Original Article

Erdheim–Chester Disease: A Diagnostic Challenge Masquerading as Lymphoma, A Case Report and Literature Review

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Abstract

Background: Erdheim–Chester disease (ECD) is a rare non-Langerhans cell histiocytosis characterized by clonal proliferation of foamy histiocytes driven by MAPK–ERK pathway mutations, most commonly BRAF^V600E. The disease exhibits wide clinical heterogeneity and frequently involves the skeletal, cardiovascular, and retroperitoneal systems, often posing diagnostic challenges. **Case Presentation:** A 79-year-old Caucasian woman came to see us for six months of exertional dyspnea, cough and neck tightness, with a decrease in appetite and fatigue. The mass in the anterior mediastinum was seen to encase the great vessels and the trachea on contrast enhanced chest CT; the left retroperineal region showed infiltration, and the classic "hairy kidney" sign, was seen on abdominal imaging, suggesting systemic ECD. ^18F-FDG PET/CT showed mild hypermetabolism in these regions. Initial FNA biopsy failed to yield a diagnosis, but a thoracoscopic biopsy of fibrotic tissue was positive for foamy histiocytic infiltration with CD68 expression and CD1a negativity and cyclin D1 positivity. There was no BRAF V600E test performed, and further molecular testing was pending. Multidisciplinary management entailed a pericardial window for effusion removal and for the planning of a mutation-specific systemic therapy. **Discussion:** This case highlights the protean presentations and diagnostic challenges of ECD and the necessity for multi-systemic clinical, radiologic and pathologic assessment. Mediastinal, pericardial, and retroperitoneal involvement is a sign of systemic involvement. **Conclusion:** A diagnosis of ECD should be suspected when there is unexplained fibrosis of the mediastinum or retroperitoneum. Imaging, histopathology and molecular testing are all crucial for proper diagnosis and treatment planning. Multidisciplinary early care provides better diagnosis and targeted therapy in this rare disorder.

Keywords: Erdheim–Chester disease; non-Langerhans cell histiocytosis; mediastinal mass; retroperitoneal fibrosis; hairy kidney sign; BRAF mutation.

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Introduction

Erdheim–Chester disease (ECD) is a rare clonal non-Langerhans cell histiocytosis that was initially described in 1930 and now falls under the "L group" of histiocytoses, along with Langerhans cell histiocytosis (LCH).[1-3] Since original description (approx 1,500 cases in modern series) less than a few thousand cases are reported, and there is a clear predilection for middle-aged adults, and with a male predominance.[1,3,4] The MAPK-ERK pathway is activated in ECD, primarily via BRAF V600E mutations (57–70%) and, less commonly, via MAP2K alterations, establishing ECD as an inflammatory type of myeloid neoplasm with therapeutic targets.

As a clinical condition, ECD demonstrates a wide range of and multiple systemic involvement. The skeletons are most commonly involved and this usually presents with bilateral and symmetrical osteosclerosis of the long bones.[1,3] The typical imaging features include periaortic "coated aorta" and perirenal "hairy kidney." Cardiovascular disease can present as a pseudo-tumor of the right atrium or pericardial thickening which is very significant regarding the morbidity and prognosis.[1,3,5,6] Central nervous system, lungs and kidneys also commonly get involved and are determinants of prognosis.[1,3,7-9]

Whole-body staging, biopsy targetting, and monitoring of disease activity benefit greatly from PET/CT, as does imaging of the brain and heart when physiologic uptake limits the sensitivity of PET/CT.[3,5,10] Xanthogranulomatous infiltrates are usually found on histopathology and are made up of foamy histiocytes that express CD68+, CD163+, factor XIIIa+, CD1a–, langerin (CD207)–, and to a variable extent, S100+.[3,11] In some patients, alterations of MAPK pathway may co-exist with LCH, so molecular testing like BRAF V600E should also be considered to confirm pathway activation and for decision-making purposes regarding targeted therapy.[1-3]

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We describe a case that exemplifies these diagnostic dilemmas and underscores the need for a multidisciplinary approach involving a team of radiologists, pathologists, hematologists/oncologists, cardiologists and neurologists to achieve a proper diagnosis and to maximize patient treatment.

