COVID-19 Reveals a Diagnosis of Erdheim-Chester Disease

Amanda Fahrenbruck OMS IV[1], Johnny Cheng, DO[2]

1 Rocky Vista University College of Osteopathic Medicine, Parker, CO 2 Critical Care and Pulmonary Consultants, Greenwood Village, CO

Introduction

Erdheim-Chester Disease is exceedingly rare with fewer than 1000 cases reported in the literature.[1] The actual incidence is unknown as there is low awareness. Additionally, as a non-Langerhans histiocytosis process, it presents with wide-ranging clinical symptoms. It was first described in 1930 and most often initially presents with long bone pain. Over the last century, it has been most often diagnosed in adult men aged 50 to 60 years old.

Clinical presentation may include:

Long bone pain	Osteosclerotic lesions
Cardiac valvular abnormalities	Arrhythmias
Myocardial Infarcts	Fibrosis of large vessels, commonly aorta
Renal Dysfunction	Exophthalmos
Seizures	Periorbital pain
Ataxia	Blindness
Cranial Nerve palsies	Pulmonary fibrosis
Pleural Effusions	Xanthelasmas

Clinical Case

A 50-year-old male presented with symptoms of myalgias and fatigue following a positive COVID-19 PCR test. He was requiring supplemental oxygen. Initial concern for hypercoagulability and venous thromboembolism led to imaging with a CT angiogram that revealed aortitis.

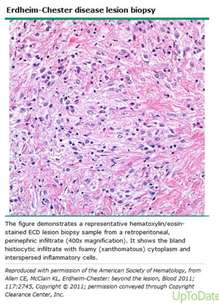
Further radiologic studies revealed soft tissue density surrounding the entire aorta as well as hyperattenuating perinephric fluid consistent with a diagnosis of Erdheim-Chester Disease (ECD).

These radiographic findings are the classic ECD findings of rind-like infiltration and inflammation of the kidneys termed **“hairy kidneys"** and **“coated aorta”** with circumferential soft tissue swelling.

This prompted further evaluation of the patient and he was discovered to have several classic findings of ECD including:

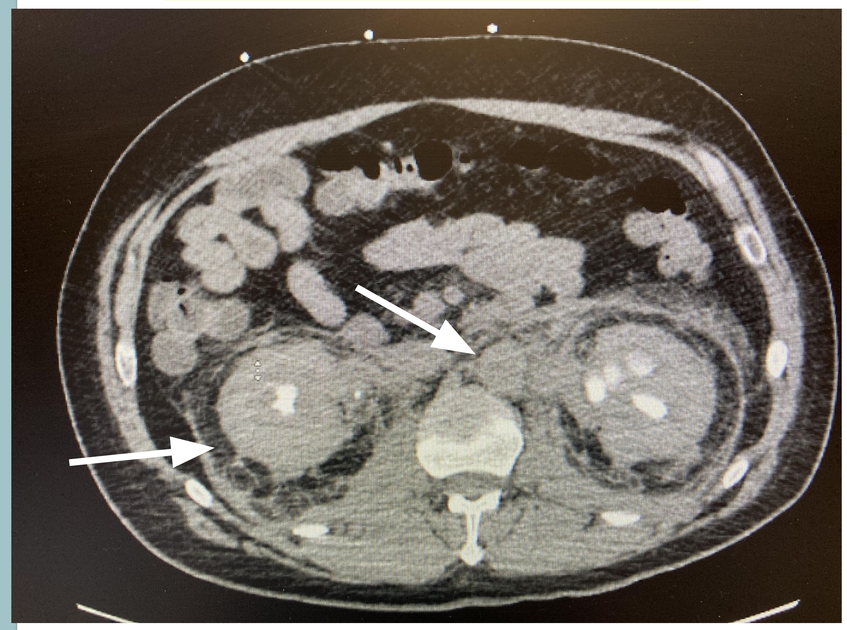
- Hypernatremia of Central Diabetes Insipidus
- Progressive Renal Dysfunction
- Myalgias that were correlated with long bone pain in the bilateral femurs

The gold standard for confirmation of Erdheim-Chester Disease is a soft-tissue biopsy. [3]
This will show lipid-laden histiocytes. However it is not always feasible to obtain such a biopsy.

