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DYSPLASIA EPIPHYSEALIS HEMIMELICA

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Four typical cases of dysplasia epiphysealis hemimelica are reported, the age when first examined varying from 18 months to 5 years. The observation periods varied from 3–20 years. After fusion between the accessory ossification centres of the osteochondroma and the regular epiphysis, no further enlargement was observed. In three cases an enlarged medial femoral condyle was resected, in two cases the resection was done through bone, leaving a non-cartilaginous surface in the knee joint. This procedure seems to have no harmful consequences; one of the patients seen 19 years later had no complaints, and the knee joint appeared almost normal on X-rays.

Key words: epiphysis; osteochondroma; exostosis; child; growth

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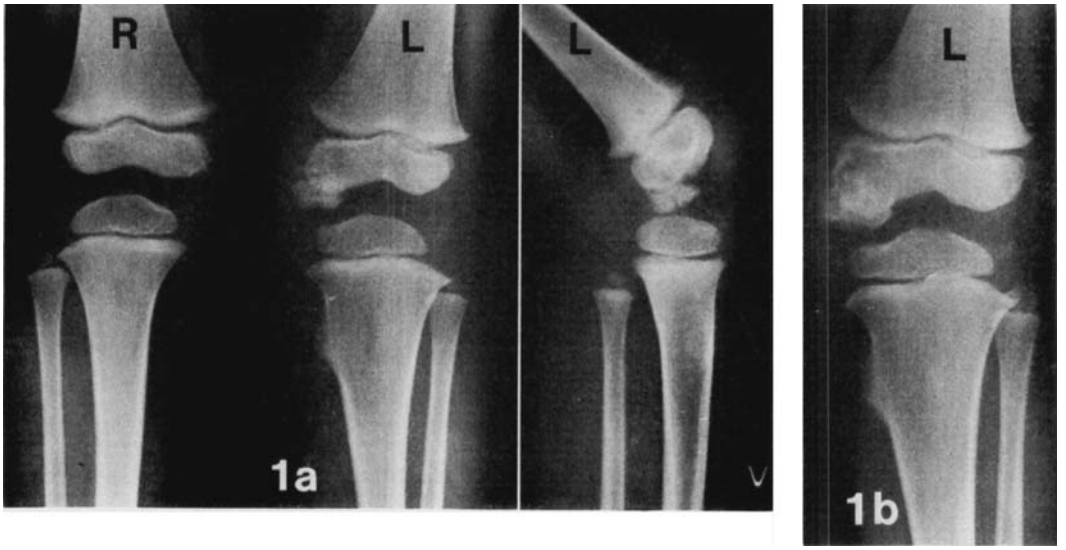
Dysplasia epiphysealis hemimelica is a growth disorder of unknown aetiology. The characteristic pathological finding is an overgrowth of the cartilaginous cells on one side of the affected limb, resulting in formation of an osteochondroma. The medial side is affected more often than the lateral. The talus, the distal femoral and the distal tibial epiphyses are most frequently involved (Theodorou & Lanitis 1968).

The disorder was first described by Mouchet & Belot (1926) using the term "Tarsomégalie". Fairbank (1956) reported 14 cases from orthopaedic centres in England and introduced the name dysplasia epiphysealis hemimelica. He presumed that endogenous or exogenous agents affect the limb buds of the foetus for a very short time in a minute area. The clinical manifestation may vary, depending on the area affected and possibly also on the time at which the agents act.

To date about 60 cases of this disorder have been reported (Barta et al. 1973). The purpose of the present paper is to add four typical cases (one having an observation period of nineteen years following surgery) and to stress some aspects of importance concerning treatment of this rare disorder.

CASE REPORTS

Case 1. A girl, 3 years old when first seen at the hospital because of increasing valgus deformity of the left knee. She had no other complaints. A bony prominence on the medial femoral condyle was found but no limitation of movement of the knee joint. X-rays (Figure 1 a) showed an enlargement of the medial femoral condyle with centres of ossification separated from the regular ossification of the epiphysis. Further the X-rays showed an irregularity in the outline of the medial side of the proximal tibial

