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**Dan Wang**

Sports Medicine & Rehabilitation

**Renhai Feng**

Sports Medicine & Rehabilitation

**Guoping Pan**

Sports Medicine & Rehabilitation

**Linrui Peng**

Sports Medicine & Rehabilitation

**Renjie Pu**

601583673@qq.com

Sports Medicine & Rehabilitation

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## Case Report

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# Diagnosis, Treatment, and Bone Remodeling of Dysplasia Epiphysealis Hemimelica of the Medial Femoral Condyle in a Child: A Case Report

Dan Wang<sup>1,3</sup> Renhai Feng<sup>1,3</sup> Guoping Pan<sup>1,3</sup> Linrui Peng<sup>2,3</sup> Renjie Pu<sup>1,3</sup>

1. Department of Radiology, Ningbo No. 6 Hospital, Ningbo, Zhejiang, China.

2. Department of Orthopaedic Oncology, Ningbo No. 6 Hospital, Ningbo, Zhejiang, China.

3. Ningbo Clinical Research Center for Orthopedics, Sports Medicine & Rehabilitation, Ningbo, Zhejiang, China.

Corresponding author: Renjie Pu 601583673@qq.com

## Abstract

Dysplasia epiphysealis hemimelica is a rare osteochondral proliferative lesion in children. It is uncommon when located in the medial femoral condyle, and clinically, it is prone to misdiagnosis. Here, we report a case of a 9-year-old boy who presented with right knee pain and restricted range of motion induced by trauma. Preoperative multimodal imaging, including digital radiography, computed tomography, and magnetic resonance imaging, revealed complex features such as articular cartilage defects, bone marrow edema, and lobulated osseous prominences, requiring differential diagnosis from osteochondritis dissecans and bone tumors. The patient underwent surgical resection of the lesion on the medial femoral condyle of the right femur, and postoperative pathology confirmed the diagnosis of DEH. A 1.5-year post-operative DR follow-up meticulously documented the entire progression of the resected region, transitioning from early bone defect to mid-term bone remodeling, and ultimately to the complete recovery of the anatomical contour in the long term. This case indicates that the integration of multimodal imaging with pathological evaluation facilitates early and precise diagnosis of DEH. For symptomatic pediatric patients with articular involvement, surgical intervention can fully leverage the self-remodeling capacity of pediatric epiphyses, thereby yielding excellent mid- to long-term clinical outcomes.

**Keywords:** Dysplasia epiphysealis hemimelica; Trevor's disease; Medial femoral condyle; Pediatric bone remodeling; Case report

## Introduction

Dysplasia epiphysealis hemimelica (DEH), also known as Trevor's disease, is characterized by asymmetrical partial epiphyseal dysplasia. Its underlying pathological alteration is the orderly hyperplastic overgrowth of epiphyseal cartilage, forming a cartilaginous exostosis that may present with multiple ossification centers that eventually coalesce, leading to hemimelic epiphyseal enlargement [1].

Recent literature has also described it as an idiopathic pathological chondrocytes proliferation, presenting as a cartilaginous osteochondroma-like exophytic mass projecting into the joint or juxta-articular region [2]. The disease predominantly occurs in childhood, commonly affecting the long bones of children under 8 years of age, whereas adult onset is extremely rare [3]. Clinical observations also indicate an average patient age of 8.5 years, with diagnosis typically occurring between 2 and 14 years [4]. Lesions frequently involve lower extremity

joints, particularly the knee and ankle [5, 6]. Although DEH is a benign entity, without timely intervention, the lesion may continue to grow until skeletal maturity, potentially predisposing patients to long-term complications such as joint degenerative changes and early-onset osteoarthritis. Previous studies have reported that when the lesion is intra-articular, MRI possesses significant diagnostic value in delineating the cleavage plane between the lesion and the normal epiphysis [7]. Therefore, in pediatric orthopedic practice, clinicians should maintain a high index of suspicion for DEH when managing pediatric patients presenting with epiphyseal masses or lower extremity joint pain.

