Original article

Diagnosis and surgical treatment of dysplasia epiphysealis hemimelica. A report of nine cases

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ABSTRACT

Background: Dysplasia epiphysealis hemimelica (DEH) is a rare developmental bone disorder with hemimelic involvement of one or more epiphysis. We report on nine new cases and discuss the clinical manifestations, the value of MRI, and the results of complete and early surgical resection of these lesions.

Materials and methods: In this retrospective study, nine patients with a diagnosis of DEH were evaluated. Age at presentation ranged from 1 year to 12 years. The main complaint at diagnosis was a swelling bony mass. Angular deformities were recorded in two patients. All patients were surgically treated and followed up clinically and by imaging. Eight patients underwent excision only.

Results: The average follow-up was 5.6 years (range, 2–10.5 years). All patients had a good outcome without related symptoms. No epiphysiodesis, angular deformity or recurrence was observed. One patient with femoral lesion involving the distal medial part of the epiphysis developed, four months after surgical excision, a calcification outside the area of total excision. This calcification did not increase in size at two years follow-up. Another patient with lateral involvement of the proximal tibial epiphysis presented a postoperative nervous complication. Spontaneous nervous recovery occurred three months after surgery.

Discussion: MRI was useful to find a potential plane of cleavage between the epiphysis and the pathological tissue. We recommend early removing ossifications when a cleavage plane is identified. Waiting a possible complication or increasing of size does not seem logical. Of course, the treatment will be not the same if no cleavage plane is found on MRI.

Level of evidence: IV.

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1. Introduction

Dysplasia epiphysealis hemimelica (DEH), also known as Trevor’s disease, is a rare developmental bone disorder with hemimelic involvement of one or more epiphysis in children [1–6] as a result of overgrowth of cartilage [7]. Although first reported by Mouchet and Belot in 1926 and called “tarsomégalie” [8], it was delineated as a distinct entity by Trevor in 1950 [4]. In 1956, Fairbank [2] described the characteristic involvement of either the lateral or medial half of a single limb. He first used the term “dysplasia epiphysealis hemimelica”, which appears to be the most logical and frequently used nomenclature. The reported incidence of DEH is 1 in 1,000,000 [7–9]. However, this estimate may be artificially low since many cases may go unrecognized because of a similar histologic appearance to osteochondroma or because the lesion may be asymptomatic [10,11]. Children or teenagers are frequently involved, mostly the males [2,4–6,12–14]. The involvement of the affected epiphysis is hemimelic, meaning that either the medial or the lateral part of the center of ossification is affected, the medial side more commonly [7]. Deformity is noted in younger children, and pain is common in older children [15]. The lower limbs are usually affected [1,4,6,16], the upper limbs [17,18] and spine [7–9] rarely so. The epiphysis grows asymmetrically and forms an osteochondral protuberance that is

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macroscopically and histologically similar to an exostosis [7]. The characteristic hemimelic pathological growth pattern results clinically in painless, bony swelling, deformities, exostoses, limb-length discrepancies, progressive stiffness and articular incongruency [1–7,10,15]. Irregularity of the articular surface may lead to early secondary osteoarthritis [9]. The lesions usually increase in size until the patient reaches skeletal maturity; however, in some cases, enlargement of the lesions was observed in adults [19]. The etiology is unknown.

The diagnosis is primarily based on plain radiographs [1]. The radiographic findings are characteristic of DEH, showing asymmetrical epiphyseal overgrowth and exostoses of osteochondroma-like appearance. At an early stage, they reveal an irregular intra-articular mass with fragmented asymmetrical ossification arising from either the lateral or frequently the medial half of the affected epiphysis without metaphyseal changes. At a later stage, this lesion fuses with the adjacent bone, like an exostosis. MRI is useful, more reliable and accurate [20–22]. It can establish the early diagnosis of DEH before the onset of ossification. It exactly defines the intra-articular osteocartilaginous mass, its extension and anatomical relations and provides detailed images of associated joint deformity. Treatment ranges in literature from simple observation of the asymptomatic lesions to surgical excision [4,7,10,23]. There have been good results with non-operative treatments in the literature [10]. Some authors [10] do not recommend excision of an articular lesion because it contains a high risk to damage the articular cartilage. In our department, we performed early systematic surgical excision before the onset of deformation even in articular localization when a cleavage plane was identified. We report nine new cases and discuss the interest of complete and early surgical resection of these lesions.

