FROM THE ORTHOPAEDIC CLINIC, VÄSTERÅS, SWEDEN
(HEAD: TORSTEN JERRE, M.D.)

DYSPLASIA EPIPHYSIALIS PUNCTATA

By

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Dysplasia epiphysialis punctata (D.e.p.) seems to be a rare disease and as far as the author can discover, only one case has been published in Sweden (*Jorup*, 1944). There may perhaps be good reason therefore to review briefly the disease and to present one case which the author recently had opportunity to observe.

This was a girl, born 29/3, 1959. She came to the Orthopaedic Clinic, Västerås for the first time on the 26/9, 1960. There was nothing of interest hereditarily. No malformations were known in the family. The mother had the customary childhood ailments but otherwise was completely healthy. There were no diseases during pregnancy. Neurosedyn or anything similar was not used. The delivery was normal. Since the birth the mother had noticed that the girl had a malposition of the right knee joint and also a shortening of the right lower extremity. Otherwise she seemed to be completely healthy and of normal development in all respects.

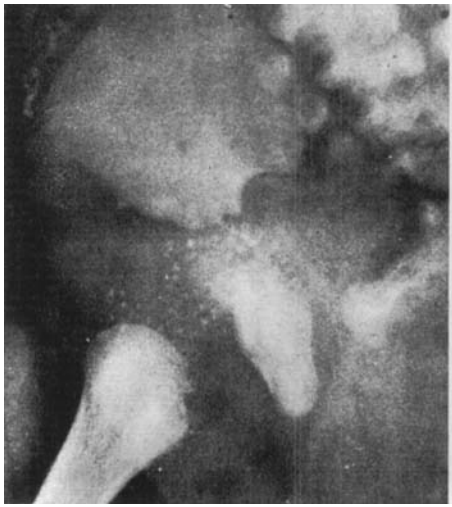
On examination at the Orthopaedic Clinic in Västerås 26/9, 1960, the patient showed at 1½ years a right-sided shortening of 4 cms., distributed equally between thigh and lower leg and also a valgus position of 20° in the right knee joint. She seemed otherwise to be healthy and normal in all respects.

The girl was earlier a patient at Pediatric Clinics and Orthopaedic Clinics at other places in Sweden.[1]

From the records and X-rays it appears that the patient already at 1 resp. 4½ months had a considerable shortening and also a valgus position in the right knee joint but seemed otherwise to be of normal development.

X-ray examination 14/4 and 29/4, 1959 (*Vänersborg*): the thorax, spinal column, skull, both upper extremities and lower left extremity were of normal condition. Within the right half of the pelvis, the right hip joint and right knee joint there were changes typical of dysplasia epiphysialis punctata (Figs. 1 and 2). The proximal

[1] Doctor *Sven Joachimsson*, head of the Pediatric Clinic, Vänersborg, Doctor *Sven Thunström*, head of the Radiological Clinic, Vänersborg, and Doctor *Folke Ståhl*, head of the Orthopaedic Clinic, Borås, were kind enough to put clinical data and X-rays at my disposal. I should like to thank them most sincerely for their help.



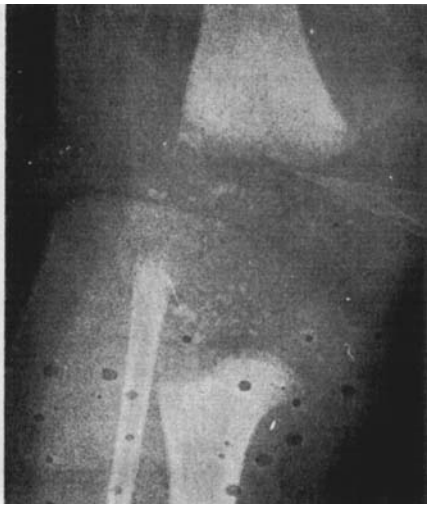


Fig. 1. *Fig. 2.*

Fig. 1. X-ray 14/4, 1959. Within the area of the right iliac crest and in the right hip joint region a large number of calcifying marks can be seen. The bone nucleus is not yet visible.

