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Case Report

Comprehensive case reports on cardiac manifestations in Erdheim-Chester disease: Imaging and clinical insights ☆,☆☆,★

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ABSTRACT

Cardiac magnetic resonance imaging (cardiac MRI) is an essential tool in the diagnosis and managing cardiac pathology. This pictorial essay discusses 3 patient case examples used in clinical practice. The cases are representative of very rarely cardiac involvement in Erdheim-Chester disease, where advanced imaging techniques play a crucial role in identifying and evaluating sporadic manifestations in the heart. Cardiac MRI is invaluable in providing detailed structural and functional information. Essential for comprehensive cardiac assessment, it is crucial in guiding effective treatment strategies and improving the patient's outcome with some of the most complex and rare cardiac conditions.

Introduction

Erdheim-Chester disease (ECD) is a rare non-Langerhans cell histiocytosis characterized by the uncontrolled creation and first accumulation of histiocytes, which has caused infiltration in many other organ systems. Described by William Chester in 1930, this disease has less than 500 reported cases worldwide [1,2]. The effect of the multiplication of lipid-laden histiocytes infiltrates many organs in ECD, which, in turn, leads to many associated manifestations of this disease [3,4]. Although cardiac involvement is rare, it represents clinically challenging problems that increase mortality and morbidity. Late diagnosis is expected due to these nonspecific symptoms and the condition's rarity, underpinning the need for increased awareness and understanding among clinicians.

☆ The authors agree that the material presented in this paper has not been published before, nor has it been submitted for publication to another scientific journal or considered for publication elsewhere.

☆☆ I attest that this work has been approved by all co-authors.

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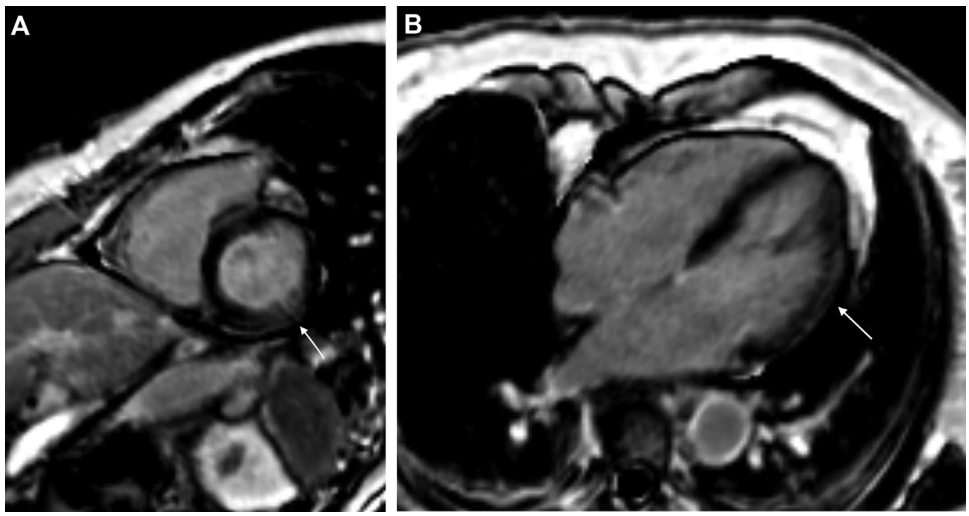

Fig. 1 and 2 – There is a minimal amount of patchy late enhancement seen in the basal, middle, and apical parts of the lateral wall of the heart.

Cardiac manifestations of ECD include pericardial effusion, myocardial infiltration, and involvement of the great vessels, among others, which may culminate in heart failure, arrhythmias, and other serious mishaps [5–7]. Most of these manifestations are identified with the help of advanced imaging techniques including cardiac magnetic resonance imaging (cardiac MRI) and 18F-Fluordesoxyglucose Positron emission tomography with computed tomography (PET-CT), providing detailed insights into the degree and nature of cardiac involvement. This pictorial article will use the cardiac manifestations observed in findings through cardiac MRI in 3 patients with ECD to allow visualization of cardiac characteristics and how each of those is correlated with clinical application.

Case presentations

Patient 1

Patient background

A 45-year-old male presented with fatigue, dyspnea, and chest discomfort. His medical history was significant for arterial hypertension, hyperlipidemia, type 2 diabetes mellitus, osteopenia, and vitamin D deficiency. The patient had been managing these chronic conditions with medications and lifestyle modifications, yet he experienced a decline in his overall health over the past year. The fatigue had progressively worsened, limiting his daily activities and quality of life. Dyspnea was initially exertional but became more persistent, occurring even at rest. The chest discomfort was intermittent and did not follow a typical anginal pattern, prompting further investigation into its etiology.