2. Materials and methods

In this retrospective study, nine Caucasian patients (eight boys and one girl) with a diagnosis of DEH were evaluated from 1999 to 2011 at the author’s institution (Table 1). We reviewed the clinical records and followed up all nine patients. All patients had a negative family history of bone dysplasia. Age at presentation ranged from 1 year to 12 years. The main complaint at diagnosis was a swelling bony mass that was increasing in size (8 cases). Among these nine patients, only two complained of pains. Angular deformities were recorded in two patients: one with severe genu valgum of 35° (Fig. 1) and another one with posterior deformity of the wrist (the extension was severely restricted) (Fig. 2).

All patients had plain radiographs, CT scans and MRI. Our patients were classified according to Azzouz et al. [16]: The “localized form” of DEH affects a single bone. The “classical form” affects more than one epiphysis in a single lower extremity, particularly the talus, distal femoral epiphysis, and distal tibial epiphysis. In the “generalized form”, the whole lower limb from the pelvis to the foot or ankle is affected. There were seven “localized form” (cases 3 to 9), one “classical form” (case 2) and, one “generalized form” (case 1). Eleven lesions were in the lower limbs, one in the upper limb. The talus was involved in six cases. CT scans or MRI determined a clear cleavage plane between the epiphysis and the abnormal cartilaginous growth in all cases (Figs. 3 and 4).

An arthroscopy was performed. All patients had a biopsy during surgery, none prior to surgery. As a clearly defined plane has been demonstrated intraoperatively for all patients, the cartilage-covered mass was excised in all patients. We did not use fluoroscopy. All patients were followed up clinically and by imaging (X-rays and MRI). Eight patients underwent excision only. No corrective osteotomy was performed. One 4-year-old boy required an excision of the mass involving the posterior distal epiphysis of the radius leading an immediate instability of the wrist. This instability was caused by lesion of carpal ligaments and dorsal capsular distension. Stabilization of the wrist was treated by ligationoplasty. In this case 1, lesions of wrist and talus were the first locations. Involvement of the calcaneus occurred one year later.

3. Results

The average follow-up is 5.6 years (range, 2–10.5 years). All patients had a good outcome without related symptoms and all of them returned to their normal activities. No epiphysiodesis, angular deformity or recurrence was observed at longest follow-up. One patient (case 5, Fig. 1) with femoral lesion involving the distal medial part of the epiphysis (with a severe valgus) developed, four months after surgical excision, a calcification outside the area of total excision. This calcification did not increase in size (X-rays and MRI monitoring) at two years follow-up. Valgus was completely and spontaneously corrected one year after surgery. Another patient (case 7) with lateral involvement of the proximal tibial epiphysis presented a postoperative nervous complication.

### Table 1

<table>
<thead>
<tr>
<th>Case</th>
<th>Age at discovery (years)</th>
<th>Gender</th>
<th>Complaints</th>
<th>Anatomic site</th>
<th>Side</th>
<th>Follow-up (months)</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>4</td>
<td>Male</td>
<td>Pain + deformation</td>
<td>Posterior distal radius R</td>
<td>R</td>
<td>126</td>
</tr>
<tr>
<td>2</td>
<td>4</td>
<td>Female</td>
<td>Pain + swelling</td>
<td>Lateral distal femoral epiphysis L</td>
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<td>75</td>
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<tr>
<td>3</td>
<td>4</td>
<td>Male</td>
<td>Swelling</td>
<td>Medial talus L</td>
<td></td>
<td></td>
</tr>
<tr>
<td>4</td>
<td>10</td>
<td>Male</td>
<td>Swelling</td>
<td>Medial talus R</td>
<td></td>
<td></td>
</tr>
<tr>
<td>5</td>
<td>1</td>
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<td>Deformation</td>
<td>Medial distal femoral epiphysis – valgus 35°</td>
<td>L</td>
<td>29</td>
</tr>
<tr>
<td>6</td>
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<tr>
<td>7</td>
<td>12</td>
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<td>Swelling</td>
<td>Lateral proximal tibial epiphysis L</td>
<td>L</td>
<td>84</td>
</tr>
<tr>
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<td>Swelling</td>
<td>Medial talus R</td>
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<td></td>
</tr>
<tr>
<td>9</td>
<td>9</td>
<td>Male</td>
<td>Swelling</td>
<td>Medial distal tibial epiphysis L</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

Fig. 3. Case 4. Ten-year-old boy. a: preoperative plain radiograph and clinical presentation; b: preoperative CT scan with a clear cleavage plane; c: postoperative plain radiograph and anatomic lesion.
The complete excision of the voluminous mass required a neurolysis of the common peroneal nerve. Spontaneous nervous recovery occurred three months after surgery.

