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Learning from errors

Dysplasia epiphysealis hemimelica: a huge articular mass with unpredictable surgical results

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Summary

Dysplasia epiphysealis hemimelica or Trevor's disease is a rare disorder of localised osteochondral overgrowth affecting the epiphysis of extremities. This paper reports a 12-year-old boy presenting with a large bony mass at the left ankle diagnosed as dysplasia epiphysealis hemimelica. The articular surface of the ankle joint of the patient was evaluated with preoperative and postoperative MRIs. The 2-year postoperative MRI showed early osteoarthritis of the ankle, therefore demonstrating the importance of early excision avoiding more complex resections of intra-articular lesions.

BACKGROUND

Dysplasia epiphysealis hemimelica (DEH) or Trevor's disease is a rare developmental epiphysial disease in children.[1] The disease was first reported by Mouchet and Belot.[2] Trevor reviewed DEH later and designated it as tarso-epiphysial aclasis in 1950.[3] The disease involves single or multiple epiphyses of extremities. The most common sites affected include the distal femur, proximal tibia and talus.[4] DEH has been reported as one in one million.[5] The disease was found to be three times more common in boys.[6] With unknown aetiology, the lesion is an osteocartilaginous overgrowth of epiphysis that mimics osteochondroma arising from the epiphysis.[7] However, the disease has not been previously reported in Thailand.

This paper reports one case of DEH with a huge intra-articular mass that originated from the talus with difficulties in surgical treatment and premature postoperative osteoarthritis.

CASE PRESENTATION

A 12-year-old boy presented with an enlarged mass on the medial aspect of his left ankle. He noted that the mass had increased in size during the 6 months prior to coming to our hospital. His mother reported that the mass appeared after he fell from a bicycle. The mass disturbed the motion of his ankle and was painful after prolonged walking and running. There was no familial history for bone dysplasia and metabolic bone disease.

On examination, there was a large bony mass of 5×6 cm over the right ankle. The palpable mass extended from the anterolateral aspect of the ankle to the posterior and medial side of the ankle (figure 1). No wound or skin lesions were found over the ankle. The movements of the ankle were limited. The boy could perform 10° of plantarflexion but could not dorsiflex the ankle. The neurovascular examination was normal.

INVESTIGATIONS

The findings on plain radiographs and CT scans of the ankle showed an excessive overgrowth of mass extruding from the anterolateral and posterior aspect of the talus (measuring 6.7×6.6×3.8 cm in antero-posterior, transverse and superoinferior diameters, figures 2 and 3). The mass demonstrated cortical and medullary continuity with the underlying bone and occupied the greater part of the ankle joint. The mass laterally displaced the distal fibula.

The MRI described a large lobulated mass occupying the left ankle joint. The mass eroded the neck and body of the talus (figure 4).

DIFFERENTIAL DIAGNOSIS

DEH, osteochondroma and parosteal osteosarcoma must be considered for differential diagnosis.

According to the radiographic appearance, DEH normally originates from the epiphysis but the osteochondroma originates from the metaphysis of the bone. Additionally, osteochondroma shows cortical and marrow continuity with the underlying bone. In contrast, the

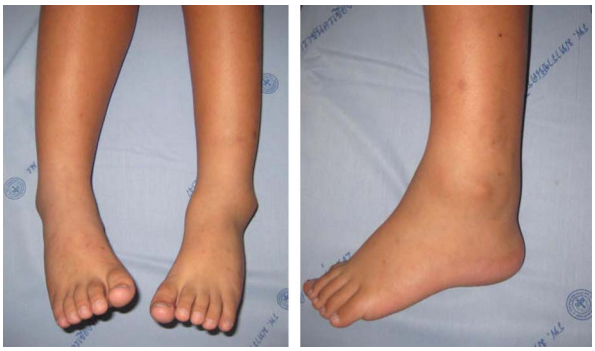

Figure 1 Left ankle shows a large mass restricting the motion of the ankle joint.

