Spondyloperipheral dysplasia

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SUMMARY Skeletal dysplasia with autosomal dominant inheritance was found in four members of one family and in one sporadic case. The syndrome comprises brachydactyly E, platyspondyly, abnormality of the sacroiliac joint, disturbance of metaphyseal modelling, epiphyseal dysplasia, and short stature. This study deals with a particular type of spondyloepiphyseal dysplasia and compares it with similar cases in two previously published papers.

In 1977 Kelly *et al*¹ described the occurrence of platyspondyly, brachydactyly, and epiphyseal dysplasia in three members of a family. The syndrome was classified as a special type of spondyloepiphyseal dysplasia and was termed spondyloperipheral dysplasia. Two years later Sybert *et al*² published a report of a family similarly affected, with brachydactyly and alteration of the vertebral bodies as well, but without epiphyseal dysplasia. As five persons were affected, autosomal dominant inheritance was proven. The great variability of individual symptoms in the family members and the relatively few persons in both the families made Sybert *et al*² uncertain whether the syndrome was the same as in the family of Kelly *et al*¹.

We had the opportunity to see a family and one sporadic case with a syndrome with the same basic findings, with some features similar to those described by Sybert *et al*² and some skeletal findings similar to those in the family of Kelly *et al*¹.

Case reports

FAMILIAL OCCURRENCE (FIG 1)

Case 1 (III.1)

The proband, a 38-year-old male, presented with pain in the knee joint owing to subchronic non-specific synovitis. Disturbance of the metaphyseal modelling of the tibia initiated a detailed x-ray and genetic examination. His height was 165 cm and weight was 57 kg. The bones of the arm, the fourth metacarpals, and the distal phalanges of the thumbs were shortened, but the feet were without evident malformation.

FIG 1 Pedigree consistent with autosomal dominant inheritance.

Case 2 (III.4)

The sister of the proband was 29 years old. Four years ago pain in the lumbar spine had started, followed by pain in the cervical spine, and later the head. Her height was 152 cm and weight was 52 kg. Brachydactyly of the hands and unequal length of the metatarsals were evident and there was pectus carinatum.

Case 3 (IV.2)

The daughter of case 2 was 10 years old. Her height was 132 cm and weight was 32 kg. Bilateral short fourth metacarpals and the feet were the same as in case 2.

A 6-year-old boy (IV.4), brother of case 3, had a thorax deformity of the same type as his mother (case 2), but the hands and feet were within normal limits. His height (108 cm) was normal for his age.

Case 4 (II.1)

The mother of the proband was 59 years old. Six years ago she experienced pain in both hip joints. Her height was 142 cm and weight was 54 kg. She

had brachydactyly of the hands and feet and external rotation contracture and restricted movement in both hip joints.

Case 5 (I.1)

The mother of case 4 died at the age of 81. She was only a little taller than her daughter. She became blind towards the end of her life. The family had noted the irregular length of her metacarpals.

SPORADIC OCCURRENCE

Case 6

A 47-year-old man had suffered from pain in the hip joints since the age of 30. He had undergone repeated surgical treatment of deformed toes. His sister, 5 years younger, had been treated for coxarthrosis for several years. The relatives could not be examined, but the patient had not noted any deformities of the hands or feet. His height was 154 cm and weight was 70 kg. The metatarsals were of unequal length as were the metacarpals, and he had limited movement of the hips with pain. Abduction in both shoulders was limited to 45°.

Radiographs

HANDS

Shortening of the fourth or fifth metacarpals or both, sometimes with a larger diaphysis, was present in all the patients except case 2. The third metacarpal was also shortened in case 6 (fig 2). Invagination of the epiphyses (cone-shaped epiphyses) of the middle phalanges of the second and fifth fingers (fig 3) appeared to be an early stage of deformity of the

FIG 2 Case 6 (sporadic). Radiograph of both hands. Note shortening of 3rd to 5th metacarpals bilaterally, short distal phalanges of thumbs and 3rd finger on the left, disturbance of metaphyseal modelling of middle phalanges, and broadening of lower epiphysis of radii with horizontal direction of articulation line.

FIG 3 Case 3 (IV.2). Radiograph of both hands. Note shortening of 4th metacarpal on the left, thicker 5th metacarpals, invagination of epiphysis of middle phalanges of forefingers, and shortening of distal phalanges of thumbs and 3rd fingers.

base as seen in the adult patient (case 4). The metaphyses of the middle phalanges in cases 1 and 6 (fig 2) revealed a bottle-like deformity. Short distal phalanges of the thumbs were found in all patients, but in case 1 only the thumbs were affected, whereas case 2 had distal brachyphalangy of all fingers.

Only cases 2 and 4 had symmetrical hand anomalies.

FEET

The usual finding was shortening of all phalanges of the toes and hyperconcavity of the bases of the proximal phalanges, resulting from invagination of the epiphyses (fig 4). In contrast to the hands, shortening of the fourth metatarsal was present only in cases 2 and 3 (fig 4). In case 6 there was also

FIG 4 Case 3 (IV.2). Radiograph of both feet. Note shortening of 4th metatarsals, invagination of epiphysis of proximal phalanges, and brachyphalangy.

unilateral shortening of the second metatarsal. In cases 2, 4, and 6 the proximal phalanges of the big toes were of unequal length.