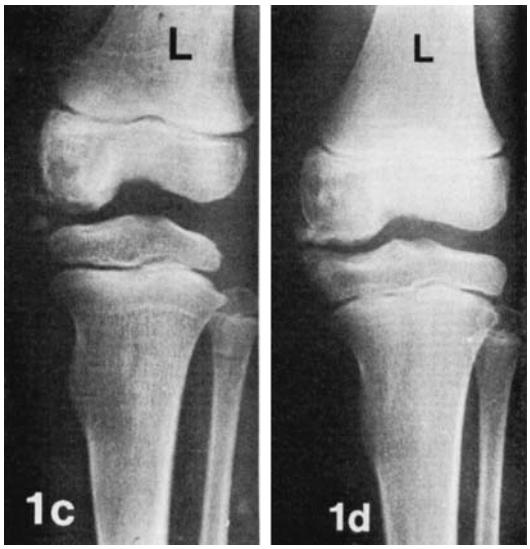


epiphysis, and an exostosis on the medial side of the proximal metaphysis. During an observation period of 1 year, the deformity increased and the separated ossification centres fused with the regular ossification centres of the epiphysis (Figure 1 b). Arthrotomy was done and the bony prominence of the femoral condyle was removed, leaving a bony joint surface in the knee. Histopathological examination showed normal cartilage and bone.

Two months after operation the patient had a normal range of movement in the knee joint and no complaints. X-rays 1 year after operation (Figure 1 c) showed that the irregular ossification centres had been only partly removed. On the medial side of the tibial epiphysis a new separate ossification centre was also seen. At the age of 8 (Figure 1 d), the irregular ossification centre of the tibia had fused with the epiphysis. The exostosis of the metaphysis had not increased in size. At the age of 13 (Figure 1 e) the joint surfaces of the tibia as well as the femur were almost normal. An increased breadth of the tibial metaphysis was observed at the place where the exostosis had been previously. At 23 years of age she had no complaints regarding the knee joint; X-rays showed some flattening of the joint surface of the medial condyle, but were otherwise normal (Figure 1 f). The lower limb involved was 2 cm longer than the other. The same observation had been made when she was 13.

Case 2. A boy, 2 years old when first seen at the hospital. He had had periods of limping on the left leg and a locking phenomenon of the knee joint. A slight valgus deformity of the knee was found, but the range of movement was normal. X-rays (Figure 2 a) showed a separate centre of ossification near the medial femoral condyle. One year later X-rays showed multiple separated ossification centres and further enlargement of the medial femoral condyle (Figure 2 b). The enlargement was removed by resection through the cartilage separating the epiphysis from the irregular ossification centres. The femoral condyle could therefore be left covered with cartilage. A corpus liberum found in the knee joint was the reason for the locking phenomenon. It consisted of hyalin cartilage covering an ossification centre and had probably become separated from the femoral condyle. The histopathological examination of the removed enlargement showed normal cartilage and bone structure. Two months after operation the patient had no complaints and the knee function was normal. At 12 years of age the patient had no problems with his knee joint, and the X-rays were entirely normal (Figure 2 c).

Case 3. A boy, 5 years old, was first seen because of pain in the right knee. He presented with a slight valgus deformity of the knee and ankle. X-rays (Figure 3 a) showed an enlargement of the medial femoral condyle with ossification centres almost totally fused with the epiphysis. Separated ossification centres were also seen around the talus, navicular and first cuneiform, some fused with the bone (Figure 3 b). Operative correction was not found necessary. At the age of 8 he had no complaints, and the knee joint movement was normal. No increase in the valgus deformity or enlargement of the femoral condyle had occurred. On the contrary the out-



