Dysplasia epiphysealis hemimelica of the ankle joint: a case report

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ABSTRACT

Dysplasia epiphysealis hemimelica is a rare non-hereditary epiphyseal disease that mimics synovial chondromatosis of the joints. The disease mainly targets long bones of the lower extremities and tarsal bones. We report one such case manifesting in the lateral tibia of a 13-year-old boy.

CASE REPORT

In February 2010, a 13-year-old boy presented with a 7-month history of pain and a bony mass (3x4 cm) at the right ankle. Pain manifested after playing football or walking a long distance. The mass restricted ankle dorsiflexion over the lateral malleolar area. It was ossific with a lobulated appearance and reached the marrow with the underlying epiphysis and adjacent bones. The mass was excised completely from the epiphysis. The detached syndesmos was sutured anatomically.

Key words: bone diseases, developmental; chondromatosis, synovial; dysplasia epiphysealis hemimelica

INTRODUCTION

Dysplasia epiphysealis hemimelica (DEH), also known as Trevor disease, is a rare non-hereditary epiphyseal disease of unknown aetiology that mimics synovial chondromatosis of the joints. Its prevalence is estimated to be one per million inhabitants and has no racial predilection. The disease mainly targets long bones of the lower extremities and tarsal bones. DEH most commonly manifests in the medial half of the epiphysis of children and adolescents, and has a male-to-female ratio of 3:1. We report one such case manifesting in the lateral tibia of a 13-year-old boy.
marrow with the underlying epiphysis and adjacent bones mimicking synovial chondromatosis in the joint (Figs. 1–2).

Surgery was indicated as the ankle was swollen and the patient had pain during physical activity.

A skin incision over the mass was made under general anaesthesia with a tourniquet applied. The anteroinferior syndesmotic ligament was detached. The mass originated from the epiphysis and connected to the fibula, which was pushed posteriorly. Osteochondral structures loosely connected to each other and looked like synovial chondromatosis (Fig. 3). The mass was excised completely from the epiphysis (Fig. 4). The detached anteroinferior tibiofibular ligament was sutured anatomically. At the 9-month follow-up, the patient had returned to his previous level of activity. The ankle range of motion was normal without pain and disability.

**DISCUSSION**

Hemimelical involvement and connection with the epiphysis are important features of DEH. Magnetic resonance imaging and computed tomography are more useful than radiography in identifying

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**Figure 1** Radiographs showing irregularities on the lateral tibia and the medial fibula and small chondromatous masses on the anterior aspect of the ankle joint (arrows).

**Figure 2** (a) Magnetic resonance imaging and (b) computed tomography showing irregular bone particles (confluent ossific mass) on the lateral distal tibial epiphysis (arrows).

**Figure 3** (a) Cartilage caps over the bones are seen. (b) After excision of the mass, the ankle joint (arrow), the distal tibiofibular corner (arrowhead), and the epiphysis (asterisk) are revealed.
such epiphyseal involvement. The usual symptom is the presence of gradually increasing osseous protuberance on one side of the knee, ankle, or other joints of the foot. The mass is painless and knobby. Radiologically, it typically appears as a non-uniform bone growth, unconnected to ossification centres and related to the epiphysis. DEH is usually mistaken for synovial chondromatosis, which is a geriatric disorder. A patient with DEH was misdiagnosed as having post-traumatic osseous fragments in the joint and was followed up for 10 years, because of a trauma history. Upper extremity involvement is rare.

Conservative treatment is recommended when there is no pain or deformity. Surgery should be considered when the lesion causes pain, loss of function, limitation of movement, and deformity. Surgery should be aggressive and early to avoid joint deformity and stiffening. Nonetheless, treatment should be customised, as patient expectations (cosmetic and wish to play sports) are important. DEH is benign and its prognosis is favourable; no malignant transformation has been reported. Excision of articular lesions is not recommended unless the articular cartilage is compromised. If resection affects the articular cartilage, as in osteochondritis dissecans, homologous osteochondral grafting should be performed, although typically this is not very successful. Correctional osteotomies may be needed in future. In our patient, surgery was performed because the lesion was very close to the loose bodies in the ankle joint.

Histopathologically, it was not possible to distinguish DEH from osteochondroma. With special molecular tests (EXT1, EXT2), genetic expressions can be analysed. Gene expressions are in normal ranges in DEH, whereas they are lower in osteochondroma (owing to a mutation). These tests are costly, hence clinical and radiological findings are important diagnostic tools.

REFERENCES