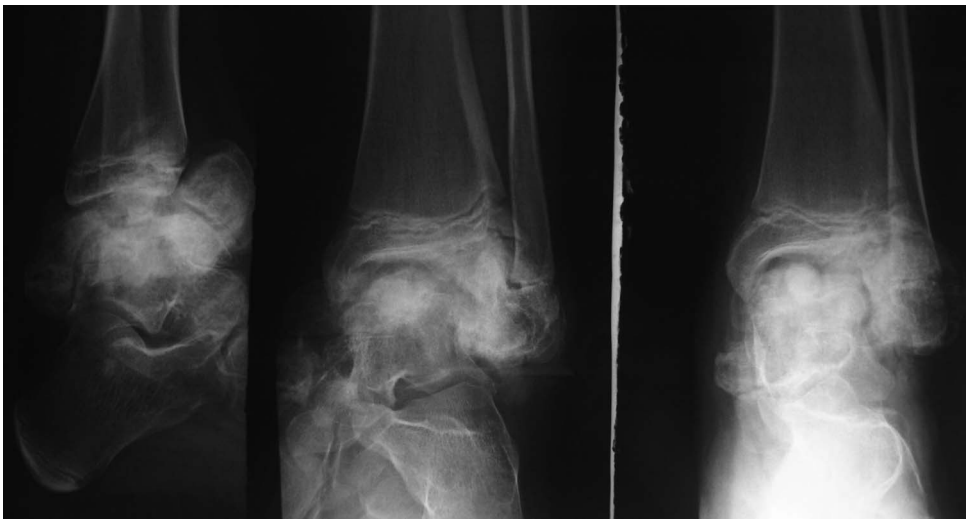

Figure 2 Plain radiographs of the left ankle show a huge lobulated mass with multiple focal calcifications on the anterolateral and posterior side of the ankle.

separation between the tumour and cortex is characteristic of parosteal osteosarcoma.[8] From the MRI findings, the lesion was a multilobulated intra-articular mass protruding from the talus and had continuity with the underlying bone.

TREATMENT

The operation was performed through the anterolateral approach to excise the articular cartilaginous mass. The mass arose from the articular surface of the talus. Also, anterior impingement between the bony mass and the articular surface of the distal tibia was identified. Because the mass obstructed the greater part of the ankle motion, as much of the mass was excised as possible to restore congruity of the ankle joint, as well as to improve the ankle motion. The excised mass demonstrated cartilaginous caps over the bones (figure 5).

OUTCOME AND FOLLOW-UP

The pathological examination performed on the cartilaginous mass revealed a benign osteochondroma with a cartilaginous cap measuring up to 0.6 cm in thickness (figure 6). Three months after the operation, the patient could perform full weight bearing on the left foot. At the 2-year postoperative examination, he could perform 10° dorsiflexion of the ankle without pain, less than the normal motion of the right ankle. The MRI scans showed marked narrowing of the ankle joint and irregularity of the articular surfaces of the distal tibia and talus. A small osteochondral defect in the middle of talar dome, measuring 0.4 cm in size, was found (figure 7).

Although the radiographic findings showed early osteoarthritis, the patient had no pain in the ankle and could perform full unrestricted activity at his 3-year follow-up.

DISCUSSION

DEH is a rare disorder of epiphysial overgrowth. Most patients with DEH seek an orthopaedist because of a painless mass or restricted joint motion. Azouz *et al*[7] classified DEH into three types: the localised form (monostotic involvement), the classical form (more than one area of osseous involvement in a single extremity) and the generalised form (disease involving an entire single extremity). This patient had a single large lesion (localised form)

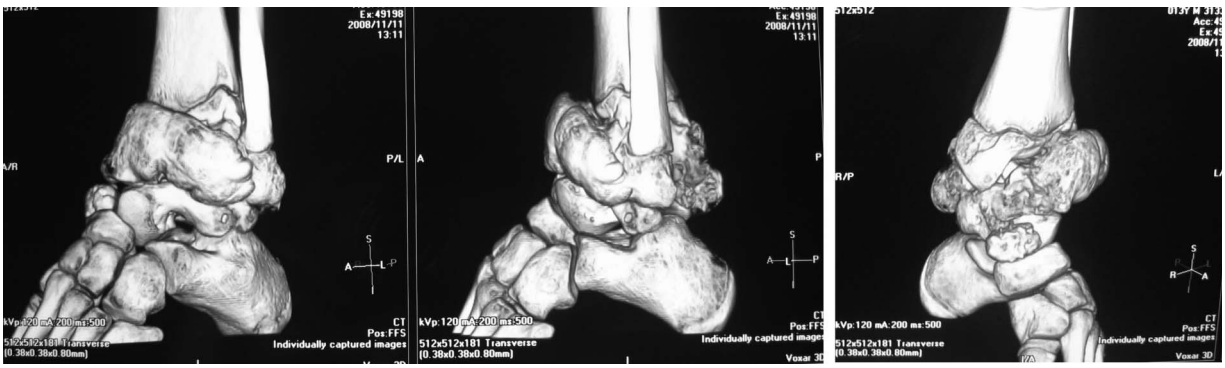

Figure 3 Three-dimensional CT of the left ankle demonstrates a huge multilobulated mass protruding from the talus and obstructing the greater part of the ankle joint.

