CASE REPORT OLGU SUNUMU

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Spondyloepiphyseal Dysplasia Tarda with Progressive Arthropathy: A Rare Cause of Disability

Progresif Artropatili Spondiloepifizyal Displazi Tarda: Nadir Bir Engellilik Nedeni

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ABSTRACT Spondyloepiphyseal dysplasia tarda with progressive arthropathy (SEDT-PA) is a hereditary skeletal dysplasia typically diagnosed in childhood. Joint pain and swelling typically emerge between the ages of 3-6, progressively leading to clinical features such as short stature, kyphoscoliosis, platyspondyly, osteopenia, secondary osteoarthritis, and joint movement restrictions. Also known as progressive pseudorheumatoid dysplasia, SEDT-PA is frequently misdiagnosed as juvenile idiopathic arthritis. Currently, no definitive treatment exists for SEDT-PA; management primarily consists of supportive care, including analgesic medications, physical therapy, exercise, and surgical interventions. In this report, we aim to highlight the case of an 18-year-old patient with SEDT-PA and its associated disability.

Keywords: Arthropathy; disability; spondyloepiphyseal dysplasia

ÖZET Spondiloepifizyal displazi tarda ile birlikte progresif artropati (SEDT-PA) genellikle çocukluk döneminde teşhis edilen kalıtsal bir iskelet displazisidir. Çoğunlukla 3-6 yaş civarında eklem ağrısı ve şişlik ortaya çıkmaya başlar; zamanla kısa boy, kifoskolyoz, platispondili, osteopeni, sekonder osteoartrit, eklem hareket kısıtlılıkları gibi klinik özellikler belirgin hâle gelir. Progresif psödoromatoid displazi olarak da bilinmekte ve sıklıkla juvenil idiopatik artrit ile tanı karmaşasına yol açmaktadır. SEDT-PA'nın şu an için kesin bir tedavisi olmamakla beraber; tedavi yalnızca destekleyici olarak analjezik ilaçlar, fizik tedavi ve rehabilitasyon uygulamaları, egzersiz ve cerrahi müdahalelere dayanmaktadır. Biz bu yazıda 18 yaşında SEDT-PA'lı bir hasta ve oluşturduğu erken yaşta ortaya çıkan engelliliğe dikkat çekmeyi amaçladık.

Anahtar Kelimeler: Artropati; engellilik; spondiloepifizyal displazi

Spondyloepiphyseal dysplasia (SED) is a rare autosomal recessive skeletal dysplasia first described by Wynne-Davies in 1982.[1,2] Abnormal collagen formation primarily affects the growth plates of the spine and extremities. SED congenita is a more severe form of the disease, typically diagnosed in infancy. The characteristic features of SED congenita include a flat face, cleft palate, myopia, and hearing loss.[3] SED tarda with progressive arthropathy (SEDT-PA) is the form typically diagnosed in childhood. It is also referred to as progressive pseudorheumatoid arthropathy of childhood or progressive pseudorheumatoid dysplasia.[4,5] Its estimated incidence in the United Kingdom is reported as 1 in 1,000,000. However, it is believed to be more common in our country, as well as in the Middle East, the Gulf States, and India.[6] Prominent clinical features include short stature, kyphoscoliosis, osteopenia, sec-

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ondary osteoarthritis, and restricted joint mobility.[7] Joint pain and swelling typically begin to appear between the ages of 3-6. The involvement of the joint cartilage leads to bone formation abnormalities, early osteoarthritis, and cartilage degeneration. Symmetric joint involvement is commonly observed, often causing diagnostic confusion with juvenile idiopathic arthritis (JIA). Given the significant differences in diagnosis and treatment, it is crucial to consider SEDT-PA in the differential diagnosis. Misdiagnosis can result in unnecessary rheumatologic tests and ineffective treatments.[8,9]

In this report, we aimed to raise awareness of SEDT-PA, a rare syndrome, and its associated disability.

CASE REPORT

An 18-year-old female patient presented to our physical medicine and rehabilitation outpatient clinic for disability assessment. She was first diagnosed with SEDT-PA at the age of 3 (a genetic analysis had been performed, but the family was unable to obtain the historical report). Additionally, her older brother had also been diagnosed with SED, and he had undergone hip and knee arthroplasty. On the current examination, the patients height was 138 cm, and her weight was 37 kg. Multiple joint deformities were observed during the physical assessment. In the locomotor system examination, cervical extension was 0˚, rotation was 30˚ bilaterally, and flexion was 50˚. The patient was unable to perform active joint movements in the thoracic region due to ankylosis. In the lumbar region, flexion was limited to 50˚, while extension was restricted to 0° (Figure 1, Figure 2).

