

# Dysplasia epiphysealis hemimelica (Trevor's disease)

## 7 of our own cases and a review of the literature

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**Background and purpose** Dysplasia epiphysealis hemimelica is characterized by irregular overgrowth of cartilage in the epiphysis, usually affecting the knee and ankle. We treated 7 children by surgery between 1980 and 2005. After reporting one child case, we summarize our cases and the cases described in the literature.

**Method** We discuss the diagnosis of this dysplasia, especially the role of radiography. We describe the suggested treatment, which could be surgical or non-surgical depending on the location and the symptoms.

**Results** After reviewing 57 cases, we found that this dysplasia occurs twice as often in males as in females. The medial side of the epiphysis is affected twice as often as the lateral side. In two-thirds of the cases, more than one epiphysis was affected. If the location of the exostosis suggests that it might lead to joint deformity, early surgical excision is recommended.

**Interpretation** Since there is often involvement of more than one epiphysis, we emphasize the importance of a skeletal survey once this dysplasia is diagnosed.

Dysplasia epiphysealis hemimelica (DEH) is a rare skeletal developmental disorder characterized by asymmetric overgrowth of cartilage in the epiphyses. It is considered to be an osteochondroma of the epiphysis. The reported incidence is 1 in 106 (Bhosale et al. 2005).

This condition was first reported as "tarsomégalie" by Mouchet and Belot (1926), which refers to the most common location. Trevor (1950) described similar cases, using the name "tarso-

epiphyseal aclasis". Fairbank (1956) reported patients with this condition and he used the term "dysplasia epiphysealis hemimelica", which is still in use today.

### Case report

A 6-year-old boy came to our department with a painless swelling on the posterior-medial aspect of his right ankle. The patient had been treated 8 months previously for what was thought to be an aseptic talus necrosis

Physical examination revealed a 3 × 3 cm tender, bony mass below and behind the right medial malleolar area. There was no limitation of ankle movements. Radiography of the right ankle showed an irregular calcified mass on the posterior side of the talus (Figure 1). The overgrowth was surgically removed from the posterior part of the talus. Path-

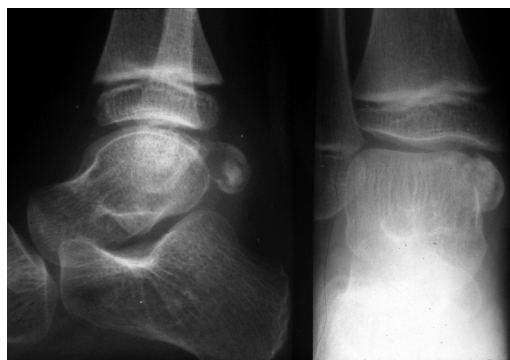


Figure 1. Lateral and anteroposterior radiograph of the right ankle showing an irregular calcified mass on the posterior side of the talus.

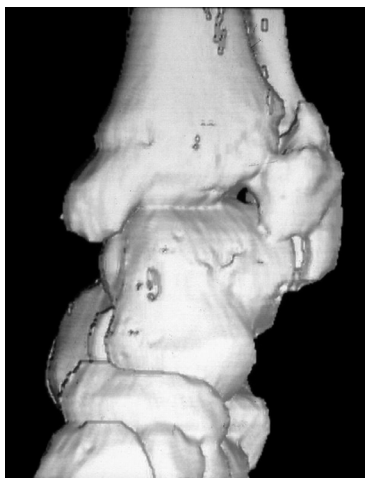


Figure 2. 3D CT scan showing an exostosis arising from the lateral side of the distal epiphysis of the tibia.

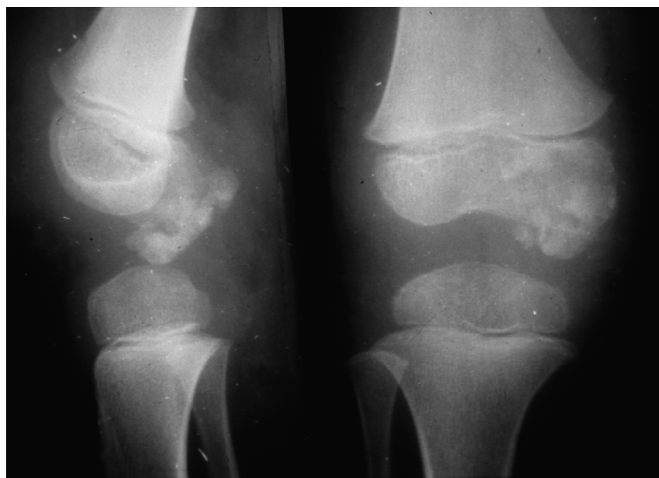


Figure 3. Lateral and anteroposterior radiographs of the knee showing a cloudy structure and exostosis with bone-like density at the medial side of the femur.

ological examination showed an osteochondroma covered with cartilage. Histologically, the trabecular bone was covered by a thick cap of hyalin cartilage.