Diagnostic findings

Cardiac MRI findings

The cardiac MRI revealed typically arranged heart chambers with no congenital or structural abnormalities. However, there was minimal patchy late enhancement in the basal, middle, and apical parts of the lateral wall. This finding was indicative of myocardial infiltration by histiocytes, which is characteristic of Erdheim-Chester Disease (ECD). Importantly, there was no myocardial edema or significant movement disorders, which suggested a chronic infiltrative process rather than an acute inflammatory 1. A small pericardial effusion measuring 15mm was observed, but it did not impair cardiac function. The patient's ejection fraction (EF) was 65%, with an end-diastolic volume (EDV) of 83.6 ml/m^2 and an end-systolic volume (ESV) of 29 ml/m^2, indicating preserved cardiac function despite the infiltrative disease (Figs. 1 and 2).

PET-CT findings

The PET-CT scan provided additional insights into the extent of disease involvement. There was wall uptake at the origin of the left subclavian artery with a maximum standardized uptake value (SUV) of 4, suggesting the presence of inflammatory activity. (Figs. 3 and 4) A small pericardial effusion and focal uptake in the pancreatic tail area (SUV 7) were observed, indicating focal pancreatitis potentially related to ECD. The "hairy kidney sign," characterized by perinephric fat infiltration with condensations surrounding the kidneys (SUV 5), was also noted, a classic finding in ECD. (Figs. 5 and 6) Additionally, there was intraspinal epidural uptake at L5/S1 with minor focal uptake in the L1 vertebral body, highlighting the extent of skeletal involvement.

Case discussion

The diagnosis of Erdheim-Chester Disease was confirmed through a bone biopsy, which revealed the presence of foamy histiocytes. Genetic testing identified the BRAF V600E mutation, a common mutation in ECD that has significant therapeutic implications. The patient was started on targeted therapy with BRAF inhibitors, which are designed to inhibit the abnormal signaling pathway caused by this mutation. The re-

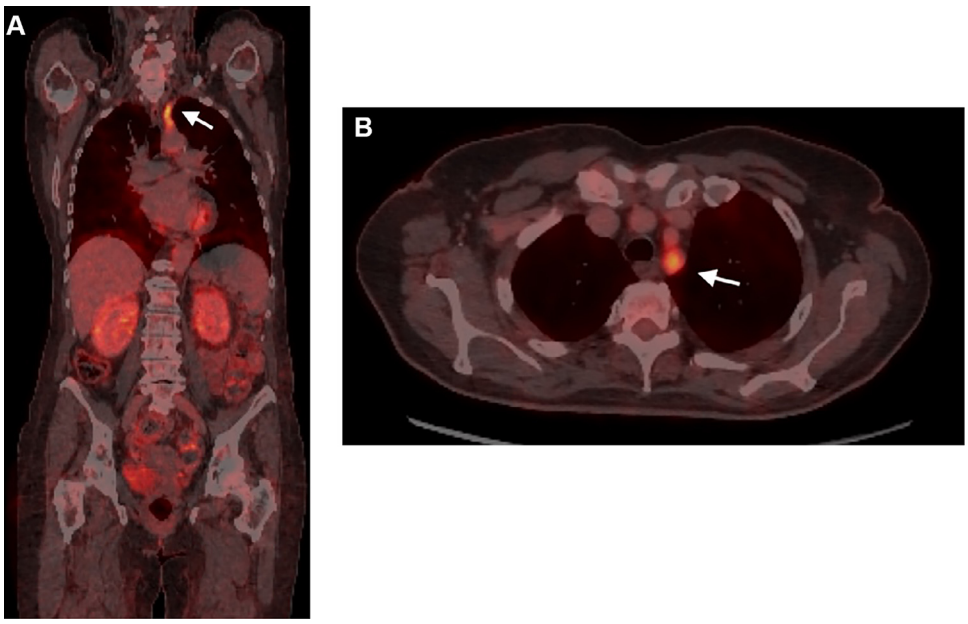

Fig. 3 and 4 – There was wall uptake at the origin of the left subclavian artery with a maximum standardized uptake value (SUV) of 4, suggesting the presence of inflammatory activity.

