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Clinical note

Bone scintigraphy as cornerstone in the diagnosis of Erdheim–Chester disease

F.J. García-Gómez[a,*], T. Cambil-Molina[a], J.J. Ríos-Martín[b], P.A. de la Riva-Pérez[a], C. Calvo-Morón[a], J. Castro-Montaño[a]

[a] Department of Nuclear Medicine, Virgen Macarena University Hospital, Seville, Spain
[b] Department of Pathology, Virgen Macarena University Hospital, Seville, Spain

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ABSTRACT

The Erdheim–Chester disease (ECD) is an extremely rare form of non-Langerhans cell histiocytosis. The main difficulty for its diagnosis lies in the wide variety of non-specific symptoms and signs that can occur in the disease process, leading, therefore, to there being no clear-cut algorithm as a guide for an optimal biopsy to confirm the diagnosis. An 81-year-old male with history of diabetes insipidus was admitted due to non-specific respiratory signs. Imaging techniques revealed osteoblastic lesions in the lumbar spine. Whole-body bone-scintigraphy (BS) was performed, in which lesions involving the axial and appendicular skeleton, with different rates of osteoblastic activity, were observed. This highlighted a symmetrical severely intense uptake in the knees, leading to an accurate biopsy specimen that enabled making the definitive diagnosis. BS is a widely available, safe, and inexpensive technique that shows a characteristic pattern of uptake for ECD, thus its use is highly recommended for screening and guiding biopsy if clinical suspicion exists. Furthermore, when the scintigraphy pattern is incidentally observed, biopsy of increased uptake areas (tibia preferably) is mandatory in order to rule out the disease.

Papel crucial de la gammagrafía ósea en el diagnóstico de la enfermedad de Erdheim–Chester

Palabras clave:
Enfermedad de Erdheim–Chester
Gammagrafía ósea
Algoritmo diagnóstico

RESUMEN

La enfermedad de Erdheim–Chester es una histiocitosis no-Larngerhans extremadamente rara. La dificultad en su diagnóstico se debe a los signos y síntomas inespecíficos que presenta, que conlleva la ausencia de un claro algoritmo diagnóstico. Reportamos el caso de un varón de 81 años con diabetes insipidus en estudio por síntomas respiratorios inespecíficos. Lesiones osteoblásticas en la columna fueron referidas en técnicas radiológicas. Mediante gammagrafía ósea (GO) se observaron lesiones osteoblásticas con diferente actividad metabólica en esqueleto axial y apendicular, destacando una actividad muy elevada y simétrica en rodillas, cuya biopsia permitió el diagnóstico definitivo. La GO es una técnica disponible, segura y barata que muestra un patrón característico, por lo que recomendamos su realización como screening y guía para toma de biopsia. Ante el hallazgo incidental de dicho patrón debería realizarse biopsia.

Introduction

The Erdheim–Chester disease (ECD) is an extremely non familiar entity, consisting of non-Langerhans cell histiocytosis, described at first by Jakob Erdheim and William Chester less than a century ago.[1] Since then fewer than 600 cases have been reported in the literature,[2] being diagnosed more frequently over recent years probably related to the greater effort in diffusion of the disease and improvements in diagnosis techniques.

The main difficulty for diagnosis of ECD lies in the wide variety of nonspecific signs and symptoms that can occur. These signs and symptoms range from bone sclerosis, orbital infiltration with proptosis, diabetes insipidus, cardiac involvement, and pulmonary infiltration. A review of the literature tell us that bone disease is the most prevalent symptom which is usually combined with infiltration of at least one more organ system.[3] This lack of specificity in symptoms and the rarity of the picture causes that there are therefore no clear-cut algorithm to guide us to obtain an optimal biopsy that confirms the diagnosis. Furthermore, optimal biopsy specimen will have important implications for the therapeutic possibilities due to new discoveries as the identification of the activating BRAF-V600 mutation.[4]

* Corresponding author.
E-mail address: javier191185@gmail.com (F.J. García-Gómez).

