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## CASE REPORT

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# Dysplasia Epiphysealis Hemimelica (Trevor-Fairbank Disease): a Case Report

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## INTRODUCTION

Dysplasia epiphysealis hemimelica (DEH) — originally described by Mouchet and Belot<sup>1</sup> — most commonly affects lower extremity joints especially the knees and ankles.<sup>2,3</sup> Usually, the body is affected on only one side, and the condition is often confined to either the medial or the lateral side of the limb.<sup>4</sup> It is a rare disorder, affecting males three times more commonly than females, with a reported incidence of 1:1 000 000, whose aetiology and pathogenesis are still unclear.<sup>1,2</sup> To the best of our knowledge, this is the first reported case of DEH in Southeast Asia. We aim to inform fellow clinicians of the specific imaging findings of DEH, such that the diagnosis can first be suggested from radiographs as well as other cross-sectional studies such as magnetic resonance imaging (MRI).

## CASE REPORT

In October 2008, a 16-month-old boy presented to the paediatric orthopaedic clinic of the Universiti Kebangsaan Malaysia Medical Centre, Malaysia, with

left ankle deformity, which had been apparent since the child had started to walk. The patient had no history of trauma, fever, or pain. There was no known family history of any joint or bone problems. On physical examination, the patient had a painless firm protrusion palpable at the medial aspect of the left ankle with calcaneal valgus of the left ankle. Initial radiographs done upon early presentation showed non-specific findings of soft tissue swelling at the medial aspect of the ankle with irregularity of the medial aspect of metaphysis of the distal tibia and talus (Figure 1). Initial impression was epiphyseal or metaphyseal dysplasia. The patient was primarily managed by observation, with serial radiographs for assessment.

Follow-up examinations over approximately 2 years showed development of mild tibial bowing, a bony prominence at the medial aspect of left knee, and macrodactyly of the left big toe with increasing length discrepancy between the big toes (Figure 2a). Serial radiographs upon follow-up showed epiphyseal dysplasia

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of the proximal and distal tibia, and dysplastic changes of the left first metatarsal, medial cuneiform, navicular, and talus (Figure 2b-d). The patient underwent percutaneous epiphysodesis of the proximal growth plate of the left

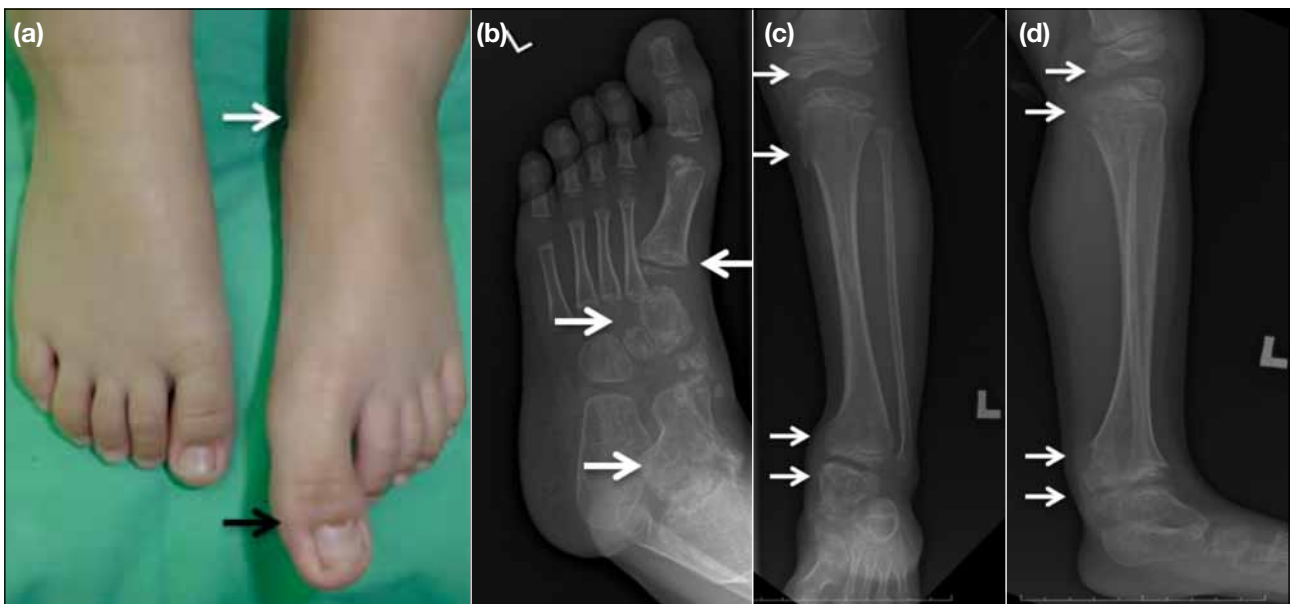
first metatarsal bone at age 3 years to reduce the length of the big toe.

