FROM THE ORTHOPEDIC AMBULATORIUM, KOLDING
(HEAD: DR. INGEBORG LOU)

DYSPLASIA EPIPHYSIALIS HEMIMELICA

By

K. RECHNAGEL

In 1926 *Mouchet & Belot* observed a disease which they named *La tarso-mégalie.* In 1950 *David Trevor* published 8 cases of the same disease, to which he gave the name *Tarso-epiphysial Aclasis,* and in 1956 *T. J. Fairbank* reported a further 12 case histories and suggested the designation *Dysplasia epiphysialis hemimelica.*

As this name indicates, it concerns a disease which is epiphyseal and has a very characteristic localisation, for changes are only found in the lateral *or* medial side of the epiphyses of one single extremity. Very often the lower extremity is affected and a varying number of its epiphyses and tarsus bones are involved. The remaining skeleton is normal.

The changes consist of irregular bone formation from multiple ossification centres, growths on the epiphyses and as a rule enlargement of the epiphysis of tarsus bone in its entirety (tarso-mégalie). These changes are easily recognized on the X-ray. Confusion is possible with dysplasia epiphysialis multiplex, perhaps also with Morquio's disease and cretinism, but the half-side localization is so characteristic that the differential diagnosis should not cause any difficulty. In osteochrondritis dissecans the changes are found within the limit of the normal epiphysis, apart from possible loose bodies, and in aseptic bone necrosis (mb Calves-Perthes) the epiphysis is certainly irregular but diminished.

The microscopic picture is on the other hand quite uncharacteristic. "Osteochondroma" or "disturbed bone formation" is the customary diagnosis, histologically, and this gives a certain guarantee against

For permisson to publish case no. 2 and 3 I am indebted to Dr. E. Hjalmar Larsen.
Presented in shorter form to the Danish Orthopedic Association 18/5/58.

malignity which may be sometimes suspected from the roentgen changes if one does not know this rare dysplasia beforehand.

No heredity or familial disposition could be demonstrated. One of the earlier reported cases was even found in a one-egg twin whose twin brother was quite normal.

Presumably the disease must therefore be considered as an embryopathy, caused by a disturbance in the 5th fetal week. This assumption is founded on different embryological experiments, to which references are made below and which will be briefly summarised.

By means of various substances, e.g., insulin, it has been possible to provoke deformities and changes of the extremities in a chicken fetus, and the extent of these deformities is exactly dependent on the time of administration.

Moreover, it is known that the ectodermal point of the rudiments of the extremity, "the apical cap", is extremely important to the development of the whole extremity. If this "cap" is exstirpated at an early stage, in a chicken fetus this means the third day, no extremity at all grows. If there is a small delay (a few hours), it can be seen that the humerus develops normally, but that the whole of the distal part of the extremity is missing. Correspondingly exstirpation of the front of the ectodermal "cap" is followed by agenesis of the radial half of the wing, and if the rear is removed the ulnar half of the wing is lacking.

One may be allowed to conclude from these experiments then that a disturbance which solely provokes changes in either the lateral or medial part of the epiphyses of an extremity must be very restricted both in duration and extent.

It seems quite natural that an effect may strike *either* the front *or* the rear of the extremity rudiment and thus, thanks to the embryonal rotation, cause changes on the medial or lateral side respectively of a lower extremity. The nature of the effect can only be guessed at, however.

According to *David Trevor's* theory the further development of the dysplasia depends on an abnormally preserved disposition towards ossification and division in the cartilage cells. Normally these lie in an epiphysis arranged in concentric layers around the ossification nucleus and some way from this there is a zone of mitosis. From this growth zone the formed, "finished" cartilage cells are displaced towards the articular surface, as they gradually move into the characteristic columns; but in the epiphysis dysplasia some of the cells keep both the

