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Case Report
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Erdheim-Chester Disease Presenting as a Massive Pericardial Effusion Confirmed by Bone Marrow Biopsy: A Case Report and Mini Review

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Abstract

Erdheim-Chester disease (ECD) is a non-Langerhans cell histiocytosis, which featured organs and tissues infiltrated with foam cells, causing lipoid granulomatosis and fibrosis. The incidence of ECD is extremely low, since it was first reported in 1930. When cardiac was involved predicting worse prognosis. Here, we report a case of ECD presenting as a pericardial effusion confirmed by bone biopsy. A 66-year-old man was admitted because of dyspnea and legs edema. The echocardiogram revealed moderate-large volume pericardial effusion. The PET/CT revealed pathognomonic coated aorta and highest uptake in both long bones, we performed a bone marrow biopsy. ECD was confirmed by foamy non-Langerhans histiocytes with CD68+CD1α-S100-.

Case Presentation

A 66-year-old woman presented with dyspnea and legs edema over 2 months, worsening of paroxysmal nocturnal dyspnea for 1 week. Her Department of Cardiology, Second Affiliated Hospital of Dalian Medical University, Dalian, Liaoning Province 116023, China only medical history was hypertension, which she had been taking medicine regularly. The patient was diagnosed with heart failure in local hospital 1 week ago. The chest radiograph and transthoracic echocardiography at the local hospital both reported massive pericardial effusion, and abdominal ultrasound showed bilateral hydronephrosis. So, she came to the Second hospital of Dalian Medical University, China for further treatment. On admission, her vital signs were: Blood pressure 193/98 mmHg, pulse rate of 88 beats/min, respiratory rate 20 breaths/min, and body temperature of 36.3°C. We didn't find arrhythmia and hypo-phonetic sounds. Her chest examination revealed minimal moist crackles at both bases. The abdomen examination showed no abnormalities and without hepatosplenomegaly. No cervical, inguinal or axillary lymphadenopathy was identified. There were no cutaneous nor neurological abnormal findings.

Methods and Procedures

Blood gas analysis showed type II respiratory failure (Under oxygen inhalation: O2 partial pressure of 69.2 mmHg, CO2 partial pressure of 69.1 mmHg). Laboratory studies revealed BNP, NT-pro BNP and myocardial necrosis markers were normal. The inflammatory markers, including Complete Blood Count (CBC), C-Reactive Protein (CRP), Erythrocyte Sedimentation Rate (ESR) were also normal. We didn't find any abnormal results in regular tests, including renal function, liver function, thyroid function, coagulation screen, total cholesterol, glucose and tumor specific antigen. As well as we detected indicators related to rheumatic immune system diseases, the autoantibody screen was negative (rheumatoid factor, and antinuclear antibody, anti-dsDNA, anti-Sm, anti-SSA (Ro), anti-RNP and anti-neutrophil cytoplasmic antibodies). Although we had suspected tuberculosis, but ultimately ruled it out.

The echocardiogram revealed normal systolic function and moderate-large volume pericardial effusion (left ventricular ejection fraction: 60%, Figure 1), then we took pericardiocentesis with total 300 ml effusion. The routine of pericardial effusion preferred some atypical cells and without tumor cells. Urinary tract CT angiography (CTU) suggested bilateral renal changes that tended to be inflammatory, but did not rule out tumor (Figure 2). Kidney biopsy were performed, and

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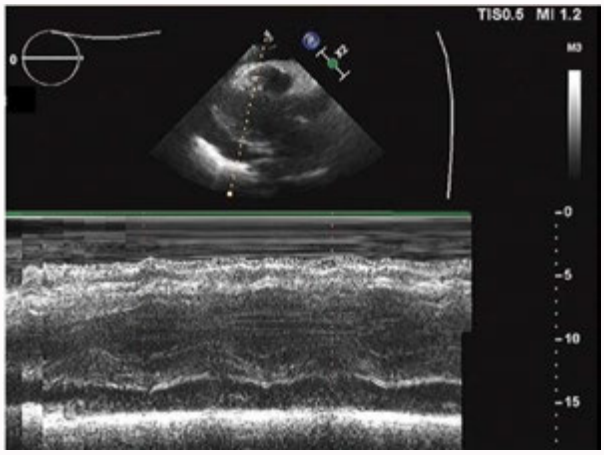

Figure 1: The echocardiogram revealed normal systolic function and moderate-large volume pericardial effusion.

