

Dysplasia epiphysealis hemimelica (Trevor disease) in the ankle

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Abstract: Dysplasia epiphysealis hemimelica, also termed Trevor disease, is a rare disorder that, although benign in nature, can be locally aggressive, particularly when affecting the ankle joint, which is the joint most frequently affected, followed by the knee. The female:male ratio is 1:3, and it is generally diagnosed between 2 and 14 years of age. Surgical treatment with complete resection is recommended before irreversible joint damage and deformity occurs. We presented a case in which dysplasia epiphysealis hemimelica was diagnosed on the medial aspect of a right ankle joint.

Keywords: dysplasia epiphysealis hemimelica, Trevor disease, ankle, osteochondroma, epiphyseal hypertrophy

Introduction

Dysplasia epiphysealis hemimelica (DEH), also known as Trevor disease, is a rare disorder with unknown etiology. Its estimated incidence is 1:1,000,000.¹⁻⁴ The ankle joint is most frequently affected, followed by the knee. The female:male ratio is 1:3, and DEH is generally diagnosed between 2 and 14 years of age.¹⁻⁴

DEH was first described by Mouchet and Belot⁵ as “tarsomegalie” in 1926. In 1950, Trevor⁶ reported a case series, including ten patients and proposed the term “tarso-epiphysal aklasis”. In 1956, Fairbank¹ reported a case series of 14 patients and used the term “dysplasia epiphysealis hemimelica”. DEH is considered a benign, asymmetrical, intra-articular mass lesion confined by epiphysis, resulting from an abnormal proliferation of cartilage tissue.^{3,4} We presented a case in which DEH was diagnosed at the medial aspect of the right ankle joint.

Case report

A 9-year-old girl was presented with swelling of the right ankle and pain during walking, for >3 months. The patient had no history of trauma, and there was pain during palpation at the medial aspect of the ankle. An osteocartilaginous mass (2.5 cm × 2.0 cm × 2.0 cm in size), localized at the distal end of the tibia and extending from the vicinity of the medial malleolus to the medial aspect of the talus, was observed using plain radiograph and computerized tomography (CT) (Figure 1).

During surgical intervention, a medial incision at the medial aspect of the medial malleolus was made, and subcutaneous tissues were explored. The lesion, of osteocartilaginous appearance and originating from the distal tibial epiphysis at medial malleolus level and extending to the medial aspect of the talus, was exposed, which destructed the medial cortex of the talus. Four large osteocartilaginous lesions were removed from the talus and ankle joint (Figure 2). Histological examination

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Figure 1 Ankle images of the patient.

Notes: Localized, irregular osseous mass at the level of anteromedial talus and medial malleolus on AP and lateral ankle radiographs (**A** and **B**). Lobulated osteocartilaginous mass appearance in the vicinity of the medial malleolus at the medial aspect of the distal tibia, right side on the axial section of CT (**C**).

Abbreviations: AP, anterior–posterior; CT, computerized tomography.

revealed that the lesion was mostly hyperplastic cartilage with little osseous tissue, and these features were identical to that of osteochondroma (Figure 3). However, the patient was diagnosed with DEH when the radiological and gross appearance of the lesion was compared with similar reports in the literature.^{1–9}

During the 14-month follow-up, the patient returned to normal activities. Range of motion at the ankle was normal without pain or disability. This study was conducted in accordance with the ethical guidelines of the Declaration of Helsinki. The patient provided written informed consent before participation. The study protocol was approved by the Kayseri Training and Research Hospital, Kayseri, Turkey.

Discussion

DEH is a rare, osseous developmental disorder manifesting with epiphyseal involvement.^{3,9} The localized form of the disease generally involves bones in the ankle and hindfoot.

