Abstract:
INTRODUCTION - Hereditary Multiple Exostoses has several synonyms like chondral osteoma, metapyseal achlasia, diaphyseal achlasia, etc. Being a rare condition the deformities disabilities need to be taken into account before planning management. Here we present a series of 4 cases with different age, sex, site, benign malignant distribution. METHODS AND MATERIAL - 2 male and 2 female patients. 2 patients presented before and 2 patients after skeletal maturity. All of them presented with multiple exostoses. First patient was operated on right elbow even though deformity was in left forearm. Second patient was operated on left proximal humerus. Third patient was advised conservative management. Fourth patient underwent wide excision in left femur for malignant transformation. Classification System Used - Masada et al. Karyotyping was done in all cases. Uncommon surgical approaches (Burger and Buckwalter for Prox. humerus, Antero Lateral Approach for Head of Radius, Medial approach for femur) were used for all the 3 cases which were operated. Even though all patients had multiple tumors only those causing symptoms, disability, malignant transformation were surgically managed. None of the patients had similar family history. DISCUSSION - Even though both our female patients had forearm deformities fit in Masada et al classification those forearms were left untouched because there was no disability there is no evidence that surgical intervention will improve forearm deformities. Biopsy prior to definitive management is absolutely essential in selected doubtful (malignant) cases. HPE was undifferentiable from solitary osteochondroma. Individualize the treatment plan or surgical approach for each patient. Masada et al guidelines may not be applicable to all cases. Karyotyping may point out only gross chromosomal aberrations. Only Gene sequencing may help to predict malignant transformation. CONCLUSION - Deformities cannot be prevented by early excision of osteochondromas. Only symptomatic lesions deserve surgical
management mere presence is not an indication for surgery. No surgical intervention improves forearm deformities. Of late less aggressive surgical approach is advised by most authors.

**Keyword**: Masada et al, Burger and Buckwalter, Karyotyping

**INTRODUCTION:**
Hereditary Multiple Exostoses is a rare developmental dysplasia at the level of growth plate which result in cartilaginous capped excrescences with an incidence of 1 in 50,000 having an autosomal dominant inheritance (1). Even though several synonyms like chondral osteoma, metapyseal achlasia, diaphyseal achlasia etc are named by various authors the one commonly used is **HEREDITARY MULTIPLE EXOSTOSES** as per Jafee in 1943 (1). Being a rare condition the deformities & disabilities may present at various degree from mild cosmetic disfigurement to severe deformities and a thought process on management need to be taken into account and individualized a per the patient since all deformities do not cause functional disabilities. Here we present a series of 4 cases with different age, sex, site, benign /malignant distribution.

**METHODS & MATERIAL:**
All these patients presented at our OP as part of the General Population.

This study was conducted in the Year 2010 - 2011. **Classification System Used**: Masada et al. **Case 1:**

**Clinical picture**

- Pre-op X-ray
- X-ray of both elbows

An Initiative of The Tamil Nadu Dr M.G.R. Medical University
University Journal of Surgery and Surgical Specialities
24 Year old female presented with complain of Pain in the Right elbow for 6 months & swelling since childhood. Examination revealed bilateral Multiple Tumor masses in the distal femur & Proximal Tibia, bilateral Genu valgum with the right elbow being fixed in supination and flexion restricted to 80 degree with bowing & shortening of 2 cms of left forearm. No other member in their family had similar swelling or complaints. Investigations confirmed the diagnosis & patient had normal karyotyping. Right elbow deformity didn’t fit into any of the classification of Masada et al whereas the deformity of Left forearm had features suggestive of Type I (4). An excision biopsy of the mass was done through an Anterolateral approach and other swellings were left untouched. Post operatively Range of movements increased to about 120 degrees with full supination & pronation.
X-ray
3D - CT scan
15 Year old male presented with complain of pain in Left arm for 10 months & multiple swelling in left arm since childhood. Examination revealed three bony swellings on the poster lateral aspect of left arm. Investigations confirmed the presence of exostoses in proximal humerus with a normal karyotyping. An excision biopsy of the mass was done using a Burger & Buckwalter approach to the proximal humerus.
Both forearms
13 Year old female presented with complain of occasional Pain in Right forearm and ankle for 6 months & multiple swellings in both arms, both forearms, knee & ankle since childhood. Examination revealed bilateral Genu valgum with Multiple palpable tumor mass around knee, ankle, shoulder with shortening of 2.5 cms & Bowing of Right forearm with a Masada et al Type II b (4). No other member in their family had similar swelling nor complaints. Investigations confirmed the presence of multiple cartilaginous outgrowths in distal femur, proximal humerus, Proximal & distal tibia with right forearm bowing and a normal karyotyping.

X-ray both forearms
A conservative approach was preferred with only occasional NSAIDS for mere pain relief.

