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DYSPLASIA EPIPHYSEALIS MULTIPLEX

REPORT OF A CASE IN A BANTU CHILD

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The above name was first introduced by Fairbank[1] in 1935, when he differentiated the condition from dysplasia epiphysealis punctata. However, Jansen[2] in 1934, reported under the name of epiphyseal dysostosis a case which Fairbank considered to be one of dysplasia epiphysealis multiplex. By 1947, 26 cases had been collected by Fairbank. From time to time in various journals other cases have been reported but the total number of cases recorded is extremely small. In the last 10 years no case has been recorded in the *South African Medical Journal* and we believe the following case report to be the first in a Bantu.

The etiology of dysplasia epiphysealis multiplex is entirely unknown. It affects both sexes, males more frequently than females. The age at which the developmental error was recognized in Fairbank's cases varied from 18 months to 14 years, with two exceptions, a girl of 20 years and a middle-aged woman. Hereditary and familial influences are not as a rule in evidence. However, two sisters were described by Gardner Hill (1937), a mother and her twin boys by Resnick[3] (1943), sisters and twins in Fairbank's series; and 3 sisters are reported by Waugh[4] (1952). Jackson *et al.*[5] (1954) trace a family tree of one of their cases which indicates a simple dominant genetic inheritance; however, they are doubtful whether this law holds in general for the condition, since most cases reported have been sporadic.

The radiological features are characterized by:

(*a*) Irregularity in contour and in bony structural pattern of the developing epiphyses.

(*b*) Multicentric epiphyseal ossification.

(*c*) Late appearance, slow development and delayed fusion of the epiphyses.

(*d*) Varying irregularity of adjacent metaphyses.

Epiphyses commonly affected are those of the hips, knees, ankles, shoulders, elbows, the metatarsal and metacarpal bones, the carpal and tarsal bones, and the phalanges of both hands and feet. The vertebrae, skull, teeth and acetabuli are singularly free of abnormality. While the metaphyseal ends of the bones adjacent to the affected epiphyses may also exhibit irregularity, the shafts of the long bones are normal in shape and structure. The lesions are characteristically bilateral, and usually symmetrical.

Information about the histological pathology has been scanty indeed, and Fairbank states that he knows of only one case in which it was investigated, when mucoid degeneration of the cartilage was found irregularly placed among scanty bone-cells.

The blood report is usually normal.

CASE REPORT

Patient M, a Bantu male child aged 10 years, complained of pain over the anterior aspect of his left hip joint. The pain was dull in nature, did not radiate, and was worse on walking and playing. This pain was related to a fall.

The patient is the 8th of 9 siblings. He was a full-term infant and was breast-fed until the age of 2 years. Teething started at the age of 6 months, crawling at 7 months and walking at 10 months. He has not had any childhood illnesses. He was born in the Cape Province, near Queenstown, but has lived in Johannes-

burg since the age of about 4 months. He attends school, and is above average intelligence, being in grade II and always coming first in class, and participates in football.

Family history: the mother, father and all the children are well.

Examination. A bright, intelligent Bantu male child presented, slightly dwarfed but no obvious deformity in spine, limbs or gait was noticed except for some broadening and slight adduction of both fore-feet. The fingers were short and stubby but relatively proportional, and hyper-extension was possible at the proximal inter-phalangeal joints. The ligaments of the other joints of the body were normal.

The patient when first seen in the out-patient department had some limitation of movement at the left hip-joint and both hip-joints were therefore X-rayed. When he was subsequently re-examined in the ward about 1 week later, no limitation of movement was found. The shape of the bones, clinically, was normal, with no widening at the metaphyses or tenderness on palpation.

Measurements. (1) Height of patient 50½ inches. (2) Arm span 53 inches. (3) Upper segment (top of head to pubis) 24½ inches. (4) Lower segment (pubis to ground) 26 inches. (5) Circumference of head 21¾ inches. (6) Left femur 13¼ inches; right femur 14 inches. (7) Left tibia 12½ inches; right tibia 12¾ inches. (8) Left humerus 9¼ inches; right humerus 9 inches. (9) Left radius 8⅛ inches; right radius 8 inches.