Case Presentation

A 95-year-old Caucasian woman, who had never smoked and was a retired nurse, presented with exertional shortness of breath and cough for 6 months, and more recently a tightness of the neck without acute respiratory distress. She also complained of loss of appetite, early fullness and tiredness. No fever, night sweats, chest pain, hemoptysis or unintentional weight loss. Her medical history was significant for gastroesophageal reflux disease, tachycardia, hyperlipidaemia and a previous COVID infection. Remarkable in the family history were the presence of breast cancer in the mother, multiple myeloma in a brother, smoking-related lung cancer in another brother and hypereosinophilia in a son. Chest computed tomography (CT) several months prior to admission showed a large mass in the anterior and superior part of the mediastinum that encased and displaced the trachea to the right and surrounded the great vessels and aorta [Figure 1]. Abdominal images showed left retroperitoneal infiltrative soft tissue mass with spiculated margins and ring-like soft tissue density along pararenal fascia, which extended to the diaphragm and involved the left adrenal gland, causing lateral displacement of the left kidney [Figure 2]. A small pericardial effusion was also observed and there were no lung nodules or parenchymal masses. There was no change in the size of a right hepatic dome lesion (approx. 2.6 cm) from several years before imaging.

Subsequent PET/CT with an 18F-FDG revealed mild hypermetabolism of the mediastinal mass, with low uptake of the retroperitoneal soft-tissue stranding, that were consistent with the perirenal infiltration or the "hairy kidney" sign typical of systemic Erdheim–Chester disease [Figure 3].[1] En sample obtained from the retroperitoneal lesion on ultrasound-guided fine-needle aspiration showed chronic inflammation and fibrotic and skeletal muscle tissue, but was not adequate for flow cytometry and did not contain any evidence of lymphoma or metastatic carcinoma.

Echocardiography performed several days later demonstrated interval increase in the size of the pericardial effusion to maximal posterior diameter of 1.5 cm without tamponade characteristics [Table 1]. The first radiologic impression suggested lymphoma and a core biopsy of an anterior mediastinal mass was performed for histopathology and flow cytometry. Biopsy did not show any sign of lymphoma, nor did the flow cytometry. An increasing diagnostic dilemma led to the collection of a larger specimen via a window in the pericardium and thoracoscopic biopsy of the mediastinal mass.

Histopathologic evaluation revealed fibrotic tissue with foamy histiocytic infiltration and the slides were submitted to the National Cancer Institute for expert review. At the last consultation, the diagnosis of Erdheim–Chester disease was favoured. Treatment options considered were: conventional first line therapy (systemic interferon-α); and targeted therapy, based on molecular analysis. To evaluate the eligibility for BRAF or MEK inhibitors which have proven to be effective in ECD, genetic studies including BRAF V600E and others are ordered.[2] Given the possibility of the presence of malignant cells, the patient was treated for pericardial effusion and mediastinal compression until the molecular results were received. With the advent of molecular profiling, the systemic therapy plan was worked out in a multidisciplinary team comprising of cardiology, oncology, and thoracic surgery.

This case has been reported in accordance with CARE guidelines.

Table 1: Timeline of Clinical Events and Diagnostic Workup

Time Period	Event / Investigation	Key Findings / Outcomes
~6 months before presentation	Onset of symptoms	Exertional dyspnea, dry cough, mild neck tightness, decreased appetite,
Initial evaluation	Medical history and family review	History of GERD, tachycardia, hyperlipidemia, prior COVID-19; family
Early diagnostic imaging	Chest CT	Large anterior–superior mediastinal mass encasing the trachea and great
Subsequent imaging	Abdominal CT	Infiltrative left retroperitoneal soft-tissue process extending to diaphragm
Functional imaging	^18F-FDG PET/CT	Mild uptake in mediastinal mass and low-grade perirenal
First biopsy	Ultrasound-guided retroperitoneal FNA	Fibrotic tissue with chronic inflammation; nondiagnostic; insufficient for
Cardiac assessment	Echocardiography	Pericardial effusion enlarged to ~1.5 cm; no tamponade physiology.
Second biopsy	Thoracoscopic mediastinal biopsy and	Foamy histiocytic infiltrates; pathology reviewed by the National Cancer
Molecular testing	BRAF V600E and MAPK pathway	Testing requested to determine eligibility for targeted therapy.
Multidisciplinary management	Cardiology, oncology, thoracic surgery	Supportive care for pericardial effusion; systemic therapy to be initiated

