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Case Report
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Trevor Disease: At Lower Tibia with Literature Review

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Abstract

Background: Trevors Disease is a rare developmental bone disorder of childhood. The bones of the ankle joint are most commonly affected. Of them Tibial involvement is less common as compared with tarsal involvement. We present a localized form Trevors Disease of lower tibial epiphysis.

Methods: A 9 year old who previously does not had any history of trauma presented with bony swelling from the anteromedial aspect of the lower tibial epiphysis with limitation of ankle dorsiflexion. The patient underwent radiological imaging followed by surgical excision and tissue biopsy for establishing the diagnosis. Additionally we collected all the reports of Trevor disease.

Results: At one year follow up there is no recurrence, restriction of movements or ankle joint dysfunction in our case.

Conclusions: The present study reports a case of localized form Trevors Disease at the lower tibial epiphysis with one year follow-up. A detailed review of Trevor disease was also performed regarding the treatment, prognosis, long term function and the areas affected.

Keywords: Ankle; Tibia; Epiphysis; Dysplasia epiphysealis hemimelica; Trevor disease

Introduction

Dysplasia epiphysealis hemimelica (DEH), also known as Trevor disease is a rare skeletal development disorder of childhood. It characterized by asymmetrical abnormal growth of epiphyseal cartilage particularly of the lower limbs. The knee and ankle joints are most commonly affected, apart from the bones of foot. The upper limbs involvement is extremely rare. Clinically and pathologically Trevor disease has much resemblance with epiphyseal osteochondroma. Mouchet and Belot was first described this condition in 1926, they called it tarsomegaly [1]. Later Trevor recognized this condition in 1950, who described 10 cases and put them into one distinct group, naming it tarso epiphysary aclasia [2]. Then Fairbank renamed Trevor disease as Dysplasia epiphysealis hemimelica to avoid confusions of the previous unsuitable designations. Trevor is characterized by the presence of irregular cartilage overgrowth occurs either in the lateral or medial part of the bone (hemimelic) usually medial. It may be a localized form (affect a single bone, classical form (multiple bones in a single limb) or generalized form (an entire limb) [3]. The ossification centers develop during growth period being individually or together. It results in a bone mass as asymmetrical epiphysis resembling exostoses [4]. The aim of our study was to perform a detailed review of this challenging disease, even though it is rare.

Case Report

A 9year old boy presented with pain and a swelling and limitation of dorsiflexion of right ankle in January 2015. He was walking normally except for uphill due to reduced ankle dorsiflexion. Other than this he was very healthy and there was no history of trauma. Physical examination showed bony hard swelling on the anterior aspect of right ankle joint which was blocking the dorsiflexion (Figure 1). There were no ankle or foot deformities.

Plain X ray of the right ankle joint shows irregular cartilaginous mass with dense areas similar to osteochondroma arising from the anterior part of the tibial epiphysis (Figure 2). The fibula, the talus, and the other foot were normal. MRI shows bony lesion from the anteroinferior part of lower tibial epiphysis with epiphyseal irregularity consistent with Trevor disease (Figure 3).

Since the clinical and radiologic findings were corroborative with Trevor's disease and the swelling was blocking dorsiflexion, patient underwent surgical procedure through the anterior

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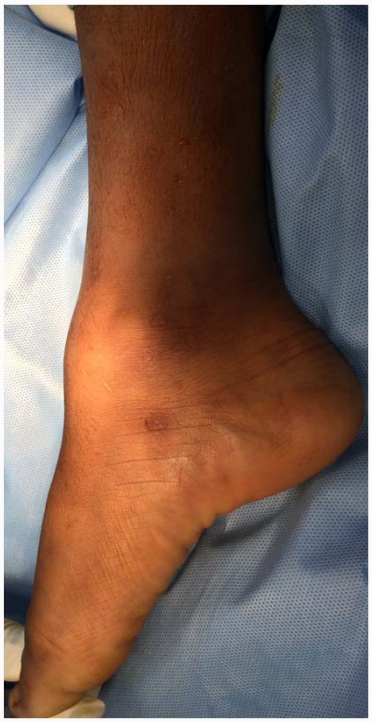

Figure 1: Swelling on the anterior aspect of right ankle joint which was blocking dorsiflexion.