The patient was followed for 1 year, and showed no recurrence of the deformity and no limitation of his ankle movements.

## Discussion

### *Etiology*

DEH is the consequence of irregular overgrowth of the epiphysis. The etiology is unknown, but there is no evidence to suggest hereditary factors and no cases of malignant transformation have been reported (Bhosale et al. 2005). Different theories have been put forward about the origin of this condition. Trevor (1950) explained it as irregular cell proliferation in the superficial zone of the articular cartilage, and Fairbank (1956) as damage of the apical cap of the limb bud in early fetal development.

Hensinger et al. (1974) reported 7 cases in 2 generations of the same family; the cases were combinations of dysplasia epiphysealis hemimelica, intracapsular chondroma, and typical osteochondroma. 6 members of the family had DEH, 3 presented as intracapsular chondroma, and 2 had typical osteochondroma. The upper and the lower limb

were affected in 3 cases and in 4 cases only the lower limb was affected. There were 5 males and 2 females; their ages at the first visit were between 12 and 37 years. 6 of the 7 patients underwent surgery for removal of the lesions that were symptomatic. According to this study, the common occurrence of DEH, intracapsular chondroma, or typical osteochondroma shows autosomal dominance with variable expression.

### *Incidence and symptoms*

Usually the age of onset is between 2 and 14 years, but it has also been described in adults (Shinozaki et al. 1999). Males are affected 3 times as frequently as females (Acquaviva et al. 2005). The most common presenting symptom is a painless mass around the affected joint. However, other reported manifestations are swelling, pain, restricted range of motion, deformity, and limb length discrepancy. According to Fairbank (1956), the most common symptom is increasing swelling over the medial or lateral aspect of the knee or ankle. The swelling feels bony in consistency, and the surrounding soft tissues are not involved. When the medial side of the lower limb is affected, knock knee or flat foot is observed only in the affected leg. The child may fall often and there may be limitation of movement at the affected joint.

Our patients also complained of a painless mass of bony consistency around the affected joint.



## Legends to Table

- A Authors
- B Sex
- C Age
- D Scapula
- E Scaphoid bone
- F Acetabulum
- G Proximal epiphysis of the femur
- H Distal epiphysis of the femur
- I Proximal epiphysis of the tibia
- J Tuberositas of the tibia
- K Distal epiphysis of the tibia
- L Distal epiphysis of the fibula
- M Talus
- N Calcaneus
- O Navicular bone
- P Cuboid bone
- Q Cuneiform bone

Mild pain and restricted range of motion was also common.

Azouz et al. (1985) have classified this condition in 3 groups: (1) localized, affecting only one epiphysis, (2) classic, affecting more than one epiphysis in the same limb, and (3) generalized, involving the whole lower limb. The most common is the classic hemimelic form and the medial side is affected twice as often as the lateral (Azouz et al. 1985). It usually affects the lower limb—in the distal femur or proximal tibia, talus, and tarsus. Upper limb involvement has been reported, in the carpal bones (Vanhoenacker et al. 1999, Takagi et al. 2000).

A summary of our patients and the cases found in the literature is given in the Table. There were 57 patients altogether (39 males). Their ages ranged from 3 months to 33 years. In 34 cases the osteochondroma was found at the medial side of the affected joint, in 18 it was found at the lateral side, and in 2 it was found on both sides. Furthermore, in 1 case the lesion was located in the acetabulum, in 1 case in the tuberositas of the tibia, and in 1 case it was located in the scapula. The ankle was affected in 36 patients, the knee in 31, the tarsal bones in 10, the hip in 8, and the upper limb in 3 (Figure 4). In 36 of the 57 patients, DEH was found in multiplex form, i.e. more than one epiphysis was involved.

### Diagnosis and pathology

Diagnosis of DEH is based on plain radiographs, which initially show an irregular calciferous mass arising from one side of the affected articular car-

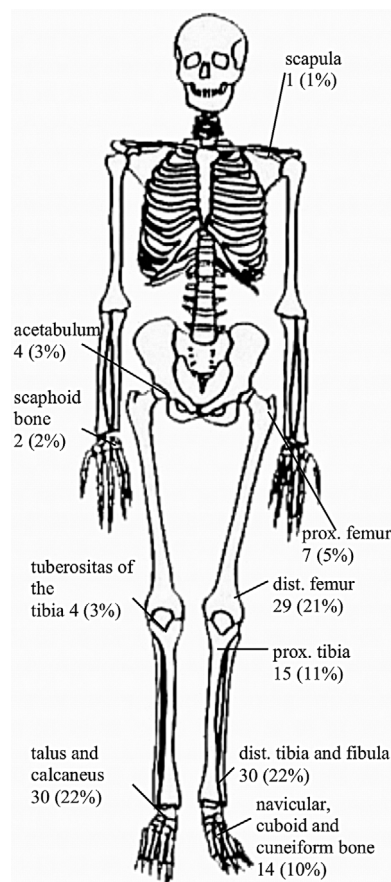


Figure 4. The most frequent locations of DEH in the skeleton. The numbers and percentages of occurrence in the different epiphyses (according to our data) are also given.