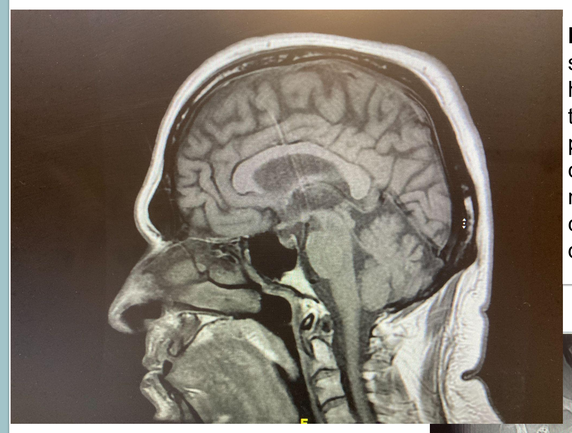


In this patient, full body imaging was performed and there was not a soft tissue mass available that would allow for safe biopsy. He was treated symptomatically and outpatient consultation with oncology was planned for further evaluation and treatment of ECD.

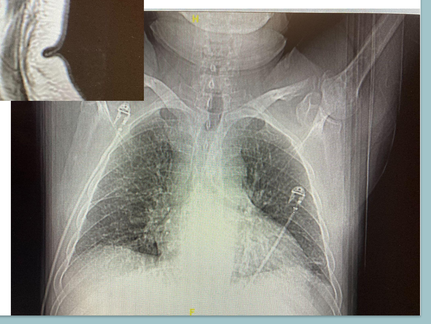
Diagnostic Imaging



- **CTA Chest/Abdomen/Pelivis WWO**: demonstrates the classic findings of “hairy kidneys” and “coated aorta.”
- There is perinephric infiltration creating a rind-like appearance circumferentially. This leads to the progressive renal dysfunction that is a central feature of the morbidity of this disease.
- The “coated aorta” is seen in approximately two thirds of diagnosed ECD cases[2]. This is seen here with a soft-tissue surrounding the abdominal aorta. It appears that this will first affect thoracic and abdominal aorta and then expand into other large vessels.



MRI of the pituitary: subcentimeter hypoenhancing nodule on the left side of the pituitary gland which could reflect a pituitary microadenoma and correlated with central diabetes insipidus.



Chest Xray: Peripheral predominant bilateral airspace opacities consistent with COVID pneumonia Unable to appreciate soft tissue infiltrations from ECD on this image.

Literature Review

Erdheim-Chester Disease (ECD) is extremely rare with fewer than 1000 cases in the medical literature since its discovery in 1930. It is as a Non-Langerhans-Cell-Histiocytosis with multi-system involvement and a poorly understood natural history. [1]

Often there is an acquired mutation in the BRAF V600E gene, which leads to a malignancy of myeloid progenitor cells and increasing production of pro-inflammatory cytokines. [4]

The mainstay of treatment is biologics including BRAF-Inhibitors, MEK inhibitors and mTOR inhibitors, although conventional chemotherapeutics also play a role in management of this disease.[1]

Conclusions

Erdheim-Chester Disease is a very uncommon condition and there is very little known about the prognosis and treatment course.

This case-presentation serves to illustrate the multi-system involvement and the need for a multi-disciplinary team-based approach to diagnosis and management. Roles included nephrology in the management of central diabetes insipidus, vascular surgery to evaluate aortitis, and oncology to manage the long-term disease modifying therapies. Further investigation is warranted to better understand this disease and be able to provide a well-coordinated multidisciplinary treatment.

Additionally, as diagnostic imaging progressives in quantity and quality, it is evident that more cases will be diagnosed based on the classic imaigning findings. In recent studies, the rates of incidental findings on imaging approaches 75%.[5]
As the barriers to imaging decrease and the rates of diagnostic imaging increase, it is expected that there will be a proportionally increasing number of rare conditions diagnosed. This necessitates that radiologists and clinicians be aware of typical findings in order to broaden their differential diagnoses to include Erdheim-Chester Disease.

Acknowledgements

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