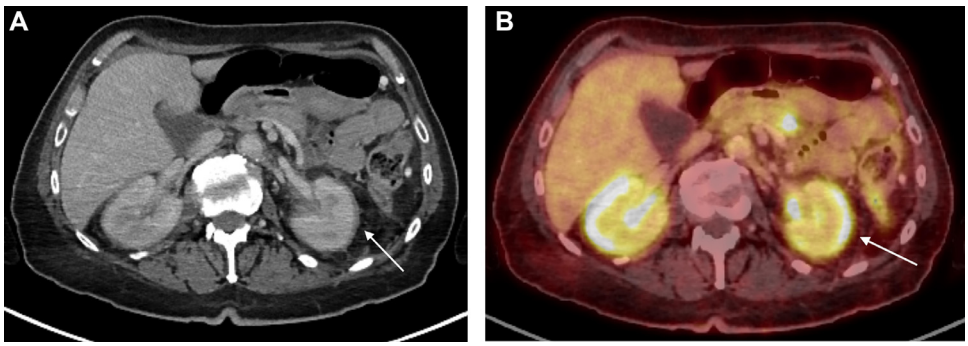

Fig. 5 and 6 – A typical "hairy kidney" sign was seen, reflecting perinephric fibrosis, a characteristic of ECD.

sponse to treatment was monitored closely through regular follow-ups and imaging studies.

The multidisciplinary approach to the patient's care was crucial for managing the complexities of ECD. The team included specialists from cardiology, endocrinology, and oncology, who worked together to address the various aspects of the disease. Cardiology focused on monitoring and managing the cardiac involvement, ensuring that the small pericardial effusion and myocardial infiltration did not progress to impair cardiac function. Endocrinology managed the patient's metabolic conditions, which were exacerbated by the systemic nature of ECD. Oncology led the treatment with BRAF inhibitors, providing targeted therapy to address the underlying genetic mutation.

Regular follow-up appointments were scheduled to monitor the patient's response to treatment and detect any signs of disease progression. Imaging studies, including repeat cardiac MRI and PET-CT scans, were conducted periodically to assess the effectiveness of the therapy and adjust the treatment plan as necessary. The patient showed promising results with the BRAF inhibitor therapy, with stabilization of the disease and improvement in symptoms.

This case underscores the importance of advanced imaging techniques in the diagnosis and management of Erdheim-Chester Disease with cardiac involvement. The use of cardiac MRI was pivotal in detecting the myocardial infiltration that was not apparent through routine cardiac evaluations. The PET-CT scan provided a comprehensive overview of the systemic involvement, highlighting the multisystem nature of ECD.

The identification of the BRAF V600E mutation was a critical factor in confirming the diagnosis and guiding the therapeutic approach. Targeted therapy with BRAF inhibitors has shown to be effective in managing ECD, offering a promising prognosis for patients with this mutation. The patient's case exemplifies the necessity of a coordinated and comprehen-

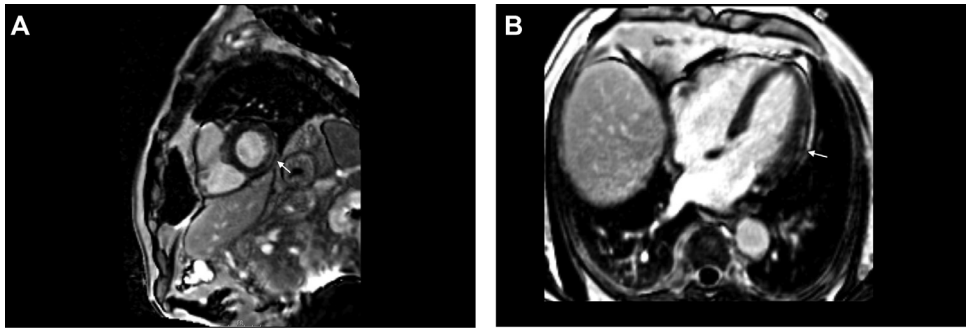

Fig. 7 and 8 – Mid-myocardial late enhancement suggests myocardial infiltration by histiocytes, which is typical of cardiac involvement in ECD.

sive approach to management, involving multiple specialties to address the various aspects of the disease.

The multidisciplinary team's involvement ensured that all facets of the patient's condition were addressed, from cardiac function to metabolic control and oncologic treatment. This integrated approach not only improved the patient's quality of life but also provided a framework for managing similar cases in the future. The regular follow-ups and imaging studies were essential in monitoring the disease progression and treatment response, allowing for timely adjustments to the therapeutic regimen.

