# Spondyloepiphyseal dysplasia tarda

## Report of a family with autosomal dominant transmission

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We present a clinical and radiographic study of a family with the autosomal dominant form of spondyloepiphyseal dysplasia tarda, in some patients associated with hearing deficiency. Although no causal treatment is possible, correct diagnosis is important to avoid unnecessary treatment and for information about prognosis, genetic counseling, and recommendation of future occupation.

Nilsonne (1927) was the first to describe a group of skeletal dysplasias that were later denoted as spondyloepiphyseal dysplasia tarda by Maroteaux et al. (1957). Initially, the heredity was described as X-linked (Jacobsen 1939, Maroteaux et al. 1957); but in later reports, both autosomal dominant and autosomal recessive transmission have been demonstrated (Spranger and Langer 1974, Wynne-Davies et al. 1982, Barber et al. 1984, Al-Awadi et al. 1984). The characteristic radiographic features are generalized platyspondyly and dysplasia of the epiphyses, especially in the femoral head, resulting in premature arthrosis (Langer 1964, Katona et al. 1985, Maroteaux et al. 1957, Barber et al. 1984, Bannermann et al. 1971, Spranger and Langer 1974, Spranger et al. 1974).

On radiographs of the spine, irregular platyspondyly, a posterior hump, and absence of ossification at the upper and lower anterior margins of the bodies are distinctive findings in the X-linked type, whereas there is no posterior hump in the autosomal dominant and recessive types (Langer 1964, Spranger et al. 1974, Barber et al. 1984, Iceton and Horne 1986). The patients have a relatively short trunk and disproportionately long limbs. The symptoms usually begin in childhood, and the condition is not associated with biochemical abnormalities or intellectual impairment. Wynne-Davies et al. (1982) and Al-Awadi et al. (1984) have presented familial studies that indicated that the autosomal recessive form may have a more severe course.

We present a clinical and radiographic study of a family with autosomal dominantly transmitted spondyloepiphyseal tarda.

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## Case reports

### *Case 1*

A 13-year-old white girl was admitted to our hospital with complaints of periodic pains in the hips and knees. She had painful limitation of hip movement and kyphosis of the thoracic spine. Neurologic examination, routine blood tests, and specific tests for rheumatoid arthritis were normal. The girl was hospitalized at the age of 20 because of sudden worsening of the pain in the right hip. The pain subsided with bed rest. At the age of 22, numerous osteocartilaginous loose bodies were removed from both knees. Arthrosis was observed, and histologic examination of the synovial membrane showed chronic inflammation. From the age of 29, the condition of her hips deteriorated, with constant pain worsened by activity, and she required daily use of analgesics and periodic use of crutches. At the age of 33, she had bilateral total hip replacement due to marked arthrosis, and also synovectomy of the left knee due to pain and swelling. At aged 33, the patient was 159 cm long and weighed 63 kg. She had a barrel-shaped chest, a short trunk, and disproportionately long limbs. She was of normal intelligence, but had a perceptive hearing deficiency. Radiographs of the hips at the age of 16 (Figure 1) and radiographs of the knees at the age of 29 (Figure 2) demonstrated progressive and severe arthrotic changes. At the age of 16, radiographs of the spine showed irregular platyspondyly and end-plate irregularities, particularly in the thoracic area (Figure 3). Severe premature arthrosis in the cervical spine was found at the age of 32 (Figure 4).

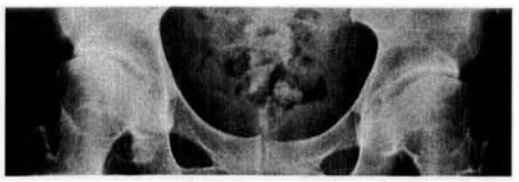

Figure 1

Figure 1. Case 1. Both hips at aged 16 years. Symmetric irregular flattening of the capital femoral epiphyses and arthrosis.

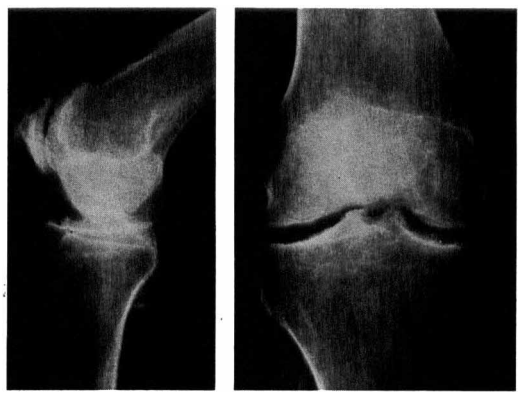

Figure 2

Figure 2. Case 1. The right knee at aged 29 years. Articular margins are irregular and sclerotic with marginal osteophytes.