At follow-up clinical examination at age 8 years, the patient's lower limbs showed a reduced discrepancy in the length of the big toes. However, there was further bowing of the tibia and bony deformity at the periarticular region of the medial aspect of the knee and ankle joints (Figure 3a). Lower limb radiographs showed more prominent epiphyseal bony outgrowths and deformity of the metatarsal, medial cuneiform, navicular, tibia, and fibula, with ankle joint ankylosis (Figure 3b-c). Findings were then more supportive of epiphyseal origin of the lesions and the diagnosis of DEH was considered. Subsequently, MRI revealed multiple osteochondral overgrowths which showed medullary and cortical continuity with the underlying epiphyses of the medial aspect of the left distal femur, left proximal and distal tibia, and medial left talus, supporting the diagnosis of DEH (Figure 4a-d).

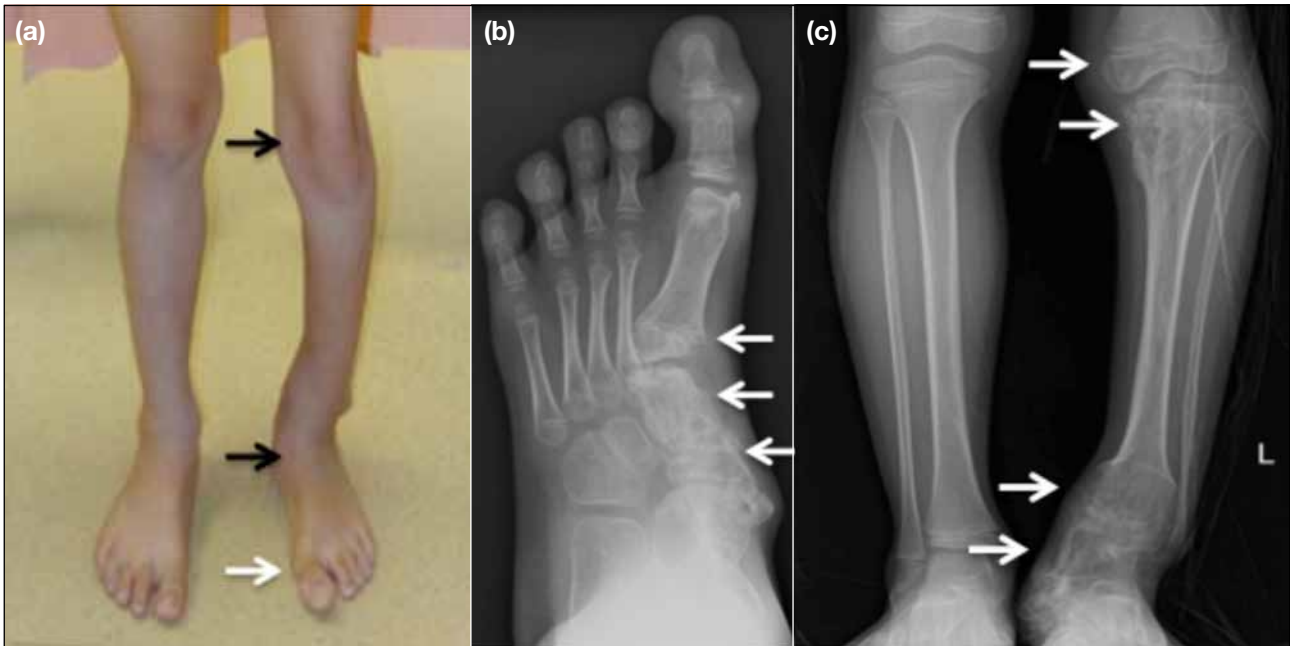
At age 10 years, the patient underwent guided growth procedures to correct the left knee and left ankle deformities. A trephine biopsy was taken from the



**Figure 1.** Patient with dysplasia epiphysealis hemimelica, age 16 months. Anteroposterior radiograph of the left tibia and fibula showing soft tissue swelling at the medial aspect of the metaphysis of the distal tibia and talus (white arrows). Initial impression was epiphyseal or metaphyseal dysplasia.