In conclusion, this case highlights the complexity of Erdheim-Chester Disease and the critical role of advanced imaging and targeted therapy in its management. The successful coordination of care across multiple specialties serves as a model for the comprehensive treatment of rare and multisystem diseases.

Patient 2

Patient background

A 62-year-old female presented with progressive dyspnea, leg swelling, and fatigue. Her medical history included hyperlipidemia and hypothyroidism. The patient's clinical symptoms prompted a detailed investigation to determine the underlying cause and to assess the extent of her condition.

Diagnostic findings

Cardiac MRI findings

The cardiac MRI revealed normally sited cardiac chambers without pericardial effusion. However, significant mid-myocardial late enhancement in multiple segments was observed, indicating myocardial infiltration by histiocytes. This finding is suggestive of Erdheim-Chester Disease (ECD), a rare form of non-Langerhans cell histiocytosis. Importantly, there was no myocardial edema present, which implies a lack of active inflammation. The mechanical function of the heart was preserved, with an ejection fraction (EF) of 67%, end-diastolic volume (EDV) of 70 mL/m^2, and end-systolic volume (ESV) of 23 mL/m^2. These values indicate that, despite the infiltration, the heart's pumping ability remained within normal limits (Figs. 7 and 8).

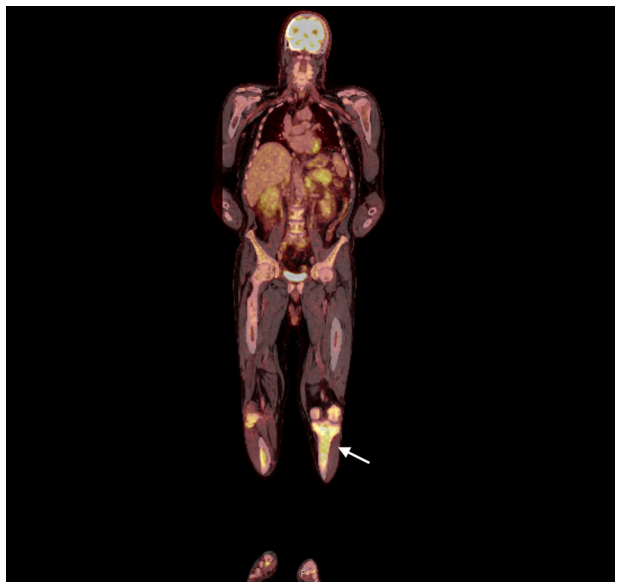

Fig. 9 – In the PET-CT scan, there was diffusely markedly increased tracer uptake in the distal femur, tibia, and tarsal bones bilaterally, with an SUVmax up to 6.0, indicating significant skeletal involvement.

PET-CT findings

The PET-CT scan showed diffusely increased tracer uptake in the distal femur, tibia, and tarsal bones bilaterally (SUVmax 6.0), signifying significant skeletal involvement, a common feature of ECD. Additionally, there was mild diffuse increased bone marrow tracer uptake and both osteolytic and sclerotic changes around the right knee. (Fig. 9) Structural changes at the proximal right femur were noted, alongside slight increased focal tracer uptake at the upper right hilus and paravertebral soft tissue proliferation in the caudal mediastinum. These findings pointed to the widespread involvement of the disease, extending beyond the skeletal system to include other soft tissues and organs.

Case discussion

ECD was confirmed with significant involvement of multiple organs including the pancreas, kidneys, bones, thoracic

and abdominal aorta, and adrenal glands. The patient was started on a combination of corticosteroids and immunosuppressive therapy to manage the systemic manifestations of ECD. Follow-up echocardiography showed moderate left atrial dilation but normal global systolic function with an EF of 60%. The patient's condition required careful and coordinated management by a multidisciplinary team, focusing on controlling the disease's systemic effects while continuously monitoring cardiac function.

This case underscores the variability in ECD manifestations and the critical role of multimodal imaging in providing a comprehensive assessment. The cardiac MRI and PET-CT scans were instrumental in delineating the extent of myocardial and skeletal involvement, thereby guiding the therapeutic approach. The absence of myocardial edema was a crucial finding, suggesting that there was no active inflammation at the time of diagnosis, which influenced the choice of treatment. Managing ECD involves addressing both the immediate symptoms and the long-term effects of organ infiltration. In this patient, the combination of corticosteroids and immunosuppressive therapy aimed to reduce histiocytic infiltration and control systemic disease activity.

