Case Reports and Series

Surgical management of dysplasia epiphysealis hemimelica of the posterior medial ankle using a medial malleolar osteotomy: A case report

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Dysplasia Epiphysealis Hemimelica (DEH) or Trevor’s disease is a rare skeletal condition that forms osteochondroma like lesions in the epiphyses of long bones. This occurs mainly in pediatric patients with a predilection for the lower extremities. The knee and the ankle joint are most commonly affected in a hemimelic distribution meaning that either the medial or the lateral part of the center of ossification is affected. We report a rare case of Dysplasia Epiphysealis Hemimelica in a active pediatric patient involving multiple tarsal bones around the left ankle that was surgically excised after using a medial malleolar osteotomy for exposure. At over four years of follow up, our patient is pain free, has no growth arrest and continues to engage in normal activities.

Introduction

Dysplasia Epiphysealis Hemimelica (DEH) is a developmental bone disease that occurs in the early years of childhood. It is characterized by an abnormal growth of cartilage arising from epiphyses of the long bones. DEH was first described by Mouchet and Belot in 1926, describing the condition as “tarsomegaly”. In 1950 Trevor described 8 cases of “tarsopiphysal aclasis” in which he believed it was a congenital error of epiphysial development, coining the name “Trevor’s disease”. Finally, in 1956 Fairbanks named the condition dysplasia epiphysealis hemimelica. The incidence of DEH has been estimated at 1 in 1,000,000 individuals in the general population.1,5

DEH is usually diagnosed in children between two and eight years old. DEH has been shown to occur three times as often in males as in females with the medial side of the epiphysis being affected twice as often as the lateral side.2,3 The knee, ankle, and parts of the foot are most commonly affected with this lesion rarely occurring in the upper limbs and spine.1,3,5 The cause is unknown and there have been no reports of malignant degeneration. Potential etiologies include hereditary and incidental epiphyseal trauma.2 Clinically these lesions can present as painless masses or swelling of one side of the affected joint. Pain can develop as the lesion gets bigger in the later stages of the disease. Other symptoms that can be seen are decreased range of motion, limb length discrepancy, and limping. CT and MRI are the imaging modalities of choice to identify the extent of joint and epiphyseal involvement of the lesion, as well as surrounding soft tissue injuries.6

Conservative treatments such as rest, bracing, and nonsteroidal anti-inflammatory medications are recommended for painless lesions while surgical resection is recommended for painful lesions that have worsened over time. Open and arthroscopic techniques have been described for the resection of the cartilaginous lesion.1 Recurrence is rare after surgical resection. We report a case of DEH arising from the posterior medial ankle involving two tarsal bones that was surgically excised.

Case Report

A seven-year-old boy with an unremarkable past medical history presented to our institution for a bony prominence on the inner aspect of the left foot that had been increasing in size and causing occasional pain. Radiographs were reviewed with the patient’s parents and they were notified of the diagnosis of Trevor’s disease. His parents were instructed to monitor symptoms and recommended surgical removal in the future if it got worse. It was recommended that surgical removal wasn’t indicated at that time given the mild symptoms and possible
recurrence with growth. Over the next year and a half, the patient was seen every sixth months for follow up X-rays and routine checkup.

At nine years of age the patient returned with increased pain with activity and running for the previous 3 months after joining a running club. Foot and ankle radiographs were obtained at this time (Figs. 1 & 2). An MRI was further ordered to assess the extent of intraarticular involvement of the lesions. The MRI demonstrated bony and cartilaginous fragments along the posterior medial talar dome measuring 2.3 cm × 2.1 cm × 1.8 cm (Figs. 3 & 4). Additional findings included a lesion at the medial navicular bone measuring 2.2 cm × 1.4 cm, tibiotalar joint effusion with synovitis, and tenosynovitis along the flexor digitorum longus tendon. At this time, the patient was placed in a cast for two weeks to rest the joint. Upon return, although there was an improvement in the pain, it was still present. Due to failure of conservative care and continued pain, a recommendation was made for left medial malleolar osteotomy, ankle arthrotomy, tenosynovectomy and resection of the large talus intraarticular cartilaginous lesion. The lesion at the medial aspect of the navicular that was noted on radiographs and MRI was asymptomatic and therefore treated non-operatively at the time.

Operative Technique

Surgery was performed under general anesthesia in the supine position with a thigh tourniquet inflated to 250 mmHg. Attention was first directed to the medial ankle. The C-arm was brought in and the level of the distal tibial physis was marked on the skin. An anteromedial ankle incision was then made, and sharp dissection was carried down to the fascia. The fascia was then incised, and the medial malleolus was exposed. The medial malleolus was predrilled in anticipation of screw fixation. A medial malleolar osteotomy was performed, and the medial malleolus was then flipped down to expose the ankle joint. The very large posteromedial intra-articular lesion was thus identified. There was extensive ankle synovitis as well as tenosynovitis of the tibialis posterior tendon and the flexor digitorum and flexor hallucis tendons on the posteromedial aspect of the ankle. The inflamed synovium was removed exposing the lesion. The large cartilaginous lesion was removed in fragments and sent to pathology for further evaluation.

Fig. 1. Anterior posterior left foot weightbearing radiograph demonstrating an osseous growth located on the medial navicular.

