

Hereditary Multiple Exostosis - Diaphyseal Aclasis

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Case Report

Abstract: Hereditary Multiple Exostosis (HME) is an inherited autosomal dominant metaphyseal overgrowth that is characterized by multiple osteochondromas. In 75% cases it occurs before age of 20 yrs with male predominance of 2:1. Most cases are asymptomatic, unless they disturb surrounding blood vessels or nerves. No treatment is necessary in most cases because lesions are clinically silent. Symptomatic, large lesions are treated by surgical excision.

Keywords: Diphyseal Aclasis, Bayonet Hand Deformity.

Introduction

Hereditary Multiple Exostosis is an inherited autosomal dominant metaphyseal overgrowth that is characterized by multiple osteochondromas. It is also been called Multiple Osteochondromatosis, External Chondromatosis and Diaphyseal Aclasis – *aclasis* referring to an alteration of modeling process.^{2,3}

Case History

15 years old female patient presented with history of multiple painless swellings around the wrist, knee and ankle joints bilaterally noted since 4-5 years. The patient was subjected for X-ray Wrist joint AP and Lateral views, Knee joint AP and Lateral views and Ankle joint AP view.

X-rays revealed

Knee joints AP and Lateral View-> Multiple cartilaginous exostoses seen arising from metaphyseal regions of distal end of femur, proximal ends of tibia and fibula, (Fig. 1, 2), growing away from joint cavity having 'coat-hanger' appearance.

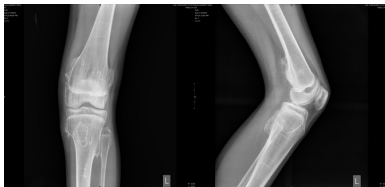


Figure 1: Left knee joint AP and lateral view: Multiple cartilaginous exostoses at distal femur and proximal tibia



Figure 2: Right knee joint AP and lateral view: Coat hanger appearance

Wrist Joints AP and Lateral views-> A large bony sessile exostoses arising from distal end of right radius growing away from joint associated with metaphyseal widening and minimal outward bowing of radial diaphysis. (Fig. 3)



Figure 3: Wrist joints AP and lateral views: A large bony sessile exostoses; at distal end of right radius

Ankle joint AP view -> A large bony sessile exostoses arising from lateral aspect of both tibia with associated metaphyseal widening. (Fig. 4)



Figure 4: Ankle joints AP view: A large sessile bony exostoses; lateral aspect of both tibia

Findings suggestive of Hereditary Multiple Exostosis (HME)

Discussion

Hereditary Multiple Exostosis is characterized by metaphyseal overgrowth and multiple osteochondromas. The number of osteochondromas varies from few to hundred, with an average of 10.

This tumor is classified with chondroid tumors as it is the cartilage cap that is actively growing.

The lesion is metaphyseal in origin but migrate to the diaphysis as normal metaphyseal growth occurs. The metaphyseal portions of long bones of the knee, ankle, shoulder, and wrist are most commonly affected; however flat bone involvement has been reported.^{4,5} The distribution is usually bilateral and may be symmetric. The elbows are usually spared. The hands are often involved in advanced cases. It is most frequently

discovered between the ages of 2 and 10 years. Both sexes are equally affected; however, the condition appears to be somewhat less severe in female patients.⁶ The chief clinical complaint is painless, lumpy joints. A characteristic bayonet hand deformity occurs about the wrist joint as a result of retardation of bone growth; the deformity is characterized by shortening of ulna, outward bowing of radius, and a subluxation of the radioulnar joint. This deformity occurs in about 30% cases.³ Malignant degeneration occurs in about 5-25 % of patients.⁷ Most lesions develop into chondrosarcoma. The pelvis and shoulder girdle are the most common sites.⁸ Definitive diagnosis is done radiologically.

Imaging Features

The lesion is metaphyseal in origin but migrate to the diaphysis as normal metaphyseal growth occurs. This progression is called 'coat-hanger' variety, pointing away from the adjacent joint. The metaphyses in this condition are also typically widened and dysplastic. The lesion is outgrowth of bone from the normal cortex, with which it is continuous. As the lesion grows, the marrow cavity extends into the exostosis. Actively growing cartilage cap can be shown by USG, CT or MRI. Cap becomes increasingly calcified with age in punctuate or nodular fashion.

Associated Features

HME tends to relate to mechanical problems, an enlarging mass, and pressure on adjoining structures or, occasionally, fracture of bony stem of the lesion. Exostoses may be larger than the solitary variety and may lead to shortening or deformity of the affected limb.

A characteristic *bayonet hand deformity* occurs about the wrist joint as a result of retardation of bone growth; the deformity is characterized by shortening of ulna, outward bowing of radius, and a subluxation of the radioulnar joint.

References

1. Kivioja A, Ervasti H, Kinnunen J, et al.: Chondrosarcoma in a family with multiple hereditary exostoses. J Bone Joint Surg 82B(2): 261,2000
2. Keith A: Studies on the anatomical changes which accompany certain growth disorders of the human body. The nature of the structural alterations in the disorders known as multiple exostoses. J Anat 54:101, 1920.
3. Ellis UH, Taylor JG: Diaphyseal aclasis. J Bone Joint Surg 33B: 100, 1951.
4. Felix NA, Mazur JM, Loveless EA: Acetabular dysplasia associated with hereditary multiple exostoses. A case report. J Bone Joint Surg 82B:555, 2000.
5. Paik NJ, Han TR, Lim SJ: Multiple peripheral nerve compressions related to malignantly transformed hereditary multiple exostoses. Muscle Nerve 23(8):1290, 2000.
6. Harsha WN: The natural history of osteochondral exostoses (osteochondroma). Am surg 20:65, 1954.
7. Flat AE: Chondrosarcoma supervening on diaphyseal aclasis. Br J Surg 43:85, 1955.
8. Garrison RC, Dahlin DC, Unni KK, et al.: Chondrosarcoma arising in osteochondroma. Cancer 49:1890, 1982.