X-ray shoulder

X-ray both knees
24 Year old male presented with complaint of pain in left thigh for 6 months, multiple hard lumps around knee since childhood with sudden increase in the pre-existing left thigh swelling for past one year. Examination revealed multiple bony swellings on the postero-lateral aspect of right thigh, knee of 5*5 cm and a scar over the swelling on the left thigh of 17*9 cm revealing an attempt for excision of the mass in another hospital which was abandoned due to unknown reasons. No other member of the family had similar complaints. Patient has undergone surgery for similar problems during childhood details of which were not available. Investigations confirmed the presence of multiple cartilaginous outgrowths in distal femur, proximal humerus, proximal & distal tibia with huge suspicious malignant lesion over left femur and a normal karyotyping. A wide excision of the mass was done through an anteromedial approach.

DISCUSSION:
The disease condition has an equal prevalence as in our case series which also had equal prevalence. Most of the cases present with multiple lumpy bumps by 20 years (5) as against our case series where 2 of our cases presented after 20 years of age. None of their family member had similar swelling or complaints. The arm span was shorter than the height in cases 1 and 3, while it was longer than the height in cases 2 and 4. The average lower:upper body segment ratio was found to be 0.97. Hence there is shortening of the lower segment when compared to the upper segment. Even though both our female patients had forearm deformities that fit in Masada et al classification, those forearms were left untouched because there was no disability & there is no evidence that surgical intervention will improve forearm deformities.
Masada et al guidelines may not be applicable to all cases.

In all our cases Karyotyping was done but couldn’t find any chromosomal aberration. All our cases had normal karyotyping against the literature review which reveals a genetic abnormality in 90% of the cases. **Chromosomes 8, 11, 19** associated with three etiological genes **EXT1**, **EXT2 & EXT3**, where malignant transformation & severe forms are common with **EXT1** (4).

Biopsy prior to definitive management is absolutely essential in selected doubtful (malignant) cases. HPE was undifferentiable from solitary osteochondroma & all cases were confirmed with histopathology- confirming the diagnosis (1).

For case 1 an excision biopsy of the mass which was continuous with radial head in the right elbow through an **Anterolateral approach** between brachioradialis and biceps (6) used against conventional posterior approach was done and other swellings were left untouched.

For case 2 excision biopsy of the mass which was done through **posterior Burger & Buckwalter approach** between the long & lateral head of triceps against the conventional posterior approach revealed three bony protuberances with a common base. For case 4 Prior to excision the radiologist confirmed the lesion as benign with cartilage cap thickness of 8mm in MRI and HPE of the biopsy specimen revealing chondrosarcomatous change from the biopsy specimen obtained only in the region of cartilaginous cap & oncopathologist confirmed the final excised mass margins as free from malignant cells. Surgical oncologist suggested that Chondosarcomatous change was primarily in cartilagenous cap only & that tumor mass was not continuous with marrow. Hence, mass on the medial side alone to be excised by wide excision which was agreed by our hospital Tumor Board also.

<table>
<thead>
<tr>
<th>CASE</th>
<th>ARM SPAN (cm)</th>
<th>HEIGHT (cm)</th>
<th>LOWER BODY SEGMENT (cm)</th>
<th>UPPER BODY SEGMENT (cm)</th>
<th>LOWER UPPER BODY SEGMENT</th>
</tr>
</thead>
<tbody>
<tr>
<td>CASE 1</td>
<td>141</td>
<td>145</td>
<td>71</td>
<td>74</td>
<td>0.96</td>
</tr>
<tr>
<td>CASE 2</td>
<td>155</td>
<td>152</td>
<td>77</td>
<td>75</td>
<td>1.03</td>
</tr>
<tr>
<td>CASE 3</td>
<td>127</td>
<td>130</td>
<td>62</td>
<td>68</td>
<td>0.91</td>
</tr>
<tr>
<td>CASE 4</td>
<td>159</td>
<td>157</td>
<td>78</td>
<td>79</td>
<td>0.90</td>
</tr>
</tbody>
</table>

**KARYOTYPING**

Karyotyping may point out only gross chromosomal aberrations. Only Gene sequencing may help to predict malignant transformation. More than 60 different kind of mutation have been reported so far in different foci. New study - novel mutation nt112delAT in EXT2 gene & EXTL (or EXT-like – sequence) gene similar to both EXT1 and EXT2 (1).
A wide excision of the mass was done through Anteromedial approach versus the routine lateral one involving the lesion, parts of quadratus femoris, sartorius, adductor muscles and the cartilage were removed in toto.

CONCLUSION:
Individualized treatment for individual patients is preferred rather than a uniform protocol. Only symptomatic lesions deserve surgical management & mere presence is not an indication for surgery. Biopsy before definitive management in doubtful cases (3). Individualize surgical approach for each patient rather than following standard approaches (2). No surgical intervention improves forearm deformities. No evidence that deformity can be prevented by early excision. But if done before maturity – recurrence is common.

Screening for disease among Family members will help to pick up asymptomatic cases. Genetic Study (karyotyping) will reveal only gross aberration & hence Gene sequencing would be helpful for those suspicious of malignant transformation.

Hence less aggressive surgical approach is preferred with excision of symptomatic lesions only. Moreover no surgical intervention improve forearm function.

REFERENCES:
1. Tachdjian's Pediatric Orthopaedics 4th edition
2. Chapman’s Orthopaedic Surgery 3rd edition
3. Campbell’s Operative Orthopaedics 11th edition
5. Turek’s Orthopaedics 6th edition