Perthes' disease and multiple epiphyseal dysplasia

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Summary

Five atypical cases were observed amongst ninety children with Perthes' disease, ten of whom had bilateral hip joint involvement. All five were boys, four being under 4 years of age. Four had bilateral hip joint disease, four presented with hip pain, three showing some degree of retardation of bone growth. In one case the hip disorder was familial, and in four there were bony abnormalities elsewhere.

Despite the absence of the classic signs of multiple epiphyseal dysplasia, a mild form of this condition is a possible alternative diagnosis for these children.

Racial and familial differences are known in the prevalence of Perthes' disease which itself may represent a dysplasia. The pathogenesis of Perthes' disease is still uncertain, although some abnormality of the blood supply to the proximal femoral epiphysis is postulated. That such a vascular defect may be engrafted on to multiple epiphyseal dysplasia is possible, with subsequent joint degeneration which may come to resemble Perthes' disease either clinically or radiologically. A plea is made for the closer study of young children presenting with what may seem to be atypical Perthes' disease.

THE study of Perthes' disease presents problems of aetiology, pathology and diagnosis. The clinical presentation varies and the outcome is uncertain. Some patients, particularly those afflicted early in life and those with no symptoms, appear to have a better prognosis. The authors suspected that in some patients this more favourable outcome might be due to their condition being in fact multiple epiphyseal dysplasia rather than true Perthes' disease.

In this series of ninety children with Perthes' disease, ten were affected bilaterally. Of these patients, one with unilateral and four with bilateral disease had atypical features including, in some, multiple epiphyseal abnormalities which did not seem to fit neatly into any consistent diagnostic pattern.

Multiple epiphyseal dysplasia is a term covering various forms of epiphyseal involvement and having

different modes of inheritance. The first description of the condition was that of Fairbank (1947), who reported twenty cases with a slight male preponderance. He particularly noted the epiphyseal changes, reduction in overall height, platyspondyly, stubby fingers and retardation of bone age. Although at that time Fairbank felt that the condition was not heritable, later reports showed that both autosomal dominant and recessive inheritance occurs.

Other forms of epiphyseal involvement have been reported. Waugh (1952) described three sisters with abnormal hips amongst a sibship of eight, but none was stated to have had hip joint symptoms. In addition, the affected members of this family were dwarfed and had short digits. Maudsley (1955) reported fourteen cases in three families, those affected having reduced height and involvement of the epiphyses of the hips, ankles, knees and spine in decreasing order of frequency. Other families with abnormalities of the knee and ankle have been described, as also has an association with osteochondritis dissecans (Ödman, 1959). Families have also been reported where the hip alone has been affected (Elsbach, 1959).

This last group is similar to or identical with the cases of 'familial Perthes' disease' described in the 1940s. In these patients transmission appeared to be as an autosomal dominant traced by Stephens and Kerby (1946) through five generations. The sex incidence is equal, contrasting with the usual five males to one female amongst patients with non-familial Perthes' disease. The proportion of bilateral cases was close to 50%, which again differs strikingly from the 10% of bilateral cases usually found amongst most series of patients with Perthes' disease.

Case reports

Case 1

A boy aged 4 years presented with a limp (Fig. 1). Little was thought of this until his father waddled in—overweight and complaining of hip pain. The boy's progress over the next 3 years was unremarkable from the Perthes' disease viewpoint, but

within 2 years of onset he began to complain of pain in his knee, and radiographs showed a defect in his distal femoral epiphysis strongly resembling osteochondritis dissecans. His father's hip radiographs (Fig. 2) showed shortened femoral necks, flattened heads and externally rotated hips. Quite by accident, the authors discovered that his cousin also had Perthes' disease, this girl being the daughter of the paternal grandfather's daughter (Fig. 3). In addition to the femoral epiphyseal changes there were abnormalities in the epiphyses of her feet, but the hands were normal. This girl's mother's hips were radiologically dysplastic. More recently another cousin has presented with osteochondritis dissecans of the knee. Paediatric and radiological colleagues of the authors have examined these two families and would rather regard their condition as familial Perthes' disease than as multiple epiphyseal dysplasia.

Case 2

The hips of a 3-year-old boy (Fig. 4) showed delay in the appearance of the capital femoral epiphyseal centres. When they did appear at the age of 7 years they were small and fragmented (Fig. 5). He also had minor changes in the proximal tibial epiphyses (Fig. 6).

Case 3

This 11-year-old boy presented with an irritable left hip, and radiography displayed bilateral Perthes'-like changes (Fig. 7). His right hip was symptomless. His height was on the 25th centile and there was reversal of the normal carrying angle of the elbow and he had mild genu varum. Of his two brothers, one whose height is on the 50th centile has normal elbows but the other whose height lies on the 10th centile has similar varus elbows but normal hip radiographs.

Case 4

At the age of 30 months this boy appeared with radiological changes in his hips and minimal symptoms (Figs 8 and 9). He also had abnormalities in the foot, with a dome-shaped talus (Fig. 10), some metatarsal epiphyseal abnormalities (Fig. 11) and minor irregularity of the distal tibial epiphysis. His toes were stubby, but his overall height and weight were normal.

