Radiology Case Reports 17 (2022) 2277–2282

Available online at www.sciencedirect.com

ScienceDirect

journal homepage: www.elsevier.com/locate/radcr

Case Report

Hemimelic epiphyseal dysplasia: a case report ☆

Chaymae Hajjar, MD[a,*], *Meriem Haloua, MD*[a], *Nizar El Bouardi, MD*[b], *Badreeddine Alami, MD*[b], *Moulay Youssef Alaoui Lamrani, MD*[b], *Mustapha Maaroufi, MD*[b], *Meriem Boubbou, MD*[a]

[a] *Department of mother and children's radiology of Fez, CHU Hassan II, Sidi Mohamed Ben Abdellah University Fes, Fes, Morocco*

[b] *Radiology department of Fez, CHU Hassan II, Sidi Mohamed Ben Abdellah University Fes, Fes, Morocco*

ARTICLE INFO

Article history:
Received 2 March 2022
Revised 23 March 2022
Accepted 25 March 2022

Keywords:
Hemimelic epiphyseal dysplasia
Trevor's disease
Epiphysis, Imaging

ABSTRACT

Hemimelic epiphyseal dysplasia HED also known as Trevor's disease is a rare pathology, characterized by a developmental disorder of an internal or external half of one or more epiphyses of a limb, mainly the lower limb, and/or of the short tarsal bones in children and young adolescents, with a male predominance. Its etiology remains unclear. Its clinical symptomatology is variable, ranging from asymptomatic involvement to orthopedic complications such as limb length inequality. As the clinic is non-specific, radiological assessment is the essential diagnostic tool for Trevor's disease, including standard radiography, MRI, CT, and possibly biopsy in some cases. The radio clinical signs make it possible to establish the diagnosis, even if it remains difficult because of the rarity of the disease and the presence of multiple differential diagnoses which are often better known such as osteochondroma and exostosis. After diagnostic confirmation, the therapeutic decision remains debated, ranging from simple observation to surgical excision. The prognosis of HED remains good, given the absence of the risk of malignant transformation. Post-therapeutic complications are dominated by recurrence or the appearance of secondary osteoarthritis. Objective: This clinical case challenges us to keep in mind the hemimelic epiphyseal dysplasia (HED) in front of a mass that originates at the level of the internal or external half of one or more epiphyses in children.

Introduction

Hemimelic epiphyseal dysplasia HED also known as Trevor's disease is a rare pathology, characterized by a developmental disorder of an internal or external half of 1 or more epiphyses of a limb, mainly the lower limb, and/or of the short tarsal bones in children and young adolescents, with a male predominance. Its *etiology* remains unclear. Its *clinical symptoma-*

Abbreviations: HED, Hemimelic epiphyseal dysplasia; MRI, Magnetic resonance imaging; CT-scan, computed tomography.

☆ Competing Interests: The authors have declared that no competing interests exist.

* *Corresponding author*.
E-mail address: chaymae.hajjar@usmba.ac.ma (C. Hajjar).

