Skeletal Radiol (2017) 46:111–115
DOI 10.1007/s00256-016-2491-7

CASE REPORT

Dysplasia epiphysealis hemimelica of the lower limb

Ruken Yuksekkaya Celıkyay[1] · Fatih Celikyay[1] · Erkal Bilgic[2] · Murat Ascı[2] · Dogan Koseoglu[3]

Received: 10 April 2016 / Revised: 27 August 2016 / Accepted: 19 September 2016 / Published online: 14 October 2016
© ISS 2016

Abstract Dysplasia epiphysealis hemimelica (DEH) is a rare developmental disorder characterized by osteocartilaginous overgrowth in one or more epiphyses. The disease usually involves a single limb or is hemimelic (lateral or medial compartment), and lower extremities are more frequently affected than upper extremities. Here we present clinical and radiological findings for a male DEH patient at 1.5 and 3.5 years of age. The radiographs obtained at first presentation showed minimal osseous overgrowth and irregularity at the epiphyses around the left knee and ankle joints, respectively. Radiographs obtained at the second presentation showed osteocartilaginous masses at most epiphyses of the left lower extremity. Two months after diagnosis (at 3.7 years old), the patient had surgery on his left knee to relieve increased joint restriction. The histopathological diagnosis was consistent with an osteocartilaginous lesion. This case report presents imaging features and age-related progression of DEH in this patient.

Keywords Dysplasia epiphysealis hemimelica · Trevor-Fairbank disease · Radiography · Magnetic resonance imaging

✉ Ruken Yuksekkaya Celıkyay
rukenyuksekkaya@yahoo.com

✉ Fatih Celikyay
fatihcelikyay99@yahoo.com

Erkal Bilgic
erkal68@hotmail.com

Murat Ascı
muratasci55@yahoo.com

Dogan Koseoglu
residdogan@hotmail.com

[1] School of Medicine, Radiology Department, Gaziosmanpasa University, Tokat, Turkey

[2] School of Medicine, Orthopedics and Traumatology Department, Gaziosmanpasa University, Tokat, Turkey

[3] School of Medicine, Pathology Department, Gaziosmanpasa University, Tokat, Turkey

Introduction

Dysplasia epiphysealis hemimelica (DEH), also known as Trevor Fairbank disease, is a rare developmental skeletal dysplasia characterized by asymmetric osteochondral overgrowth of epiphyseal cartilage. DEH has an approximate incidence of one in one million and predominates in males (3:1 males:females) [1]. DEH can affect either a single joint or multiple joints. The involvement is usually in a single limb or hemimelic (lateral or medial compartment). Radiographically, DEH is characterized by asymmetrical overgrowth of epiphyses with irregular ossifications. Imaging studies play an important role in DEH diagnosis due to its characteristic appearance on radiographs [2]. This case report presents the imaging features and age-related progression of a male DEH patient diagnosed at 3.5 years of age. Also the report highlights that, although DEH diagnosis is usually straightforward in terms of having a typical appearance, location, and distribution, the findings in the early stages can be subtle.

Case report

A 3.5-year-old male presented to our institution with a 2-year history of leg curvature, gait disturbance, and swelling on his left leg. According to his medical history, 2 years earlier he was

treated with vitamin D for a diagnosis of rickets that was based on low serum levels of 25 OH(D). Radiographs obtained at that time showed a genu varum deformity and minimal osseous overgrowth and irregularity at the epiphyses around the left knee and ankle joints, respectively (Fig. 1). However, there were no radiological findings of rickets such as growth plate widening and metaphyseal flaring. At the first presentation, CT or MR examinations were not performed. At the second presentation, a physical examination revealed a prominent genu varum deformity and a mass in the left knee. The latest serum biochemistry values were normal. The last radiographs demonstrated apparent asymmetrical mass-like osseous hypertrophy in the epiphyses of the left hip, knee, and ankle. The findings were especially pronounced in the lateral femoral epiphysis, which had irregular exophytic ossification (Fig. 2). The epiphyses of the other extremities (right and left upper) were normal. The CT and MR imaging examinations were performed for a more detailed evaluation and for treatment planning. The patient did not receive anesthesia or sedation for these examinations. A CT exam of the left knee clearly demonstrated osseous hypertrophy on the lateral side of the femoral and tibial epiphyses (Fig. 3). MR imaging of the knee showed asymmetrical osteocartilaginous masses and continuity with epiphyses of the distal femur and proximal tibia (Fig. 4). The cartilaginous parts of the lesions had intermediate signal intensity on a fat saturated proton-density sequence, amd hypointense signal intensity on a T1-weighted sequence. The radiological diagnosis was consistent with DEH. The patient underwent surgery to relieve increased joint restriction and deformity. The femoral osteocartilaginous mass partially was resected to avoid of native cartilage damage. Histologic examination of the resected lesion chondrocyte showed clusters in deep areas of hyaline cartilage and fissure formations on articular cartilage (Fig. 5). The 6-month-follow-up radiograph of the knee exhibited increased size of the osteocartilaginous masses that was consistent with progression (Fig. 6.)

