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Dysplasia Epiphysealis Hemimelica of the Knee Joint: A Case Report

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Abstract

Background: Dysplasia epiphysealis hemimelica is a rare non hereditary epiphyseal disorder characterized by irregular overgrowth of cartilage in the epiphysis. The disease mainly targets distal long bones of the lower extremities e.g. ankle joint and tarsal bones. The guidelines for treatment of dysplasia epiphysealis hemimelica are sparse in literature due to the rarity of the syndrome. We report one such case manifesting in the right knee of a 5-year-old girl, a site which is not commonly reported and therefore prone to missed diagnosis and hence recurrence, along-with its surgical challenges , addressing all the anatomical and biomechanical derangements along with limb length discrepancy and to restore movements.

Keywords: Dysplasia epiphysealis hemimelica , irregular overgrowth , epiphysis, rarity.

Introduction

Dysplasia epiphysealis hemimelica (DEH) is a rare non-hereditary epiphyseal outgrowth of unknown etiology that mimics synovial chondromatosis of the joints [1]. The disease mainly targets long bones of the lower extremities and tarsal bones[1-3]. We report one such case from our institution manifesting in the right knee of a 5 year-old girl. Literature shows approximately 70 cases reported around the knee region since its first description 88 years ago.

Case Report

In February 2013, a 5-year-old girl presented with a 1½ year history of a gradually increasing swelling [Figure 1a] at the right knee and painless limp. There was no other remarkable history: family or past history. Patient had a history of a previous excision on the lateral side of the same knee with recurrence of deformity within six months. However, there was no history of loss of movement at the time of previous surgery. On examination, there was an irregular bony swelling 15 cm by 10 cm by 10 cm on the antero medial aspect of right distal femur and proximal tibia. It was associated with a 1.5 cm shortening of each of the right femur and right tibia. There were no warmth, tenderness or skin changes over the swelling and the knee had 60º fixed flexion deformity [Figure 1b] with further painless motion up to 120º associated with crepitus. The patient also had a genu valgum of 20 degrees which was chiefly femoral. The patella was small and dislocated laterally. There was no distal neurovascular deficit. There were no deformities at the ipsilateral hip and ankle nor at the contralateral hip, knee and ankle. Radiographs [Figure 1c] showed an ossified mass over anteromedial aspect of right knee. 3D CT[Figure 1d] and MRI [Figure 1e] showed a distinct plane of separation between the lesion and the normal epiphyseal bone.

Arthroscopy revealed that menisci and both the cruciate ligaments were intact and medial proximal tibial articular surface in the weight bearing domain was involved. The mass was too large for piecemeal excision and hence open surgery was performed immediately. Through an anterior mid line approach the right knee extensor mechanism was exposed [Figure 2a] which was found to be dislocated laterally by the mass. A 5cm by 3cm mass was found protruding out of the inter-condylar notch [Figure 2b] which was excised. The excised material [Figure 2c] was a globular bony mass covered by a smooth white glistening cartilage surface. There was involvement of the medial tibial articular surface as well in the form of 1.5cm by 1cm small mass [Figure 2d] which was shaved off. Lateral release and medial plication [Figure 2e] was done to realign the extensor mechanism before closure [Fig 2f]. A back slab was given post operatively for 15 days and then physiotherapy was started. The patient was discharged 22 days after surgery. At the time of discharge there was a 20º flexion contracture with further ROM upto150º and active knee extension from 90º to 60º was achieved. At 2 years follow up the patient was found to be almost completely relieved from the deformity [Figure 3a] and further correction of limb length discrepancy [Figure 3b] is planned.

Discussion

The incidence of this entity was reported as 1 in1000,000[4]. It was first described as tarsomegalie in 1926 by Mouchet and Belot[5]. Trevor used the term tarso-epiphyseal aclasis in 1950[6]. Since then this abnormality is commonly referred to as Trevor's disease. The term dysplasia epiphysealis hemimelica (DEH) was coined by Fairbank[1] in 1956. The word hemimelica is derived from 2 Greek words, hemi (half) and melos(limb). According to Fairbank, DEH is confined to the medial or lateral half of an epiphysis of a single limb it has a male-to-female ratio of 3:1[4].

The etiology of DEH is unknown. There is no strong evidence to suggest a hereditary component. It has been

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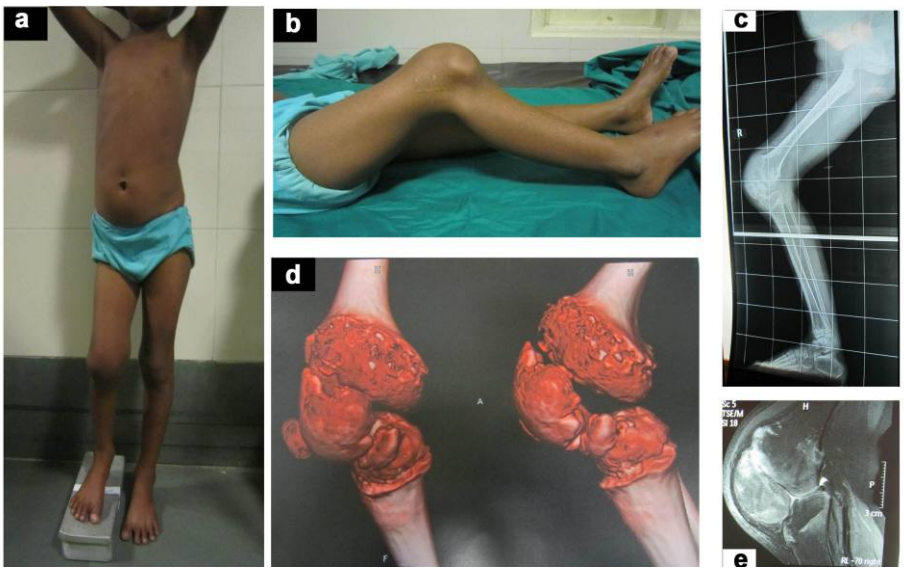

