Case report

The Imaging Findings of Erdheim–Chester Disease: A Multimodality Approach to Diagnosis and Staging

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Abstract

Erdheim–Chester disease (ECD) is a rare, non-Langerhans histiocytic disorder. The most common manifestations consist of polyostotic sclerotic lesions with the majority of cases also demonstrating soft tissue involvement of the sinuses, retroperitoneum, large vessels, heart, lungs, and central nervous system. Nuclear medicine can play an important role in assessing the extent of the disease with bone scintigraphy and fluorodeoxyglucose (FDG)-positron emission tomography (PET). We present the case of a middle-aged female who initially presented with tooth pain. She subsequently underwent imaging including plain film, bone scan, computed tomography (CT), magnetic resonance imaging (MRI), and FDG-PET/CT, which showed characteristic bony involvement. Biopsy results confirmed the diagnosis of ECD.

Keywords: Erdheim–Chester disease (ECD), non-Langerhans histiocytic disorder, sclerotic lesions

Introduction

Erdheim–Chester disease (ECD) is a non-Langerhans cell histiocytic disorder with a highly variable clinical presentation. Almost all patients diagnosed with ECD have some degree of bone involvement (95%) with the majority also demonstrating soft tissue disease.[1] Sites of extraosseous disease include the sinuses, large vessels, retroperitoneum, heart, lungs, central nervous system (CNS), skin, pituitary gland, and orbits.[2]

Osseous lesions are characteristically symmetric, involving the diaphysis and metaphysis of the long bones in nearly all patients with ECD[3] and involvement of the facial bones is frequently seen.[4] Cardiac involvement can manifest as valvular abnormalities or conduction problems[5] and is associated with high morbidity and mortality.[6] CNS symptoms are highly variable with exophthalmos, diabetes insipidus, ataxia, and headache reported.[7]

Imaging findings of ECD can be highly variable. Nuclear imaging frequently plays a role in the assessment of ECD. In particular, bone scintigraphy[8] and fluorodeoxyglucose (FDG)-positron emission tomography (PET)[9] have been shown to be useful. Numerous authors have also examined the use of conventional imaging in the evaluation of ECD but to date, no optimal approach to the imaging of these patients has been proposed.

Case Report

A 59-year-old female complaining of chin numbness and jaw discomfort was seen by her dentist. An

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orthopantomogram demonstrated lucencies in the mandible [Figure 1]. A follow-up computed tomography (CT) was performed and showed multiple lytic lesions [Figure 2], with evidence of osseous erosion and destruction of the mandibular cortex. These findings were considered suspicious for osteomyelitis and correlation with bone scan was suggested.

The bone scan demonstrated increased uptake in the mandible as well as in the right iliac, along with symmetric uptake in the femurs and tibia [Figure 3]. Mild uptake was also present in multiple other locations in the axial and appendicular skeleton. The particular distribution of uptake was described as highly suggestive of ECD.

A skeletal survey was performed following the bone scan [Figure 4]. Subtle sclerotic lesions were noted involving the cervical spine, pelvis, and right proximal femurs. These findings were noted to be much less striking than on the bone scan.

Subsequent whole body magnetic resonance imaging (MRI) redemonstrated the multiple known sclerotic foci in the humeri, femurs, and iliac bones [Figure 5] but did not identify soft tissue involvement. It was concluded that on the basis of these findings, ECD could be neither confirmed nor excluded. CT scans of the thorax, abdomen, and pelvis performed around this time were not contributory.

CT-guided biopsy of the right iliac was performed. Histological analysis revealed clusters of foamy histiocytes within the bone marrow, which were CD68-positive, compatible with ECD.

The patient was subsequently referred for FDG-PET/CT for staging [Figure 6]. The findings on PET correlated closely with those of the bone scan and showed increased FDG uptake in multiple locations in the axial and appendicular skeletons, with prominent, symmetric uptake in the lower extremities (arrows). No soft tissue lesions were identified.

Two years prior to her workup for ECD, the patient had been diagnosed with diabetes insipidus of unknown etiology. Given her diagnosis of ECD, the patient underwent MRI of the head [Figure 7] and was found to have bulky adenohypophysis with associated thickening of the pituitary stalk, in keeping with ECD. In addition, circumferential epidural enhancement centered at C7-T1 was present but demonstrated no appreciable spinal cord compression.

