

A Case of Hereditary Multiple Exostoses: Role of FNAC in diagnosis

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ABSTRACT

Hereditary Multiple Exostoses (HME) is a rare autosomal dominant bone disease. It is characterized with numerous benign osteochondromas, which grow outward from the metaphyses of long bones. In many cases it is asymptomatic but can lead to considerable number of complications, like pressure symptoms, limb deformities, and can cause psychosocial problems. Malignant transformation is rarely seen. Surgery is the main modality of treatment. This paper aims to highlight the role of FNAC in diagnosis of HME.

KEYWORDS: Hereditary Multiple Exostoses, FNAC, Complications

INTRODUCTION

Hereditary Multiple Exostosis (HME) syndrome, also known as Hereditary multiple osteochondroma (HMO), familial osteochondromatosis, diaphyseal aclasia, hereditary deforming dyschondroplasia, and multiple cartilaginous exostoses, is an autosomal dominant disorder which is characterized by the development of multiple osteochondromas (exostosis) and is frequently associated with progressive skeletal deformities. This disorder was first reported by Boyer in 1814. [1] Though the condition is usually asymptomatic, osteochondromas can cause compression of nerves, blood vessels and tendons due to their location, size, number and causing consequent pain and impairment of movements. We report a case of multiple hard swellings on limbs in a 14-year old male who was diagnosed as a case of HME with help of FNAC.

CASE REPORT

A 14-year-old male presented with a history of multiple swellings in his upper arm, knees, and ankles for the past 10 years (Figure 1). The patient first developed swelling in the right arm and soon noticed similar swellings in the

lower limbs. The swellings were painless and gradually increased in size. There was no history of trauma or any other constitutional symptoms. The patient did not have any pressure symptoms, a history of fractures, or vascular and neurological manifestations.

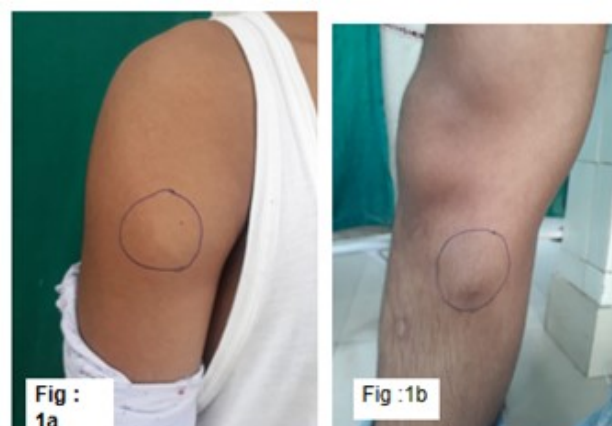


Figure 1: 1a and 1b: Bony swelling in the arm and leg

His father also had a similar history of bony outgrowths, which were asymptomatic. On examination, the swellings were nontender, measuring from 2 to 6 cm in size, and appeared to be continuous with the underlying bone structure. They were not attached to the overlying skin and were immobile with no signs of inflammation.

A radiographic examination was advised for a skeletal survey of both knees, ankles, shoulder, and chest. X-ray demonstrated multiple bony swellings involving the right humeral head, right tibia, and right ankle joint (Figure 2).

Fine-needle aspiration cytology (FNAC) of the swellings in the right upper arm and right knee was performed using a 22-gauge needle mounted on a 10ml syringe. Slides stained with Giemsa stain showed fragments of magenta-

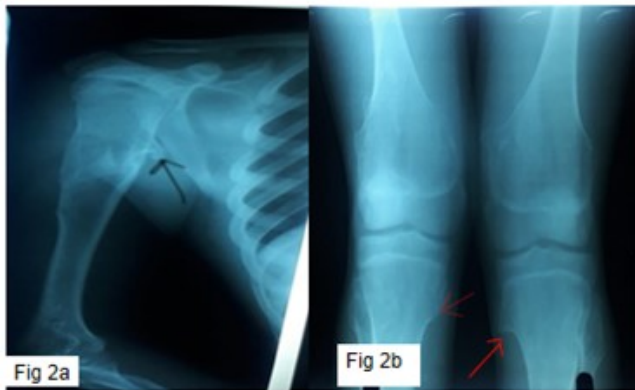


Figure 2: X-ray showing exostoses of : Fig 2a: Humerus and Fig 2b Tibia

colored matrix with enmeshed spindle cells having plump to elongated nuclei (Figure 3). Foci of calcification were also noted. FNAC findings were suggestive of a benign cartilaginous tumor.

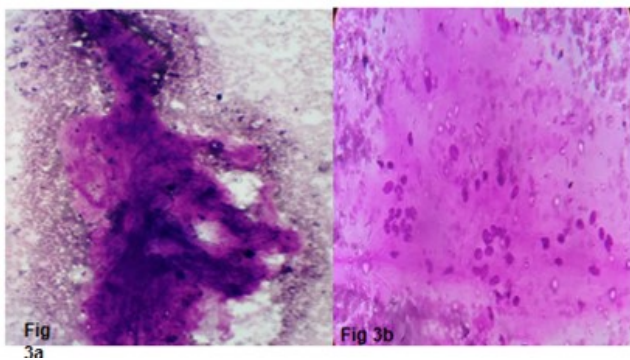


Figure 3: FNAC smears 3a, 3b : showing chondroid stroma with Oval to spindle-shaped cells distributed as chondromyxoid fragments and singly scattered in the smears (Giemsa Stain)

Based on clinicoradiological correlation, cytologic findings, and a positive family history, a diagnosis of hereditary multiple exostoses (HME) was rendered, and a follow-up was advised.