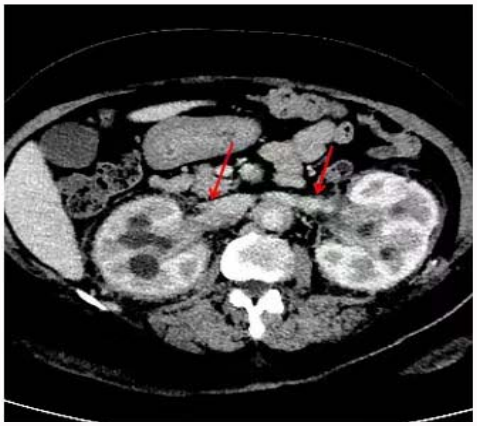

Figure 2: Urinary tract CT angiography (CTU) suggested bilateral renal changes.

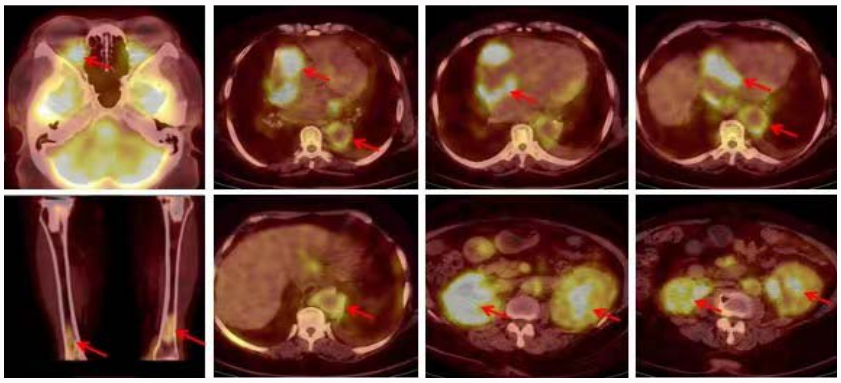

Figure 3: Kidney biopsy were performed, and the pathology results showed inflammatory change.

the pathology results showed inflammatory change (Figure 3). The whole-body Positron Emission Tomography (PET-CT) was performed furtherly, and it revealed fluorodeoxyglucose highest uptake in multiple organs, including all levels of blood vessels, optic nerve, bones, kidney and other organs (Figure 4). The whole-body bone scan revealed increased uptake in multifocal bone lesions, special in both distal femurs. Cardiac and aortic lesions involvement were identified. The results indicated a systemic disease.

After a multi-disciplinary treatment, Erdheim-Chester disease was highly suspected. The characteristic pathologic feature of ECD is histiocytosis. Due to the limitations of the myocardial biopsy, we performed a bone marrow biopsy furtherly. The bone marrow biopsy showed diffuse infiltration by foamy non-Langerhans histiocytes surrounded by fibrosis (Figure 5). Immunohistochemical staining with CD68 showed positive result, and negative results for CD1a and S100, compatible with ECD (Figure 6). Eventually the ECD was confirmed by biopsy with bone marrow despite with negative pathologic findings in kidney biopsy. Then the patient went to the department of hematology of Peking Union Medical College Hospital for further medical treatment and participated in clinical trial of

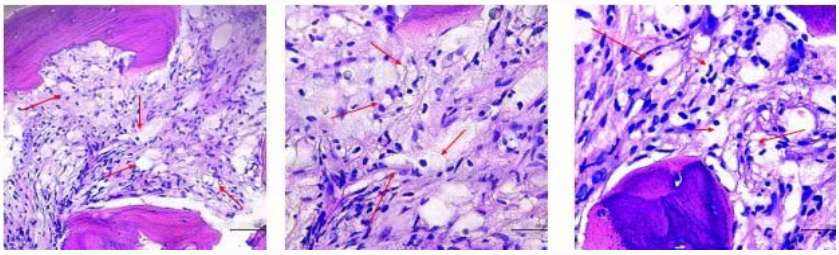

Figure 4: ^{18}F-FDG PET/CT scans revealed fluorodeoxyglucose highest uptake in multiple organs, including all levels of blood vessels, optic nerve, bones, kidney and other organs. The highest uptake lesions of cardiovascular were mainly around aorta and right coronary.