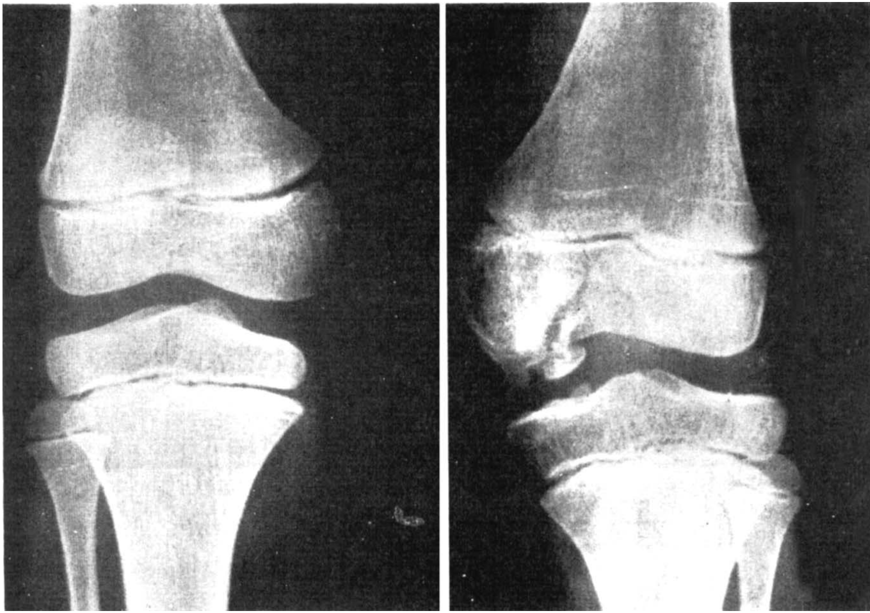


Fig. 1.
Case 1. Irregularities in the medial femur and tibial condyle.

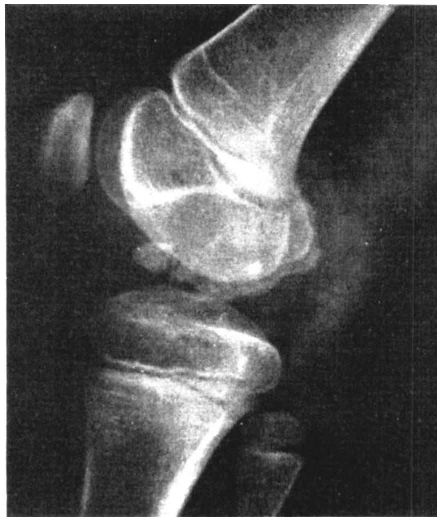


Fig. 2.
Case 1. Lateral projection.

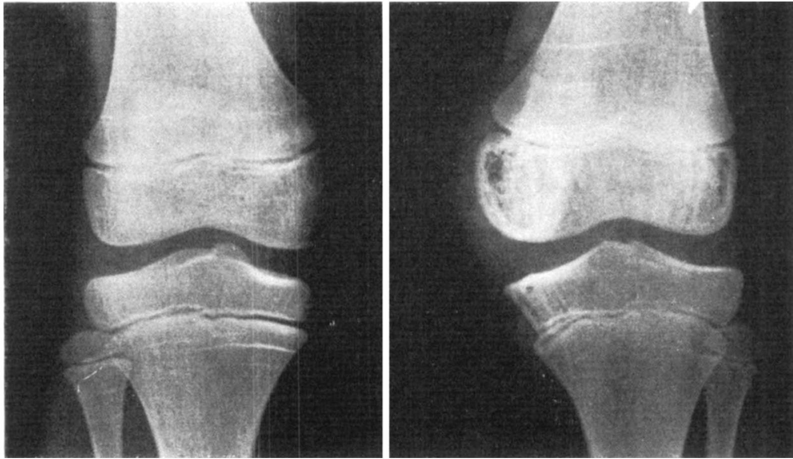


Fig. 3.
Case 1. Epiphyses smoothed after operation.