However, in daily clinical practice, the diagnosis and management of DEH present significant challenges. The early clinical manifestations lack specificity, predominantly presenting as joint pain, restricted range of motion, joint contour deformity, or local swelling. A study reported that among 6 patients, joint limitation accompanied by pain was the primary symptom in 66%, joint contour deformity accounted for 34%, and a systematic review indicated that a palpable mass and pain were observed in 54% of cases [8]. Radiological findings are likewise heterogeneous: plain radiographs typically demonstrate irregular ossification at the involved epiphysis, while CT scans reveal irregularly fragmented osteochondral masses extending intra-articularly [9,10]. These features render it difficult to differentiate DEH from conditions such as osteochondritis dissecans and osteochondroma. Cases have also been reported where DEH was misdiagnosed as osteochondritis dissecans of the medial femoral condyle, leading to delayed or improper management [11]. Regarding treatment, standardized guidelines or specific treatment algorithms are currently lacking [8]. Therapeutic strategies are tailored individually based on lesion site, size, and symptom severity, ranging from conservative observation to surgical resection. Literature has documented varying outcomes between conservative management and surgical intervention, emphasizing that an accurate diagnosis can prevent unnecessary procedures [12]. In a systematic review of DEH involving the foot and ankle, 92% of patients underwent surgical treatment, with 95% receiving mass excision, yielding a recurrence rate of 9% [8]. Other studies have noted that simple excision yields favorable outcomes for extra-articular lesions, whereas intra-articular involvement may necessitate corrective osteotomy, with deformity recurrence being relatively common [13]. The aforementioned diagnostic ambiguity and therapeutic heterogeneity underscore the critical necessity for individualized clinical decision-making in DEH.

In this context, by presenting a detailed case involving diagnosis, treatment, and an 18-month follow-up, this article aims to summarize the multimodal imaging characteristics of DEH and analyze the clinical biological process of articular surface reconstruction and bone remodeling following lesion resection.

## **Case Presentation**

### **1. Patient Information and History**

A 9-year-old boy was admitted to our hospital presenting with "right knee pain and restricted range of motion for over two months following a fall, which

had worsened for one week." More than two months prior to admission, the patient experienced pain over the anteromedial aspect of his right knee after a fall, without skin breach; no medical intervention was sought at that time. One week ago, the pain intensified, and a magnetic resonance imaging (MRI) scan of the right knee performed at a local hospital suggested "possible osteochondritis dissecans of the medial femoral condyle and medial meniscal injury." Seeking further management, the patient was admitted to our department with the outpatient working diagnoses of: (1) lesion of the right medial femoral condyle (osteochondritis dissecans? bone tumor? dysplasia epiphysealis hemimelica?); and (2) injury of the right medial meniscus. Since the onset of symptoms, the child had no fever and experienced no significant weight loss.

## 2. Specialized Physical Examination and Laboratory Evaluation

- **Physical Examination:** Physical examination revealed a firm, well-demarcated mass measuring approximately 3cmx2cm over the medial aspect of the right knee, with marked tenderness upon palpation. No obvious swelling, skin breach, or active bleeding was observed in the remaining areas. Range of motion (ROM) during flexion and extension of the right knee was limited. Neurological sensation of the right lower extremity was intact, with good dorsalis pedis artery pulsations, satisfactory capillary refill, and normal motor function in the toes.
- **Functional Assessment:** The Visual Analog Scale (VAS) pain score was 1. The Barthel Index for activities of daily living was 60, indicating moderate functional impairment.
- **Laboratory Examination:** Complete blood count (CBC) revealed an elevated platelet count of  $377 \times 10^9/L$  and a mean corpuscular volume (MCV) of 80.5 fL. The erythrocyte sedimentation rate (ESR) was slightly elevated at 17 mm/h. Urinalysis showed a high specific gravity ( $>1.030$ ). Coagulation profiles and antithrombin III activity were within normal limits. Rheumatic immune markers and serum alkaline phosphatase (ALP) levels were unremarkable.

