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Dysplasia epiphysealis multiplex — Problems in Diagnosis and Treatment of the Hip

Dysplasia epiphysealis multiplex — problemy diagnostyczne i lecznicze dotyczące biodra

INTRODUCTION

Dysplasia epiphysealis multiplex (DEM) is a generalized affection of the skeletal system (1, 2, 4) involving the hips, knees, feet, shoulders, elbows and hands as well as the spine. It presents complex diagnostic problems, especially in babies and small children, in whom the symptoms are not dinstinctly developed. From the clinical point of view, DEM has to be distinguished from Perthes' disease.

TERMINOLOGY OF THE DISEASE, PATHOGENESIS

Dysplasia epiphysealis multiplex was first described by Barrington-Ward in 1912 (cit. by 5). The term *dysplasia epiphysealis multiplex,* however, comes from Fairbank (cit. by 5), who described the first case of this disease in 1935 and further 20 cases in 1947. In 1937, Ribbing (cit. by 5) described abnormal development of the epiphysis in 7 members of a family.

DEM is usually inherited as a dominant trait but recessive forms have been also reported. The single cases represent most likely new mutations.

In ca 50% cases, the disease occurs within a family (3, 5). Hurley and Asling (cit. by 5) claim that a disorder of manganese metabolism is the cause of *dysplasia epiphysealis multiplex.*

CLINICAL MATERIAL

In the years 1970—1990 at the Lublin Orthopaedic University Children's Clinic, 36 children (28 boys and 8 girls) were treated for DEM (0.36% of total number of 8700 patients). Most of them reported difficulty of their hands, only a few complained of knee and foot pain and all had problems with the hips.

In 14 children, the symptoms were previously attributed to Perthes' disease, in 7 to *coxitis fugax* and in one to rheumatoid inflammation of the hip. In the remaining 14 cases, DEM was the primary diagnosis. Perthes' disease is ten times more frequent than DEM. Our patients' age ranged from 3 to 16 years (average 9.5). In 26 cases, the height of patients was insufficient and in 10 it was excessive, 19 children were underweight, 5 were overweight and 12 remained within normal limits.

CLINICAL AND RADIOLOGICAL SYMPTOMS CONCERNING HIPS

Clinical symptoms

Clinical symptoms may appear at different ages. They seem to be connected with overburdening of the limbs and depend on the intensity of the disease.

Among our patients, 3 groups were distinguished according to age and intensity of the disease:

group I — small children (3—5 years old) reported early;

group II — older children (6—9 years old) reported late;

group III — children of 10—16 years old reported at the stage of fixed hip contraction.

The group I (15 children) reported mild symptoms such as: pain, limping, limb fatigue and temporal limitation of hip movement.

Patients in the group II (13 children) had advanced clinical and radiological changes. They had not been treated till their admission to the Clinic or had been treated for Perthes' disease. Case histories revealed limping or waddling gait, general weakness, easy fatigue and muscular atrophy in buttocks and thighs. Children complained of pain in the groin and upper parts of the thigh. Some suffered night pain in the hip or in the whole lower limb. Examination revealed limitation of hip movement.

To the group III (8 children) belonged patients admitted to the Clinic with significantly limited hip movement, limping and decreased functional ability. Apart from these involving the hip, other signs were also found: scoliosis, lumbar-hyper-lordosis, limited spinal flexion, flexed knee contraction, crooked knee, even habitual patellar subluxation, shortened and enlarged phalanges in the feet and hands. Some children had limited movement of the shoulders and elbows.

X-ray findings

In young children — group I (Figs. 1, 1a, 1b, 1c) 3—5 years old, slight changes in the head of the femur and in its ossification were observed. The head has oval shape, becoming flattened in a multisegmented contour. The observed changes were similar to these characteristic of Perthes' disease but, in contradistinction to Perthes' disease, were not actively progressive.

In older children — group II (Figs. 2, 2a, 2b, 2c, 2d) 6—9 years old, who had been improperly treated, changes in the head of the femur resembled the fragmentation stage in Perthes' disease.

Identical permanent radiological features, typical of Perthes' disease, existed for a long time: months and even years.

