Erdheim-Chester disease: a case report with rare presentation of a catastrophic disease

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ABSTRACT

Background: Erdheim-Chester disease (ECD) is a non-Langerhans cell histiocytoses with multiorgan xanthomatous infiltration of tissues by histiocytes, surrounded by fibrosis.

Case presentation: A 68-year-old Caucasian female was admitted to the emergency department for dyspnea. The patient was given empiric antibiotic therapy with Piperacilin/Tazobactam, but after 10 days the patient underwent respiratory failure accompanied with worsening of the renal function, noninvasive ventilation and diuretics for perfusion were given, but the treatment was unresponsive. Kidney Computed Tomography showed enlargement of both kidneys, suggesting an inflammatory process. A renal biopsy was performed which confirmed the diagnosis of ECD.

Conclusion: It is an overlooked diagnosis due to its rarity, variable presentation, and the subsequent manifestations may also develop after several years.

Keywords: End-stage renal disease, Erdheim-Chester disease, hemodialysis, histiocytosis, non-Langerhans cell, case report.

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Background

Erdheim-Chester disease (ECD) is a non-Langerhans cell histiocytoses with multiorgan xanthomatous infiltration of tissues by histiocytes, surrounded by fibrosis. These foamy histiocytes are stained positive for CD-68 and lack CD1a expression [1,2]. It mostly presents in the fifth decade of life with a high prevalence in males [3].

It is a multisystemic disease which can affect any organ but skeletal involvement is the most common with pain in the bones. The extent and distribution of the disease determines the clinical course.

Case Presentation

A 68-year-old Caucasian female was admitted to the emergency unit for dyspnea and confused mental state. Medical history of the patient included arterial hypertension, diabetes mellitus type-2, obesity, dyslipidemia, hypothyroidism, obstructive sleep apnea, pulmonary hypertension with fibrosis, lymphoma (22 years ago with an orbital mass), and loss of visual acuity that was treated only with corticosteroids but without clinic follow-up since.

At admission, laboratory data revealed hemoglobin level 7.8 g/dl (normal range, 12.5–15.0 g/dl), leukocytes 23.900 × 10^9/l (normal range, 4.000–10.000 × 10^9/l) with neutrophils predominance, C-Reactive Protein 7.28 mg/dl (normal range <0.1 mg/dl), urea 121 mg/dl (normal range, 25–50 mg/dl), creatinine 1.7 mg/dl (normal range, 0.5–1 mg/dl), albumin 1.8 g/dl (normal range, 3.5–4.5 g/dl).

Other relevant data included normal serum protein electrophoresis, normal serum immunoglobulins; antinuclear antibodies, Anti-Neutrophil Cytoplasmic Antibodies and anti-dsDNA antibodies were negative. Computerized tomography (CT) of the thorax showed diffused pleural thickening with right pleural effusion, intra-lobular and sub-pleural thickening, bronchiectasis, interstitial fibrosis, and circumferential sheathing of the thoracic aorta. CT of kidney displayed enlargement of both kidneys with hypodense regions, suggesting an inflammatory process, hydronephrosis was absent.

The patient was given empiric antibiotic therapy with Piperacilin/ Tazobactam 3.375 g every 6-hours; however, after 10 days, the patient underwent respiratory failure, anasarca, worsening of renal function with anuria, and atrial fibrillation, and was transferred to Intensive Care Unit (ICU). In the ICU, the patient was treated with non-invasive ventilation and diuretics for perfusion, but the treatment was found unresponsive and it became necessary to start renal replacement therapy.

When hemodynamic stabilization was achieved, the patient was transferred to the Nephrology department. An ischemia of the left lower limb (grade IV) was also present with the necessity to amputate, and maintain hemodialysis dependency. Immunoglobulins dosing were also made to exclude IgG4 disease.

The renal function was normal one month before the admission. A renal biopsy of the patient was performed that revealed numerous histiocytes, which were positive for CD68 and FXIIIa, and negative for S100 and CD1a, which suggested the diagnosis of ECD.

Before the stage of the disease could be graded or any effective therapy could be offered, the patient passed away.

Discussion

The present case presented with renal, pulmonary, cardiovascular, and orbital involvement. As in this case, end stage renal disease requiring hemodialysis due to ECD is very rare [4]. Approximately, one-third of ECD cases present with pseudo retroperitoneal fibrosis, in some cases it is complicated by bilateral hydronephrosis causing obstructive renal failure [5]. Sometimes, renal failure is caused directly by infiltration of the renal parenchyma.

Dyspnea accounts for a large majority of respiratory symptoms. As evident from the present case's CT scan, there was involvement of the pleura and lung parenchyma, this accounts for more than 50% of the cases and it is associated with a poor prognosis [1]. The current patient presented cardiovascular involvement, and the most frequent cardiovascular sign is the circumferential sheathing of the thoracic and/or abdominal aorta [4]. One in four patients with ECD develops exophthalmos caused by infiltration of the retro-orbital soft tissues. The ophthalmic mass that the present patient had 22 years ago was probably compatible with this [4].

ECD, although rare, is an overlooked diagnosis because it has a variable presentation and its manifestations may develop after several years [3]. The correlation between clinical, radiological, and histological findings is mandatory for the diagnosis of the disease. The histological analysis involves the analysis of histiocytes in tissue biopsies which are typically foamy, CD68+, CD1a–, and may be found in almost any tissue [4].

Conclusion

ECD is an overlooked diagnosis due to its rarity, variable presentation, and the subsequent manifestations may also develop after several years. All the end-stage renal disease cases reported in literature with ECD had obstructive renal disease and ureterohydronephrosis, but in the present case there was no evidence of an obstructive cause. Since, it is a rare diagnosis, it is always necessary to have clinical suspicion to make an early detection.

List of Abbreviations

CT	Computerized tomography
ECD	Erdheim-Chester disease
ICU	Intensive care unit

Consent for publication

Informed consent to publish this case report was obtained from the patient's family.

Ethical approval

Ethical approval is not required in Centro Hospitalar do Médio Tejo to publish an anonymous case report.

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Summary of the case

1	**Patient (gender, age)**	68 years old, Female
2	**Final diagnosis**	Erdheim-Chester disease
3	**Symptoms**	Dyspnea, anasarca, anuria,
4	**Medications**	Diuretics, hemodialysis,
5	**Clinical procedure**	renal biopsy, CT scan
6	**Specialty**	Hematology; Nephrology