Dysplasia Epiphysealis Hemimelica

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Abstract

Dysplasia epiphysealis hemimelica is extremely rare condition. Asymmetrical abnormal cartilage proliferation and associated enchondral ossification in an epiphysis is characteristic. It is limited to the medial or lateral half of a single limb. A child with dysplasia epiphysealis hemimelica of the knee is reported.

Key words: Dysplasia, tarsomelic, epiphyseal osteochondroma

Introduction

Dysplasia epiphysealis hemimelica (DEH) is an uncommon skeletal development disorder affecting the epiphysis in young children. More than 120 cases have been reported in literature since the first report of Mouchet and Belot in 1926, who described the condition as a tarsal bone disorder and coined the name "tarsomelic". In 1956 Fairbank described 14 additional cases and suggested the term "dysplasia epiphysealis hemimelica". This term stresses the occurrence in epiphyses and round bones in joints other than the ankle and differentiates the condition from other forms of epiphyseal dysplasia. It also refers to the involvement of half of the epiphysis.

The medial sites of epiphysis are affected two or three times more frequently than the lateral site. Some patients have disturbance of an entire epiphysis rather than a medial or lateral portion and others have had a medial lesion at one level in an affected limb and lateral at another. Involvement of almost all epiphyses on one side of the body including the pelvis, scapula, ribs, and vertebra has been reported.

DEH has been reported in other anatomical sites, including the capitul femoral epiphysis and acetabulum, carpal bones, shoulder, wrist, calcaneus, patella.

Dysplasia epiphysealis hemimelica also has been described as a developmental affection of the skeleton that is characterized by unilateral, asymmetrical abnormal proliferation of cartilage, which is associated with enchondral ossification in an epiphysis. The growth of the lesion usually ends at the time of epiphyseal closure as in metaphyseal exostosis.

This disorder has received a variety of other names in the world literature: epiphyseal osteochondroma, unilateral epiphyseal dysplasia, chondrodystrophic epiphysaire, fragmentation osseous hypertrophique. Aznau et al., who reported the variable manifestation of DEH, have subdivided it into localized, classical and generalized forms. This rare osteochondrodys trophy is not easily recognized and thus misdiagnosis and incorrect treatments often occurs. It should be differentiated from the multiplex and punctate forms of epiphyseal dysplasia. A case report as well as a brief literature review regarding DEH is presented here.

Case report

At the age of two years and seven months, the patient was brought in for consultation because of slight limping and swelling on inner side of the left knee. X-rays showed considerable bony involvement and irregular cartilaginous tissue with calcification (Figure 1).

Figure 1: Considerable bony involvement and irregular cartilaginous tissue with calcification are detected on X-ray.

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addition, instances have been reported in which the entire epiphysis rather than only half of it was affected.3

Any bone of the body may be affected, but no involvement of the skull or ossa faciei has been reported.6 The commonly involved bones in order of frequency are the distal femur, proximal tibia, talus, tarsal navicular, and first cuneiform. Other reported sites of involvement include:

- **Upper limb:** scaphoid, lunate, capititate, hamate, trapezium, trapezoid, metacarpals, phalanges, proximal radius,6 proximal and distal humerus, glenoid cavity, and coracoid process of the scapula.

- **Lower limb:** calcaneus cuboid, middle and lateral cuneiforms, metatarsals, phalanges, proximal tibia,6 distal tibia, fibula and proximal femur.

- **Pelvis:** sacroiliac joint, acetabulum, pubic and iliac bones.3

The etiology is unknown. The epiphysial dysplasia is inborn errors of epiphysial development and are presumed to be caused by an abnormality of epiphysial ossification. The process is progressive until at the time of epiphysial plate closure. The prognosis depends on the severity of the involvement. It is a benign lesion and malignant degeneration has never been reported.3

Clinically, patient with dysplasia epiphysialis hemimelica exhibits hard swelling which is usually painless, with slow progressing at the site of involvement. Usually, deformities (varus, valgus, flexion-contracture, pes planus, pes equines) are apparent and range of motion is limited. Pain is not a frequent complaint but patient may complain of aching pain. Other symptoms may include lengthening or shortening of the affected limb and regional muscle atrophy.3

The earliest radiographic sign of this dysplastic disorder is the appearance of secondary centers of ossification adjacent to an epiphysis. These centers slowly enlarge, eventually merging with each other and then with the epiphysis. In some patient, the normal centers of ossification in the affected limb may appear prematurely and in time become larger than that of opposite side. The metaplastic area also be involved, exhibiting widening, streaking like that in Ollier's disease, or spur formation as in osteochondroma. Radiologically, mature lesions show enlargement of one side of an epiphysis, simulating an osteochondromatous mass or enlargement of one side of an ossification center. The epiphyses are irregularly calcified and ossified. The joint is usually deformed. Once the abnormality is identified in one bone, a radionuclide bone scan or skeletal survey should be performed to detect additional sites of involvement. The lesion continues to enlarge usually stops with skeletal maturity. Not uncommonly, however, clinical
symptoms may worsen due to deforming and secondary osteoarthritis.\(^7\)

Double-contrast arthrography can be used to determine the configuration of the articular surface and the extent of the cartilaginous mass if MRI is not available.

Fluoroscopy during the arthrogram can help demonstrate the joint dynamics. CT helps in making the correct diagnosis by accurately demonstrating the relationship between the bones, soft tissue, and cartilaginous mass. It can provide information about the extent of the lesion and the condition of the articular cartilage.

MRI\(^{10,11}\) provides detailed view of the unossified cartilaginous mass as well as the status of the articular cartilage. MRI has the advantage over double-contrast arthrography of being non-invasive.

MRI, which provides more accurate information than CT, can be used to differentiate the abnormal epiphyseal growth from the main epiphysis. Depending on the stage of development of the ossific nucleus, MRI may cause the normal nuclear signal intensity for T1 and T2 weighted images to be different in the cartilaginous mass overgrowth and the epiphysis. Plain radiographs supplemented with MRI provide the most information.

Biopsy is not needed for diagnosis as the radiological appearance is very specific in most cases. Once the diagnosis of DEH is made, a skeletal survey needs to be done to check for other sites of involvement. This survey should include the pelvis, and upper and lower extremities with the hand and feet. For the appearance of new lesions, patient or parent awareness and clinical surveillance till puberty is needed.\(^5\)

DEH should be differentiated from other entities having a similar radiographic appearance, such as myositis ossificans, tumoral calcinosis, dysplasia epiphysealis multiplex, and chondroplasia punctata, primary aseptic necrosis.

Treatment should be undertaken if the lesion is causing deformity, pain or interference with function. Treatment directed at correction of the deformity and improvement of function consists of excision of the mass or corrective osteotomy. If the cartilaginous overgrowth is not in the weight-bearing articular surface, simple excision of the mass is indicated. If mass is intraarticular, it has been policy to evaluate the joint by arthrogram. If the arthrogram showed that the joint surface was smooth, extraarticular osteotomy was indicated for correction of varus-valgus deformities. Recurrence of the angular deformity after corrective osteotomy may be anticipated if the growth plate at the affected joint is open and active.

References