

Hereditary Multiple Exostoses: A Case Report.

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ABSTRACT

Background: Exostoses are bone growths that are abnormal or different from underlying architecture of bone. It starts near growth centres close to joints. Face and skull are severally unaffected. Hereditary Multiple Exostoses (HME) is a rare genetically transmitted bone dysplasia that is inherited in an autosomal dominant manner.

Aim: This paper presents the clinical and radiological features of a six year old boy from South Eastern Nigeria with Hereditary Multiple Exostoses seen in our hospital.

Method: The case record of the patient with Hereditary Multiple Exostoses is presented to highlight the clinical presentation and management options of the condition.

Results: A six year old boy manifested with the features of Hereditary Multiple Exostoses at the age of two. He presented with painless progressive multiple swellings on the trunk, both lower and upper limbs. No deformities were noted. He had no pressure symptoms. Clinical features and Radiographs were diagnostic.

Conclusion: Hereditary Multiple Exostoses with multiple bone spurs exists in our environment, though rare. Treatment still remains watchful waiting, till pressure symptoms develop.

Key words: Hereditary, Multiple Exostoses, Rare Disease.

INTRODUCTION

Hereditary Multiple Exostoses (HME) is a rare autosomal dominant disorder affecting the endochondral skeleton during growth¹. 10-20% can arise spontaneously. It is a cartilage capped bony projection found primarily at the juxta-epiphyseal regions of the most rapidly growing ends of bone².

The long bones of the legs, arms, fingers, toes and shoulder blades are commonly affected. Face and skull are severally unaffected. Exostoses grow as the child grows. It is the most common bone tumour seen in children³⁻⁴.

It is considered a hamartoma and as such stops growing at the end of the growth of the affected bones. It is estimated to occur in about 1 in 50,000 people. It causes asymmetrical retardation of longitudinal bone growth with subsequent deformity and discrepancy in

limb-length (very common). Significant inequality of more than or equal to 2cm has been reported. Malignant transformation is in order of 5% of all cases⁵. The femur is twice affected as the tibia⁶.

Mutations in three genes (EXT 1, EXT 2 and EXT 3) have been implicated in the aetio-pathogenesis.

These lesions have the tendency to cause mechanical interference with normal function of the soft tissues passing over them.

The pressure of the exostoses causes irritation and occasional damage to nerves, arteries and muscles, hence, the presence of pain. A second clinical setting, presents as multiple lumps, pain or deformity, while the third setting will present with multiple lumps, pain, and deformities. Management will depend on the stage of presentation.

In the absence of pain and deformities, masterly inactivity is the rule. However, surgical intervention is paramount in the presence of deforming complications. We hereby present a 6 year old boy with clinical and radiological features of Hereditary Multiple Exostoses (HME) seen in our hospital. This is the only case seen in our hospital in the last ten years.

CASE REPORT

O.Ca 6 year old pupil, presented at the surgical outpatient department of Nnamdi Azikiwe University Teaching Hospital (NAUTH) with a four year history of multiple swellings on the trunk and both upper and lower limbs. The Swellings were noticed by the parents. They were multiple and on the anterior chest wall, back and both lower limbs. Initially, they were small sized (0.5cmx1cm) but progressively increased in size over time. No history of pain, change in skin colour or ulceration. No history of trauma. No history of weight loss, headaches, anorexia, bone pain, jaundice, cough or breathlessness, abdominal swelling or any other constitutional symptom. Pregnancy was carried to term. Perinatal period was uneventful. No family history of similar swelling.

General examination was unremarkable. On examination of the musculo-skeletal system, there were multiple non-tender hard swellings over the anterior chest wall (largest 3 – 6cm; smallest 1-2cm in widest diameters). There were also multiple non-tender hard swellings on the antero-medial surface of the proximal third of both arms. There were similar swellings on the distal third of both forearms.

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On both lower limbs, there were similar bony hard swellings on the medial and lateral surfaces of the distal third of both thighs and medial side of the proximal third of both legs (figure I). However, the lesions on the right lower limb are bigger. There were no limb length discrepancies.

There was no change in colour of overlying or surrounding skin. No differential warmth or lymphadenopathy. No loss of distal neurovascular function. A diagnosis of hereditary multiple exostoses was made

The result of haemoglobin estimation, genotype, erythrocyte sedimentation rate, and full blood count were within normal ranges for his age. Radiograph of the chest showed bony out-growths from the sternum and ribs. Radiograph of both knees showed bony out-growths in the distal femoral and proximal tibial metaphysis, growing away from the physis (Figure II). In the upper limbs, radiograph of the shoulders and wrists revealed multiple bony outgrowths. Biopsy of one of the lesions showed Osteochondroma on histopathology.

Parents and patient were counselled on outcome and natural history of the ailment. He is presently on three monthly checks- up visits.

Figure I: Clinical picture of patient's knee.



Figure II: AP radiograph of the patient's knees.



DISCUSSION

Hereditary Multiple Exostoses is an autosomal dominant hereditary disorder. 10-20% arises spontaneously. There are mutations in three genes: EXT 1 which maps to chromosome 8q24.1, EXT 2 which maps to 11p13, and EXT 3 maps to short arm of chromosome 19. HME affects both sexes equally⁷.

Mutations in these genes cause synthesis of truncated EXT protein with abnormal function. EXT protein is important in Heparan Sulfate synthesis. It is thought that normal chondrocyte proliferation and differentiation may be affected, leading to abnormal bony growth. Pre-implantation genetic testing and prenatal diagnosis are available for new couples. HME has 95% penetrance. Expressivity is variable. In a patient with a negative family history of the disease, the patient may be the first to clinically express the trait, just like in our patient. This may be due to sporadic mutation.

Exostoses are initially recognised and diagnosed in the first decade of life in over 80% of individuals with HME. Tibia and scapula are often most noticeable locations⁸. Clinical and radiological findings are usually diagnostic. Clinical features will depend on time of presentation. Pathology will depend on site, size and extent of physal involvement.

Multiple bony swellings of the proximal humerus and distal radius and ulnar, with swellings of the distal femur and proximal tibia are the hallmark of presentation. The scapula and pelvis can be involved⁹. Late presentations are usually accompanied by complications such as pain and deformities. Peripheral

Figure III: AP radiograph of the patient's ankles.



nerve compression symptoms can occur in up to 22.6% of patients. Peroneal Neuropathy in children is an example¹⁰. Malignant changes do occur with transformation from osteochondroma to chondrosarcoma. The incidence of malignant transformation ranges from 0.5-2.5%¹¹.

In our patient, there was an early presentation at the age of two. Symptoms were mainly multiple swellings on the proximal humerus, distal radius, distal femur and proximal tibia.

As corroborated by other works^{6,7,9}, early presentations are not usually associated with pain and deformity. Hence, he has no pain or deformity. There was no asymmetrical growth retardation or limb length discrepancy. However, atypically he had chest wall bony swellings. This has not been documented as a common predilection site.

CONCLUSION

Hereditary Multiple Exostoses is a rare disease. Sporadic mutations still occur in patients with no family history. The treatment of non-complicated HME is watchful waiting, with the aim of intervening surgically when complications arise.

ETHICAL CONSIDERATION

Written informed consent was obtained from the patient and parents for publication of this case report and accompanying images.

CONFLICTING INTERESTS

The authors declare that they have no competing interests.

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