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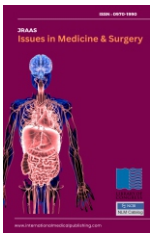

Case Report

Section: Pathology

Erdheim-Chester Disease Presenting as Refractory Anemia: A Rare Case Report from Central India

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HIGHLIGHTS

- Rare congenital uterine anomaly.
- MRI confirmed uterus didelphys.
- Successful pregnancy management
- Favorable maternal neonatal outcomes.
- Early diagnosis improves outcomes.

Key Words:

Erdheim-Chester disease
Non-Langerhans histiocytosis
Bone marrow biopsy
Immunohistochemistry
Cd68
BRAF mutation

ABSTRACT

Introduction: Erdheim–Chester disease (ECD) is a rare non-Langerhans cell histiocytosis characterized by multisystem infiltration of foamy histiocytes and progressive fibrosis. Owing to its heterogeneous clinical presentation and rarity, diagnosis is often delayed and requires histopathological and immunohistochemical confirmation. **Aim & Objectives:** To report a rare case of Erdheim–Chester disease presenting with refractory anemia, highlighting the role of bone marrow biopsy and immunohistochemistry in diagnosis, and emphasizing the importance of early molecular evaluation for targeted therapy and improved patient outcomes. **Case Presentation:** A 47-year-old female presented with generalized weakness, easy fatigability, intermittent diarrhea, insomnia, and refractory anemia. Routine laboratory and radiological investigations were inconclusive. Peripheral smear revealed normocytic normochromic anemia. Bone marrow aspiration was inadequate due to fibrosis, following which a trephine biopsy was performed. Histopathological examination showed diffuse infiltration by spindle-shaped and foamy histiocytes associated with stromal fibrosis. Immunohistochemistry demonstrated positivity for CD68 and CD163, while CD1a and Langerin (CD207) were negative, excluding Langerhans cell histiocytosis. CD3 highlighted admixed reactive T-lymphocytes. Based on histomorphological and immunophenotypic findings, a diagnosis of Erdheim–Chester disease was established. The patient received supportive treatment and was advised molecular testing for BRAF mutation analysis and further systemic evaluation for targeted therapy planning. **Results:** Bone marrow biopsy with immunohistochemistry confirmed Erdheim–Chester disease, showing CD68 and CD163 positivity with negative CD1a and CD207. The patient received supportive treatment and was advised BRAF mutation analysis for targeted therapy. **Conclusion:** This case highlights the importance of considering Erdheim–Chester disease in patients presenting with unexplained constitutional symptoms and infiltrative marrow pathology. Bone marrow biopsy with immunohistochemistry plays a pivotal role in diagnosis. Early recognition and molecular evaluation are essential for timely initiation of targeted therapy and improved clinical outcomes.

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INTRODUCTION

Erdheim–Chester disease (ECD) is a rare, non-familial, non-Langerhans cell histiocytosis characterized by the infiltration of lipid-laden (foamy) histiocytes into multiple organ systems, leading to progressive tissue damage and fibrosis. First described by William Chester in 1930, ECD has since been recognized as a distinct clinicopathological entity within the spectrum of histiocytic disorders. According to the revised classification by the World Health Organization (WHO), ECD is categorized under the "L" (Langerhans-related) group of histiocytoses due to shared molecular features, despite its distinct immunophenotypic profile [1,2].

The pathogenesis of ECD has evolved from being considered a purely inflammatory condition to a clonal hematopoietic disorder driven by mutations in the mitogen-activated protein kinase (MAPK) pathway. The most frequently identified mutation is BRAF V600E, present in approximately 50–60% of cases, followed by mutations in MAP2K1, KRAS, and NRAS genes [3-5]. These discoveries have not only enhanced understanding of disease biology but also opened avenues for targeted therapies. Clinically, ECD exhibits a highly heterogeneous presentation, often involving the long bones, cardiovascular system, retroperitoneum, central nervous system, and lungs. Skeletal involvement, particularly bilateral symmetric osteosclerosis of long bones, is considered a hallmark finding, although extra-skeletal manifestations frequently dominate the clinical picture [6,7].