https://doi.org/10.1016/j.radcr.2022.03.103

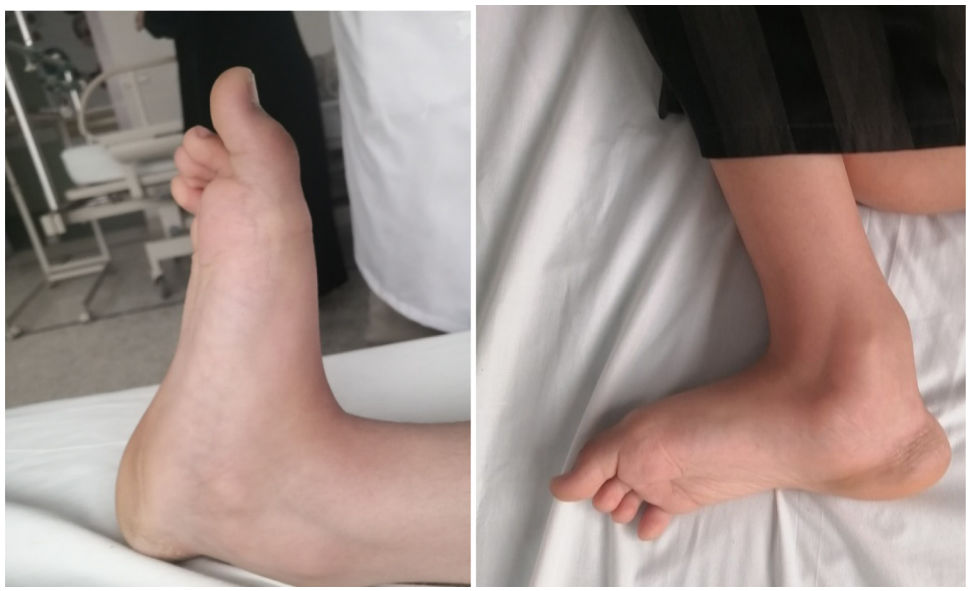

Fig. 1 – Clinical picture: axial deformity of the right ankle in varus with swelling of its internal face.

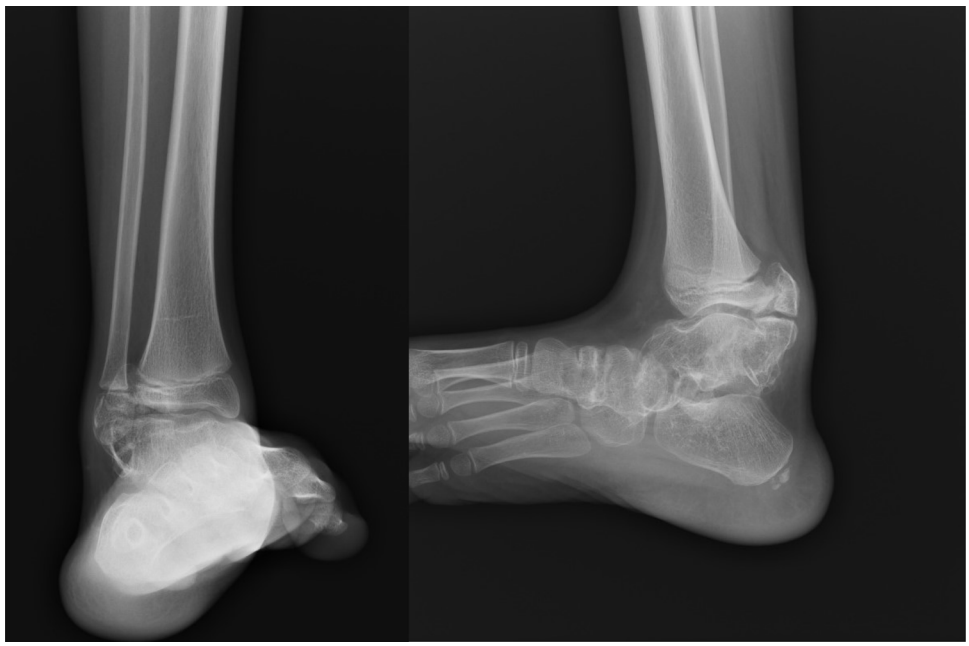

Fig. 2 – Standard radiograph of the right ankle showing an exophytic, intra-articularly developed bony growth located in the anteromedial portion of the lower end of the tibia and talus.

tology is variable, ranging from asymptomatic involvement to orthopedic complications such as limb length inequality.

As the clinic is non-specific, *radiological assessment* is the essential diagnostic tool for Trevor's disease, including standard radiography, MRI, CT, and possibly biopsy in some cases.

We report an observation of hemimelic epiphyseal dysplasia in a child with an axial ankle deformity, with the contribution of imaging in the diagnosis of this anomaly.

