CLINICAL VARIATION IN DYSCHONDROSTEOSIS

A REPORT ON 13 INDIVIDUALS IN 8 FAMILIES

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Thirteen patients with dyschondrosteosis from eight families are reviewed and their clinical and radiographic variation noted. Inheritance is likely to be autosomal dominant but with only 50 per cent penetrance. Stature was moderately reduced, due to shortening of the bones of the leg. Radio-ulnar shortening could either involve both bones equally or the radius predominantly, in which case a typical Madelung deformity was seen. Tibio-fibular disproportion was present in half the patients, two of them having severe deformity associated with tibia varum and a long fibula. The treatment of one of these patients is described. It is recommended that patients with dyschondrosteosis should be kept under surveillance during the growing period. Problems in the limbs, especially the legs, may require operations to equalise the length of the two bones.

Dyschondrosteosis is a form of mesomelic dysplasia first described over 50 years ago by Léri and Weill (1929). Patients are of short stature due to shortening of the limbs and the most obvious sign is a Madelung deformity at the wrist.

Anton, Reitz and Spiegel (1938) and Dannenberg, Anton and Spiegel (1939) reviewed the literature and identified 171 cases of non-traumatic Madelung's deformity, gave detailed criteria for diagnosis and preferred to name the disorder "dyschondroplasia of the distal radial epiphysis". They noted a hereditary influence as well as the short stature of their patients and it seems likely that most of their cases had dyschondrosteosis. Maroteaux and Lamy (1959) described two families and noted shortening of the pubis to heel segment and deformity of the upper end of the tibia. More recently Langer (1965) described seven cases in three families, with deformed forearms as the predominant feature, although some were short-legged. Inheritance was noted to be dominant, with a sex ratio of four affected females to one male and no male to male transmission, suggesting possible X-linked dominance. However, male to male transmission was present in the family described by Carter and Currey (1974). Herdman, Langer and Good (1966) reported five cases in two families and Sensenbrenner (1974) reported two families with one instance of tibio-fibular disproportion. Martinelli and Campailla (1975) described three cases in one family and proposed a "major" or complete

form of the condition and a "minor" or incomplete form.

Henry and Thorburn (1967) classified Madelung's deformity into four groups: post-traumatic, those with bone dysplasia (diaphysal aclasis), those with Turner's syndrome and "idiopathic", and noted premature fusion of the growth plate of the radius with consequent overgrowth of the ulna. Golding and Blackburne (1976) writing from Jamaica, described 26 female patients with primary Madelung's deformity with no shortening of the lower limb and with no affected relatives. Their male patients with "Madelung's deformity" were all found to have dyschondrosteosis. They noted two variants—congenital short ulna and a reversed Madelung's deformity with anterior bowing of the radius and anterior dislocation of the ulnar head. Funderbunk *et al.* (1976) described a family with mesomelic shortening closely resembling that of dyschondrosteosis, but in addition the family had hereditary nephritis. Lichtenstein, Sundaram and Burdge (1980) described a family of an affected father and two daughters where the females had dyschondrosteosis including Madelung's deformity, but the father had only shortening of the forearm and lower leg.

No previous report stresses the tibio-fibular disproportion, nor comments on mechanical problems at the knee or ankle. A patient with dyschondrosteosis treated by one of the authors (GEF) had a deformity of the lower limb which required operation and it was decided to review the range of physical abnormality and complica-

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tions of this condition. Height, body proportions and radiological features were studied with particular reference to radio-ulnar and tibio-fibular disproportion.

MATERIAL AND METHODS

Eight index patients and five affected relatives, five male and eight female, were reviewed. All were attending Skeletal Dysplasia Clinics at the Princess Margaret Rose Orthopaedic Hospital, Edinburgh (eight cases), the Western Infirmary, Glasgow (three cases), or the Robert Jones and Agnes Hunt Orthopaedic Hospital, Oswestry (two cases). The age of the patients at the time of examination was from 4 to 43 years, four being between four and seven years, five between 12 and 15, and four between 32 and 43.

Pedigrees were obtained and clinical and radiographic examination carried out of index patients and of affected relatives. The standing height, the span, the head to pubis and pubis to heel measurements taken with the patient lying down, and the natural history and complications with limb growth were recorded.