Patients may present with non-specific symptoms such as fatigue, weight loss, or endocrine abnormalities, contributing to diagnostic delay.

Histopathologically, ECD is characterized by xanthogranulomatous infiltration composed of foamy histiocytes, fibrosis, and occasional Touton giant cells. Immunohistochemistry plays a critical role in differentiating ECD from other histiocytic disorders, with typical positivity for CD68 and CD163 and negativity for CD1a and Langerin (CD207) [8]. This immunophenotypic distinction is essential to exclude Langerhans cell histiocytosis.

Radiological evaluation, including computed tomography (CT), magnetic resonance imaging (MRI), and positron emission tomography (PET), is indispensable in assessing the extent of systemic involvement. Characteristic findings such as "coated aorta" and "hairy kidney" sign further aid in diagnosis [9,10].

Given its rarity and diverse manifestations, ECD often poses significant diagnostic challenges. Early recognition is crucial, as therapeutic strategies have evolved considerably, with interferon-alpha historically used as first-line therapy, and targeted agents such as BRAF and MEK inhibitors now demonstrating substantial clinical benefit [11-13]. Bone marrow biopsy showing histiocytic infiltration and fibrosis **(Figure 1)**.

This case report highlights the importance of a multidisciplinary diagnostic approach integrating clinical, histopathological, and immunohistochemical findings in identifying Erdheim–Chester disease, particularly in atypical presentations.