Patient and observation

A 10 years old boy, presented with a slowly progressive right ankle deformity. On examination, he had a history of a common injury to the right foot at the age of 5 years, causing pain on the medial side of the joint of the right ankle with limping, for which he received undocumented or-

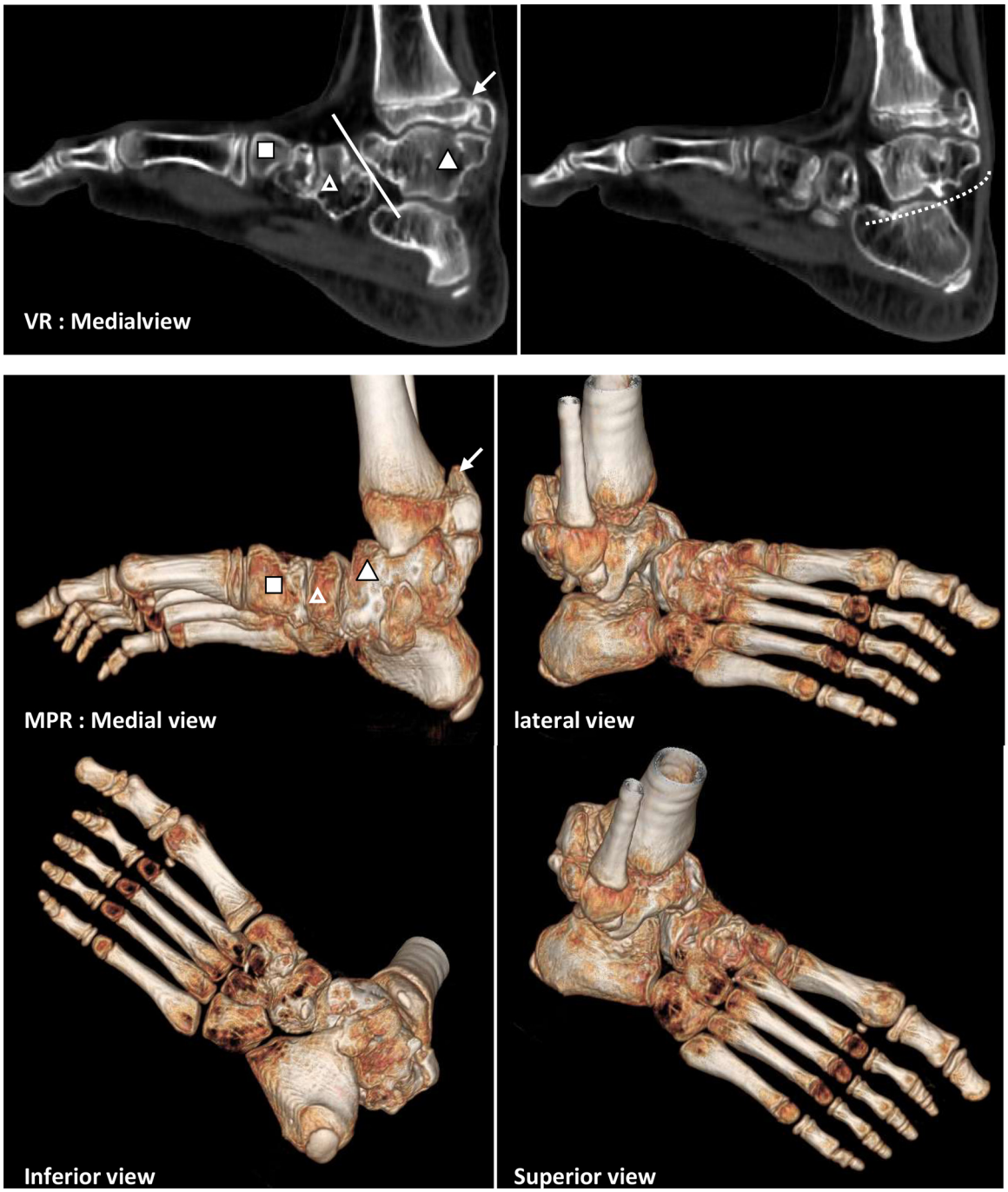

Fig. 3 – CT scan with multi-planar reconstructions showing multiple epiphyseal osteo-cartilaginous growths, some with a pedicle base and others with a large base, involving the medial part of the ankle bones, in particular the talus (white triangle), the scaphoid bone (hollow triangle), the medial cuneiform bone (white rectangle) as well as the inferomedial end of the tibia (white arrow), with intra- and juxta-articular development, in particular talocalcanean (dotted line) and taloscaphoid (white line), suggesting a hemimelic epiphyseal dysplasia.

thopedic treatment. The clinical symptomatology worsened with the appearance of an axial deformity of the medial aspect of the ankle, motivating his family to consult in our training.