RESULTS

Clinical features. The presenting sign in seven of the 13 patients was short stature or limbs which were noted to be disproportionately short in relation to the trunk (Fig. 1). This was usually noticed under the age of eight years and in one case as early as nine months, possibly because her mother was similarly affected. Four patients presented with pain or deformity of the wrist, all between 12 and 15 years old. One boy presented at the age of 12 years with a painful foot, and one girl at six years old with short stature and "genu varum" (in fact tibia varum), which had increased over the previous three years. She was one of the only two patients with marked tibio-fibular disproportion, both of whom had minimal signs in their upper limbs. The 43-year-old father of one patient had noticed no abnormality, although his body proportions were altered and his forearm deformity obvious. Although only one-quarter of patients presented complain-

Fig. 1

A six-and-a-half year old patient with dyschondrosteosis. Her height was 96.5 centimetres and her limbs disproportionately short in relation to her trunk.

ing of a deformity of the forearm, all showed it to some degree, with prominence of a dorsally subluxated or dislocated ulnar head, posterolateral bowing of the radius and slight limitation of wrist movements. There were no other abnormal findings in the upper limb apart from a 30-degree loss of supination in two patients. No wrist needed specific treatment for pain or deformity.

Body proportions. The standing height varied from near normal to a moderately short stature (Fig. 2), three of the

Fig. 2

Fig. 3

The height and span of 13 patients. Figure 2—Height is compared with standards from Tanner, Whitehouse and Takaishi (1966). Figure 3—Span is compared with standards from Provis and Ellis (1955).

13 patients having heights at the third percentile and five below it. Arm span was reduced in all but three patients (Fig. 3). The head to pubis measurement was within normal limits in all patients, while pubis to heel measurements were reduced in all cases, when compared with the standards given by Tanner, Whitehouse and Takaishi (1966) and by McKusick (1972).

Radiological features. The appearances of the forearm and wrist varied from near normality, with slight bowing and reduction in absolute length in three patients, to shortening of one or both forearm bones with the typical changes of Madelung's deformity. Figures 4 to 6 show the failure of development of the medial half of the lower radial epiphysis which was obvious in the earliest available radiograph at four years of age. Figure 7 shows a normal 12-year-old wrist for comparison. Absence of the epiphysal line medially was sometimes accompanied by rarefaction at the lower medial border of the radius and in one case the epiphysis extended upwards between the radius and ulna. In some patients both forearm bones were equally reduced in length with thickened shafts and minimal subluxation or bowing, but in others the ulna appeared to continue growing and dislocated posteriorly at its lower end (Figs 8 and 9). There was no case with a reversed Madelung's deformity, and all patients had bilateral involvement.

Lower limb changes were less severe and six of the 13 patients were normal, except with regard to absolute length of the tibia and fibula. Tibial changes were usually minor, with only three patients showing beaking on the

medial aspect of the upper end. The fibula, by contrast, was more variable and was relatively too long in four patients and too short in two patients (Figs 10 to 12). This disproportion could be apparent at either the upper or the lower end, or at both, and could be asymmetrical in the two limbs. In two patients the disproportion between the tibia and fibula was of greater severity and was accompanied by tibia varum at the uppermost third of the bone, with problems at the knee. In contrast, although the ankle mortise appeared abnormal in some radiographs, only one patient had developed symptoms and they were of minor degree.

Bone maturation, apart from the lower radial epiphysis, was not delayed and there were no notable changes in other parts of the skeleton, or associated anomalies in other systems.

Fig. 8

Fig. 9

Radiographs of the wrist of an adult patient showing the relative lengthening of the ulna with posterior dislocation.

Fig. 4

Fig. 5

Fig. 6

Fig. 7

Figures 4 to 6—Radiographs of the forearm of a patient with dyschondrosteosis at four years, 12 years and as an adult. Figure 7—Radiograph of the wrist of a normal 12-year-old for comparison.

Fig. 10

Fig. 11

Fig. 12

Radiographs of different patients to show disproportion of leg bones. Figure 10—Normal upper end, but fibula appears too long at the lower end. Figure 11—Fibula appears short at both upper and lower ends. Figure 12—Fibula appears long at both upper and lower ends.

