Trevor's disease (dysplasia epiphysealis hemimelica) in a 9 years-old child: a case report

Doença de Trevor (displasia epifisária hemimélica) em criança de 9 anos: relato de caso

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ABSTRACT

We report a rare case of a 9 years-old boy with dysplasia epiphysealis hemimelica in the topography of the sustentaculum tali of the right ankle associated with pain and episodes of loss of balance. Treatment included surgical resection that resulted in complete improvement of pain and balance disorders.

Keywords:

Ephiphises/pathology; Bone diseases, developmental/diagnosis; Case reports

RESUMO

Relato de um caso raro de displasia epifisária hemimélica em uma criança do sexo masculino de 9 anos de idade, em topografia do sustentáculo do tálus em tornozelo direito, associada à dor e a episódios de perda de equilíbrio. O tratamento realizado foi baseado em ressecção cirúrgica, evoluindo com melhora completa das queixas álgicas e do défice de equilíbrio.

Descritores:

Epífise/patologia; Doenças do desenvolvimento ósseo/diagnóstico; Relatos de casos

INTRODUCTION

Trevor disease was initially reported by Mouchet and Belot in 1926 under the name of tarsomegaly,[1] and in 1950 it was described in the form of eight case reports by David Trevor, who called the disease tarso-epiphyseal aclasis.[2] In 1956, Thomas Fairbanks coined the term dysplasia epiphysealis hemimelica (DEH),[3] as the disease is known to date.

It is a rare benign developmental disorder that affects half of the epiphysis in children aged up to 14 years, with incidence of 1:1 million.[4] It is characterized by a localized osteochondral defect containing asymmetric proliferation of cartilage with endochondral ossification, most commonly affecting epiphyses of the knee and ankle.[5]

Histologically, the lesion is indistinguishable from the osteochondroma. It is essential not to confuse osteocartilaginous growth with DEH, which arises from the epiphysis, with exostosis originating from the metaphysis. Differential diagnoses include chondrodysplasia punctata and multiple epiphyseal dysplasia, which occur bilaterally.[6]

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This case report aimed to describe a clinical case of a 9-year-old male child monitored in a foot and ankle surgery outpatient clinic and discuss the pathology in question based on clinical and radiological findings.

CASE REPORT

A 9-year-old male child was referred to the Foot and Ankle Surgery Outpatient Clinic in September 2014 by another Orthopedic Department. Symptoms began in December 2013, with complaints of pain on the medial surface of the right ankle that worsened with the use of closed shoes, associated with frequent falls. The patient had no history of previous injuries, morbidities, or reports of hospitalization for any other pathology in recent years.

Clinical evaluation revealed the formation of a superficial medial tumor of firm consistency, adhered to deep planes, painful on palpation and no signs of neurovascular deficit (Figure 1). The American Orthopaedic Foot and Ankle Society (AOFAS) score for ankle and hindfoot came to 74 points. The additional tests requested were plain radiograph, computed tomography, and magnetic resonance imaging, important for defining the extent of the tumor, establishing its topography, and verifying the possibility of malignancy (Figure 2).

The plain radiographs showed increased medial volume with apparent involvement of the body of the talus, with no signs of tibiotarsal or subtalar joint involvement.

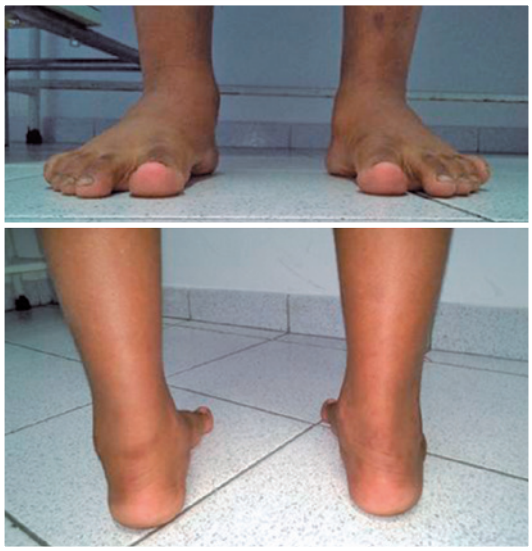

Figure 1 | A lump can be seen on the medial surface of the right ankle in the preoperative stage.