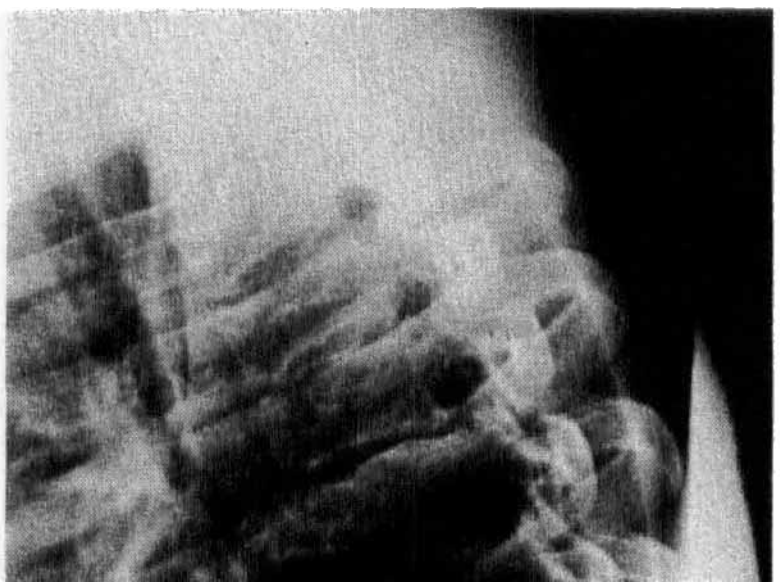

Figure 3

Figure 4

Figure 5

Figure 3. Case 1. Thoracolumbar spine at aged 16 years. Irregular platyspondyly, particularly in the thoracic area, and end-pate irregularities.

Figure 4. Case 1. Cervical spine at aged 32 years. Severe premature arthrosis.

Figure 5. Case 2. Thoracolumbar spine at aged 12 years. Platyspondyly with anterior wedging and irregularity of the vertebral end plates with some disc space narrowing.

### *Case 2*

A 12-year-old boy, the elder son of Case 1, consulted our orthopedic department with year-long complaints of pain in the hips and knees, especially following activity. He had slight reduction in hip movement, a barrel-shaped chest, a short trunk with disproportionately long limbs, and slight scoliosis. His hearing and intelligence were normal. Radiographic examination of both hips, knees, and ankles showed epiphyseal dysplastic changes indistinguishable from multiple epiphyseal dysplasia tarda at a similar age. In the thoracolumbar spine (Figure 5), platyspondyly with anterior wedging and irregularity of the end plates were found.

### *Case 3*

The second son of Case 1 was examined at our orthopedic department shortly after birth because of bowed legs due to an arcuate varus position of both tibiae. At the age of 5 years, he had a valgus position of the knees

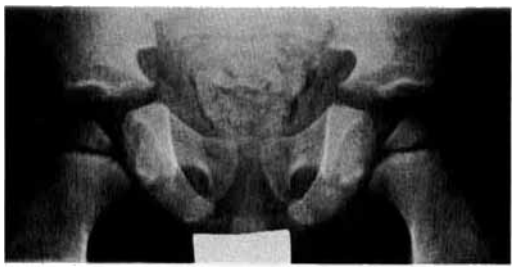

Figure 6. Case 3. Both hips at aged 3 years. Epiphyseal dysplasia and delayed bone maturation.

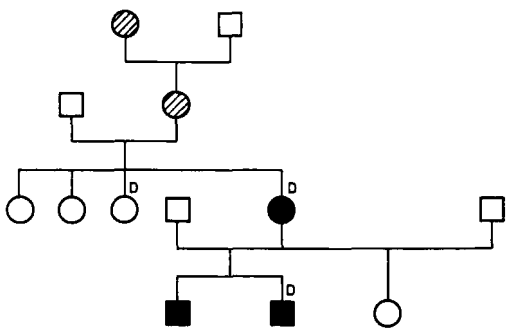


Figure 8. Pedigree of autosomal dominant spondyloepiphyseal dysplasia tarda.