In children (group III) 10—16 years of age (Figs. 3, 3a, 3b) we observed deformed hips with an increased CCD angle of the neck, small heads showing signs of chronic necrosis, dysplastic condyles in the tibiae and femur, with patellar fragmentation and disorders in the development of the metatarsals, metacarpals and the phalanges of the hands.

TREATMENT

Children in whom DEM was detected at an early stage of clinical and radiological change, were treated by means of burden loosening, termo-, hyro-, kinesi-, and balneotherapy. Similar treatment was administered to children with advanced clinical and radiological hip signs but results were worse. All children were treated in the Sanatorium Department of the Lublin Orthopaedic University Children's Clinic.

Conclusions

1. In children with the symptoms characteristic of Perthes' disease, cases of *dysplasia epiphysealis multiplex* can occur.
2. From the clinical point of view, it is important to distinguish DEM from Perthes' disease. In all patients with bilateral Perthes' disease DEM should be excluded.
3. In DEM, abnormalities of the shoulders, elbows, hands, knees, feet and spine and characteristic slow evolution of changes within the hip can be observed.
4. DEM, affecting the hip, is characterized by discrepancy between the advanced radiological and less marked clinical changes within the hip can be observed.
5. Contractions of the hips and other joints develop with time and are *signum male omnis* for patients, their locomotor and life functions.
6. Clinical and radiological examination of children's parents and other children in the same families revealed changes (sometimes subclinical) in 50% members.
7. Prophylaxis of lower limbs, especially protection of hips against necrotic and deformative changes, is particularly important.

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STRESZCZENIE

Dysplasia epiphysealis multiplex (DEM) jest uogólnioną chorobą układu kostnego. Z klinicznego punktu widzenia DEM wymaga różnicowania z chorobą Perthesa. W latach 1970—1990 w Klinice Ortopedii Dziecięcej w Lublinie leczono 36 dzieci (28 chłopców i 8 dziewczynek) z powodu DEM. Stanowi to 0,36% z ogólnej liczby 8700 leczonych pacjentów. Wiek naszych chorych wynosił 3—16 lat (średnia 9,5 lat).

Pod względem radiologicznym pacjentów zaszeregowano do 3 grup. Grupa I to małe dzieci w wieku 3—5 lat z wczesnymi objawami bolesności i ograniczeń ruchów bioder. Grupa II to dzieci 6—9-letnie z objawami klinicznymi zgłaszanymi dość późno. Grupa III to dzieci 10—16-letnie zgłaszane do Kliniki w fazie ciężkich przykurczów bioder i dużej niewydolności statyczno-lokomocyjnej kończyn dolnych.

Omówiono leczenie fizykalne i postępowanie profilaktyczne, które ma chronić biodra przed zmianami, zniekształceniami i przykurczami.

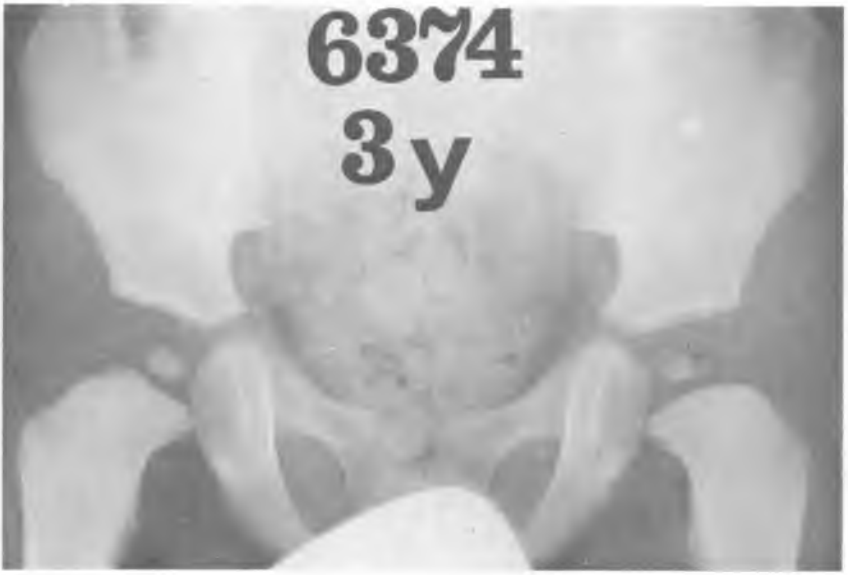

Fig. 1. Tomasz P., No. 6374, 3 years old. Retarded development of the femoral heads. Good movement of the hips