Fig. 2. a: Anterior posterior left ankle weightbearing radiograph demonstrating an osseous growth located on the medial talus. b: Lateral left ankle weightbearing radiograph demonstrating an osseous growth located on the talus expanding posteriorly.
After a copious irrigation, no evidence of additional intra-articular loose fragments was found. The ankle joint was shown to be stable at this time. The medial malleolus osteotomy was then repaired using two 4.5 mm cannulated screws (Fig. 5). This provided stable fixation. After fixation, no restriction to ankle movement in dorsiflexion nor plantarflexion was seen and the ankle was stable in the coronal plane. Closure was performed in a layered fashion. The patient was placed into a short leg cast.

Histopathological exam revealed DEH of the tarsal bones, showing the normal bone surrounded by a cartilage cap covered by perichondrium. Endochondral ossification was demonstrated at the bone-cartilage interface (Fig. 6). Following surgery, the patient was non-weightbearing for 4 weeks in a cast. Then the patient was transitioned to a CAM boot and underwent a course of physical therapy for 6 weeks.

At three months post-operative, the patient was pain-free and returned to normal activity as tolerated. At ten months post-operative, both the hardware used for fixation of the medial malleolus as well as the navicular lesion were removed due to pain. At the final follow up of over 4 years post-operative, the patient has continued pain-free normal activities and normal growth plates (Fig. 7).

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**Fig. 3.** Axial T1 weighted image demonstrating a mixed bony and cartilaginous lesion along the posterior medial talar dome extending into the surrounding soft tissues.

**Fig. 4.** Coronal T2 weighted image demonstrating an increased signal intensity along the medial ankle with the bony cartilaginous lesion extending into the ankle joint.

**Fig. 5.** Post-operative anterior posterior projection of left ankle demonstrating fixation of medial malleolar osteotomy and clearance of cartilaginous and osseous lesions.

**Fig. 6.** a: Low power microscopic appearance demonstrating normal bone with a rounded, thick cartilage cap, covered by a thin layer of perichondrium (magnification x 40, hematoxylin-eosin, [H&E] stain). b: High power microscopic appearance demonstrating endochondral ossification at the interface between bone and cartilage cap; the cartilage cap is composed of disorganized, benign chondrocytes (magnification x 100, H&E stain).
DEH is a benign developmental condition that produces cartilaginous lesions commonly in the lower extremity. Although different theories like inheritance have been proposed, the etiology of DEH remains unknown. There has been only one report in the literature regarding two generations of family members having DEH. Even with the incidence reported as 1:1,000,000, some believe it is more common. In 2005 Bhsoale et al. reported the incidence as 1 in 106 patients and stated that it is underreported due to improper diagnosis.

DEH usually presents in children aged 2–8 years old as a mass that may or may not be accompanied by pain. These masses are three times more likely to occur in males than females. The most affected bones are distal tibia and fibula (22%), talus and calcaneus (22%), distal femur (21%), proximal tibia (11%), navicular, cuboid, cuneiforms (10%), scaphoid (2%) and scapula (1%). These masses are usually unilateral with a predilection for the medial aspect of the ankle joint. Pain may occur due to irritation of soft tissue from shoe wear from underlying mass or due to the osseous mass affecting the joint. Osseous masses affecting the joint can cause not only pain but also gait abnormalities, limb length discrepancies, and angular deformities.

Multiple classification systems have been described for DEH. Most commonly, the system by Azouz broke down DEH into 3 distinct groups: localized, classic and generalized. Localized affects only one epiphysis, classic involves more than one epiphysis in the same limb and generalized affects the entire lower extremity. Recently another classification was described by Clark et al 2016 which is based on whether the lesion was described by Clark et al 2016 which is based on whether the lesion is intra-articular or extra-articular. Clark argues that this is the single most important factor affecting long-term morbidity and therefore should be the basis of the classification system.

Standard radiographs are often the first step in management following a thorough physical exam. The radiographic findings are characteristic. In early stages it presents as an irregular lesion rising from the metaphysis, and under a microscope the cartilage contains bands of cartilage separating area of cancellous bone which are not present with osteochondromas.

If nonoperative treatment is chosen, close follow up is necessary to evaluate the progression of the lesion. Consequences of non-operative treatment include early osteoarthritis of the ankle and ossification of the hypertrophic cartilaginous areas resulting in limb length discrepancy or angular deformities. Surgical intervention for the treatment of DEH is indicated when the lesion causes pain, loss of function, and deformity. The goal of surgical intervention is to fully remove the lesion without injuring the epiphysis. Following adequate surgical excision, the majority of patients regain full ROM and have equal limb length. The most common complications include muscle wasting and osteoarthritis of the involved joint.

This case was unique in that DEH involved multiple tarsal bones and had other adjacent soft tissue findings that were further discovered after using a medial malleolar osteotomy for exposure. Surgeons must be careful when placing screws across a growth plate in children, as it can potentially lead to growth arrest. After trying conservative treatment for two years, our patient opted for surgical intervention. At the current time of over 4 years following surgery, our patient continues to be without pain, recurrence, growth arrest or activity restrictions.

In conclusion, DEH is a rare condition that affects mainly pediatric patients with predilection for the lower extremity. A thorough clinical workup with appropriate imaging is key to the diagnosis. Although conservative care may be successful in patients, surgical intervention is often warranted to prevent premature arthrosis of the foot and ankle, limb length discrepancy, angular deformities and or gait abnormalities.

Patient Informed Consent Statement

The authors declare that informed patient consent was taken from all the patients.

Declaration of Competing Interests

The authors declare no conflicts of interest.

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