CASE REPORT

Trevor - Fairbanks Disease: Dysplasia Epiphysealis Hemimelica of the Knee – A Case Report and Review

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ABSTRACT

Dysplasia Epiphysealis Hemimelica (DEH) is a rare skeletal developmental disorder involving epiphyses of long bones. It primarily emerges during childhood and is characterised by an osteocartilaginous overgrowth involving epiphyses of long bones. Although the exact aetiology of DEH remains elusive, it can be treated conservatively or surgically via excision or corrective osteotomies, depending on the patient's clinical presentation. Radiological imaging must be carried out before surgery to prevent expected intraoperative complications if the patient is planned for surgical intervention. We are reporting a case of an 8-year-old child who presented with left knee pain with imaging demonstrating a DEH. The patient then underwent surgery. The patient had a rapid recovery following several rehabilitation sessions, and no recurrence was reported at the follow-up. The surgical outcome of DEH is unpredictable and largely dependent on several factors. Surgical options should be offered only to those with impacted symptoms.

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INTRODUCTION

Trevor disease, also known as Dysplasia Epiphysealis Hemimelica, was first documented by Mouchet and Beloit as "tarsomegalie" in 1926 [1,2]. The term "Dysplasia Epiphysealis Hemimelica" was only introduced later by Fairbank in 1956, describing the occurrence of epiphyses in joints other than the ankle and is confined to the lateral or medial half of an epiphysis [3]. It is characterised by osteocartilaginous overgrowth involving the epiphyses of long bones [4]. The reported incidences are mostly patients between the ages of 2 to 14, and the occurrences in boys are three times more than in girls [5]. Patients usually present with asymmetric, painless swelling affecting one side of the joint of the lower limb. In some cases, the patient may have limping, limited range of motion, varus or valgus deformity or limb length discrepancy [6].

Hence, management strategies varied from observation to surgical intervention depending on the severity of the symptoms and joint functions. This case report aims to review the literature with attention to its diagnosis and treatment modalities for the disease.

CASE REPORT

An 8-year-old boy presented with left knee swelling 2 years ago, complaining of limited range of motion of the left knee and pain during ambulation. Further history revealed a history of trauma 5 years ago and claimed a sustained hairline fracture over the left distal femur, which was treated conservatively with a cast.

On physical examination, he had a genu valgum with a limitation to range of motion between 30 to 100 degrees on flexion-extension and a fixed flexion deformity of 30 degrees and pain on terminal flexion. A firm-to-hard swelling was palpable over the anteromedial joint line. A scannogram of the bilateral lower limb revealed valgus deformity of the left knee. Otherwise, there is no overt limb length discrepancy (figure 3). Left knee radiographs revealed cartilaginous enlargement over the posterior medial epiphysis of the proximal tibia with several loose bodies (figure 4). MRI of the left knee revealed multiple predominantly ossified and coalesced loose bodies at the posterior medial aspect of the femur fused to the medial femoral condyle with a lobulated lesion at the infrapatellar fat pad with intercondylar extension (figure 5).

A left femur biopsy was performed, and the findings were minimal tissue composed of cartilaginous, bone, and fatty marrow. There were no malignant cells detected. The patient eventually underwent open exploration, resection of medial femoral condyle chondromatosis, resection of synovial chondromatosis, impaction bone grafting, hyalofast implantation, biceps femoris tendon and iliotibial tract release.

