Trevor’s Disease: Management Difficulties and Proposed Classification

DAINE O. CLARKE, MBBS, BMEDSc (HON)

abstract

Dysplasia epiphysealis hemimelica, also known as Trevor’s disease, is a rare developmental disorder with osteocartilagenous overgrowth of the epiphysis or epiphyseal equivalent. The condition bears similarities to osteochondroma in terms of its radiographic appearance, but differs in its pathobiology and geographic occurrence. Unlike the metaphyseal occurrence of osteochondromata, it arises from the epiphysis. The clinical presentation is wide and varied, but mechanical symptoms and deformities predominate. Early reports of the condition suggested involvement of the lower limbs only. However, since then, numerous reports have indicated a more generalized distribution. Difficulties in management and recurrence rates seem to hinge on whether its origin is intra-articular or extra-articular. A new classification system to include these parameters is discussed. [Orthopedics.]

Trevor’s disease, also known as dysplasia epiphysealis hemimelica, is a rare, nonhereditary hemimelic developmental disorder of the epiphysis or other ossification center that is characterized by osteocartilagenous overgrowth, usually affecting the medial side. The incidence is approximately 1:1,000,000.\(^1\) The range of clinical manifestations is wide and varied, but mechanical symptoms and deformities predominate.\(^2\)

The natural history of dysplasia epiphysealis hemimelica is continued lesion growth until skeletal maturity, with associated early physeal closure.\(^3\) Intra-articular lesions tend to be complicated, with recurrence and fixed deformities.\(^4\) Smith et al\(^5\) recommended counseling parents with respect to the progressive nature of the disease and the need for follow-up until skeletal maturity. The condition, although benign, can have debilitating consequences.

This article describes a case of classic dysplasia epiphysealis hemimelica with intra-articular involvement in a female patient in Jamaica, an island with a population of 2,700,000. This report highlights the natural history of the condition, the unique challenges it presents, and the complications associated with inadequate follow-up.

Case Report

A 7-year-old girl with no known previous illnesses initially presented to the authors’ institution at 3 years of age with a history of a painless, hard mass of the left popliteal fossa and deformity of the left foot. Radiographs showed a calcified mass (Figure 1). Magnetic resonance imaging showed a 3.2 × 2.9 × 3.5-cm epiphyseal mass projecting toward the popliteal fossa (Figure 2). The lesion had a chondroid-type matrix, and a similar lesion was noted within the dome of the talus. A diagnosis of Trevor’s disease was made, and the natural history of the dis-

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ease, management options, and the need for follow-up were discussed with the patient’s parents.

The patient subsequently underwent open excision of both lesions. The postoperative period was uneventful, and the patient was discharged with a plan for follow-up in the outpatient department. When she was examined 3 months postoperatively, range of motion of the left knee was 0° to 100° and the left ankle had a 10° flexible equinus deformity. The patient was subsequently lost to follow-up and presented 4 years later with a 90° fixed flexion deformity of the left knee and rigid equinus deformity of the left ankle.

**DISCUSSION**

Dysplasia epiphysealis hemimelica was first described as a disease affecting the tarsus by Mouchet and Belot in 1926 under the appellation “tarsomegalie.” In 1950, in a review of 8 cases with postscript mention of a further 2 cases, Trevor coined the term “tarsoepiphyseal aclasis” after concluding that the disease was not confined to the talus but was more widespread. In 1956, Fairbank described the tendency of the condition to involve the medial or lateral half of the affected limb and coined the current term “dysplasia epiphysealis hemimelica” to describe the distribution.

The incidence is approximately 1:1,000,000. The male-to-female ratio is 3:1. Dysplasia epiphysealis hemimelica usually presents in the first 2 decades of life and is rare in adults. The lower limbs are affected more often than the upper limbs. The condition is usually confined to 1 aspect of the epiphysis, with the medial aspect affected more often than the lateral aspect.

