CASE REPORT

Dysplasia epiphysealis hemimelica of the ankle: a fracture-like rare developmental disorder

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SUMMARY

Dysplasia epiphysealis hemimelica is a rare developmental disorder which affects the epiphyses. We report a case of the disease located in the ankle joint, referred to our clinic with the initial misdiagnosis of a Salter-Harris 3 type fracture of the distal epiphysis of the tibia. After correct diagnosis, the patient was treated surgically with the excision of the cartilaginous masses. Taking an accurate medical history and performing adequate imaging studies is essential in diagnosing and treating this disease. Fracture-like epiphyseal configurations in patients with no history of injury should raise suspicion of periarticular developmental disorders.

BACKGROUND

Dysplasia epiphysealis hemimelica (DEH), or Trevor's Disease, is a rare developmental disorder of the epiphyses in children.¹ The disease results in the formation of benign cartilaginous tumours, arising either from the growth plate or the articular cartilage.¹ The lesions are most commonly seen in the distal tibia and the distal femur and often involve one side of the epiphysis,¹ which is why the term 'hemimelica' is used to describe the disease. The incidence of DEH is 1 in 1 000 000² and the male to female ratio is 3:1.³ The disease appears to be non-hereditary.⁴ The lesions are not premalignant.¹ The radiographic appearance of DHE may be confused with an epiphyseal fracture resulting in inappropriate treatment. We outline diagnostic and radiological criteria along with the indicated treatment in a case of distal tibial DHE.

CASE PRESENTATION

We report a case of DEH affecting the ankle joint which was treated in our department. We are a tertiary referral centre for paediatric orthopaedics associated with an academic institution. The study was approved by the Ethics, Research and Development Committee of our Institution and consent was obtained by the patient for reporting the case. The patient, an 11-year-old boy, was referred to our department from a provincial hospital with the initial diagnosis of a Salter-Harris 3 fracture of the distal tibial epiphysis, evident only on the lateral (figure 1) X-ray of the ankle and not on the anteroposterior (figure 2). There was a history of pain in the ankle after running or prolonged walking, but there was no trauma. During the initial assessment, the patient had a limitation of dorsal extension and plantar flexion of at least 10° compared with the non-affected joint.

INVESTIGATIONS

The lateral X-ray of the ankle gave the impression of a Salter-Harris 3 fracture of the distal epiphysis of the tibia, and therefore a CT scan with three-dimensional (3D) reconstruction was carried out, revealing the presence of three bony masses in the anterior of the ankle joint (figures 3–5). Owing to the obscurity of the radiographic appearance, the case was referred to our paediatric orthopaedic unit for diagnosis and treatment. Further investigation by MRI was not necessary due to the diagnosis being reached based on symptoms, location, age of the patient and CT scan images.

DIFFERENTIAL DIAGNOSIS

The diagnosis of DHE was based on the non-traumatic occurrence of the event, the anatomic location of the lesion and the congruency of the epiphyseal plate.

TREATMENT

The patient was treated surgically for the removal of the masses (figures 6 and 7). A below-knee cast was placed after the surgery and removed in a

Figure 1 Lateral X-ray of the ankle.

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Figure 2 Anteroposterior X-ray of the ankle.

month's time. X-rays were taken after surgical procedure (figures 8 and 9), confirming the removal of all masses. A confirmation biopsy was sent to exclude malignancy.

OUTCOME AND FOLLOW-UP

Two months after the surgery, the boy had returned to his full activity. Three years postoperation, on clinical examination, although there was no restriction in plantar flexion, there was a

Figure 3 Three-dimensional CT scan reconstruction; anteroposterior view of the ankle.

Figure 4 Three-dimensional CT scan reconstruction; lateral view of the ankle.

10° reduction in dorsi flexion. The patient, however, reports no restriction in his activities. An anteroposterior and a mortise view X-ray of the ankle (figures 10 and 11) revealed a recurrence of a small mass lesion. Since the patient has no serious symptoms, a new follow-up was suggested in a year's time.

