Hereditary Multiple Exostoses: Report of a Family from Riyadh

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Hereditary multiple exostoses (diaphyseal aclasis) is a hereditary condition in which, during the period of bone growth, multiple cartilage-capped exostoses appear especially involving long bones near joints. A Saudi family from Riyadh is reported in which the males of three generations are affected, two of whom underwent surgery for disfiguring benign tumours. The lesions of hereditary multiple exostoses appear to carry a small risk of sarcomatous transformation. The exact figure for this occurrence has varied widely in the literature from 3% up to 25% of patients according to various reports. It seems prudent that affected family members should be counselled to seek medical attention promptly if one exostosis demonstrates rapid growth especially after the age when bone growth is usually completed.

Hereditary multiple exostoses (diaphyseal aclasis McKusick Catalog No 13370) is a hereditary condition of bone in which cartilage-capped exostoses appear during the period of bone growth and may continue to enlarge until the epiphyses fuse. The long bones, pelvis, scapula and ribs are frequently affected. The condition is inherited as an autosomal dominant trait. Individuals often present with bony subcutaneous nodules appearing during childhood and adolescence, most commonly located at the ends of long bones around knee, ankle, elbow and wrist joints or involving the scapula, ribs or iliac crest. Two Saudi males from an affected Riyadh family are presented.

Case 1
A 22-year-old man presented to the King Fahad Hospital orthopedic clinic with a history of multiple swellings of different portions of the skeleton present since youth. The nodules were painless and did not interfere with general activity. The patient requested their removal for cosmetic reasons. The family history included similar nodules affecting males including the father, brothers, grandfather and cousins. Females in the family were unaffected to the patient’s knowledge. However, examination of the female family members was not possible. Physical examination of the patient revealed a well developed, well nourished Saudi man without abnormal findings except for multiple bony deformities of the skeleton including a 7 cm hard subcutaneous mass arising from the proximal portion of the right humerus, a 1 cm mass arising from the distal portion of the radius, 1 cm mass of the left rib, a 3 cm mass of the distal right femur, a 2 cm mass of the proximal right tibia and a 2 cm mass of the proximal left tibia. At surgery, pedunculated bony nodules having a cartilaginous cap were removed from the right humerus and adjacent to the knee joints bilaterally.
Case 2
The 27-year-old brother of the above described patient presented in a similar fashion. The lesions which most concerned the patient were large masses located in both thighs, one on the anterior surface of the left thigh and the other on the posterior aspect of the right. The masses had been present for many years without history of sudden growth. Surgical removal was performed on 30 October 1988. Histologically, the lesions were confirmed to be benign.

Discussion
Hereditary multiple exostoses (HME) is characterized by the appearance of cartilage-capped exostoses during the period of bony growth of the skeleton. The long bones, pelvis, scapula and ribs are frequently affected with rare involvement of the vertebrae, carpal and tarsal bones. The disease is inherited as an autosomal dominant trait. However, several families have been reported to show a distortion of the male to female ratio with more males affected. A milder form of the disease in women and transmission by apparently unaffected women has been previously recorded.

Crandall et al.\(^2\) reported 85 members of one family comprising six generations dating back to 1832 in which 33 members were known to be affected by the condition. In 70% of the individuals there were associated deformities such as short stature, bowing of the forearm with ulnar deviation of the wrists and valgus deformity of the ankles and knees. As is characteristic, the exostoses typically pointed away from the joint. These authors classified 61% of the men and 31% of the women in that family as affected with the degree of involvement from mild to severe according to the scheme below:

1. Mild: 1–3 nodules without shortening or deformity
2. Moderate: multiple nodules and deformity of the forearm without limitation of movement
3. Severe: deformity limiting movement or disturbing gait

The incidence of the disease is not known, however, Solomon\(^3\) estimated that three new cases were diagnosed each year at an orthopaedic hospital at which 7000 new patients were seen annually. Stocks & Barrington\(^4\) collected 1124 case histories from the literature but other conditions may have been included most notably Ollier's disease. Ollier's disease may rarely have small exostoses but is characterized by enchondromatoses, a cartilagenous tumour growing within bone rather than on an exostosis at the surface.

The frequency of sarcomatous change in exostoses of HME has been reported to vary from 3% to
Figure 2. Classical appearance of hereditary multiple exostoses lesions arising adjacent to the knee. Note the manner in which bony lesions point away from the joint.

25% in different reports. Dahlin\(^3\) reviewed 272 cases and estimated the risk of malignancy at 10% while Solomon\(^3\) and Gordon \textit{et al.}\(^5\) thought the risk was less than 3%. No cases of chondrosarcoma were reported in the study by Crandall\(^2\) which involved 33 individuals over six generations. Nevertheless, it would appear reasonable to counsel affected family members to seek medical attention promptly in the event that an exostosis demonstrates rapid growth especially after the end of the usual bone-growth years. Chondrosarcomas when they occur in HME are typically diagnosed
in the fourth decade of life and most often occur proximally in the long bones or in the bony girdle.

Suggested therapy for exostosis in HME is surgical removal if the lesion causes limitation of motion or deformity or fractures or if vital structures are compressed. Cosmetic reasons are often the indication, however, as in the first case presented here. In the second case, the swellings interfered with motion of the extremities and were also cosmetically unacceptable.

This is the first report of hereditary multiple exostoses to the authors' knowledge from Saudi Arabia. Personal communications from medical colleagues in Riyadh indicates that several other cases have been diagnosed in Riyadh but not reported in the literature.

References