FEMUR WITH MULTIPLE EXOSTOSES
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ABSTRACT: This study describes a left femur bone which was found with multiple exostoses. Both the upper and lower ends of the bone had multiple exostoses. Multiple hereditary exostoses (MHE) is one of the most common benign bone tumors. It commonly involves long bones, affecting their normal growth. Generally doesn’t need any treatment. However in some cases it may compress upon nerves or blood vessels demanding treatment. It may also produce bone deformities, shortened stature, limb length discrepancies, decreased range of movements, impaired function and premature osteoarthritis. Rare cases of malignant transformation have been reported. The knowledge of exostoses is important for a clinician as it may resemble other medical conditions with similar symptoms.

KEYWORDS: Osteochondromatosis, Multiple cartilaginous exostoses, Diaphyseal Aclasis, Premature osteoarthritis, Medullary compression, Chondrosarcoma, Growth disturbances.

INTRODUCTION: Multiple hereditary exostoses (MHE) is an autosomal dominant developmental disorder. It is characterized by the presence of multiple osseous prominences with cartilage caps arising most commonly from the metaphysis of long bones. In simple words, it is the formation of a new bone on the surface of a bone. It affects men three times more common than women and most commonly involves distal femur, proximal tibia & proximal humerus, rarely may develop in a joint. It is also referred to by additional names such as, osteogenic disease, chondral osteogenic dysplasia, chondral osteoma, dyschondroplasia, deforming chondrodysplasia, multiple hereditary osteochondromata, multiple cartilaginous exostoses or simply exostotic dysplasia¹.

It is estimated to occur in 1 in 50,000 people. If one parent has MHE, there is a 50% chance that any children they have will also develop the disorder. Spontaneous mutation is responsible for approximately 10% of the cases of MHE.

MHE may produces bone deformities, shortened stature, limb length discrepancies, decreased range of movement, impaired function and premature osteoarthritis. It can stretch or compress nerves, causing motor and sensory problems and pain. The knowledge of exostoses is very important for early recognition, treatment and prevention of complications.

MATERIALS & METHOD: During the regular osteology demonstration class conducted for the first year MBBS students of JSS Medical College, Mysore, a left femur bone (fig 1) was observed with multiple exostoses. The femur bone had exostotic growth at both the upper and lower ends.

OBSERVATION & DISCUSSION: Many growths were nodular in relation with the greater trochanter, lesser trochanter (fig. 2 & 3), neck of the femur (fig. 3) and supracondylar part of the shaft of the femur. Sharp spicule-like exostoses (fig. 4 & 5) were observed more in the lower end.

In multiple hereditary exostoses, bone growths are usually observed in relation to the metaphysis of the bone and characterized by the growth of cartilage capped benign bone tumors.
around areas of active bone growth, particularly the metaphysis of the long bone. But in the present specimen, bone growths were observed in epiphysis in addition to metaphysis. The number of exostotic nodules were more in the neck region. Many nodules were observed in the lower end also.

Osteochondromas are the most common benign bone tumors, developing in bones of endochondral origin. About 85% are solitary, rest are seen as a part of Multiple hereditary exostoses. Solitary ones are diagnosed in late adolescence and early adulthood, but MHE become apparent during childhood\(^2\).

MHE lesions usually continue to grow until closure of the growth plates at the end of puberty. No new lesions develop thereafter, although regrowth of an exostoses after surgical removal is possible\(^1\).

The exostoses are either pedunculated or sessile. Pedunculated tumors have a pedicle which arises from the cortex; commonly directed away from the growth of the plate of the bone. Sessile tumor presents a more broad based attachment to the cortex of the bone. In the present specimen all nodules were sessile and no pedunculated nodule was observed\(^3\).

Imaging of osteochondromas can provide information about the size and location of the lesions, for determination of malignant potential as well as for preoperative planning. Magnetic resonance imaging, with its superior soft tissue contrast, can identify the morphology of the osteochondromas and potentially associated soft tissue masses, suggesting malignant transformation\(^4\).

Subungual exostoses may develop on a distal phalanx, especially of the great toe. Often there will be history of trauma. Excision is indicated when elevation of the nail produces pain.

Surgery (en bloc resection) is indicated when the lesion is large enough to be unsightly or produce symptoms from pressure on surrounding structures, or when imaging fractures suggest malignancy. Biopsy may be indicated for diagnosis of sessile lesions. MHE may require osteotomies to correct deformity. Recurrence is rare and probably is due to failure to remove the entire cartilaginous cap\(^5\).

**TRANSMISSION OF MHE:** MHE is an autosomal dominant developmental disorder, with 96% penetrance. It is linked with mutations in 3 genes:

1. EXT1- chromosome 8q24.1
2. EXT2- chromosome 11p13
3. EXT3- chromosome 19p

Mutations in these genes typically lead to the synthesis of a truncated EXT protein which does not function normally. EXT proteins are important enzymes in the synthesis of heparan sulfate. Normal chondrocyte proliferation and differentiation may be affected, leading to abnormal bone growth\(^6\).

**CLINICAL SIGNIFICANCE:** Multiple hereditary exostoses may produce bone deformities, shortened stature, limb length discrepancies, decreased range of motion, fatigue, pain, muscle irritability and premature osteoarthritis.

There is a lot of variations in clinical presentation as well as, the age of presentation. Most of them are diagnosed in the second decade of life\(^7\). The abnormalities caused by MHE range from functional to aesthetic problems.
The common symptoms include pain, muscle irritation, decreased range of movements at joints, fractures & numbness due to compression on nerves.

CONCLUSION: Being aware of this kind of rare condition will help the practitioner to suspect the case in early stage and manage accordingly.

REFERENCES:

Fig. 1: Left femur with exostoses

Fig. 2: Upper end femur (anterior view)
Fig. 3: Upper end femur (posterior view)

Fig. 4: Lower end femur (posterior view)

Fig. 5: Lower end femur (anterior view) showing exostotic growth with spicule

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