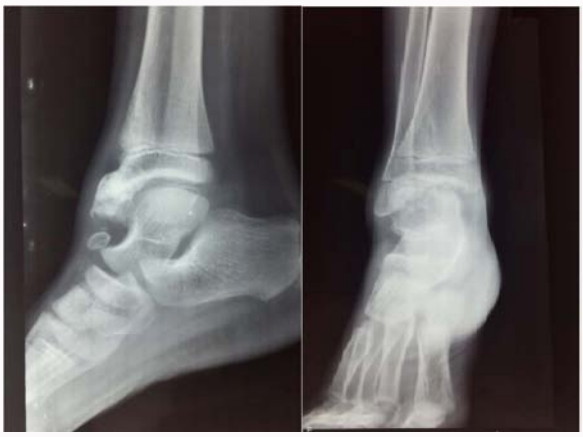

Figure 2: X ray right ankle joint AP and Lateral view shows irregular cartilaginous mass with dense areas arising from the anterior part of the tibial epiphysis.

approach, under general anaesthesia and tourniquet (Figure 4).

The lesion was removed using a broad osteotome from the anterior and antero medial part, near to the medial malleolus (Figure 5). There was a cartilaginous loose body obtained near to the tibialis anterior tendon.

Intraoperative dorsiflexion and plantar flexion were checked and found to be perfectly normal. Articular surfaces were also intact.

The histopathological examination report of the removed osteochondral fragments was consistent with benign osteochondroma. Mitotic figures or cellular changes of malignant transformation were absent.

In the post operative period weight bearing was prohibited for 4 weeks, followed by physiotherapy. No intra-operative or immediate

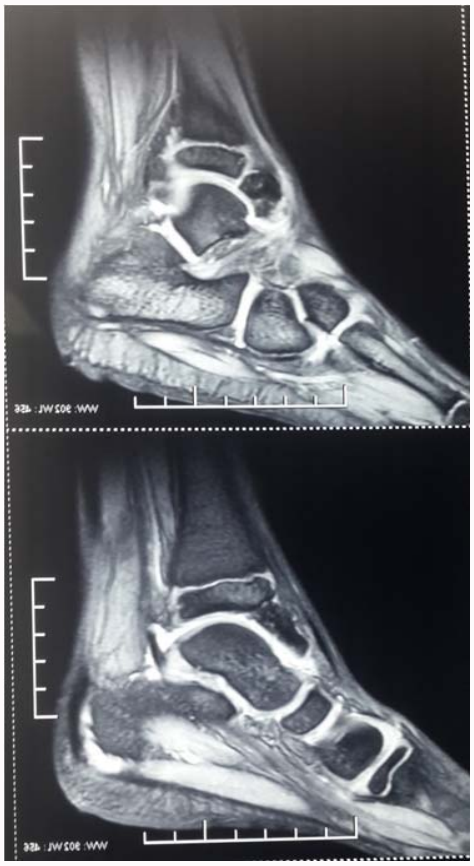

Figure 3: MRI shows bony lesion from the anteroinferior part of lower tibial epiphysis with epiphyseal irregularity.

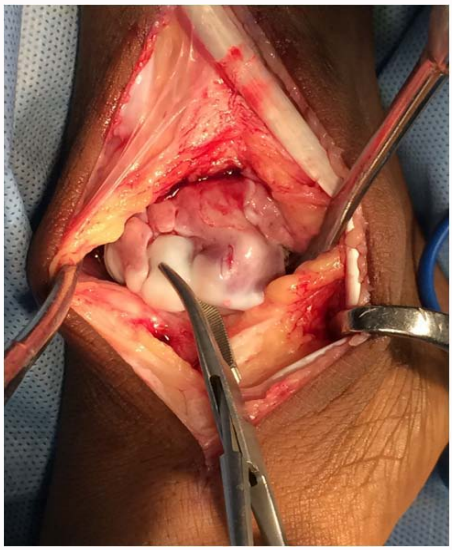

Figure 4: The irregular cartilaginous mass exposed through anterior approach.

post-operative complications were noted. At 1 year follow-up, the boy presented with no pain, no disease recidivism and complete joint function (Figure 6).

