

Concurrent Occurrence of Dysplasia Epiphysealis Hemimelica involving Lateral Malleolus and Sinus Tarsi: Rare Case with Review of Literature

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Learning Point of the Article:

Concurrent involvement of the distal fibular epiphysis, talus, and sinus tarsi is a rare presentation of dysplasia epiphysealis hemimelica; careful clinicoradiological evaluation and complete surgical excision can result in excellent functional outcomes.

Abstract

Introduction: Dysplasia epiphysealis hemimelica (DEH), also known as Trevor's disease, is a rare developmental disorder characterized by asymmetric osteochondral overgrowth arising from the epiphysis. It commonly involves the lower limb, particularly the ankle and knee, and may mimic osteochondroma both clinically and histologically.

Case Report: We report an unusual case of an 8-year-old boy presenting with pain, swelling, and progressive restriction of ankle motion. Imaging revealed osteochondral lesions arising from the distal fibular epiphysis and the inferolateral aspect of the talus extending into the sinus tarsi. Surgical excision of both lesions was performed through an anterolateral approach. Histopathological examination showed features of a benign osteochondral lesion consistent with DEH. At 12-month follow-up, the patient demonstrated near-full ankle range of motion, no recurrence, and satisfactory functional recovery.

Conclusion: Concurrent involvement of the distal fibula, talus, and sinus tarsi represents an unusual presentation of DEH. Comprehensive imaging evaluation and complete surgical excision can provide favorable functional outcomes.

Keywords: Dysplasia epiphysealis hemimelica, Trevor's disease, ankle, sinus tarsi, osteochondral lesion, pediatric.

Introduction

Dysplasia epiphysealis hemimelica (DEH), also known as Trevor's disease, is a rare, non-hereditary developmental disorder of the epiphysis characterized by asymmetric osteochondral proliferation, typically confined to one side of an epiphysis or a comparable structure [1,2,3]. Its prevalence is estimated at approximately 1 in 1,000,000, with a male predominance of about 3:1, and most cases are identified in early childhood, usually before the age of eight, when swelling, pain, or deformity prompts clinical evaluation [4,5,6]. Fairbank

introduced the currently accepted name, DEH, in 1956 [3].

The abnormal cartilage forms an irregular nodular mass located on either the medial or lateral part of the bone, with the medial side more frequently affected [5,6]. DEH may involve a single epiphysis (localized form), multiple epiphyses in one limb (classical form), or an entire limb (generalized form), most often the lower extremity from the pelvis to the foot [5,6]. Approximately two-thirds of affected children present with multiple lesions. Clinically, DEH appears as a nodular bony mass that can cause deformity, restricted motion, or recurrent sprains

Author's Photo Gallery



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Figure 1: Pre-operative clinical photographs showing a visible bony swelling over the anterolateral aspect of the ankle (a and b). Intraoperative image demonstrating exposure of the osteochondral mass arising from the distal fibula and talus within the sinus tarsi region (c).

[6]. Histologically, the lesion resembles an osteochondroma, making radiological correlation critical for diagnosis [4,5,6].

We report an unusual case of DEH involving both the distal fibula and talus, with outgrowth into the sinus tarsi region, highlighting the diagnostic challenges and emphasizing the importance of individualized surgical management [5,7,8].

Case Report

An 8-year-old boy presented to the orthopedic clinic with a 1.5-year history of pain and swelling in the right ankle associated with progressive difficulty in walking. There was no history of trauma, fever, weight loss, or family history of joint or bone disorders.

On physical examination, a firm, non-tender bony swelling was palpable anterior to the lateral malleolus. The overlying skin was normal, and there were no inflammatory signs (Fig. 1). Range of motion at the ankle and subtalar joints was restricted, and gait was limited due to discomfort.

Radiographs (Fig. 2) and computed tomography (Fig. 3) of the right ankle and lower leg showed a well-defined osseous overgrowth arising from the distal fibular epiphysis, projecting anterolaterally, along with an additional irregular bony prominence arising from the inferior surface of the talus in the sinus tarsi region.

Magnetic resonance imaging (MRI) demonstrated lobulated osseous masses arising from the distal fibular epiphysis and



Figure 2: Pre-operative radiographs of the right ankle – Anteroposterior view showing osseous overgrowth from the distal fibular epiphysis (a), Oblique view demonstrating the extent of the lesion (b), Lateral view showing an osteochondral prominence from the talus extending into the sinus tarsi (c).