DEH is benign and its prognosis is favorable; no malignant transformation has been reported.^{4,7–9} The etiology of DEH is uncertain. Potential causes include a congenital error affecting the limb bud during early fetal life or the presence of abnormal chondrocytes that continue to proliferate in an unregulated manner.⁹

In Trevor disease, histopathological diagnosis is not pathognomonic.³ However, *EXT1* and *EXT2* gene expressions can be studied by additional molecular assays.⁹ Gene expressions are within 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.⁷ CT and magnetic resonance imaging can confirm the diagnosis, aid surgical planning, and define the relationship of the mass to local structures.^{3,4,9,10} Specifically, CT can help define the anatomical relationship between the mass and the host bone, and magnetic resonance imaging can show the extent of epiphyseal involvement, joint deformity,

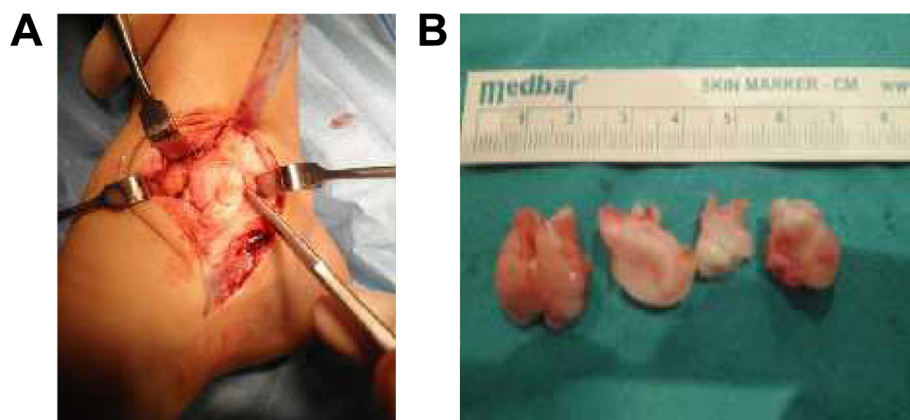


Figure 2 Intraoperative view of patient.

Notes: Intraoperative appearance of osteocartilaginous lesion at medial malleolus and anteromedial aspect of talus in the ankle (**A**). View of the excised osteocartilaginous lesion (**B**).

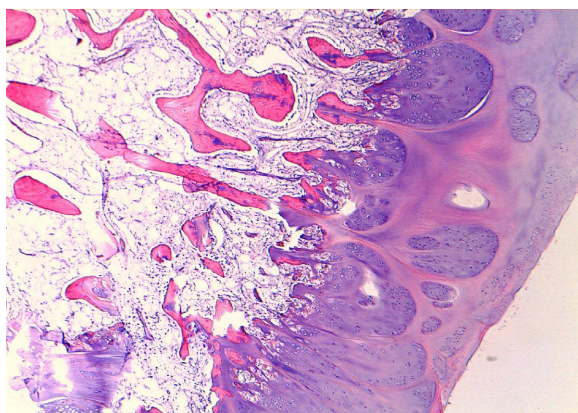


Figure 3 Histopathological findings in Trevor disease.

Notes: Superficial hyaline cartilage layer, columnar chondrocytes (right side), and mature bone trabeculae containing osseous spicules and bone marrow (left side) (hematoxylin–eosin stain, magnification 400×).

and the status of the articular surface.^{1,3,4,9} Radiographs usually demonstrate a partially ossified, lobulated, cartilaginous mass arising unilaterally from the affected epiphysis with or without an osseous connection.^{3,4,9,10}

DEH was classified by Azouz et al¹¹ as having three distinct presentations: localized, classic, and generalized. In Trevor disease, histopathological findings are similar to those in benign osteochondroma,^{3,4,9,10} and it is also difficult to discriminate DEH from benign osteochondroma clinically and pathologically.^{4,10} Both diseases are specific to the developmental period. Thus, radiological findings are important in differential diagnosis. Irregular bone growth originating from epiphysis and a distinct ossification focus that can be seen in radiological evaluation are the most important discriminative features of Trevor disease.^{2–4,9,10} In addition to osteochondroma, intra-articular loose body, myositis ossificans, or synovial chondromatosis can be considered in differential diagnosis.^{3,4,9}

Management options for Trevor disease include observation and surgical excision. Asymptomatic lesions may be treated nonoperatively,^{3,4,7,8} but surgery should be considered

when the lesion causes pain, loss of function, limitation of movement, and deformity.^{3,4,7,8}

Conclusion

In conclusion, DEH is an uncommon, benign disease. Although histopathological findings in Trevor disease are similar to those in osteochondroma, it has differential features that can be distinguished clinically and radiologically. However, molecular analysis has to be done for the definitive diagnosis. Early diagnosis and treatment are necessary to prevent articular dysfunction.

Disclosure

The authors report no conflicts of interest in this work.

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