The case:
An 11-year-old boy presented with a 2-year history of left ankle pain and abnormal gait, worsened with sports activities such as football and wrestling.

Figure: Standing anteroposterior radiograph of the left ankle (A) with lateral radiograph of the left foot (B).

Your diagnosis?

For answer see page 269
Diagnosis:

Dysplasia Epiphysealis Hemimelica (Trevor Disease) of the Ankle

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An 11-year-old boy presented with a 2-year history of aching-type left ankle pain. The pain was not present at night but occurred with activities, especially recreational football and wrestling. Physical examination denoted absence of an antalgic-type gait, but he walked with his left foot externally rotated. There was tenderness along the medial left ankle joint and talus without appreciable edema in the ankle joint region and no mechanical joint findings on dynamic examination. There were no signs of planovalgus foot with standing.

Anteroposterior (AP) and lateral radiographs of the ankle and foot demonstrated a partially ossified, lobulated intra-articular mass within the medial gutter of the ankle joint (Figure 1). Computed tomography (CT) and magnetic resonance imaging (MRI) of the ankle were obtained to further define the lesion along with its position in relation to surrounding structures. Computed tomography denoted an irregularly shaped, protuberant bony lesion connected to the anteromedial talus and a contiguous component of partially calcified soft tissue that demonstrated a cleavage plane with the talus (Figure 2). Magnetic resonance imaging demonstrated protuberant bone and soft tissue adjacent to the medial talus. The signal in the anterior component was mostly isointense to bone, whereas the signal in the posterior component was partially isointense to cartilage, with areas of low signal corresponding to the areas of calcification (Figure 3A). A thin cartilage cap was also present, isointense to articular cartilage (Figure 3B). There was mild edema in the bone and soft tissues, and also within the deep deltoid ligament fibers. Displacement of the posterior tibialis and flexor digitorum tendons was present, and there was irregularity of the medial talar dome and overlying articular cartilage suggesting chondromalacia and osteochondral injury. No synovitis was appreciated within the ankle joint.

The patient was taken to the operating room, where the...
Lesion was exposed through a medial malleolar osteotomy distal to the physeal plate (Figure 4A). Two large osteocartilaginous lesions were removed from the talus and ankle joint (Figure 4B). Of note was the presence of a small osteochondritis lesion (5x5 mm) of the talar dome that was subsequently curetted to stable cartilage and microfractured with a drill bit. Pathology specimen was read as osteochondroma. Immediate (Figure 5A) and 16-month postoperative (Figure 5B) AP views of the ankle after smooth pin removal are shown. The patient was pain free with all chosen activities at final follow-up.

**Discussion**

Dysplasia epiphysealis hemimelica (DEH), also known as Trevor disease, is a rare disorder of unknown etiology with an estimated incidence of 1:1,000,000.1-4 There is a male:female ratio of 3:1.2,5 Most cases occur in the first decade of life.2 The ankle joint is most often affected, followed by the knee, with 54% of cases affecting the ankle joint.1,3 Fairbank originally described the condition as cartilaginous overgrowth of a long bone epiphysis that occurs on one side of an affected limb, usually medial.1 Dysplasia epiphysealis hemimelica was later classified by Azouz as having 3 distinct presentations: localized (1 epiphysis involved), classic (more than 1 epiphysis involved in the same extremity), and generalized (the entire limb affected).1,6 Patients most commonly present with...
a painless limp and a localized mass that may increase in size, with or without adjacent joint deformity and stiffness.

Radiographs usually demonstrate a partially ossified, lobulated, cartilaginous mass arising unilaterally from the affected epiphysis with or without an osseous connection. The diagnosis is usually made with radiographs, but the lesion can be mistaken for an intra-articular loose body, osteochondromatosis, or synovial chondromatosis. Computed tomography and MRI can confirm the diagnosis, aid surgical planning, and define the relationship of the mass to local structures. Specifically, CT can help define the anatomic relationship between the mass and the host bone, and MRI can show the extent of epiphyseal involvement and joint deformity and the status of the articular surface. There may be a cleavage plane demonstrated between the lesion and host bone that gradually ossifies as the lesion matures. The lesion in this case had a portion confluent with the medial talus, whereas another portion had a cleavage plane with the adjacent talus.

Treatment for DEH depends on the status of the affected joint. Conservative management can be successful in early lesions, but surgical excision is recommended if progressive size, joint deformity, or pain occur. Complete resection of all pathologic tissue is essential because any remaining tumor will continue to grow. When present in the ankle, DEH is best treated aggressively with surgical excision at a time when surgical treatment will be easier and young joint cartilage less damaged, with best results occurring when joint deformity and persistent synovitis are not present. Hemi-articular joint involvement, a hallmark finding in DEH, especially in a growing child, may lead to joint malalignment, specifically angular deformity, which may cause progressive symptomatology. Mild growth disturbance and osteochondral injury of the medial talus was demonstrated in the current patient with pain, suggesting a need for treatment. Bakerman et al suggested yearly MRI follow-up after excision to monitor for recurrence.

The etiology of DEH is currently unknown, although it has been hypothesized that it is due to a defect in the regulation of cartilage proliferation. The underlying pathology of DEH resembles an osteochondroma. Lesions are benign with normal cancellous bone and varying degrees of endochondral ossification and a hyaline cartilage cap. No malignant degeneration has been reported to date.

**CONCLUSION**

Dysplasia epiphysealis hemimelica is a rare disorder that, although benign in nature, can be locally aggressive, especially when affecting the ankle joint. Aggressive surgical treatment with complete lesion resection is recommended before irreversible joint damage or deformity occurs.