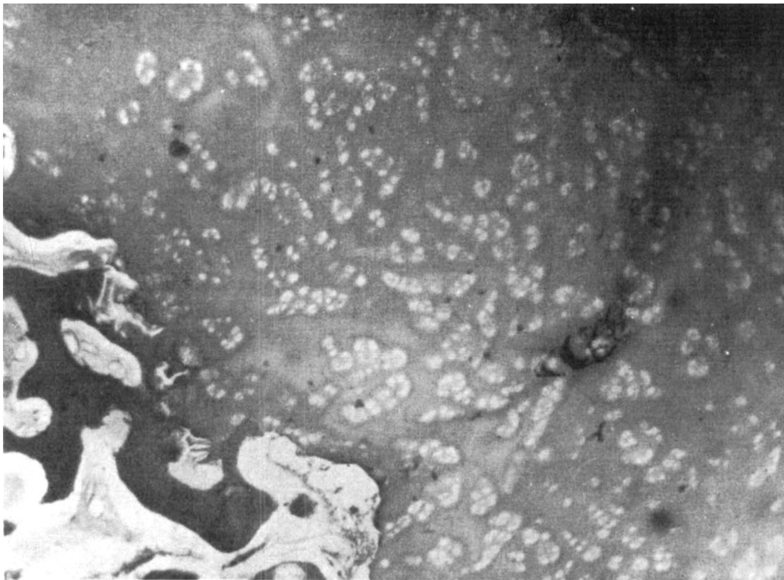
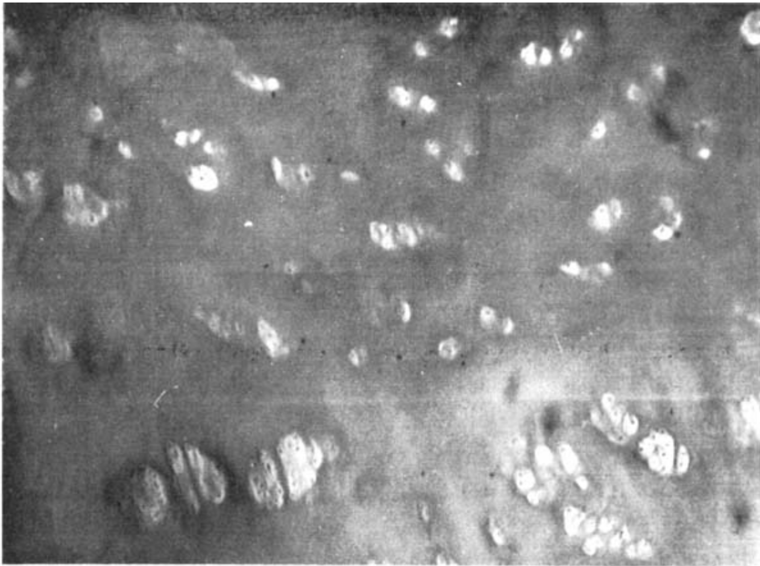


Fig. 4.



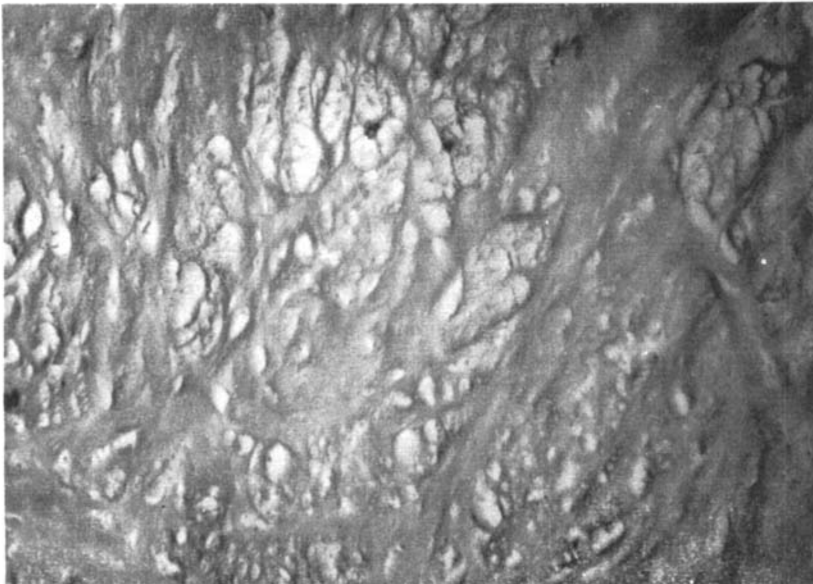


Fig. 4.

