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Erdheim-Chester Disease: A Case of a Cardiac Involvement

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Abstract **Case Report**

Erdheim-Chester disease (ECD) is a rare non-Langerhans histiocytic multisystem disorder due to xanthogranulomatous infiltration of tissues by spumous histiocytes, It typically presents with diffuse bone pain however the cardiac manifestation are common and occur to 75% of the patients. It is frequently asymptomatic and detected incidentally on radiological imaging and it most commonly manifests as pericardium and myocardium infiltration We report the case of a 66-year-old woman patient, admitted for diffuse bone pain, asthenia and a left arm weakness who presented and infiltration of the aorta with a coated aspect, a thickened right atrium and a small pericardial effusion.

Keywords: Erdheim-Chester disease, cardiovascular involvement, right atrium infiltration, pericarditis, pericardial effusion, coated aorta sign.

INTRODUCTION

Erdheim-Chester disease (ECD) is a rare non-Langerhans histiocytic multisystem disorder of unknown etiology commonly manifest as multifocal sclerotic lesions due to xanthogranulomatous infiltration of tissues by spumous histiocytes, lipid-laden macrophages or histiocytes, surrounded by fibrosis, within multiple tissues and organs damaging them and leading in certain cases to life-threatening organ failure [2, 6].

ECD can be distinguished from Langerhans cell histiocytosis by the immunohistologic and microscopic characteristics of histiocytes. ECD patients are constantly positive for CD68 and negative for CD1a, and ultrastructural studies show no Birbeck granules [1].

Since 1930, when ECD was first described by Erdheim and Chester, fewer than 1000 cases have been reported in the medical literature [12] but the actual incidence is unknown.

The signs and symptoms of Erdheim-Chester disease varies depending on the affected organ and usually appear in the adulthood between the ages of 40 and 70, the disorder may also occur at any age [11]. It presents as a multi-systemic disease with manifestations affecting the skeleton, skin, respiratory, cardiovascular, renal and nervous system [13].

CASE PRESENTATION

Herein , we present a case of a 66 year old woman whose medical history is a diabetes insipidus since 12 years put on desmoprissin and three successive miscarriages , whose symptomatology seems to go back to 2 months of diffuse bone pain, a dysarthria and a left arm weakness On admission The clinical examination showed a conscious patient with stable hemodynamic and respiratory status, bilateral xanthelasma and nodular lesions around the eyes, edema of the lower limbs reaching mid-leg and flaccid hypotonia of the left arm.

The blood results during admission revealed an inflammatory syndrome made of normocytic anemia with a hemoglobin of 90 g/L and a mean corpuscular volume of 95.6 femtoliter, ferritinemia at 311ug/L hyperleukocytosis at 22480/uL predominantly neutrophilic polynuclear 19880/uL, thrombocytosis at 504000/uL and a CRP at 86 mg/l renal function tests, and liver function tests, including alkaline phosphatase (ALP), were normal (apart from low albumin, 22.6 g/L). The coagulation screen and glucose were also normal.

Due to the inflammatory syndrome in addition to asthenia and weight loss, a CT scan of the chest, abdomen, and pelvis were requested to rule out infection and malignancy, a cerebral MRI to determine the neurological status and echocardiography to rule in/out the cardiac origin of the oedematous syndrome.

The findings within the chest were a combination of interlobular septal thickening and bilateral pulmonary micronodules. The kidneys showed diffuse perinephric soft tissue making the hairy kidney sign and also within both kidneys responsible of bilateral hydrocalices. There was also a peri-aortic infiltration involving the thoracic aorta and the sub-renal abdominal aorta giving it an appearance of "coated aorta", associated with a right peri-atrial infiltration, a small pericardial effusion. The echocardiography showed a small pericardial effusion and a thickened and non-dilated right atrium,(inferior and lateral wall 16mm/10mm) the ejection fraction was preserved (65%) Brain MRI showed an arachnoidocele and no acute intracranial abnormality

The case was forwarded to the internists where the diagnosis of ECD was made and confirmed on The Immunohistochemical staining: the skin biopsy demonstrated a dermal fibrosis with non-Langerhans histiocytosis with cells that are CD68+ ,S100+ and CD1a - , the bone marrow biopsy objectified a non Langerhans medullary histiocytosis with cells that are CD20+ ,CD5+ PS100+ ,CD34 - and CD1a - .

The patient was managed on corticosteroid therapy and albumin perfusion. Symptoms improved after optimizing doses of medical treatment and then referred to hematology for chemotherapy where the VNCOP-B protocol was started.