Fig. 1a. 7-years-old patient. Fragmented heads, as in Perthes' disease. Limited movement of the hips

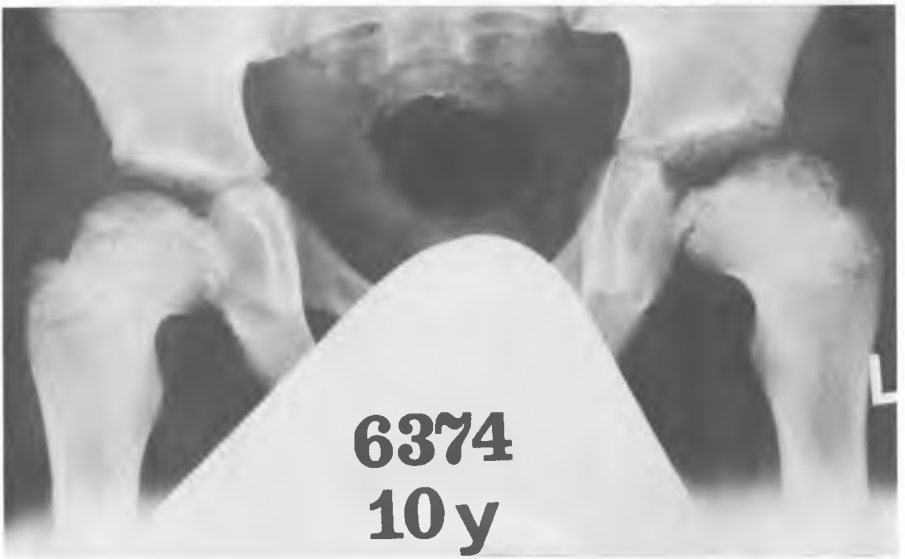

Fig. 1b. 10-years-old patient. Systematically rehabilitated, training in swimming pool

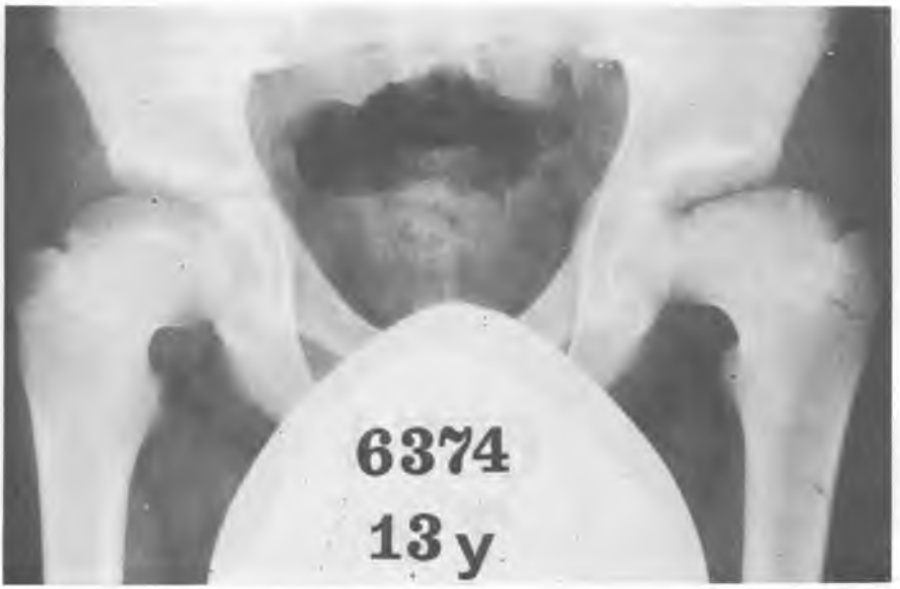

Fig 1c. 13-years-old patient. *Coxa vara bilateralis*. Sufficiently developed femoral heads. Good movement of both hips