Microphotos from case 1. Basic cartilage substance of varying colour, the cartilage cells are arranged somewhat irregularly.

ability to divide and to ossify and so lead to the formation of several ossification centres and enlargement of the epiphysis.

There is no proof of the pathogenesis stated and *D'Angio et alii* point out that other possibilities also exist. In their cases from 1955 good correspondence was found between the innervation area from a part of the 5th lumbar segment and the localization of the epiphysis changes so that the ossification changes may possibly be explained as local hyperemia, nervously conditioned.

The treatment of the dysplasia is restricted to operative removal of the growths, if these lead to important deformities, limitation of mobility and other kinds. The interventions are usually very rewarding, at any rate in the first stage. In the longer view one may indeed expect early arthrosis of the affected joints; but a fundamental tendency to recurrence or malignant change has not yet been reported.

In all, 28 cases have hitherto been found in the literature. A further 3 case histories are reported below. Of this total of 31 patients, 7 are girls and 24 are boys.

Finally it must be added that where blood investigations were undertaken these continually showed normal conditions.

CASE HISTORIES

1. (OAS 22960). Boy of 9 years, who, aged 7, began to complain of tiredness in walking and who gradually limped a little on his left leg. Otherwise always healthy. His birth and the preceding pregnancy were free from complication.

At the first examination the left knee was about 170° valgus, while the right was almost the same. There was no accumulation or tenderness. The left knee could be moved 170/30 ,the right knee 180/30. Te left leg was about 1½ cm. longer than the right.

The X-ray examination showed deformation of the left medial femoral condyle projecting bone and irregular bone definition. There were similar but lesser changes in the medial tibial condyle (Figs. 1 and 2). The remaining skeleton was normal.

In arthrotomy the intumescences presented themselves as knobs of cartilage with an osseous centre. They were removed and the articular surfaces were smoothed off.

Microscopy: hyaline cartilage with a basic substance of varying colour. The cartilage cells were not arranged regularly. Bone formation seemed to proceed in an irregular manner as there were different ossification lines which did not run quite regularly. No sign of malignity. *Histological diagnosis:* interrupted bone formation. Signed K. Schourup (Fig. 4).

The post-operative course was complication-free; it was only a few months before the movements of the knee were natural. At the last check he walked without limping, moved the left knee exactly like the right, without any valgus position. The X-ray check demonstrated the very handsome shape of the condyles, and no sign of recurrence (Fig. 3).

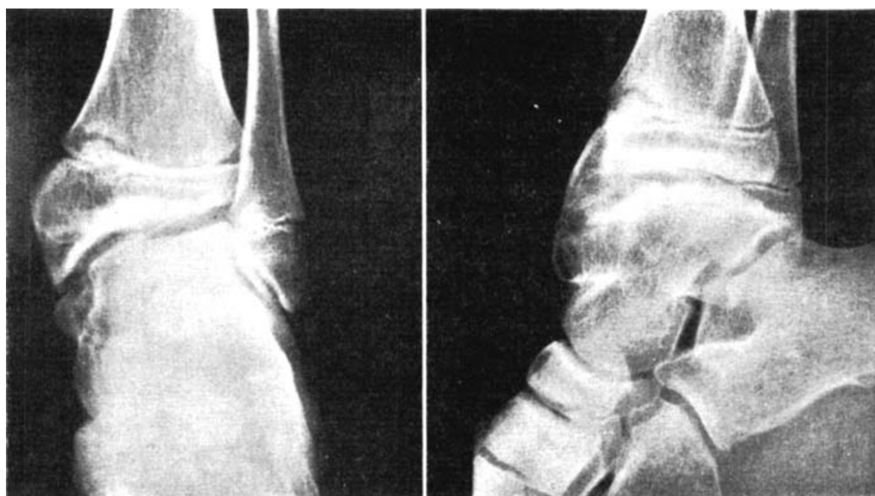


Fig. 5.
Case 2. Considerable enlargement and deformation of the talus and medial part of the tibial epiphysis.

