Analysis of dysplasia epiphysealis hemimelica (Trevor’s disease) of the ankle

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ABSTRACT
Dysplasia epiphysealis hemimelica or Trevor’s disease is an osteocartilaginous deformity in the bone epiphyseal area. It is an uncommon condition prevailing in males. It develops in childhood, with growth plates still open, and it affects mainly patients’ ankles and knees. It is a condition of unknown origin.

We present three cases with different degrees of severity, and also the therapeutic alternatives for each. There is only one case subject to surgical treatment due to irreducible equinus deformity. We provide details about surgical techniques, postoperative management and results in histologic analyses. We recommend operating on only those patients with some functional limitation or severe deformity due to the high recurrence rates they are associated with.

Key words: Trevor’s disease; epiphyseal osteochondroma; ankle; endochondral ossification.

Level of evidence: IV

ANÁLISIS DE LA DISPLASIA EPIFISARIA HEMIMÉLICA (ENFERMEDAD DE TREVOR) DE TOBILLO

RESUMEN
La displasia epifisaria hemimelica o enfermedad de Trevor es una deformidad osteocartilaginosa en la región epifisaria. Es poco frecuente y predomina en el sexo masculino. Se desarrolla en la infancia cuando los cartílagos de crecimiento están abiertos, y afecta principalmente el tobillo y la rodilla. Su origen es desconocido.

Se presentan tres casos con distinto grado de compromiso y las alternativas terapéuticas. Un solo caso quirúrgico por equino irreductible. Se detallan la técnica quirúrgica, el manejo posoperatorio y el resultado de anatomía patológica. Se recomienda operar sólo a pacientes con alguna limitación funcional o severa deformidades por el alto índice de recidiva.

Palabras clave: Enfermedad de Trevor; osteocondroma epifisario; tobillo; osificación endocondral.

Nivel de Evidencia: IV

Conflict of interests: The authors have reported none.
Introduction

Dysplasia epiphysealis hemimelica is a very uncommon entity, with a 1:1,000,000 incidence, which prevails in males (3:1 male: female ratio) and whose onset if between 2 and 14 years old. The ankle joint is the most frequently affected (54%), followed by the knee joint.1-5

This condition is characterized by osteocartilaginous overgrowth from one side of the epiphysis (lateral or medial sides, what defines the condition as “hemimelica”). The medial compromise of the epiphysis doubles its lateral compromise. These lesions are considered as epiphyseal osteochondromas; up to now, there are no reports on malign degeneration.6

Depending on their location, lesions can be articular or juxta-articular lesions. The latter are associated with better prognosis.7

Reasons for consultation are usually tumor, deformity, mobility limitation and joint rigidity.

Nowadays there are no standardized treatment guidelines, but most authors agree on surgical treatment as the indication in cases of pain, deformity or mobility limitation. We should bear in mind that these lesions are associated with high recurrence rates after resection, and they may take several interventions.8

The aim of this article is to describe onset characteristics and therapeutic suggestions in this condition.

Case 1

It is the case of a sixteen years old male who consults for painless ankle deformity, with no functional limitation (Figures 1-3).

▲ Figure 1. Case 1. Left ankle valgus deformity.

▲ Figure 2. Case 1. AP and lateral X-rays of both ankles.
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Case 2

It is the case of a forty-two years old male with no symptoms who consults for a 13-year history of ankle deformity; without progressive increase in size (Figures 4 and 5).

Figure 3. Case 1. Radiographic check-up two years later.

Figure 4. Case 2. Left ankle oblique, lateral and AP X-rays. There is talar exostoses.

Figure 5. Case 2. CT scan. Lateral marginal exostoses at the level of the talar bone in association with the anterior talofibular ligament, which is incompletely outlined in this study. There is another area of dorsal exostoses in relationship with talonavicular and calcaneocuboid joints.
Case 3

It is the case of a twelve years old male with right knee retroversion while walking, and ankle deformity. Right elbow disorders which do not limit mobility, but elbow deformity. He required ankle surgery (Figures 6-8).

Surgical technique

Approximately 8-cm-long AM approach focused on the apex of the deformity. The osteochondroma is reached and resected with safety margins of 8 mm both in talar bone and the AM margin of the tibial pilon. We carry out percutaneous lengthening of the calcaneal tendon (Figure 9).

Figure 6. Case 3. Ankle deformity with no inflammatory signs.

Figure 7. Case 3. Right ankle AP and lateral X-rays. Altered tibial pilon and talar bone.