## 3. Imaging Findings

- **Digital Radiography (DR):** Plain radiographs demonstrated rough margins along the surfaces of the right medial trochlea and medial femoral condyle. A lobulated, well-circumscribed osseous prominence with heterogeneous ossification was observed protruding into the joint cavity. The base of the lesion was continuously fused with the distal femoral epiphysis (Figure 1.A1, A2).
- **Computed Tomography (CT):** CT scans revealed structural deformation and local abnormal enlargement of the posterior aspect of the right medial femoral condyle, along with an irregular, uneven trochlear articular surface. The lesion exhibited heterogeneously increased bone density, containing multiple isolated patchy and granular ossification centers, accompanied by mild adjacent soft tissue swelling (Figure 1.B1–B3).
- **Magnetic Resonance Imaging (MRI):** MRI demonstrated marked thinning and

d localized defects of the cartilage beneath the medial femoral condyle articular surface, with patchy areas of subchondral bone marrow edema characterized by prolonged T1 and T2 relaxation signals. A small joint effusion was observed within the joint space. High signal intensity (Grade I–II) was identified in the posterior horn of the right medial meniscus (Figure 1.C).

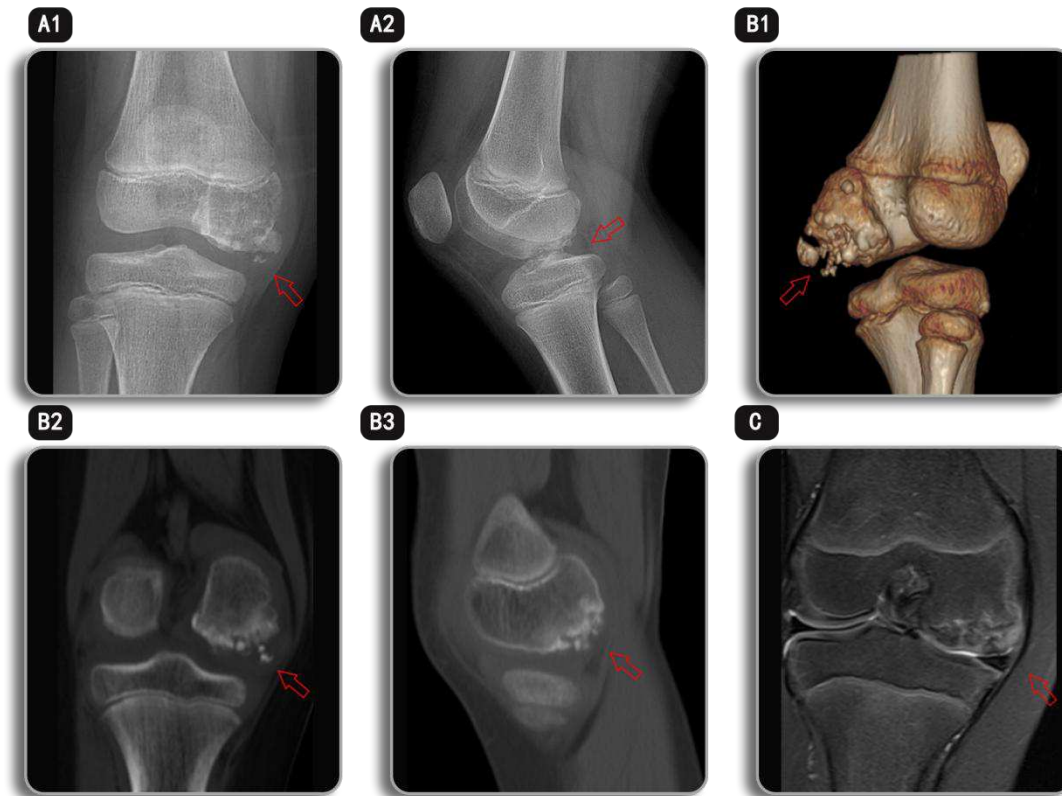


Figure 1. Preoperative radiological findings. Anteroposterior (A1) and lateral (A2) radiographs of the knee joint, demonstrating irregular bone destruction of the distal femur accompanied by peripheral punctate and patchy ossifications. (B1–B3) CT images of the knee joint, including a 3D reconstruction image (B1) as well as coronal (B2) and sagittal (B3) views, showing bone hyperplasia and fragmented/separated osseous structures within the lesion site. (C) Coronal MRI scan of the knee joint, revealing signal abnormalities in the lesion area along with surrounding soft tissue involvement/edema. *Note:* The red hollow arrows indicate the location of the lesion.