Case Discussion:

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Epidemiology of ECD: Erdheim–Chester disease (ECD) is a rare type of non-Langerhans cell histiocytosis that occurs in about 1 out of every 1 million people. It predominately occurs in the middle age group, mainly in men (1:2–3).[1,2] Our patient, however, was on the older end of the disease spectrum and does illustrate that this can happen without any known classic risk factors. While not commonly recognised in the past, the diagnostic yield has progressively improved over the years due to increased awareness and the use of more advanced imaging techniques; most diagnoses have been made in referral centres within the field of histiocytic disorders.[2]

Pediatric cases are very unusual and may have overlapping features with juvenile xanthogranuloma and are often BRAF^V600E mutated. Some clustering is reported in the South

of Italy and the centre of France, and in some cases this clustering is correlated with lower Human Development Index.[2] In about 50–70% of cases, right atrial pseudomass, pericardial thickening and the characteristic "coated aorta" appearance are the most clinically important findings in cardiac involvement. Pericardial effusion together with a large anterior mediastinal mass were the most prominent findings in our case, illustrating ECD's forte in the cardiovascular compartment.

Cardiac involvement occurred in 48% of patients in a large French cohort; cardiac involvement had no significant impact on overall survival, but appeared to be more common in patients with BRAF^V600E mutations.[1] What is interesting is that although ECD tends to be more prevalent in males, there are approximately one third of patients that are female, as was observed in this patient, and this should not reduce the suspicion for ECD.

Pathophysiology: Recent studies have redefined ECD as not solely an inflammatory disease but rather as a clonal, hematopoetic disease.[4] It occurs when abnormal cells in the body multiply in an uncontrolled way in response to recurrent activating mutations of the MAPK pathway, most commonly BRAF^V600E, followed by MAP2K1, ARAF, KRAS and NRAS.[4,6] This molecular understanding has revolutionised diagnosis and therapy and has led to targeted drugs such as BRAF or MEK inhibitors being used to treat our patient, which would otherwise be proposed for therapy following a mutation analysis.

Histopathologically, ECD lesions consist of foamy CD68+ histiocytes in a fibrotic and inflammatory stroma, typically located in the long bones, retroperitoneum, cardiovascular system, orbits, lungs and brain.[4,5]

The ooze of these histiocytes causes the progressive damage, fibrosis and organ dysfunction depending on the site of disease. Osteosclerosis, retroperitoneal fibrosis and infiltration of the pericardium or eye have been common features reported.[4–6] However, ECD can be clonal with myeloid neoplasms, such as AML, indicating a shared progenitor in the bone marrow lineage.[4,6] The clinical trajectories could be different due to this overlap and the rare overlap with hematologic disorders could explain.

In summary, ECD is a complicated disorder involving neoplastic histiocytic proliferation, fibrosis and immune dysfunction. Our case, with extensive mediastinal involvement and retroperitoneal fibrosis along with pericardial effusion is illustrative of the multifaceted and systemic nature of the disease.

Diagnostics and Differentials: The diagnosis of Erdheim–Chester disease (ECD) can be difficult because of the rarity and commonality of the imaging and histopathologic findings with other systemic disease states. Possible differentials before diagnosis of ECD include IgG4-related disease, fibrosing mediastinitis, sarcomas, lymphomas, metastatic carcinomas and Langerhans cell histiocytosis.[3,6,12] In this patient, cross-sectional imaging revealed a large anterior mediastinal mass encasing the great vessels and a pericardial effusion, along with retroperitoneal and perirenal soft-tissue thickening showing the characteristic "hairy kidney" appearance on PET/CT. These findings strongly suggested multisystem involvement consistent with ECD.[3,6,12]

Histopathologic examination remains the gold standard for confirmation. The initial fine-needle aspiration yielded fibrous tissue with chronic inflammation but was nondiagnostic. A subsequent excisional biopsy of pericardial fat showed foamy, vacuolated histiocytes within a fibrotic stroma, a pattern typical of non-Langerhans cell histiocytosis. Immunohistochemistry demonstrated CD68-positive, CD1a-negative histiocytes, rare S100 reactivity, and nuclear cyclin D1 positivity, supporting the diagnosis of ECD.[13,14] Molecular testing for BRAF V600E was negative, and further next-generation sequencing was recommended to assess for other MAPK/ERK pathway mutations (MAP2K1, KRAS, NRAS).

