Erdheim-Chester Disease with Revealing Mandibular Localization: About A Case

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Abstract

Erdheim-Chester disease is a rare systemic Langrehanian histiocytosis. It is diagnosed on the basis of a bundle of clinical, radiological and histological arguments. Its confirmation is based on a precise immunological profile. The bone involvement is almost constant, characterized by osteocondensing lesions more often at the metaphyso-diaphyseal level of the long bones of the lower limbs. We report the case of a 37-year-old female patient with chronic left jugular swelling. The CT scan revealed a mandibular osteolytic lesion including the cortex without extension towards the soft parts. The diagnosis of EDC was established based on the histological aspect of the lesion and confirmed by the immunohistochemical study.

Key words: Erdheim-Chester Disease, Histiocytosis, Mandible, Osteolysis.

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INTRODUCTION

Erdheim-Chester disease is a rare multi-systemic non-Langerhansian histiocytosis, characterized by almost constant bone involvement and a rich and diverse symptomatology making its diagnosis difficult which is essentially based on histopathological analysis and a specific immunohistochemical profile [1]. The bone involvement can be suggestive in its classic form, which is manifested by bilateral and symmetrical osteocondensation of the metaphyso-diaphyseal cancellous bone of the long bones, with predominance in the lower limbs. Nevertheless it can be atypical with lytic lesions taking on a tumor aspect [2]. The mandibular localization is exceptional and rarely described in the literature, which prompted the publication of this clinical-radiological case.

CASE REPORT

We report the case of a 37-year-old woman with a history of chronic bilateral knee and ankle arthralgia and two episodes of self-resolving redness of the eye who consults for a left jugular swelling progressively developing over 6 months.

Clinically, we assess a solid left jugular mass, fixed relative to the deep plane without signs of local inflammation, associated with a bilateral erythematous conjunctivitis. The rest of the clinical examination was of no particularities.

Blood tests requested initially were normal with a leukocyte count at 8500 U / ml, an erythrocyte sedimentation rate at 6 mm / h and a CRP at 1.2 mg / dL.

A craniofacial CT scan was carried out and objectified an osteolytic lesion of the left hemi-mandible affecting the ramus and the horizontal branch with blowing and rupture of the cortex in places (Fig 1 - 3) with no extension to the soft parts surrounding it and deep spaces of the face (fig 2).

Subsequently, a phosphocalcic blood test was requested showing a low level of serum vitamin D (15 ng / ml) with normal values for: alkaline phosphatases (79 U / L), LDH (207 U / L), CK (51 IU / L), Calcemia (95 mg / L) and Phosphoremia (26 mg / L). The tumor markers CA15-3, CA125 and CA 19-9 were negative.

Surgical biopsy of the mandibular lesion with anatomopathological study, showed layers of foaming histiocytes with enlarged nuclei associated with some inflammatory elements such as lymphocytes, plasma cells and neutrophils without specific granuloma with mature bone tissue at the periphery of the lesion composed of thick osseous spans sheltering Haversian...
canals, as well as large hematic layers and heaps of focal atypical histiocytes without mitosis. The complement by immuno-histochemistry was in favor of a non-langerhansian histiocytosis with an anti-CD68 positive, anti-CD1a and anti-protein S-100 negative.

The diagnosis of Erdheim-Chester disease was therefore retained based on the clinical manifestations and histological and immunological confirmation.

In addition, the mandibular location as well as the radiological aspect of the bone involvement are not characteristic of this disease, but can be found in its atypical form.

As part of the assessment of this disease, x-rays of the two lower limbs were requested as well as a bone scan, an abdomino-pelvic ultrasound and a thoraco-abdomino-pelvic CT scan and which did not reveal any other location or visceral involvement.

**DISCUSSION**

Erdheim-Chester disease was first described in 1930 by Viennese pathologist Jakob Erdheim, a pupil of William Chester [1, 3, 4]. It is a non-Langerhansian histiocytosis characterized by specific immunostaining and by the absence of the Birbeck granule found in Langerhansian histiocytosis [5].

