MULTIPLE EPIPHYSIAL DYSPLASIA

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In 1935 Fairbank described a patient with irregular ossification of several epiphyses, but without the features of any condition then recognised. In 1947 he grouped twenty similar cases under the name of dysplasia epiphysialis multiplex, gave a full description of the clinical and radiological features, and discussed the differential diagnosis from cretinism, dysplasia epiphysialis punctata, chondro-osteodystrophy of the Morquio-Brailsford type, dyschondroplasia, osteopoikilosis, osteopetrosis and pituitary gigantism.

The chief characteristics of this dysplasia are irregularity in density and outline of several developing epiphyses, stubby digits and short stature. The number of epiphyses affected is variable, but usually includes those for the femoral and humeral heads. Deformity of the epiphyses is usually permanent, and often causes osteoarthritis later in life. In addition, changes other than in the epiphyses are often seen. Sixty-three patients, whom he assigns to this group, are known to Fairbank (1954), excluding those whose affected epiphyses are too few to be called multiple. Whereas the cases first collected showed little evidence of hereditary or familial influence, many of those now known do so. Those showing hereditary influence are a mother and four children (three brothers and one sister) (Hirsch 1937); a father and son, reported as exhibiting chondro-osteodystrophy of the Morquio-Brailsford type, but without the spinal changes typical of this condition, and considered by Fairbank to be cases of multiple epiphysical dysplasia (Wood and Robertson 1943); a mother and twin boys reported as exhibiting punctate dysplasia (Resnick 1943); a mother and son (Fairbank 1947); a mother, two sons and a daughter (Jackson et al. 1954); three generations of a family (Maudsley 1955); and a father and son (Maudsley 1955). Those showing familial influence alone are a brother and sister (Barrington-Ward 1912), seven members of one family (Ribbing 1937), two brothers (Emr 1952), three sisters (Waugh 1952), a brother and sister (Watt 1952) and a brother and sister (Maudsley 1955). In addition, unpublished cases known to Fairbank (1954) are a brother and sister, and a second brother said to be affected, two brothers, twin brothers, a girl whose two sisters are said to be affected, and two sisters of a family of ten, of which two others are said to be affected.

The present three cases occur in three generations of one family. In addition, the history indicates that three further members of the family have been affected, one in a fourth generation.

CASE REPORTS

Case 1—A boy aged fourteen. For six years he had limped slightly and the left hip had ached. At the age of ten he had spent some time in bed because of this pain. For the last year it had become more troublesome and the right hip had begun to ache.

Stature 5 feet 3 inches. The left lower limb was half an inch shorter than the right above the knee. Each hip flexed through 130 degrees, and abduction of the left hip was restricted by 10 degrees. The fingers were somewhat stubby (Fig. 16) and there was a slight flexion contracture of the little fingers.

Radiographic examination—Epiphysical or other abnormalities were seen in the hips (Fig. 1), knees (Figs. 4 and 5), feet (Figs. 10 to 12), shoulders (Fig. 13) and hands (Fig. 18). At the elbow the upper end of the radius and the capitulum were irregular. The eleventh thoracic vertebral body was slightly irregular. At the ankle the tibial epiphysial line was convex distally. The skull showed no abnormality.
Case 1—Hips. The left femoral head is mottled and very flattened, and its upper part appears crushed and fragmented. The right femoral head is flattened, but much less, and its texture is normal. The femoral necks are widened. Shenton's line is broken on each side. The left acetabulum is deformed to the shape of the femoral head.

Case 2—Hips. The femoral heads are grossly flattened and irregular, and the acetabula are shaped correspondingly. The right femoral head contains cysts. Mottled opacities below the femoral necks probably represent loose bodies in the joints.

Case 3—Hips. The femoral heads are somewhat flattened and the joints very osteoarthritic.
Case 1. Figure 4—Right knee. The lateral femoral and tibial condyles are mottled. Rounded lips arising from the femoral condyles at the lateral and medial margins of the joint give the condyles an angular shape. The distal end of the femur shows vertical striation. The left knee is similar. Figure 5—Left knee. The patella shows a dense central nucleus within less dense bone. The distal femoral epiphysis bears rounded lips, adjacent to the epiphysial line, more marked in front than behind, and slightly overlapping the metaphysis. The right knee is similar, except for the patellar changes.

