseptic manner with the inclusion of a sterile ankle tourniquet. A linear incision was made directly superficial to the crest of the lesion on the lateral aspect of the foot. The subcutaneous fat was sharply and bluntly dissected and retracted to reveal the lesion. The superficial portion of the lesion was glistening white and smooth, and the base was contiguous with the calcaneus (Fig. 5). It measured $4 \times 3 \times 3$ cm. The lesion was removed \textit{in toto} and sent for histopathologic analysis. A cortical deficit of the lateral plantar calcaneus was observed and curettage of the area was performed to ensure removal of the neoplastic tissue. Bone wax was used for hemostasis of the exposed cancellous bone. The wound was aseptically lavaged and closed in a layered fashion. The patient was immediately placed in a removable walking cast, with partial weightbearing for 4 weeks. His convalescence was unremarkable.

The gross pathology report revealed multiple hard, lobulated portions of calcified, cartilage-like tissue of the aforementioned dimensions. The surface of the lesion was semitranslucent. The microscopic evaluation showed fragments of bone partially covered by a thick layer of disorganized cartilage (Fig. 6). There was a zone of endochondral ossification and a deeper zone of sclerotic bone with osteoblastic activity. There was no evidence of cellular atypia. These features indicated benign osteochondromatous exostosis.

**Discussion**

This presentation of hereditary multiple exostoses is particularly rare in two respects: the age of the patient when the lesion became symptomatic, and the location of the lesion. Several authors have reported that approximately 80% of people with hereditary
multiple exostoses begin to manifest symmetrical exostoses by the age of 2 years. The patient described here first noticed his lesions at the age of 14. He noticed the calcaneal lesion at age 35. Osteochondromas are usually discovered in the upper tibial epiphysis and the scapula. Presentation of osteochondromas in the calcaneus, as in this patient, is exceedingly rare. Exostoses of the tarsus have a frequency of 12% to 25% in the foot. Most are in the calcaneus. Patients with hereditary multiple exostoses have an average of six lesions in the long bones of the upper and lower extremities. The most common locations for the neoplasm are the distal femur (70% to 90%), the proximal tibia (70% to 95%), the proximal fibula (30% to 90%), and the distal fibula and tibia (25% to 85%). Thus the knee and ankle joints are commonly afflicted.

The family history in this case is especially noteworthy. There had been no diagnosis of hereditary multiple exostoses in the family prior to that of the patient's father. Although this remains a mystery, it can be assumed that the apparent "skipping of generations" occurred in one of the paternal grandparents or that there was a lack of penetrance in the gene of the paternal grandmother, as seemingly evidenced by the absence of the disorder in one of the patient's sisters. Lack of access to the health-care system may also explain why previous generations went undiagnosed.

Moreover, the patient described here had several distinguishing characteristics that may have assisted in diagnosis. The disorder often results in differential bone growth, as seen here, that leads to short stature, skeletal disproportion, limb-length discrepancies, forearm bowing, and ankle valgus. These characteristics are observed in as many as 50% of patients.

A specific manifestation of differential bone growth is the presentation of brachymetatarsia and brachymetacarpia involving the third, fourth, or fifth segments. As shown in the radiograph in Figure 4, both the third and fourth metatarsals in this patient are short. Joint deformities such as ankle and genu valgum or large periarticular lesions restricting normal range of motion may culminate in premature osteoarthrosis.

Radiographically, the exostosis is a diffuse club-shaped thickening of the metaphysis. It may be a pedunculated mass with the apex facing away from the nearest growth plate. When the lesion is viewed on end, it has a lucent appearance. The cortex and the spongiosa of the exostosis are contiguous with the cortex and cancellous bone of the parent bone. On radiographic views, this characteristic results in an attenuated but intact cortex containing the expansive cancellous bone.

The prognosis for a patient with hereditary multiple exostoses is usually good. The lesions require excision only when they impinge on vital organs or cause unmitigated pain. Cellular dysplasia of osteochondroma to chondrosarcoma is rare, with a frequency of between 3% and 25%.

Direct trauma to longstanding lesions has been implicated as a possible precursor to dysplasia. Suspicion for chondrosarcoma should be high if the exostosis is poorly defined in outline on radiographs. Bone destruction is often present. Fortunately, metastasis occurs late in the course of the disease. The treatment is local excision. There was no history of dysplasia in this patient's family.

Conclusion

A rare case of hereditary multiple exostoses in the calcaneus of a 35-year-old man has been presented. Given the location and symptomatic nature of the lesion, the patient opted for surgical excision of the lesion. His awareness of a positive family history coupled with the knowledge of his own diagnosis of hereditary multiple exostoses since the age of 14 allowed the authors to make the presumptive diagnosis of osteochondroma of the calcaneus and provide appropriate treatment.

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References