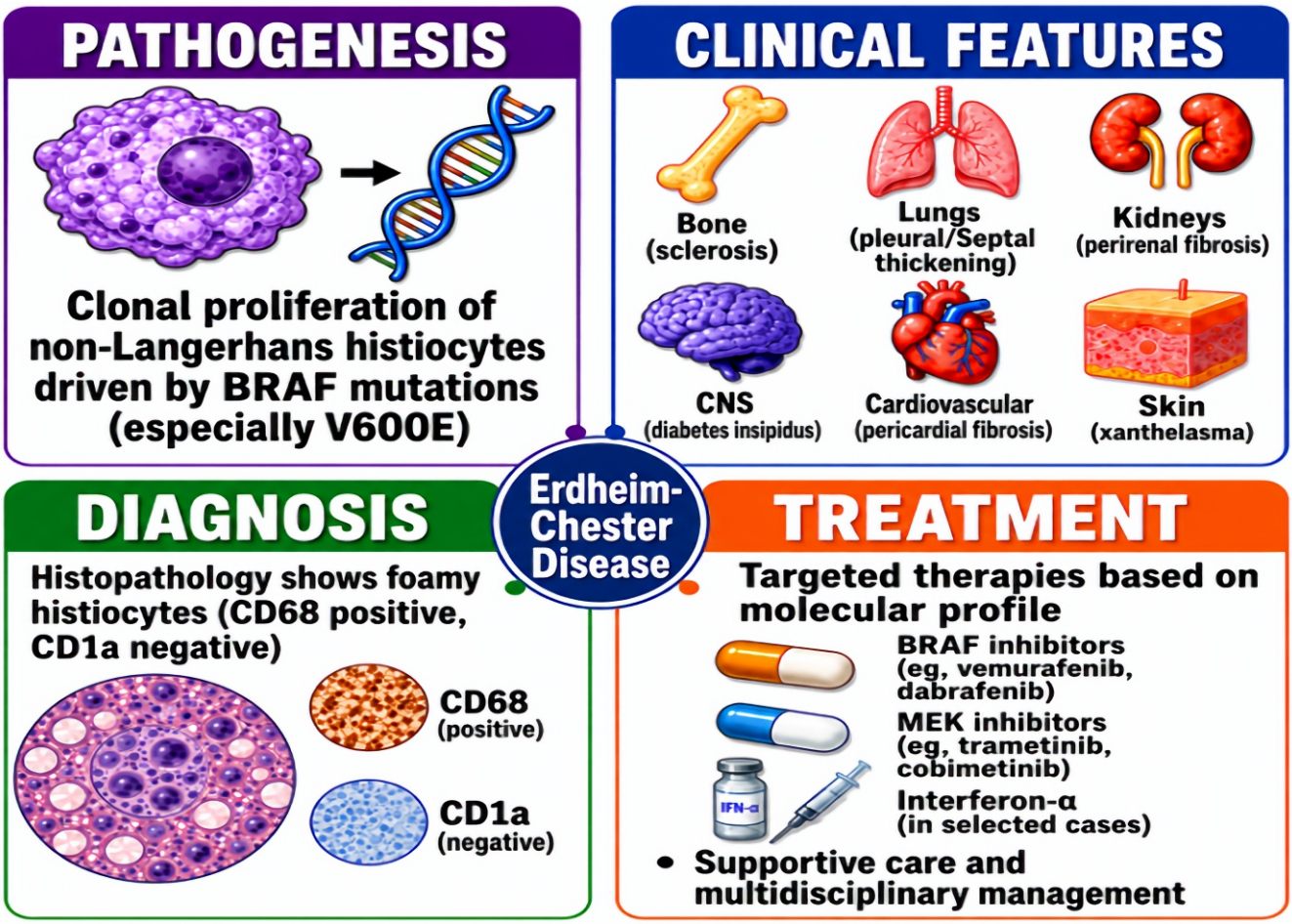

Figure 1: Bone marrow biopsy showing intertrabecular histiocytic infiltration with associated fibrosis.

CASE PRESENTATION

A 47-year-old female presented to the outpatient department with complaints of generalized weakness, easy fatigability, intermittent episodes of loose stools, and persistent insomnia for several months. The patient also reported progressive malaise and reduced exercise tolerance, which had gradually worsened over time. There was no significant history of fever, chronic cough, weight loss, night sweats, bleeding manifestations, or known autoimmune disease. The patient denied any history of tuberculosis, malignancy, chronic liver disease, alcohol abuse, or exposure to toxic agents. Her past medical history was unremarkable, and there was no significant family history of hematological or autoimmune disorders.

On physical examination, the patient appeared pale and mildly cachectic. Vital parameters were stable at presentation. Systemic examination revealed pallor without significant lymphadenopathy or hepatosplenomegaly. Cardiovascular, respiratory, and neurological examinations were largely unremarkable. Initial clinical suspicion included chronic inflammatory disease, hematological malignancy, or infiltrative bone marrow disorder. The patient had previously undergone multiple radiological and laboratory investigations at peripheral healthcare centers to evaluate refractory anemia; however, no definitive diagnosis could be established. Due to persistent symptoms and unexplained anemia, she was referred for further hematological evaluation.

Investigations

Baseline hematological investigations revealed persistent anemia with reduced hemoglobin levels. Peripheral smear examination showed normocytic normochromic red blood cells with no evidence of hemolysis or abnormal circulating cells. Total leukocyte count and platelet counts were within normal limits. Biochemical investigations, including liver function tests, renal function tests, serum electrolytes, and inflammatory markers, were non-contributory.

Radiological investigations performed previously did not reveal any definitive malignant or infective pathology. In view of persistent unexplained anemia and constitutional symptoms, bone marrow examination was planned.

Bone Marrow Examination

Bone marrow aspiration yielded inadequate material due to marrow fibrosis, and subsequently a trephine biopsy was performed. Histopathological examination of the biopsy revealed normocellular marrow spaces with diffuse infiltration by sheets and clusters of histiocytes associated with stromal fibrosis. The infiltrating cells exhibited abundant pale eosinophilic to foamy cytoplasm and small round to oval nuclei without significant atypia. No granulomas, metastatic deposits, or features suggestive of hematological malignancy were identified. Bone marrow biopsy showing intertrabecular histiocytic infiltration with associated fibrosis **(Figure 2)**.

Bone marrow biopsy showing spindle-shaped histiocytes with stromal fibrosis **(Figure 3)**.