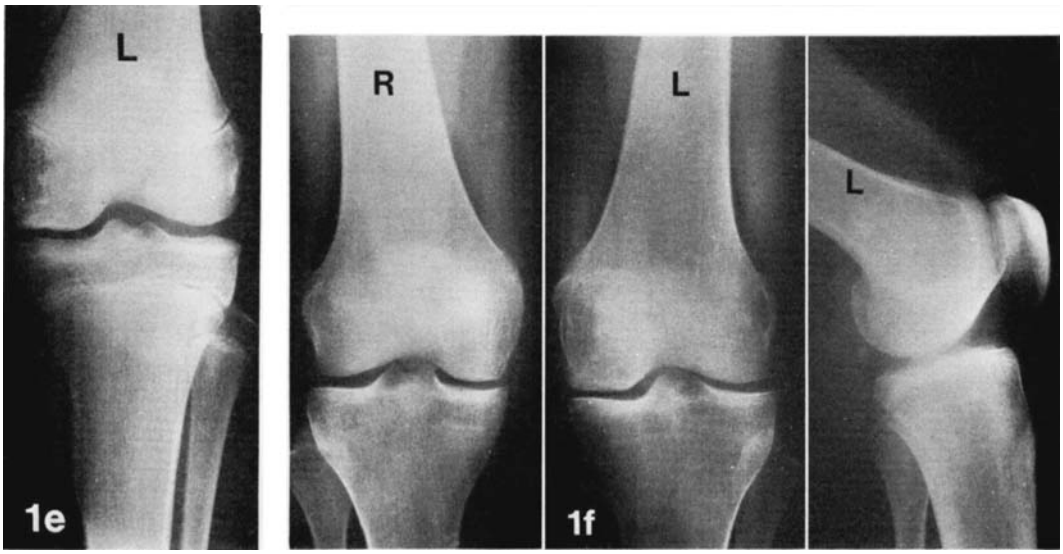
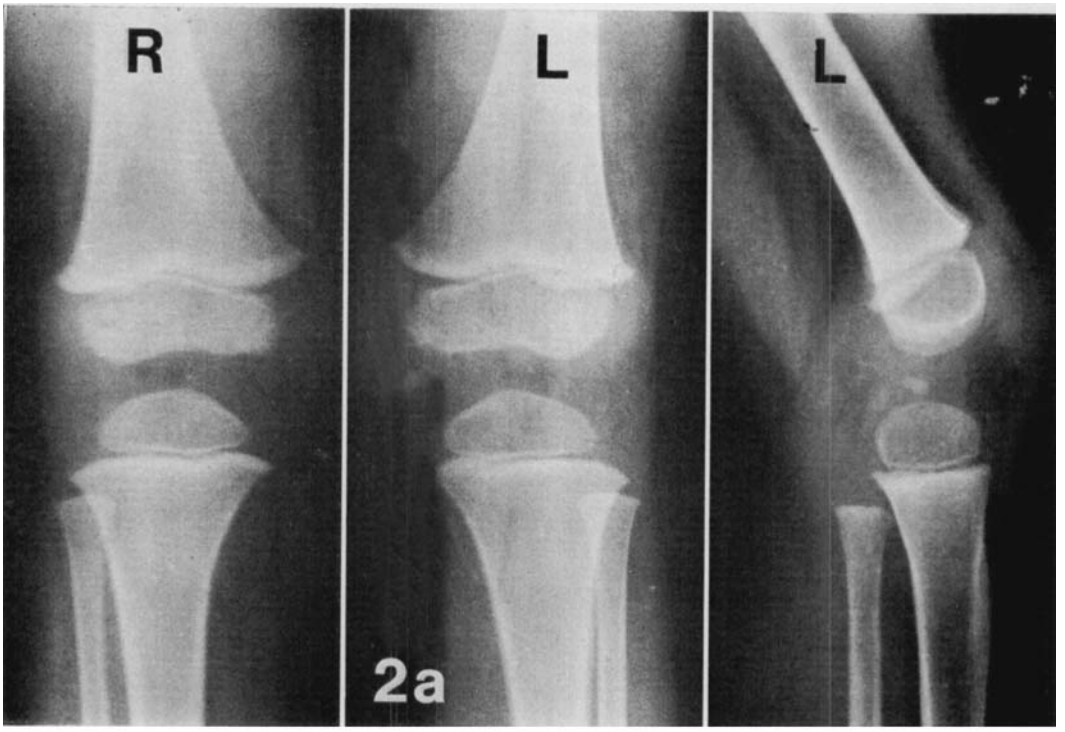


Figure 1. Case 1. a) X-rays of the knee joints of a girl 3 years of age. Separated ossification centres are seen at the left medial femoral epiphysis. An exostosis is present in the tibial metaphysis. b) The osteochondroma was fused to the epiphysis at 4 years. c) The knee at 5 years, 1 year after resection of the femoral condyle. A new irregular ossification centre is seen at the medial tibial epiphysis. The X-rays of the left knee at 8 (d), at 13 (e) and at 23 years (f) show a spontaneous development towards an almost normal joint.

growth was fully fused with the epiphysis and seemed relatively small because of the increase in the size of the epiphysis. The shape of the condyle was more normal (Figure 3 c). There was likewise no worsening in the ankle region.