**Figure 2.** Same patient with dysplasia epiphysealis hemimelica, age 3 years. (a) Perioperative photograph of foot and ankle showing macrodactyly of the big toe (black arrow), causing length discrepancy, and bony prominence of the medial aspect of left ankle (white arrow). (b) Corresponding anteroposterior radiograph of the left foot showing enlargement and irregularities of the proximal epiphysis of the first metatarsal, medial cuneiform, navicular, talus, and distal tibial epiphysis (white arrows). (c and d) Anteroposterior and lateral radiographs of the left tibia and fibula, showing irregular bony outgrowths arising from the epiphyseal surfaces of the distal posteromedial femoral condyle and the proximal posteromedial tibial condyle. The epiphysis of the distal tibia and talus are irregular (white arrows). There is remodelling and mild expansion of the adjacent metaphysis of the proximal and distal tibia (white arrows).



**Figure 3.** Same patient with dysplasia epiphysealis hemimelica, age 8 years. (a) Photograph of the lower limbs showing reduced degree of big toe discrepancy post epiphysiodesis (white arrow). There is now bowing of the tibia and bony deformity at the periarticular region of the medial aspect of the knee and ankle joints (black arrows). (b) Anteroposterior foot radiograph showing more prominent epiphyseal bony outgrowths and deformity of the metatarsal, medial cuneiform and navicular bones (white arrows). (c) Anteroposterior bilateral tibia fibula radiograph showing more prominent epiphyseal osseous lesions (white arrows). There is now gross deformity of the ankle joint with ankylosis (white arrows). Findings are now more supportive of epiphyseal origin of the lesions.

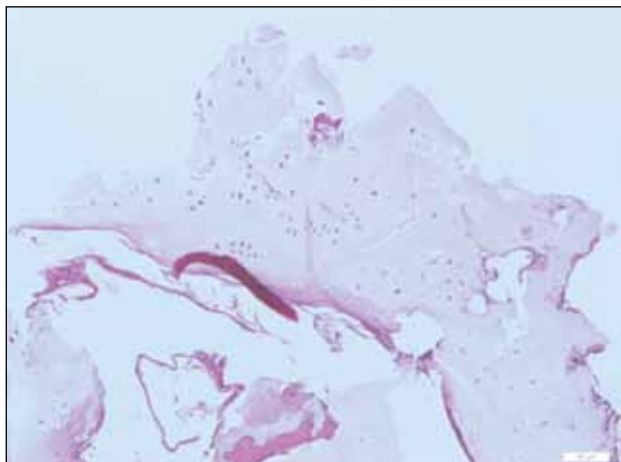


**Figure 4.** Same patient with dysplasia epiphysealis hemimelica, age 8 years. (a) Coronal T1-weighted, (b) coronal proton density T2-weighted fat saturation, and (c) sagittal views in T1-weighted magnetic resonance imaging (MRI) scans of the left knee showing medullary and cortical continuity between the bony outgrowth and the underlying epiphysis of the distal femur and proximal tibia (white arrows). There is remodelling of the adjacent metaphysis of medial and posterior part of proximal tibia (white arrows). (d) Coronal T1-weighted MRI scan of the left ankle showing similar lesions arising from the medial aspect of talus and distal tibia with marrow continuity and osseous mass (white arrows). There is ankylosis of the ankle joint. Findings are supportive of and consistent with dysplasia epiphysealis hemimelica.

left distal tibia epiphysis which showed presence of cartilaginous cap lacking endochondral ossification with disorganised cartilaginous growth, suggestive of the underlying DEH (Figure 5).

## DISCUSSION

Previous studies have emphasised the difference between the osteocartilaginous overgrowth of DEH originating from the epiphysis and the bony exostosis originating



**Figure 5.** Biopsy of the left distal tibia showing presence of cartilaginous cap lacking endochondral ossification. The disorganised cartilaginous growth may suggest the underlying dysplasia epiphysealis hemimelica (haematoxylin and eosin stain; x 20).

from the metaphysis.<sup>2,4,5</sup> Clinical manifestations are variable, with the most common presenting complaint being painless deformity around a joint to a painful joint with mechanical symptoms.<sup>6</sup> Different degrees of involvement are observed in this disease, subdivided into localised, classical, and generalised forms. The classical form, affecting more than one epiphysis in a hemimelic distribution of the limb, is the most common.<sup>6</sup> Our patient had disease involving the distal femur, proximal and distal tibia, talus, medial cuneiform, navicular, and first metatarsal, in a hemimelic distribution, and is thus classified under the classical form.

