Turkish Journal of Medical Sciences

Turkish Journal

of

Medical Sciences

Spondyloepiphyseal Dysplasia Tarda with Progressive Arthropathy with Delayed Diagnosis

Esra TUĞ[1], Efsun ŞENOCAK[2]

[1] Department of Genetics, İzzet Baysal Faculty of Medicine, Abant İzzet Baysal University, Bolu - TURKEY

[2] Department of Radiology, Faculty of Medicine, Hacettepe University, Ankara - TURKEY

Keywords
Authors

medsci@tubitak.gov.tr

Scientific Journals Home Page

Abstract: Spondyloepiphyseal dysplasia tarda with progressive arthropathy (SEDT-PA) is a rare autosomal recessive hereditary skeletal disease, and mutations in WISP3 are responsible for its onset. WISP3 is essential for maintaining cartilage integrity mainly by regulating the expression of collagen II, and mutations of WISP3 linked to SEDT-PA can compromise this function and lead to cartilage loss, which is frequently misdiagnosed as juvenile idiopathic arthritis. It is characterized by arthralgia, joint contractures, bony swelling of metacarpophalangeal and interphalangeal joints and platyspondyly. Clinical and laboratory signs of joint inflammation such as synovitis, a high erythrocyte sedimentation rate and an elevated C-reactive protein level are usually absent. Although the disease begins early in life (usually between 3 and 8 years of age), the diagnosis may be delayed. In the present case report, we describe a female patient diagnosed with SEDT-PA at the age of 40 years, although she had been exhibiting the typical radiological and clinical features of the disease since the age of 8 years. Genetic disorders like SEDT-PA may also have rheumatological involvement, and thus should be kept in mind in the differential diagnosis of inflammatory joint diseases.

Key Words: Spondyloepiphyseal dysplasia, pseudorheumatoid arthropathy, progressive arthropathy, osteochondrodysplasias

Turk J Med Sci 2008; **38**(1): 83-89.
Full text: pdf
Other articles published in the same issue: Turk J Med Sci,vol.38,iss.1.