4. Discussion

Treatment ranges in literature from simple observation of the asymptomatic lesions to surgical excision [4,7,10,23]. There have been good results with non-operative treatments in the literature [10]. The treatment should be individualized depending on the clinical findings. Asymptomatic lesions can be treated non-operatively, as there is no known of malignant transformation [7]. Surgical treatment is usually indicated when the lesion produces pain, deformity or interferes with the joint motion [24]. Surgery should also be undertaken to correct the deformation or to treat the damages of the articular cartilage: triple arthrodesis of the ankle, epiphysiolysis or extra-articular osteotomies, chondroplasty of the knee [15,23]. Kuot et al. [10] had excellent results with excision of juxta-articular lesions, but only fair or poor results with excision of articular lesion. So, they did not recommend excision of an articular lesion unless it became a loose body. Excision of an intra-articular lesion is not recommended because it contains a high risk to damage the articular cartilage and to induce early osteoarthriti. If the joint surface is smooth, any associated deformity can be corrected through extra-articular osteotomy. We wish to modulate these assertions. Osteoarthritis secondary to joint surface deformity may also develop with a conservative treatment. Keret et al. [23] showed that when the lesion was intra-articular, recurrent deformities may occur following repeated distal osteotomies. Before complete ossification, especially in the first decade of life, there is often a cleavage area of cartilage between the ossification center in the lesion and that of the epiphysis [25]. In our series, we determined a clear cleavage plane between the epiphysis and the abnormal cartilaginous growth, in all cases, even in the three cases with intra-articular involvement (Fig. 3). In our department, we performed early systematic surgical excision, even with asymptomatic lesion, before the onset of deformation or invasion of the epiphysis, even in articular localization (as in our cases 1 and 5). Waiting a possible complication or increasing of size seems not logical. Our treatment would certainly not have been the same in case of no cleavage planes found on MRI (Fig. 5). In our series, surgical resection was performed following the cleavage planes and avoiding any damage of the growth plate or tarsal bones. A minimal two years follow-up seems to be sufficient to eliminate secondary complications of the treatment as growth disturbance. The specific benchmarks and cleavage planes predefined by MRI are very useful to guide and to allow the complete resection of the tumor. For Keret et al. [23], in the three patients which developed genu valgum, no plane between the accessory ossification center and the main epiphysis could be demonstrated intraoperatively, even though the MRI clearly showed the plane of separation in all three patients. We have no explanation for this finding, except perhaps the young age of children (8, 9 and 13 months). Nevertheless, even in good results, clinical and imaging survey until skeletal maturity must be performed to detect recurrences or the secondary appearance of other epiphyseal damage as lesions do not always present synchronously [15].

We did not encounter difficulties in the diagnosis. Although the diagnosis of DEH can be presumed based on history, physical examination, and imaging studies, a biopsy is necessary for definitive diagnosis. Cartilaginous nodules characterize the histological appearance of the lesions with osseous metaplasia, like those of an oseochondroma picture (Fig. 6). After complete ossification, the lesion is microscopically indistinguishable from an oseochondroma: a base of normal bone and a cap of hyaline cartilage are observed [24]. Oseochondroma may occur in any bone that is preformed from cartilage, but the most common locations are the long bones at the metaphyseal region and none are epiphyseally centered [24]. Because of the similar patho-anatomy and histologic features of DEH and oseochondroma, the decision to classify a lesion as DEH is based primarily on the intra-articular location of a lesion [2,16]. However, differential diagnosis remains difficult especially with synovial oseochondromatosis, myositis ossificans, tumoral calcinosis, soft tissue sarcoma, injury-related soft tissue calcification. Synovial oseochondromatosis represents cartilaginous nodule formation by the synovial membrane. It is invariably a monoarticular disease. Radiographically, the nodules usually appear as multiple intra-articular radiodense lesions. Myositis ossificans is a tumor-like heterotopic ossification in soft tissue, usually muscles, but also tendons, ligaments, and joint capsules [24]. It becomes a firm mass attached to the adjacent bone and soft tissues during maturation. Within a few months, it usually becomes mature bone with a cortical shell surrounding a center of cancellous tissue. Tumoral calcinosis is a rare condition, consisting of calcium salt deposition in extracapsular soft tissues about joints. Radiographically, the masses of tumoral calcinosis appear as homogeneous calcific dense nodules about the joints [24]. Chondrodyasplasia punctata is a form of multiple epiphyseal dysplasia that is characterized by calcification of unossified cartilaginous epiphyses during the first year of life. The clinical findings are short
Fig. 5. On this case (out of our series because he was seen as an out clinic patient for a second opinion), we did not suggest removing the lesion because no cleavage plane was found. A. Coronal plain radiograph. B. Coronal MRI T2 TSE sequence.