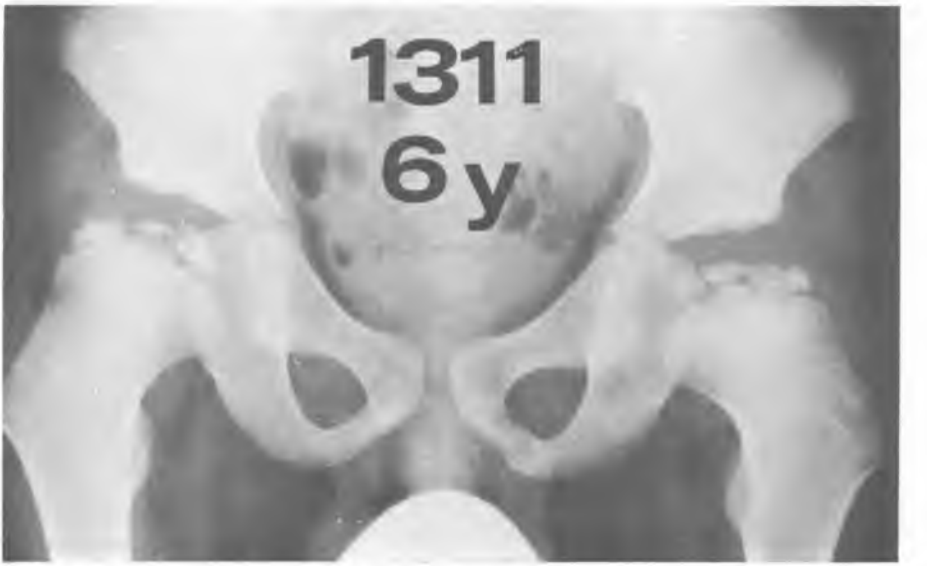

Fig. 2. Piotr K., No, 1311, 6-years-old patient. Fragmentation of femoral heads, widening of femoral neck. Almost full movement of the hips. Initially diagnosed as Perthes' disease

Fig. 2a. 7 years of age, beginning of remodelling of both heads and thier lateralization

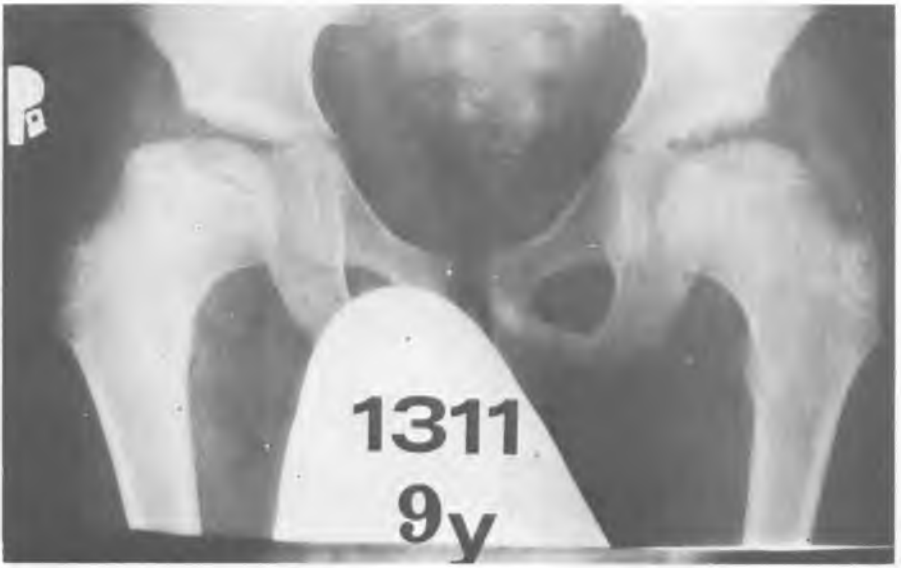

Fig. 2b. 9-years-old patient, further remodelling of the heads. Full movement of the hips