Blood Investigation. (1) White blood-cells 8,200 per c.mm. (2) Haemoglobin 14·3 g. (3) ESR 12 mm. in 1 hour. (4) Calcium 5·2 mEq/litre. (5) Phosphorus 4·0 mg./100 c.c. (6) Alkaline phosphatase 31 units. (7) Urea 17 mg./100 c.c. (8) Potassium 5·1 mEq/litre. (9) Sodium 14 mEq/litre. (10) Chlorides (as Na Cl) 540 mg./100 c.c. (11) CO_2 combining power 46 m 1/100 c.c.

Urinary Analyses. (1) Microscopic—nothing abnormal detected. (2) Chemical—nothing abnormal detected. (3) The total volume of urine for 24 hours was 2,570 c.c. and the content of 17-Ketosteroids (estimated as dehydro-iso-androsterone) 4·5 mg.

It should be noted that only the alkaline phosphatase was raised in the above estimations.

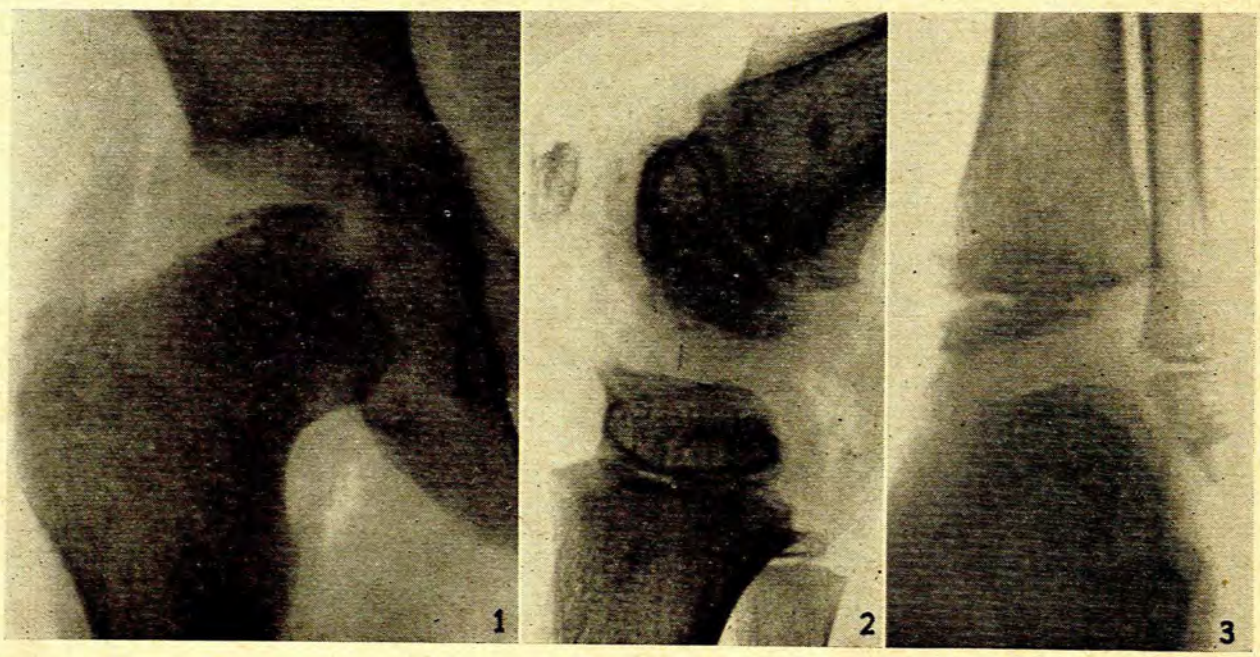
Histological Report. Section of this specimen from the lower epiphysis of the right fibula, shows the presence of atypical hyaline cartilage, in which the cells are irregular and scattered and have little pattern of orientation. Small fragments of osseous tissue are present within the cartilage, but there is little evidence of endochondral ossification. The margins of the cartilage are lined by flattened cells, lying adjacent to loose fibrous connective tissue, containing blood vessels.

Radiological Report

The findings on radiological examintation of the skeleton were as follows:

Hip Joints (*Fig.* 1). The right capital epiphysis shows marked flattening, increased density, fragmentation and several ossific centres. On the left side the capital epiphysis is somewhat larger, slightly flattened, and irregular in outline. Medically a few small ossific centres are shown. The adjacent metaphyseal outline on both sides is irregular and both femoral necks are widened. The epiphyses of the greater trochanters show slight irregularity in outline. The acetabuli are normal.