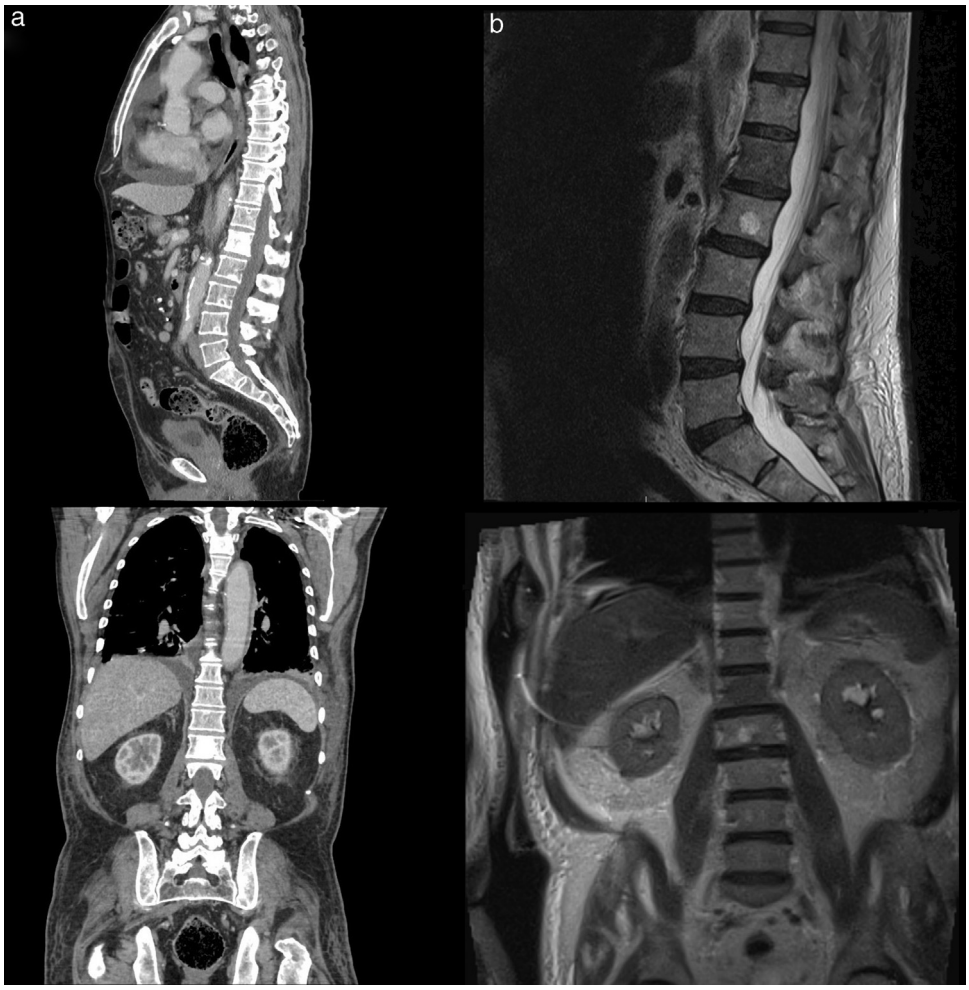

Fig. 1. (a) Thoracoabdominal CT with IV contrast, sagittal and coronal views, revealing blastic lesion in the L1 vertebral body that respects the pedicles, of uncertain significance and consistent with metastatic lesion. (b) MRI of the lumbosacral spine with and without contrast enhancement, sagittal and coronal views, showing a discrete hypointense signal in both T1- and T2-weighted sequences with slight internal heterogeneous enhancement involving the bone marrow of the vertebral body of L1. Despite its nonspecific appearance, it is suggestive of moderately osteoblastic vertebral metastasis.

Whole-body bone scintigraphy produces planar images of the skeleton, including anterior and posterior views of the axial and appendicular skeleton. Also additional spot views (planar images of a selected portion of the skeleton) or SPECT (tomographic image of a portion of the skeleton) are obtained as needed.

Clinical case

We present a new case recently diagnosed in the wake of the findings in bone scan. This is an 81-year-old male patient with history of diabetes insipidus who was admitted because respiratory symptoms (dyspnea and tachypnea). Severe pericardial effusion and bilateral pleural effusion were observed in the initial examination and leading to place a chest tube to drain up to 1800 ml of exudative pleural effusion with no evidence of malignant cells. Both thoracoabdominal CT and spinal MR scans (Fig. 1) were performed and revealed a moderately blastic lesion of uncertain significance in the vertebral body of L1 while pedicles were unaffected, compatible with metastatic bone disease of unknown origin. In order to skeletal mapping and according to their wide availability, a ^{99m}Tc-HDP whole-body scan was performed (Fig. 2) demonstrating severe lesions with increased uptake of the tracer in the axial and appendicular skeleton, located at L1 body, left clavicle, hummers, radius and bilateral costal arches. These lesions presented different intensity of radiotracer uptake. Furthermore, a bilateral severely intense uptake in the diaphysis and metaphysis of both femurs, metaphysis of both tibiae and heel were observed. According to our previous experience,[5,6] we described the images as highly suggestive of ECD and indicating the need for histopathological confirmation from knee biopsy. Bone biopsy was initially reported as absence of neoplastic cells. Because of the high suspicion of ECD, the review of biopsy specimen was requested in order to specifically discard such entity (Fig. 3). Foamy histiocytes positively stained for CD68 marker but S100 and CD1a negative were demonstrated after a thorough examination of samples. BRAF-V600E mutation was also confirmed. These findings allowed confirming the definitive diagnosis of ECD and discarding the neoplastic process of unknown origin which until then was the suspected diagnosis.

Discussion

ECD is a non-Langerhans cell form of histiocytosis characterized by multiorgan xanthomatous infiltration of tissues by CD68-positive, CD1a-/S100-negative foamy histiocytes. Individuals are more frequently affected in their fifth decade and there is a slight male prevalence.[7] The etiology of ECD has not been elucidated. Because of its rarity and diverse presentations, it can be extremely difficult to diagnose. In most cases, patients are not properly diagnosed and managed as polypathological or cancer patients