Case 4. A boy, who had been treated since the age of 18 months because of a valgus deformity of the left knee. He had no complaints. X-rays (Figure 4 a) showed enlargement of the medial femoral condyle with multiple centres of ossification, and on the tibial side an exostosis was seen on the proximal tibial metaphysis. At 5 years of age the patient was first referred to hospital because of a marked valgus deformity of the knee. The affected limb was 2 cm longer than the other. He also had a swelling of the medial malleolus. X-rays (Figure 4 b) showed that the irregular ossification centres of the femur had fused with each other and also with the ossification centre of the epiphysis. The exostosis of the tibial metaphysis was seen only as an increased breadth of the proximal metaphysis. X-rays of the ankle region (Figure 4 c) showed separate centres of ossification in the medial malleolus. At operation the cartilage covering the enlarged femoral condyle was found to be very thin and bluish. The prominence of the femoral condyle was resected leaving a raw bony surface in the joint. No cartilaginous space was found between the ir-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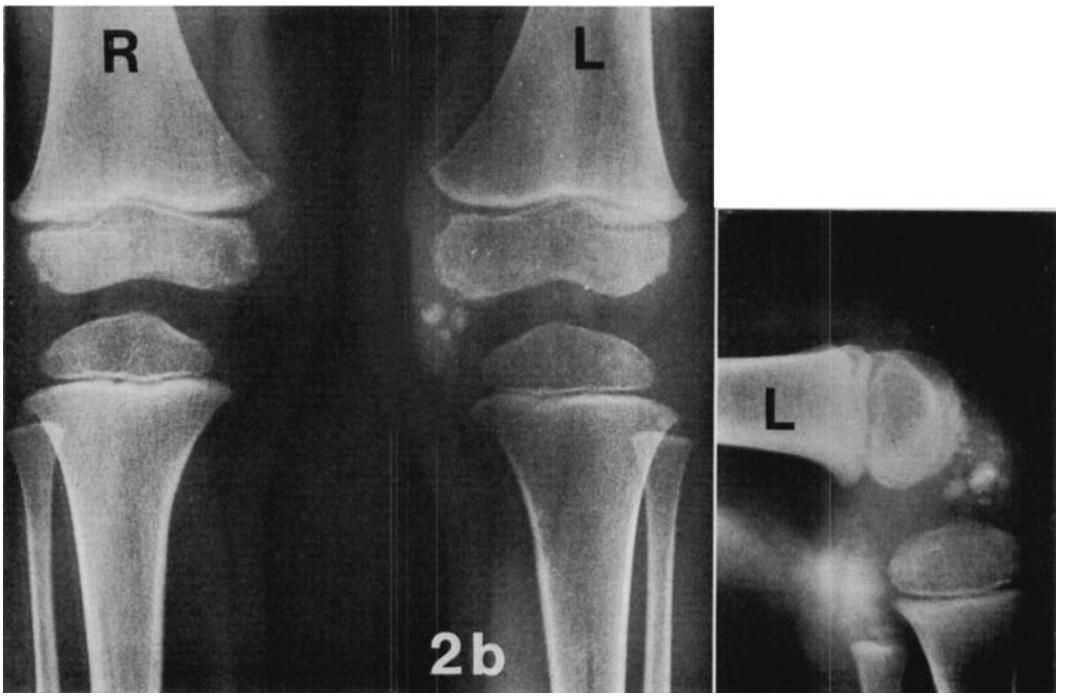
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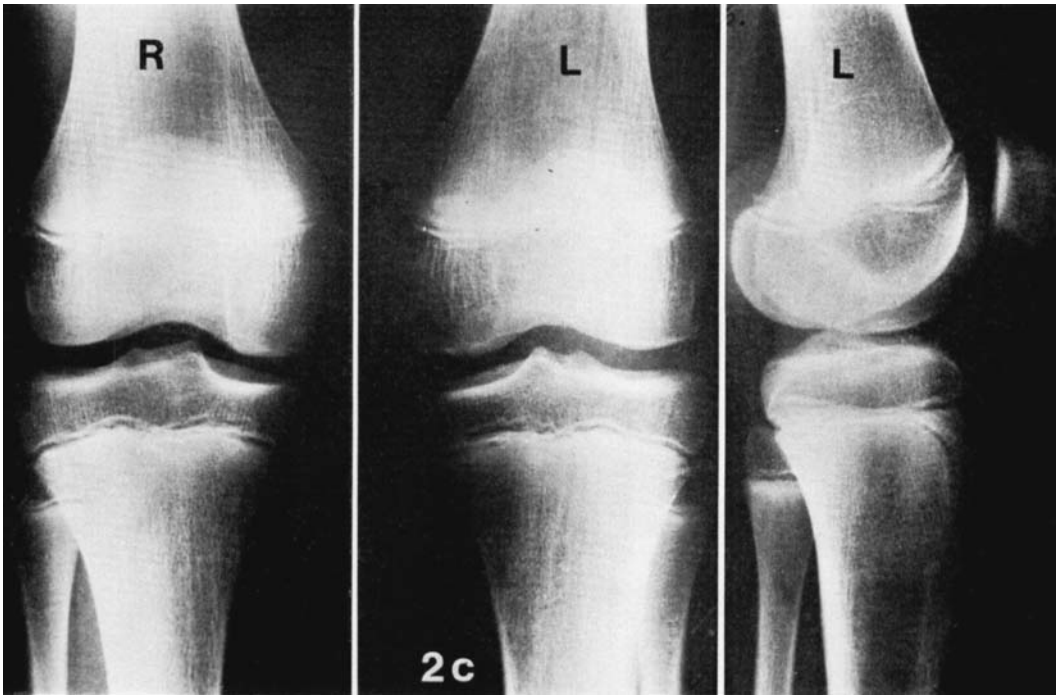


