Interesting Image

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^{99m}Tc-MDP Bone Scintigraphy in a Case of X-Linked Spondyloepiphyseal Dysplasia Tarda

X'e Bağlı Spondiloepifizyal Displazi Tarda Olgusunda ^{99m}Tc-MDP Kemik Sintigrafisi

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Abstract

Spondyloepiphyseal dysplasia tarda (SEDT) is a rare X-linked skeletal disorder affecting the spine and long bones, leading to short stature, spinal deformities, and joint stiffness. It is caused by genetic mutations, and primarily affects males. Diagnosis is confirmed by imaging and genetic testing. We report the case of a 33 years old patient with a history of X-linked SEDT, who presented with pain in the lower limbs and pelvis, accompanied by limited mobility. Bone scan with ^{99m}Tc-methylene diphosphonate (MDP) revealed a polyostotic SEDT involvement, periarticular ossifications, and bony bridges in the active phase. The authors highlight the role of bone scan with ^{99m}Tc-MDP in diagnosing this rare disease.

Keywords: Bone scan, ^{99m}Tc-MDP, spondyloepiphyseal dysplasia tarda, polyostotic involvement

Öz

Spondiloepifizyal displazi tarda (SEDT), omurga ve uzun kemikleri etkileyen, kısa boy, omurga deformiteleri ve eklem sertliğine yol açan nadir bir X'e bağlı geçiş gösteren iskelet hastalığıdır. Genetik mutasyonlardan kaynaklanır ve öncelikli olarak erkekleri etkiler. Tanı, görüntüleme ve genetik testlerle doğrulanır. Alt ekstremitelerde ve pelviste ağrı ve hareket kısıtlılığı ile başvuran, X'e bağlı SEDT öyküsü olan 33 yaşında bir hastayı bildiriyoruz. ^{99m}Tc-metilen difosfonat (MDP) ile yapılan kemik taraması, poliostotik SEDT tutulumunu, periartiküler ossifikasyonları ve aktif fazda kemik köprüleri gösterdi. Yazarlar, bu nadir hastalığın tanısında ^{99m}Tc-MDP ile yapılan kemik taramasının rolünü vurgulamaktadır.

Anahtar kelimeler: Kemik taraması, ^{99m}Tc-MDP, spondiloepifizyal displazi tarda, poliostotik tutulum

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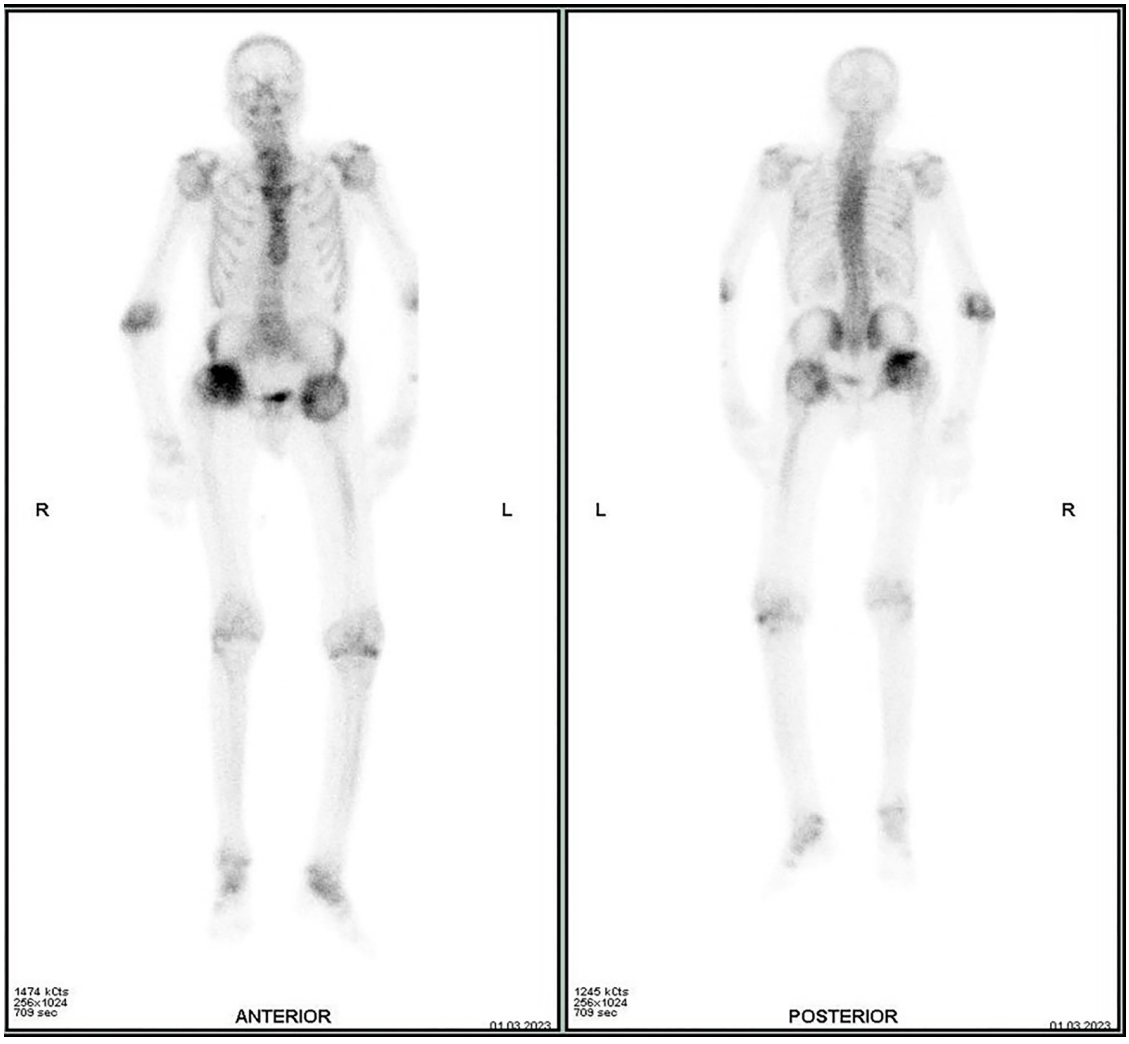