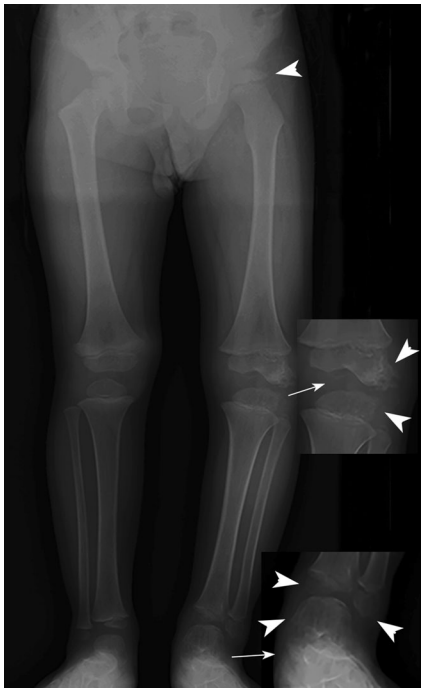


Fig. 2 Radiograph obtained when the patient was 3.5 years old showed osseous masses that were continuous with the epiphyses at the left femur, tibia, distal fibula and medial part of the talus (*arrows*)



Fig. 1 Radiograph obtained when the patient was 1.5 years old showed minimal osseous overgrowth and irregularities at the epiphyses around the left knee and ankle joints (*arrowheads*). The left capital femoral epiphyses was irregular and smaller than the right femoral epiphyses (*short arrow*)

Discussion

Dysplasia epiphysealis hemimelica, which is also called Trevor-Fairbank disease, was first reported in 1926 by Mouchet and Belot [3], who described the condition as tarsomegalie based on its location [4]. In 1950, Trevor reported eight cases of the disease and named it tarso-epiphyseal aclasis. In 1956 Fairbank used the term dysplasia epiphysealis hemimelica (DEH) to describe this skeletal disorder because the disease also involves other joints and only half rather than the entire epiphysis [5, 6].

DEH results in asymmetric osteocartilaginous overgrowth of the epiphysis, and presents as skeletal deformity, pain, and restricted joint movement. DEH is considered to be a non-hereditary disorder. Although the etiology of DEH remains unclear, it may be associated with a congenital defect that affects the early stages of limb development during the fetal period or while chondrocytes proliferate without regulation [1]. Malignant transformation of DEH has not been reported. Lower extremity bones such as the femur, tibia, fibula, and

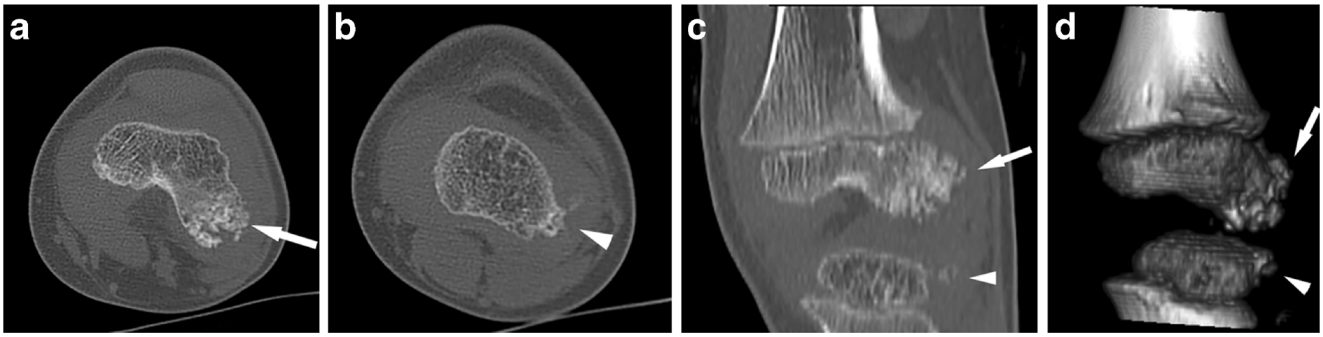


Fig. 3 Axial (**a**–**b**) and oblique MPR (**c**) with 3-D volume rendering images (**d**) of the left knee showing a osseous mass with irregular ossification at the lateral aspect of the femoral (*arrows*) and tibial (*arrowheads*) epiphyses when the patient was 3.5 years old

tarsus are the most frequently affected by DEH, although involvement of other bones such as the pelvis, carpus, and phalanges has also been reported [7, 8]. The involvement is usually unilateral and hemimelic (lateral or medial compartment). Bilateral involvement or cases where entire epiphyses are affected have rarely been reported [9–11] With single joint involvement, the tarsal or knee joint is most typically affected.

