INTRODUCTION

Dysplasia epiphysealis hemimelica (DEH), or Trevor disease, is a rare developmental disorder affecting the epiphyses in young children. The first report of DEH in the literature was by Mouchet and Belt in 1926, who described the condition as a tarsal bone disorder and used the term tarsomegalie. In 1950, Trevor reviewed 10 cases of DEH and used the term tarsoepiphyseal aclasis. In 1956, Fairbank reported 14 cases and coined the term dysplasia epiphysealis hemimelica.

DEH is thought to be a variant of osteochondroma arising within a joint.
**Problem:** DEH is a benign disorder, and no cases of malignant transformation have been reported. The natural history of DEH is a continuous increase in size of the lesion until skeletal maturity. Therefore, the long-term prognosis for untreated lesions involving the weightbearing surface of the joint, although unreported, is no doubt a progression toward pain and arthrosis.

**Frequency:** The incidence of DEH has been estimated at 1 case per million population. It is a rare disorder, both in the United States and internationally.

No race predilection is known to exist. DEH is most commonly found in males, with a male-to-female ratio of 3:1. It usually manifests in childhood and early adolescence.

**Etiology:** The etiology is unknown; the disease does not appear to be genetically transmitted.

**Pathophysiology:** The cause of DEH is unknown, but many theories exist. In a 1983 report, Connor et al suggested that the fundamental defect seems to be an abnormality of the regulation of cartilage proliferation in the affected epiphysis, resulting in cartilaginous exostosis. Trevor considered DEH to be a congenital error of epiphyseal development; the etiologic factor affecting the limb buds during early fetal life was thought to involve an altered process of cell proliferation at the superficial zone of articular cartilage, allowing for persistent proliferation and production of a large cartilaginous mass. In 1956, Fairbank suggested that the disorder was due to a localized disturbance of the preaxial or postaxial part of the apical cap of the limb bud in early fetal development.

**Clinical:** Most patients present with painless swelling or a mass on one side of the joint, limitation of motion, and occasionally, recurrent locking of the joint.

DEH most commonly occurs around the knee, talus, and tarsal navicular and first cuneiform joints. The medial side of the epiphysis is most commonly affected. A painless swelling occurs on the affected joint, with occasional decreased range of motion, angular deformity, and concomitant regional muscle wasting.

Differential diagnoses include chondroblastoma, osteochondroma, and enchondroma.

DEH should be treated if the lesion is causing deformity, pain, or interference with function. Most cases are treated surgically.
**Relevant Anatomy:** Refer to *Surgical Exposures in Orthopedics: The Anatomic Approach*, by Stanley Hoppenfeld, MD.

**Contraindications:** Surgery is contraindicated if no medical symptoms or no mechanical block is present.

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**WORKUP**

**Lab Studies:**
- No distinctive laboratory abnormalities are associated with DEH.

**Imaging Studies:**
- Plain radiography may reveal early focal calcification of the affected site with later appearance of an irregular bony enlargement, which becomes fused to the affected epiphysis.
- Computerized tomography scanning assists in defining the anatomic relationship between the mass and its parent bone and in evaluating the condition of the articular cartilage and soft tissue.
- MRI depicts the unossified cartilaginous mass in great detail, as well as the status of the articular cartilage, and helps to differentiate the abnormal epiphyseal growth from the main epiphysis.
- Double contrast arthrography assists in delineating the joint space deformity and the extent of cartilaginous mass and the joint dynamic, but it is an invasive diagnostic tool.

**Histologic Findings:** Macroscopically, the bone enlargement is consistent with an exostosis and normal hyaline cartilage.

Microscopically, the histologic appearance of the lesion is that of a well-defined cartilage cap over projecting bone that is contiguous with the underlying normal bone and is indistinguishable from that of an osteochondroma. It is consistent with a benign osteochondroma. The basic pathologic process appears to be abnormal cartilage proliferation in an epiphysis with associated enchondral ossification before complete ossification. A cleavage area of cartilage is present between the ossification center in the lesion and that of the epiphysis.

**Staging:** In 1985, Azouz et al classified DEH into the following categories:
- Localized, involving only one epiphysis
- Classic (most common), affects more than one area in a single limb
- Generalized, affects the whole lower limb from pelvis to foot
Medical therapy: Supportive joint care, consisting of short-term splinting of the joint, may be of benefit.

Surgical therapy: Most of the reported cases of DEH in the literature have been treated surgically, in the form of excision of the mass while preserving the integrity of the affected joint as much as possible and correction of angular deformity.

Follow-up care: After surgery, the affected limb requires a short period of immobilization with a cast or splint followed by active range of motion of the joint.

### COMPLICATIONS

- Recurrence of the angular deformity after the corrective osteotomy - May be anticipated if the growth plate at the affected joint is open and active and the lesion itself has not been removed
- Degenerative osteoarthritis
- Fixed deformity
- Leg-length discrepancy
- Loose bodies
DEH, although an uncommon entity, results in considerable disability because of direct involvement of the articular surface of the joint. Surgical excision offers the best results, but corrective osteotomy and reoperation for recurrent lesion may also be required.

Caption: Picture 1. Dysplasia epiphysealis hemimelica. Anteroposterior and lateral radiographs of the right ankle of a child aged 3.5 years showing a lesion in the medial dome of the talus and epiphyseal part of fibula. It causes ankle pain and deformity.
Caption: Picture 2. Dysplasia epiphysealis hemimelica. Anteroposterior radiograph of the patient from Image 1 at age 7 years. Radiograph depicts an increase in the size of the lesion where it was excised, but it did recur at age 12 years, at which time the lesion was removed.

Caption: Picture 3. Dysplasia epiphysealis hemimelica. Anteroposterior and lateral radiographs of the right ankle of a child aged 3.5 years revealing a lesion in the medial dome of the talus and epiphyseal part of fibula. The lesion causes ankle pain and deformity.

BIBLIOGRAPHY

Dysplasia Epiphysealis Hemimelica excerpt

