



## Hereditary Multiple Exostoses a case report

### KEYWORDS

osteochondroma, hereditary multiple exostoses, deformities

**Dr Ujjwala S Keskar**

**Dr Sanjay A Natu**

M.D(Pediatrics) .Associate Professor,Dept of Pediatrics, Smt Kashibai Navale Medical College ,Off Mumbai-Banglore bypass,Narhe,Pune

M.D(Pediatrics) . Professor,Dept of Pediatrics, Smt Kashibai Navale Medical College ,Off Mumbai-Banglore bypass,Narhe,Pune

### ABSTRACT

*Hereditary Multiple Exostoses (HME) is an autosomal dominant disorder affecting rapidly growing bones. It usually presents after the age of two years as multiple bony growths on the appendicular skeleton. Although exostoses are benign, they are often associated with characteristic progressive skeletal deformities and may cause clinical symptoms because of mechanical pressure on surrounding tendon, nerves, and vessels. The most common deformities include short stature, limb-length discrepancies, valgus deformities of the knee and ankle, asymmetry of the pectoral and pelvic girdles, bowing of the radius with ulnar deviation of the wrist and subluxation of the radiocapitellar joint . Malignant transformation to chondrosarcoma is seen in 0.5 to 5 % cases [1, 2, 7, 8].It is a rare condition (incidence of 1/50000) , not commonly seen in our country .*

### Introduction:

Exostoses is a cartilage capped bony projection found primarily at the juxta-epiphyseal regions of the most rapidly growing ends of long bones. HME is manifested by the presence of multiple exostoses also called dyschondroplasia, multiple congenital osteochondromata, deforming chondrodysplasia, diaphyseal aclasis (which means that the modelling of the entire affected bone area is abnormal).

Although exostoses are benign lesions, they are often associated with characteristic progressive skeletal deformities and may cause clinical symptoms because of mechanical pressure on surrounding tendon, nerves, and vessels.

### Case Report

A 9-year-old Hindu female child of non-consanguineous marriage presented with multiple bony projections on chest, around knee, ankle and wrist since last three years. Girl was short [height 116.5 cm (<3<sup>rd</sup> centile)] and had deformity of right wrist and left ankle. She complained of tingling numbness sensation and pain while writing with right hand. There was no history of similar swellings in any of the family members. She had multiple hard, non – tender, fixed bony swellings 0.5 cm to 1 cm in size around both wrists, knees, left ankle and on fifth rib bilaterally. There was shortening of the right upper limb by 5 cm with deformity of wrist and ulnar deviation of forearm (Fig.1).In the left lower limb there was shortening by 3 cm and deformity of ankle.

Skeletal survey showed multiple osteo-cartilagenous tumours at the metaphysis of the distal femur, proximal and distal tibia and fibula; right distal radius, ulna and costochondral junction of fifth rib. HRCT chest showed small sessile, bony, verrucous lesions from multiple ribs & few vertebral bodies. There was no intrathoracic extension. Lungs and mediastinum were normal.

She got operated for right hand deformity. Right distal ulnar exostoses was excised with placement of external fixator in right ulna. Histopathology of the lesion revealed osteochondroma. The postoperative period was uneventful and post-operative x-rays showed complete excision. The patient has remained pain-free since then and is still being followed up in the outpatient clinic.



(Fig .1)

### Discussion

HME is an autosomal dominant disorder with 100% penetrance but with variable expression and male predominance 1.5:1[1, 8, 10].Family history of similar lesions is present in >90 % cases [10].Our patient had no family history. Therefore in a family with a negative history, the patient may be the first person to demonstrate the clinical expression or it could be a sporadic mutation.HME is a genetically heterogeneous disorder and is associated with mutations in EXT1 or EXT2 genes which are both tumour suppressor genes. EXT genes encode glycosyltransferases, termed 'exostosins', which are involved in the biosynthesis of heparan sulphate (HS). MHE affected individuals are HS-deficient that causes a local error in the normal negative feedback loop regulating chondrocyte proliferation and maturation that, consequently, results in premature differentiation and abnormal bone growth at the growth plate(exostoses) [1,4,5].

Osteochondromas develop and increase in the size in first decade of life and cease to grow when the growth plates close at puberty [3, 7, 8]. A diagnosis of HME can be made when radiologically at least two osteochondromas of the juxta-epiphyseal region of long bones are observed [8]. It can be confirmed by histological evaluation.

The characteristic radiographic appearance of exostoses may vary from broad based (sessile) or pedunculated. The cortex of exostoses is continuous with that of the underlying bone. They originate in the metaphysis in long bones and migrate to the diaphysis as growth continues in the epiphysis. The cartilagenous cap is usually radiolucent, but after puberty irregular zones of calcification may appear. Extensive calcification with irregularities of the cap should suggest the possibility of malignant change [6]. It is suspected if there is growth or pain in the tumour after puberty or if there is increased thickness of the cartilagenous cap (over 1cm) in adults [8].

Individuals with HME are frequently of short stature, with most having heights 0.5 to 1.0 SD below the mean. Sitting height is generally less abnormal than total height, indicating that the limbs are involved disproportionately as compared to the spine [1, 6]. In our child height was affected more (<3rd centile). It could be because of individual bones which were more severely affected causing severe short stature [1, 6].

Forearm deformities are found in 40%–60% of patients with HME [1, 7]. Most common observation is disproportionate shortening of the ulna. It is attributed to its greater growth relative to that of the radius and to its smaller cross-sectional diameter (one fourth diameter of radius). (Fig. 2).



(Fig. 2)

Valgus knee deformities are found in 8%–33% of patients with HME [1, 3]. Fibula with smaller cross sectional area has been found to be shortened disproportionately as compared to the Tibia and this is likely responsible for the con-

sistent valgus direction of the deformity [4]. The anatomic proximity of the lesion to the physis is responsible for its tethering effect on one side of the physis, thus producing an asymmetrical growth of the bone. This latter effect is the basis of the limb-length discrepancy and the observed forearm varus, ulnar deviation, genu and ankle valgum in HME [2]. Prevalence of clinically significant limb-length discrepancy that is inequality (2 cm or greater) has been reported to be 10%–50% [1, 7]. Our child had significant limb-length discrepancy and deformities at right wrist and left ankle.

The implication of the progressive pathology of the condition is that the clinical presentation may be in three stages. In the first stage, patients will present with multiple lumps at the end of the long bones without any significant pain or deformity. The second stage will feature multiple lumps with pain or deformity while the third stage will consist of multiple lumps, pain and multiple deformities. Management therefore depends on the presentation. In the presence of uncomplicated exostoses, management is essentially by masterly inactivity as these patients have a normal life expectancy.

However, surgical intervention (corrective osteotomy, epiphyseodesis, excision, limb lengthening) is indicated when the exostoses exhibit pressure symptoms, cause deformity or undergo malignant transformation [1]. The mean age of onset of malignant degeneration is 31 years; it seldom occurs before the 10th or after the 50th year [6]. The upper end of the femur and the pelvis are the main locations, but they may also be found in the shoulder girdle and the ribs [3, 7, 8].

The study on 283 patients with HME in Netherlands by Goud et al. found that in children with HME 30% had stopped playing sports as they grew. Those attending school 50% had complaints, mostly related to physical activities but also about difficulty in writing and using computers or being bullied. Approximately 60% of the children had pain (usually associated with a more negative perception of their disease), problems at school, and a need for greater number of surgical procedures [9].

In conclusion HME is a progressive disease of the bones that causes pain, deformities and pressure symptoms. It affects their cosmetic appearance and compromises the quality of life.

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