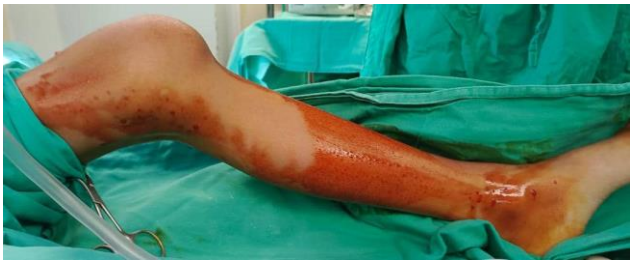


Figure 1

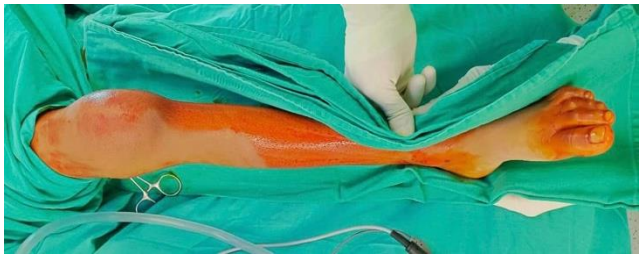


Figure 2

Figure 1 and 2 Examination shows fixed flexion deformity of left lower limb

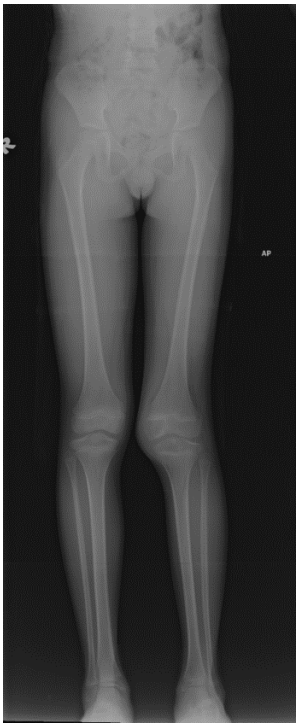
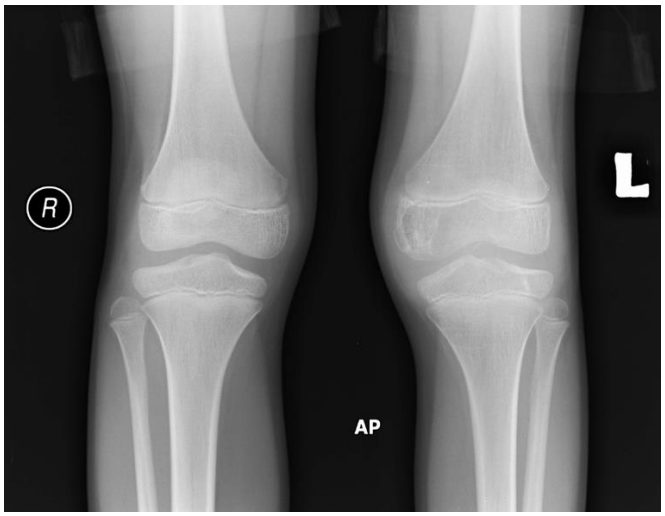


Figure 3 Scannogram shows valgus deformity over left lower limb with no limb length discrepancy



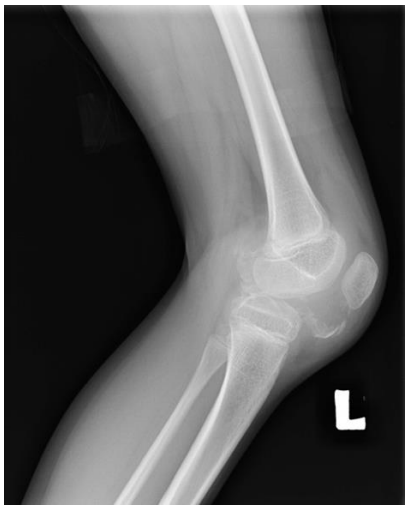
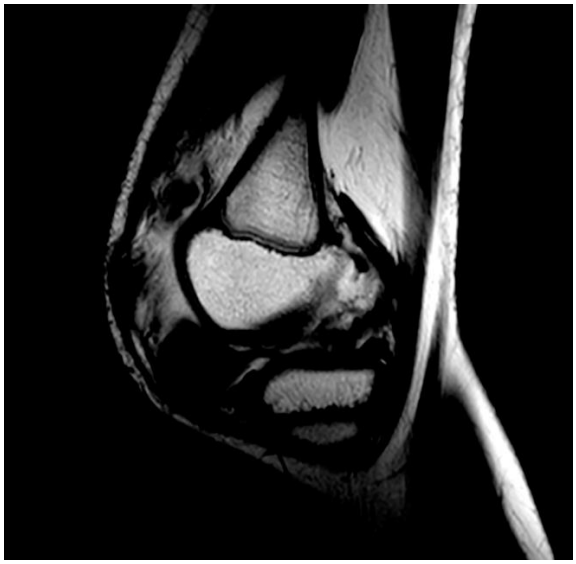


Figure 4A Figure 4B

Figure 4A and 4B: X-ray of the left knee shows cartilaginous enlargement over the posterior medial epiphysis of the proximal tibia with the presence of several loose bodies.