This case also highlights the importance of a multidisciplinary approach in managing complex conditions like ECD. Cardiologists, radiologists, and other specialists must work together to interpret the findings from various imaging modalities and to develop a cohesive treatment plan. Monitoring the patient's response to treatment and adjusting the therapeutic strategy accordingly is essential for optimizing outcomes. In summary, the detailed imaging findings and coordinated care approach were pivotal in managing this patient's ECD, emphasizing the necessity for a thorough and integrated diagnostic and therapeutic strategy in such rare and multifaceted diseases.

Patient 3

Patient background

A 50-year-old male presented with new-onset chest pain and palpitations. He had a significant medical history, including chronic back pain, fatigue, and recurrent respiratory infections. His symptoms of back pain had been persistent for several years, often managed with analgesics without a definitive diagnosis. The fatigue was progressive and debilitating, affecting his daily activities and quality of life. The recurrent respiratory infections, characterized by frequent bouts of bronchitis and sinusitis, had led to multiple courses of antibiotics over the years. These recurrent infections raised concerns about an underlying immune dysfunction, but previous evaluations had not identified a specific cause.

Diagnostic findings

Cardiac MRI findings

The cardiac MRI revealed a normally arranged cardiac chamber configuration without any congenital or structural abnormalities. However, there was patchy late gadolinium enhancement observed in the lateral wall of the heart. This finding was indicative of myocardial infiltration by histiocytes, a hallmark of Erdheim-Chester Disease (ECD). The ejection fraction (EF) was measured at 66%, indicating that the patient's cardiac function was within normal limits. The end-diastolic volume (EDV) was 75 ml/m^2, and the end-systolic volume (ESV) was 26 ml/m^2, further suggesting preserved cardiac function. Importantly, there were no signs of myocardial edema or movement disorders, which would have suggested acute inflammation or significant myocardial compromise. (Figs. 10 and 11).

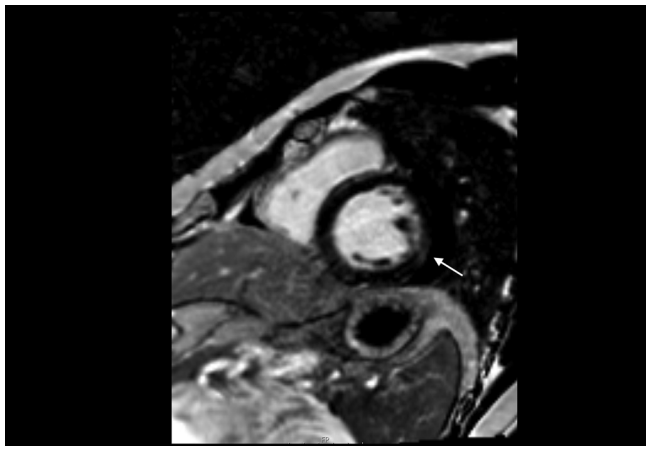

Fig. 10 – There is also patchy late enhancement in the lateral wall of the heart, a significant finding strongly indicative of ECD.

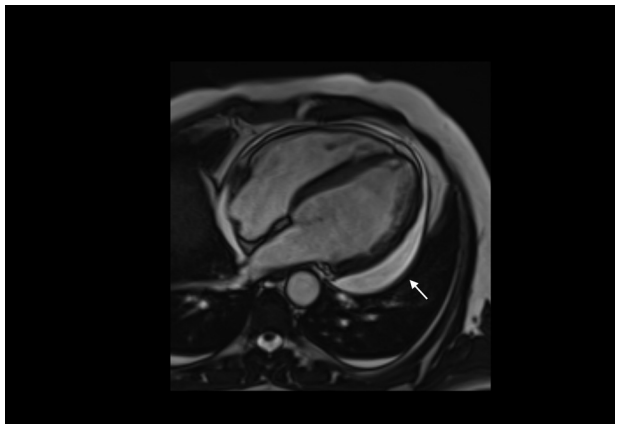

Fig. 11 – Cardiac manifestations of ECD include pericardial effusion.

