Utility of multimodality imaging in a curious case of crippled child

Babu Peter Sathyanathan and Gopi Bagya S. K.

*Barnard Institute of Radiology, Madras Medical College, Chennai, Tamil Nadu, India

Abstract

Here we are reporting a case of Trevor's disease a.k.a dysplasia epiphysealis hemimelica (DEH) in a 13 year male child, who initially presented with swellings on his right knee and ankle joint, causing severe progressive pain on walking. Clinical diagnosis of chondrosarcoma was given. CT investigations of right knee and ankle joints were done and the findings remained inconclusive to arrive at a diagnosis. Patient was advised for MR evaluation of right ankle swelling to delineate soft tissue involvement. MR ankle was done and it was suggestive of multiple benign lesions arising from right ankle joint involving multiple bones predominantly arising from their medial aspect of proximal and distal epiphyses. The radiograph and CT images were reviewed again and based on MRI findings, the diagnosis of classic type of Trevor's disease was drawn at the end. Thus saving the child from the stress of unwanted aggressive surgical resection of tumor and aiding in tailoring the treatment strategy for this benign crippling lesion.

Keywords: Trevor's disease, Dysplasia Epiphysealis Hemimelica, Osteochondromas, Magnetic Resonance Imaging
Introduction
Trevors’ disease is a rare and severe debilitating disease affecting the epiphysis (1-3) of long bones causing bony outgrowths (osteochondroma like) with cartilaginous cap, when involving joint surface can result in severe disabling pain to the patient. Epiphyseal involvement can result in limb shortening and limb length discrepancy.

Case Report
The patient, a 13-year old male child, was referred to our radiology department as a radiologically diagnosed case of chondrosarcoma of right knee joint, with imaging done elsewhere. The patient had presented with a swelling over right knee joint developing slowly over a period of 2 years. The patient presented to the orthopaedic department with above mentioned complaints and after local examination, provisional diagnosis of chondrosarcoma of right knee joint was given. Clinical examination and detailed history taking revealed the chronic and benign course of the

Fig.2: (a) T1 & (b) PD fat saturated sequence of right ankle joint showing multiple osteochondromas arising from the epiphysis of right tibia, talus and navicular bones. (c) radiograph of right ankle joint. (d) & (e) CT showing multiple osteochondromas arising from bones of right foot

The diagnosis is usually achievable with computed tomography and X rays, but MRI helps in visualizing the cartilaginous cap, compression of adjacent neurovascular bundle and other soft tissue.
swelling. Upon closer examination, it became evident that the deformities also involved his right ankle joint. The patient was apparently healthy otherwise and had no antecedent history of trauma or systemic illnesses. The patient was then subjected to CT of right knee and ankle joints and MR right ankle joint.

**Imaging**

- CT of right knee joint revealed multiple sessile bony outgrowths arising from distal femoral epiphysis, Patella and Proximal tibial epiphysis involving articular surface. Florid pedunculated bony lesion with irregular surface arising from medial condyle of distal femur noted with multiple loose bodies seen within the right knee joint. (Fig. 1 a-d)

- CT of right ankle joint revealed multiple sessile bony outgrowths arising from distal tibial epiphysis, articular surfaces of talus involving tibiotalar, talocalcaneal and talonavicular joints, navicular and cuneiform bones. (Fig. 2 d,e)

- MR ankle showed medullary and cortical continuity between the lesions and the adjacent bones with medial predominance of the lesions and no surrounding edema. (Fig. 2 a,b)

**Radiological diagnosis**

Initially misled by the history, the approach was in terms of a malignant lesion. But after a series of discussions, we centered in on the obvious diagnosis. Summary of the findings from CT and MRI were:

- Irregular epiphyseal bony outgrowths arising from multiple bones of right knee and ankle joints
- Showing cortical and medullary continuity with adjacent normal bones
- Favoring predominantly the medial aspect of joints involving a single limb.

All these factors point towards the diagnosis of "Dysplasia Epiphysealis Hemimelica" of classical type.

**Biopsy**

Biopsy from distal femoral epiphyseal florid bony outgrowth was done and the report came out to be osteochondroma favouring the diagnosis of Dysplasia Epiphysealis Hemimelica. (Fig. 3)

**Discussion:**

Plain Radiography:

The typical appearance is of asymmetric epiphyseal cartilaginous overgrowth, containing multiple ossific centres. The pattern of epiphyseal chondral calcification may be stippled, irregular, or dense. Metaphyseal widening and remodelling may occur. The ossification centres of affected bones typically appear early, and premature closure of the affected epiphysis may occur. The resulting limb length inequality, joint incongruity, and articular surface irregularity predisposes to early onset osteoarthritis.

**Computed tomography (CT):**

CT demonstrates the typical chondroid nature of the calcifications, medullary continuity and will identify cortical irregularity of the lesion, or that secondary to degenerative changes within the joint. The multiplanar reformattting capability of CT allows detailed evaluation of the extent of joint involvement and is a valuable tool for surgical planning.

**Magnetic Resonance Imaging (MRI)**

MRI is the technique of choice for identifying the epiphyseal origin of the osteochondroma like lesions in Trevors' and confirming the medullary continuity of the bony lesions. The condition of the articular cartilage and degree of joint deformity can be evaluated by MRI, as can any neurovascular or ligamentous involvement. The cartilage cap of the lesion has T2 hyperintensity and intermediate signal intensity on T1-weighted sequences. Focal areas of dense calcification within the marrow may be seen as signal voids on T1 and T2-weighted sequences.

**Conclusion:**

With a prevalence of 1:1,000,000 and a male predominance in children of age group around 3–4 years, only Ollier's disease and hereditary mul-
tiple exostoses can present with multiple bony lesions as seen in our case. But the rarity of the lesion with its atypical features make it difficult to diagnose and the complications arising out of the lesions are much debilitating to the patient with resultant reduction in mobility and pain during young age. Plain radiograph plays a vital role in diagnostic evaluation of osteochondroma like lesions, though CT can be more sensitive in identifying and localising the lesions. In most cortical based lesions, MR imaging prior to CT evaluation can result in diagnostic errors (4-6). Hence it is essential to follow a protocol in imaging of bony lesions to delineate the nature and extent of the lesions.

License Policy:

This work is licensed under a Creative Commons Attribution 4.0 International License. This license requires that reusers give credit to the creator. It allows reusers to distribute, remix, adapt, and build upon the material in any medium or format, even for commercial purposes.

References


Conflict of interest:

The authors declare that there were no conflicts of interest within the meaning of the recommendations of the International Committee of Medical Journal Editors when the article was written.

Disclaimer/Publisher’s Note:

The statements, opinions and data contained in all publications are solely those of the individual author(s) and contributor(s) and not of Swiss J. Radiol. Nucl. Med. and/or the editor(s), Swiss J. Radiol. Nucl. Med. and/or the editor(s) disclaim responsibility for any injury to people or property resulting from any ideas, methods, instructions or products referred to in the content.