Several other diseases were systematically ruled out during evaluation. Fibrosing mediastinitis was considered due to extensive mediastinal fibrosis, but the absence of dense keloid-type collagen and the presence of foamy histiocytes excluded this diagnosis. IgG4-related disease was ruled out because storiform fibrosis, plasma-cell–rich infiltrates, and IgG4-positive staining were not present. Lymphoma (T-cell, B-cell, or Hodgkin type) was excluded based on negative CD20, CD30, TdT, and flow cytometry results. Sarcoma and metastatic carcinoma were dismissed due to the absence of cytologic atypia and negative epithelial and mesenchymal markers. Langerhans cell histiocytosis was excluded based on negative CD1a and Langerin staining.[3,6,12]

Taken together, the integration of imaging findings, immunophenotypic profile ($CD68^+/CD1a^-/cyclin\ D1^+$), and expert pathologic review by the NIH Hematopathology Branch confirmed Erdheim-Chester disease as the unifying diagnosis.

Clinical Features: The long bones, especially of the femur and tibia, are usually affected by bilateral, symmetrical osteosclerotic lesions, and patients typically have ongoing bone pain in the skeletal sites affected, which is one of the most common complaints.[2,6,7] The patient did not complain of bone pain, but it is the most frequent skeletal symptom of ECD. Endocrine abnormalities also occur regularly.

Central diabetes insipidus is one of the most early and typical findings, which is associated with the involvement of the hypothalamic–pituitary axis.[7,15] Patients often have perirenal fibrosis (hairy kidney") and periaortic involvement (coated aorta"), suggestive of retroperitoneal and vascular tropism of the disease.[7,8] The PET/CT of our patient demonstrated classic perirenal infiltration with the "hairy kidney" sign which support these features.

Involvement of the cardiovascular system, namely pericardial fibrosis, effusion, myocardial and perivascular infiltration is seen in up to 75% of cases, and is a significant cause of morbidity and mortality.[8] Likewise, our patient had a large anterior mediastinal mass around the great vessels and was found to have a large effusion in the pericardium, reflecting ECD's cardiovascular system predilection.

The neurologic manifestations are variable and include cerebellar dysfunction, ataxia, cognitive impairment, cranial neuropathy and visual disturbances.[6,9] Almost half of the patients have CNS involvement, which is associated with worse outcomes and is responsible for many of the ECD related deaths.[8,10]

Other include bilateral exophthalmos, which may be associated

with xanthelasma around the eyelids.[3] In addition there may be systemic or "constitutional" symptoms of illness (fatigue, fever, muscle aches, and weight loss), especially in more advanced disease.[3,6] Fatigue and diminished appetite were the main early clinical features in our patients, similar to the other non-specific systemic features.

Complications: Erdheim–Chester disease (ECD) is a multisystem disorder, and the complications are dependent on the pattern and degree of involvement of the organs. Cardiovascular complications can be serious such as "coated aorta" (periaortic fibrosis), thickening of the pericardium, recurrent effusions and conduction defects and/or arrhythmias. Our patient had developed a large pericardial effusion which made it necessary to create a window and therefore cardiac involvement in ECD had the potential to be life-threatening.

Renal complications are also common and are usually due to a progressive hydronephrosis or obstructive uropathy caused by the perirenal fibrosis with the typical "hairy kidney" sign on imaging.[3,6,16] This discovery was clearly illustrated in our case, with a finding of infiltration of the retroperitoneum and perirenal infiltration on PET/CT scan.

Depending on central nervous system infiltration, neurological symptoms can develop such as central diabetes insipidus, cerebellar dysfunction, cranial neuropathies or stroke-like episodes. Note that our patient did not have any obvious neurological deficits, but will be monitored for CNS progression, a well-established cause of morbidity in ECD.[6,16] Lung involvement may cause pleural effusion, interstitial fibrosis, or restrictive lung disease, and although bones are rarely involved, any disease of the skeleton usually presents as symmetrical sclerotic lesions, causing a slow developing bone pain and mobility impairment.

