A Case of X-linked Recessive Spondyloepiphyseal Dysplasia Tarda

X'e Bağlı Resesif Spondiloepifizyal Displazi Tarda Olgusu

X'e Bağlı Resesif Spondiloepifizyal Displazi Tarda / X-linked Recessive Spondyloepiphyseal Dysplasia Tarda

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This case report has been presented at 24th National Congress of Physical Medicine and Rehabilitation (27 - 31 March 2013, Maxx Royal Hotel, Antalya, Turkey).

Özet

X kromozomuna bağlı resesif spondiloepifiziyal displazi tarda (SEDT) çoğunlukla erkekleri etkileyen çok nadir görülen bir hastalık olup boy kısalığı, tipik olarak boyu aşan kulaç uzunluğu ve fıçı göğüs deformitesi ile karakterizedir. Etkilenen hastalarda, vertebral epifiz merkezi tutulur ve epifiz oluşumu gecikir ve zamanla platispondili (yassılaşmış vertebral cisimler) ortaya çıkar. Bunlara ilave olarak, erken dönemde başlayan osteoartrit ve osteoporoz nedeniyle dejeneratif süreçli ve ilerleyici eklem ve sırt ağrıları söz konusu olabilir. Bu hastalarda motor ve bilişsel fonksiyonlar normal bulunur. X'e bağlı resesif SEDT tanısı, halen klinik ve radyolojik özellikler ve soy ağacı analizinin bir arada değerlendirilmesine dayanmaktadır. Moleküler analize ise, özellikle şüpheli durumlarda, sadece doğrulayıcı tanı için ihtiyaç vardır. Bu yazı, X'e bağlı resesif SEDT'nin tanısal bulguları üzerine odaklanan bir olgu sunumudur.

Anahtar Kelimeler

X'e Bağlı Resesif; Displazi Tarda; Platyspondyly; Soy Ağacı

Abstract

X-linked recessive spondyloepiphyseal dysplasia tarda (SEDT) is a very rare disorder that mostly affects males and is characterized by short stature, arm span typically exceeding height, and barrel chest deformity. In affected patients, the center of the vertebral epiphysis is involved, the formation of the epiphysis is delayed, and platyspondyly (flattened vertebral bodies) occurs over time. In addition, progressive joint and back pain with degenerative process due to early onset of osteoarthritis and osteoporosis may ensue. Motor and cognitive functions are normal in such patients. The diagnosis of X-linked recessive SEDT is still based on a combination of clinical and radiographic features, and pedigree analysis. Molecular analysis is only needed for the confirmatory diagnosis, especially in suspected cases. This article is a case report focusing on the diagnostic findings of X-linked recessive SEDT.

Keywords

X-Linked Recessive; Dysplasia Tarda; Platyspondyly; Pedigree

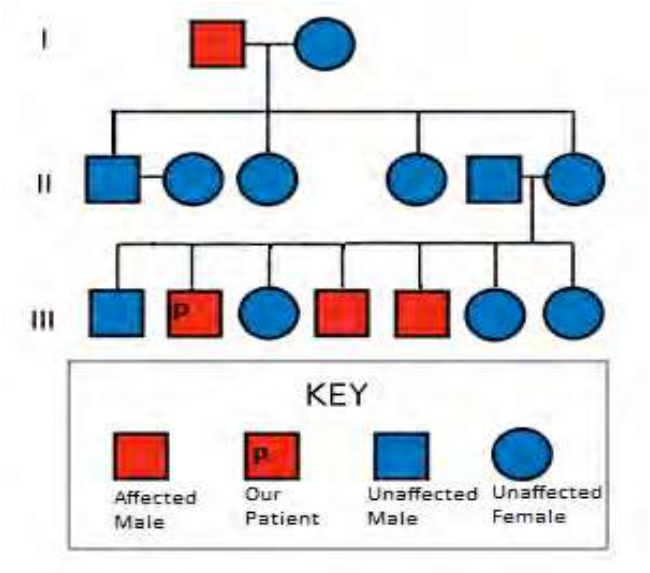
DOI: 10.4328/JCAM.1944 Received: 22.06.2013 Accepted: 04.07.2013 Printed: 01.09.2013 J Clin Anal Med 2013;4(suppl 1): 27-9
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Introduction

X-linked recessive spondyloepiphyseal dysplasia tarda (SEDT) is a very rare hereditary disorder that mostly affects males, and is characterized by short stature, arm span typically exceeding height, and barrel chest deformity [1,2]. At birth, the affected patients have normal lengths and body proportions [2]. In affected patients, the centre of the vertebral epiphysis is involved, the formation of epiphysis is delayed, and platyspondyly (flattened vertebral bodies) and hump-shaped central portions of the vertebral bodies, which are the typical diagnostic radiographic features of X-linked SEDT, occur over time [2,3]. In addition, progressive joint and back pain with degenerative process due to early onset of osteoarthritis and osteoporosis may ensue [4]. Motor and cognitive functions are normal in such patients [2]. This article is a case presentation focusing on the diagnostic findings of X-linked recessive SEDT.