DEH symptoms frequently emerge during childhood, and often present as a hard painless mass adjacent to the affected joint, with deformities developing at a later stage [1]. Extremity length may also be affected due to overgrowth or premature closure of the epiphysis. Radiographically, DEH is characterized as a partially calcified-ossified, lobulated mass that involves the epiphysis with or without an osseous attachment [12–14]. Usually only one side of the epiphysis is involved, with the medial side being more frequently affected [15]. In our case, the patient had involvement of the lateral compartment of the hip and knee joint, while the medial compartment of the tibia and talus was affected along with the lateral side of the fibula in the ankle joint. The mass showed stippled, irregular, or dense calcification [1]. CT is useful for defining the relationship of the mass to local structures such as host bone and joint. Because CT has strong sensitivity for identifying chondroid calcification in the mass, unossified cartilaginous components can be underestimated [1]. As such, MRI is the best approach for evaluating unossified cartilage masses, epiphyseal involvement, and complications such as joint degeneration, bursitis, and neurovascular compression [1, 16]. The signal intensity of the cartilaginous regions of the lesion may be similar to that of the normal epiphyseal cartilage or have slightly higher signal intensity on T2-weighted sequences due to disorganized collagen fibers and a somewhat increased proteoglycan content in the lesion [1, 16, 17]. Some cases can show a cleavage plane between the cartilage of the unossified mass and the normal epiphyseal cartilage [2], although a clear cleavage plane was not seen for this case. This condition can be associated with disease progression after resection.

DEH shares similar histological features with osteochondromas such as normal looking chondrocytes lying in a cartilaginous matrix, which occasionally appears as cellular foci with mild to moderate atypia. However, osteochondroma is a benign osteocartilaginous tumor that arises from the metaphyses and is due to a mutation in the EXT gene family [18]. Unlike osteochondromas, genetic studies have not identified EXT1 and EXT2 gene mutations in DEH patients [19]. Moreover, chondrocyte clumping in a fibrillary matrix has been shown and only focal

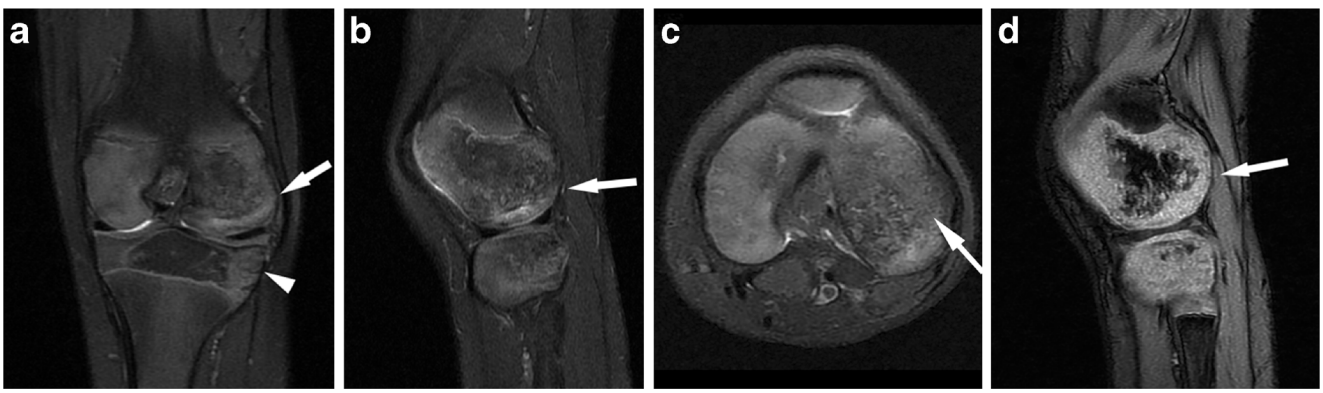


Fig. 4 Coronal (**a**), sagittal (**b**) and axial fat-sat PD (**c**) weighted and sagittal GRE (**d**) images demonstrate osteocartilaginous masses at the left distal femur (*arrows*) and proximal tibia (*arrowhead*) epiphyses, which have signal features that were similar to the unaffected regions of the epiphysis. It was not seen a clear cleavage plane between normal epiphyseal cartilage and osteocartilaginous lesions