Case 5

This child had an abdominal radiograph after swallowing a pin. Abnormal femoral epiphysis were noted at the age of 3 years but he had no hip symptoms either then or 1 year later. His family history was unrevealing, but his height and weight lay well

below the 3rd centile. There was no sign of hypothyroidism.

Four of these five patients presented with pain in the hip. If these are, in fact, minor manifestations of multiple epiphyseal dysplasia it is interesting to speculate on the reason for the development of hip pain early in childhood. Is there really a common pathogenesis between this and Perthes' disease?

Discussion

The postulated pathology of Perthes' disease and the relevance of Trueta's work (1957) in the blood supply of the femoral head is well known. Anatomical variations in vasculature of the proximal femoral epiphysis may account for the appearance of Perthes' disease in certain subjects and such abnormalities or variations may have a heritable background. There is certainly racial variation in prevalence. Fisher (1972) found that black races are less often afflicted than white, although it has been said that Perthes' disease is common amongst Nigerians. Kemp (1973) has suggested that superficially placed blood vessels supplying the femoral head may be obliterated by a rise in intra-articular pressure such as may occur in the effusion of a non-specific synovitis of the hip. His views are based upon experimental work with dogs and it is perhaps significant that he found some breeds susceptible to joint changes, e.g. miniature poodles, whilst beagles and Jack Russell terriers were not. In Perthes' disease there may be delayed incorporation of retinacular vessels into the substance of the femoral neck. It is possible that avascularity could be superimposed upon epiphyseal dysplasia in some cases and, indeed, there may be predisposing factors in the relationship of the retinacular vessels to the femoral head and neck.

The available pathological studies on Perthes' disease show frequent fissuring and fibrillation of the articular cartilage often associated with subsidence of the subchondral bone (Mizuno *et al.*, 1966). But such changes may well be secondary. It is conceivable that structural failure of the epiphyseal cartilage could occur in the hip joint in minor degrees in epiphyseal dysplasia, although the report of Hunt *et al.* (1967) described thickening of the articular cartilage. Their material was obtained following through-hip amputation for osteosarcoma in a child with epiphyseal dysplasia. They described, in addition to the thickened articular cartilage, disarray of epiphyseal ossification with bars of hyaline cartilage extending into the diaphysis. In the epiphyseal cartilage itself vertical clefts, focal calcified deposits and irregular matrix staining were also noted.

A further feature common to Perthes' disease and

FIG. 1. Perthes'-like changes in the right hip joint at age of 4 years.

FIG. 2. Father of case no. 1. Bilateral changes in hip joints.

FIG. 3. Cousin of case no.1. Bilateral epiphyseal changes.

FIG. 4. Case no 2, aged 3 years. Note the broad femoral necks, irregular metaphyses and failure of appearance of the ossific nuclei.

FIG. 5. Case no. 2, hips at age of 7 years.

FIG. 6. Case no. 2, knees at age of 5 years. The tibial epiphyses slope steeply medially and laterally.

FIG. 7. Case no. 3, aged 9 years. There is loss of epiphyseal height in both hips although symptoms were confined to the left side.

FIG. 8. Case no. 4. Hips at the age of 33 months: the capital epiphyses are small and flattened.

FIG. 9. Case no. 4. Hips at the age of 10 years showing epiphyseal irregularity and flattening.

Fig. 10. Case no. 4, aged 10 years: note abnormal dome shaped talus.

multiple epiphyseal dysplasia is the delayed appearance of epiphyseal ossification centres which, however, in the latter, fuse normally on time. Jacobs, Harrison and Turner (1974) found that of 157 children with Perthes' disease, 95% of boys and 85% of girls had significantly retarded bone age in comparison with 20% of boys and 4% of girls in a suitable control group. Delayed appearance of the affected epiphyseal centres has been described in

many of the varieties of multiple epiphyseal dysplasia, and was a feature of cases 2, 4 and 5 in the present series.

The five patients here described comprise four of the ten bilateral cases amongst ninety patients with Perthes' disease. This incidence suggests the possible value of more careful investigation of patients with bilateral hip disease, particularly the younger ones. Case 1 had features in common with the so-called 'familial' Perthes' disease and case 5 with a limited type of epiphyseal dysplasia. Cases 2 and 4 had changes approaching the milder forms of a more generalized epiphyseal dysplasia and patient no. 3 may well have had Perthes' disease engrafted on a pre-existing hip epiphyseal dysplasia.

Conclusions

The similarities between Perthes' disease and multiple epiphyseal dysplasia may be fortuitous and nothing more than architectural in that they reduce the competence of the femoral head as a weight-bearing structure. Either may lead to early cartilage failure and subsequent secondary changes. Perhaps one might be justified in considering that Perthes' disease itself may be a form of mild skeletal dysplasia, in which aberrant features of vascularity, or of growth and maturation predispose to epiphyseal infarction. Identifying patients with a constitutional epiphyseal abnormality in addition to Perthes'-like changes may be useful in determining ultimate prognosis, although one suspects in the long term, disability in the hip joints is due to secondary changes and that these can only be made worse by the presence of other epiphyseal abnormalities.

FIG. 11. Case no. 4, aged 10 years: the toes are shortened. The 1st metatarsal has an extra capital epiphysis.

Acknowledgment

The authors wish to express their gratitude to Dr I. R. S. Gordon for help and advice in the preparation of this paper.

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