PET-CT findings

The PET-CT scan provided a comprehensive assessment of the extent of disease involvement. There was increased tracer uptake noted in the distal femur, tibia, and tarsal bones, indicating marked skeletal involvement typical of ECD. This skeletal uptake suggested active disease with significant bone marrow infiltration. No abnormal liver uptake was observed, effectively ruling out hepatic involvement. However, there was increased uptake at the of the pancreas, a finding consistent with ECD-related pancreatic involvement. (Fig. 12) Additionally, there was intraspinal epidural uptake at the L5/S1 level and focal uptake in the L1 vertebral body, indicative of spinal

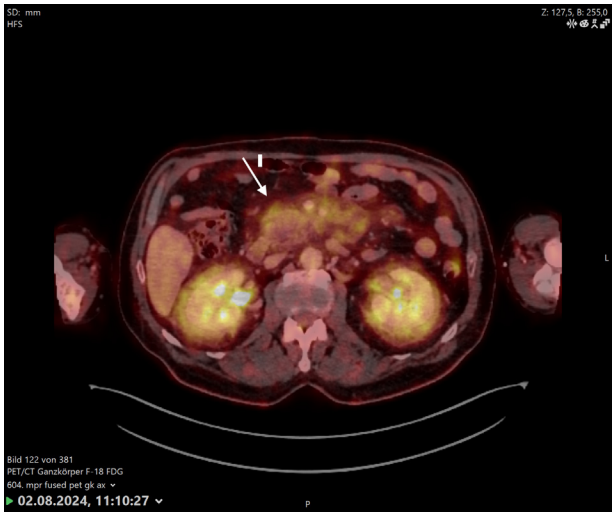

Fig. 12 – There was increased uptake at the of the pancreas, a finding consistent with ECD-related pancreatic involvement.

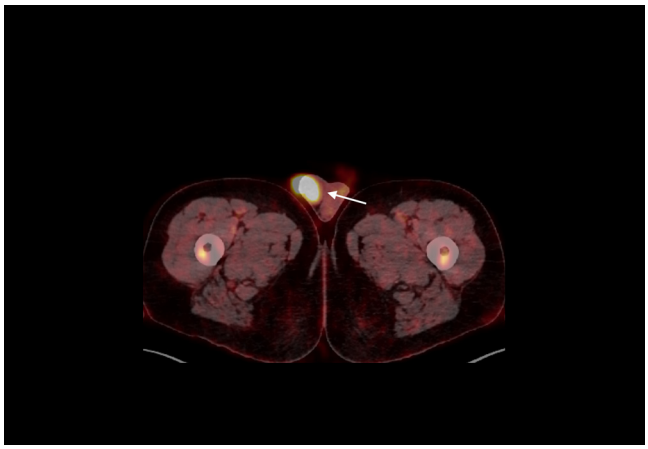

Fig. 13 – Increased tracer uptake in the testes demonstrated another characteristic feature of ECD.

involvement by the disease. Increased tracer uptake in the testes demonstrated another characteristic feature of ECD. (Fig. 13)

Case discussion

Erdheim-Chester Disease with prominent cardiac and skeletal involvement was confirmed through histological examination. The biopsy revealed infiltration with foamy histiocytes and areas of fibrosis, consistent with the pathology of ECD. The presence of these histiocytes, which are lipid-laden and have a foamy appearance, confirmed the diagnosis. The patient was started on a combination of corticosteroids to reduce inflammation and kinase inhibitors targeting the identified mutations. These targeted therapies were essential in managing the disease's progression and mitigating its multisystem effects.

Echocardiography, used to complement the imaging findings, showed normal valve morphology and function. However, there was moderate left atrial dilation, which could be attributed to the chronic cardiac strain imposed by the infiltrative process. Despite this, the global systolic function of the heart remained preserved, underscoring the importance of early intervention and management.

This case highlights the critical role of early diagnosis and continuous monitoring in patients with Erdheim-Chester Disease, especially those with cardiac involvement. Advanced imaging techniques, such as cardiac MRI and PET-CT, were instrumental in providing a comprehensive evaluation of the disease's extent and guiding the therapeutic approach. The cardiac MRI was particularly valuable in identifying myocardial infiltration, which is often challenging to diagnose due to the nonspecific nature of symptoms like chest pain and palpitations.

The PET-CT scan's ability to detect widespread skeletal and organ involvement emphasized the systemic nature of ECD. The characteristic imaging findings, such as the "hairy kidney" sign and focal skeletal uptake, provided critical diagnostic clues that corroborated the histological examination results.