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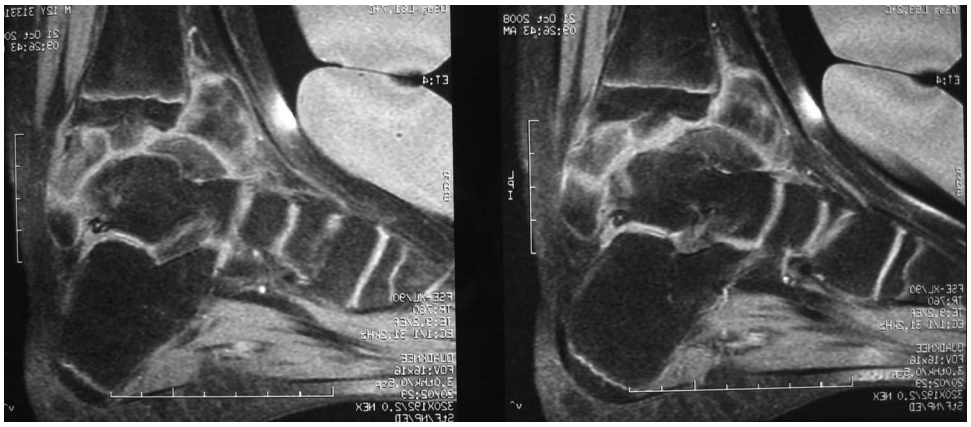

Figure 4 MRI of the left ankle. T2-weighted images demonstrate a heterogeneous mass arising from the talus and occupying the greater space of the joint.

involving the talus and the ankle joint. Radiographic features of Trevor's disease are characterised by a lobulated mass protruding from one side of the epiphysis of the long bone or tarsal bones. The mass has irregular calcification and metaphyseal changes may be found.[1 9]

CT scanning and MRI were helpful in demonstrating the extent of the mass as well as the relationships between the lesions, bones and surrounding soft tissues. Additionally, an MRI can identify the irregularity of the articular surface and subchondral bone. Histological examination of the lesions resembles a benign osteochondroma, consisting of normal bone and hyaline cartilage with abundant enchondral ossification.[1 10]

Treatment of DEH may use observation or excision depending on the location, the extent of lesion and the individual problems of the patient. Keret *et al*[11] showed good results of non-surgical treatment among patients with Trevor's disease. Surgical intervention by excision was considered for those patients with pain or functional limitation of the joint. Orthopaedic surgeons may have to be concerned with an incomplete or complete resection of the mass, depending on the site and extent of the mass.

The outcome of surgical intervention for DEH is variable depending on the location and size of the mass. If the mass is large and intra-articular, excision may increase the risk of early osteoarthritis. Kuo *et al* reviewed nine patients with the diagnosis of DEH. They showed excellent results with excision of juxta-articular lesions but found fair and poor results with excision of articular lesions.[12] They reported local recurrence after partial excision of the articular mass and found the cases had joint stiffness after excision. Additionally, surgical treatment

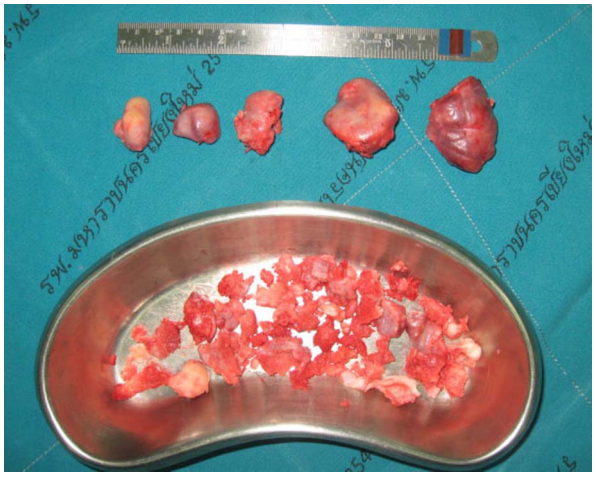

Figure 5 Histopathological appearance demonstrates cartilaginous cap covering the medullary bone which looks similar to osteochondroma.

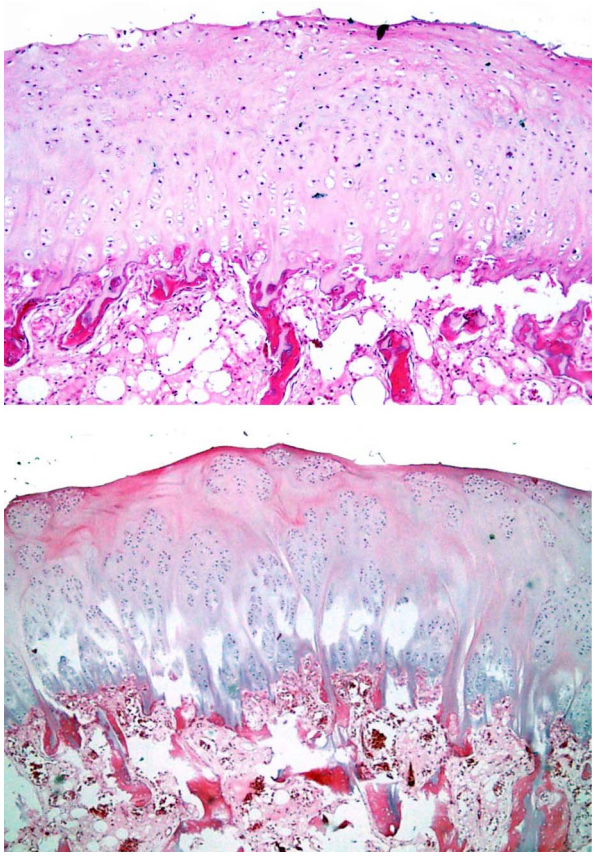

Figure 6 Excised mass shows cartilaginous caps covering the bones.