Discussion

ECD is difficult to diagnose on imaging due to its variable manifestations, generally nonspecific findings, and its

Figure 1: Orthopantomogram (panorex) demonstrating multiple lucencies (arrows) in the left mandible

Figure 2: CT of the mandible. There are well-defined lytic lesions with evidence of cortical destruction (arrow). These findings were initially considered suspicious for osteomyelitis

Figure 3: Bone scan demonstrating multiple areas of abnormally increased uptake including the mandible and right iliac as well as symmetric uptake in the femurs and tibia (arrows). Mild uptake was also present in multiple other locations in the axial and appendicular skeletons. This distribution is highly suggestive of Erdheim–Chester disease

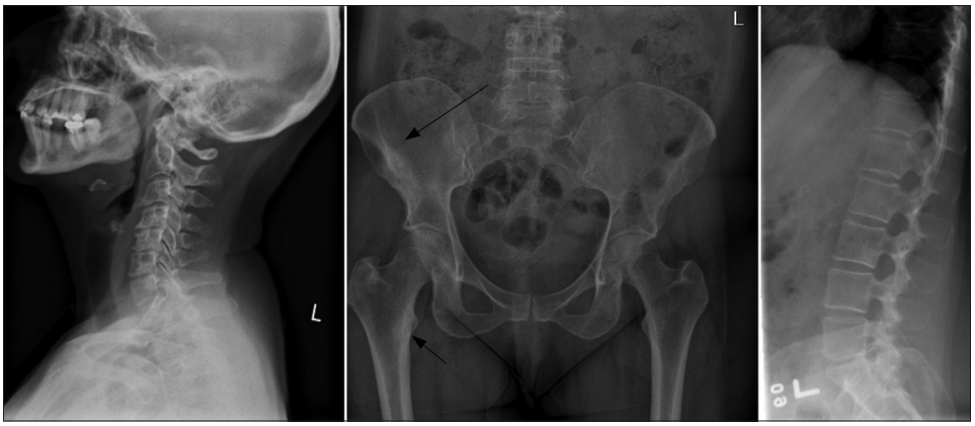

Figure 4: A skeletal survey was performed. Some mild, ill-defined sclerosis was noted in the proximal femora, right iliac (arrow), and cervical spine. The thoracic and lumbar spine were noted to be unremarkable

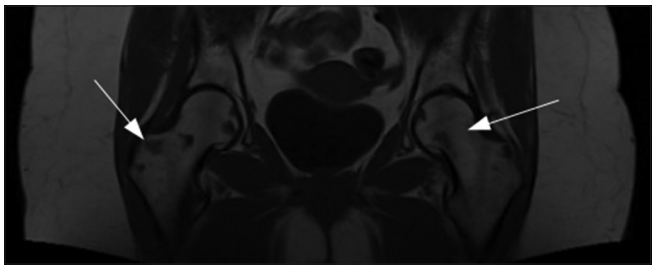

Figure 5: MRI findings demonstrating multiple bilateral and symmetric sclerotic foci in the humeri, femurs, and iliac bones (arrows)

Figure 6: Findings on PET were very similar to those of the bone scan and showed increased FDG uptake in multiple locations in the skeleton. As in the case of the bone scan, the symmetric uptake in the lower extremities (arrows) is strongly suggestive of ECD

rarity. The optimal use of imaging in the diagnosing, staging, and follow-up of this unusual condition has not yet been determined.

Our experience shows that the most useful modalities for the evaluation of ECD are those that best delineate osseous lesions as the strikingly symmetric pattern of activity seen on both the bone scan and PET is highly suggestive of the diagnosis.

In this patient, FDG-PET appeared to be at least as sensitive as bone scan for the evaluation of bony involvement. Furthermore, the ability to evaluate the soft tissues with PET-CT may offer a great advantage in those patients with extraosseous involvement.

Our experience also suggests that whole body MRI could play a role in the staging of ECD. For the evaluation of neural involvement, MRI remains the modality of choice. However, no studies have directly compared the utility of MRI and PET for the diagnosis and staging of this condition.

As we see from this case, CT and radiography have the least to offer in terms of diagnosis and staging due to the subtle findings that often accompany these conditions. As such, we believe that in cases where the diagnosis of ECD is suspected based on plain film or CT findings, further evaluation with bone scan or preferably PET and/or MRI should be performed in order to accurately identify the sites of disease involvement.

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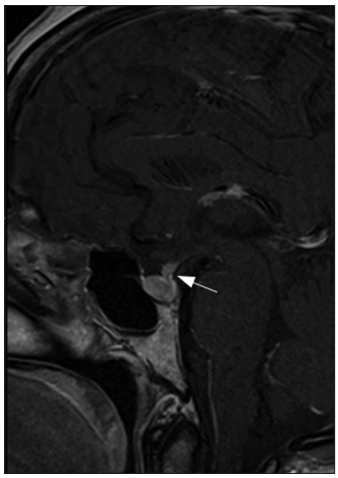

Figure 7: MRI of the head, which demonstrated a bulky adenohypophysis with thickening of the pituitary stalk (arrow), a typical finding in ECD

Conflicts of interest

There are no conflicts of intrest.

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