The treatment plan, including corticosteroids and kinase inhibitors, was tailored based on the imaging and histological findings. The presence of foamy histiocytes and fibrosis on histological examination not only confirmed the diagnosis but also provided insight into the disease's chronicity and the extent of tissue damage. This information was crucial in determining the intensity and duration of the treatment regimen.

Overall, this case underscores the importance of a multidisciplinary approach in managing Erdheim-Chester Disease. The collaboration between cardiology, radiology, pathology, and oncology was pivotal in ensuring comprehensive care for the patient. The advanced imaging techniques facilitated an accurate diagnosis, while the targeted therapies addressed the underlying genetic mutations, improving the patient's prognosis and quality of life.

Early recognition of cardiac involvement in ECD, as demonstrated in this case, is vital for preventing complications such as heart failure and arrhythmias. Continuous monitoring through regular follow-ups and imaging studies is essential to assess the treatment response and adjust the therapeutic strategy accordingly. This approach ensures optimal management of this rare and complex disease, highlighting the advancements in imaging and targeted therapy that have significantly improved patient outcomes.

Discussion

Erdheim-Chester disease is a genetic disorder associated with rare, non-Langerhans cell histiocytosis. It features the abnormal development and succeeding proliferation of an unusual population of histiocytes, a white blood cell variety. Clinical manifestations are highly relative due to the histiocytes infusing a relatively broad range of tissues and organs in the body [8]. Although cardiac involvement in ECD may be rela-

tively rare, it represents a crucial portion of multiorgan participation in the disease, requiring a meticulous and comprehensive evaluation with advanced imaging techniques like cardiac MRI and PET-CT [9,10]. We discuss the varied manifestations of ECD on the heart, the role of advanced cardiac imaging as part of diagnosis and ECD management, and the treatment implications of ECD driven by specific genetic mutations.

The cardiovascular manifestations of ECD are highly variable and complex, including myocardial infiltration, massive pericardial effusion, and aortitis. Cardiac symptoms are of importance since they usually have implications for the final prognosis and quality of life of the patients. In this scenario, cardiac MRI and PET-CT are becoming essential advanced imaging techniques in the detection and characterization of such changes in cardiomyopathy changes.

It is characterized as myocardial infiltration with histiocytes. The late gadolinium enhancement in this region could happen because of fibrosis or active inflammation. Myocardial late enhancement in this region reflects significant histiocytic infiltration in these patients. This myocardial involvement can lead to restrictive cardiomyopathy, which is a condition where the walls of the heart become rigid, impairing it from dilating and filling blood typically. Thus, heart failure and arrhythmia may result; these underpin the need for regular monitoring of the heart in such patients.

Pericardial involvement in ECD can be severe cardiac involvement, with effusion usually being the uppermost presentation of the disease [10]. For the cases that are being discussed, both Patient 1 and Patient 2 presented with a pericardial effusion in the context of a patient who had experienced a series of clinical imperfections due to this diagnosis. The presentation of pericardial effusion may start mildly, but on the amount and rate of the accumulation, it may necessitate fungal contamination or even surgery for severe cases [11]. Timely diagnosis of pericardial effusion towards proper intervention and management of the condition to avert further serious complications, such as cardiac tamponade, is thus essential by cardiac MRI or echocardiography. Cardiac tamponade is a life-threatening condition in which fluid accumulation leads to pressure on the heart, therefore reducing its ability to work correctly.

Advanced imaging studies can demonstrate another manifestation of ECD with aortitis [12,13]. If not diagnosed and treated, the potential emergencies involve aneurysms or aortic dissection. In addition, the “hairy kidney” sign for perinephric fibrosis was present under imaging studies in all patients. This is even detectable on PET-CT due to increased perinephric tracer uptake and is pathognomonic for ECD. Perilymphatic infiltration into the renal capsule with a denuding appearance due to the end-stage desquamation of the overlying cortex is referred to as the “hairy kidney” sign [14,15]. Since using potassium compounds by the septum pellucidum allows for more detail in histological sections, an oblique slice may reveal local thickenings of the leptomeninges that are of great use in targeting the biopsy candidate.

Echocardiography is valuable in the initial assessment and follow-up of cardiac involvement by ECD [16]. The described cases have an echocardiographic complement to MRI results with further insights into cardiac function and morphology. The moderate left atrial dilation and normal systolic function in these patients agree with cardiac manifestations known with ECD histories. Echocardiography is of particular value in estimating valvular function and chamber size, being noninvasive and easily applicable for routine assessments in ECD patients.