Affected by history
D Hearing deficiency
Affected

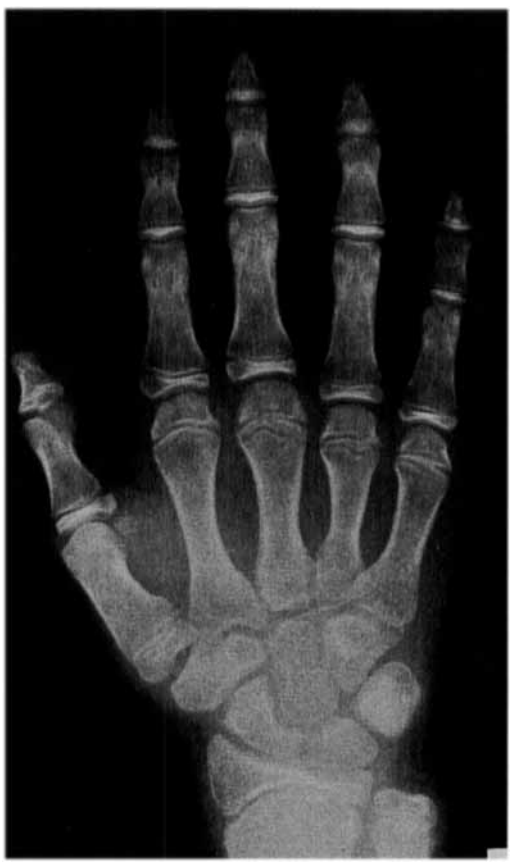

Figure 7. Case 3. The right hand at aged 11 years. Short metacarpals and terminal phalanges.

with a distance between the malleoli of 9 cm; the condition subsided with growth. He was admitted to the hospital at the age of 8 because of acute pain in the left hip, which worsened during abduction and internal rotation. The pain subsided with bed rest. At the time of our study, at the age of 11, he had no complaints. He had a barrel-shaped chest and a short trunk with disproportionately long limbs. He was of normal intelligence and had a perceptive hearing deficiency. At radiographic examination of the skeleton, relatively small iliac bones and epiphyseal dysplastic changes of the hips were seen at the age of 3 (Figure 6). The metacarpals and terminal phalanges were short (Figure 7) as in his elder brother. Platyspondyly and anterior wedging mainly in the thoracic region were found.

A genetic history of the family was obtained (Figure 8). The mother and grandmother of Case 1 had both suffered from crippling pain in the extremities. They were not available for examination. Three older siblings of Case 1 were unaffected, but an older sister had a perceptive hearing disorder. The third child of Case 1, a 6-year-old girl of her second marriage, was examined by us, and no clinical or radiographic signs of disease were found. There was no consanguinity between the parents.

## Discussion

The pedigree is consistent with a dominant inheritance pattern. X-linked dominant inheritance has never been described in spondyloepiphyseal dysplasia. Hearing deficiencies, as in our study, have been reported in association with epiphyseal dysplasia (Bannermann et al. 1971, Pfeiffer et al. 1973, Fraser et al. 1969). The coherence is inconstant, and its significance is unknown. Wynne-Davies and Gormley (1985) estimated the population of spondyloepiphyseal dysplasia tarda in Great Britain to 9.4 per million, whereas Andersen

Jr. et al. (1987) found 7 per million in an area of Denmark.

Multiple epiphyseal dysplasia tarda, which is often transmitted as an autosomal dominant, is neither associated with biochemical abnormalities nor intellectual impairment, and has only little or no spinal involvement (Langer 1964, Spranger et al. 1974, Crossan et al. 1983, Barber et al. 1984). However, several familial studies have been presented with traits of both spondyloepiphyseal and multiple forms (Diamond 1970, Kelly et al. 1977, Tehranzadeh et al. 1978).

To avoid unnecessary treatment, it is important to differentiate spondyloepiphyseal dysplasia tarda from bilateral Perthes´ disease (Crossan et al. 1983, Andersen Jr. et al. 1987). A familial history and radiographic examination of the spine and joints will facilitate the diagnosis. Further, during the active phase of bilateral Perthes´ disease, the involvement of the femoral epiphyses is almost always asymmetric (Crossan et al. 1983).

In hereditary conditions with several modes of transmission, a familial study is necessary to provide adequate genetic counseling. Because of the possibility of premature arthrosis, the patients should be recommended sedentary occupation and informed about the prognosis and possible financial support.

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