Clinical examination revealed swelling of the medial aspect of the right ankle with a varus deformity of the foot. Physical examination revealed limited joint movement with limping walking. Palpation revealed severe pain on the medial side of the right tibio-astragalar joint. The rest of the musculoskeletal examination was normal; the rest of the somatic examination was without abnormalities (Fig. 1).

A standard radiograph of the right ankle was performed, showing an exophytic and intra articularly bony growth lo-

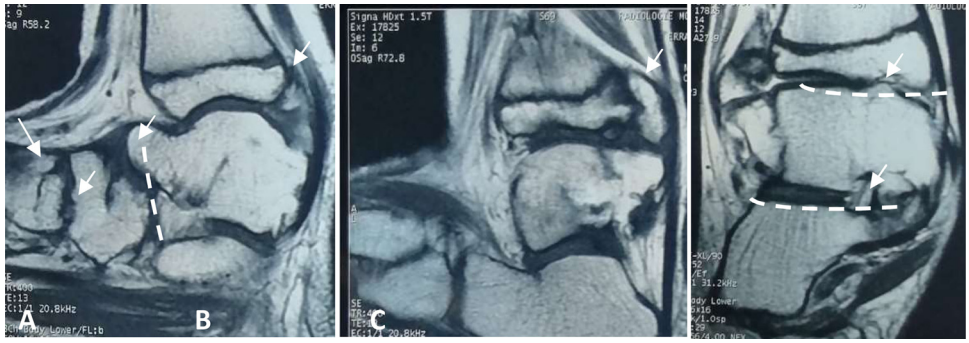

Fig. 4 – *Magnetic resonance imaging* with sagittal (A-B) and coronal (C) sections showing the presence of osteochondromatous growths with poly-lobed contours and a signal identical to that of the bone, some of which are intra- and juxta-articular, notably tibio-talar, talo-calcaneal and talo-scaphoid (dotted lines), opposite the postero-medial edge of the lower tibial extremity and medial bones of the ankle (white arrow) No abnormalities of the synovium ligaments and no signs of malignancy.

cated in the anteromedial part of the lower end of the tibia and talus (Fig. 2).

It was therefore a symptomatic intraarticular mass responsible for functional discomfort. To better define the nature of this mass and its anatomical relationships, a locoregional extension assessment was necessary. *A CT* with multi-planar reconstructions was performed showing multiple epiphyseal osteocartilaginous growths, some with a pedicle base and others with a large base, involving the medial part of the ankle bones, in particular the talus, the scaphoid bone, the medial cuneiform bone, and the lower medial extremity of the tibia, with intra and juxtaposed joint development, in particular the talo-calcaneal and talo-scaphoid bones (Fig. 3).

Magnetic resonance imaging was performed, with sagittal, axial, and coronal sections. Radiological analysis of the sections showed osteochondromatous growths with polylobed contours and a signal identical to that of the bone, opposite the posteromedial border of the lower tibial extremity and the medial bones of the ankle. The synovium did not thicken. The ligaments were intact. Therefore, magnetic resonance imaging confirmed the intraarticular location of certain masses. It determined their osteocartilaginous nature, and above all, eliminated the presence of signs of malignancy (Fig. 4).

Discussion

Hemimelic epiphyseal disease HED also known as Trevor's disease is a rare pathology. It is a disorder of skeletal development in children that affects the epiphyseal cartilage unilaterally, producing an osteocartilaginous outgrowth associated with the presence of accessory epiphyseal ossification centers [1].

It is described in children between the ages of 2 and 14 years of age [2], appearing to be more frequent during the first decade of life [3], and remains rare in adults. It is more common in men, with a sex ratio of 3 boys to 1 girl. [4].

There is no evidence of hereditary transmission and no cases of malignant degeneration have been described in the literature [5,6].