Figure 2. Case 2. a) X-rays of the knee joints of a boy at the age of 2 years showing separate ossification centres near the medial femoral epiphysis. b) Pre-operative X-rays at 3 years. c) Normal joints at 12.

regular ossification centres and the epiphysis. Histopathological examination showed normal bone and cartilage. One year after operation the boy had no complaints regarding the knee or the ankle joint.

DISCUSSION

Four cases of dysplasia epiphysealis hemimelica have been treated at the Sophies Minde Orthopaedic Hospital. The age when first observed medically varied from 18 months to 5 years. Three of the patients were male and one female. In two cases the left and in two cases the right lower limbs were affected, in all cases on the medial side. The medial femoral condyle was affected in all cases. In one case this was the only localisation. The upper tibial condyle, the lower tibial epiphysis, the talus, the navicular and the medial cuneiform, were each affected in one instance. The distribution according to sex, side, and affected epiphysis is in agreement with other publications (D'Angio et al. 1955, Theodorou & Lanitis 1968).

Two of the cases reported here also had metaphyseal affection, in both instances as an exostosis of the tibia metaphysis well separated from the epiphysis. This must be interpreted as a true metaphyseal abnormality and not as a secondary manifestation as stated by Kettelkamp et al. (1966). After some years the metaphyseal exostosis in our cases was seen only as an increased breadth of the metaphysis (Cases 1 and 4). Metaphyseal affection has earlier been reported by several authors (Ingelrans & Lacheretz 1953, Fairbank 1956, Mosely 1957, Saxton & Wilkinson 1964, Trevor 1950). This metaphyseal affection, to date observed in 11 typical cases seems to be a part of the growth disorder. The term "epiph-