#### 4. Surgery and Histopathology

Under general anesthesia, the patient underwent "resection of the lesion in the right medial femoral condyle." Following successful anesthesia induction, the patient was placed in a supine position, and the surgical field was routinely disinfected, draped, and a tourniquet was applied. An 8-cm incision was made along the medial aspect of the right knee. Tissues were dissected layer by layer, and the vastus medialis muscle was retracted anteriorly to expose the medial femoral condyle. Abnormal bone proliferation was observed at the medial femoral condyle, with the articular surface presenting an irregular, cauliflower-like elevation measuring approximately 2cmx2cm (Figure 2.A1). The boundary between the lesion and the surrounding normal articular cartilage was relatively clear. A periosteal elevator was used to dissect along the cleavage plane, resulting in

complete detachment of the mass. Following detachment, a cartilage defect measuring approximately 2cmx2cm was noted on the weight-bearing surface of the medial femoral condyle (Figure 2.A2). The lesion within the medial femoral condyle was then accessed through the cartilaginous defect. A curette was used to scrape the lesion, and multiple sclerotic foci were palpated within the cancellous bone. These sclerotic foci were meticulously removed using a high-speed burr. Inspection confirmed that the lesion was thoroughly debrided, and the wound was extensively irrigated with normal saline (Figure 2.B). After changing sterile gloves and instruments, two sheets of absorbable hemostatic gauze (Beiling) were applied for hemostasis. The surrounding tissues were closed layer by layer using absorbable sutures, and the skin incision was secured with surgical tissue adhesive. The operation was completed smoothly with minimal intraoperative blood loss, and the patient returned safely to the ward with continuous saline irrigation. Postoperative histopathological examination revealed hyperplastic cartilaginous tissue with focal calcification, consistent with dysplasia epiphysealis hemimelica (Figure 2.C).

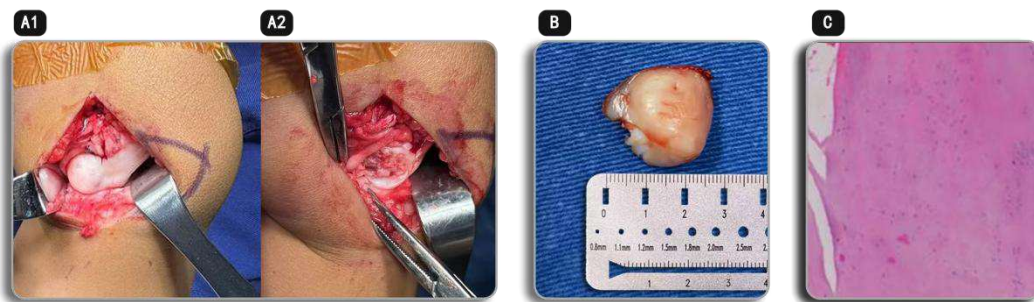


Figure 2. Intraoperative findings, gross specimen, and histopathological examination. (A1) Intraoperative view showing the exposed lesion. (A2) The underlying wound surface following lesion resection. (B) Gross appearance of the completely resected lesion tissue. (C) Postoperative histopathological section showing hyperplastic cartilaginous tissue accompanied by focal calcification.