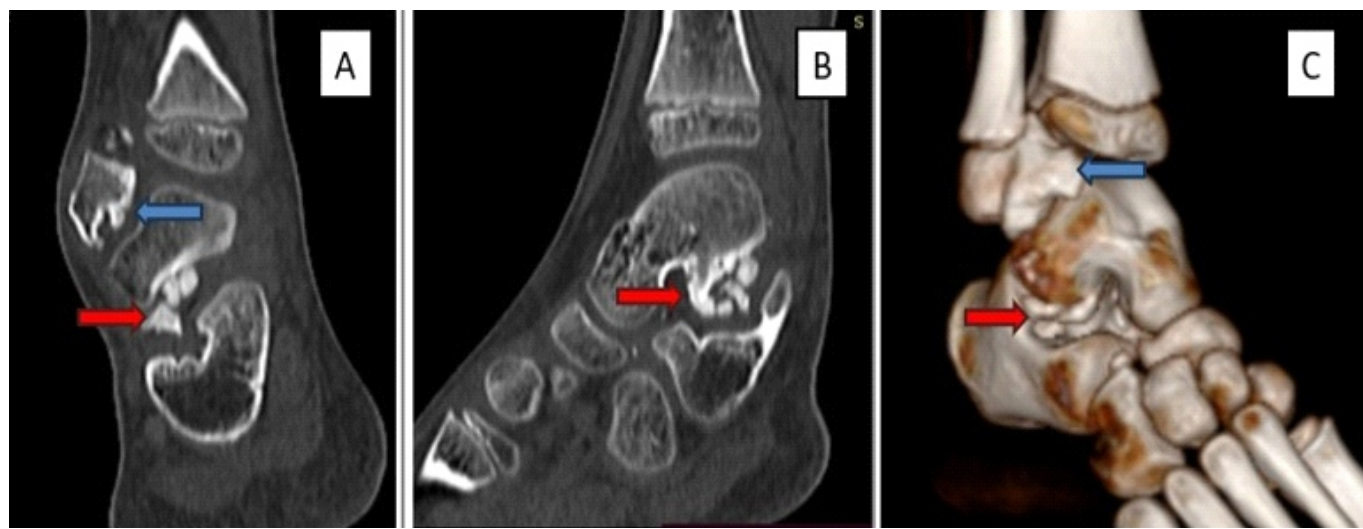


Figure 3: Computed tomography images of the right ankle – coronal (a), sagittal (b), and 3D reconstruction (c) – demonstrating irregular osteochondral overgrowth arising from the distal fibular epiphysis (blue arrow) and the inferolateral aspect of the talus projecting into the sinus tarsi (red arrows).

from the inferolateral aspect of the talus, forming a sessile growth in the sinus tarsi region measuring approximately 1 cm in thickness and <1.5 cm in height and width. Both lesions showed cortical and medullary continuity with their parent bones. There was no cortical destruction, marrow edema, soft-tissue invasion, or joint effusion, although the subtalar joint space was reduced (Fig. 4).

Differential diagnoses included osteochondroma, synovial chondromatosis, intra-articular loose bodies, and epiphyseal dysplasia. The epiphyseal origin of the lesion with intra-articular extension and cortical-medullary continuity on imaging favored the diagnosis of DEH.

The patient was positioned supine under general anesthesia with a pneumatic tourniquet applied to the thigh. Through an

anterolateral approach, both lesions were exposed. Intraoperative fluoroscopy was used to localize the lesions and confirm complete excision. Careful dissection was performed to identify and protect the superficial peroneal nerve. The subtalar joint was inspected intraoperatively, and the articular cartilage was preserved as much as possible while excising the osteochondral masses. Both lesions were excised en bloc, and the raw surfaces were curetted to normal cancellous bone. Histopathological examination (Fig. 5) demonstrated mature cartilage and trabecular bone without cellular atypia. Although histological findings overlapped with osteochondroma, clinicoradiological correlation and gross findings supported the diagnosis of DEH [1,2,3,4,5,6,7,8].

The patient was discharged with a below-knee cast for 2 weeks.