Fig. 2. X-ray 14/4, 1959. In the right knee joint there are the same calcifying marks as in the right hip joint. The bone nuclei are not yet visible. The fibula projects proximally about 1 cm. past the tibia.
(The many small light patches in the lower third of the picture are due to film defects.)

epiphyses of the femur were not yet visible bilaterally. The distal epiphyses of the femur and the proximal epiphyses of the tibia were of normal size for this age on the left side, but on the right side had not yet appeared.

X-ray examination 12/8 and 9/9, 1959 (*Borås*): the thorax, both upper extremities and the left lower extremity were normal. The small stippling marks within the right iliac crest area and within the region of the right hip joint had diminished considerably in number. Within the femur's distal and within the tibia's proximal epiphysis areas there was a status quo. Within the right ankele joint (not previously X-rayed) there were the same changes as within the hip and knee joint. All the epiphyses in the lower left extremity were normally developed for the age, on the right side they were not visible as yet, either in the hip, knee or ankle joint.

X-ray examination 26/9, 1960: the thorax, spinal column, skull and left lower extremity were normal. Within the lower right extremity there were changes still typical of dysplasia epiphysialis punctata (Figs. 3 and 4).

This disease was described for the first time by *Conradi* in 1914 under the name of chondrodystrophia foetalis hypoplastica. Later it was reported under various headings such as epiphysical dysplasia puncticularis, chondrodystrophia calcificans congenita, chondroangio-

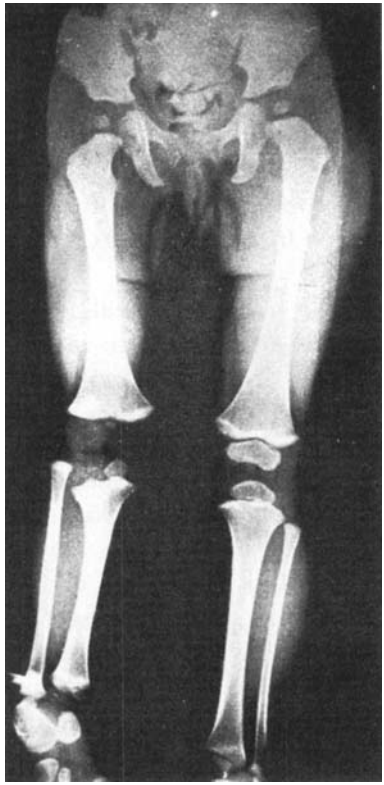


Fig. 3.
X-ray 26/9, 1961. The right lower extremity is underdeveloped with a shortening of the femoral and tibial diaphyses of 2 cms. each. The fibula the shortening of which scarcely reaches 1 cm., is abnormally long in relation to the tibia, above all proximally. The marks observed earlier in the area of the right iliac crest have now completely disappeared. The marks in the right hip joint region have further considerably diminished both in number and in calcification density, but still appear plainly on the original film. All the bone nuclei also appear now on the right side but are much lesser in size than on the left side. The difference in this respect was most pronounced between the distal femoral epiphyses. There was a moderate valgus position in the right knee joint.

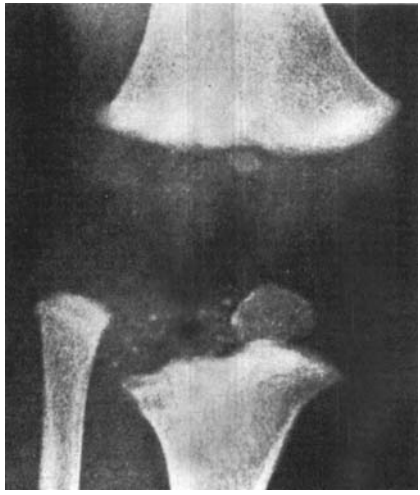


Fig. 4.
X-ray 26/9, 1961. In the femur's distal epiphysis area the marks have now diminished both in number, calcification density and size. In the upper epiphysis area of the tibia the status is unchanged.

pathia calcarea seu punctata, stippled epiphyses, dysplasia epiphysialis punctata.