Hip joint rotations were restricted to 5˚ bilaterally, while hip flexion was limited to 90˚. There was no restriction in knee flexion; however, knee extension was restricted by 40˚ at the end of the range of motion. In both ankles, ankylosis developed in the neutral position.

The patient could ambulate independently on flat surfaces but required single-point support for stair navigation and prolonged walking.

Regarding the upper extremities, no movement limitations were observed in the fingers; however, the Heberden and Bouchard nodes had developed bilaterally. Wrist flexion was measured at 60˚ bilaterally, whereas extension was completely restricted at 0˚. Elbow flexion was limited to 90˚, while extension was restricted by 30˚ at the end of the range of motion. Shoulder movements were restricted at the mid-range of motion bilaterally.

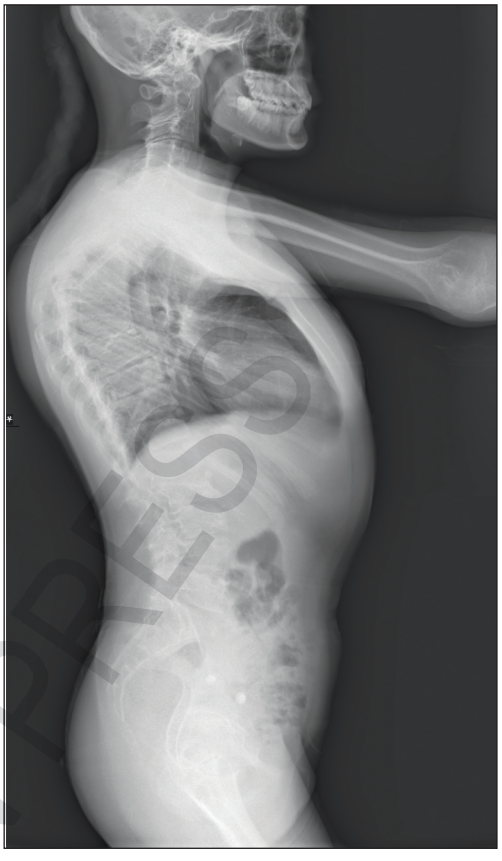

FIGURE 1: Lateral spinal radiograph shows increased kyphosis in the thoracic region, generalized platyspondyly, loss of vertebral body height, and increased anteroposterior diameter.

Upon taking the patient's history, it was noted that she had never previously visited a physical therapy and rehabilitation department and had no history of physiotherapy or exercise intervention. The study was carried out according to the Helsinki declaration. Written and verbal informed consent was obtained from the patient for the case report.

DISCUSSION

SEDT-PA is a rare autosomal recessive hereditary disorder that leads to joint involvement and progressive deformities in childhood.[4] It results from a mu-