Figure 8. Case 3. CT scan. Hypertrophy in an ossification nucleus with adjacent mass of calcified soft tissues.
We immobilize patients’ ankle with 90º-casts during 15 days and, at postoperative day 15, we implement walker boot. Patients initiate articular amplitude exercises with physiotherapists depending on pain tolerance. There are no complications and patients progress to up to 90º-mobility ankles, decrease in hip and knee mobility, and walking comfortable status. We submit the resected material to histological analysis—cartilaginous tissues lined by a dense fibroconnective layer, with endochondral ossification and mature bone tissues, which represent osteochondroma (Figure 10).

Ethology is unknown. Connor et al. suggest that defects are due to the abnormal regulation of cartilage growth in the compromised epiphysis. There is no evidence of hereditary factors.

From the clinical point of view, this condition shows as tumor, deformity, mobility limitation and joint rigidity. Other symptoms are limping and limbs discrepancy. The overgrowth in limbs can be attributed to the growth of multiple epiphyseal centers or to the increase in blood supply to the areas operated on. In general, limb shortening is subsequent to the premature closure of growth plates, or it can occur as a surgical complication.

Discussion

Dysplasia was described in 1926 for the first time by Mouchet and Belot as “tarsomegaly”, making reference to its most frequent location. In 1950, Trevor coined the term “tarso-epiphyseal achalasia”. In 1956, Fairbank used the expression “dysplasia epiphyseal hemimelica”, which today is the commonest denomination.

It is considered an epiphyseal osteochondroma; from a histologic point of view it resembles osteochondroma from the epiphysis. There are no reports on malignant degeneration.
Nowadays, what is used is the Azouz et al.’s classification, which divides it into three groups: Group 1 or located lesion—compromise of just one epiphysis; Group 2 or classic lesion (the commonest one)—compromise of more than one epiphysis in the same limb; Group 3 or generalized lesion—the whole lower limb is compromised.

Keret et al. divide lesions into juxta-articular or articular lesions, instead of extra-articular or intra-articular lesions. “Extra-articular” suggests that the lesion is outside the capsule but, in their series of cases, all injuries were epiphyseal and intra-capsular; some of them were adjacent to the joint (juxta-articular lesions) whereas others compromised the articular surface downright (articular lesions).

Simple X-rays are usually sufficient to make diagnosis, if doctors are familiarized with this condition. X-rays show unilateral, lobular, partially ossified masses from the affected epiphysis with or without bone connection. Some radiologic images can be confusing, suggesting intra-articular free body, osteochondromatosis or synovial chondromatosis.

Among differential diagnoses we can mention multiple epiphyseal dysplasias, osteochondroma, multiple hereditary exostoses, and chondrosarcoma.

CT scan is mainly useful in preoperative planning, allowing surgeons to evaluate the size of the mass, its relationship with nearby bone and the joint status.

MRI allows us to evaluate the relationship between the mass and vital elements; moreover, it is very useful to determine the size of the mass, articular deformity and the status of the joint surface.

Most authors agree on surgical treatment for pain, deformity and mobility limitation.

If the option is resection, it should be complete; otherwise, all remaining affected tissues will continue growing and they will result in recurrence.

In case of deformity, mass resection can be supplemented by corrective osteotomy.

Authors such as Struijs et al. believe that, in view of the slightest functional limitation or moderate pain, the patient should be subject to resection. In their experience, this condition makes progress to mass growth with subsequent articular deformity. Complete resection of all the affected tissues is essential; otherwise, the residual tumor will continue growing with joint deformity.

Keret et al. report excellent results in the resection of juxta-articular lesions and bad results in intra-articular lesions resection. On the basis of their experience, they do not recommend the resection of articular lesions; at least they show as free bodies. They advise resection in cases of symptomatic juxta-articular lesions, and articular free bones.

Acquaviva et al. classify these lesions in extra-articular and intra-articular on the grounds of their location. In extra-articular lesions, treatment should consist of resection, depending on the case, with favorable results. In intra-articular lesions, osteotomy may be necessary to achieve angular correction in deformities.

According to Gökkus et al., if the injury is intra-articular, especially in knee, arthroscopy may be indicated to evaluate the surface of the intra-articular mass; if the mass is adapted to the articular curve it should not be resected, and hemiepiphysiodesis would be an option to correct deformity.

Masquijo and Baxter conclude that so far the treatment for this condition has not been defined, and the only ones that should be subject to surgical treatment are symptomatic lesions that interfere with function.

Bakerman et al. suggest yearly MRI for follow-up after resection to evaluate likely recurrence.

Conclusions

Dysplasia epiphysealis hemimelica is a very uncommon entity with ankle compromise. Treatment is usually conservative, unless articular function, two-feet weight-bearing or near joints are compromised.

In the patient we operated on, surgical indication was based on the patient’s irreducible equinus foot, and his gait impairment with flexed knee and hip.

Bibliography

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