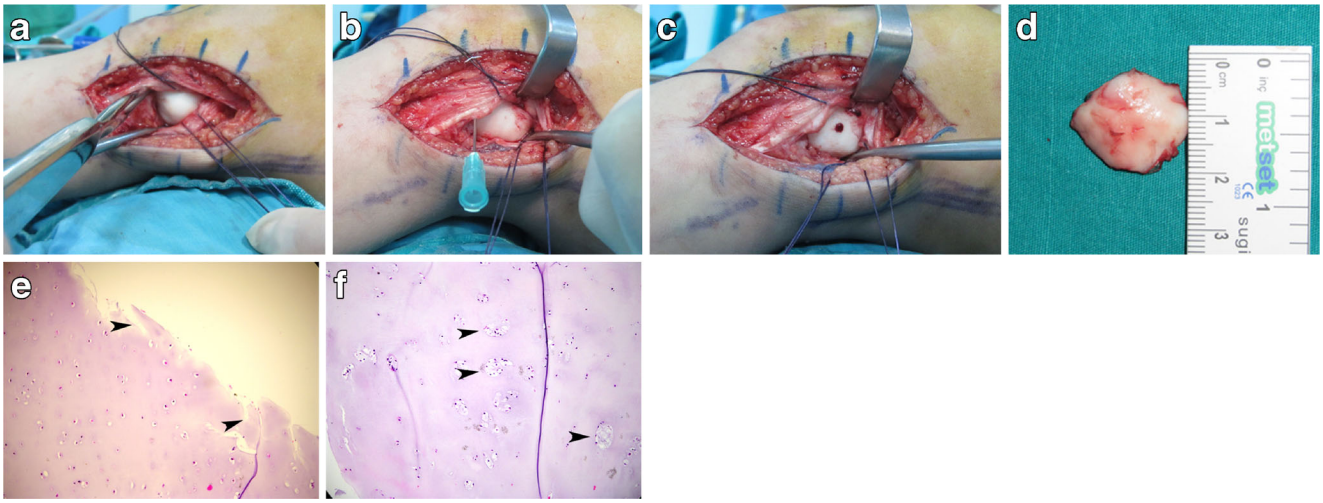


Fig. 5 Surgical and histological findings of the lesion resected from the left femur distal epiphysis. Intraoperative findings showed a cartilaginous mass of the knee epiphysis (**a**–**c**). The excised lesion was 1.8 cm × 2 cm and was white and firm (**d**). Degenerative changes were characterized by fissure formations (*arrowheads*) on articular cartilage (**e**) and chondrocyte clusters (*arrowheads*) in deep areas of the hyaline cartilage (**f**) (HE, 10×)

endochondral ossifications can be distinguishing features of DEH [1, 19, 20]; these histopathological findings were not seen in our case.

Although there is currently little consensus about DEH treatment, clinical symptoms usually dictate therapeutic approaches. DEH can be conservatively managed during the early disease stages and when there are fewer symptomatic lesions, but surgical treatment is recommended for progressive lesion size, joint deformity, or pain [1, 2, 20]. Surgical management options include correction osteotomies, surgical excision, arthrodesis, or chondroplasty [2]. Excision of an intraarticular lesion does carry the risk of damage to native cartilage and early osteoarthritis, but incomplete excision can result in disease recurrence [13, 14]. Prognosis depends on the location and size of the lesion, the degree of involved articular surfaces.

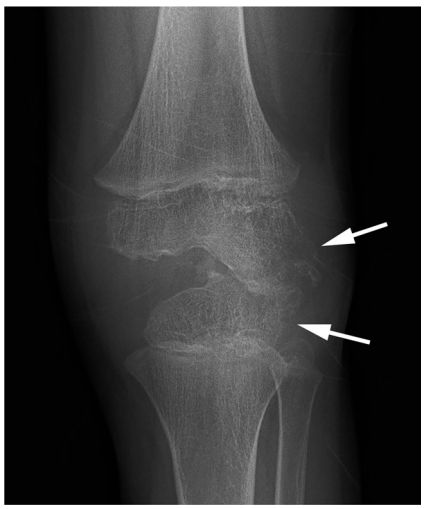


Fig. 6 At the 6 month post-op, there was increase in the size of osseous lesions at the distal femur and proximal tibia (*arrows*)

Epiphyseal cartilage overgrowth can occur during infancy or early childhood; however, diagnosis at these young ages may be difficult due to the smaller sizes of the epiphyses and insufficient ossification. Indeed, in our case, the radiographic findings when the patient was 1.5 years-old were not obvious because of insufficient ossification. MR imaging may be helpful at early ages and early stages of DEH due to superior soft tissue resolution afforded by this method.

