FAMILIAL PERTHES' DISEASE RESEMBLING MULTIPLE EPIPHYSIAL DYSPLASIA

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Although familial Perthes' disease has been described by various authors it is still not generally accepted as an entity and no case has yet been described in Great Britain. Kehl (1925) and Brill (1927) reported a family in which twenty-six members in six generations were affected by hip joint changes suggestive of Perthes' disease. Perthes himself (1913) reported two affected brothers, and Stephens and Kerby (1946) described a family which showed a dominant inheritance pattern with marked penetrance. The condition has been described in twins by Giannestras (1954) and Soderberg (1957). Very recently Barrie, Carter and Sutcliffe (1958) described the condition of multiple epiphysial dysplasia, which has a definite familial tendency, and it is possible that some of the earlier cases described came under this category.

The family which is to be described here shows four generations, in three of which one or more members have been examined and found affected by an osteochondritis involving the hip joint. The pedigree is shown in Figure 1.

**CASE REPORTS**

**First generation—**I. Subject I/1—No clinical details are available about this woman, but she was said by her children to have walked with a limp all her life, because of hip trouble. She died at eighty-six years. Subject 2 was normal as far as is known, but was not examined.

**Second generation—**II. Subjects 1, 2, 6 and 7 of this generation were examined and had a radiographic skeletal survey of the hips, hands, wrists, knees and ankles. Of these subjects 1, 2 and 6 showed no clinical or radiological abnormality. Subject II/7, a man aged sixty-three years, first began to have pain in both hips in his late forties. His radiograph (Fig. 2) showed flattening of both femoral heads with some osteoarthritic changes. No other skeletal abnormalities were found.

**Third generation—**III. All the members of this and the successive generation have been examined and had skeletal surveys. Subjects 1 and 3 were clinically and radiologically normal.
Radiograph of the hips of subject II 7 (aged sixty-three years) showing flattening of both femoral heads with osteoarthritic changes.

Radiograph of the hips of subject III 2 (aged twenty-four years) showing symmetrical deformity of femoral heads and necks.

Figure 4—Radiographs of the hips of subject IV/1 (aged five years) showing severe flattening of both femoral capital epiphyses. Figure 5—Radiograph of right ankle of subject IV/1 (aged five years) showing irregularity and thinning of lateral side of lower tibial epiphysis. The left ankle showed similar changes.
Subject III/2, a man aged twenty-four years, had pain in the left hip from the age of seven, and a troublesome right hip from the age of twelve. For the past eighteen months he had severe pain, worse with exercise, especially in the left hip. The radiograph (Fig. 3) shows flattening of both femoral heads, with a symmetrical deformity of the femoral necks. Skeletal survey showed no other abnormality.

**FIG. 6**
Radiograph of the hips of subject IV 2 (aged three years) showing irregularity of the epiphysial line in the right hip.

**FIG. 7**
Radiograph of the hips of subject IV/3 (aged two years) showing a small and irregular femoral capital epiphysis on the left side.

**Fourth generation—IV.** All three members of this generation were examined. Subject IV/1, a girl of five years, was first noticed to walk with a limp when she was two and a half years old and she has been treated by relief from weight bearing at home ever since. The radiograph (Fig. 4) shows severe flattening of the femoral capital epiphyses, the left one being reduced to a very thin central portion. Both tibiae (Fig. 5) show irregularity and thinning of the lateral side of the lower epiphysis and there is some reduction in the ossification of the bones of her hands.
Subject IV/2, a boy of three years, began to walk at fifteen months and has never shown any clinical abnormality. The radiograph of his hips (Fig. 6) shows irregularity of the epiphysial line on the right.

Subject IV/3, a boy two years old, is walking normally and no trouble has been noted. On the radiograph (Fig. 7) it can be seen that the left femoral capital epiphysis is smaller than the right one and more irregular in outline.

Skeletal surveys on the above two subjects revealed no other abnormalities.

DISCUSSION

This family shows an osteochondritis of the hip suggesting a dominant inheritance pattern with complete penetrance in the fourth generation.

In all cases described the hip has been the site of osteochondritis. In subject II/7 the original lesion was diagnosed as bilateral osteoarthritis of the hip. Subject III/2 was previously diagnosed as suffering from bilateral Perthes' disease, as was his daughter, subject IV/1, who, however, also had abnormal epiphyses in the ankles and hands and must therefore be considered to be a case of multiple epiphysial dysplasia. The other two children (IV/2 and 3) are so young that it is difficult to give a definite diagnosis.

Is this, therefore, a family with Perthes' disease or one with multiple epiphysial dysplasia in which the hip joint has been singled out to be affected bilaterally in three cases?

Bilateral Perthes' disease is relatively uncommon and in these patients, particularly the one shown in Figure 3, the deformity is remarkably symmetrical on both sides. On the other hand, the changes in multiple epiphysial dysplasia have a radiological distribution which is often most severe in the epiphyses of the wrists, hands and ankles (Barrie et al. 1958). The little girl (IV/1) does show that her hips, ankles and hands have been affected, which gives strength to the diagnosis of a multiple dysplasia. The two youngest children (IV/2 and 3) show only one epiphysis affected and may very well prove to be typical examples of Perthes' disease.

SUMMARY

1. An account is given of a family in which five members in three generations were affected by osteochondritis involving the hips, in three cases bilaterally.
2. One patient showed aseptic osseous necrosis of the epiphyses of the ankles and fingers.
3. The differential diagnosis between Perthes' disease and multiple epiphysial dysplasia is discussed, but it is not certain into which category these patients fall.
4. The assistance of a family history and skeletal survey in diagnosis is illustrated.

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REFERENCES