

# Gastrointestinal Erdheim-Chester Disease

## Abstract:

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## Abstract

We report a rare case of Erdheim-Chester Disease, a non-Langerhans cell histiocytosis. A 60-year old female presented with a seven-month history of vague abdominal symptoms. A large retroperitoneal mass was detected on computed tomography (CT), but multiple CT-guided biopsy samples were inconclusive. Laparoscopy revealed a mass in the distal ileum, which was resected. Histology and immuno-histochemistry supported a diagnosis of Erdheim-Chester Disease.

## Case Report

A 60-year-old female was referred by her general practitioner with a seven-month history of dyspepsia and an eight-week history of anorexia, abdominal distension and a stone weight loss. Clinical examination was unremarkable. Routine haematological investigations, oesophago-gastro-duodenoscopy and colonoscopy were normal. A computed tomography (CT) scan of the abdomen and pelvis illustrated a large retroperitoneal mass, 5x1.6cmx8.5cm, extending from above the diaphragm to the bifurcation of the aorta. A CT-guided biopsy was performed and histopathology showed a reactive pattern with histiocytes and an associated fibrous reaction of non-caseating granulomata. In the presence of an essentially negative biopsy, a PET-CT was performed which showed increased FDG uptake at the site of the retroperitoneal mass. Thus, a second CT-guided biopsy was performed, out of concern that the initial biopsy was not representative, which again showed similar pathologic findings. Overall, the imaging, however, was suggestive of a malignant lymphoma.

A CT abdomen and pelvis, repeated three weeks later, illustrated doubling of size of the right-sided mesenteric nodes. This strongly suggested presence of an aggressive neoplasm, inconsistent with the two prior pathologic interpretations. Laparoscopy revealed peritoneal seedings suggestive of carcinomatosis, which had inconclusive pathology at frozen section. A mass was detected in the distal ileum with minor obstruction, and was resected with side-to-side (GIA 80) anastomosis. Histological analysis of the resected tissue showed a diffuse proliferation composed of large atypical cells with abundant pink cytoplasm, vesicular chromatin and rare mitoses. Scattered Touton-like giant cells were identified. Immunohistochemical studies confirmed a histiocytic neoplasm, (CD68 positive, CD163 positive, Factor XIII positive, CD1a negative). These combined features were most suggestive of Erdheim Chester disease. Our patient is currently undergoing CHOP (cyclophosphamide, doxorubicin, vincristine, prednisolone) chemotherapy.

## Discussion

Histiocytic neoplasms are derived from histiocytes or macrophages and are rare tumours. Since many of these tumours were poorly recognized prior to the widespread use of immunohistochemistry, the incidence is uncertain. Histiocytic neoplasms include histiocytic sarcoma, Langerhans cell histiocytosis Langerhans cell sarcoma and disseminated juvenile xanthogranuloma/Erdheim Chester disease. Although nearly a century has passed since histiocytoses were recognized, their pathophysiology remains an enigma, and treatment is nonspecific. Erdheim-Chester disease (ECD) is a rare non-Langerhans cell histiocytosis, first described in 1930<sup>2</sup>. It is a systemic, heterogeneous disease mainly involving the bones, lungs, skin, retro-orbital tissues, central nervous system (CNS), pituitary gland, vessels, kidneys, <sup>5</sup> retroperitoneum, and heart. It has an unknown incidence, with fewer than 500 cases published in the literature.

Gastrointestinal involvement, as occurred in our patient, is extremely rare<sup>4,6</sup>. In a series of thirty-seven patients, the mean age at diagnosis was fifty-two years. Two criteria, of which one should be fulfilled, were proposed as a requirement for diagnosis of ECD: (i) Typical histological findings with foamy histiocytes and polymorphic granuloma and fibrosis or xanthogranulomatosis with CD68-positive and CD1a-negative immunohistochemical staining and (ii) Typical skeletal findings with a) radiographs showing bilateral and symmetric osteosclerosis of the diaphyseal and metaphyseal regions in the bones and/or b) symmetric and abnormally increased labeling of the distal ends of the long bones, and sometimes, the upper limbs, on bone scan. The clinical presentation of ECD is largely dependent on the distribution of disease, which may range from asymptomatic bone lesions to multisystemic, life-threatening forms with poor prognosis, especially with CNS or cardiovascular involvement. The most common presenting symptom of ECD is bone pain, mainly affecting the lower limbs.

Typical radiological and pathological features may suggest the diagnosis, but there is a broad clinical spectrum, ranging from asymptomatic tissue infiltration to fulminant multisystem organ failure. Numerous therapies are proposed in the management of ECD, including corticosteroids, chemotherapy, radiotherapy, calcineurin inhibitors and alpha-interferon therapy. Prognosis of disease is largely dependent on the extent of extra-skeletal involvement and in particular, involvement of central nervous or cardiac systems. Mortality rate is quoted as forty percent in the first forty months.

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