Immunohistochemistry

Immunohistochemical (IHC) studies were carried out to characterize the histiocytic infiltrate and exclude other histiocytic disorders. The infiltrating histiocytes showed strong cytoplasmic positivity for CD68 and CD163, confirming their macrophage/histiocytic origin. CD3 positivity highlighted admixed reactive T-lymphocytes within the lesion. The lesional cells were negative for CD1a and Langerin (CD207), thereby excluding Langerhans cell histiocytosis. Based on the characteristic histomorphological and immunophenotypic profile, a diagnosis of Erdheim–Chester disease was established.

Further clinicoradiological correlation suggested multisystem involvement, consistent with the known heterogeneous presentation of ECD. The patient was subsequently advised molecular testing for MAPK pathway mutations, including BRAF V600E mutation analysis, along with multidisciplinary evaluation for systemic disease assessment and therapeutic planning.

Histopathological and immunohistochemical features of Erdheim–Chester disease **(Figure 4).** Negative immunohistochemical markers in Erdheim-Chester disease **(Figure 5).** Summary of Investigations in the Present Case **(Table 1).**

Treatment, Management, and Outcome

Following diagnosis, the patient received supportive treatment including correction of anemia and symptomatic management. She was advised comprehensive systemic evaluation and molecular testing for BRAF V600E mutation to assess eligibility for targeted therapy. In view of suspected multisystem involvement, the patient was referred to a tertiary care center for further management with consideration of interferon therapy and targeted agents such as BRAF/MEK inhibitors. On follow-up, the patient showed symptomatic improvement with stable clinical status.

RESULTS

A 47-year-old female presented with generalized weakness, refractory anemia, diarrhea, insomnia, and constitutional symptoms. Routine laboratory and radiological investigations were inconclusive, while peripheral smear showed normocytic normochromic anemia. Bone marrow aspiration was unsuccessful due to fibrosis, necessitating a trephine biopsy. Histopathological examination demonstrated diffuse infiltration by foamy and spindle-shaped histiocytes with associated stromal fibrosis. Immunohistochemistry revealed strong positivity for CD68 and CD163, with negative staining for CD1a and Langerin (CD207), confirming the diagnosis of Erdheim–Chester disease and excluding Langerhans cell histiocytosis. The patient received supportive treatment and was advised BRAF V600E mutation analysis and further systemic evaluation to facilitate targeted therapy planning.

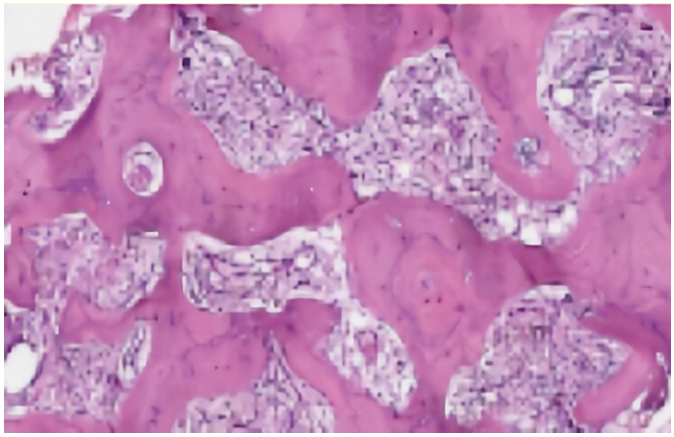

Figure 2: Bone marrow biopsy section showing trabecular bone with intertrabecular infiltration by sheets of histiocytes and associated fibrosis (H&E stain, ×100).

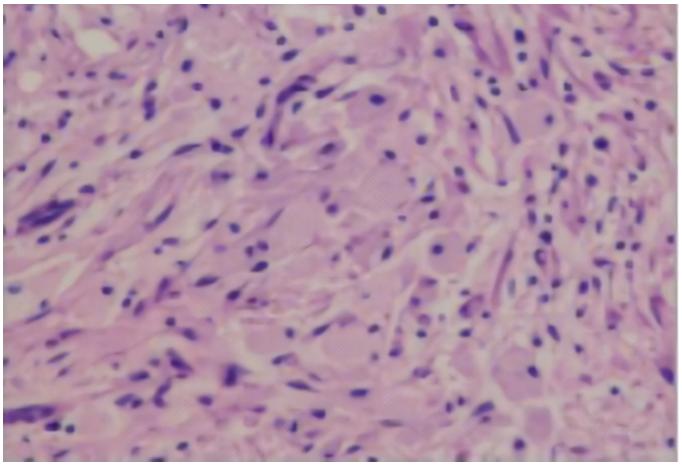

Figure 3: Photomicrograph showing spindle-shaped histiocytic cells with stromal fibrosis in the bone marrow biopsy specimen (H&E stain, ×400).

