CASE REPORT

Dysplasia Epiphysealis Hemimelica or Trevor's Disease: A Case Report

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Trevor's disease, or dysplasia epiphysealis hemimelica (DEH), is an uncommon skeletal developmental disorder representing an osteochondroma arising from 1 or more epiphyses. We present the case of a 21-year-old female who had suffered from a hard protruding mass of the right ankle for more than 10 years. It was mistakenly thought to be posttraumatic osseous fragments by a local practitioner since her childhood. However, DEH was diagnosed with the characteristic image findings and proven by the pathology report after an operation. Review of the related literature shows that this disease is rare in Taiwan. The imaging studies, which are helpful in making a correct diagnosis and proper preoperative plan, are also discussed.

Key words: **Trevor's disease, Dysplasia epiphysealis hemimelica, Epiphysis, Osteochondroma**

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Trevor's disease is a rare developmental lesion that is histologically identical to an osteochondroma affecting 1 or more epiphyses. The reported incidence is 1 in 1,000,000 [1]. Historically, it has been referred to by many names. It was originally described as a "tarsomegalie" in 1926 by Mouchet and Belot [2]. Trevor described 10 patients in 1950 and used the name tarso-epiphyseal aclasis, and this abnormality is also commonly referred to as Trevor's disease [3]. In 1956, Fairbank reported on 14 patients and renamed the condition as the currently used term, *dysplasia epiphysealis hemimelica* (DEH) [4].

DEH usually becomes evident in children and young adults. It is more common in men than in women in a ratio of approximately 3 to 1 [1]. There is no definite evidence that it is hereditary [7, 10], and no cases of malignant transformation have been reported [5, 7, 8]. Typically, clinical manifestations include swelling and, less commonly, pain and deformity. The common sites of involvement are the distal tibia and femur. Upper extremity involvement is rarely reported [5, 10-12]. The disease is usually restricted to the medial or lateral side of the limb (hemimelic).

Herein we report a case of DEH with a typical location and imaging findings. A history of trauma at an early age, however, misled the local practitioner to the diagnosis of fracture fragments. To the best of our knowledge, only 1 case has been reported in Taiwan [9]. By introducing this case, we hope to inform clinicians of its specific imaging findings. In addition to conventional imaging examinations, a three-dimensional (3D) reconstruction computed tomography (CT) scan was also used for this case.