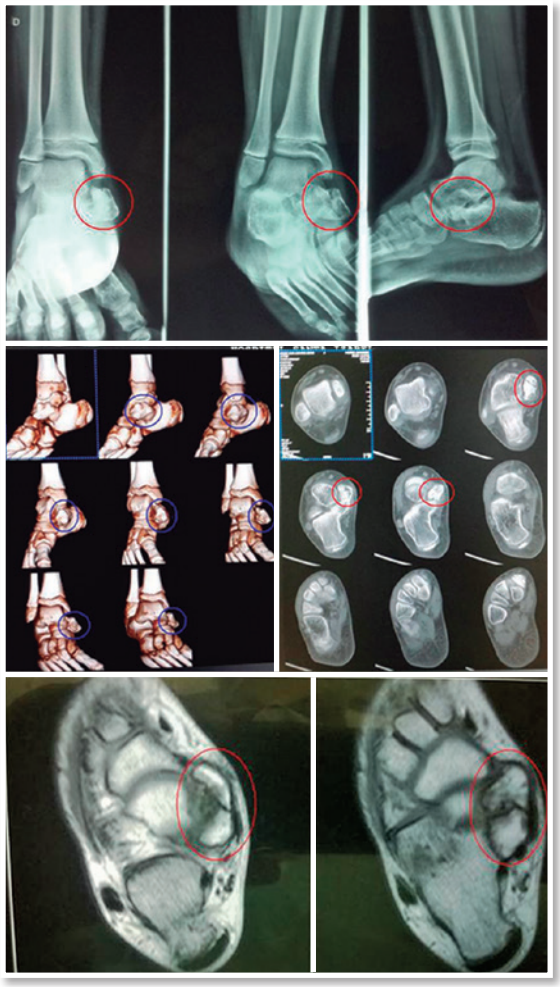

Figure 2 | Radiography, computed tomography and nuclear magnetic resonance.

The CT scan revealed that the lesion originated in the sustentaculum tali, with small areas of calcification adjacent to the lesion.

The MRI scan revealed an oval, juxta-articular tumor lesion, without involvement of the subtalar joint surface, not adhered to deep planes, without signs of expansile process and clearly delimited.

The clinical and radiological characteristics of the lesion were compatible with expansile bone lesion with osteochondroma-like features in the topography of the sustentaculum tali.

The diagnostic suspicion was of Trevor's disease, because it is a probable osteochondroma lesion located in the sustentaculum tali. As the patient was experiencing pain and having difficulty walking, we opted for surgical treatment in November 2014 with resection of the tumor and submission of a specimen for anatomical pathology testing.

In the intraoperative phase (Figure 3), we observed a juxta-articular tumor originating from the sustentaculum tali, which underwent bloc resection, with preservation and total release of the subtalar joint. The measurements of the anatomical specimen were 3.3x2.7x1.8cm. Histopathological analysis revealed bone tissue without atypia with a peripheral coating comprised of cartilaginous tissue also without atypia. The conclusion reached was that it was an osteochondroma lesion.

The patient progressed satisfactorily with complete improvement of pain, no motor or sensory abnormalities, and no impairment of the range of motion or gait, as evidenced in the last review performed in the third postoperative month (Figure 4). The postoperative AOFAS score for ankle and hindfoot was 100 points.

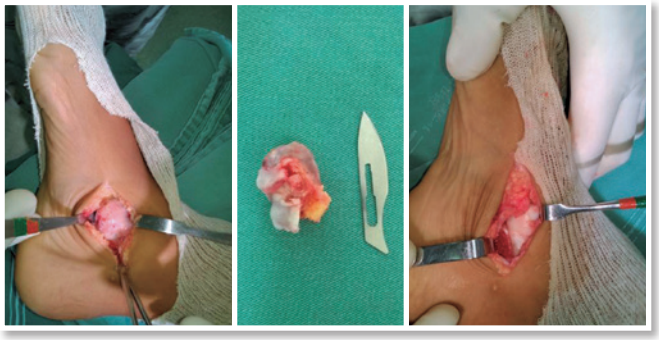

Figure 3 | Surgical resection. Medial approach to the sustentaculum tali, with complete resection of the tumor, without involvement of adjacent structures.