The number of earlier published cases was according to *Fairbank* (1949) 16, according to *Mosekilde* (1952) 39, and according to *Weil* (1957) fully 50. *Mörch* (1944) estimates the incidence in Denmark to be 1 in 500,000 newborns or 1 case every 7 years. According to the majority of authors the disease is considerably more common in girls than in boys. Prevalence in a family was reported by *Maitland* (1939), *Raap* (1943) and *Vinke & Duffy* (1947).

The diagnosis is made radiologically and at least in typical, early cases the changes are so characteristic that no differential-diagnostic deliberations are necessary. The X-rays show a varying number of small calcifying marks of unequal size. These appear as a rule much before the time when ossification of the epiphysis in question normally takes place (*Brogden & Crow*, 1958), indeed, their presence has even been confirmed prenatally in the foetus (*Frank & Denny*, 1954). In those cases of D.e.p. observed over a fairly long period of time, it was proved that the stippling again disappeared within the course of the first years of life either in such a way that they were gradually resorbed or also in such a way that they were incorporated in the normal ossification centre (*Selakovich & White*, 1955).

At the same time as the stippling of the epiphyses it is usual radiologically to be able to observe a retardation in the development of the bone nuclei while growth disturbances in the affected extremities are the rule in D.e.p.

The epiphyses to which the disease is most commonly localised are according to *Fairbank* (1949) the upper and lower ends of the femur, upper end of the tibia and upper end of the humerus. It also occurs in the lower end of the tibia, both ends of the fibula, the lower ends of the radius and ulna, the bones of the hand and foot, the acetabulum, ischium and sacrum, the patella, the ribs, the sternum, the hyoid, the intervertebral discs and vertebrae and even in the synovial membrane of different joints (*Fairbank*, 1949, *Mosekilde*, 1952, *Swoboda*, 1956 and *Weil*, 1957). The disease may be localised to one part of the body only but within the same patient may also be distributed over a large number of the above localisations.

Various cases of D.e.p. have been examined pathologo-anatomically and histologically.

Conradi (1914) studied in detail and described the pathologo-anatomical and histological findings in a girl, who died at the age of 1 month.

From the histological research in this case he drew the conclusion that the radiologically demonstrated calcifications represent bone nuclei appearing too early.

Harris (1933) reported patchy mucoid degeneration and cystic spaces in the cartilaginous epiphyses, particularly near the articular surfaces. In some places the areas of degeneration were invaded by blood vessels, and a core of fibrous tissue had formed. In the vertebral bodies which ossified from two centres, there was failure of the usual orientation of cartilage cells, and of normal calcification and ossification. *Harris* insists that the fundamental error is similar to that which he found in achondroplasia.

Burckhardt (1938) examined the histologic picture after test excision on a child 6 months old with radiologically typical dysplasia epiphysialis punctata. He considers the fundamental point to be morbid changes in the cartilage, a softening of the cartilage or cartilage malacia. This author regards the calcified layers characteristic of the disease as a sign of a healing process within necrotic parts of the cartilage.

Hässler & Schallock (1940) described the pathologo-anatomical findings and the histological pictures in great detail. Like earlier authors *Hässler & Schallock* were also able to observe both a great number of calcification foci in separate epiphysis cartilage and also necrotic processes here. These authors consider that it is not a question of a primary necrosis with secondary calcification but instead that it concerns a primary calcification with secondary necrosis. According to these authors the cause of the primary calcifications would be some form of disturbance in the calcium metabolism.

Lund (1942) found that the muscles of the limbs were composed mainly of tough white fibrous tissue and were very short. This was the cause, according to him, of the frequently occurring joint contractures.