## 5. Postoperative Radiological (DR) Follow-up

- 1 Day Postoperatively: Compared to the preoperative status, the main lesion and surrounding loose osseous fragments were substantially resected. Irregular bone defects along the excision margin remained visible, with no residual free osseous fragments observed within or around the joint space (Figure 3.A).
- 1 Month Postoperatively: The hyperplastic lesion of the right medial femoral condyle was thoroughly cleared. The margins of the resected region appeared rough and serrated, with heterogeneous local bone density, and a prominent anatomical defect and concavity were present on the articular surface (Figure 3.B).
- 3 Months Postoperatively: The defect margins in the resected region transitioned from serrated to blunted. Postoperative localized stress-induced sclerosis was gradually absorbed, and early new bone formation was visible, filling the defect zone (Figure 3.C).
- 1 Year Postoperatively: The morphology of the medial femoral condyle de-

monstrated significant improvement. The original surgical excision area fused seamlessly with the adjacent normal bone. The articular surface concavity became shallower, the bone density tended toward uniformity, and no signs of lesion recurrence were observed (Figure 3.D).

- 1.5 Years Postoperatively: The anatomical contour of the medial femoral condyle completely regained a smooth and symmetrical appearance with rounded margins. The medial knee joint space restored equal width, and the bilateral lower extremities exhibited essentially equal length (Figure 3.E).

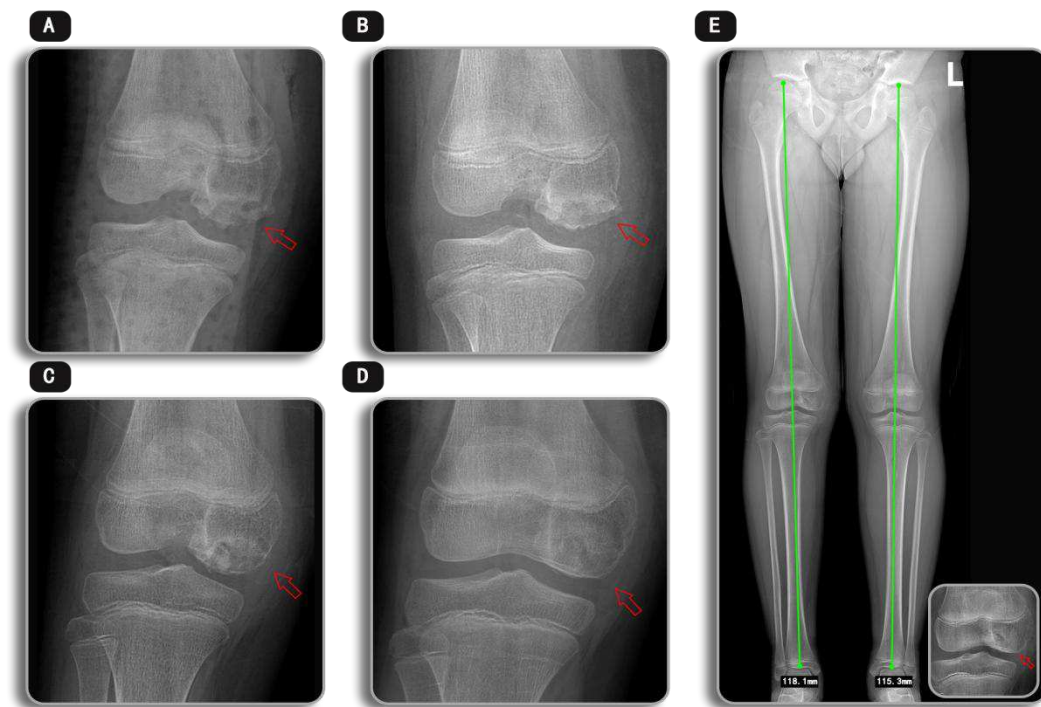


Figure 3. Postoperative radiological (X-ray) findings. (A–E) Serial anteroposterior radiographs of the affected knee during follow-up, demonstrating the progressive healing process of the lesion site over time (indicated by red hollow arrows). (A) At 1 day postoperatively, showing complete resection of the lesion; (B) at 1 month postoperatively, with the lesion margins becoming progressively distinct; (C) at 3 months postoperatively, showing gradual filling and bone formation within the osseous defect zone; (D) at 1 year postoperatively, showing repair of the lesion site and restoration of a smooth articular surface. (E) Full-length anteroposterior radiograph of both lower extremities at 1.5 years postoperatively (green lines represent the mechanical axes of both lower limbs), demonstrating equal limb lengths and no significant deviation in lower limb alignment.