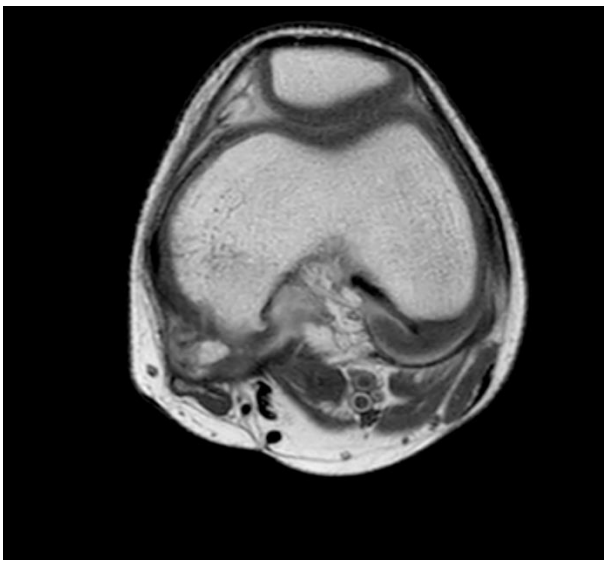


Figure 5A Figure 5B

Figure 5A and 5B MRI T1 show hyperintensity at the posterior medial condyle of the left knee at the epiphysis.

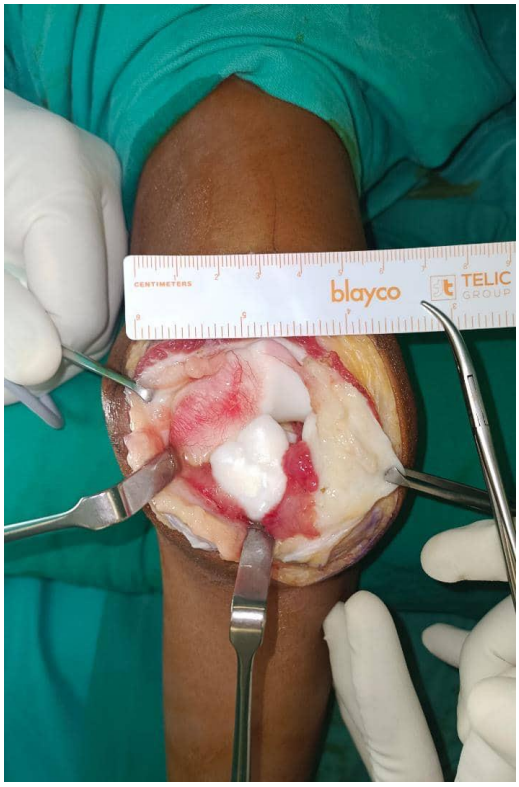


Figure 6

Figure 6 Intra-op knee joint chondromatosis adhered to ligamentum mucosum.

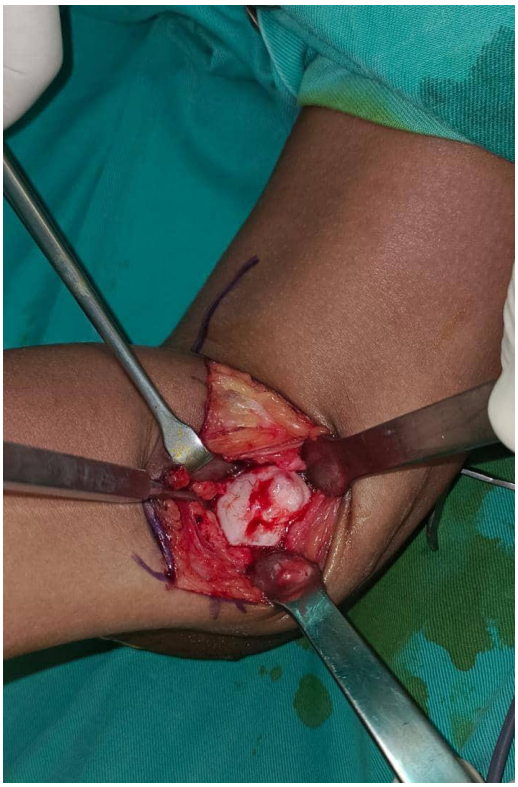
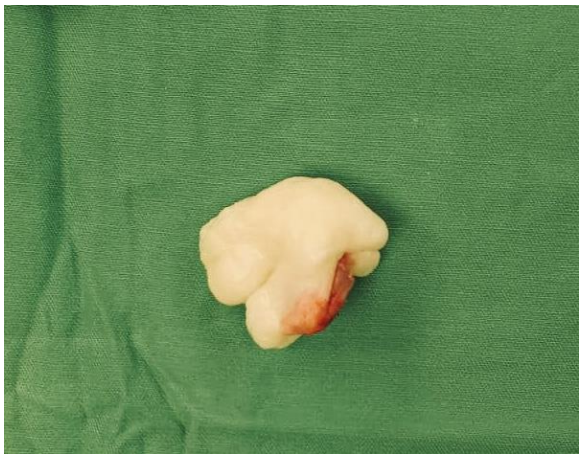


Figure 7

Figure 7 Intra-op posterior medial chondromatosis partly adhered to innermost fibers.