Overall, cardiovascular or central nervous system involvement is the most common cause of death in ECD, and molecular diagnostics and multidisciplinary management are essential for early diagnosis.[3,16]

Treatment: In the past, interferon alpha and pegylated interferon have been used as first-line therapy in patients with multisystem Erdheim–Chester disease (ECD).[16] Without actionable mutations these agents have been shown to stabilize disease and extend lifespan. In recent years, however, improvements in molecular analysis have led to the development of targeted therapy as the preferred standard for therapy when genetic alterations are discovered.

Since our patient was BRAF V600E negative, there was a recommendation to perform next generation sequencing to check for other mutations of the MAPK/ERK pathway; including KRAS, NRAS, and MAP2K1.[13,14] Identifying these mutations is important because when they are identified in a patient, treatment decisions can be made which take the molecular findings into account. BRAF inhibitors - vemurafenib or dabrafenib in combination with trametinib - can be used for BRAF-mutant disease, while MEK inhibitors - cobimetinib or trametinib - can be used for MAP2K1-mutant ECD.[13,14]

Since the mutation profile was pending, our patient was treated conservatively, with symptomatic and supportive treatment. A pericardial window procedure was performed, and the effusion was found to be enlarged; the procedure was successfully performed to help by relieving the compression symptoms. They were delayed for systemic therapy until molecular diagnosis was confirmed, and the care was coordinated with oncology and cardiology services.

Adjunctive measures can also be of great value in improving the quality of life and may consist of short courses of treatment with corticoids for symptom control, bisphosphonates for bone pain and targeted surgical or percutaneous procedures to clear organ obstruction such as the ureteral stenting used for hydronephrosis. For the management of a multisystem disease such as ECD, multidisciplinary care with the participation of oncology, cardiology, nephrology and thoracic surgery is still necessary to achieve the best outcome and for long-term monitoring.[3,17]

Prognosis and Follow-up: The prognosis of Erdheim–Chester disease (ECD) is highly dependent on the level of organ involvement and the presence of mutations that can be targeted and used to treat. Those who have skeletal involvement but no other organ disease tend to have a better prognosis, while those who have cardiovascular or central nervous system involvement are at greater risk of morbidity and mortality.[3,14,16] Cardiac and CNS complications remain as the top causes of death, highlighting the need for early molecular testing and selection of therapy based on individual patients.

Overall survival has been significantly improved with the introduction of targeted agents like BRAF and MEK inhibitors, especially in those BRAF or MEK mutation positive. Long-term disease stabilization has also been reported in interferon-treated patients who are negative for the mutations.[3,14,16]

Our patient has been monitored closely with repetitive clinical, echocardiographic and follow up PET/CT imaging to assess disease activity, and he remains under active follow-up. Late relapse or progression is possible and lifelong monitoring is recommended, preferably in a multi-disciplinary team with working experience in histiocytic disorders.

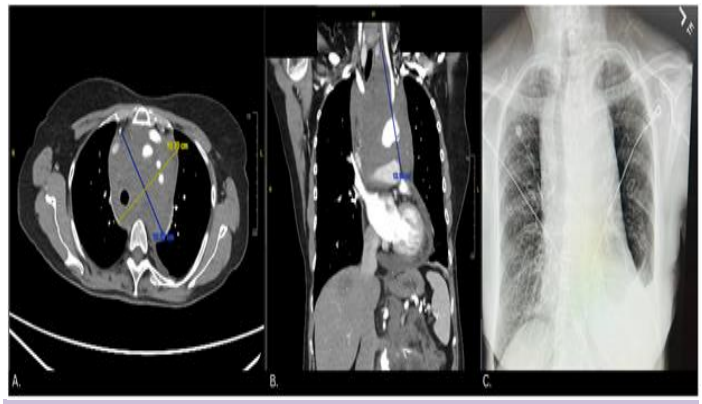

Figure 1: Thoracic Imaging Findings.

Axial (A) and coronal (B) contrast-enhanced computed tomography (CT) images of the chest show a large anterior and superior mediastinal soft-tissue mass encasing the great vessels and trachea with rightward deviation. The lesion measures approximately 10.7 × 10.5 × 13.1 cm in maximal dimensions. A chest radiograph (C) demonstrates a markedly widened mediastinum with clear lung fields, corresponding to the mediastinal mass.