2. *(OHK 4533/44)*. A boy aged 16. Treated with built-in support from the age of 2 for flat foot. When 13 he developed a tendency to walk a little on the toes of the left foot, since pain occurred on the medial side of the ankle joint, if he stepped on the complete planta.

The medial malleolus was enlarged and prominent. There was a plain valgus position of the left ankle joint and limited dorsal flexion (130/100), while the right ankle joint could be moved freely (135/85). There was free subtalar mobility. No pain on movement.

The X-ray examination showed medial and forward enlargement of the tibial epiphysis and a prominence on the talus, somewhat irregular structure (Fig. 5). The knee joints, hips and upper extremities were normal.

In arthrotomy "exostoses" were found covered with irregular, granulated cartilage. As far as the joint cartilage in the talo-crural joint could be surveyed this was also irregular, most closely recalling osteochrondritis. The prominent parts were chiselled away.

At a follow-up fully a year after operation he walked without awkwardness and moved the left ankle joint 150/100.

3. *(OHK 707/59)*. A boy aged 3, who was previously completely healthy. Six months ago he stumbled on the right foot and could not support himself on it for some days. Afterwards he was symptom-free but about 1 month ago the mother noticed a prominence on the right medial edge of the foot. No pain.

Objectively a firm, indolent prominence, the size of a hazelnut, was found on the medial side of the right foot in front of the talo-navicular joint, together with a slightly irregular, firm prominence behind the right medial malleolus. The basic joint of the right large toe was a little more prominent than the left. The lower

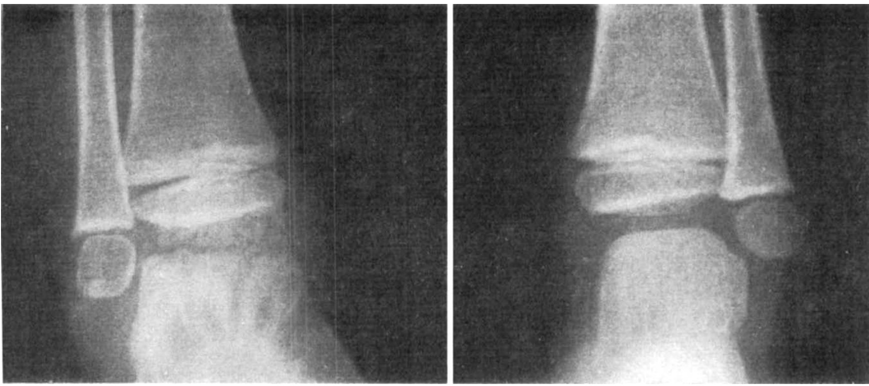


Fig. 6.
Case 3. Changes in the tibial epiphysis, talus and os naviculare.

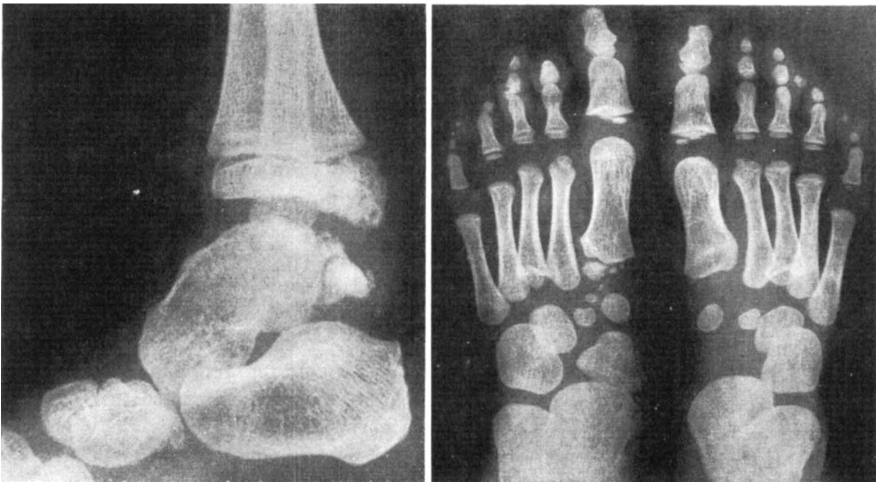


Fig. 7.
Case 3. Right os naviculare enlarged and irregular,—left still not developed.

extremities were equally long. The right ankle joint could be moved 80/140, and the foot was freely supinated.