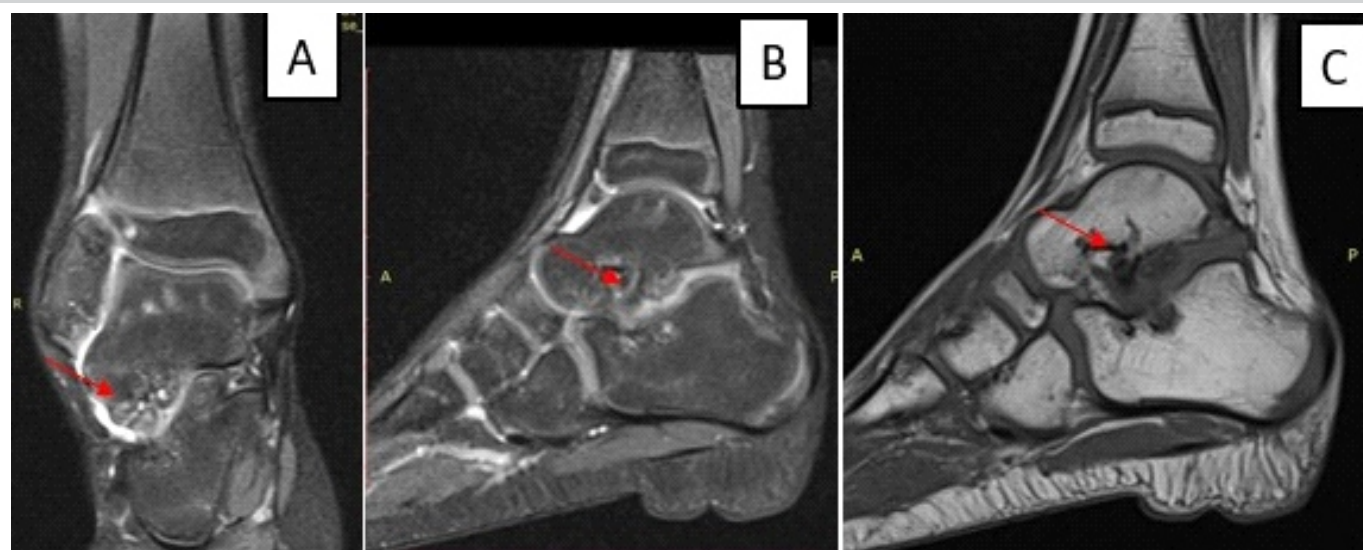


Figure 4: Magnetic resonance imaging of the right ankle. Coronal T2-weighted (a), sagittal T2-weighted (b), and sagittal T1-weighted (c) images showing a lobulated epiphyseal lesion with cartilage signal intensity.

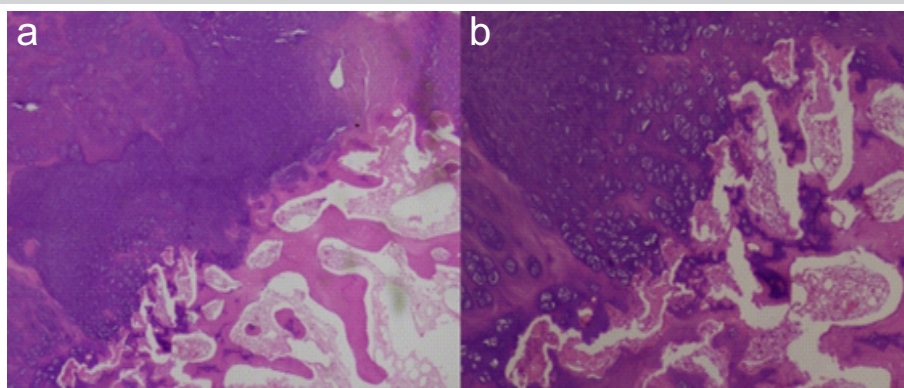


Figure 5: Histopathological examination. (a) Scanner view showing an osteochondral lesion with a cartilaginous cap overlying mature bony trabeculae (Hematoxylin and Eosin [H&E], ×40). (b) Low-power view demonstrating orderly distribution of bland chondrocytes without atypia (H&E, ×100).

Following suture removal, gradual weight-bearing was initiated with an ankle boot walker for 4 weeks. Follow-up at 3, 6, and 12 months showed no recurrence, and the patient remained asymptomatic at the 12-month review. At final follow-up, the patient had resumed normal activities with near-full ankle motion and no pain (Fig. 6). The American Orthopaedic Foot and Ankle Society ankle-hindfoot score was 92/100, with a Visual Analog Scale pain score of 1/10.

No limb-length discrepancy, angular deformity, progression of hindfoot malalignment, or evidence of physeal disturbance was observed during follow-up. Recurrence assessment during follow-up was performed clinically and with serial radiographs; post-operative MRI was not considered necessary because the patient remained asymptomatic with no radiographic evidence of recurrence.