The etiology of this disease is largely unknown [6] and can occur at any age, with a predominance in male subjects aged 40-70 years [7].

The clinical manifestations are varied given the ubiquity of histiocytic involvement with non-specific symptoms [8], which makes its positive diagnosis difficult. However, Erdheim-Chester disease can manifest itself most often by the classic triad made...
of aspecific, moderate bone pain with no particular rhythm prevailing in the lower limbs, painless bilateral exophthalmos and by diabetes insipidus which can precede the other symptoms of a few years. Therefore, the disease is suspected before a bundle of clinical and radiological arguments, and can only be confirmed by an anatomopathological, immunohistochemical and molecular study.

Histologically, it is characterized by the presence of foamy histiocytes with a high lipid content or of giant polynucleated cells loaded with lipids (Touton cells) and which can be associated with fibrosis plaques, gigantocellular granulomas or a Polynuclear and lymphocytic inflammatory infiltrate. These histiocytes express CD68 and CD163 but not CD1a with variable expression of the protein S100 and factor XIIIa, which makes it possible - in addition to the absence of the Birbeck granule - to differentiate non-Langerhansian histiocytosis from Erdheim –Chester of Langerhansian histiocytosis [10, 11]. Molecular analysis can reveal the BRAFV600E mutation [12].

Classic bone involvement is characterized on x-ray and on computed tomography by bilateral and symmetrical metaphyseal-diaphyseal bone marrow osteocondensation of the long bones (distal ends of the femurs and proximal of the tibias) with respect for the epiphyses, sub chondral regions, parts soft and extremities. An endosteal cortical reaction is noted giving a dimmed appearance of the cortico-medullary junction. It is sometimes associated with a periosteal reaction in 1/3 of the cases or foci of osteonecrosis by peri-adventitious histiocytic infiltration (micro-angiopathy) [2, 13-15].

In atypical forms, the bone involvement can be a type of pseudo-tumor focal osteolytic lesions with soft tissue damage, or mixed combining osteosclerosis and bone lysis [16-20].

In our patient, osteolytic involvement of the ramus and the horizontal branch of the left hemimandible was the only bone lesion found and indicative of the disease. This localization and its osteolytic nature remain exceptional in Erdheim Chester disease unlike Langerhansian histiocytosis where it is usually found.

MRI confirms the medullary infiltration which appears in hyposignal T1, hypersignal T2, with heterogeneous enhancement involving the diaphyseal-metaphyseal and partially epiphyseal regions, sparing the subchondral regions and thus making it possible to determine the clear boundaries between healthy zones and pathological areas. It can be associated with a periostitis which is manifested by a T2 hypersignal or by a linear peri-cortical enhancement [21, 22].

Bone scintigraphy, SPECT-CT with 18mTc diphosphonates or PET-scanner with 18FDG are sensitive to the early phase of the disease showing bilateral and symmetrical diaphyseal and metaphyseopiphysial hyperfixation [23].

Bilateral and symmetrical peri-renal involvement represents the most frequent extra-osseous location, giving a so-called hairy kidney appearance on CT with sheathing of the renal sinuses and lumbar ureters or mega-calicosis. The radiological distinction with idiopathic retro peritoneal fibrosis (Ormond disease) or with a disease associated with IgG4 is sometimes difficult without a complete histology [24].

Pulmonary involvement may manifest as dyspnea, cough or even respiratory failure with interstitial chest CT syndrome caused by thickening of the interlobular septa, thickening of the scabs, micronodular centerlobular opacities or lymphangitic distribution sometimes associated with effusion plural. The study of the LBA fluid can reveal CD68 + CD1a-spumous histiocytes [25].

Cardiovascular involvement is most often manifested by peri-aortic infiltration in the “aortic mantle” (coated aorta), circumferential, homogeneous, regular, rather peri-adventitious than parietal, which may extend

REFERENCES