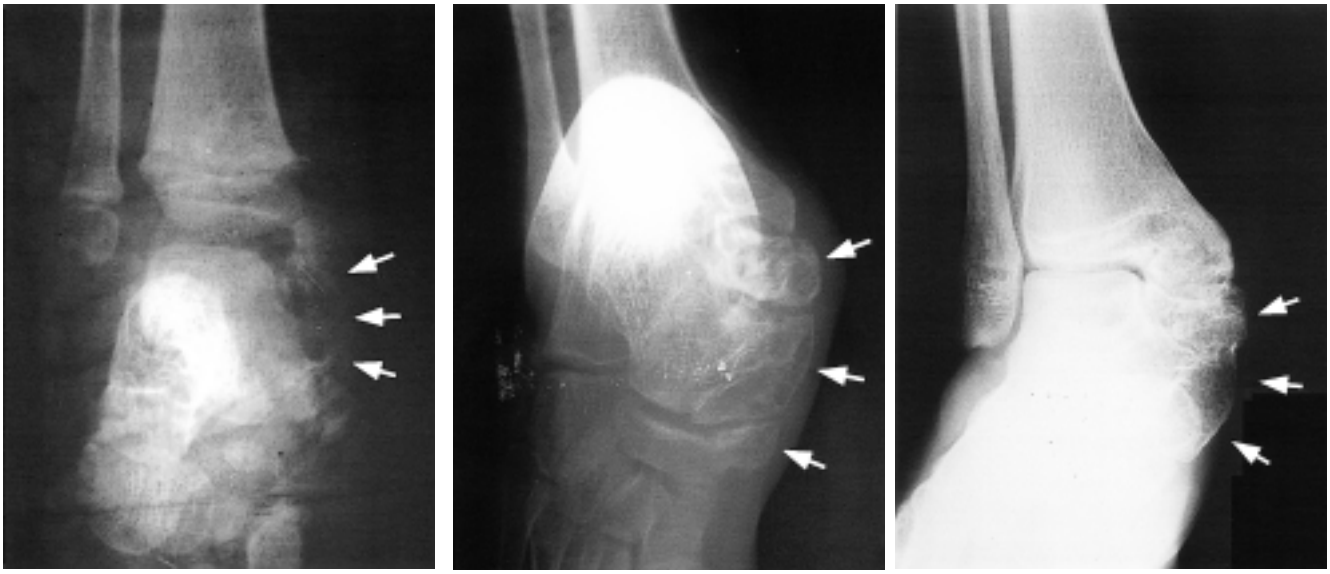

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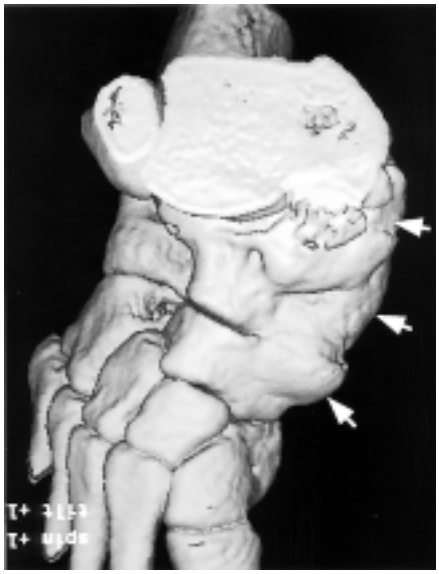
Figure 1. Serial lateral radiographs of right ankle in the case of Trevor’s disease discloses progressive enlargement and confluence of the bony masses. **a.** At 3 years old, small faint osseous bodies over the medial aspect of the hind foot were noted (arrows), which were interpreted as posttraumatic bone fragments then. **b.** At 10 years old, confluences of the osseous bodies with tarsal bones (arrows) were found. **c.** At 21 years old, confluence and enlargement of the bony mass resulted in hind foot valgus.

CASE REPORT

A 21-year-old woman was admitted to our hospital because of right ankle bony mass for more than 10 years. She had had a lesion over the right medial ankle since she was 3 years old, which had been noted after an accident. The mass became progressively larger as she grew up. There was no associated painful disability or marked tenderness except for occasional aching pain. Ankle radiographs were taken twice (Fig. 1a, b), and she was diagnosed as posttraumatic dislocation or large displaced osseous fragments. No specific treatment was given during these years. Some doctors suggested that she continue observation and clinical assessment until she reached skeletal maturity. Because of the unpleasant appearance, she was eager to resolve the problem, and admission was arranged for further evaluation and treatment.

Physical examination revealed a palpable hard mass about 8×8 cm^2 over the right medial malleolar area with limited eversion and inversion of the ankle. Valgus deformity with mild lateral rotation of the ankle and mild muscle atrophy on the right were also noted. Bilateral lower limbs were of equal length. Plain radiographs showed a prominently enlarged medial malleolar osseous mass filling the medial space of the right hindfoot (Fig. 1c). On spiral computed tomography scan with 3D shaded surface display (SSD) reconstruction, a confluent ossific mass was found which had a lobulated appearance and communicated from the marrow with the underlying epiphysis and adjacent bones (Fig. 2). Subsequent magnetic resonance scan for preoperative evaluation of the soft tissue and cartilage demonstrated a bony mass protrusion with marrow continuity from the medial aspect of the distal tibial platform, medial malleolus, talus, and also the navicular bone (Fig. 3). The mass had normal cortical and trabecular signal intensity with incomplete confluence, and was localized over the medial aspect of the right ankle and hindfoot. The resultant enlargement of the medial malleolus and talus caused compression and displacement of the surrounding soft tissue and valgus deformity of the right ankle. The medial tendons, including the posterior tibialis tendon, flexor digitorum longus, and flexor hallucis longus, were preserved. Degenerative change of the right ankle joint with thinning of the articular cartilage was evident.

According to the radiographic findings, a preliminary diagnosis of Trevor's disease was made, and excision was performed. A bony mass



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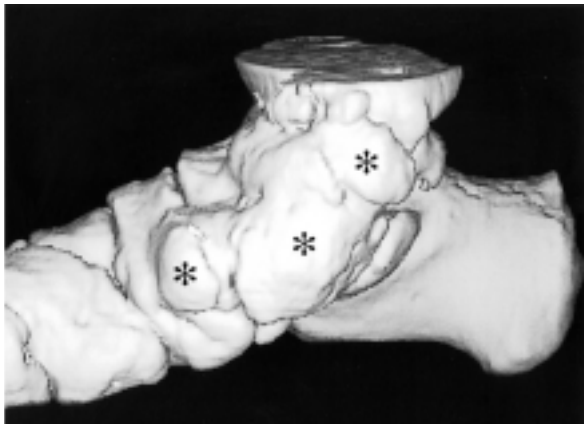

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Figure 2. Craniocaudal **a.** and lateral **b.** projections of the 3D reconstructed CT images, shaded surface display (SSD), well delineate the lesion for surgical planning (arrows and ∗).

with cartilage caps over the medial malleolus and talus medial site was found during the operation. It was removed with an oscillating saw, and the deltoid ligament was repaired and re-attached to the bony surface. The postoperative condition was stable, and the patient was discharged with good wound healing.

