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## Spondyloepiphyseal dysplasia tarda in a child with severe and an adult with mild clinical features

Sirs,

Spondyloepiphyseal dysplasia (SED) refers to heritable abnormalities in which the primary skeletal disturbance occurs in the spine and in the epiphyses of the long bones. SED may be categorised in 3 broad groups as follows: the congenital type (SED congenita), with abnormalities recognisable at birth; the pseudoachondroplastic type, which presents in infancy and early childhood; and the tarda type, recognised later in childhood (1). Two cases of spondyloepiphyseal dysplasia tarda (SEDt), one a child with severe and one an adult with mild clinical features, are presented here. The radiologic progression of the disease and the differential diagnosis are discussed.

The first case was a 12-year-old boy referred with progressive dorsal kyphosis, joint deformities, leg pain with activity, and gait

disturbance. His physical development was normal up to age 8. His parents were first-degree cousins with no family history of a skeletal disease. His posture was hyperlordotic with an increased base width during gait. His dorsal kyphosis, lumbar lordosis and the antero-posterior diameter of the chest were increased. Moderate limitation of motion was present in both shoulders and hips. There were bony deformities in the medial aspects of the elbow joints, knees and ankles. A comparative radiographic examination at age 8 and 12 revealed progression and expansion of the platyspondyly towards the dorsal region with increasing anterior wedging, irregularities, subchondral sclerosis and Schmorl's nodes at the end-plates of the T8-11 vertebrae. The humeral and femoral heads were flattened and irregularities in the proximal epiphyses were noted.

The second case was a 29-year-old male who played soccer as a recreational activity, who was admitted with the complaint of pain in the right hip which became evident on physical activity. On examination he had a relatively short trunk with mild scoliosis and increased dorsal kyphosis. Paravertebral muscle spasm and pain in motion of the lumbar spine were observed, although the spinal mobility was not limited. The range of motion in the hips was limited and painful. The radiographic findings resembled those of the first case with increased degenerative changes in the spine (Fig. 1) and hips.

Spondyloepiphyseal dysplasia tarda, first described by Nilsson in 1927 (2,3), is a developmental skeletal disorder that usually presents clinically late in the first or early in the second decade of life with progressive involvement of the spine and epiphyses. The diagnosis of SEDt is difficult in a sporadic case without a positive family history, but when suspected clinically radiographic findings are usually sufficient for the diagnosis. Hyperostotic new bone formation on the posterior two-thirds of the articular surfaces of the vertebral bodies is usually distinctive for the X-linked type (3, 4). Degenerative spine and hip disease characterize the later stages of the disorder (3). Dysplastic changes may be observed in other major joints (1,3).

Scheuermann's disease, juvenile lumbar osteochondrosis and spinal manifestations of the mucopolysaccharidoses resemble the spinal findings of SEDt (5). A subtype of pauciarticular juvenile chronic arthritis (JCA) with late onset may also affect the hips (6). JCA and ankylosing spondylitis might have been considered in the differential diagnosis of our cases. The results of the extensive laboratory workup were within normal ranges for the two cases.

We believe that case 1 represents a case of SEDt with severe and progressive features, beginning at an early age. The second case was a fairly mild one clinically. There was no back pain despite the striking radiographic findings. The late clinical presentation and the delay in diagnosis (at the age of 28), despite his physically active lifestyle seems unusual.

We would like to emphasize that SEDt should be considered in the differential diagnosis in patients who have complaints of back and hip pain and gait disturbances even in adulthood. After the diagnosis of SEDt, patients should be advised to engage in appropriate exercises and moderate activity and should be informed about the prognosis and genetic aspects. Overtreatment should be avoided.

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## Pachydermodactily as a cause of painful swelling of the knuckles: Successful treatment with intralesional steroids

Sirs,

Pachydermodactily is a fibrosing condition characterized by symmetrical, diffuse swelling of the proximal interphalangeal (PIP) joints of the fingers (1-3). The lesions are usually painless, but may cause pain and stiffness of the PIP joints, which has in



**Fig. 1.** T1 and T2-weighted sagittal magnetic resonance images of the lumbosacral spine in case 2, demonstrating platyspondyly, Schmorl's nodes and irregularity at the end-plates of the lumbar vertebrae with narrowing of the intervertebral disc spaces, emphasizing the findings in the plain radiographs.

# Letters to the Editor

some instances led to confusion with inflammatory arthritis (4-6).

A 36-year-old former cleaner was referred to a rheumatologist by his family doctor, due to a long history of swollen and painful knuckles, with a query whether he had rheumatoid arthritis. The patient's father had had some disease of the tendons of his hands (possibly Dupuytren's contracture), and his brother definitely had Dupuytren's contracture, but none of them had any abnormality of the knuckles. The patient was a heavy smoker, and had always been employed in various kinds of manual labour. He gave a history of 9 years of painful and swollen knuckles, with some morning stiffness. The pain was inflammatory rather than mechanical in nature. He had tried diclofenac and indomethacin as analgetics, but they had limited effect and he had to stop them due to GI side effects.