In 1985, Azouz et al classified dysplasia epiphysealis hemimelica into 3 forms according to the phenotypic distribution: (1) localized, affecting a single bone, usually of the hind foot, but any epiphysis; (2) classic, affecting more than 1 bone of a single limb; and (3) generalized, involving an entire lower limb from pelvis to foot.

Since then, the wide distribution of the condition has been documented and published, and it is well recognized that dysplasia epiphysealis hemimelica is not confined to the lower limb. In addition, the location of the lesion (ie, intra- or extra-articular) affects clinical decision making. Bilateral cases also have been reported. The natural history of dysplasia epiphysealis hemimelica is continued growth until skeletal maturity, with associated early pathologic physeal closure. Intra-articular lesions tend to be complicated, with recurrence and fixed deformities. Therefore, the authors rec-

![Figure 1: Plain lateral radiograph of the left knee.](image1)

![Figure 2: Sagittal magnetic resonance image of the left knee.](image2)

**Table**

<table>
<thead>
<tr>
<th>Intra-articular</th>
<th>Extra-articular</th>
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<tbody>
<tr>
<td>Type A: Affecting the lower limb only</td>
<td>Type A: Affecting the lower limb only</td>
</tr>
<tr>
<td>1. Localized: Affecting a single bone</td>
<td>1. Localized: Affecting a single bone</td>
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<tr>
<td>2. Classic: Affecting more than 1 bone of a single limb</td>
<td>2. Classic: Affecting more than 1 bone of a single limb</td>
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<tr>
<td>Type B: Affecting the upper limb</td>
<td>Type B: Affecting the upper limb</td>
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<tr>
<td>Type C: Affecting both the upper and lower limbs</td>
<td>Type C: Affecting both the upper and lower limbs</td>
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<tr>
<td>Type D: Bilateral involvement/involvement of the spine</td>
<td>Type D: Bilateral involvement/involvement of the spine</td>
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ommend an alternate classification system (Table).

The management of Trevor’s disease is controversial. Because of the rarity of this condition, there is no level I evidence, and the literature consists of case reports and retrospective studies with limited numbers of patients. Treatment options range from simple observation to radical excision of lesions. The natural history of dysplasia epiphysealis hemimelica is cessation of growth once skeletal maturity is reached, and malignant transformation has not been documented to date.

Observation of asymptomatic lesions can be justified on these premises and has had good outcomes. The question remains which patients should be observed and which patients should undergo surgical intervention. The stratification of dysplasia epiphysealis hemimelica into intra-articular and extra-articular lesions may simplify this dilemma.

Excision of extra-articular lesions has led to much more favorable outcomes than excision of intra-articular lesions. Extra-articular lesions often can be excised safely. Because the natural history of dysplasia epiphysealis hemimelica is continued growth until the physis closes, all symptomatic extra-articular lesions should be removed. For this reason, removal of asymptomatic extra-articular lesions should be considered.

The approach to intra-articular lesions has no simple algorithm. Management of this subset of lesions must be tempered. Some authors have suggested observation only, extra-articular osteotomy to correct deformities, and even excision. Reports of outcomes after excision of intra-articular lesions vary. Kuo et al reported a high rate of complications after excision of intra-articular lesions, whereas Bosch et al reported excellent results with early intervention. The only clear indication for excision of intra-articular lesions is the presence of symptomatic loose bodies.

**CONCLUSION**

The pathology of dysplasia epiphysealis hemimelica continues to be clarified with advances in molecular techniques. The incidence may be higher than previously reported. The pathology is not confined to the lower limbs, as previously thought. Regardless of the anatomic location, the single most important factor affecting long-term morbidity seems to be whether the lesion is intra-articular or extra-articular. A classification system that includes these parameters may be useful as knowledge of this condition expands. The natural history is for continuous growth of the lesion until cessation of physal growth. Regular follow-up, regardless of the subtype, is needed.

**REFERENCES**