Compliance with ethical standards

Conflict of interest The authors declare that there is no conflict of interest regarding the publication of this paper.

References

1. Tyler PA, Rajeswaran G, Saifuddin A. Imaging of dysplasia epiphysealis hemimelica (Trevor's disease). Clin Radiol. 2013;68(4):415–21.
2. Bosch C, Assi C, Louahem D, Alkar F, Mazeau P, Delfour C, et al. Diagnosis and surgical treatment of dysplasia epiphysealis hemimelica: a report of nine cases. Orthop Traumatol Surg Res. 2014;100(8):941–6.
3. Mouchet ABJ. Tarso megalie. J Radiol Electrol. 1926;10:289–93.
4. Trevor D. Tarso-epiphyseal aclasis: a congenital error of epiphysial development. J Bone Joint Surg (Br). 1950;32-B(2):204–13.
5. Fairbank TJ. Dysplasia epiphysialis hemimelica (tarso-ephiphysial aclasis). J Bone Joint Surg (Br). 1956;38-B(1):237–57.
6. De Smet L. Dysplasia epiphysealis hemimelica of the hand: two cases at the proximal interphalangeal joint. J Pediatr Orthop B. 2004;13(5):323–5.
7. Karam AR, Birjawi GA, Saghieh S, Tawil A, Khoury NJ. Generalized dysplasia epiphysealis hemimelica with contralateral sacro-iliac joint involvement. Skelet Radiol. 2008;37(12):1147–52.
8. Gregory Jr PR, Rooney RJ. Bilateral dysplasia epiphysealis hemimelica: a case report. Foot Ankle. 1993;14(1):35–7.

9. Merzoug V, Wicard P, Dubousset J, Kalifa G. Bilateral dysplasia epiphysealis hemimelica: report of two cases. Pediatr Radiol. 2002;32(6):431–4.
10. Wiart E, Budzik JF, Fron D, Herbaux B, Boutry N. Bilateral dysplasia epiphysealis hemimelica of the talus associated with a lower leg intramuscular cartilaginous mass. Pediatr Radiol. 2012;42(4):503–7.
11. Bahk WJ, Lee HY, Kang YK, Park JM, Chun KA, Chung YG. Dysplasia epiphysealis hemimelica: radiographic and magnetic resonance imaging features and clinical outcome of complete and incomplete resection. Skelet Radiol. 2010;39(1):85–90.
12. Struijs PA, Kerkhoffs GM, Besselaar PP. Treatment of dysplasia epiphysealis hemimelica: a systematic review of published reports and a report of seven patients. J Foot Ankle Surg. 2012;51(5):620–6.
13. Nowicki PD, Borders H. Dysplasia epiphysealis hemimelica (Trevor disease) of the ankle. Orthopedics. 2015;38(4):216. 69–71.
14. Haddad F, Chemali R, Maalouf G. Dysplasia epiphysealis hemimelica with involvement of the hip and spine in a young girl. J Bone Joint Surg (Br). 2008;90(7):952–6.
15. Lang IM, Azouz EM. MRI appearances of dysplasia epiphysealis hemimelica of the knee. Skelet Radiol. 1997;26(4):226–9.
16. Iwasawa T, Aida N, Kobayashi N, Nishimura G. MRI findings of dysplasia epiphysealis hemimelica. Pediatr Radiol. 1996;26(1):65–7.
17. Perl M, Brenner RE, Lippacher S, Nelitz M. Dysplasia epiphysealis hemimelica: a case report with novel pathophysiologic aspects. Clin Orthop Relat Res. 2009;467(9):2472–8.
18. Bovee JV, Hameetman L, Kroon HM, Aigner T, Hogendoorn PC. EXT-related pathways are not involved in the pathogenesis of dysplasia epiphysealis hemimelica and metachondromatosis. J Pathol. 2006;209(3):411–9.
19. Pazzaglia UE, Beluffi G, Marchi A, Bozzola M, Savasta S, Bonaspetti G. Poly-epiphyseal overgrowth: description of a previously unreported skeletal dysplasia. Pediatr Radiol. 2007;37(10):1025–30.
20. Douira-Khomsi W, Louati H, Mormech Y, Saied W, Bouchoucha S, Smida M, et al. Dysplasia epiphysealis hemimelica: a report of four cases. Foot Ankle Surg. 2011;17(1):37–43.