On pathologic examination, the lesion consisted of multiple pieces of bony tissue. The surface was capped by cartilage and covered by fibrous tissue with endochondral ossification. This finding is consistent with dysplasia epiphysealis hemimelica.

DISCUSSION

The etiology of DEH is still unclear. In Trevor's report, he believed that the lesion was congenital and was related to an insult during the formation of the limb bud [3]. Fairbank believed the process to be "a true dysplasia or faulty growth of part of the epiphysis itself" [4].

Typically, the disease occurs over the epiphysis or the tarsal or flat bones, in a single lower extremity. The ankle is most commonly affected. Although asymmetrical involvement has been reported [13], the lesions typically affect only one side of the joint, with the medial side affected twice as frequently as the lateral [6].

There may be single lesion or multiple lesions; 2/3 of patients have multiple lesions [1, 5, 11, 12]. This leads to designation of a localized form (monostotic involvement), a classic form (more than 1 area of osseous involvement in a single extremity), and a generalized or severe form (involvement of an entire single extremity) [14]. The localized form of DEH usually affects bones of the hindfoot or ankle. The classic form shows characteristic hemimelic distribution and accounts for more than 2/3 of cases. It typically involves more than one epiphysis within a single lower extremity, especially around the knee and

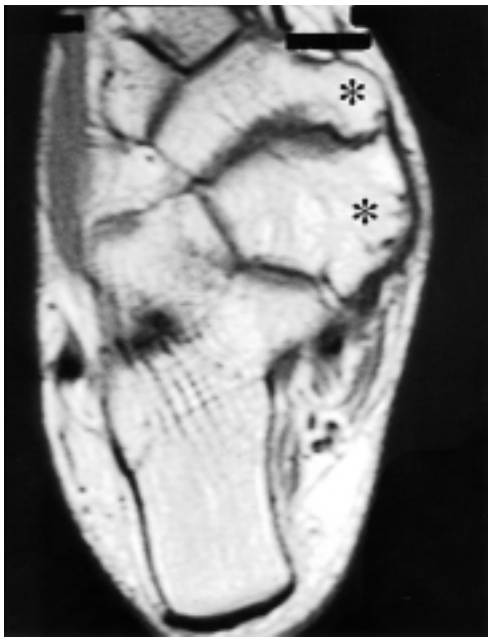

Figure 3. Axial (a, TR/TE/excitation=550/11/2) T1-weighted MR images (Signa 1.5T, GE Medical system) of the ankle demonstrate osteochondromas arising from the talus and navicular with marrow continuity and protruding osseous mass (∗).

ankle (talus, distal femoral, and distal tibial epiphyses). In the generalized or severe form, the entire lower extremity may be involved. The affected location in our patient was over the right medial ankle, which is the most common site reported. The involved range was the classic form.

In our patient’s history, plain films were taken twice. One was taken at the age of 3 just after the accident, and small faint osseous bodies can be observed (Fig. 1a). The other was taken when she was about 10 years old; multiple ossified masses can be noted protruding from the medial malleolus, talus, and navicular bone (Fig. 1b). The latest film taken in our hospital shows that all the separate ossifications join to give a lobular appearance with valgus deformity of the ankle joint (Fig. 1c). These radiographic findings have been well described in multiple reports [5, 6]. The lesion is often not visible on plain radiographs initially, as it may not begin to ossify. Once mineralization has begins, it appears as at a secondary ossification center. In infant or young child, small, multifocal, irregular ossifications are seen adjacent to 1 side of an ossifying epiphysis (or carpal or tarsal bone). The adjacent metaphysis may be widened. Subsequently, the ossifications become confluent with the adjacent bone, eventually appearing as a lobulated osseous mass protruding from the epiphysis (or carpal or tarsal bone) [15]. The final appearance resembles that of osteochondroma with the affected area (or areas) remaining large and irregular.

Premature closure of the physis may occur along with its consequence of limb deformity and limb length discrepancy. The articular surface is often irregular and, combined with the angular deformity, frequently results in premature secondary osteoarthritis. At an early stage, this finding may be confused with either loose bodies or synovial osteochondromatosis. Synovial osteochondromatosis occurs primarily in later adulthood and is extremely rare in children. A misdiagnosis of loose bodies was previously made for our patient due to her traumatic history. But, subsequent examinations allowed us to make the definite diagnosis because bony fragments should not enlarge as a patient grows up.

