SPONDYLO-EPIPHYSEAL DYSPLASIA TARDA

THE X-LINKED VARIETY IN THREE BROTHERS

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Three brothers with the X-linked variety of spondylo-epiphyseal dysplasia tarda are presented. Their clinical courses differed: one was relatively free of symptoms, the second had slowly progressive osteoarthritis of the hip, and the third had an acute, disabling arthritis necessitating total hip arthroplasty. The clinical features of this disorder are reviewed.

Spondylo-epiphyseal dysplasia tarda is a form of short-trunked dwarfism. It now appears that this condition can be inherited in one of three modes: X-linked (Jacobsen 1939; Maroteaux, Lamy, Bernard 1957; Langer 1964; Branford, Beveridge and Wynne-Davies 1982), autosomal recessive (Wynne-Davies, Hall and Ansell 1982; Al-Awadi et al. 1984) or dominant (Felman 1969; Connor, Evans and Sardharwalla 1982; Barber, Gow and Mayo 1984). In 1927 Nilsonne first described the condition in three brothers. Jacobsen (1939), in the first of the few papers in the English literature, described 20 affected males in five generations with what he called hereditary osteochondrodystrophy deformans. Maroteaux et al. (1957), in the French literature, coined the term spondylo-epiphyseal dysplasia tarda.

In 1968 Specht reported a patient with low back pain, and radiographic irregularity of vertebral endplates with bi-convex lower lumbar vertebral bodies seen in anteroposterior views. There was disc space narrowing and apparent calcification of the intervertebral cartilage. No hereditary pattern was discernible, nor were there any complaints of peripheral joint pain. A computer-assisted literature search revealed only four other references to this type of the condition (Diamond 1970; Wynne-Davies et al. 1982; Crossan, Wynne-Davies and Fulford 1983; Wynne-Davies and Gormley 1985), none of which described the classic X-linked disease.

We describe a family in which all three brothers had the condition, with two unaffected parents and two unaffected sisters. The differing ways in which these patients presented is emphasised and the condition is reviewed, with particular attention to its orthopaedic aspects.

CASE REPORTS

Case 1. The eldest of the three brothers presented at the age of 29 years with an 18-month history of right hip pain and a much longer history of low back pain. Hip replacement had been recommended at a previous consultation in Australia. He worked seven days a week as a cabinet maker and had had no time off work, being able to walk 3 to 4 km without symptoms. He had significant night pain and some pain at the end of the day.

His height was 147 cm (vertex to pubis 66 cm, pubis to floor 81 cm) with the typical appearance of a short trunk and slight kyphosis. His right hip had reduced abduction and lateral rotation with normal flexion and no flexion deformity. Radiographs of his hips and spine were similar to those of his two brothers. He was not sufficiently disabled to warrant arthroplasty, and a course of anti-inflammatory medication gave relief of symptoms.

Case 2. The second brother, aged 27, seemed fit and had no musculoskeletal complaints. He was of short stature, this having been noticed when he was 10 or 11 years of age. He played squash regularly, though he had had a brief episode of mild hip discomfort two years before.

His height was 153.5 cm with a typically short torso. Range of hip movement was equal bilaterally with 100° of flexion, 25° of both abduction and adduction, and 30° and 10° of medial and lateral rotation respectively. His leg lengths were unequal, 83.5 cm on the right and 81.5 cm on the left, giving him a slight lumbar scoliosis. There were no other clinical abnormalities and his radiographs are shown in Figures 1 and 2.

Case 3. The 24-year-old youngest brother had suffered congenital amputation above the left elbow. He was of short stature but had no musculoskeletal complaints until at the age of 15 years he had collapsed with unexplained pain in the right hip which had necessitated three weeks of traction in hospital. After this he had only minor episodic catching sensations until the age of 23 when he noted more frequent catching in the right hip. During the last game of the soccer season he had an acute onset of severe pain which again necessitated traction in hospital. This pain did not resolve despite prolonged bed rest in hospital. When seen by the senior author he was using crutches, being unable to take weight on his right leg. He also complained of progressive non-radiating low back pain for three months, associated with movement but unaffected by coughing or sneezing.

He was 159.0 cm tall (vertex to pubis 71 cm; pubis to floor 88 cm) and was in considerable pain. His lumbar spine had reduced flexion and extension but spinal rotation did not exacerbate the pain, though lateral flexion did. Movements of the left hip were normal but on the right, although there was no fixed flexion deformity, only 20° to 30° of flexion could be tolerated. No rotation was possible at the right hip, and only slight and painful abduction and adduction. There was 1.5 cm...
Case 2. Figure 1 – Radiograph of the pelvis showing deep acetabula and short femoral necks. Figure 2 – Lateral radiograph of the spine showing typical changes in the vertebral bodies.

Case 3. Figure 3 – Anteroposterior radiograph of the pelvis showing cystic changes in the right femoral head. Figure 4 – Lateral radiograph of the lumbar spine showing characteristic changes in the vertebral bodies.
of wasting in the right quadriceps and right calf, with slight weakness of the right gluteal muscles, gastrocnemius and the long toe flexors. There was also a sensory deficit to pinprick in the L2-3 dermatome on the right.