Discussion

The etiology of Trevor disease is still unclear. Insult during the formation of the limb bud was the reason as reported by David

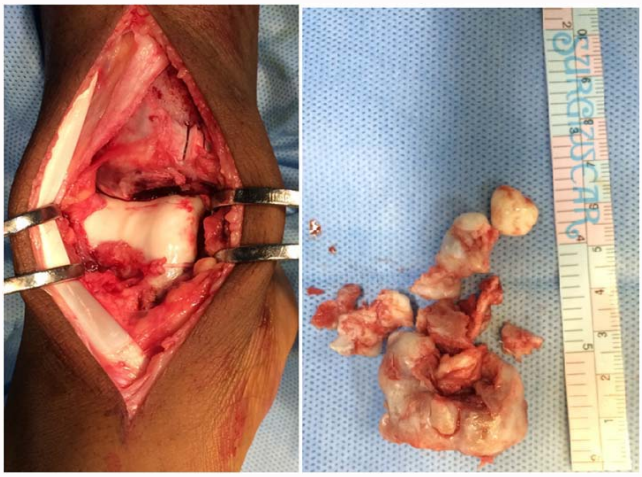

Figure 5a: Per operative view after the excision from the anterior and antero medial part.
Figure 5b: Removed cartilaginous mass.

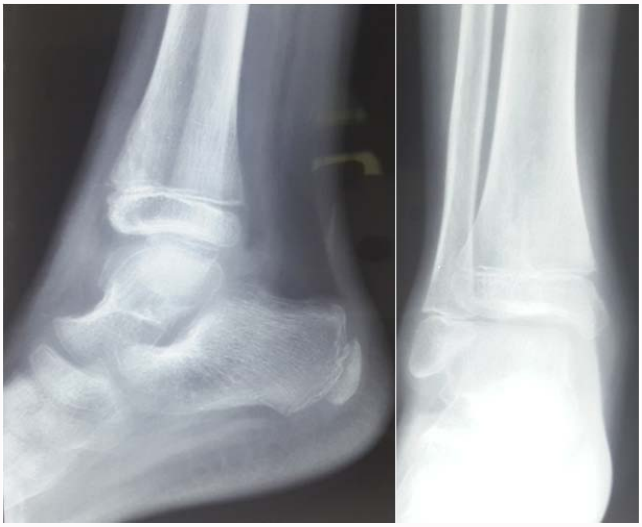

Figure 6: X ray right ankle joint AP and Lateral view shows no disease recidivism.

Trevor's [2]. Faulty growth of a part of the epiphysis or "a true dysplasia" itself was the aetiology believed by Fairbank [5].

It results from an abnormal control of cell proliferation from the affected epiphysis. The irregular cell proliferation occurs in the superficial zone of articular cartilage which causes changes in apical development button of lower limb and blood epiphysary vessel arrangements [2,5]. It also causes an imbalance between proliferation and cellular death mechanism. [2] There are changes in fetal limb apical ectoderm, resulting in pre or post-axial disorders [2].

Hence there is overgrowth with a cartilage cap that projects into the adjacent joint [6]. Even though Trevor disease belongs to the group of skeletal osteochondromas its incidence is rare. The reported incidence of it is 1: 1,000,000 [7]. Apart from a single report of familial incidence by Hensinger, Trevor disease is considerd neither as genetic nor any common environmental factor in its aetiology [6,8]. It is believed to affect the limb bud in early fetal life. Generally it affects children of 2 and 14 years and is most frequent in boys.

The descending order occurrence in the bones are distal tibia and fibula (22%), talus and calcaneus (22%), distal femur (21%), proximal tibia (11%), navicular, cuboid, cuneiforms, (10%), scaphoid (2%) and scapula (1%)[5]. Hemimelic denomination came due to its involvement only in one limb. Medial epiphyseal involvement is two times more than the lateral [5].