SPINE

The deformity of the spinal column in cases 2, 4, and 6 was mainly the result of hyperconcavity of both horizontal surfaces of the vertebral bodies, accompanied by diminished height. The most severe changes in these three patients were seen in the lumbar region, showing the so-called 'fish vertebra' pattern (fig 5a-c). In the thoracic region the surface concavity of the vertebral bodies was situated more dorsally (fig 5d). The cervical vertebrae were changed

only in the lower half which showed slight biconcave platyspondyly.

These findings were less marked in case 3 and present only in the lumbar spine. In case 1 the biconcavity was not present (fig 5a), but the pedicles were short, and on the anteroposterior radiograph the interpedicular distance did not narrow distally, so the spinal canal was relatively smaller than normal.

PELVIS

Cases 1 and 6 had an identical abnormality in the sacroiliac joint (fig 6) which was not present in the other cases. Sacralisation of the fifth lumbar vertebra was seen only in case 3 (fig 7).

FIG 5 Lateral radiograph of lumbar spine. (a) Case 1 (III.1), (b) case 2 (III.4), (c) case 3 (IV.2) + thoracic spine, (d) case 6. (a) Slight wedge-like deformity of L2 only, short pedicles in all visible vertebrae; (b-d) platyspondyly with biconcave deformity.

FIG 6 Case 1 (III.1). Radiograph of pelvis. The hyperplastic posterior parts of both iliac bones are unusually extended over the sacral bone and reach nearly to the margin of the vertebral body of L5. The overlaps in the sacroiliac joints are greatly increased. There is asymmetry of both ischia and flattened femoral heads with marginal osteophytes.

FIG 7 Case 3 (IV.2). Radiograph of pelvis. Note bilateral sacralisation of L5 and low epiphysis of the femur with steep growth line.

HIP JOINTS AND LOWER LIMBS

In case 3, the only child in the series, there was hypoplasia of the upper femoral epiphyses (fig 7) with slight flattening of the head. There was slight coxarthrosis in case 1 and severe coxarthrosis in cases 4 and 6. In cases 1 and 2 the lower ends of the femora and upper ends of the tibiae were broadened

(fig 8). The femora were spindle-like, whereas in the tibiae the broadening was present only on the medial side, with a slight exostotic prominence.

SHOULDER JOINTS

Epiphyseal dysplasia of the upper end of the humerus was only present in two cases. In case 3 there were no clinical signs, but case 6 had limited abduction. There was a shallow defect on top of the greater tubercle, probably owing to pressure by the acromion on abduction.

ELBOW JOINTS

These were normal.

WRIST JOINTS

The articulation area of the distal epiphyses of the radius in cases 2, 4, and 6 were only slightly slanted, and in case 6 the adjacent lower shaft was broadened (fig 2). In cases 1 and 2 the ulna was $\frac{1}{2}$ cm shorter on the right, and the styloid process in cases 2 and 4 showed independent ossification bilaterally.

Discussion

The abnormalities of the hands and feet in these cases correspond to brachydactyly E showing one or more short and broad metacarpals or metatarsals, usually excluding the first digit, short middle phalanges in the fifth finger, and short distal phalanges in the thumb. Some have short middle phalanges of the second digit and short distal phalanges of the other digits. Affected subjects are short in stature and have normal facies. Involvement of vertebrae or long bones, as seen in our family and

FIG 8 Case 1 (III.1). Anteroposterior radiograph of left knee joint. Note disturbance of metaphyseal modelling of tibia with prominence (arrow) on the inner side and spindle-like broadening of the lower metaphysis of the femur.

the sporadic case, has not been considered part of the spectrum of brachydactyly E.² Only two other similar families have been reported so far.^{1,2}

The bone changes in our patients are similar to those described by Sybert *et al.*² The findings in the hands are the same, even in the broadening of the metaphyses of the middle phalanges, as can be seen in their published radiograph of case 5 (fig 6), but which was not mentioned by the authors. The similarity is also evident in the metaphyseal changes of the lower femora and upper ends of the tibiae in case 1 of their study. The spinal abnormalities are also of the same nature as in our patients. Sybert *et al.*² do not refer to pelvic abnormalities, except in their case 3 they mention bilateral congenital dislocation of the hips, although radiographs were not available. In contrast to our findings they did not note any epiphyseal dysplastic changes, although invagination of an epiphysis of the index finger can be seen in the radiograph of their case 5. Nevertheless, the authors suggest that the syndrome belongs

to the spondyloepiphyseal dysplasias. Their pedigree clearly shows autosomal dominant inheritance.

The second family of three persons in the study of Kelly *et al.*¹ differs from those of Sybert *et al.*² and our patients in having severe epiphyseal changes in the elbows and shortening of the ulnae.

The great variability and limited number of patients makes it uncertain whether the abnormalities observed in the other two reported families and in our patients represent the same syndrome.

References

- ¹ Kelly TE, Lichtenstein JR, Dorst JP. An unusual familial spondyloepiphyseal dysplasia: 'spondyloperipheral dysplasia'. *Birth Defects* 1977;**13**:149-65.
- ² Sybert VP, Byers PH, Hall JG. Variable expression in a dominantly inherited skeletal dysplasia with similarities to brachydactyly E and spondyloepiphyseal-spondyloperipheral dysplasia. *Clin Genet* 1979;**15**:160-6.

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