Discussion

DEH is an uncommon developmental disorder of the epiphysis

with heterogeneous clinical presentations. Although the exact etiology remains unclear, proposed mechanisms include abnormalities in epiphyseal cartilage proliferation during skeletal development. The condition predominantly affects the lower limb, with a predilection for the medial side; however, lateral involvement, as observed in the present case, is distinctly uncommon [1,7].

A major diagnostic challenge is distinguishing DEH from osteochondroma. Although both lesions share histological similarities, their anatomical origin is the key distinguishing

factor. Osteochondromas arise from the metaphysis and are extra-articular, whereas DEH originates from the epiphysis and frequently demonstrates intra-articular extension [1,5,9]. Advanced imaging modalities, particularly MRI, are essential for assessing lesion extent, articular involvement, and for pre-operative planning [9]. Azouz et al. classified DEH into three forms: Localized (single epiphyseal involvement), classical (multiple epiphyses in a single limb), and generalized (entire limb involvement) [10]. Based on this classification, the present case can be categorized under the classical form, as it involves multiple epiphyses within the same anatomical region.

A review of previously reported cases involving the ankle and sinus tarsi region (Table 1) demonstrates that most cases present with pain, swelling, and restricted range of motion, with the talus being the most commonly involved site [11,12,13,14,15,16,17,18,19]. Sinus tarsi involvement is relatively rare and has been reported in only a limited number of cases, such as those described by Graves [11] and Baumfeld et

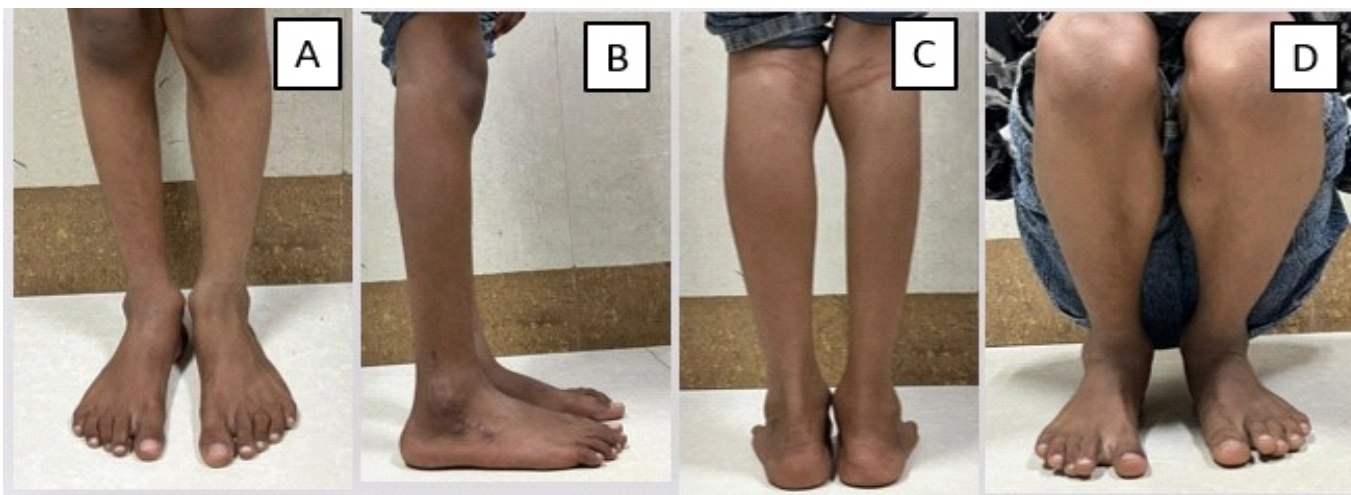


Figure 6: Post-operative clinical photographs of the ankle and foot – anterior (a), lateral (b), posterior (c), and squatting position (d) – demonstrating good functional recovery and maintained hindfoot alignment.