Management of the tibio-fibular disproportion. One patient had minimal signs in the upper limb but developed increasing tibia varum from the age of six years with progressive sloping of the ankle mortise (Fig. 1 and Figs 13 to 15). At the age of 10 years tibial osteotomy and lengthening was combined with epiphysiodesis of the distal fibular epiphysis. This gave a five-centimetre increase in tibial length, and resulted in satisfactory correction of the tibial deformity (Fig. 16). The other patient with marked tibio-fibular disproportion was 19 years old when first seen, and refused treatment.

18). The diagnosis is made obvious by multiple exostoses in other parts of the skeleton.

In Ollier's disease there is usually asymmetrical involvement of the limb bones with enchondromata and islands of cartilage in the shafts of long bones (Fig. 19). The ulna is predominantly affected, and the disorder is non-genetic.

Hypochondroplasia is more difficult to distinguish from dyschondrosteosis, since it is also of autosomal dominant inheritance, patients have the same moderate degree of short-limbed short stature and frequently also

Further radiographs and clinical photographs of the patient shown in Figure 1. Figure 13—Radiograph taken at six-and-a-half years of age showing minor beaking at the upper end of the tibia, with relative lengthening of the fibula apparent at the lower end. Figure 14—Radiograph at nine-and-a-half years showed fibular lengthening apparent at both ends of the bone. Figure 15—Appearance at nine-and-a-half years shows increased tibia varum. Figure 16—Clinical appearance one year later, six months after bilateral tibial osteotomy and lengthening with epiphysiodesis of the distal fibular epiphyses.

Fig. 13

Fig. 14

Fig. 15

Fig. 16

Genetics. Of the eight index patients, two were male and six female. There was unequivocal evidence of the disorder in only two of the 10 parents for whom medical evidence could be obtained but it is likely from the histories that the disorder was present in at least five of the 16 parents. Three of 13 sibs were affected, and none of four children. No effect of paternal age could be demonstrated and there was no consanguinity of the parents. Father to son transmission was definite in one case and probable in a second. It seems therefore that this disorder is of autosomal dominant inheritance, although not fully penetrant, since only about 25 per cent of first-degree relatives were clinically affected, rather than the 50 per cent expected of full dominant inheritance.

Differential diagnosis. Trauma or infection with secondary effects on the growth plate may cause similar deformities, but these are unlikely to be bilateral or to affect both upper and lower limbs.

Diaphysal aclasis is a developmental disorder also of autosomal dominant inheritance, but the abnormalities of the forearm bones differ from dyschondrosteosis, shortening being predominantly of the ulna (Figs 17 and

tibio-fibular disproportion (Wynne-Davies, Walsh and Gormley 1981). Madelung's deformity does not develop in hypochondroplasia, metaphyses may be splayed (Fig. 20) and changes such as short pedicles and scalloping of the posterior border of vertebral bodies may be apparent in the spine.

DISCUSSION

Dyschondrosteosis is clearly a distinct condition of autosomal dominant inheritance and the most usual feature appears to be failure of development of the lower radial epiphysis on its medial side. This leads in some cases to a typical Madelung deformity with shortening and bowing of the radius and dorsal dislocation of the ulnar head. However, nearly equal shortening of both radius and ulna may occur, with only minimal subluxation and bowing. No patient in this series has required operation for symptoms or deformity of the wrist.

Tibio-fibular disproportion has not previously been stressed, but in this series half the patients had obvious involvement of the lower limb, two of them with marked deformity. As in the forearm, the length of the two leg bones may be reduced, equally or disproportionately, but

Fig. 17

Fig. 18

Fig. 19

Fig. 20

Differential diagnosis. Figure 17—Diaphyseal aclasis. The lower end of the radius is not markedly sloping and the ulna is short. Figure 18—Diaphyseal aclasis. The radius is curved, but the carpus is not pyramidal in shape and the ulna is obviously short. Figure 19—Ollier's disease. Cartilaginous islands are seen in the shafts of radius and ulna. There is radial bowing but the ulna is short. Figure 20—Hypochondroplasia. Radiograph of wrist which is normal apart from splaying of the metaphyses (compare with Fig. 7).

unlike the ulna, the fibula may appear to be too long or too short. Operation may be required to correct the varus deformity of the tibia or the ankle, perhaps by tibial lengthening or by arresting the growth of the fibula by

epiphysiodesis. It is recommended that patients with dyschondrosteosis should be kept under review during the growing period, particularly for growth problems affecting the lower limbs.

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