The particularity of the disease is that it affects the epiphyses unilaterally, and the medial side is twice as affected as the lateral side. Typically, it affects one or more epiphyses of a lower limb, especially the medial femoral condyle, the distal and proximal end of the tibia, the tarsal bones, and the talus [7,8].

The hip joint may be affected in the acetabulum [9] or in the femoral head [10] Involvement of the patella is less common [11].

The involvement of the upper extremity and the bilateral distribution appears to be exceptional [12].

Thus, Azzouz et al. [1] have classified HED into 3 main groups. A localized form: in the epiphysis of a single joint of a limb. A classic form; the most frequent, where the damage is hemimelic in distribution in more than 1 epiphysis of a single limb. Then lastly, the generalized form characterized by the attack of all the joints of the lower limb, from the pelvis to the foot.

Clinically, HED can take several forms, ranging from simple asymptomatic lesions (discovered late after a revealing trauma), paucisymptomatic (exacerbated pain on walking, progressive unilateral medial or lateral swelling, lameness when the lesion is located in the hip joint) to orthopedic complications (axial deviations, abnormal joint movements, inequality of limb length), as was the case for our patient.

Since the clinic is nonspecific, the *radiological assessment* is the indispensable diagnostic tool for Trevor's disease.

Standard radiographs may be normal at a very early stage of the disease, especially in very young children. At an advanced stage, multiple secondary ossification centers appear in the epiphysis, in the form of irregular masses of hemimelic location that originate on one side of the affected epiphysis. This is accompanied by asymmetric epiphyseal enlargement [13].

With maturation, the ossification centers will unite and fuse with the rest of the epiphysis. The lesion will become ossified, calcified, and confluent with the underlying bone [14].

In our case, we report a case of epiphyseal dysplasia of the right ankle with the presence of a bony outgrowth of the posterior aspect of the talus on standard radiograph.

CT can be used to define the anatomy of the affected joint. It shows the location of the lesion in the epiphysis, as well as its relationship with adjacent bone and soft tissues, which allows its extension to be defined, but is less effective than MRI. Sometimes it can determine the cleavage plane between the lesion and the normal epiphysis [14]. Three-dimensional CT reconstruction can be useful to better guide the surgical decision and eventually judge the therapeutic result, and to allow subsequent monitoring, but MRI remains more sensitive for better early detection of complications or recurrences [15].

Radiographic data are often characteristic, but the use of magnetic resonance imaging allows better recognition of the lesion, its precise its location, size, and, in particular, its osteoarticular and soft tissue relationships, and helps to develop a surgical strategy by specifying the locoregional extension.

The advent of magnetic resonance imaging has allowed a significant advance in the positive diagnosis of HED, even at an early stage before the onset of ossification. It is performed in T1 and T2 sequences with axial, coronal, and sagittal slices, without and with gadolinium injection. The lesion presents an intermediate signal in T1 and a hyper signal in T2 with a hypo signal corresponding to the calcified foci. The non-ossified part has the same signal as normal articular cartilage [16]. In addition to the morphological characteristics of the lesion, MRI assesses its extension, its relationship with bone, the adjacent articular cartilage, tendons, ligaments, and muscles due to the cuts made in the various planes. This provides a complete assessment of the locoregional extension of the lesion. It also determines the plane of cleavage between the lesion and the normal epiphysis. [17,18], which will help guide the therapeutic decision, particularly if the proposed treatment is surgical.

Subsequently, magnetic resonance imaging provides follow-up and monitoring. It detects recurrences and the occurrence of complications in early stages, allowing proper management [19].

Biopsy is recommended for atypical or unusual disease locations, for early stages of the disease where radiological features are still insufficient to confirm the diagnosis, or for strong suspicion of other differential diagnoses requiring different treatment [20].

Pathological study can rule out most differential diagnoses of HED, except osteochondroma, as it only shows benign cartilage proliferation with changes similar to those seen in osteochondroma [20]. However, the initial clinical and radiological presentation must be taken into account, even after the histological result has been obtained. Rather, it is the collection of all criteria that often leads to the correct diagnosis.