Knee Joints (Fig. 2). The epiphyses at the lower ends of the femora are large, flattened and markedly irregular, and show some spiky projections. Several small additional centres are present. The tibial epiphyses



Figs. 1, 2 and 3

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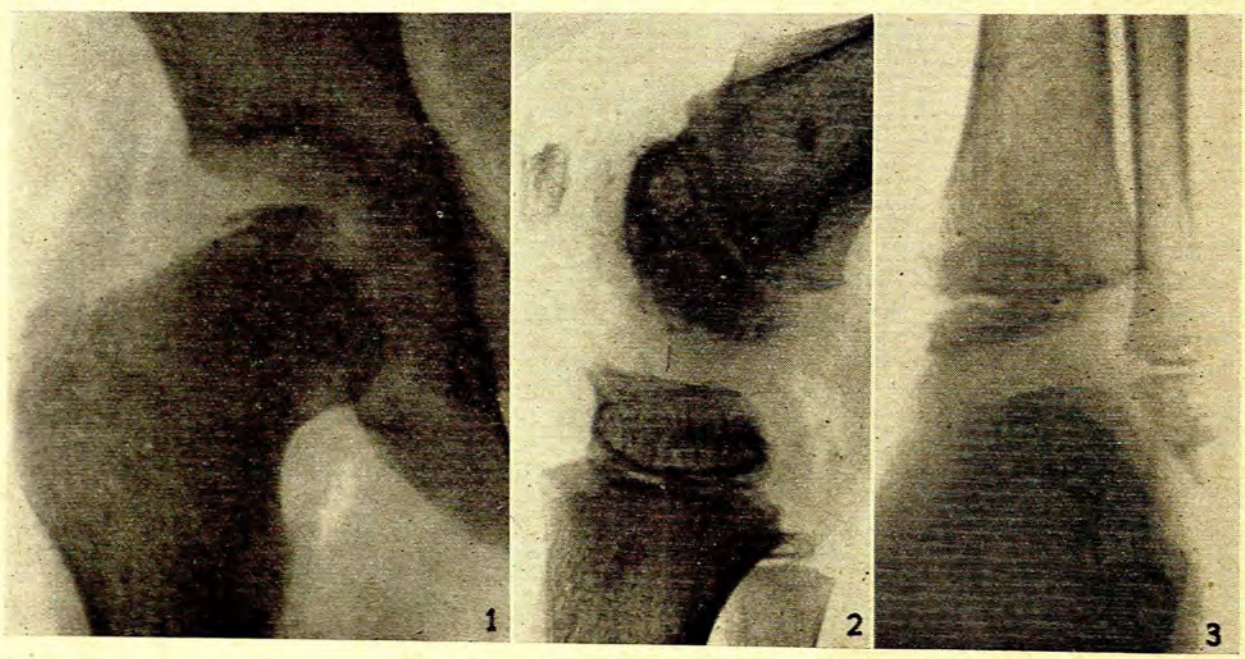


Figs. 1, 2 and 3

are triangular in shape and slightly irregular in outline. Lateral views reveal small irregular patellae with spur formation. The fibular epiphyses are not remarkable, but all the epiphyses at the knees show coarse trabecular structure.