Figure 1. A 33-year-old patient with a known history of X-linked spondyloepiphyseal dysplasia tarda (SEDT) is presenting with lower limb and pelvic pain, accompanied by limited mobility. Standard radiographs showed hump-shaped deformities in the central portion of the vertebrae, narrowed intervertebral disc spaces, and moderate epiphyseal dysplasia of the long bones. These findings were associated with bony bridges in the hips and knees.

A bone scan with ^{99m}Tc-methylene diphosphonate ^{99m}Tc-(MDP), performed 3 hours after the intravenous injection of 666 MBq (18 mCi) of ^{99m}Tc-MDP, revealed heterogeneous and abnormally increased activity with deformities of the femoral heads, elbows, knees, and ankles, a short spine, and marked thoracolumbar scoliosis, consistent with polyostotic SEDT involvement. Focal and linear areas of increased uptake were seen in the knees and hips, corresponding to periarticular ossifications and bony bridges in the active phase (Figure 1).

SED is a subgroup of bone dysplasias that affects the spine and the epiphyses of long bones. It includes three major forms: congenital SED, late-onset (tarda) SED, and SED associated with progressive arthropathy (1). More recently, at least four types of SEDT, some with autosomal recessive inheritance and others with autosomal dominant inheritance, have also been described (2). The classical form of SEDT, as illustrated in this case, is a genetic disorder inherited in an X-linked recessive pattern, manifesting exclusively in males. It results from mutations in the *SEDL* gene located on Xp22.12-p22.31 and manifests around puberty with back pain, a short stature and a short trunk, while the extremities and face remain unaffected. Early onset degenerative joint disease, particularly affecting the spine and hips, usually develops in early adulthood. Radiologic abnormalities of the vertebral bodies include platyspondyly and a central hump (1,3). Bone scan is rarely indicated in SEDT; however, the skeletal involvement observed in this condition is unusual, requiring a thorough understanding of the distribution pattern of bone abnormalities for accurate interpretation and to avoid confusion with other entities, thereby ensuring appropriate management (4).

Ethics

Informed Consent: Informed consent was obtained from the patient.

Footnotes

Authorship Contributions

Surgical and Medical Practices: I.Z., Concept: I.Z., S.O.N., M.A., A.D., Data Collection or Processing: I.Z., S.O.N., A.D., Analysis or Interpretation: I.Z., M.A., Literature Search: I.Z., S.O.N., M.A., Y.B., O.A.S., A.D., Writing: I.Z., S.O.N., A.D.

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