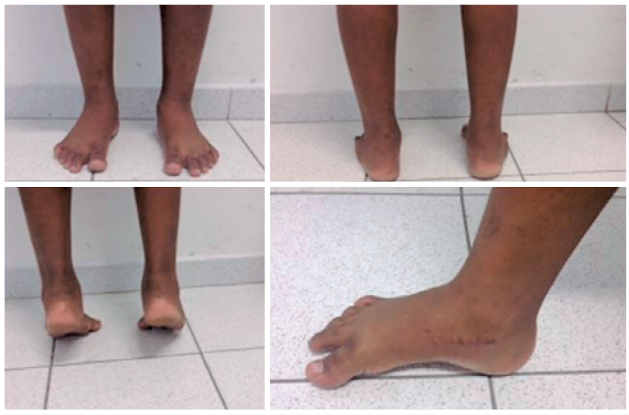

Figure 4 | Third month of postoperative follow-up.

DISCUSSION

DEH is uncommon and there are few case reports in the national and international literature. It is estimated that about 250 cases of the disease have been documented to date, yet this figure may be underestimated by undiagnosed cases because most of them are asymptomatic or due to the simple diagnosis of osteochondroma not associated with the unique characteristics of the disease. The highest rate of findings of this pathology is in the 2-14 year age group, predominating in men over women at a ratio of 3:1.[7]

The knee joint (distal femur and proximal tibia) has a greater number of occurrences followed by the talus and the joint between the navicular and the medial cuneiform bones.[8] In the literary review performed by the authors, no descriptions of DEH were found in topography of sustentaculum tali, as described in this report. Azous et al. described a classification for DEH, with three variants, based on clinical presentation: localized (in cases where only one bone is affected); classic (where there is involvement of more than one bone in the same limb) and generalized (in which there is involvement from the pelvis to the foot). In the case in question, we classified the pathology as localized. It is important to determine its articular relationship, which may be intra- or juxta-articular, a fact that interferes in the post-resection prognosis, since joint involvement is associated with chondral lesion and early arthrosis. The medial side is more frequently affected than the lateral side, but without clinical relevance.[9]

The etiology of the disease is still unknown.[10] Different theories not yet proven attempt to explain the appearance of the pathology as irregular cell proliferation in the superficial zone of the articular cartilage; changes in the epiphyseal vascular arrangement; imbalance between the mechanism of proliferation and cell death; abnormalities in the apical portion of the ectoderm in fetal development, resulting in pre- and post-axial disorders, which, according to Trevor, would determine the medial or lateral involvement of the disease. There are no reports of genetic transmission.[8]

The most frequent clinical symptom of the disease is pain related to the slow growth of the tumor mass most commonly located in the medial portions of the knee and ankle. Fairbank et al., in their reports, observed edema, joint weakness, and angular deformities of the affected joints.[3] Growth of the lesion is interrupted when there is epiphyseal closure and skeletal maturity is reached.[4,7]

The diagnostic methods used most frequently are plain radiography, computed tomography, and magnetic resonance imaging. The radiograph shows one or more irregular masses near the border of one of the halves of the epiphysis in addition to epiphyseal enlargement. Tomography helps to determine the anatomical relationships between

the tumor mass and bone, as well as whether there is continuity with the cortical bone, but it is inferior to MRI, which defines the bone and cartilaginous structures better in multiple planes. Histopathological study is only imperative in cases where surgical treatment is recommended or in cases with atypical radiological presentation.[7,8]

Surgical treatment is indicated in the presence of pain, joint deformities or limited mobility. The identification and complete excision of the tumor are essential for the prevention of its recurrence, common to osteochondromatosis cases, but without statistical data when related to DEH. Extra-articular tumors have a better prognosis than those resected on joint surfaces - these may have poor outcomes such as osteoarthrosis. There are no reports of malignization of this type of lesion. Anatomical pathology testing corroborates typical findings of chondromatosis lesions.

CONCLUSION

Trevor disease or dysplasia epiphysealis hemimelica is a rare disease characterized by asymmetric epiphyseal growth of the epiphyseal cartilage.

The low incidence of the pathology prevents the execution of clinical studies with a greater number of patients. Thus, case reports should allow further knowledge of the disease, allowing diagnosis and treatment protocols to be established for the pathology.

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