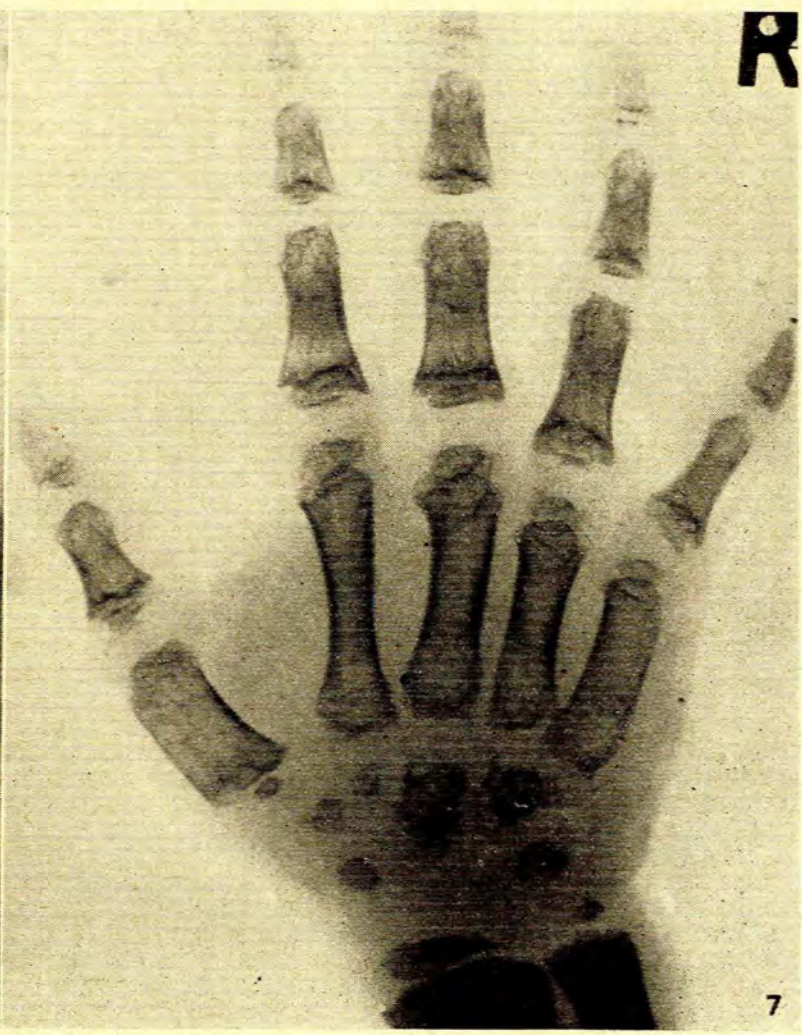
Ankle Joints (Fig. 3). The lower tibial epiphyses are dense and unduly wedge-shaped with marked tapering laterally and producing characteristic obliquity of the articular surface (this triangular appearance is said to occur in 50% of cases studied). The lateral malleoli are low in position. On either side there is a large irregularly-shaped ossific centre, below which several small elongated subsidiary centres are present. There is striking irregularity of the metaphyseal ends of both tibiae.

Feet (Fig. 4). All the epiphyses show marked irregularity in contour and in degree of mineralization. In many instances multiple centres are seen. The metatarsals show numerous discrete punctate centres. These bones are short and broadened, with irregular and rather notched ends, and coarse trabeculation can be seen both proximally and distally. The phalanges are broad and stumpy, with coarsely woven pattern throughout, especially in the big toes. The navicular bones show increased density and antero-posterior compression, presenting the appearance of osteochondritis (Kohler's disease). The other tarsal bones also show slight irregularity in form, and coarse trabeculation. The striking feature in both feet is the symmetry in the disposition and character of the lesions.

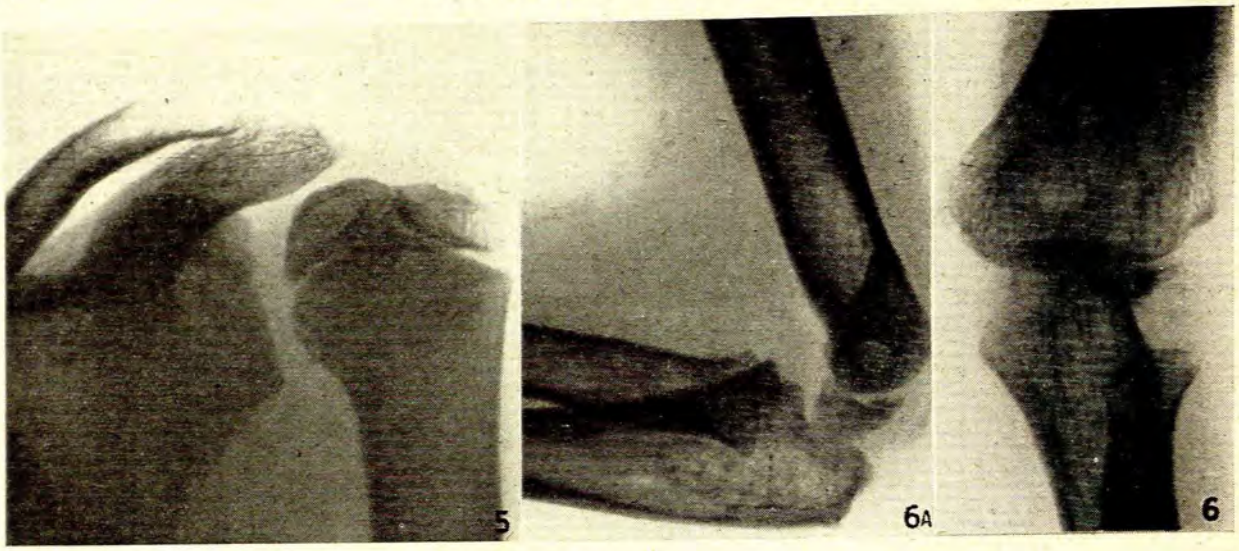
Shoulder Joints (Fig. 5): These show flattened epiphyses arising from several separate centres.