The two *main differential diagnoses* of Trevor's disease are osteochondroma (osteogenic exostosis) and exostotic disease. [21,22] These two entities are similar to HED in their histologic characteristics, and distinct in their clinical presentations (HED occurs in children and young adolescents between the ages of 2 and 14 years, whereas osteochondroma is more common between the ages of 10 and 30 years); and also distinct in the epiphyseal location in the case of HED and metaphyseal location in the case of osteochondroma.

The risk of multiple hereditary exostosis degeneration into chondrosarcoma is possible and is nil in the case of HED.

However, based on radio clinical data, HED can be differentiated into several entities, mainly: [23]

- √ *Synovial chondromatosis*: characterized by multiple cartilaginous nodules reaching the joints, originating from the synovium and which can detach constituting intraarticular foreign bodies, with a normal aspect of the epiphysis, which differentiates it from the HED.
- √ *Myositis ossificans*: characterized by the presence of calcification in soft tissues (muscles, tendons, and fascia) without bone involvement, which differentiates it from HED [24].
- √ *Tumoral calcinosis*: characterized by the presence of a multinodular mass of variable size, but without involvement of the adjacent joint, the epiphysis appears normal, which is not the case in HED [25].
- √ *Vascular or parasitic calcifications*, which can sometimes be mistaken for a HED lesion given the presentation as periarticular swelling [26].
- √ *Ollier's disease*: characterized by the presence of osteocartilaginous growths that impede normal bone growth. All bones can be affected, even short bones. There is no hemimelic localization of the lesions, which differentiates it from HED [27].
- √ *Legg Calve Perthes disease*: The fragmentation phase of primary osteochondritis of the hip may give the appearance of multiple centers of ossification. The cyclic evolution of this pathology definitely supports the diagnosis.
- √ *The capsular or para-articular chondroma* is a benign cartilaginous lesion, presented on radiography as an osteolytic image with fine calcifications. The lesion is not related to the epiphysis. This appearance helps to differentiate it from HED [28].

The treatment of HED is not well defined and remains debated. It varies from simple surveillance to surgical removal of the lesion and correction of complications. The therapeutic decision is made on a case-by-case basis, depending on the stage of the diagnosis, the location of the lesion, its extension, in particular the intra or extraarticular location of the lesion, and the pathological consequences on the child's physical activity.

Overall, the prognosis of HED remains good; in particular, there is no risk of malignant degeneration, but there is a risk of recurrence of the disease as long as the organism is growing, hence the interest in regular monitoring until bone maturity [29].

Conclusions

Hemimelic epiphyseal disease should be considered in the presence of an epiphyseal mass in children. Early diagnosis ensures proper management, preventing the occurrence of orthopedic complications. The development of molecular biology techniques can facilitate its diagnosis, thus excluding other differential diagnoses, notably osteochondroma.

Patient consent

Written informed consent was obtained from the parents. They consented to the submission of the case report to the journal.