tilage. Multiple ossification centers appear at one side of the epiphysis. Later on, these ossification centers become confluent with the underlying bone. Sometimes—especially in the early stages of DEH—identification by radiography may be difficult, as the lesion can be mistaken for a tumor. CT can be helpful to demonstrate the relationship between bones, soft tissues, and cartilaginous mass. Three-dimensional reconstructed images (Figure 2) may be of value for surgical planning (Wenger and Adamczyk 2005). MRI appears to be very useful for determination of the extent of epiphyseal involvement, joint deformity, and soft tissue changes. Teixeira et al. (2001) reported the role of bone scintigraphy, which shows increased uptake in the pathological epiphyseal area. They suggested that once DEH is diagnosed, a whole body skeletal scintigraphy should be done to check

for other sites of involvement. Although biopsy is not necessary, it might be useful early on when the radiographic changes are not yet typical or when masses are unusually located. Histologically, the lesion resembles osteochondroma that is continuous with the underlying normal bone and with a cartilage cap over it. The osteochondroma arises from the metaphysis or the diaphysis of the bone, while DEH usually arises from the epiphysis. Radiography of our patients also showed irregular calcification around the affected joint (Figure 3).

Maroteaux et al. (1993) described 2 patients who were mother and son. In this study, the authors emphasized the similarities and differences between DEH and dominant carpotarsal osteochondromatosis (DCO), especially in the radiographic findings, localization, and incidence. The radiological features of both diseases are very similar: irregular cartilage surface, calcification, and detached ossification center. In DCO, some affected bones show disorganized development; their volume is increased, their contours are irregular, and they frequently have a fragmented appearance. The simultaneous involvement of the upper and lower limbs is characteristic of DCO; furthermore, the osteochondroma can be asymmetrical and bilateral also. Bilateral involvement has also been reported in DEH (Merzoug et al. 2002) and the lower limb is more often affected. According to Maroteaux et al. (1993), DCO is transmitted with autosomal dominant inheritance, while DEH occurs sporadically.

#### *Treatment and prognosis*

The treatment of DEH is still discussed in the literature and is different depending on the location. Surgery should be undertaken if the lesion is causing pain, joint deformity, or limited range of motion. Asymptomatic patients may not require treatment since there is no known risk of malignant transformation (Kuo et al. 1998).

Acquaviva et al. (2005) classified the lesions as extraarticular and intraarticular, depending on the location of the osteochondroma. With extraarticular localization, simple excision of the mass yields favorable results. If the osteochondroma is intraarticular, osteotomy might be needed to correct angular deformity. Kuo et al. (1998) divided the lesions into juxtaarticular and articular forms. With juxta-

articular localization, when the lesion was adjacent to the articular surface they had excellent results with excision of the osteochondroma. They did not recommend excision in the case of lesions directly involving the joint surface, with articular localization, unless the lesion becomes a loose body. Excision of the lesion with articular localization may lead to early osteoarthritis, which might require arthrodesis.

The patients treated for DEH at our department presented with painful or painless swelling and limited range of motion. The localization of the lesions was juxtaarticular or articular, and the osteochondroma was excised in all the patients.

The prognosis of DEH is variable, and depends on the location and size of the lesion. If the lesion is intraarticular, resection may increase the risk of degenerative joint disease even further. In all cases, there is a risk of recurrence of this disease until the epiphyses are closed. Continuous monitoring may be needed.

#### *Conclusions*

To our knowledge, this study is the most extensive investigation of DEH patients to date. According to our data, males are twice as prone to this disease as females and the medial side of the epiphysis is affected twice as commonly as the lateral side. In two-thirds of the cases, more than one epiphysis was affected. Once DEH is diagnosed, we recommend that a skeletal survey should be done, looking for multiple locations. If the location of the exostosis could lead to joint deformity, early excision is recommended to avoid early degenerative joint disease. If resection affects articular cartilage, implantation of an autologous or homologous osteochondral graft may be indicated, similar to the treatment used in osteochondritis dissecans.

#### *Contributions of authors*

GS contributed with ideas and set down the basics of the study. KK helped with the radiographs. SK, TT, and VMR documented the patients who were treated surgically at our department, searched the literature, and processed all the data. VMR and SK wrote the manuscript.

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