A diagnosis of ECD in all 3 patients was confirmed on histological examination that showed characteristic features of the disease, including foamy histiocytes and fibrosis [17]. Of particular interest is identifying the BRAF V600E mutation in patient 1—this mutation is noted in about 50% of ECD cases and has critical therapeutic implications for targeted therapy. Vemurafenib, for example, which inhibits this oncogene, has been very inspiring in treating ECD via direct targeting of such aberrant signaling. Therefore, confirmation of the proper diagnosis with a BRAF V600E mutation presents a unique opportunity with a significant impact on treatment options and leads to improved patient outcomes.

There is no doubt that imaging will play the most prominent part in the diagnosis and management of ECD. While cardiac MRI yields images of excellent resolution for the structure and function of the heart, the technique enables the assessment of myocardial infiltration, fibrosis, and pericardial effusion. On the other hand, PET-CT provides metabolic information that may guide the recognition of active inflammation and subsequent assessment of disease activity. Used together, imaging with Single-photon emission computed tomography (SPECT) and PET overall assesses cardiac involvement by ECD, allowing early detection and an accurate diagnosis, with the prospects of successful management of the disease.

In summary, cardiac involvement with ECD entails an uncommon but essential aspect for multiorgan participation in this disease. The multifaceted nature of cardiovascular manifestations—myocardial infiltration, pericardial effusion, and aortitis—together reflects the necessity for due imaging in the diagnostic and management process of ECD. Appropriately, this is the guide for making informed clinical decisions toward better outcomes using cardiac MRI and PET-CT, 2 critical images for the modern age that diagnose these manifestations. So, cardiac follow-up is of prime importance for managing all the potential complications that cardiac involvement can cause in ECD, ensuring timely interventions for the optimal care of such patients with such a rare and troublesome disease. Genetic mutations, for instance, BRAF V600E, further pinpoint a potential target for curative therapy and bring new hope for better treatment strategies and improved quality of life in affected individuals.

Multidisciplinary treatment for cardiac involvement in ECD includes cardiologists, radiologists, oncologists, and pathologists. Therapeutic options for ECD include interferon-alpha, targeted therapies such as BRAF and Mitogen-aktivierte Proteinkinase-Kinase (MEK) inhibitors, corticosteroids, and cytotoxic agents; however, treatment decisions are according to the extent of disease involvement, the presence of the BRAF mutation and patient comorbidities [18–20].

In conclusion, ECD is a sporadic disorder and is, hence, diagnostically challenging, with very heterogeneous manifestations. Cardiac involvement is uncommon, but the clinical outcomes might be terrible. Thus, this pictorial essay underscores cardiac MRI and PET-CT are essential for early diagnosis and comprehensive assessment of cardiac involvement

in ECD. The detailed imaging and clinical correlation of the present report will add to the existing knowledge of cardiac involvement in ECD and thus enable timely diagnosis with appropriate management of this rare disease.

In other words, the importance of recognition of cardiac manifestations of ECD lies in timely and appropriate treatment. Advanced imaging modalities like cardiac MRI and PET-CT identify the degree of cardiac involvement and hence guide therapeutic decisions. Our understanding of this enigmatic disorder continues to evolve; therefore, further research and clinical collaboration are needed if we hope to ameliorate patient prognosis and find better ways to treat them.

Management of ECD with cardiac involvement requires a multidisciplinary approach, involving cardiologists, radiologists, oncologists, and pathologists. Treatment options for ECD include interferon-alpha, targeted therapies such as BRAF and MEK inhibitors, corticosteroids, and cytotoxic agents. The choice of therapy depends on the extent of disease involvement, presence of the BRAF mutation, and patient comorbidities.

Conclusion

Erdheim-Chester disease is a sporadic disorder and is, hence, diagnostically challenging, with very heterogeneous manifestations. Cardiac involvement is uncommon, but the clinical outcomes might be terrible. Thus, this pictorial essay underscores cardiac MRI and PET-CT are essential for early diagnosis and comprehensive assessment of cardiac involvement in ECD. The detailed imaging and clinical correlation of the present report will add to the existing knowledge of cardiac involvement in ECD and thus enable timely diagnosis with appropriate management of this rare disease.

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Patient consent

We confirm that written informed consent was obtained from all individual participants (or their legal guardians) involved in the study for the publication of this manuscript and any accompanying images, data, or anonymized details.

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