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Incidental Discovery of Perirenal Erdheim-Chester Disease During a Post-Traumatic CT Scan: A Case Report

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ABSTRACT

Background: Erdheim-Chester disease (ECD) is a rare non-Langerhans cell histiocytosis characterized by multisystem infiltration by CD68+/CD1a− foamy histiocytes. Retroperitoneal and perirenal involvement, known as "hairy kidney," is a hallmark of the disease, present in 50–68% of patients. Diagnosis is often delayed due to its nonspecific clinical and radiological presentation.

Case presentation: We report the case of a 72-year-old male who underwent an abdominal computed tomography (CT) scan for evaluation following a motor vehicle accident (MVA). Imaging incidentally revealed a well-defined, bilobed, soft-tissue density lesion abutting the left kidney, isodense to the renal parenchyma without contrast enhancement. The renal parenchyma appeared non-infiltrated with normal portal-phase enhancement. Lymphoma was initially suspected. Ultrasound-guided percutaneous biopsy was performed, revealing infiltration by a non-Langerhans histiocytic neoplasm with morphological and immunophenotypic features consistent with Erdheim-Chester disease.

Conclusion: This case illustrates the incidental discovery of an ECD with an atypical presentation on an abdominal CT scan and highlights the importance of considering this rare diagnosis in cases of perirenal soft tissue lesions. A biopsy remains essential for establishing a definitive diagnosis, and a comprehensive multisystem evaluation is required once the diagnosis is confirmed.

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KEYWORDS:

Erdheim-Chester disease; Abdominal CT scan; histology proven; atypical hairy kidney presentation

I. INTRODUCTION

Erdheim-Chester disease (ECD) is a rare non-Langerhans cell histiocytic neoplasm, first described by Jakob Erdheim and William Chester in 1930.[1] It predominantly affects adults with a median age at diagnosis of 55 years and a male-to-female ratio of approximately 3:1.[1][2] By 2020, approximately 1,500 cases had been reported worldwide, although improved recognition has led to a rising incidence over the past decade.[3][4]

ECD is classified among the "L" group of histiocytoses alongside Langerhans cell histiocytosis (LCH) and is now recognized as a clonal myeloid neoplasm driven by activating mutations of the MAPK and PI3K-AKT signaling pathways.[3][5] The BRAF V600E mutation is found in 50–70% of cases, followed by MAP2K1 mutations in approximately 20%.[6][3]

The clinical manifestations of ECD are highly variable, ranging from asymptomatic incidental findings to severe multisystem disease. Common sites of involvement include long bones (80–95%), retroperitoneum/perinephric region (40–68%), cardiovascular system (40–70%), central nervous system (40%), and endocrine organs (50–70%).[6] The perirenal soft-tissue infiltration, known as "hairy kidney," is a hallmark radiological feature observed in approximately 50–68% of patients.[6][7][1]

We report the case of an incidental discovery of atypical perirenal lesion subsequently diagnosed as ECD during a post-traumatic CT evaluation.

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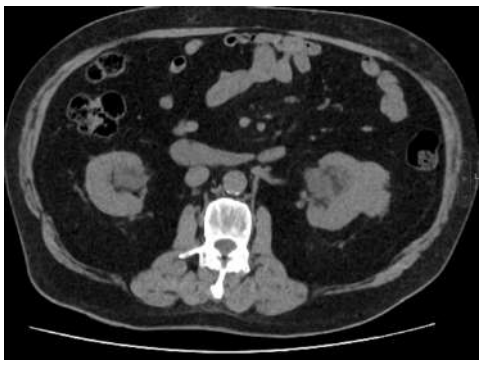
II. CASE PRESENTATION

A 72-year-old male (born in 1952) presented to the emergency department following a motor vehicle accident. As part of the trauma workup, a contrast-enhanced abdominal CT scan was performed.

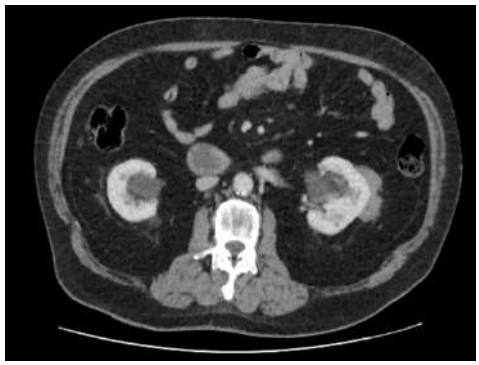
A. Imaging Findings

The CT scan revealed an incidental finding of a well-circumscribed, bilobed soft-tissue density lesion abutting the left kidney in an perirenal space location. The lesion was isodense relative to the renal parenchyma on unenhanced images and showed no significant contrast enhancement after intravenous contrast injection. The renal parenchyma appeared uninvolved, with normal enhancement during the portal venous phase. No hydronephrosis, periaortic infiltration, or other retroperitoneal abnormalities were identified.

Given this presentation and the focal infiltration of the perirenal space, lymphoma was initially considered the leading diagnostic possibility.



a



b

Fig.1 Axial-section CT scan without contrast (a) and after contrast administration in the portal phase (b).