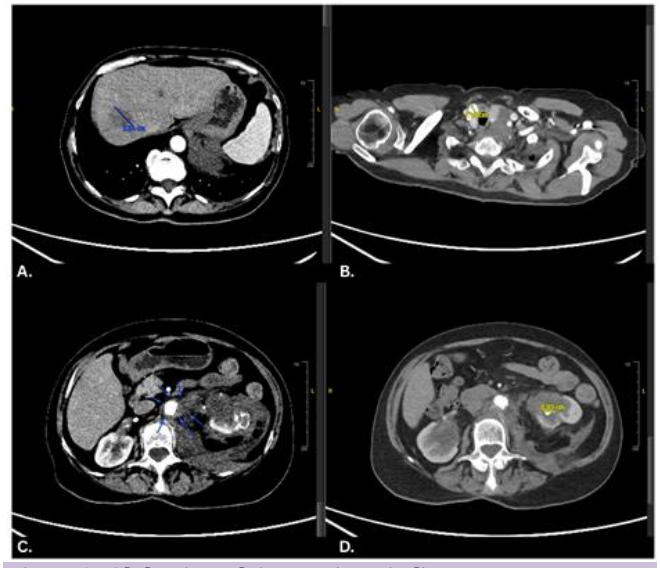

Figure 2: Abdominopelvic Imaging Findings.

Axial CT images reveal an infiltrative soft-tissue process along the left adrenal region and pararenal fascia, extending to the diaphragm and displacing the left kidney laterally (A). A small left supraclavicular lymph node measuring 1.0 cm is seen (B). Additional axial slices show perirenal soft-tissue stranding and ring-like enhancement (C) with thickening of the perirenal fascia up to 0.9 cm (D), findings consistent with the "hairy kidney" sign of systemic Erdheim–Chester disease (ECD).

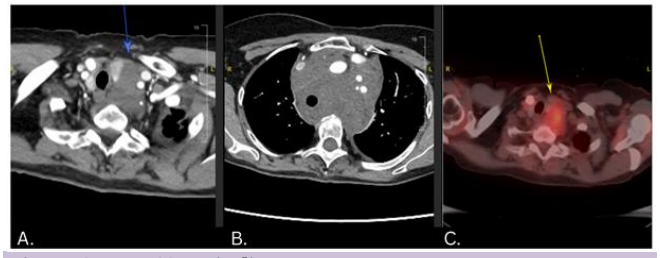

Figure 3: PET/CT Findings.

Axial contrast-enhanced CT (A, B) and fused ^18F-fluorodeoxyglucose positron emission tomography/CT (C) images demonstrate mild hypermetabolic activity in the left supraclavicular soft-tissue lesion (yellow arrow) and low-grade uptake in the mediastinal mass, consistent with metabolically active but indolent histiocytic infiltration in ECD.

Conclusion

This case highlights the value of the consideration of ECD as a cause of unexplained multisystems involvement, especially in the presence of characteristic radiological features, such as infiltration of perirenal soft tissue or pericardial effusion. Interpretation of multidisciplinary evaluation is crucial for an accurate diagnosis and appropriate management and should include imaging, histopathology and molecular testing. Supportive care, in the absence of mutation, can help prevent complications and enhance patient results when suffering from the disease is timely recognized.

Limitations: This report is the single case, which cannot be used as a general rule for the patients with Erdheim–Chester disease (ECD). The variety of presentations of ECD and its low prevalence further limits the interpretation of individual clinical observations. There was a lack of complete assessment of treatment outcomes due to pending molecular results and initiation of targeted therapy at the time of reporting and the absence of longer term follow-up data to assess stability or recurrence of treatment. Further, the case was handled as an inter-disciplinary clinical case, and some differences in the interpretation of imaging and/or histopathology interpretation were not standardized. In addition, there was no histopathology (HPE) image to include and visually demonstrate the typical ECD histiocytic infiltration and fibrosis. However, despite these constraints, the case adds to the available literature that includes many insights about the complexity of the diagnosis and clinical spectrum of ECD, especially when the clinical presentation is dominated by thoracic and retroperitoneal involvement.

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Nil.

Conflicts of interest

There are no conflicts of interest.

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