In our series, the most common symptom of DEH was an asymptomatic joint swelling, increasing in size with the growth. Angular deformity was related to intra-articular lesions of the wrist (case 1) and the knee (case 5). The two patients complaining of pain had atypical location: posterior half of the distal radial epiphysis and lateral half of the distal femoral epiphysis. In our series, DEH affected the boys (except one girl) and occurred in the first decade in all patients except one. Talus was involved in six cases, making it the most frequent site.

Fig. 6. Histological sections. a: cartilaginous nodule with osseous metaplasia in the center making an osteochondroma-like appearance; b: cartilaginous nodules within partially ossified connective tissue (×20); c: cartilaginous nodule almost completely ossified with a persistent cartilaginous cap external appearance forming an osteochondroma-like appearance.

The diagnosis is usually based on plain radiographs, which show the characteristic hemimelic pattern of asymmetrical epiphyseal overgrowth and exostoses of osteochondroma-like appearance [2,4,7,16,23,26]. Typically, radiographs initially show an irregular mass with focal ossification arising from one side of the affected epiphysis [7]. On maturation, the lesion ossifies and becomes confluent with the underlying bone [7]. The final appearances may be similar to those of an osteochondroma or the affected area remains enlarged and irregular. Premature closure of the physis may cause deformity or a limb-length inequality [7]. CT imaging is sometimes insufficient to show the early cartilage lesions before the
ossification and to exactly define the cleavage planes [27,28]. MRI provides a much more accurate image, can establish the diagnosis of DEH at an early stage (before the onset of ossification) and can be helpful in determining a potential plane of cleavage between the epiphysis and the pathological tissue [7,20–23]. It is important to analyze different weighted gradient-echo image with and without gadolinium in order to find the cleavage plane (Fig. 4). MRI may also show any joint involvement, abnormalities of soft tissue around the involved joints and has a role in distinguishing DEH from tumors [21]. Iwasawa et al. [22] and Peduto et al. [29] described bone and soft tissue abnormalities in dysplasia epiphysealis hemimelica. The osteochondral overgrowth of the epiphysis has a signal intensity similar to that of normal epiphyseal cartilage and bone on all imaging sequences [29]. Multiple additional small foci of enchondral ossification can be seen within the overgrown epiphysis [29]. The fused irregular osseous components of the osteochondromas continuous with the epiphysis and those that had been separated or fragmented can be readily identified and characterized in multiple planes by MR imaging [29]. The cartilaginous cap is depicted as a mottled area of high intensity on a T2-weighted image [22]. On T1-weighted MRI scan, low signal intensity of the cartilaginous region and high signal intensity of the marrow fat portion of the tumor can be observed. T2-weighted MRI scan show that protruding intermediate to high signal intensity lesions (cartilage and bone marrow) are divided by low signal calcified matrices. MRI was used for our nine patients. MRI showed the most accurate details of the unossified cartilaginous mass as well as the status of the articular cartilage. We determined a clear cleavage plane between the epiphysis and the abnormal cartilaginous growth, in all cases. Our patients were young and we did not observe fusion between the pathologic mass and the epiphysis or tarsal bones. MRI can also detect early recurrences [10,20–23].

5. Conclusion

Even if some authors do not recommend excision of an articular lesion because of a high risk to damage the articular cartilage, we recommend early removing ossifications when a cleavage plane is identified. Waiting a possible complication or increasing of size does not seem logical. Of course, the treatment will be not the same if no cleavage plane is found on MRI.

Disclosure of interest

The authors declare that they have no conflicts of interest concerning this article.

References