Case Report

A 36-year-old male patient was admitted to our outpatient clinic with low back pain caused by walking for about two years. He had a disproportionately short stature, arm span exceeding trunk, and barrel chest. In his physical examination, his height and arm span were 137.7 cm and 149.4 cm, respectively. A three-generation pedigree analysis revealed an X-linked recessive pattern of inheritance because his grandfather (his mother's father) was affected while his parents and sisters were unaffected (Pedigree 1). His cervical, lumbar and dorsal spine X-rays showed lumbar scoliosis and loss of height in the vertebral bodies at all levels of the spine (Figures 1,2,3). Spinal magnetic resonance imaging (MRI) showed platyspondyly, and irregular and narrow inter-vertebral spaces at all levels (Figures 1,2,3). Bone density was measured by dual X-ray absorptiometry, and osteopenia was determined at the lumbar spine (L2-L4, BMD: 0.855 gr/cm2; T score: -2.12). His routine laboratory tests were found to be in the normal range. In the light of the present findings, the patient was diagnosed with X-linked recessive SEDT, and recommended calcium and vitamin D supplementation, swimming, and dorso lumbar corset.



Pedigree 1. The patient's three-generation pedigree analysis

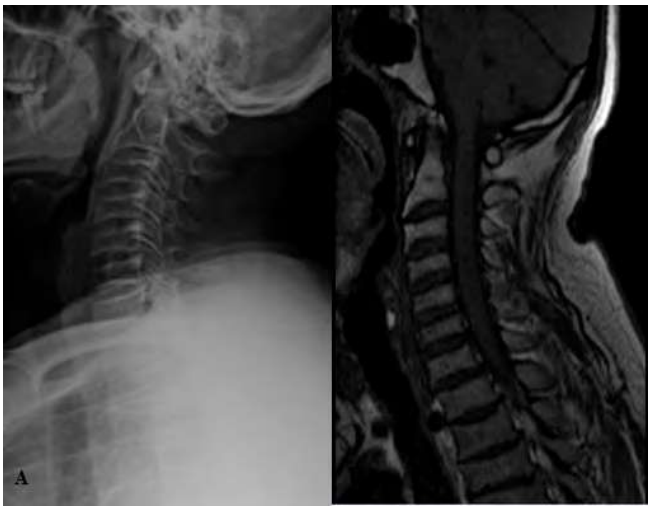


Figure 1. X-ray of the cervical spine in lateral projection (A), T1 weighted sagittal cervical spine MRI (B).

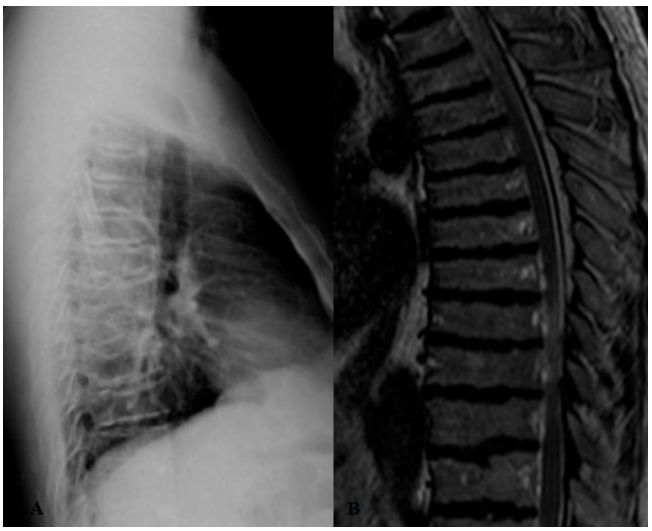


Figure 2. X-ray of the dorsal spine in lateral projection (A), T1 weighted sagittal dorsal spine MRI (B).



Figure 3. X-ray of the lumbar spine in anterior-posterior projection (A) T1 weighted sagittal lumbar spine MRI (B).

Discussion

Spondyloepiphyseal dysplasia comprises a rare group of skeletal dysplasias, and it is classified into congenita and tarda forms according to the age of onset and clinical severity [2,4]. In SEDT, autosomal dominant, autosomal recessive, and X-linked

recessive patterns of inheritance have been reported [2,3]. The diagnosis of X-linked recessive SEDT is still based on a combination of clinical and radiographical features, and pedigree analysis with absence of male to male transmission of the phenotype. Molecular analysis is only needed for the confirmatory diagnosis, especially in suspected cases [2].

X-linked recessive SEDT is usually diagnosed in early adolescence, but in this period radiographic findings may be confused with other diseases including mild Morquio's disease, multiple epiphyseal dysplasias with vertebral changes, and ado¬lescent kyphosis [2]. Therefore, the diagnosis may be delayed. Given that our patient was 36 years old, had normal laboratory values, and an X-linked recessive pedigree, and typical physical and radiological manifestations, he was diagnosed with X-linked recessive SEDT. Thus, the patient was informed about the disease and treatment options, and he was not referred to a tertiary center for molecular analysis.

Patients with X-linked recessive SEDT usually present with low back pain and short stature [4,5]. In addition, these patients are prone to early onset of osteoarthritis and osteoporosis, and spine curvature disorders such as scoliosis and/or kyphosis [2,4,6]. However, these symptoms and findings may appear in the course of several diseases including metabolic, rheumatic, and genetic disorders. Therefore, a differential diagnosis should be carefully done in all patients with nonspecific symptoms without typical characteristic features of X-linked SEDT, which are radiographic findings including platyspondyly and hump-shaped central portions of the vertebral bodies and pedigree analysis showing an X-linked recessive mode of inheritance.

In conclusion, X-linked recessive SEDT should be kept in mind in low back pain patients with short stature to eliminate unnecessary tests and procedures. Knowledge of the typical diagnostic features described above is required to yield a quick and definitive diagnosis.

Competing interests

The authors declare that they have no competing interests.

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How to cite this article:
Alpaycı M, Delen V. A Case of X-linked Recessive Spondyloepiphyseal Dysplasia Tarda. J Clin Anal Med 2013;4(suppl 1): 27-9.