Camurati-Engelmann Disease: A Late And Rare Sporadic Case With Vertebral And Pelvic Involvement

Camurati-Engelmann Hastalığı: Vertebral Ve Pelvik Tutulumla Seyreden Geç Ve Nadir Sporadik Olgu

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Abstract

Camurati-Engelmann disease (CED), or progressive diaphyseal dysplasia, is a rare, autosomal dominantly inherited bone disease. Progressive expansion and sclerosis predominantly affecting the diaphyses of the long bones are the characteristic hallmarks of this disease. The vertebral and thoracic-cage involvement is seen only in the most severe cases. This case is of interest because of its markedly involvement of vertebral bodies, ribs and pelvis at the same time, severe form and sporadic presentation. Also we were not able to find a report such a kind of diffuse involvement in the literature.

Key Words: Camurati-Engelmann disease, Bone dysplasia, Hyperostosis.

Özet

Camurati-Engelmann hastalığı ya da ilerleyici diafizyel displazi, nadir, otozomal dominant geçişli bir kemik hastalığıdır. Özellikle uzun kemiklerin diafizlerini etkileyen ilerleyici genişleme ve skleroz bu hastalığın karakteristik özelliğidir. Vertebra ve toraks kafesi tutulumu sadece ciddi vakalarda görülmektedir. Direk grafi ve BT incelemelerinde, bilateral femur ve tibia diafizlerinde, vertebralarda, pelvis ve kostalarda simetrik genişleme ve sklerozis izlendi. Lomber BT incelemelerinde bu hastalık için tipik olan özellikle arka elemanlardaki tutulum dikkat çekici idi. Vertebra korpuslarının, kostaların ve pelvis kemiklerinin eş zamanlı ve belirgin tutulumu ile sporadik oluşu bu olguyu dikkat çekici kılmaktadır. Literatürde bu kadar diffüz tutulumla karakterize olguya rastlamadık.

Anahtar Kelimeler: Camurati-Engelmann hastalığı, Kemik displazisi, Hiperostozis.

Introduction

Camurati-Engelmann disease (CED) or progressive diaphyseal dysplasia (PDD) is a rare autosomal dominantly inherited bone disease characterized by progressive endosteal and perosteal bone formation in the diaphyses of the long bones. This results in cortical thickening, narrowing of the medullary cavity and a sclerotic and expanded diaphyseal segment. The metaphyses can be affected as well, but the epiphyses are typically spared. Axial skeleton is less affected. There may be sclerosis of the posterior elements of the spine and some sclerosis of the pelvis. Leg pain, easy fatigability, muscle weakness and waddling gait are the most frequent symptoms. Radiographic changes are diagnostic for this disease. We reported the late sporadic form with a rare vertebral, costal and pelvic involvement at the same time.

Case Report

A 52-year-old woman with a chronic history of bilateral leg and back pain, easy fatigability, muscular weakness and bone tenderness was referred our department for radiologic investigation. No members of the patient's family complained about similar symptoms. She had never had any radiographic examination before.

On physical examination she was at 1.58-m-tall with normal body muscle mass. There was no abnormality in neurologic examination. There was no history of trauma, infection or systemic illness. Blood analysis and total urine tests were normal.

Radiographs showed a bilateral symmetrical diaphyseal enlargement and sclerosis of the tibia, femora, vertebrae, pelvis and ribs.

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Diaphyseal and metaphyseal parts of long tubular bones were affected without epiphyseal involvement (Fig. 1). Muscle mass was normal. There was no other cortical thickening such as in clavicles or skull base. Lumbar and pelvis CT showed similar sclerosis pattern of bone (Fig. 2). Cervical and thoracic MRI showed diffuse hypointensity correlated with sclerosis on bodies and posterior elements of vertebrae.

Radiographic findings and clinical history with a normal chest x-ray and the absence of any urine and blood abnormality suggest the diagnosis of PDD.

Discussion

Camurati-Engelmann disease can be familial or sporadic. Familial forms are inherited by autosomal dominant. The existence of sporadic forms is disputed, since other members may not have been screened radiologically. Our patient has a negative family history and shows typical radiological appearances in the femora, tibiae and pelvis. Her children have not presented any typical radiographic changes on their plain roentgenograms.

