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Spondyloepiphyseal dysplasia tarda simulating juvenile chronic arthritis

Sirs,

We describe three patients with spondyloepiphyseal dysplasia tarda with progressive arthropathy (SEDT-PA) in two Turkish families who had earlier been mistakenly diagnosed as having juvenile chronic arthritis (JCA).

Case 1, the sister of Case 2, first developed pain and stiffness in her finger joints at the age of 13 years. She underwent corrective surgery for coxa vara at age 15. Moderate effusions developed in both knees when she was 16. Therefore, she was diagnosed to have polyarticular JCA by a paediatrician, with 20 mg prednisolone being added to her treatment. On examination, her height was 146 cm (5th percentile) with an arm span of 158 cm. The fingers showed mild flexion deformities in the proximal interphalangeal (PIP) joints. The wrists showed dorsal swelling and tenderness, and the right and left elbow lacked 15% of the expected range of extension. Both knees showed soft tissue swelling with moderate effusions. Synovial cell count was 800 mm^3.

Case 2, the sister of Case 1, was diagnosed as having genu varus and operated on in another hospital at the age of 5. Involvement of the hand joints, knees and ankles ensued with time. Accordingly, she was diagnosed as having JCA by a paediatrician and was put on non-steroid anti-inflammatory drugs (NSAID). She was referred to our clinic at the age of 15. On examination, she had a short neck and prominent chest. Her height was 143 cm (5th percentile) with an arm span of 158 cm. The fingers showed mild flexion deformities of the PIP joints. The wrists showed dorsal swelling and tenderness, and the right and left elbow lacked 30% of the expected range of extension. She also had a moderate effusion of her knee joints besides synovial hypertrophy. Her synovial cell count was 1300 mm^3.

The parents of case 3 were first degree cousins with a negative family history of precocious osteoarthritis. The patient developed fusiform swelling of the metacarpophalangeal and interphalangeal joints with moderate limitation of extension of the fingers at the age of 8. She was diagnosed as having JCA in another hospital, and put on methotrexate (7.5 mg/ per week), prednisolone (10 mg/day), and NSAID. She discontinued these drugs after 6 months and received only NSAID. On examination, she had a relatively short trunk with scoliosis, increased dorsal kyphosis and barrel shaped chest. Her height was 138 cm (5th percentile) with an arm span of 152 cm. Her wrist showed mild dorsal swelling and both elbows lacked 40% of the expected range of extension.

The laboratory findings of all the 3 cases were negative or within normal ranges. All the 3 cases had generalised flattened vertebral bodies (platyspondyly) of varying degrees at the endplate irregularity (Fig. 1). The phalangeal epiphyses were enlarged along with osteoarthritic changes being present in the interphalangeal joints.

Spondyloepiphyseal dysplasia tarda with progressive arthropathy (SEDT-PA) was described as an inherited skeletal dysplasia with striking progressive impairment of joint mobility, osseous swelling of the joints, best seen in the fingers and short stature in older patients (1, 2). Autosomal recessive, autosomal dominant, and X linked recessive patterns of inheritance in SED have been reported (3). Gedeon *et al*. showed that the X-linked form of spondyloepiphyseal dysplasia tarda (SEDL) is caused by mutations in the SEDL gene (4). El-Shanti *et al.* showed strong evidence for localisation of a gene for SEDT-PA to chromosome 6q (5). Although we could not performed any genetic analysis in our cases, based on clinical, laboratory and radiological findings, our cases were concluded to bear typical features of SEDT-PA.

Although the first two cases had non-inflammatory synovial fluid and synovial hypertrophy in both knees, we were unable to support the presence of an inflammatory condition in our cases. Previous reports also emphasised that SEDT-PA may present with soft tissue swelling in association with effusions (6). Effusions are usually reported to be non-inflammatory unless calcium pyrophosphate dihydrate crystals are present (1). We were unable to show any crystal in synovial fluid in our cases. The disorder is more frequent in Arabic countries,the reason being the large family sizes and high consanguinity rate (7). There have been several reports of cases with SEDT and SEDT-PA in Turkey up to the present time (8, 9), even though we are unsure of its prevalence yet. Still, because consanguinity rate is also rather high in our country (1-47%) (10), prevalence of SEDT-PA might actually be more common than it is thought in Turkey. So, we could expect more cases of SEDT-PA provided that atypical rheumatoid arthritis cases are fully reviewed and more precise diagnoses are made.

SEDT-PA might be mistaken for JCA, which could result in overtreatment with immunosuppressive drugs. Therefore, we suggest that SEDT-PA also be considered for the differential diagnosis of JCA particularly in countries with high consanguinity rate.

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