Trevor disease is characterised by an asymmetrical limb deformity because of the localized overgrowth of cartilage. Histologically it resembles osteochondroma. Ankle or knee regions are the most commonly affected areas and it always confined to a single limb. This usually involves only the lower extremities and on medial side of the epiphysis as in our case. Painless mass or swelling on one side of an affected joint, particularly the medial side with mechanical limitation of movement is the most common symptom. Pain usually occurs at a later stage of the disease. Additional symptoms in each child vary depending on the size and location of the cartilage mass, like joint deformity, limb length discrepancy, locking of the joint and muscle wasting, which often results limping. After the epiphyseal closure, the disease stops its progression.

In untreated case the cartilage mass in the joint will increase in size and undergo ossification to form a osteocartilaginous mass and ultimately results degenerative arthritis. Degenerative findings are more common in older patients and children usually present with pain, edema and joint stiffness in Fairbank findings [5].

Several epiphyseal diseases are presenting with the same radiological findings so final diagnosis of HED without more specific examinations is difficult. Irregular and multicentric epiphyseal opacities are common for both. Normal bone mass covered by abundant cartilaginous foci of endochondral ossification, resembling osteochondroma is the histological finding. Histological it is not possible to differentiate Trevor disease from osteochondroma [5].

Genetic expression analysis of EXT1, EXT2 genes are very much helpful. These genes are within normal ranges in Trevor disease, while they are lower in ostechondroma (owing to a mutation) [9]. Since these genetic tests are expensive, the diagnosis is often made on clinical and radiological findings. The differential diagnosis of Trevor disease includes posttraumatic osseous fragments, synovial chondromatosis, ostechondroma, or anterior spur of ankle [9]. Synovial chondromatosis can be ruled on the basis that it occurs in much older age group [9]. Multiple epiphyseal changes can be produced by other conditions like punctate epiphyseal dysplasia, achondroplasia and aseptic necrosis which is also comes in the differential diagnosis. Osteochondroma and carpotarsal dominant osteochondromatosis are the articular calcifying tumors which most resemble Trevor disease [10]. There is no report of malignant transformation in the literature [11].

Computed tomography-scan is an excellent modality to define the boundary between pathological and normal bone tissues. It is very much helpful in the surgical planning [12]. Magnetic resonance imaging offer better soft tissue delineation and helps to identify the degree of soft tissues and epiphysis involvement. It has a great role in differentiating from other tumoral diseases [13]. Eventhough bone scintigraphy shows nonspecific findings; it can identify several HED affected loci.

The treatment of Trevor disease is not clearly described. It depends on the location, evolvement intensity and degree of functional incapacity. Asymptomatic patients are just followed up since malignant transformation is nonexistent [14]. Most reported

cases of Trevor disease in the literature have been treated surgically. Surgical excision is indicated in cases of pain, articular deformity or incongruence, or limitation of motion. Simple excision of the mass and correction of the deformity if present, while preserving the integrity of the affected joint surface as much as possible is considered as the standard treatment [9]. Acquaviva et al. [15] classified the injuries in Trevor disease as extra or intra-articular. A simple excision of extra-articular lesions offers good results than intra-articular lesions. A study done by Kuo et al. [14] concludes poor outcomes in intra-articular tumors. Arthrosis was a frequent complication in such patients. Complete excision of the lesion is mandatory for the prevention of tumor recidivism. Therefore, Irrespective of the anatomopathological type, it is essential to follow-up the patient until complete skeletal maturity [14].

Conclusion

Trevor disease is a rare developmental bone disorder characterized by an osteocartilaginous tumor arising from the epiphysis of long bones in children. Involvement of lower tibia is even rarer. Even though there is no gold standard treatment described in the literatures, the general conclusion is surgical treatment as the best.

We have described a case of Trevor disease of lower tibia in a 9 year old boy who was treated by simple surgical excision with a general review of this uncommon disease to help the orthopedic surgeon on the Trevor disease management.

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