On examination, the patient had a moderate scoliosis, but was in good general health. There were large and tender knuckle pads over PIP II-V bilaterally. These lesions clearly involved skin structures only, and there was no synovitis of the joints themselves, as judged by joint palpation and provocation. There was a variable degree of contracture of the flexor tendons, indicative of Dupuytren's contracture. Plain X-rays of the hands and wrists, a technetium bone scan, and routine blood tests including ESR and CRP were all normal. Rheumatoid factor and anti-nuclear antibodies were not detected. A skin biopsy showed hyperkeratosis and acanthosis, and considerable thickening of the dermis with increased number of irregular collagen bundles.

The diagnosis thus was pachydermodactily, due to the typical clinical findings, and absence of joint disease. A literature review

in 1998 identified 60 cases of pachydermodactily (or knuckle pads) (3), a distinctive form of digital fibromatosis that is most common in young males, and four times more common in patients with Dupuytren's contracture. It is interesting that the patient had a family history of Dupuytren's contracture, and that he himself had contracture of the flexor tendons. Some cases have been associated with repeated trauma to the hands and knuckles, as in live-chicken hangers (7) or boxers (8), or a compulsive habit of rubbing the fingers (2,3), but others have not (3,6). The patient had been a manual labourer most of his working life, but not engaged in activities involving excessive trauma to his knuckles. The unusual aspect of this case is that the patient was considerably troubled by his painful, swollen knuckles, and was unable to work as a cleaner, particularly as this work involved using his hands a great deal. Most earlier cases (1-3), even those referred to a rheumatologist for suspicion of joint disease (4-6) have been more or less asymptomatic even on long-term follow-up.

The majority of cases of pachydermodactily have given the patients little trouble, and have not required any form of therapy. Topical steroid ointments has been ineffective in several instances (1,9), and there is no generally accepted treatment. In some instances of very large or painful knuckle pads, surgical resection of the fibrotic tissue in the reticular dermis has been tried with success (10), but the current patient was unwilling to undergo surgical therapy. There are also some reports upon record of intralesional corticosteroid injections leading to benefit in patients with pachydermodactily (9,11), with a variable degree of reduction of lesion size. It was decided to

try an intralesional injection of depomedrone into one of the lesions, taking care to infiltrate the tissue properly. On follow-up a month later the knuckle pad was much decreased in size, and less painful to the touch (Fig. 1). All the other lesions were treated in a similar manner, with prompt and sustained benefit. One year later, the patient experienced recurrence of six out of his eight knuckle pads, which were then treated in the same manner, again with swift (within 2 weeks) resolution of the lesions. In addition, physiotherapy involving exercises to increase hand and finger mobility proved valuable, and the tendon contracture decreased notably. This enabled the patient again to seek employment.

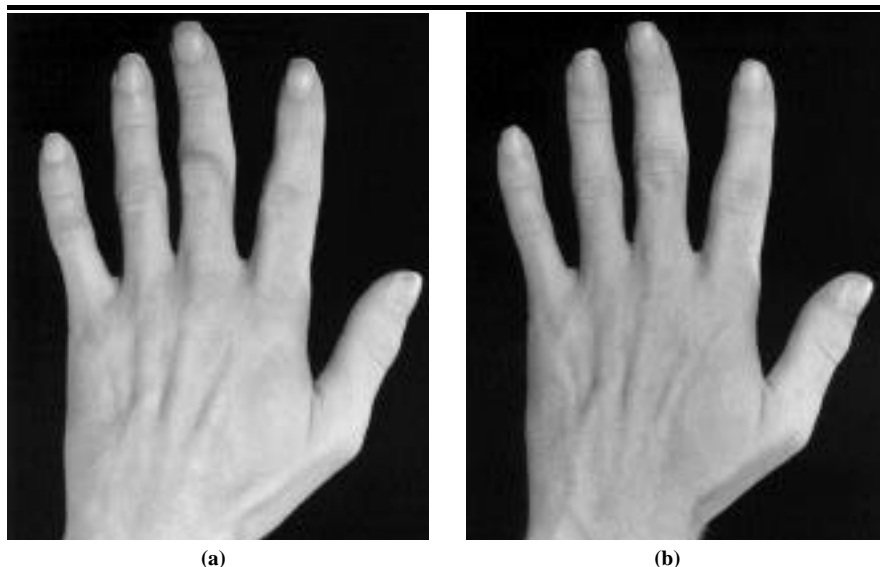
Rheumatologists should be aware of the existence of pachydermodactily, and that it may occasionally be mistaken for proximal interphalangeal joint synovitis. The present case indicates that these knuckle pads are not always painless, and this may complicate the differential diagnosis further. If the clinical appearance of the lesions is atypical, an ultrasound examination or a bone scan may prove useful. Importantly, this case also strongly supports the notion that intralesional steroid injections are a safe and relatively long-lasting way of treating these lesions.

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**Fig. 1.** The patient's left hand four weeks after injection of the knuckle pad on the second finger (a), and four weeks after injection of all four knuckle pads (b).