Plain radiographs of the spine and pelvis showed the typical features of spondylodephyseal dysplasia tarda (Figs 3 and 4). A bone scan revealed an enlarged left kidney and right renal agenesis, which was confirmed by an intravenous pyelogram. A myelogram was normal except for slight anterior indentation of the contrast column caused by the shape of the vertebral bodies.

Arthroscopy of the right hip was performed with synovial and bone biopsy and, at the same time, decompression of the femoral head. Culture of the synovial fluid and synovium was negative but the bone biopsy revealed avascular necrosis. This operation failed to relieve his severe pain and one month later a muscle release was performed, with postoperative mobilisation of the hip on a continuous passive movement exerciser. Again there was no significant improvement and about three months after admission a total hip arthroplasty was performed, with satisfactory relief of pain.

**DISCUSSION**

In a recent review of skeletal dysplasias in Great Britain (Wynne-Davies and Gormley 1985) the prevalence of the X-linked form of spondylodephyseal dysplasia tarda (males only) was estimated at 1.7 per million, as opposed to 7.7 per million for the other forms.

The other forms, autosomal recessive and dominant, have received more recent attention. In both these types there is only mild shortening of the trunk. Symptoms are noted in childhood, often between the fourth and tenth years of life, and progressive arthropathy is common, most often involving the proximal interphalangeal joints of the hands. Radiographs of the spine show platyspondyly, with wedging of the vertebral bodies and end-plate irregularities. The thoracic spine is most involved.

In contrast, the X-linked form has more obvious short-trunk dwarfism, with adult heights ranging from 127 cm to 160 cm, averaging 140 cm. Symptoms are rare before the age of 12 or 13 years, and their development before the age of 10 is distinctly unusual. The first symptom in adolescence is usually back pain of a vague pattern, while at a later stage osteoarthrosis of the large proximal joints may develop. The hips are most frequently involved (Langer 1964; Bannerman, Ingall and Mohn 1971; Bailey 1973, pp. 503–17; Rimoin and Hollister 1979), with non-specific abnormalities in childhood becoming diagnostic by the early teens (Jacobson 1939; Maroteaux et al. 1957; Poker, Finby and Archibald 1965; Harper, Jenkins and Laurence 1973).

In the sex-linked form, the most characteristic radiographic changes occur in the lumbar spine. There is flattening of the vertebral bodies with a built-up hump of bone in the central and posterior part of both superior and inferior end-plates and there is no visible bone in the areas of the ring apophyses. The disc spaces themselves show marked narrowing, usually in their posterior portions, with Schmorl's nodes anteriorly giving a fish-mouth appearance. The appearance of disc calcification is not a true one but is caused by part of the vertebral body itself (Langer 1964).

The pelvis is small and markedly disparate with the rib cage in size. The acetabula are deep and the femoral necks are short. Mild degenerative changes occur frequently in the hips and less commonly in the shoulders, but extensive cyst formation may develop in the femoral head. The suggested has been made that joint involvement is milder in the X-linked form than in the other variants, and that arthroplasty is rarely indicated (Connor et al. 1982; Wynne-Davies et al. 1982; Kaibara et al. 1983; Al-Awadi et al. 1984). Bannerman et al. (1971) stated that secondary osteoarthrosis became troublesome in the fifth decade and disabling by the seventh decade.

In the family we report, neither of the parents was involved, nor were the two sisters of the three index cases. A maternal grandfather was of excessively short stature and had severe hip disease. This male-only pattern, with characteristic radiographs, confirms that this family suffers from a classic X-linked form of spondylodephyseal dysplasia tarda.

The three brothers demonstrate the different clinical courses the condition can take. The eldest has slowly progressive premature osteoarthrosis of the hip, and it appears inevitable that he will eventually need an arthroplasty. The middle brother, apart from a short episode of hip pain several years ago, is symptom-free but there are cystic changes within one femoral head. The youngest brother developed severe arthritis of the hip of acute onset which disabled him. Despite a series of other operations, pain continued and total joint replacement was performed at the age of 24 years.

The underlying cause of the condition has not yet been elucidated, all reported laboratory and metabolic studies having been normal (Thompson et al. 1982). In our Case 3 the excised femoral head showed avascular necrosis and it seems likely that the acute onset of his pain was associated with a subchondral fracture. Of our other cases, one had a congenital amputation and absence of one kidney. The other had a skin lesion of a type which has been mentioned in two other cases (Specht 1968; Franford et al. 1982).

Low back pain is common in patients with this disorder but usually responds to conservative management. Secondary osteoarthrosis of the hip, however, is not always mild (Weinfeld, Ross and Sarasohn 1967; Bailey 1973, pp. 503–17) and patients with sex-linked spondylodephyseal dysplasia may need total hip arthroplasty before the age of 40 years.

**REFERENCES**


Jacobsen AW. Hereditary osteochondrodystrophy deformans: family with 20 members affected in 5 generations. JAMA 1939;113:121-4.