The onset of symptoms has been described as early as three months and as late as the sixth decade. Muscle atrophy and waddling gait is noted in approximately %50 of patients, pain and muscle weakness occurs in approximately 33% . Fatique, abnormal deep tendon reflexes and bone tenderness are reported less often. Our patient did not have any waddling gait but presented with muscle weakness, leg and back pain and bone tenderness.

There are no abnormality in laboratory findings such as hematological profile, electrolytes, parathyroid hormone, serum calcium and phosphorus, calcitonin, alkaline phosphates, uric acid and thyroid hormones like in our case.

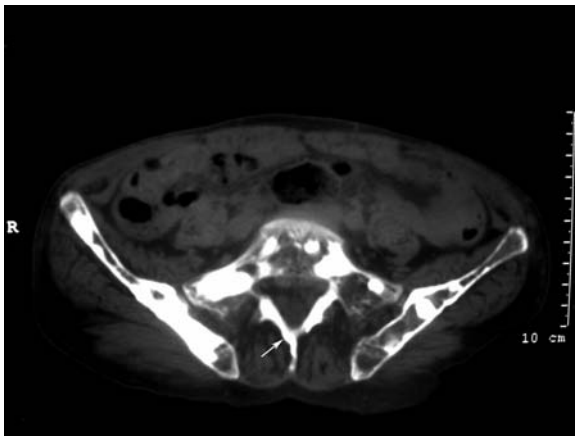

Figure 1. Plain radiograph of the bilateral legs shows symmetrical thickening and enlargement, hyperostosis of the diaphyseal portions of the tibia (white arrows), with metaphyseal undermodeling and vertical striations.

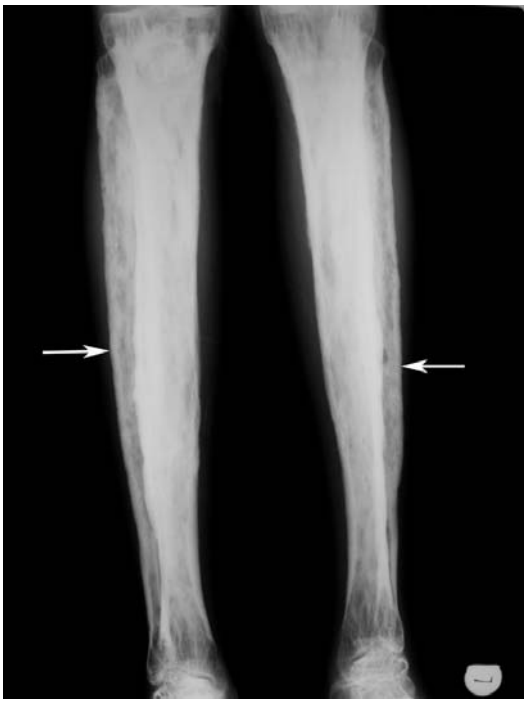

Figure 2. Axial CT scan of pelvis showing diffuse and severe sclerosis of the posterior parts of lumbar vertebrae (white arrow) and iliac bones.

Radiographic abnormalities of this disease are remarkably constant and distinctive. Cortical fusiform thickening of the diaphyses of the long bones is bilateral and symmetrical and usually starts at the diaphyses of the femora and tibiae. Fibulae, humeri, ulnae and radii may be affected. In progress the metaphyses may become affected as well, but the epiphyses are spared. Sclerotic changes at the skull base may be present. In our case there was no involvement in the skull base. According to the literature vertebral (confined to the posterior part of the vertebral body and arches) and thoracic-cage involvement is seen only in the most severe cases, in which practically the whole skeleton is affected. In our case lumbal vertebrae, posterior elements of lumbal vertebrae and costae are affected markedly. The diagnosis of PDD can be made on the basis of roentgenographic findings. Our case is similar to those in the literature in the distribution of bone involvement. But our case is of particular interest because of its severe form and wide involvement presenting with vertebral, pelvic and costal involvement at the same time.

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