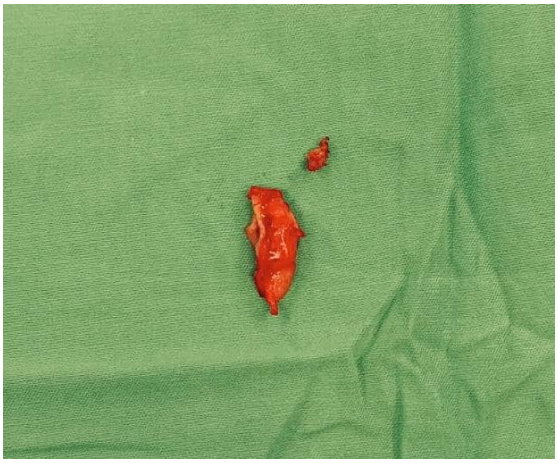


Figure 8A and 8B HPE: Synovial chondromatosis with synovial ossification.

DISCUSSION

Dysplasia Epiphysealis Hemimelica is a rare skeletal developmental disorder that is one of the variants of skeletal osteochondromas. It is characterised by osteochondromas involving the epiphysis of the long bones.[4]

Although the aetiology of DEH is still unclear. There were a few proposed theories:[1]

1. Insult to the fetal limb apical ectoderm.
2. Changes in the arrangement of blood vessels at the epiphysis.
3. Failure of peripheral cartilaginous cells to degenerate.
4. Abnormal proliferation of cartilaginous cells.

Depending on the patient's clinical symptoms, DEH can be treated conservatively or through surgical intervention. For asymptomatic patients, these patients should be under observation until skeletal maturity to evaluate disease progression.[4] In cases where the lesion is causing pain, deformity or interference with function, surgical intervention should be offered to improve and reverse function via mass excision with or without corrective osteotomy.[6] However, excision of articular lesions is not recommended as it may cause postoperative degenerative joint changes.[7]

Surgical intervention provides better outcomes in these cases:[8]

1. Exostoses that are compressing neurovascular structures or tendons.
2. Exostosis that interferes with extremities growth.
3. Varus or valgus deformities of joint.
4. Functional limitations of the joint.
5. Pain.

In preoperative planning, magnetic resonance imaging (MRI) is the gold standard compared to computed tomography (CT) scans. MRI can evaluate cortical and medullary continuity between the accessory ossification centre and parent bone, identify intraoperative complications such as fractures and osseous deformity, and assess the relationship between the neurovascular bundle.[4] MRI can also be used to differentiate other tumoral disease.[7]

The latest literature has suggested a whole-body MRI (WBMRI) as a relatively fast imaging method for Trevor's disease [9]. WBMRI can demonstrate additional lesions throughout the affected limbs without the use of ionizing radiation, revealing the typical pattern of overgrowth seen in DEH. Hence, it was proposed to add WBMRI in centres equipped with those services to the routine MRI examination performed to evaluate the lesion of Trevor's disease. WBMRI have clear therapeutic advantages; if a distribution pattern typical of DEH is found, it constitutes a strong argument to withhold the need for a biopsy.[9]

CONCLUSION

Trevor's disease is a rare skeletal developmental disorder characterised by cartilaginous overgrowth involving the medial or lateral half of the epiphysis. If a patient present with a bony lesion over the epiphysis region, we must raise suspicion of DEH instead of osteochondroma. Several studies have shown that DEH can be treated either conservatively or with surgical intervention depending on the patient's clinical symptoms; however, currently, there are no standard guidelines on the treatment of DEH.

Preoperatively, the patient should be assessed for functional limitations, deformity, or pain. MRI should be done before surgical resection to prevent intraoperative complications. The option of a WBMRI, if available, should be considered as a management protocol for Trevor's disease for surveillance of other joints and acquiring an imaging diagnosis, avoiding the need for a biopsy before the commencement of intervention.

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