Case Report

Dysplasia Epiphysealis Hemimelica with Bony and Soft-Tissue Abnormalities: A Case Report and Review of the Literature

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ABSTRACT—

We report a case of a 3-year-old boy with dysplasia epiphysealis hemimelica (DEH) who presented with a hard painless swelling on the medial side of his left ankle joint. Plain X-rays, CT and MRI showed a bony tumor and associated soft-tissue abnormalities. At age one year, he had been diagnosed with an osteochondroma and underwent incomplete surgical excision at a local hospital. Due to joint impingement and restricted movement, we chose to completely excise the tumor. Pathologically, the specimen showed evidence of an osteochondroma. From its clinical, imaging, and pathological features and reports in the medical literature, a diagnosis of DEH was established. DEH is a rare variant of osteochondroma, resulting in many patients being incorrectly diagnosed or diagnosed with osteochondroma alone. It is important to differentiate low-grade malignant tumor or osteochondroma-like parosteal osteosarcoma from DEH of the talus, especially when accompanied by soft tissue abnormalities. Surgical treatment is mandatory in patients with symptoms such as pain, joint impingement and deformation. Incomplete excision of an articular lesion may lead to recurrence and additional surgery, making it less effective than complete excision.

KEY WORDS: dysplasia epiphysealis hemimelica (DEH), osteochondroma, soft-tissue abnormalities, talus, Trevor’s disease

INTRODUCTION

Dysplasia epiphysealis hemimelica (DEH), or Trevor’s disease, is a rare skeletal developmental disorder characterized by osteocartilaginous overgrowth involving single or multiple epiphyses on one side of the body, especially in children1-3. DEH can be classified into “localized”, affecting a single bone; “classical”, affecting more than one area in a single lower extremity; or “generalized”, involving an entire lower limb from the pelvis to the foot4. These lesions can also be classified as juxta-articular (i.e., adjacent to the articular surface) or articular (i.e., directly involving the joint surface forms)5, or as intra or extra-articular variety6.

We report a case of a 3-year-old boy with DEH, who presented with a recurring hard painless swelling of his left ankle, with MRI showing associated soft-tissue abnormalities. At age one year, he had been diagnosed with an osteochondroma at a local hospital and underwent partial excision. However, it recurred after two years.

CASE REPORT

A 3-year-old boy presented at Xiang-Ya Second Hospital with a painless swelling and gradually enlarging deformation on the medial aspect of his left ankle. Two years earlier, at age one year, he had presented with deformation and abnormal gait, for which he underwent incomplete excision at a local hospital (Fig. 1A). Pathologic findings showed an osteochondroma.

Physical examination showed a 4-cm-long surgical scar on the medial side of his left ankle, along with a 2 X 3 cm swelling on the posterio-medial side. The mobility of the ankle was reduced, with medial and lateral rotations of 10° - 0° - 15° and dorsiflexion and...
plantar flexion of $10\degree - 0\degree - 20\degree$. There was no evidence of wasting and no discrepancy between the lengths of his lower limbs.

A plain X-ray showed a bony tumor on the posteriomedial side of the distal epiphysis of his left tibia and talus, with swelling of surrounding soft tissues (Fig. 1, B - C). Computerized tomography (CT) showed an irregular, expanding bony tumor, with osteoepiphysis of the left distal tibia and the left medial and posterior ankle. The tumor was of heterogeneous density, including cystic low density and high-density spotted state focus (Fig. 2A). Three-dimensional reconstructed images showed that his talus-ankle articular surface was ossified and the local joint space was narrowed, with osteoporosis and a cystic euphotic zone in the left talus (Fig. 2B - C). These findings suggested tumor recurrence in the medial epiphysis of the left tibia and talus; moreover, the presence of soft tissue lesions suggested a low-grade malignant tumor.

MRI showed a bony tumor on the medial side of the distal epiphysis of the tibia and talus (Fig. 3). T1-weighted MRI showed a mass of low to intermediate signal intensity, whereas T2-weighted MRI showed a mass of intermediate to high signal intensity. Swelling of the surrounding soft tissues, increased effusion, and heterogeneous signals in the left calcaneus and talus were observed, again suggesting tumor recurrence in the medial epiphysis of the left tibia and talus, although epiphyseal dysplasia of the left talus could not be ruled out. Plain X-rays of the left knee and

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**Fig. 1:** Clinical course in our patient. (A) At one year of age, this patient had undergone incomplete excision at a local hospital. (B - C) Plain X-rays of his left ankle at three years of age, showing a bony tumor on the posterior-medial side of the distal epiphysis of the tibia and talus, with an irregular and thickened structure and swelling of the surrounding soft tissue. (D) X-ray six months after complete excision, showing no recurrence of tumor, although the local joint space of the talus-ankle articular had become narrower.