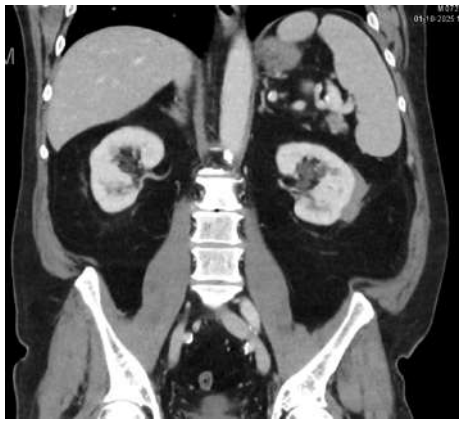


Fig.2 Coronal scan after contrast injection during the portal phase

B. Biopsy and Histopathological Analysis

An indication for percutaneous biopsy was established to characterize the lesion. The patient was referred to the interventional radiology department, where an ultrasound-guided core needle biopsy of the perirenal lesion was performed.

Histopathological examination revealed tissue infiltration by a non-Langerhans histiocytic neoplasm. The morphological and immunophenotypic features, interpreted in the clinical context, were consistent with a diagnosis of Erdheim-Chester disease. The neoplastic cells displayed characteristics of foamy histiocytes with an immunohistochemical profile expected in ECD: CD68+, CD163+, CD1a−, and Langerin (CD207)−.[6][8]

III. DISCUSSION

This case illustrates several important clinical and diagnostic considerations regarding ECD.

A. Incidental Discovery and Diagnostic Challenge

ECD is frequently diagnosed with significant delay due to its nonspecific clinical and radiological presentation.[5][9] The diagnosis requires integration of clinical, radiological, and histopathological findings.[2][8] In this case, the patient was asymptomatic regarding the perirenal lesion, which was discovered incidentally during trauma imaging. This presentation is consistent with the known spectrum of ECD, which can range from incidental asymptomatic findings to life-threatening multisystem disease.[9]

B. Imaging Characteristics

The perirenal infiltration in ECD, classically described as "hairy kidney," is one of the most characteristic radiological features of the disease and is present in 50–68% of patients.[6][7][10][1] CT typically reveals soft-tissue density infiltration surrounding the kidneys, with moderate enhancement on contrast-enhanced scans.[7] The bilobed, unilatéral, and well-defined morphology described in our case is somewhat atypical compared to the classic diffuse wispy perirenal infiltration pattern; however, focal or

mass-like presentations of ECD have been reported and may represent early or localized disease.[10][5]

The perirenal involvement in ECD may extend to the renal sinuses (56%), proximal ureters (61%), renal arteries (49%), adrenal glands (48%), and the aorta (43%), potentially leading to hydronephrosis, renal artery stenosis, and chronic kidney disease.[11][10][12]

C. Differential Diagnosis

The initial diagnostic hypothesis of lymphoma is a common consideration when encountering perirenal soft-tissue masses.[5] The differential diagnosis of perirenal soft-tissue lesions includes:

- **Lymphoma:** Typically presents as homogeneous soft-tissue infiltration that may encase the kidney.
- **Retroperitoneal fibrosis:** May mimic ECD but lacks the characteristic histiocytic infiltrate.
- **Other histiocytic disorders:** Including Rosai-Dorfman disease, which can involve the perinephric space but typically presents as hilar masses or subcapsular infiltration.[6]
- **Metastatic disease and sarcoma.**

Histopathological analysis with immunohistochemistry remains the gold standard for definitive diagnosis.[6][8]

D. Pathological Diagnosis

The diagnosis of ECD relies on the demonstration of tissue infiltration by foamy histiocytes with a characteristic immunophenotype: CD68+, CD163+, factor XIIIa+, CD1a−, and Langerin−.[6][8] Touton giant cells may be present. Molecular testing for BRAF V600E and other MAPK pathway mutations is recommended by the NCCN guidelines for all confirmed cases, as it has direct therapeutic implications. [6]

E. Recommended Workup After Diagnosis

Following histopathological confirmation of ECD, the NCCN guidelines recommend a comprehensive multisystem evaluation including:[6]

- Whole-body FDG-PET/CT scan including distal extremities (vertex to toes)
- MRI of the brain with and without contrast
- Complete laboratory evaluation
- Molecular testing for BRAF V600E and other MAPK pathway mutations (NGS panel)
- Cardiac evaluation (echocardiography, cardiac MRI) as clinically indicated
- Technetium-99m bone scintigraph

This comprehensive workup is essential to determine the extent of disease involvement, as CNS involvement is an independent predictor of mortality, and cardiovascular involvement occurs in 40–70% of patients.[1][3][6][13]

F. Treatment Considerations

Treatment of ECD depends on the extent and severity of organ involvement. Interferon-α has been the historical first-line therapy.[1][3] Targeted therapies, particularly BRAF inhibitors (vemurafenib, dabrafenib) and MEK inhibitors (cobimetinib, trametinib), have transformed the management of ECD, with robust and reproducible efficacy.[3][5] Molecular profiling is therefore essential to guide therapeutic decisions.

For patients with isolated or asymptomatic disease, close monitoring may be considered, although the multisystem nature of ECD generally warrants active treatment.[3]

IV. CONCLUSION

This case highlights the importance of considering ECD in the differential diagnosis of perirenal soft tissue lesions, even when they are discovered incidentally and present with a more atypical appearance. The “hairy kidney” appearance is a distinctive feature that can present in various forms and should prompt further investigation. A percutaneous biopsy followed by immunohistochemical and molecular analyses is essential for establishing a definitive diagnosis. Once the diagnosis is confirmed, a comprehensive multisystem evaluation is essential to guide treatment decisions and assess the prognosis.

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