Karlén & Cameron (1957) described as the important histological abnormality the presence, throughout the tissue of the epiphyseal cartilages studied, of abnormal circumscribed areas reaching one or two millimetres in size. These areas contrasted with the surrounding cartilage in staining darkly and irregularly, and they were presumed to indicate sites of calcification. Some of them showed proliferation of cartilage cells, whereas others showed advanced degeneration of the tissue with complete loss of cells.

As possible etiologies individual authors have put forward lues, A- and D-hypervitaminosis, hypothyroidism and disturbances in the cal-

cium metabolism. Later authors were not able to confirm these hypotheses, however, and the etiology of D.e.p. must still be regarded as completely unknown.

Very often D.e.p. is combined with malformations and morbid changes within other organic systems, such as multiple joint contractures not solely within those regions which are sites for D.e.p., congenital cataract, dyskeratosis, hip joint dislocation, foot deformities, heart complaints, cleft palate, saddle nose, general weakness and mental disturbances.

Owing to the simultaneous malformations and diseases a large number, according to *Fairbank* (1949) half the number of the published cases, were born died or died at a very young age. *Mosekilde* (1952) states in this connection that 16 out of 42 known cases died, 15 during the first year of life and 1 aged 3½. The cause of death in 5 cases was pneumonia, in 5 cases other infectious diseases, in 4 cases heart complaints and in 2 cases it was unknown.

The author has now followed-up his case of D.e.p. for 1½ years with quite frequent clinical and radiological checks and the patient recently at the age of 3 was given a renewed, painstaking examination. The mother states that the girl has always appeared completely normal in all respects with the exception of the present deformity and the shortening of the lower right extremity.

At objective examination the patient still displayed a shortening of the lower right extremity of 4 cms., equally distributed between the thigh and lower leg, and also a valgus position in the right knee joint of 20°. The mobility in the hip, knee and ankle joints is normal. Otherwise the girl has a quite normal physique and seems happy, awake and normally developed for her age both psychologically and somatically. Length 90 cms. Weight 14 kgs. (Fig. 5).

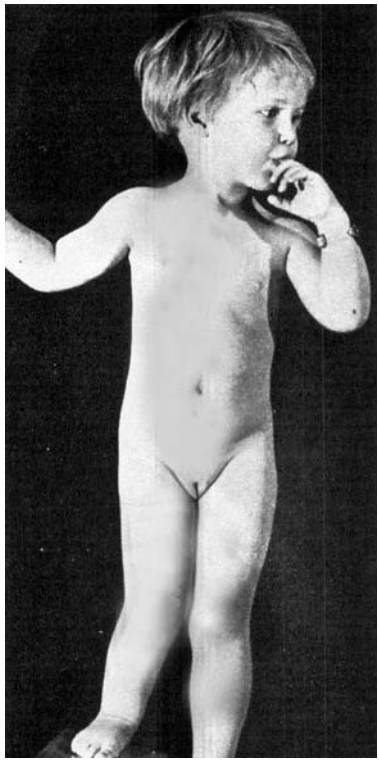
X-ray examination 5/4 and 6/4, 1962: the left lower and both the upper extremities were normal. There was still 2 cms. shortening of both right femur and tibia. The previously described stippling marks no longer appear within the proximal epiphysis area of the femur but still are visible within the femur's distal and the tibia's proximal and distal epiphysis areas (Figs. 6 and 7). The femur's proximal bone nucleus is now of normal size. There is still a valgus position in the right knee joint of 20°.

During April, 1962, the girl was submitted to specialist examinations by ophthalmologist, otolarryngologist, pediatricians and child psychiatrists, but no malformations or morbid changes could be discerned in the respective organic systems.

Laboratory tests: SR 5 mms.
Hb 84. Red 4.82.
White 7100 $<^{3,800\ p}_{3,300\ m}$

Calcium in serum	10.2 mg. %
Phosphorus in the serum	5.5 mg. %
Calcium in the urine	0.1 gm.
PBI	5.9 gamma per cent.

Fig. 5.
The girl aged 3.