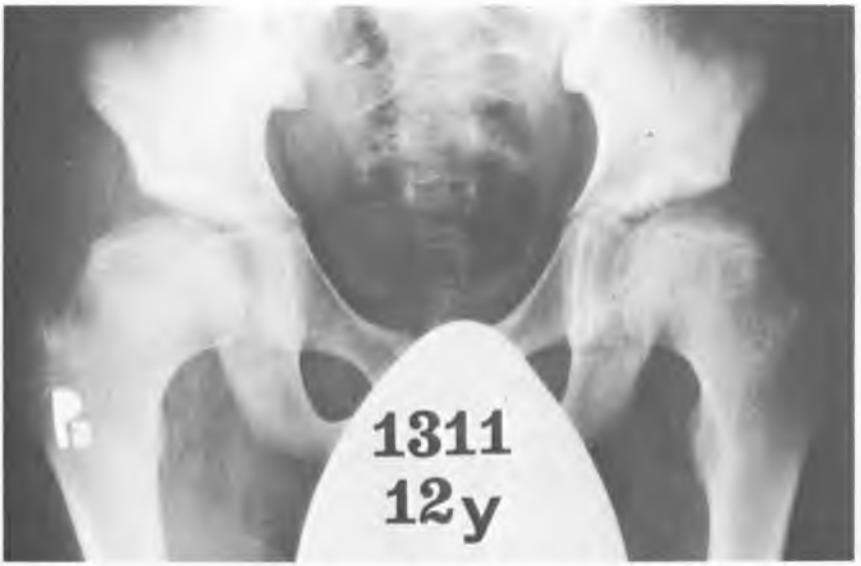

Fig. 2c. 12-years-old patient. Oval shaped of femoral heads. Good movement. Patient still training by swimming

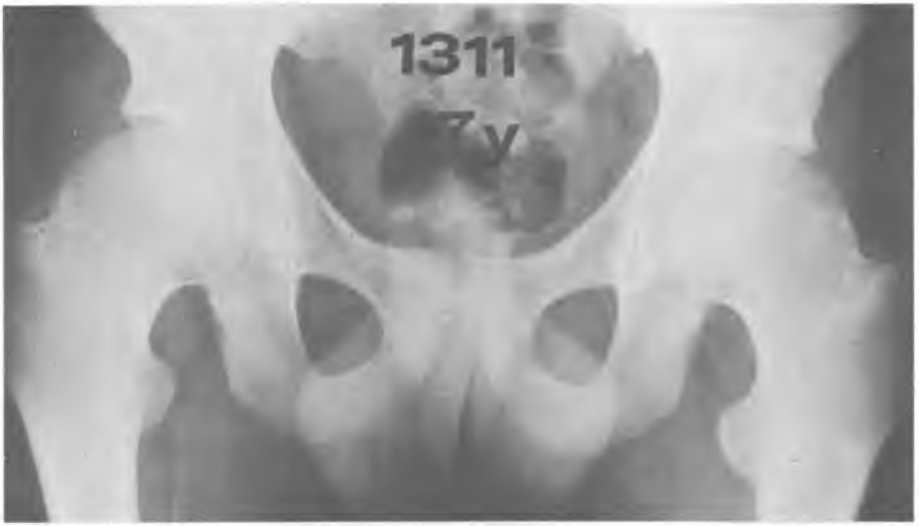

Fig. 2d. 17-years-old patient. Dysplasic hips. Good movement

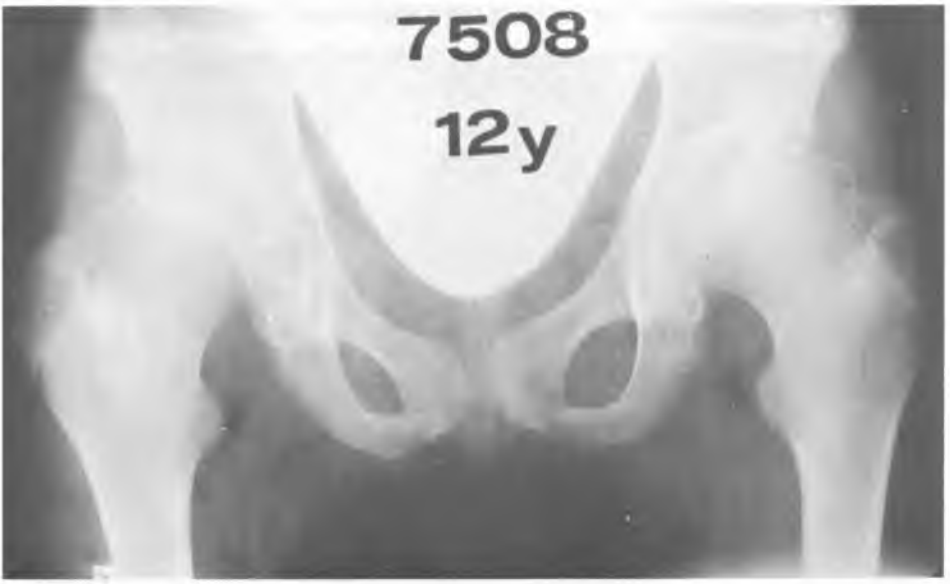

Fig. 3. Violetta D., No. 7508, 12-years-old patient. Heads atrophic and distinctly lowered, neck short, *coxa valga*. Pain in knees, hips, easy fatigue. Limitation of movement of the hips