DISCUSSION

The cause of DEH is unknown. It results from an abnormal control of cell proliferation in the epiphysis leading to bone overgrowth with a cartilage cap that projects into the adjacent joint.^{5 6} The most commonly affected lower limb joint is the ankle (43.2%).⁷ According to Azouz *et al*,⁸ DEH is classified

Figure 5 CT scan; lateral view of the ankle.

Figure 6 Surgical approach of ankle.

into 3 groups: localised, that affects only one epiphysis; classic, that affects more than one epiphysis in the same limb; and generalised, that involves the entire lower limb. Symptoms of DEH include functional impairment and limitation of range of movement, deformity and swelling, and in some cases, pain and wasting of the muscles that move the affected joint. Gait abnormalities or limb length discrepancy can also be present.^{9–11} In simple radiographs, DEH, in its early stages, presents as an irregular lesion rising from the affected epiphysis which gradually ossifies and usually becomes confluent with the underlying bone.^{12–13} Asymptomatic lesions can be treated non-surgically.¹⁴ Surgical removal of the lesions has good long-term results, although in a recent review article an incidence of 15% knee or ankle osteoarthritis was reported, 7.7% of which will need arthrodesis of the ankle joint.⁷

Correct diagnosis is most important in DEH. In most cases, diagnosis is delayed due to the fact that the symptoms are non-specific and also because the diagnosis is not made during the first visit to the orthopaedic clinic.¹⁵ DEH has been misdiagnosed as Perthes disease, chondroblastoma, calcified post-traumatic haematoma and idiopathic hypertrophy of the femoral condyle.¹⁵ DEH has also been mistaken for synovial chondromatosis, which is an adult disorder.^{4–16–18} A patient with DEH was misdiagnosed as having post-traumatic osseous

Figure 7 Tumours from ankle after surgical removal.

Figure 8 Postoperative mortise view X-ray.

fragments in the joint and was followed up for 10 years, because of a trauma history.¹⁸

To the best of our knowledge, no case in the literature concerning DEH to date has been reported to have been misdiagnosed as a recent fracture and specifically as a Salter-Harris

Figure 9 Postoperative lateral X-ray.

Figure 10 Follow-up mortise view X-ray.

3 type fracture of the distal epiphysis of the tibia. In the case of the 11-year-old boy, the lesion gives the impression of a Salter-Harris 3 type fracture, specifically in the lateral radiograph of the ankle. In the CT scan performed, followed by 3D reconstruction, it became evident that there was no fracture in the ankle, but rather three lesions in front of the tibial epiphysis.

Figure 11 Follow-up lateral X-ray.

Therefore, taking an accurate medical history and also using complementary imaging studies as well as simple radiographs, are essential in diagnosing DEH and treating it. A 3D reconstruction of the CT scan is also useful in preoperative planning.¹⁹ Correct diagnosis is very important as it may save the patient from unnecessary surgery and, if the surgery is necessary, it may help in performing it correctly.¹⁵

It is argued that with MRI, it is easier to evaluate the epiphyseal osteochondral growth and its probable effects on adjacent structures, allowing for a better definition of bone and cartilage structures at multiple planes, but scarce reports describing MRI findings of DEH are found in the literature.^{9 20 21} MRI is indicated as a useful investigation in the early phases of the disease, when a small mass with calcifications inside is observed; this, however, was not the case in our patient.^{20 21} Additional drawbacks concerning MRI are an inability to perform on an emergency basis as in this case of initial wrong diagnosis of a fracture requiring immediate treatment and, furthermore, the need for anaesthesia when performing MRI in younger patients.

Learning points

- ▶ DEH is a rare disease which may cause a disability in ankle function. Although benign, it is not easy to diagnose and in several cases, it is primarily misdiagnosed.
- ▶ Diagnosis is made possible after taking an accurate medical history as well as performing supplementary imaging studies apart from simple radiographs.
- ▶ A CT scan accompanied by three-dimensional reconstruction can be an adequate imaging study in order to make the correct diagnosis and also evaluate the completion of treatment, when surgical removal of the lesion is warranted. MRI can be required in early stages of the disease when the mass is mostly cartilaginous.
- ▶ Surgical removal of masses, biopsy to exclude malignancy and follow-up of the patient is advised.

Competing interests None declared.

Patient consent Obtained.

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