Case 2. Figure 6—Left knee. The femoral condyles are flattened and angular, and the lateral condyle shows a shallow depression. There is lipping of femur and tibia laterally. The right knee is similar. Figure 7—Right knee. The lateral femoral condyle is flattened. In the left knee this change is less marked.
Case 2—The boy’s mother, aged thirty-five. At the age of five, in the gymnasium, it was noticed that she could not sit cross-legged. Since the age of eleven the hips had been painful, but lately had troubled her less. Her activities were not materially restricted.

Stature 5 feet 1½ inches. The left lower limb was half an inch shorter than the right above the knee. Each hip flexed through 100 degrees. Abduction was nil in the right hip, and 20 degrees in the left. Rotation was very restricted. Shoulder abduction was restricted by 30 degrees. There was a slight flexion contracture of the little fingers, but the fingers were not abnormally short.

Radiographic examination—The femoral heads were grossly deformed (Fig. 2), and the femoral condyles were flattened and angular (Figs. 6 and 7). The humeral heads were flattened (Fig. 14). The hands and ankles showed no abnormality.

Case 3—The boy’s grandmother, aged seventy-one. While she was at school pain began in the right knee and later affected the left knee and both hips. The patient used one stick, limped slightly and could not climb stairs.

Stature was 4 feet 10½ inches. The left lower limb was half an inch shorter than the right. The hip flexed through 90 degrees, and abduction and rotation were very restricted. There was genu valgum. Shoulder abduction was restricted by 30 degrees. The fingers were somewhat stubby, and there was a slight flexion contracture of the little fingers (Fig. 17).

Radiographic examination—The femoral heads were flattened and the hips osteoarthritic (Fig. 3). The knees were osteoarthritic and the lateral femoral condyles flattened (Figs. 8 and 9). The ankle joints were oblique (Fig. 19). The humeral heads were flattened (Fig. 15).
Case 1. Figure 10—Right foot. Changes are most marked in the great toe. The epiphysis for the base of the proximal phalanx is irregular, and contains a dense central nucleus. Towards its medial limit is a depression, opposite which lies a depression in the metatarsal head. At each margin of the head of the proximal phalanx a tiny island of bone lies within a depression in the phalangeal head. The lateral island is partly fused with the phalanx. In the fourth toe the epiphysis for the base of the terminal phalanx appears to be annular with a central nucleus. All the metatarsal heads are somewhat flattened. Figure 11—The same foot nine months later. The tiny bone island lateral to the head of the proximal phalanx is completely fused to the phalanx, and the medial island is almost completely fused. The first metatarsal head is lipped medially and laterally, having the appearance of early hallux rigidus.

Case 1—Right foot. A depression occurs in the talar head towards the lower limit of its navicular facet. Other tarsal bones are somewhat irregular. The left foot is similar.
FIG. 13
Case 1—Shoulders. The humeral heads are irregular and the glenoids shallow.

FIG. 14
Case 2—Shoulders. The humeral heads are flattened.

FIG. 15
Case 3—Shoulders. The humeral heads are flattened.
The metacarpal heads were somewhat flattened and the metacarpo-phalangeal joints osteoarthritic. Very large osteophytes arose from the adjacent parts of the second and third lumbar vertebral bodies on the left.

The history of three other members of the family is as follows (Fig. 20).

4. Woman aged eighty. Joint trouble since early childhood. At the age of six she was treated in hospital with lower limb traction and cod liver oil. Stature a little under 5 feet.


6. Man. Onset of hip trouble at boarding school, ascribed to damp bed. Diagnosis of "congenital hip joint." Led an active seafaring life, and remained at sea as Master until the age of fifty-seven, when he died of carcinoma of the lip. The first member of the family known to have been affected.