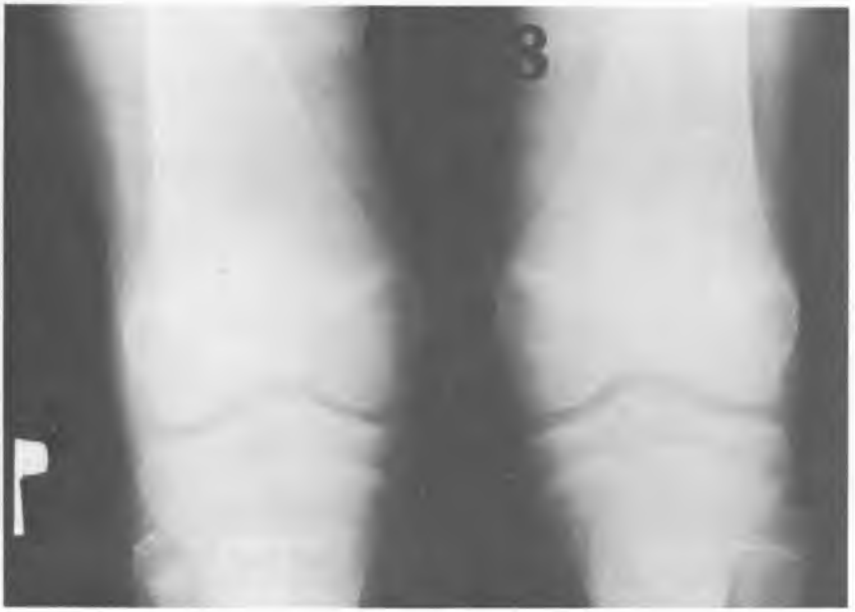

Fig. 3a. Proximal epiphyses of tibia and distal epiphyses of femur extremely shallow

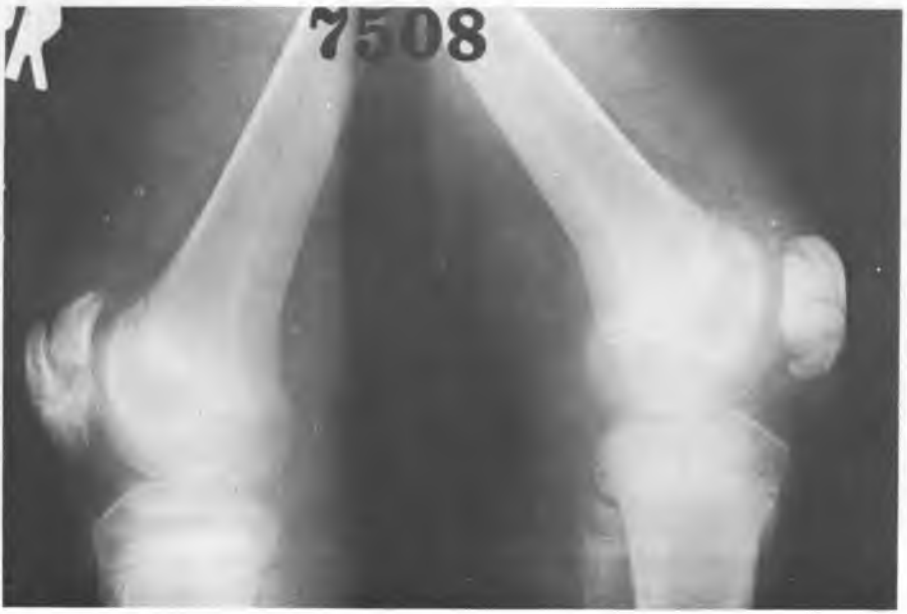

Fig. 3b. Fragmentation of patella