CT is used to assess the continuity of the lobulated mass with the underlying epiphysis. It also demonstrates similar attenuation of tissue between the two structures, both composed of cartilage and osteoid. However, the relationship in different section planes is better evaluated with MR imaging. In addition to conventional CT imaging, we further studied this case with 3D reconstruction. The appearance of the mass and the 3-dimensional relationships between the lesion and adjacent bones (distal tibia, talus, and navicular) were observed more clearly in the reconstructed images (Fig. 2).

MR imaging is extremely useful for identifying the extent of epiphyseal involvement, joint deformity, and the effect on surrounding soft tissue, as with osteochondromas at other sites. Details of the unossified cartilaginous mass and the status of the articular and meniscal cartilage and growth plate are optimally assessed with MR imaging. The lesion and involved epiphyseal cartilage have similar signal intensities, with intermediate signal intensity on T1-weighted and high signal intensity on T2-weighted MR images. Areas of low signal intensity on T1- and T2-weighted images indicate areas of calcification or ossification that increase as the lesion and patient's skeletal maturity progress. Often there is a clear plane of separation between the lesion and the normal epiphysis. Complete skeletal maturity of our patient was noted at admission, and the lesion was shown to be totally ossified with marrow continuity to adjacent bones on MR imaging. According to the findings on her early radiographs, we believe that the cartilaginous component and the separated plane between the accessory ossification center and the normal epiphysis could have been observed if she had been examined at a younger age.

According to the pathology report, the lesion is a lobulated mass protruding from the epiphysis with a cartilaginous cap. The lesion is sometimes indistinguishable from the normal epiphysis. Its histologic features are identical to those of an osteochondroma, consisting of a base of normal bone and hyaline cartilage with abundant enchondral ossification. The histologic findings support Trevor’s hypothesis that the disease is a result of abnormal cellular activity at the cartilaginous ossification center. The normal cartilage sequence follows an orderly fashion of cell proliferation, maturation, senescence, and disintegration that does not occur in DEH. Trevor used the term aclasis in describing this failure of normal cellular breakdown.

Biopsy is typically not necessary because of diagnostic radiologic features. However, a

skeletal survey should be performed to exclude additional areas of involvement, particularly of the lower extremities. Surveillance and clinical assessment are usually continued until the patient reaches skeletal maturity. Surgical intervention is more frequently required for these lesions than for solitary osteochondromas because the epiphyseal location is often associated with pain, deformity, or loss of normal mechanical function [11]. Excision, even incomplete resection, may result in a reduction of symptoms. Surgery is often directed at improving joint congruity to lessen subsequent development of secondary osteoarthritis; thus, treatment at an early stage of disease improves the outcome [3, 11]. Corrective osteotomies may also be needed to treat residual deformities. The prognosis of this disease is generally good.

It is reported that plain films supplemented with MRI provide the greatest diagnostic information in cases of DEH. Though MRI is helpful in defining the site and extent of the osteocartilaginous mass and providing detailed images of the associated joint deformity, we think it is not routinely required. If there isn't much cartilaginous, component the exact location and extent of lesion can be fully evaluated by CT imaging. With 3D reconstruction, the lesion can be "seen" through the covering soft tissue from any direction without excision.

SUMMARY

Clinicians must be aware of DEH if a patient has a growing hard mass over one side of single lower extremity. Imaging can help to make a correct diagnosis. If bony fragments are suspected, a subsequent imaging examination will be helpful. CT and MR can show the relationship between the lesion and the underlying bone marrow or surrounding soft tissue. MRI is not needed as a routine examination especially when the patient has passed puberty. Sectional and reconstructed 3D CT can be used for making a proper evaluation and surgical plan. ◆

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Trevor氏病：病例報告

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Trevor氏病是一種少見的骨骼發展疾病，現在較常使用的名稱為半側肢體骨骺發育不良，即是長在骨骺板上的骨軟骨瘤，我們報告一位21歲的女性，從3歲起就因右踝關節內側逐漸腫大而困擾，在外面的醫院都被當作是受傷後的變化來治療，直到來我們醫院後才根據其獨特的影像表現而診斷為Trevor氏病，此診斷也獲得開刀後的病理報告證實；回顧過去的相關文獻，我們發現此病例在台灣地區極為罕見，值得提出來討論，同時我們發現經由影像檢查不但能獲得正確診斷，對手術前評估也有幫助。

關鍵詞：Trevor氏病、半側肢體骨骺發育不良、骨骺板、骨軟骨瘤