DISCUSSION

Hereditary Multiple Exostoses (HME) is a skeletal disorder affecting bones formed by endochondral ossification. HME is characterized by the formation of ectopic cartilage growth plate-like exostoses near the growing ends of long bones and other skeletal elements. HME is caused by mutations in either of two genes: exostosis (multiple)-1 (EXT1), located on chromosome 8q24.11–q24.13, or exostosis (multiple)-2 (EXT2), located on chromosome 11p11–12.^[1] However, the exact mechanism for multiple osteochondromas remains to be fully elucidated.

Approximately 65% of patients with multiple osteochondromas have a positive family history^[2], as seen in our case.

While previously thought to have male predominance, HME now appears to affect both sexes with equal incidence.^[3] Most patients are diagnosed by the age of 5 years, and virtually all are diagnosed by the age of 12 years, as found in our case.^[4] The late presentation of our case can be attributed to factors such as ignorance related to illiteracy and poor socioeconomic status.

The most commonly involved bones include the distal femur (90%), proximal tibia (84%), fibula (76%), and humerus.^[1] Our case presented with exostoses involving the right humeral head, right tibia, and right ankle joint.

In HME, a noticeable swelling may be the first presenting symptom, as seen in our case. Many osteochondromas are asymptomatic and are only discovered as an incidental finding on X-rays.^[5] Multiple deformities can be seen in osteochondromas around the knees, ankles, shoulder, elbows, and wrist. Genu valgum (knock knees), ankle valgus, ulnar bowing and shortening, and radial head subluxation are common deformities. Other complications include pressure symptoms, fractures, vascular compromise, neurological sequelae, cosmetic deformity, and overlying bursa formation.^[6] No deformities or complications were seen in our case.

Morphologically, osteochondromas are either sessile or pedunculated and range in size from 1 to 20 cm. The cap of an osteochondroma is composed of benign hyaline cartilage of varying thickness and is covered by perichondrium. The hallmark of radiographic diagnosis of HME is the "presence of osteochondromas at the juxtaphyseal region of long bones in which the cortex and medulla of the osteochondroma represent a continuous extension of the host bone".^[4] Suspicion of HME can be established by the presence of ≥ 2 typical radiological lesions with or without a positive family history.^[6] In our case, X-rays showed lesions in the right tibia, right humerus, and bones of the right ankle joint, along with a positive family history.

Fine-needle aspiration cytology (FNAC) is an important technique in the diagnosis of bone tumors. In six cases of osteochondroma reported by Jayshree K. and Jayalaxmi^[7], FNAC showed a combination of mature osteoid and chondroid fragments, with one case showing hematopoietic marrow elements. Garg et al reported similar FNAC findings in a case of HME.^[3] In a study by Chhabra et al^[8], cytospins of a case of osteochondroma yielded particulate material difficult to spread, similar to our case. The smears in their study were also hypocellular and composed mainly of stromal material. The cells were oval to spindle with well-defined cytoplasm and oval nuclei and were distributed as chondromyxoid fragments and as singly scattered in their smears. Few foci of calcification and few osteoclasts and osteoblasts were also noticed in the smears of the above study. Similar findings were also seen in our case, with the FNAC smears showing fragments of magenta-colored matrix with enmeshed spindle cells having plump to elongated nuclei. Foci of calcification were also noted.

While the tumors sometimes regress spontaneously, the importance of studying this tumor lies in the fact that a very small proportion of solitary tumors and multiple lesions evolve into chondrosarcomas.^[7] The suspicion of secondary chondrosarcoma is indicated by growth of the tumor after puberty and the presence of pain. Measurement of the cartilage cap by MRI is an important method to predict malignant transformation of osteochondromas.^[2] Kok has stated that "a threshold value of 2 cm is enough to diagnose malignancy".^[9] Other radiological features of malignant transformation include irregular margins of the cap, osteolytic foci inside the exostosis or near the bone, the presence of a soft tissue mass surrounding the exostosis with scattered or irregular calcifications.^[2] None of these features were seen in our case.

Osteochondromas currently do not have a medical treatment. Surgical removal of exostoses is advised only when there are symptomatic exostoses or there is a suspicion of malignant transformation.^[2] Surgical treatment is often directed at correcting the associated deformities rather than restricted to the exostosis alone. In our case, only follow-up was advised as there were no indications for surgery.

The importance of FNAC as a diagnostic tool can be gauged by the fact that in a majority of cases, the modality of treatment was decided solely on the basis of FNAC without any need for confirmation by biopsy.^[10] Biopsy is indicated only to assess malignant degeneration if radiologically indicated.

CONCLUSION

This case highlights the role of fine-needle aspiration cytology as an adjunct to radiography for the diagnosis of HME. FNAC is a simple, minimally invasive, cost-effective procedure that obviates the need for biopsy to diagnose these lesions.

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