The X-ray examination showed enlargement and irregular ossification of the right, distal tibial epiphysis, talus and os naviculare (Figs. 6 and 7). Knee joints, hip joints and the remaining skeleton were normal.

There is no grounds for therapy at the present time.

SUMMARY

A short survey of previous reports on cases of the rare disease of the epiphyses *dysplasia epiphysialis hemimelica* and reflections on its etiology; presumably it is to be regarded as an embryopathy. 3 new cases of the disease are presented.

ZUSAMMENFASSUNG

Eine kurze Übersicht von früheren Berichten über eine seltene Epiphysenerkrankung, genannt *dysplasia epiphysialis hemimelica* und Betrachtungen über die Äthiologie der Erkrankung. Wahrscheinlich handelt es sich um eine Embriopathie. 3 neue Fälle der Erkrankung werden vorgestellt.

RESUME

Un court aperçu de rapports précédents sur les cas de la rare maladie des épiphyses, *dysplasia epiphysialis hemimelica,* avec considérations sur son étiologie; il semble qu'il faille la considérer comme une embryopathie. 3 nouveaux cas de la maladie sont présentés.

REFERENCES

1. *Mouchet, A., & Belot, J.: La* Tarsomégalie. J. de Radiologie et d'Electrologie. 10, 289,1926.
2. *Streeter, G. L.:* Focal Deficiences in Fetal Tissues and their Relation to Intrauterine Amputation. Contributions to Embryology. 22. No. 126 (Publication 414 of the Carnegie Institution of Washington) 1930.
3. *Harris, H. A.:* Bone Growth in Health and Disease. London: Humphrey Milford, Oxford University Press. 143, 1933.
4. *Saunders, J. W.:* The proximo-distal Sequence of Origin of the parts of the Chick Wing and the Role of the Ectoderm. J. of Experimental Zoology. 108, 363, 1948.
5. *Trevor, David:* Tarso-epiphysial Aclasis. A Congenital Error of Epiphysial Development. J. of Bone and Joint Surgery. Vol. 32B, 204, 1950.
6. *Duraiswami, P. K.:* Experimental Causation of Congenital Skeletal Defects and its Significance in Orthopaedic Surgery. J. of Bone and Joint Surgery. Vol. 34B, 646, 1952.
7. *Donaldson, J. S., Sankey, H. H., Girdany, B. R. & Donaldson, W. F.:* Osteochrondroma of the distal femoral epiphysis. J. of Pediatrics. 43, 212, 1953.
8. *Ingelrans, P. & Lacheretz, M.:* A propos d'un cas de chrondrodystrophie epiphysaire. Rev. chir. orthop. 39, 242, 1953.

9. *Tschumi, P.:* Konkurrenzbedingte Rückbildungen der Hinterextremität von Xenopus nach Behandlung mit einem Chloraethylamin. Rev. Suisse de Zoologie. 61, 177, 1954.
10. *D'Angio, G. J., Ritvo, M., & Ulin, R.:* Clinical and Roentgen Manifestations of Tarso-epiphyseal Aclasis. Am. J. Roentgenol. 74, 1068, 1955.
11. *Caffey, J.:* Pediatric X-ray Diagnosis. Chicago. The Year Book Publishers, Inc., 3rd ed. 1956.
12. *Fairbank, T. J.:* Dysplasia epiphysialis hemimelica. J .of Bone and Joint Surgery. Vol. 38B, 237, 1956.
13. *Keats, Theodore E.:* Dysplasia epiphysialis hemimelica. Radiology. Vol. 68, 558, 1957.