## Discussion

This case primarily reports a rare presentation of dysplasia epiphysealis hemimelica causing right medial femoral condyle pain in a 9-year-old boy. Preoperative imaging demonstrated ambiguous features that could not exclude osteochondritis dissecans (OCD), bone tumors, or hemimelic epiphyseal dysplasia, thereby rendering a definitive preoperative diagnosis challenging. Surgical intervention was undertaken for the lesion, during which intraoperative exploration confirmed an exophytic mass arising from the hyperplastic epiphysis of the medial femoral condyle, followed by complete excision. Postoperative histopathology de

definitively established the diagnosis of DEH. Subsequent serial radiological follow-up demonstrated progressive repair of the osseous defect, successful bone remodeling, restoration of normal articular contours, and excellent functional recovery, with no evidence of lesion recurrence during the follow-up period. Findings from this case demonstrate a direct correlation between meticulous surgical excision of the lesion and a favorable long-term prognosis. This outcome not only underscores the efficacy of the surgical approach but also provides valuable insights and guidance for clinical decision-making in similar rare pediatric cases.

The clinical presentation of this case exhibits remarkable uniqueness across three distinct dimensions—anatomic location, clinical manifestations, and radiological features—which directly contributed to the preoperative diagnostic uncertainty. Regarding its anatomical site, although previous literature has reported the distal femur and distal tibia as common predilection sites for DEH [14], and isolated cases involving the posteromedial distal femur have been documented [15], a retrospective study involving 9 patients identified only 1 case with a lesion situated in the medial epiphyseal region of the distal femur [16]. This indicates that involvement of this specific region remains relatively rare among DEH predilection sites. In contrast to the more commonly reported sites such as the talus or distal tibia, the localization of the lesion to the medial femoral condyle in this case highlights its anatomical singularity. In terms of clinical manifestations, the present case presented with trauma-induced pain that progressively intensified, accompanied by a palpable local mass and restricted joint motion, yet without marked joint effusion. Previous studies have documented that periaarticular masses and localized pain represent cardinal features of DEH, and instances have been reported where patients presenting with knee pain were initially misdiagnosed with osteochondritis dissecans of the medial femoral condyle [11]. This aligns closely with our initial working diagnosis and reflects how easily DEH can be confounded with other knee pathologies during early clinical evaluation. Radiologically, this case demonstrated a complex, mixed multiphasic presentation across three modalities: plain radiography revealed a lobulated, well-defined osseous prominence with heterogeneous ossification protruding into the joint cavity; CT demonstrated multiple isolated patchy and granular ossification centers; and MRI identified subchondral cartilage defects along with bone marrow edema. In contrast, the classic MRI hallmark of DEH reported in previous literature is an osteochondral mass exhibiting signal intensity similar to normal cartilage containing low-signal foci, separated from the normal epiphysis by a distinct cleavage plane [17]. Compared with these typical features, the prominent mixed characteristics observed in this case—including bone marrow edema, multiple fragmented osseous elements, and exophytic bony ridges—far exceeded the classic radiological spectrum of DEH. This rendered a definitive preoperative diagnosis challenging, thereby emphasizing the diagnostic complexity and clinical reporting value of this case.

