Dominant carpotarsal osteochondromatosis

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Abstract
Dominant carpotarsal osteochondromatosis is a particular disorder of the wrist and tibiotalar joints with abnormal bone proliferation and osteochondromas. Two patients, a mother and son, are described here; a similar condition has previously been described in seven affected members of a family. The upper and the lower limbs are affected in the same patient and the lesion can be bilateral. Autosomal dominant inheritance is a further criterion allowing the diagnosis of dysplasia epiphysealis hemimelica.

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In the group of chondrodysplasias with disorganised development of cartilage, multiple cartilaginous exostoses, enchondromatosis, and other entities have already been defined, but a number of disorders remain insufficiently characterised. In the following description we delineate a particular disorder which has been confused with dysplasia epiphysealis hemimelica; we propose to name this condition 'dominant carpotarsal osteochondromatosis'. We report two cases in one family.

Case reports
Case 1
Case 1, a boy, was seen for the first time at the age of 16 years for swelling and restricted mobility of his left ankle. The deformity was limited to the medial and posterior part of the ankle. Flexion and extension at the tibiotalar joint and inversion and eversion at the subtalar joint were restricted. The initial radiological examination of the foot showed a fragmented, irregularly shaped, osteocartilaginous tumour behind the calcaneus and the talus. This mass, which could not be delimited from the adjacent bones, partially occupied the subtalar joint, expanding medially under the malleolus (fig 1). A second, round, irregular, ossified structure was noticed next to the fibula. A lateral radiograph from the right foot was normal. Six months later these osteochondromas were removed surgically. After six months the patient presented with a swelling on the palmar surface of the right wrist. Additionally he complained of limited flexion of the right knee with one episode of locking.

A radiograph of the right hand showed an osteocartilaginous mass on the proximal end of the second metacarpal expanding laterally towards the trapezoid (fig 2). X rays of the right knee showed a sharply delimited peripheral defect of the medial femoral condyle. MRI showed an osseous sequestrum from the medial portion of this condyle. These findings were in accordance with osteochondritis disseccans (fig 3).

When he was last seen at the age of 17 years, growth and development were normal (173 cm, 69 kg). Minor asymmetry of the lower limbs with a shortening of 2 cm of the left leg was noticed. Flexion of the right knee was slightly limited. There were hard, deforming swellings under the medial malleolus of the ankles on both sides, particularly on the left (fig 4). A small, hard, and painless protuberance was observed on the palm of the right hand. The radiographs of the feet confirmed the recurrence of the tumours in the left foot. On x rays of the right foot, an osteocartilaginous mass was visible in front of the tibia just above the talus (fig 5).

X rays of the right hand showed no change from the previous result and the left hand showed no abnormality. Radiologically the
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rest of the skeleton was normal apart from minor deformities of the 10th to 12th dorsal vertebrae, which showed slight irregularities of the end plates, most probably a result of unnoticed Sheuermann's disease.

An axial CT scan of the left ankle and foot (Dr M Roussey) confirmed the alteration of the tarsal bones, particularly the talus and the calcaneus, which showed an irregular shape and newly formed ossifications, especially in the central part of the bones (fig 6). There was partial synostosis of the calcaneoscapoid joint. The lateral part of the scaphoid and the cuboid were also involved, whereas the first cuneiform appeared normal. The sagittal reconstructions showed the existence of disorganised ossification in front of and behind the ankle as well as a deformity of the neck of the talus of the tibiotarsal joint.

CASE 2
Case 2, aged 52 years, was the mother of case 1. She told us that a deformity of her right wrist was first noticed in adolescence but has not developed since then. Clinical examination showed normal growth (168 cm). The ankles were normal. However, a swelling about 1.5 cm in diameter could be palpated on the palmar side of the left wrist.

Radiographs taken at the age of 33 years showed an osteocartilaginous proliferation involving the lateral part of the carpus trapezium, trapezoid, and capitate. This swelling extended to the palmar surface of the wrist. In addition, an isolated, triangular shaped ossified structure could be seen distal to the carpus and a second smaller one between the fourth and fifth metacarpal bones (fig 7). There had been no progression over the last 20 years as shown by recent x-rays. On the left foot an irregularly shaped proliferation was seen on the proximal end of the first metatarsal bone.
Discussion
The features described here have previously been observed by different authors but have been attributed to dysplasia epiphysealis hemimelica, as in a family with seven affected members in two generations reported by Hensinger et al.2 The lesions in these patients were the same as those described in our patients. Their distribution was also very similar. A family was previously reported in which one patient had epiphyseal lesions of the knee comparable to those seen in dysplasia epiphysealis hemimelica.1 The patient’s father had similar changes of the wrist and the tibiotalar joints. However, the author did not mention bilateral involvement and, in spite of the inheritance, it is impossible to affirm that Silverman1 described the same disorder as we report here.

The case reported by Hensinger et al2 and our cases certainly represent a distinct entity, clearly different from dysplasia epiphysealis hemimelica as illustrated by the radiological features, the localisation of the lesions, and the autosomal dominant inheritance. The nature of the lesions is multiform and they can be very similar to those observed in dysplasia epiphysealis hemimelica; initially such a diagnosis was proposed in our first patient. In addition, the abnormalities of the wrist in our second case resemble the lesions of the upper limbs observed in dysplasia epiphysealis hemimelica. This applies to several cases in the report of Hensinger et al.2

Some affected bones show disorganised development; their volume is increased, their contours are irregular, and they frequently have a fragmented aspect and small ossification points in the periphery.

As well as these abnormal proliferations intra- and extra-capular osteochondromas can be found. They appear completely independent from the adjacent skeletal structures. In our first patient such osteochondromas were found in the subtalar region and in our second patient between the fourth and fifth metacarpals. They were also described in some of the patients of Hensinger et al.2

The development of the skeletal lesions progresses during the growth period but stops after puberty as seen in our second patient. Comparison of the radiographs at the age of 52 years with those at the age of 33 showed no obvious modifications of the lesions.

The localisation of the lesions should be emphasised: as in dysplasia epiphysealis hemimelica, the bones of the carpus and the tarsus are exclusively involved. However, the involvement, although asymmetrical, can be bilateral as was observed in our first patient who presented with lesions of both feet. Bilateral involvement was also reported in cases 2, 3, and 5 in the report of Hensinger et al.2

The simultaneous involvement of the upper and the lower limbs should also be noted. The knee can also be involved and the presence of osteochondromas in the knee was noted by Hensinger et al2 (case 4). Furthermore, osteochondritis dissecans was found in our first patient. We think that most probably this is not a coincidental occurrence.

Though dysplasia epiphysealis hemimelica may have variable manifestations,4 its mode of inheritance is a further criterion allowing one to exclude this diagnosis with confidence: it is usually sporadic whereas carpotarsal osteochondromatosis is transmitted in an autosomal dominant manner with variable expression in both our family and that of Hensinger et al.2

No other autosomal dominant disorder with disorganised cartilaginous development, such as metachondromatosis and multiple cartilaginous exostoses, can be confused with the form presented here. These disorders present typical exostoses of the fingers which are quite different from the lesions described here. We therefore think it is useful to distinguish this rare disorder from dysplasia epiphysealis hemimelica with which it should not be confused and for which genetic counselling will be quite different.