So far as can be ascertained, the remainder of the family, including the boy's sister, who is a ballet dancer, is unaffected.
Fig. 18
Case 1—Hands. Many of the phalangeal epiphyses show a dense nucleus lying within a ring. Several metacarpal and phalangeal heads show shallow depressions, partly formed by small lips at the margins of the heads. The heads of the proximal and middle phalanges have not arrived at their normal bicondylar form. The metacarpal bases, carpus, and radial and ulnar epiphyses are somewhat irregular.

Fig. 19
Case 3—Ankles. The joint lines are inclined obliquely upwards towards the fibulae. The tali are shaped accordingly.
DISCUSSION

Corresponding joints were not affected to the same extent in the three patients examined. The mother's (Case 2) hips were very deformed and contained loose bodies. The boy's (Case 1) left femoral head was grossly flattened. The grandmother's (Case 3) femoral heads showed only moderate deformity, but the hips were very osteoarthritic. The shoulders were affected in each, although the boy's humeral heads did not show the typical flattening. The knees were affected in each, and the lateral femoral condyles of the two older patients were flattened, to the extent of causing genu valgum in the grandmother. Slight flexion contracture of the little fingers was found in all three patients. Otherwise the mother's hands differed little from normal. The other two patients had stubby fingers. Obliquity of the ankles was seen in the grandmother only. The boy's tibial epiphyseal lines at the ankle were convex distally.

The stature was short in the three patients examined, but only the grandmother was less than five feet tall.

![Family tree](image)

**Fig. 20**

Family tree. Conjugal partners are omitted. Members 1, 2 and 3 are the boy, his mother and his grandmother. Members 4, 5 and 6 were not available for examination.

Rounded lips arising from the boy's femoral condyles, at the medial and lateral margins of the knee (Fig. 4), gave the condyles the angular shape described by Fairbank (1947). Between each lip and the condyle's normal curve a small depression was formed. A similar appearance was seen in the boy's hands, in the heads of some of the metacarpals and of the proximal phalanges (Fig. 18). The talar heads also showed small depressions (Fig. 12). These changes in hands and feet were extra-epiphysial. All these small depressions were probably filled with cartilage, and they do not necessarily represent irregularities in the joint surfaces.

Small depressions in the heads of the proximal phalanges of the boy's great toes were each occupied by a tiny bone island (Fig. 10), presumably embedded in cartilage. Those islands lying on the lateral sides of the phalangeal heads were beginning to join the phalanges. Nine months later all these islands had fused (Fig. 11). Shallow depressions were noted in the femoral condyles by Ribbing (1937), who also found that they were sometimes occupied by bone islands. Barrington-Ward (1912) noted, in the knee of a girl of nine years, "an apparent loose body with a corresponding depression in the femoral condyle." Watt (1952) described a similar appearance in the superior articular surface of the talus, which he called osteochondritis dissecans, but which might perhaps represent a bone island which had not joined the main bone, rather than a fragment which had separated.

The distal ends of the boy's femora showed vertical striation (Fig. 4). Such localised striation has been seen in several conditions (Fairbank 1951) but apparently not in this dysplasia.
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The boy's left patella showed a dense central nucleus surrounded by less dense bone (Fig. 5), an appearance similar to that noted by Fairbank (1947) in the capitular epiphyses of three patients. Several phalangeal epiphyses in the boy's hands (Fig. 18), and those of the proximal phalanges of his great toes (Fig. 10) presented the appearance of a dense central nucleus lying within a ring of less dense bone.

The three patients examined, and the other three affected members of this family, all began to have painful hips in childhood. Fairbank (1947) reported others who had suffered similarly. Further, although function is better than might be inferred from the radiographs of the hips, it is sometimes restricted. This experience is contrary to Waugh's (1952) statement that "in no case of dysplasia epiphysialis multiplex have there been symptoms referable to the changes in the femoral heads," and that "function is invariably good." However, the deformable state of the femoral heads probably lasts too long to make treatment by relief from weight bearing practicable, even if the patient were to seek advice before much deformity had occurred.

SUMMARY

Three cases of multiple epiphyseal dysplasia in three generations of one family are described. The history indicates that three further members, one in a fourth generation, have been affected.

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