An eleven-year-old female Turkish patient with progressive pseudorheumatoid dysplasia mimicking juvenile idiopathic arthritis

Sirs.

Progressive pseudorheumatoid dysplasia (PPD) is a rare arthropathy of childhood. It is also known as spondyloepiphyseal dysplasia tarda with progressive arthropathy (SEDT-PA). It is a progressive, noninflammatory connective tissue disorder with autosomal recessive inheritance and has a more crippling course than spondyloepiphyseal dysplasia tarda (SEDT), which is X-linked. The primary site of involvement is the articular cartilage. The first symptoms generally begin between the ages of 3 and 8 years, with progressive joint stiffness that first affects the hips. Further evolution to joint contractures is observed. An initial diagnosis of juvenile idiopathic arthritis (JIA) is often made with clinical symptoms of morning stiffness and increasing stiffness of the limb joints and spine; therefore, it is also known as progressive pseudorheumatoid arthritis of childhood (PPAC).

An 11-year-old Turkish girl was admitted to our hospital with complaints of progressive joint stiffness. She was the first child of third degree consanguineous Turkish parents, who originally came from Syria. Her past and family history were unremarkable. Her first symptoms began as stiffness of the hips at the age of 3. She was diagnosed with bilateral Legg-Calvé-Perthes disease. She suffered progressive stiffness of all joints and fingers. At the age of 7 she was diagnosed as seronegative polyarticular JLA in a different medical center. Sulfasalazine and ibuprofen therapy was instituted for 4 years. She continued to suffer progressive joint involvement despite physical and medical therapy. At the age of 11 she was admitted to our hospital. Clinical examination revealed a normal facial appearance, prominent kyphoscoliosis, pectus excavatum and X-bain deformity of the legs. The mobility of the spine, elbows, wrists, fingers, bilateral hip joints, knees and ankles were reduced. The hand fingers had flexion contractures with marked deformity of the proximal interphalangeal joints.

Hematological and immunological investigations showed no signs of a rheumatic disease. Radiologic examinations revealed S-shaped kyphoscoliosis, flattened vertebrae (platspondyly) and anterior end plate abnormalities with defective ossification (Fig. 1). Osteoarthritides and irregularity in the articular surfaces of all the large joints were noted. There were no signs of bone erosions. The metacarpophalangeal and proximal interphalangeal joints of the hands were markedly enlarged, with irregular epiplyses and enlarged metaphyses. A diagnosis of PPD was made based on the clinical and specific X-ray findings (1).

PPD was first described by Wynee-Davies et al. (2) and Spranger et al. (3-5) independently. The disorder is more frequent in the Middle East and Gulf States and is common among the Arab population (6). Al-Awadi et al. (7) described an Arab family from Jordan with 8 affected members in 5 sibships and included a family from Syria. Ours is the first reported case from Turkey in a patient of Syrian origin, supporting previous evidence that this rare genetic disorder may be more common in this race.

Chondral abnormalities were demonstrated in the iliac crest by Spranger et al. (4,5) with abnormal nesting chondrocytes and defective columnization of the cells in growth zones. Chondrocytes had large Golgi bodies, intracytoplasmic vesicles and variably dilated cisterns of the endoplasmic reticulum. El Shanti et al. mapped the PPD locus to 6q, the mapping made COL10A1, a candidate gene (8). By linkage analysis of 11 patients affected with PPD, Fischer et al. showed localization to interval 6q22 between D6S1594 and D6s432 (9). Hurwitz et al. examined the WISP3 gene, which maps to the same region of 6q, and identified WISP3 mutations in affected individuals, indicating that the gene is essential for normal postnatal growth and cartilage homeostasis (10).

The differential diagnosis between PPD and seronegative polyarticular JIA may indeed be challenging. Radiologic examinations indicating bone dysplasia, platspondyly, spondylosis of the spine, enlarged and irregular epiphyses, a history of consanguinity, normal blood tests, absence of systemic signs of inflammation and lack of response to anti-rheumatoid drugs should raise the possibility of a diagnosis of chondrodysplasia. In this way inappropriate immunosuppressive therapy can be prevented and genetic counselling can be offered.

References