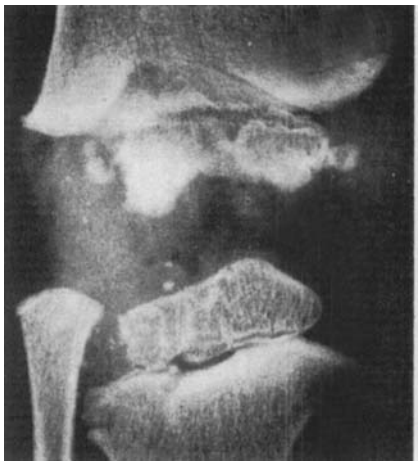


The continuning X-ray checks showed that the changes typical of the disease gradually diminished. Regression began in the regions most proximally situated and since then were observed to take a peripheral direction with increasing age. Even at 4½ months they had thus become less marked within the iliac crest area and hip joint region. At 1½ years they had completely disappeared in the area of the iliac crest, had further diminished within the hip joint region and also begun to recede in the distal epiphysis area of the femur. At 3 years the changes were still quite absent in the area of the iliac crest and had now also completely disappeared in the hip joint region. In the distal epiphysis area of the femur they had further receded and now had also begun to be less marked in the proximal epiphysis areas of the tibia, but remained without change in the distal epiphysis area of the tibia.

That the stippling marks typical of D.e.p. usually recede and disappear with increasing age is a fact known and pointed out by several authors. Whether this regression normally occurs in a certain order is not stated in previously published cases nor has it been discussed in earlier literature. I wished to point out the course of the regression in my case, naturally without drawing any conclusions whether this course would be generally applicable.

Taking into consideration the available examination results one may find every justification in this case in expecting a good prognosis for life in the future. As the valgus position and shortening has not shown any tendency to increase during 1½ years of observation, one may well continue to dare to hope for a really stationary condition.

Until now the valgus position in the knee joint has been treated with a non-articulated knee cap. At a later date prevalent orthopaedic therapy



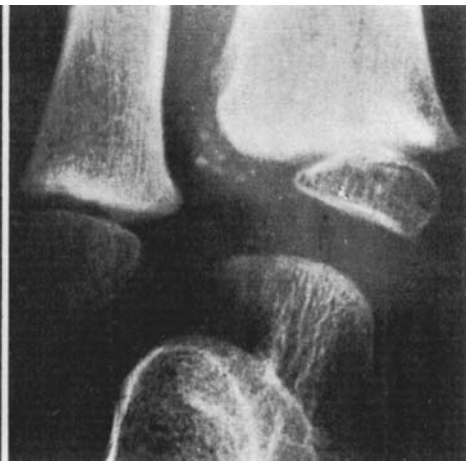


Fig. 6. *Fig. 7.*

Fig. 6. X-ray 6/4, 1962. The previously described calcifying marks still appear in in the femoral and tibial epiphyses, on the original films, however, in both places they are lesser in number than before. The distal bone nucleus of the femur and the proximal bone nucleus of the tibia have grown considerably but are still lesser in size than on the left side. The fibula projects still about 1 cm. past the tibia.

Fig. 7. X-ray 6/4, 1962. The calcifying marks still emerge above all laterally in the epiphysis area in approx. the same number, calcification density and size as before. The distal bone nucleus of the tibia has grown considerably but is still lesser in size than on the left side.

will in all probability be required in the form of epiphyseodesis and/or corrective osteotomy, shortening osteotomy and/or lengthening osteotomy.

SUMMARY

The author describes a case of dysplasia epiphysialis punctata and gives in connection with this an account of this rare disease.

RESUME

L'auteur décrit un cas de dysplasie épiphysialis punctata et donne à cette occasion un compte rendu de cette maladie rare.

ZUSAMMENFASSUNG

Der Verfasser beschreibt einen Fall von Dysplasia epiphysialis punctata und bespricht im Zusammenhang damit diese seltene Erkrankung.

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