Elbow Joints (Fig. 6). Only the epiphyses of the capitellum, the internal epicondyle and the head of the radius are present. The capitellar epiphyses are dense, and present an irregular stippled appearance.

Hands and Wrists (Fig. 7). Only 6 ossific nuclei are present in the wrists. Bony age is thus retarded. The carpal bones are grossly irregular in form, and this is shown particularly in the capitate and hamate, where sharp spiky projections are present. The distal radial epiphysis is flattened, dense and irregular, and the ulnar centre is very small. Some increased density and



Figs. 4 and 7



Figs. 5, 6 and 6a

irregularity of the adjacent metaphyses is shown. The metacarpals and phalanges are short and broad, with irregular ends and abnormal pattern of cancellous structure. The irregularity of the epiphyses is well demonstrated in the bones of the hand, similar to the changes noted in the feet.

Radiographs of the spine, skull and teeth did not reveal any abnormality.

DIFFERENTIAL DIAGNOSIS

Perthes' Disease. When the signs of bilateral Perthes' disease are found in a patient below the average height it is advisable to radiograph some of the other joints, e.g. shoulder, hips and ankles. Perthes' disease occurs bilaterally in 15% according to Howorth[7] and 7·5% according to Morris.[6] The pathology is different in the two conditions. At any one stage the epiphyses in dysplasia epiphysealis multiplex tend to improve in radiographic appearance, in contrast to osteochondritis, in which the early changes become progressively worse before improvement commences. The abnormal epiphyses in dysplasia epiphysealis multiplex eventually become normal in density, but the outline, though usually smooth, remains permanently irregular. The age at which this permanent definition becomes apparent seems to vary considerably in different cases.

Cretinism is excluded on the general examination. In cretinism the stubby fingers are approximately of equal length. Delay in fusion at the epiphyseal lines in cretins is associated with a band of sclerosis in the terminal layer of the metaphyses.

In *Dysplasia Epiphysealis Punctata* the abnormalities generally are much grosser. The shafts of the long bones are short and thick. Many of these patients die before the age of 1 year and half the cases have congenital cataracts.

In *Morquio-Brailsford Disease* (*Osteochondrodystrophy*) the acetabuli appear markedly enlarged and irregular on X-ray. The spine exhibits special features, viz. the shape of the bodies and in many cases kyphosis. The central prolongation of the bodies with the forward-projecting tongue is quite distinctive and diagnostic.

In *Dyschondroplasia* (*Ollier's Disease*) the epiphyseal changes are comparatively insignificant and are entirely overshadowed by the changes in the metaphyses, which contain unossified cartilage. The changes tend to be unilateral.

In *Osteopoikilosis* the epiphyses are of normal shape. The discrete dense spots characteristic of this condition are not confined to the epiphyses.

In *Osteopetrosis* some epiphyses occasionally show irregularity in density but changes in the shafts dominate the picture.

Pituitary Gigantism. Mottled epiphyses have been described in association with pituitary gigantism.

SUMMARY

A case of dysplasia epiphysealis multiplex is described, believed to be the first case reported in a Bantu. The clinical and radiographic appearances are typical. The results of blood and urinary investigations are recorded and the histological pathology of the case described. The differential diagnosis of this condition is discussed.

We should like to thank Dr. Dorfman, of the South African Institute of Medical Research, for his interest in the case and for the histo-pathological description. For permission to publish the case, we are grateful to the Superintendent of the Coronation Hospital.

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