Table 1: Review of the literature on dysplasia epiphysealis hemimelica involving the ankle and sinus tarsi region

No.	Author (year [References])	Age/sex	Site involved	Sinus tarsi involvement	Clinical presentation	Imaging	Treatment	Follow-up	Outcome
1	Graves (1991) [11]	Pediatric	Subtalar joint	Yes	Pain, limited subtalar motion	Radiograph	Excision	18 months	Recurrence reported
2	Bakerman <i>et al.</i> (2005) [12]	Children	Ankle epiphysis	No	Pain, limited dorsiflexion	Radiograph, CT	Surgical excision	1–3 years	Improved ROM
3	Bhosale <i>et al.</i> (2005) [13]	8 F, 12 M	Talus	No	Hard swelling, limited motion	X-ray, CT	One excision; one conservative	12–24 months	Surgical case improved
4	Rosero <i>et al.</i> (2007) [14]	Pediatric series	Talus/ankle epiphysis	Not specified	Swelling, pain, restricted ROM	X-ray, CT, MRI	Excision (symptomatic)	2–5 years	Good functional recovery
5	Azzoni (2009) [15]	Child	Talus (hindfoot)	No	Hindfoot valgus deformity	X-ray, CT	Excision±osteotomy	2 years	Deformity maintained
6	Baumfeld <i>et al.</i> (2014) [16]	12F	Talar head extending into sinus tarsi	Yes	Hindfoot mass, inversion/eversion block	X-ray, CT, MRI	Surgical excision	12 months	Pain-free, good ROM
7	Ionescu <i>et al.</i> (2021) [17]	Pediatric	Talus/distal tibia	No	Painful ankle mass	X-ray, CT, MRI	Excision	6–12 months	Good early outcome
8	Sato <i>et al.</i> (2021) [18]	7 years	Talus+loose bodies	Possible	Instability, swelling	MRI, CT	Arthroscopic/open excision	1 year	Stability restored
9	Sharma <i>et al.</i> (2022) [20]	8 M	Ankle epiphysis	No	Swelling, pain	Radiograph	Excision	1 year	Symptomatic relief
10	Meena <i>et al.</i> (2024) [5]	13 M	Talus/distal tibia	Not specified	Swelling, mild pain	X-ray, CT, MRI	Excision	1–2 years	Favorable outcome

M: Male, F: Female, CT: Computed tomography, ROM: Range of motion, MRI: Magnetic resonance imaging

al. [16]. Furthermore, most reported cases involve a single epiphysis, whereas multifocal involvement within the same joint region is uncommon. A similar unusual presentation involving multiple epiphyses in the upper limb has been reported by Kantiwal *et al.* [20], highlighting the spectrum of extensive disease involvement.

In comparison, the present case is unique due to the simultaneous involvement of the distal fibular epiphysis and the talus with extension into the sinus tarsi. To the best of our knowledge, simultaneous involvement of the distal fibular epiphysis, talus, and sinus tarsi in DEH has rarely been reported in the literature and highlights the importance of comprehensive imaging evaluation to identify multifocal disease.

Management strategies are guided by symptom severity and functional limitation. Asymptomatic lesions may be managed conservatively with observation [5, 7]. However, symptomatic cases require surgical excision, particularly in the presence of pain, deformity, or joint dysfunction [2,5,19,21]. Literature review suggests that surgical excision generally results in good functional outcomes, although recurrence has been reported, especially in skeletally immature patients [11,22,23]. In our case, complete excision resulted in excellent functional recovery without recurrence at 12 months. Arthroscopic excision was not preferred because the lesion involved both the

distal fibular epiphysis and the sinus tarsi region, requiring wider exposure for complete excision and adequate protection of surrounding neurovascular and soft-tissue structures. Despite its benign nature, DEH carries a risk of recurrence due to persistent epiphyseal activity in growing children [5, 22, 23]. Although no recurrence was observed at 12-month follow-up, the patient has been advised periodic long-term clinical and radiological surveillance until skeletal maturity to monitor for recurrence, physeal disturbance, deformity progression, or secondary degenerative changes. This case expands the existing spectrum of DEH by demonstrating concurrent multiepiphyseal involvement within the ankle joint, which has rarely been reported.

Conclusion

DEH is a rare epiphyseal disorder that can present with atypical multi-site involvement, as seen in this case, affecting both the distal fibula and talus with sinus tarsi extension. Differentiation from osteochondroma is essential and relies on clinicoradiological correlation. Symptomatic lesions require surgical excision to relieve pain and restore joint function. Careful resection with preservation of joint congruity results in favorable outcomes; however, long-term follow-up is necessary due to the risk of recurrence in skeletally immature patients.

Clinical Message

Dysplasia epiphysealis hemimelica should be considered in children presenting with epiphyseal osteochondral lesions around the ankle. Unusual involvement of both the distal fibula and talus with sinus tarsi extension can occur. Early surgical excision in symptomatic cases provides excellent functional recovery and prevents long-term joint dysfunction.

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given the consent for his/ her images and other clinical information to be reported in the journal. The patient understands that his/ her names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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