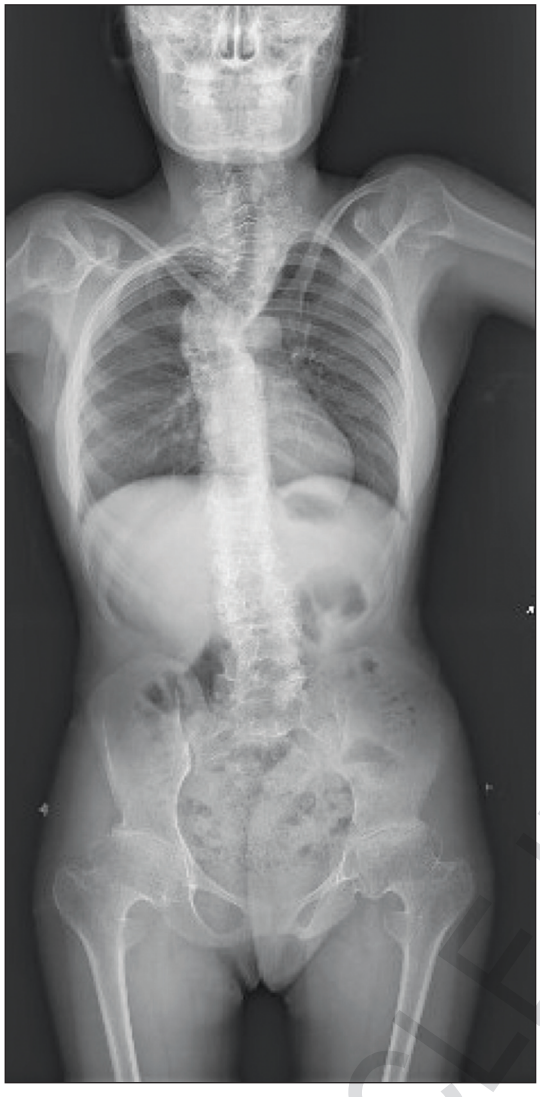

FIGURE 2: Anteroposterior spinal radiograph shows scoliosis in the thoracic region, widespread degenerative changes; bilateral hip joints within the imaging field demonstrate narrowed and irregular joint spaces, subchondral sclerosis, and flattened femoral heads

tation in the WNT1-inducible signaling pathway protein 3 (WISP3) gene on chromosome 6q22. WISP3 belongs to a gene family that encodes growth factors involved in multiple roles within connective tissue, including the regulation of cell proliferation, differentiation, and migration.[10]

The characteristic features of the disease include involvement of the spine and bone epiphyses, osteopenia, short adult stature (below the 3rd percentile), thoracic kyphoscoliosis, and progressive joint involvement. This typically manifests as pain, swelling, stiffness, and contracture development in the hips, metacarpophalangeal joints, proximal and distal interphalangeal joints, wrists, elbows, knees, shoulders, and ankle joints.[9,10] In infancy, height is usually within the normal range. However, as the disease progresses, skeletal changes in the spine can lead to spinal canal stenosis and spinal cord compression, resulting in the emergence of neurological symptoms.[6,8]

Conditions considered in the differential diagnosis include Scheuermann's disease, mucopolysaccharidosis, myopathies, other dysplastic syndromes, hypothyroidism, and JIA.[6,9] Difficulty in walking, muscle weakness, joint swelling-particularly in the small joints of the hands-knee deformities, and contractures are common presenting symptoms in affected patients.[9,10] Patients may present in early childhood with progressive stiffness and flexion contractures in the bilateral interphalangeal joints, accompanied by metaphyseal bone overgrowth in the metacarpals and phalanges. Due to these findings, the condition can mimic polyarticular JIA, leading to misdiagnosis. However, in these cases, inflammatory serum markers were not elevated, and tests for rheumatoid factor, antinuclear antibodies, and anti-citrullinated protein antibodies were negative. In addition, anti-rheumatic medications do not provide therapeutic benefit. Radiological examinations are expected to reveal dysplastic changes, epimetaphyseal widening, and platyspondyly rather than arthritic changes or destructive erosion. A timely and accurate diagnosis is crucial to avoid unnecessary immunosuppressive treatments.[10,11]

Degenerative changes, such as osteophyte formation and periarticular calcifications, can be observed in the hands, shoulders, knees, feet, and elbows of adolescent and adult patients. These findings tend to appear at a younger age in individuals with SEDT-PA compared with those with non-genetic osteoarthritis. In adulthood, radiographic findings become nonspecific, and secondary changes associated with aging can further complicate the diagnosis.[12,13] It has been reported that the diagnosis can be delayed for many years. However, the time between symptom onset and diagnosis has been found to be shorter in our country. This has been attributed to the greater awareness of the disease and the higher

prevalence of consanguineous marriages, which increases the suspicion of genetic disorders.[5,6]

There is currently no definitive treatment for SEDT-PA; management is primarily supportive and includes pain-relieving medications, physical therapy, and surgical interventions. In the literature, there are cases of SED patients who have experienced symptomatic and partial functional relief through physical therapy and exercise programs.[6,14] Bal et al. reported that in four patients diagnosed with SEDT, physical therapy and exercise interventions provided symptomatic relief for several months and reduced the need for analgesics.[7]

SEDT-PA is a significant genetic disorder that leads to severe disability at an early age. Although preventing the progression of arthropathy may not be possible, appropriate physical therapy and exercise programs can help reduce disability and improve the patient's quality of life. Therefore, we believe that evaluating these patients for physical medicine, rehabilitation interventions, and exercise programs could provide additional benefits.

Source of Finance

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Conflict of Interest

No conflicts of interest between the authors and / or family members of the scientific and medical committee members or members of the potential conflicts of interest, counseling, expertise, working conditions, share holding and similar situations in any firm.

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