### Imaging Features

Often, radiology findings of DEH are characteristic. Radiographs typically show an irregular mass with focal ossification, arising from one side of the affected epiphysis or sesamoid bone. The pattern of epiphyseal chondral calcification may be stippled, irregular or dense. The lesions may be either single or multiple and usually enlarge with skeletal growth and may later be associated with joint deformity. After maturation, the lesions slowly ossify to become confluent with the underlying epiphysis. Premature physal closure with resultant deformity and occasional limb discrepancy may also be observed. Although the metaphysis is usually not involved directly, widening and remodelling of the metaphysis may occur as sequelae of the epiphyseal lesion. Irregularity of the articular surface may lead to early secondary osteoarthritis.<sup>5,7</sup> In our patient is was

initially unclear if the origin of the periarticular osseous lesions was epiphyseal or metaphyseal. The epiphyseal origin became more obvious after further follow-up examinations over the 6 years after the initial radiograph.

MRI is accurate in identifying the lesion and determining its anatomical relations, and can show the extent of the tumour, providing the status of bone and associated soft tissue changes. MRI is also important to demonstrate the signal intensity of the tumour and its continuity with the primary site, as well as secondary changes in adjacent menisci, tendons, ligaments, and muscles. In some patients, it is possible to observe a cleavage plane between the cartilage of the unossified mass and the normal epiphyseal cartilage.<sup>6,8</sup> As the lesion matures, MRI may demonstrate cortical and medullary coalescence between the ossification centre of the lesion and the primary epiphysis, with marrow signal intensity similar to that of the underlying bone. Gadolinium contrast may occasionally play a role in cases of a complication or a rapidly growing osteochondroma. Subcutaneous soft tissue enhancement may be seen in cases with secondary inflammatory changes.<sup>5,7</sup> In our patient, MRI confirmed the diagnosis by demonstrating the continuity of the osseous lesions with the underlying epiphysis.

### Pathology

The DEH lesions were initially thought to be multiple epiphyseal osteochondromas. Genetic studies show that DEH lesions lack the EXT1 and EXT2 gene mutations seen in osteochondromas, and therefore is considered as a different entity. Although DEH arises from the epiphysis, osteochondromas are typically metaphyseal in origin. DEH is composed of epiphyseal osteocartilaginous nodules, which resemble secondary ossification centres, which then matures into an osteochondroma-like cap over time. These nodules may form bands of cartilaginous tissue, which separate islands of cancellous bone, a feature that is not observed in an osteochondroma. Furthermore, endochondral calcification or ossification, which is a usual prominent feature in maturing osteochondromata, is only focally seen in DEH.<sup>7</sup> The histopathological findings are consistent with growth plate dysplasia, which is not specific for, but does not exclude, the diagnosis of DEH.

### Differential Diagnosis

The differential diagnosis of a peri- or intra-articular mass can be narrowed down by a multi-imaging approach in combination with a clear and accurate history. DEH should be differentiated from other osteocartilaginous

lesions that include synovial osteochondromatosis, capsular or para-articular chondromas, and particularly osteochondromata.<sup>9</sup> Other differential diagnoses include myositis ossificans, infection, chronic infantile neurologic, cutaneous and articular syndrome, tumoural calcinosis, and vascular or parasitic calcification.<sup>5</sup> In our patient, the other most likely initial differential was osteochondromata, owing to the initial difficulty in determining the epiphyseal or metaphyseal origin of the lesion on the then-16-month-old child's radiograph.

### Treatment

The optimal treatment of DEH remains controversial, and depends on the location, involvement intensity, and degree of functional incapacity. Asymptomatic patients are periodically reviewed, with serial radiographic follow-up examinations. Surgical procedure is indicated in cases of pain, joint deformity or incongruence, or motion limitation.<sup>5</sup> Our patient was initially observed, then managed surgically by percutaneous epiphysodesis of the left first metatarsal, later followed by the left knee and ankle owing to progressive deformity.

### CONCLUSION

A rare cause of bony peri- or intra-articular swelling, DEH may be difficult to differentiate from other skeletal dysplasia on conventional radiography. Therefore, MRI

is recommended, on which DEH can be identified by the medullary and cortical continuity of the lesion with the adjacent epiphysis. Distinct clinical and radiologic findings and histopathological confirmation are crucial in the diagnosis of DEH, to ensure optimal management to minimise associated skeletal deformity and function.

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