REFERENCES

[1] Azouz EM, Slomic AM, Marton D, Rigault P, Finidori G. The variable manifestations of dysplasia epiphysealis hemimelica. Pediatric Radiol 1985;15:44–9.
[2] Kuo RS, Bellemore MC, Monsell FP, Frawley K, Kozlowski K. Dysplasia epiphysealis hemimelica:clinical features and management. J Pediatr Orthop 1998;18:543–8.
[3] Merzoug V, Wicard P, Dabosset J, Khalifa G. Bilateral dysplasia epiphysealis hemimelica:report of two cases. Pediatr Radiol 2002;32:431–4.
[4] Shinozaki T, Ohfachi T, Watanabe H, Aoki J, Fukuda T, Takagishi K. Dysplasia epiphysealis hemimelica of the proximal tibia showing epiphyseal osteochondroma in an adult. Clin Imaging 1999;23(3):168–71.
[5] Trevor D. Tarso epiphysial aclasis.A congenital error of epiphysial development. J Bone Joint Surg (Br) 1950;32:204–13.
[6] Fairbank TJ. Dysplasia epiphysealis hemimelica (Tarso epiphysial aclasis). J Bone Surg (Br) 1956;38:237–57.
[7] Kettelkamp DB, Campbell CJ, Bonfiglio M. Dysplasia epiphysealis hemimelica.a report of fifteen cases and a review of the litterature. J Bone Joint Surg (Am) 1966;48:746–66.
[8] Fasting OJ, Bjerkreim I. Dysplasia epiphysealis hemimelica. Acta Orthop Scand 1976;47:217–25.
[9] Bleshman MH, Levy RM. A unusual location of an osteochondroma. Radiol 1978;127:456–63.
[10] Sherlock DA, Benson Mk. Dysplasia epiphysealis hemimelica of the hip:a case report. Acta Orthop Scand 1986;57:173–5.
[11] Cyrillo R, Montandon S, Frederico B, Moraes MD. Dysplasia epiphysealis hemimelica of the patella. Radiographics 2006;26:581–6.
[12] Viviana MR, Sandor K, Thomas T, Katalin K, Gyorgy S. Dysplasia epiphysealis hemimelica (Trevor's disease) 7 of our own cases and review of the litterature. Acta Orthop 2007;78(6):856–61.
[13] Skripitz R, Lussenhop S, Ludwig MS. Wedge excision chondroplasty of the knee in dysplasia epiphysealis hemimelica report of 2 cases. Acta Orthop Scand 2003;74(2):225–9.
[14] Gerscovich EO, Greenspan A. Computed Tomography in the diagnosis of dysplasia epiphysealis hemimelica. J Can Assoc Radiol 1989;40:313–15.
[15] Dennis R, Wenger MD, Mark J. Evaluation, imaging,histology and operative treatment for dysplasia epiphysealis hemimelica (Trevor disease) of the acetabulum:a case report and review. The Iowa Orthop J 2005;25:61–4.
[16] Iwasawa T, Aida N, Kobayashi N, et al. MRI findings of dysplasia epiphysealis hemimelica. Pediatr Radiol 1996;26:65–7.
[17] Peduto AJ, Frawley KJ, Bellemore MC, Kuo RS, Foster SL, Onikal E. MRI of dysplasia epiphysealis hemimelica:bony and soft tissue abnormalities. AJR Am J Roentgenol 1999;172(3):819–23.
[18] Starck DD, Bradeley WJ. Dysplasia epiphysealis hemimelica.MRI.Musculoskeleton. St Louis,Mosby 1999;917.
[19] Acquaviva A, Municchi G, Marconcini S, Mozzarella F. Dysplasia epiphysealis hemimelica in a zoung girl role of MRI in the diagnosis and follow up. Joint Bone Spine 2005;72:183–6.
[20] Mihir M, Thacker E, Azouz M, Scully S, Pitcher JD, Temple HT. Dysplasia epiphysealis hemimelica of the tibial tubercle. Pediatr Radiol 2006;36:244–6.
[21] Donaldson JS, Sankey HH, Girdany BR. Osteochondroma of the distal femoral epiphysis. J Pediatr 1953;43:212–16.
[22] Heiple KG. Carpal osteochondroma. J Bone Surg 1961;43A:861–4.
[23] Dorfman HD, Czerniac B. Synovial lesions. St Louis, Mosby: Bone tumors; 1998. p. 1041–57.
[24] Dorfman HD, Czerniac B. Synovial lesions. St Louis, Mosby: Bone tumors; 1998. p. 1041–57.
[25] Benchekroun T, Benjelloun B, Jorio M, EL Malki Tazi A. La calcinose tumorale à propos d'un cas et revue de la litterature. Medecine du Maghreb 1999;76:18–20.
[26] Dietz FR, Mumford JE. A hamartomatous joint mimicking dysplasia epiphysealis hemimelica of the talus. Iowa Orthop J 1994;14:166–70.
[27] Oestreich AE, Mitchell CS, Akuson JW. Both Trevor and Ollier disease limited to one upper extremity. Skeletal Radiol 2002;31:230–4.
[28] Steiner GC, Meushar N, Normal A, Present D. Intracapsular and para articular osteochondromas. Clin Orthop 1994;303:231–6.
[29] Bovee JVMH, Hameetman L, Kroon HM, Aigner T, Hogandroorn P. EXT related pathways are not involved in the pathognosis of dysplasia epiphysealis hemimelica. J Pathol 2006;209:411–19.