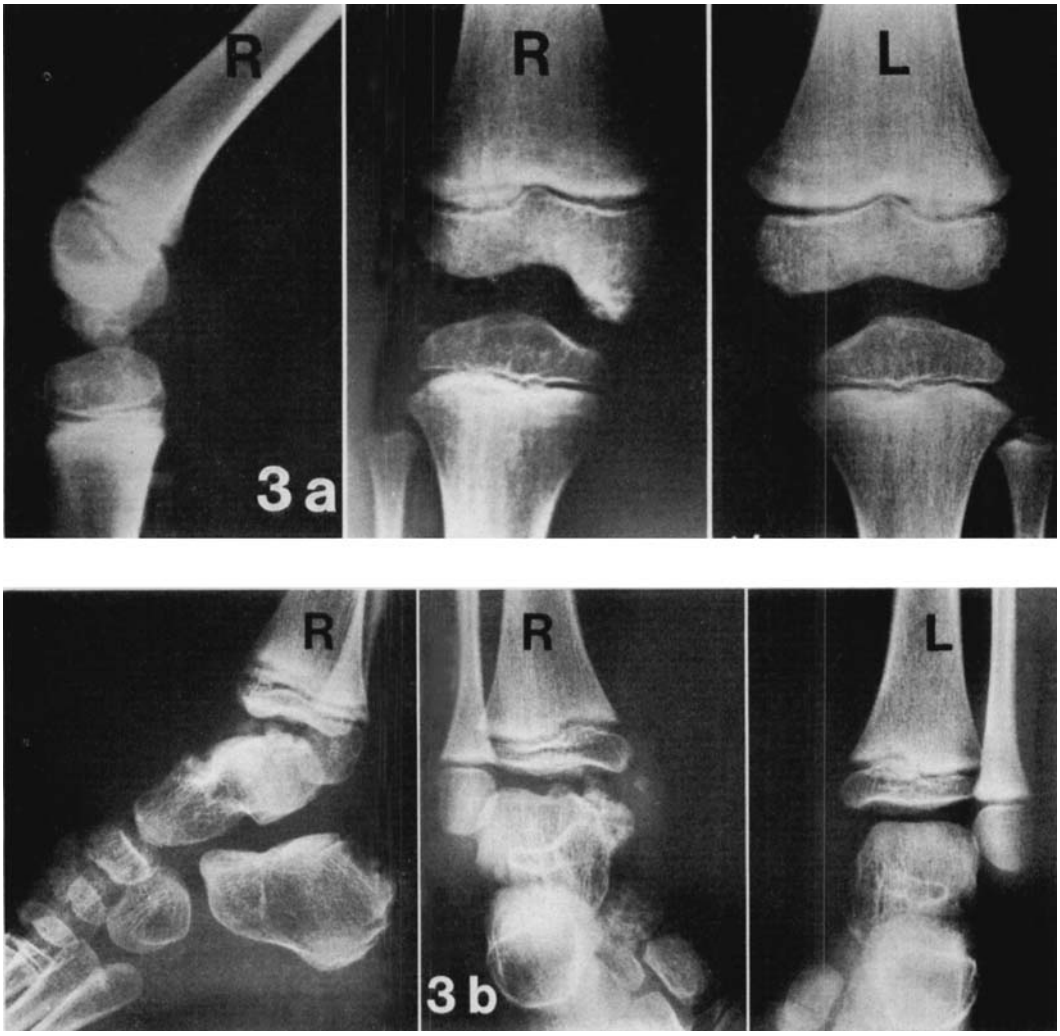
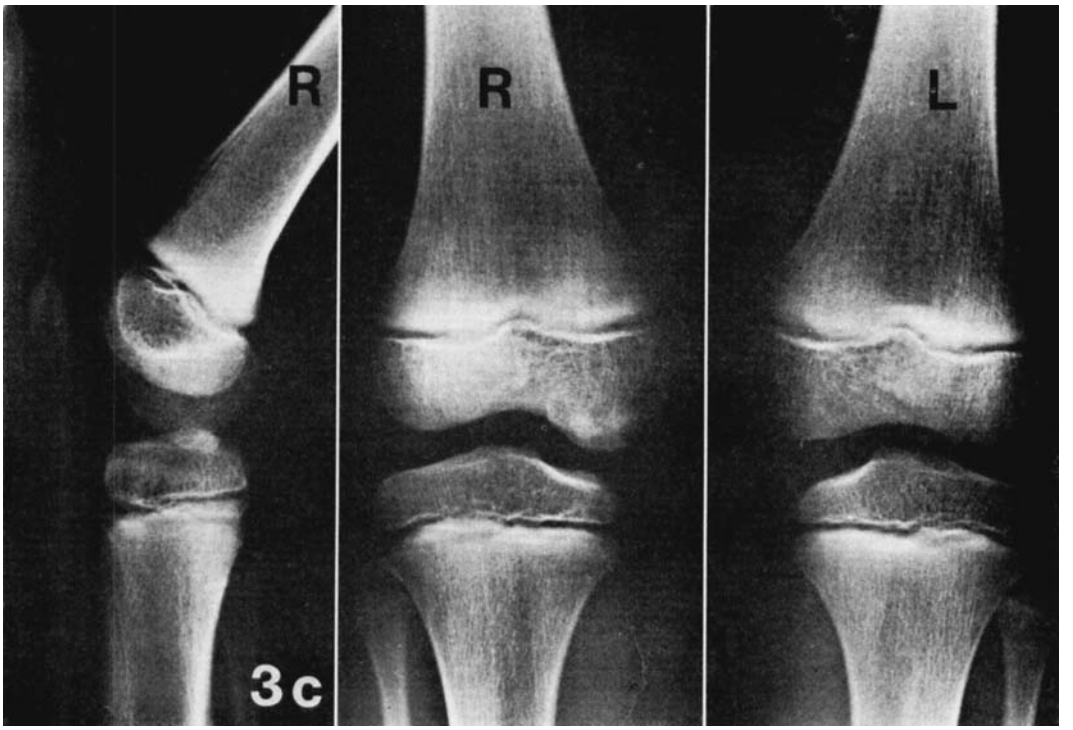


Figure 3. Case 3. a) X-rays of the knee joints of a boy at the age of 5 showing an enlargement of the right medial femoral condyle. b) X-rays of the ankle and foot of the same patient showing multiple centres of ossification around the talus, navicular and first cuneiform. c) X-rays at the age of 8 show some spontaneous improvement of the knee affection.

yseal", used in describing this condition, is therefore not strictly correct. In two of our cases the affected limb was found to be longer than the unaffected one. This finding has also been reported by others (D'Angio et al. 1955, Rechnagel 1960, Saxton & Wilkinson 1964) and probably reflects an increased growth rate of the whole limb.

