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**Case Report**

# Dysplasia Epiphysealis Hemimelica

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**Abstract:** Dysplasia epiphysealis hemimelica (DEH) is a developmental disorder with a very low incidence. It is characterized by overgrowth of bone and cartilage in one or more epiphyses. The usual presentation of disease is in a single limb and is hemimelic (involving either lateral or medial compartment). Lower extremities are more frequently affected. Here we present a case of a male patient at 2.5 years of age presenting with bilateral involvement of knee joints by DEH. The diagnosis was confirmed on both radiological and histopathological findings.

## Introduction

Dysplasia EpiphysealisHemimelica (DEH) also known by the name of Trevor Fairbank disease is an exceptionally rare disorder. The estimated incidence is 1:1,000,000. This disease usually affects children between ages of 2 to 14 years. Males are more commonly affected with a male to female ratio of 3:1. Most commonly affected site is ankle followed by knee joint. The etiology of this disorder is unknown. The disease is characterized by cartilaginous growth at the epiphyseal region. Grossly and microscopically the lesion resembles an osteochondroma. The classic form of DEH is asymmetrical and hemimelic.

## Case Report

This 2.5-years-old boy was admitted with a complaint of slowly growing swelling on both knees of 1 year duration. On physical examination painful swelling was palpable on the inner aspect of both knees (Fig. 1 & 2). Motion was restricted at the both knee joints. The laboratory findings were normal. X-ray showed bilateral effusion in knee joints. Patient was initially managed for 8 months as a case of septic arthritis. Patients symptoms failed to subside despite therapy. The lesion was biopsied and sent for histopathology. On gross inspection four irregular white colored osteocartilaginous pieces measuring 2x2 cm were present. They were decalcified and submitted entirely in two cassettes. Microscopically lobules of mature, benign and hyperplasic cartilage were seen with focal areas of ossification (Figures 3 & 4). Based on morphology a diagnosis of benign cartilaginous lesion was given.

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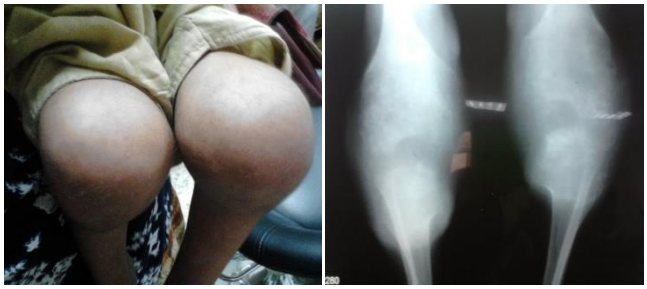
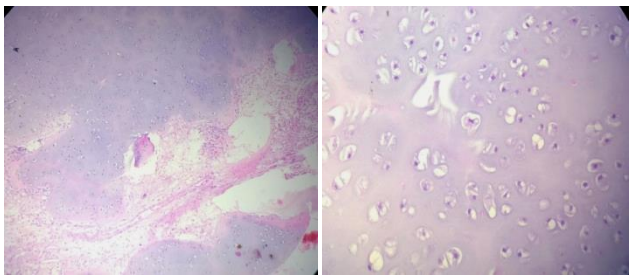
A repeat X-ray was also done. The X-ray picture showed expansion of epiphysis of bilateral distal femur, proximal tibia and patella. They appeared to be partially calcified with secondary metaphyseal irregularities. (Fig.3). It was therefore concluded that these findings are in favour of dysplsia epiphysealis hemimelica (DEH) keeping in view the biopsy proven diagnosis of cartilaginous lesion.



**Figures 1 & 2: Gross and radiological appearance of lesion, showing bilateral enlargement of knee joint.**



**Figures 3 & 4 : Lobules of mature, benign cartilage (H&E X 40 & 400 respectively)**

## Discussion

Dysplasia epiphysealis hemimelica (DEH) synonymously named Trevor-Fairbank disease, was first reported in 1926 by Mouchet and Belot.[1] The term dysplasia epiphysealis hemimelica (DEH) was introduced in 1956 by Fairbank because this skeletal disorder also involves other joints and only half of the

entire epiphysis.[2] The manifestations of the disease are asymmetric osteocartilaginous overgrowth of the epiphysis. Patient usually presents with skeletal deformity, pain, and restricted joint movement. The etiology of DEH is still unclear. It has been postulated that the disease might be associated with a congenital defect affecting the early stages of limb development during fetal life.[3] The bones most frequently affected by DEH are of lower extremity including femur, tibia, fibula, and tarsus.[4] The pattern of involvement is usually unilateral and hemimelic (affecting either lateral or medial compartment). Bilateral involvement does occur but is exceedingly rare. [5,6]

DEH presents clinically as a hard painless mass contiguous to the affected joint, with progression of disease deformities develop[3]. On radiography , DEH is seen as a partly calcified or ossified mass that is lobulated and involves the epiphysis.[7] Either the medial or lateral side of epiphysis is affected, but medial side is more frequently affected.

Histological features of DEH are similar to osteochondromas. The lesion is composed of normal looking chondrocytes lying in a cartilaginous matrix, with occasional cellular foci exhibiting mild to moderate atypia. Osteochondroma differs in being a benign osteocartilaginous tumor arising from the metaphyses and harboring a mutation in the EXT gene family.[8] No EXT1 and EXT2 gene mutations have been found in DEH patients [19]. Some morphological distinguishing features of DEH are chondrocyte clumping in a fibrillary matrix and focal endochondral ossification.[9,10]

Therapeutic approaches to DEH are dictated by clinical symptoms. Management is conservative during early stages, surgical treatment is recommended for progressively increasing lesion size, joint deformity, or pain.[3,10,11]. Surgical management includes correction osteotomies, surgical excision, arthrodesis, or chondroplasty.[2]. Prognosis depends on the location and size of the lesion and the degree of involved articular surfaces. Diagnosis of DEH at a very young age may be difficult due to the smaller sizes of the epiphyses and insufficient ossification.

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