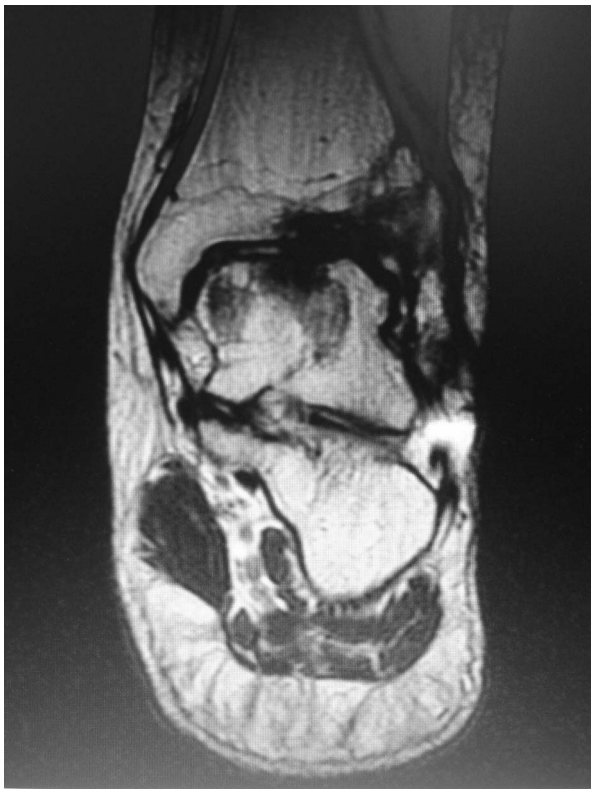

Figure 7 MRI of left ankle at 2-year postoperative exam. T1-weighted images demonstrate premature arthritis. *Note*: small articular defect at the distal tibia and the dome of the talus.

may not be necessary for asymptomatic patients. According to the literature, no malignant transformation of the mass was reported.

This patient had an intra-articular lesion protruding from the talus and obstructing the range of motion. With concerns of pain and limitations of the ankle joint, an attempt was made to remove the large mass from the ankle joint. However, it was difficult to manage this patient who had a huge lump arising from the articular surface and obstructing ankle joint motion. Unfortunately, the 2-year postoperative MRI showed early painless secondary osteoarthritis although the patient had increased ankle motion. The researchers believed that premature osteoarthritis of the patient may result from iatrogenic intra-articular excision.

Bhosale *et al*[13] suggested excision if the lesion caused disability and arthrodesis of the ankle may be appropriate with extensive involvement of the talus. Trevor[3] and Kettelkamp *et al*[14] suggested an arthrodesis for patients developing degenerative joint disease. For this patient with early osteoarthritis, continuous clinical symptoms and the normal activity of daily living must be monitored to determine radiological changes to progressive osteoarthritis.

This patient had improvement of ankle joint motion and pain was eliminated during the 3-year postoperative period. According to the MRI scans, there was no recurrence of the mass. The patient could perform 10° dorsiflexion the ankle and had no limitations of normal daily living. The patient was recommended to continue follow-up at regular intervals.

Owing to the delayed medical examination, this patient had an enlarged intra-articular mass, making the decision for surgical treatment more difficult. Early excision is recommended with no resection of a painless intra-articular mass unless the mass obstructed motion of the joint owing to the risk of early degenerative joint disease. With a large intra-articular lesion and the risk of cartilage injury, the large mass should be partially resected to improve joint motion and avoid cartilage injury. Afterward, the patient needs to be monitored continuously with plain radiographs and an MRI after excision of the mass.

In summary, early identification, adequate radiological investigations and a well-planned operation for a child with DEH can achieve the good surgical outcome.

Learning points

- It is essential to differentiate dysplasia epiphysealis hemimelica from osteochondroma or parosteal osteosarcoma with radiological investigations and histological examinations to clarify diagnosis.
- MRI is very useful for preoperative evaluation of the lesion and postoperative follow-up.
- Dysplasia epiphysealis hemimelica with a painless intra-articular mass should not be excised unless the mass interferes with the joint motion.

Competing interests None.

Patient consent Obtained.

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