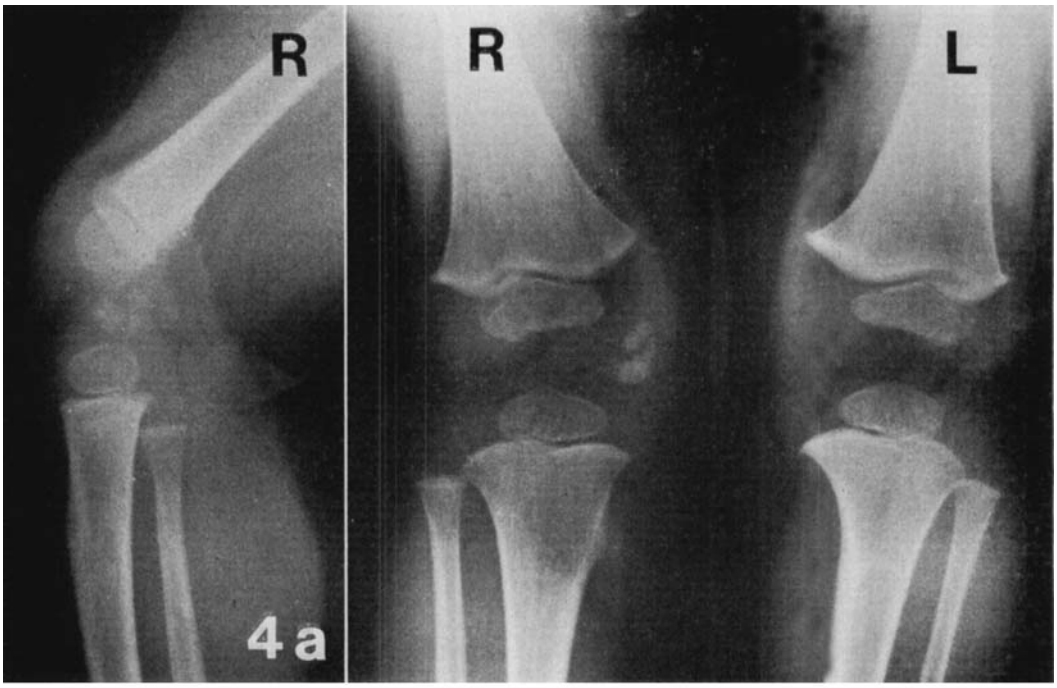
The initial complaints were deformity of the joint, limping, pain, and in one patient a locking phenomenon in the knee joint caused by a corpus liberum. In all cases a valgus deformity of the knee was found. One patient had a valgus deformity in the ankle joint, caused by affection of the medial malleolus. Another only had an enlargement on the inside of the ankle, caused by affection of the talus.



Histopathological examination showed, in all cases, an osteochondroma with groups of proliferating normal cartilaginous cells and centres of ossification. This is in agreement with the findings of other authors (Donaldson et al. 1953, Rechnagel 1960, Barta et al. 1973).

The four cases reported here have been followed up for from 3 to 20 years. In the femoral condyle the separated ossification centres fused with the regular epiphysis in two cases at an age of 3–4 years. In one case, first seen at the age of 5 years, the fusion had already taken place. The enlargement of the femoral condyle increased in this period before fusion (Cases 1, 2 and 3). In two cases there was no further enlargement of the condyle after fusion, in fact some normalisation was seen (Cases 1 and 3). A similar course was also found with regard to the tibial epiphyseal affection and the tibial metaphyseal exostosis. This observation is of importance with regard to treatment of this growth disorder. An explanation for this course could be found in the observation of Barta et al. (1973). They described an almost normal growth cartilage between the epiphysis and the separated ossification centres. Probably most of the growth of the enlargement takes place in this "growth plate"; accordingly, after fusion, the osteochondroma does not increase in size to any appreciable extent.