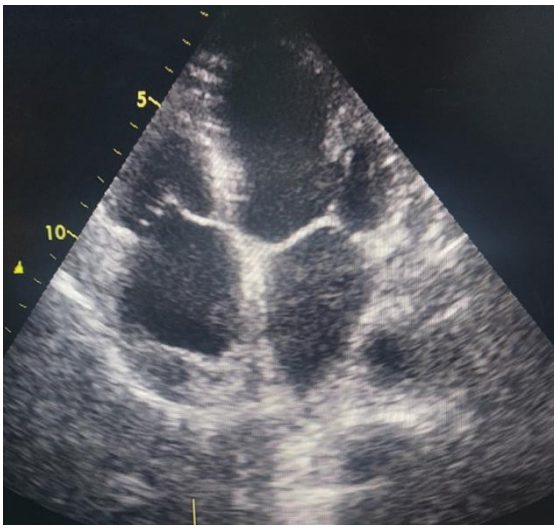

Figure 1: An Apical four-chamber view of the heart showing the thickened RA

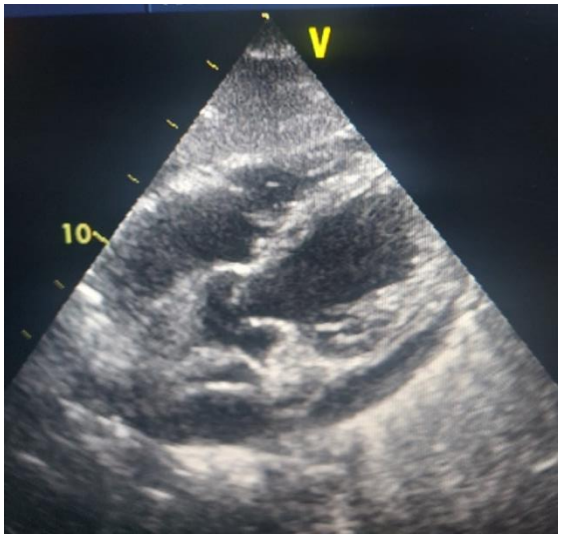

Figure 2: A Subcostal five chamber view showing the posterior pericardial effusion

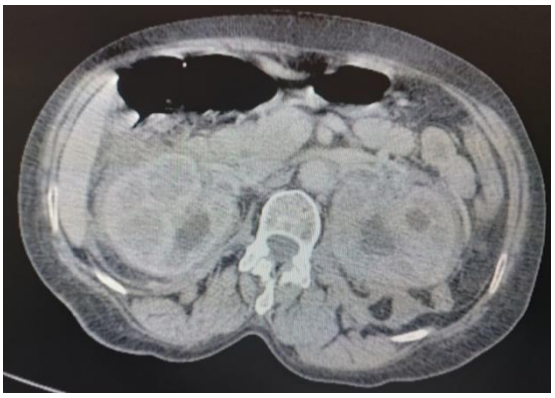

Figure 3: CT Scan showing the abdominal aortic infiltration with the kidneys hydrocalices

DISCUSSION

75% of ECD patients suffer from cardiovascular involvement[8] It most commonly manifests as infiltration of the pericardium and myocardium with a tendency to involve the right atrial myocardium [2], with a pseudotumoral aspect [6, 9] It can also present as pericarditis and pericardial effusion, which may lead to cardiac tamponade [2].

Extensive vascular involvement of the aorta and it's branch has also been reported [6, 10] this periaortic infiltration is pathognomonic of ECD. known as the "coated aorta" phenomenon [5] When faced with this type of periaortic infiltration, the diagnosis of ECD shout be considered , this particular aspect seems to be specific to ECD, as it has not been reported in other forms of histiocytosis particularly, Langerhans cell histiocytosis [5].

SVC disease is less common [6] unlike arterial involvement, it was generally due to a thrombotic complication of a venous compression by the xanthomatous process rather than to direct infiltration of the vessel wall [2].

Electrocardiographic abnormalities that have been reported include shortening of the PR interval, sinoatrial block, sinus bradycardia, Q-wave abnormalities, and ST-T wave abnormalities [7].

Identifying cardiac involvement is crucial because cardiovascular complications, including hemodynamically significant arrhythmias, cardio-myopathy, myocardial infarction, and severe valvular insufficiency, are frequent causes of death in patients with ECD [7].

The cardiac infiltration in our case has extended from the right atrium to involve surrounding vasculature and specially multiple segments of the aorta. fortunately the patient did not present any cardiovascular symptomatology except for edema which was correlated to hypoalbuminemia since the echocardiography showed a preserved ejection fraction estimated at 65% without significant valvular disease or dilatation of the inferior vena cava, the BNP/Pro BNP were also negative.

CONCLUSION

To conclude, The ECD cardiovascular involvement is frequent and most commonly manifests as infiltration of the pericardium and myocardium with a tendency to involve the right atrial myocardium , Clinical manifestations, are numerous and polymorphous that may lead in certain case to death, therefore the Cardiovascular involvement in ECD patients should be sought systematically.

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