The aforementioned preoperative diagnostic process fully underscores the cl

inical complexity of this entity. DEH is easily confused with osteochondritis dissecans radiologically, making their differential diagnosis essential: Radiographic Differentiation: OCD on plain radiographs typically presents with localized osteochondral fragments, a surrounding sclerotic rim, and a radiolucent line of separation, often accompanied by intra-articular loose bodies. In contrast, DEH maintains structural epiphyseal integrity without isolated bone fragments, sclerotic margins, or loose bodies, exhibiting only slightly decreased local bone density within the epiphysis. MRI Differentiation: MRI serves as the primary tool for distinguishing between the two entities. In OCD, MRI reveals a localized osteochondral lesion with fluid signal traversing the osteochondral interface, often associated with cartilage disruption, a low-signal sclerotic border surrounding the lesion, and bone marrow edema strictly localized around the fragment. Conversely, DEH presents with patchy bone marrow edema within the epiphysis, an intact and continuous osteochondral interface lacking fluid-filled clefts or separated bone fragments, and preserved articular cartilage; moreover, the epiphyseal marrow edema in DEH can completely resolve during follow-up without leaving bone defects or loose bodies.

Postoperative radiological follow-up fully validated the efficacy of the surgical strategy while illustrating the distinct characteristics of bone defect repair and epiphyseal remodeling. At one month postoperatively, the rough articular margins, heterogeneous bone density, and focal concavity represented typical features of the early healing phase following lesion resection, rather than signs of surgical failure or recurrence. Vashisht et al. [18] reported a case of an 11-year-old male with a mass originating from the superior pole of the patella and extending into the suprapatellar pouch; intraoperatively, the lesion was predominantly cartilaginous and underwent complete surgical excision, though no long-term postoperative status was described. In contrast, the present case provides a comprehensive account of the entire healing continuum—from early postoperative repair to long-term symmetrical restoration of the articular contour—over a 1.5-year radiological follow-up period, thereby offering a valuable clinical reference for the surgical management of this condition.

This study is limited by its design as a single case report, which cannot fully account for individual variations, alternative treatment regimens, or the potential confounding effects of individual bone healing dynamics on long-term prognosis. Furthermore, although a 1.5-year follow-up period was conducted, the possibility of late disease recurrence cannot be definitively ruled out. Future research should leverage multicenter case accumulation to establish comprehensive DEH databases incorporating clinical, radiological, and histopathological parameters. Exploring the correlations between histological indicators and imaging parameters will provide a more objective foundation for accurately evaluating the biological behavior of lesions and guiding optimal clinical decision-making.

## **Conclusion**

Through the diagnostic and therapeutic course of dysplasia epiphysealis hemimelica of the right medial femoral condyle in a 9-year-old boy, this case serves

s as a critical reminder that this rare entity should not be overlooked in the routine evaluation of pediatric knee joint pathologies. For femoral condylar DEH involving the articular surface and presenting with prominent clinical symptoms, serial radiological follow-up demonstrated that surgical excision achieves highly satisfactory mid- to long-term bone remodeling and functional recovery. The clinical value of this case lies not only in confirming the feasibility and efficacy of surgical management, but also in demonstrating how integrating clinical, radiological, and histopathological findings enables precise diagnosis and individualized treatment for this rare condition.

#### **Abbreviations**

DEH: Dysplasia epiphysealis hemimelica

DR: Digital radiography

CT: Computed tomography

MRI: Magnetic resonance imaging

OCD: Osteochondritis dissecans

VAS: Visual analog scale

CBC: Complete blood count

MCV: Mean corpuscular volume

ESR: Erythrocyte sedimentation rate

ALP: Serum alkaline phosphatase

ROM: Range of motion

#### **Ethics approval and consent to participate**

Ethics approval was waived by the Ethics Committee of Ningbo No. 6 Hospital due to the retrospective and non-interventional nature of this case study. Written informed consent to participate was obtained from the patient's parents (legal guardians).

#### **Consent for publication**

Written informed consent for the publication of this case report, including all personal clinical details and accompanying clinical images, was obtained from the patient's parents (legal guardians). A copy of the written consent is available for review by the Editor-in-Chief of this journal.

#### **Availability of data and materials**

The datasets used and/or analyzed during the current study are available from the corresponding author upon reasonable request.

#### **Competing Interests**

The authors declare that they have no competing interests.

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#### **Authors' contributions**

DW and RH.F collected the data and drafted the original manuscript. R.J.P performed the radiological evaluation and image analysis. L.R.P performed the surgery and critical revision of the manuscript. All authors read and approved the final manuscript.

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