**Fig. 2:** CT findings in our patient, showing (A) an irregular and expansional bony tumor (osteoepiphysis in the left distal tibia, left media and posterior ankle) with heterogeneous density (cystic low density and high-density spotted state focus). The talus-ankle articular surface had ossified and the local joint space had narrowed. Swelling of local soft tissue was observed, as well as osteoporosis and a cystic euphotic zone in the left talus. (B, C) Three-dimensional reconstructed images are shown.
right ankle and knee showed no abnormalities. Blood biochemistry showed elevated concentrations of alkaline phosphatase (290.51 µl; normal range: 30.0-110.0 µl) and acid phosphatase (7.3 µl; normal range: 0.0-5.55 µl), while other tests, including routine blood tests, electrolytes, erythrocyte sedimentation rate and C-reactive protein concentrations were all within normal range.

Because this patient presented with painless swelling, joint deformity and limited range of motion, we performed surgery. The medial and posterior aspects of the epiphysis of the left distal tibia were

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**Fig. 3:** MRI findings in our patient, showing (A - B) a bony tumor measuring 2 x 3 cm on the medial side of the distal epiphysis of the tibia and talus. T2- weighted MRI showed intermediate to high signal intensity, with swelling of the surrounding soft tissues, increased effusion, and a heterogeneous signal in the left calcaneus and talus.

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**Fig. 4:** Histopathological examination, showing a benign osteochondroma, composed of cortical and medullary bone with overlying hyaline cartilage.
completely excised, along with the overgrowth of the posterior talus. Histopathological examination of the specimen revealed a benign osteochondroma, composed of cortical and medullary bone with overlying hyaline cartilage (Fig. 4). The patient was followed-up for six months, during which he showed no recurrence of pain or deformity and no limitations in ankle movements (Fig. 1D).

DISCUSSION

DEH is a rare skeletal developmental disorder, usually diagnosed at age 2 - 14 years, but rarely in adults[7]. Its reported incidence is 1:1,000,000[6], but it may be much higher[8], since many patients may be improperly diagnosed, including the diagnosis of an osteochondroma[8-10]. DEH is two-fold more common in men than in women[11]. Most lesions affect one side of the joint, with a 2:1 medial-to-lateral ratio. In two-thirds of patients, more than one epiphysis is affected[11]. DEH usually involves a single lower extremity, especially around the knee and ankle. Although asymptomatic in most patients, DEH may cause mechanical symptoms, including swelling, reduced mobility, stiffness, articular deformity, limb length discrepancy, and early secondary osteoarthritis, depending on size and location.

Imaging, by X-ray, CT and MRI, has a major role in the diagnosis of DEH. Radiographically, DEH presents with asymmetrical overgrowth on one side of an epiphysis, with irregular or premature calcifications. Following a diagnosis of DEH, a skeletal survey should be performed to determine whether there are multiple locations, using methods such as scintigraphy and whole-body MR imaging[12].

Bony and associated soft tissue abnormalities are extremely rare in DEH[13,14]. Despite the imaging features of DEH, it must be differentiated from low-grade malignant tumors and osteochondroma-like parosteal osteosarcomas when the talus is involved, especially when associated with soft tissue abnormalities. The CT and MRI findings in our patient suggested tumor recurrence, and the presence of soft tissue lesions suggested the possibility of a low-grade malignant tumor.

Pathologically and histologically, DEH lesions are similar to benign solitary osteochondromas[1]. The lesion may be a pedunculated mass with a cartilaginous cap or an enlarged irregularity of the articular surface. Gene expression assays, including EXT1 and EXT2, are all within normal ranges in DEH, but are lower in osteochondromas due to a mutation[15,16]. Because these tests are costly, clinical and radiological findings are important diagnostic tools[17]. Based on the clinical, imaging and pathological features of the specimen and reports in the medical literature, a diagnosis of DEH was established.

Treatment of DEH ranges from simple observation to surgical excision, depending on the location and extent of involvement. In the absence of articular symptoms, simple observation is recommended[5]. Patients who exhibit symptoms such as pain, joint deformity, or limited range of motion should be treated surgically. Although excising a juxta-articular lesion generally yields excellent results, excising an intra-articular lesion may lead to early secondary osteoarthritis, which may then require arthrodesis. Excision of lesions directly involving the joint surface, with articular localization, is generally not recommended unless the lesion becomes a loose body. The risks of recurrence until the epiphyses are closed suggest the need for continuous monitoring[10]. Little is known about the results of incomplete excision of articular DEH. Although two patients who underwent incomplete excision without additional surgery were reported to be doing well, with no evidence of local recurrence or physical problems and slow resolution and disappearance of the residual DEH masses[14], others have shown poor clinical outcomes, including local recurrence[5]. DEH lesions increase in size until skeletal maturity, without malignant transformation[5,10].

CONCLUSION

DEH is a rare variant of osteochondroma, resulting in an incorrect diagnosis in many patients. It is important to differentiate low-grade malignant tumors and osteochondroma-like parosteal osteosarcomas from DEH of the talus, especially when accompanied by soft tissue abnormalities. Surgical treatment is mandatory in patients with symptoms such as pain, joint impingement and deformation. Incomplete excision of an articular lesion may lead to recurrence and additional surgery.

REFERENCES