Figure 1: preoperative photographs showing clinical pictures (a,b), X ray (c), CT (d), MRI (e) scans.

hypothesized that this condition represents a fundamental defect in the regulation of cartilage proliferation in the affected epiphyses. Azouz et al [8] introduced a 3-group classification: group 1, localized, in which only one epiphysis is affected; group 2 (most common), classic, in which more than one epiphysis in the same limb is affected; and group 3, generalized, in which the whole lower limb is affected. Our patient in discussion fits into group 2.

Epiphyseal cartilage capped benign over growth is the unique feature of DEH. Struijs et al [9] showed the rarity of this disease in this location in a systematic review. They found a total of 48 studies having 138 patients with 255 lesions. Most lesions were located in the ankle or foot (139 of 255), and the talus was the most frequently affected bone. Rosero et al [10] showed only 21% distal femur and 11% proximal tibia were involved in their study of 57 patients.

The most common presenting symptom is a painless mass around the affected joint. The diagnosis can be guided by imaging studies. Initially radiography shows stippled calcification at epiphysis region. Eventually it looks like exostosis. It is possible that the earlier surgeon in this case confused it as a simple exostosis and excised it through a lateral approach. 3D CT scan should be done to assess the continuity of the lobulated mass with the underlying epiphysis. MRI is mandatory for identifying the extent of epiphyseal involvement, joint deformity and any effects on surrounding soft tissues. Teixeira et al. (2001) [11] reported the role of bone scintigraphy as increased uptake in the pathological epiphyseal area. It is useful check for other sites of involvement.

DEH is benign and its prognosis is favourable; no malignant transformation has been reported [6, 12].

Histopathologically, it was not possible to distinguish DEH from osteochondroma [3]. But genetic expressions (EXT1, EXT2) can be helpful [13].

The literature shows evidence that recurrences are more likely in patients with open physes at the time of surgery or after incomplete resection [1, 4]. There is no literature support to guide the management of limb-length discrepancy in this disease either by same limb lengthening or contralateral limb epiphysiodesis. It is also not described how to manage residual deformity. So the patient was counseled regarding the need for a long follow up with the possibility of further intervention.

Conclusion

Dysplasia epiphysealis hemimelica is a relatively rare disease, but the numbers of reported cases are gradually increasing. Hence, during the examination of a paediatric patient with a swelling of either the medial or lateral half of a joint, or a swelling that appears bony, painless and intra articular it is necessary to include DEH in the differential diagnosis. Provisional diagnosis can be made by clinical examination, surgical treatment is mandatory when symptoms like pain, joint impingement or deformation are present. Surgical prognosis is favourable when the mass is juxtaarticular or extraarticular. When the mass is intraarticular, early surgery may cause secondary osteoarthritis.

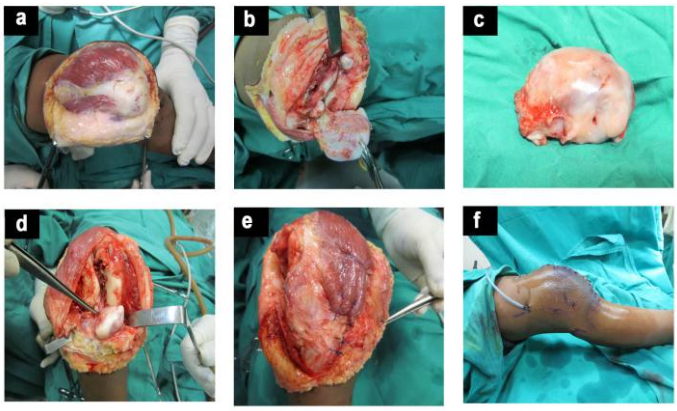

Figure 2: Surgical steps showing exposure (a), femoral mass (b), excised mass (c), tibial mass (d), medial plication (e) and closure (f).

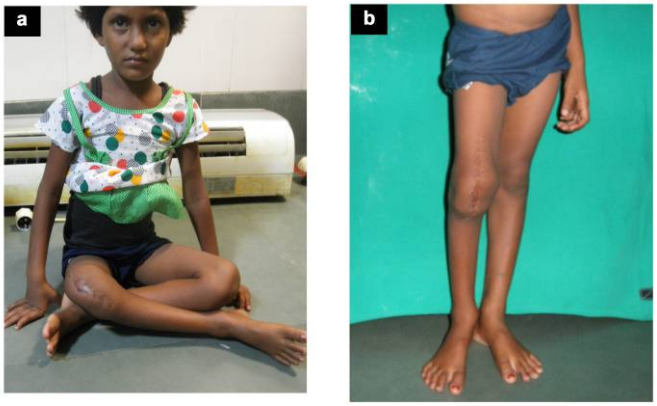

Figure 3: Follow up pictures (a,b).

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