Surgery was found necessary in three cases, all because of an increased valgus deformity in the knee joint. Arthrotomy was done and the enlarged medial femoral condyle was resected. In one case the operation was done at the age of three, before fusion, leaving a cartilage-covered surface in the joint. In two cases the operation was done after fusion, leaving a bony surface in the knee joint. One of these patients was reexamined 19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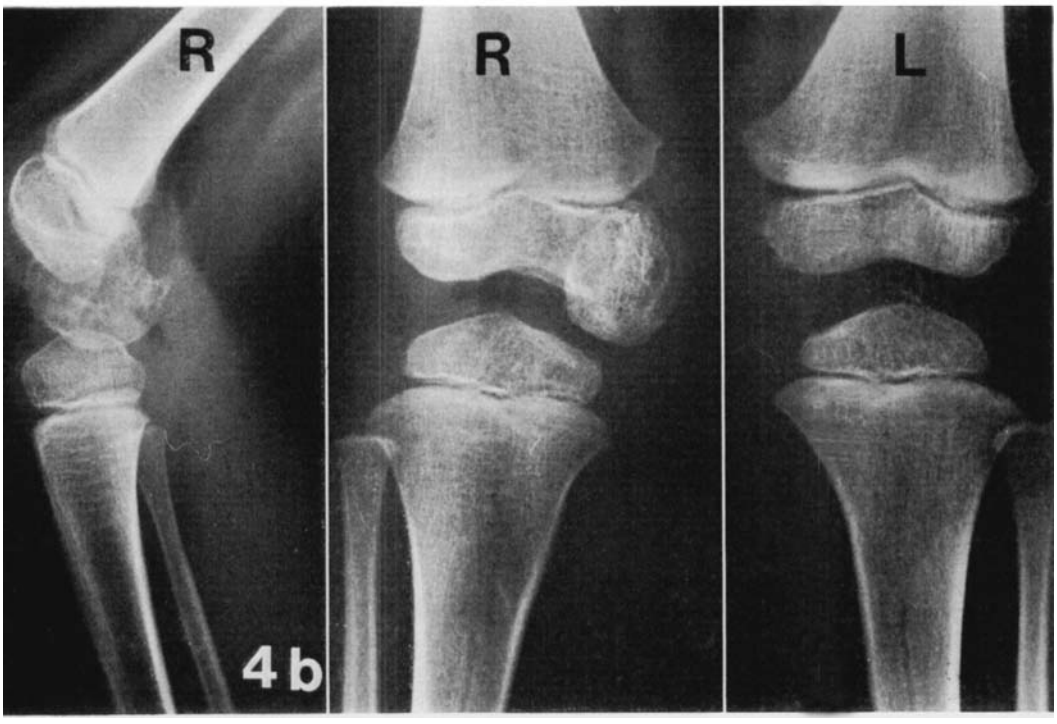
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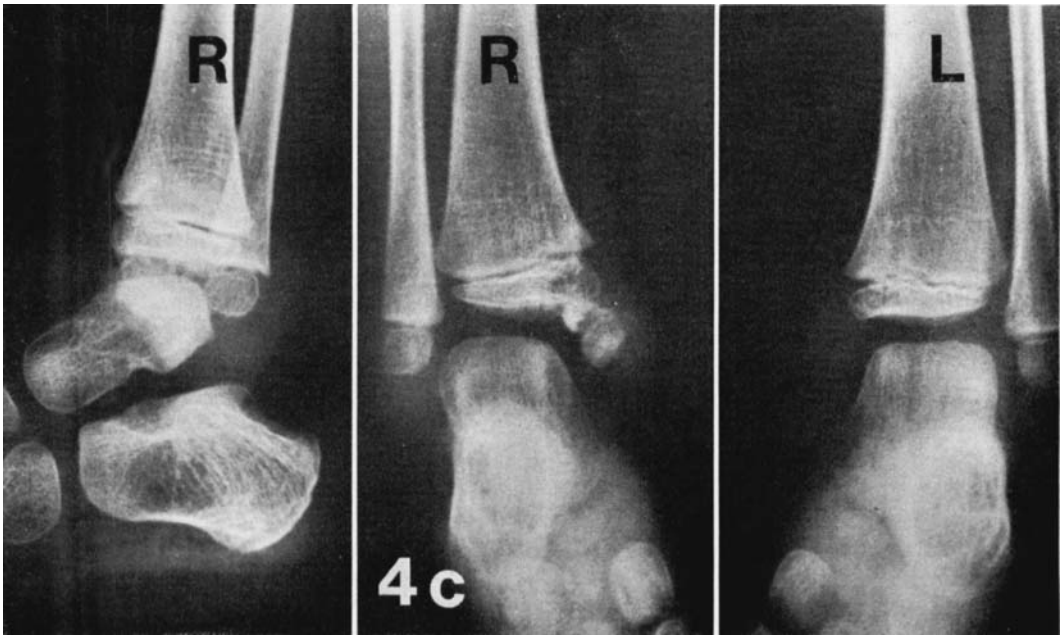

Figure 4. Case 4. a) X-rays of the knee of a boy at the age of 18 months showing enlargement and separated ossification centres in the medial femoral condyle and an exostosis of the tibial metaphysis. b) Pre-operative X-rays at the age of 5 years show a bony tumour fused with the epiphysis. c) X-rays of the ankle at the same age show eccentric ossification centres in the medial malleolus.

years later. She had no complaints. On clinical examination the knee joint was normal, and X-rays did not reveal any obvious pathological findings. However, if surgery is necessary in the knee region, the optimum time is probably before fusion between the accessory ossification centres and the rest of the epiphysis, i.e. before 4 years of age. Resection through cartilage is then possible.

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