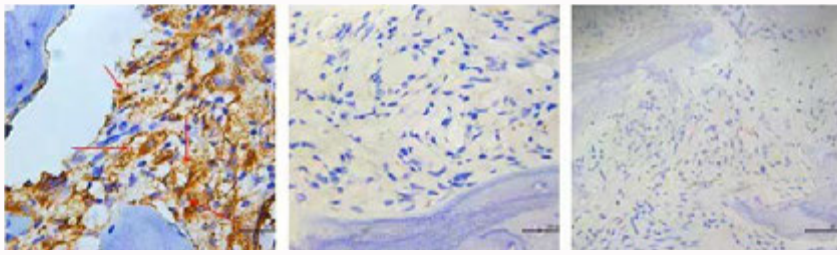

Figure 5: The bone marrow biopsy was performed showing diffuse infiltration by foamy non-Langerhans histiocytes surrounded by fibrosis.

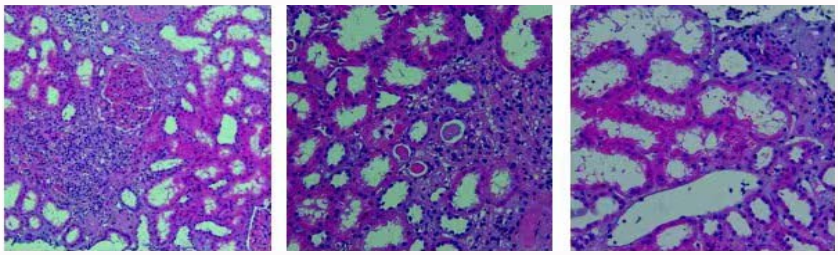

Figure 6: Immunohistochemical staining with CD68+CD1a-S100-.

BRAFV600E inhibitor.

Discussion

Erdheim-Chester Disease (ECD) is a non-Langerhans cell histiocytosis, which featured organs and tissues infiltrated with foam cells, causing lipoid granulomatosis and fibrosis. The incidence of ECD is extremely low, since it was first reported in 1930. Until 2019, there are only 1,500 known cases worldwide [1]. The median age at diagnosis of ECD was 60 years, the male/female patient ratio was 3:1 for pure ECD [2]. ECD often involves in multiple systems, such as lung, kidney, bone and nervous system. With the development of imaging, the positive rate of the cardiovascular system in ECD has been improved. The specific cardiovascular changes include the fibrosis in periaortic, pericardial, myocardial, or less often peri-coronary fibrosis [3], and cardiovascular involvement occurs in up to 75% of patients [4]. The incidence of diffuse thickening and/or pericardial effusion is 13% to 24%, but cardiac tamponade hardly occurs [5,6]. When cardiac was involved predicting worse prognosis, the mortality rate is as high as 40% [7]. At present, characterized histological changes are still the gold standard for ECD diagnosis, at assistance of radiologic examinations such as 18F-FDG PET/CT scans. In this case, we found foamy and CD68+CD1α-histiocytes in bone marrow, even though the kidney biopsy was negative. In this case we observed pericardial effusion, and we also observed pathognomonic coated aorta through PET/CT. We did not find the other characterized feature, such as right atrium pseudotumor, whose incidence rate is 30% [5].

The pathogenesis of ECD is still unclear, and treatment is mainly related to gene mutations. The RAS-RAF-MEK-ERK pathway is a cellular signaling pathway, which plays a major role in tumors. BRAFV600E mutation results in an activation of RAS-ERK pathway, independently of RAS activation. BRAFV600E mutations have been detected in patients with LCH. The study showed that ECD and LCH share similar oncogenic pathways. A BRAFV600E mutation was detected by pyrosequencing in 13 of 24 (54%) patients with ECD. In addition, immunohistochemical analysis of ECD tissue samples using BRAFV600E-selective antibody showed that the mutation was only expressed in typical foam histiocytes and Touton giant cells, but not in lymphocytes, fibroblasts, and endothelial cells [8]. The BRAFV600E mutation was also positive in this patient, and the treatment is based on the special gene mutation (the result was not supplied). This reported patient has been participating in the clinical trial of BRAFV600E inhibitor for half a year. During follow-up, pericardial effusion and other clinical symptoms have not appeared again, such as weakness, dyspnea and legs edema.

Conclusion

ECD is a rare histiocytic neoplasm, cardiovascular involvement is frequent and indicates a poor prognosis. Herein, we report a case of ECD mainly presenting as a pericardial effusion diagnosed through a combination of clinical, radiologic, and pathologic findings. Significant challenges remain in the diagnosis and treatment of ECD.

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