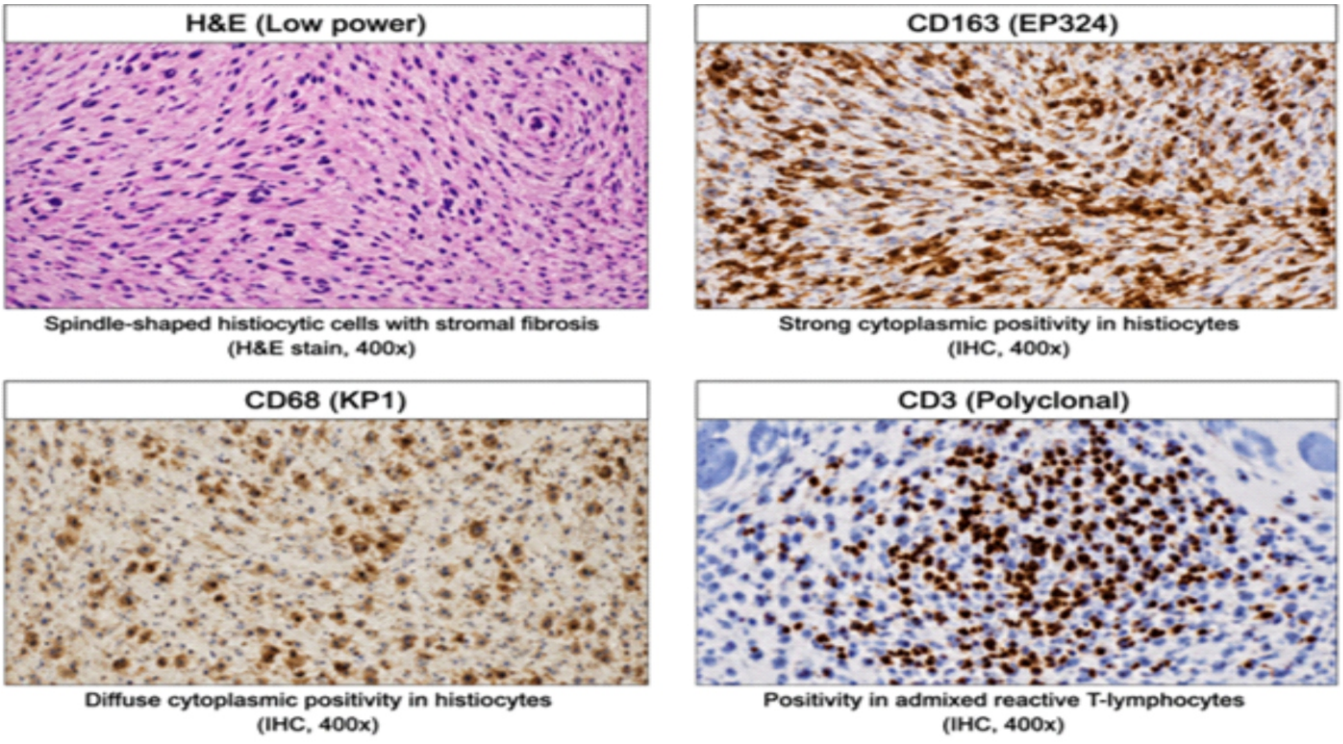

Figure 4: Histopathological and immunohistochemical findings in Erdheim–Chester disease. (A) H&E-stained section showing spindle-shaped histiocytic infiltration with stromal fibrosis. (B) CD163 immunostain demonstrating strong cytoplasmic positivity in histiocytes. (C) CD68 immunostain showing diffuse positivity in infiltrating histiocytic cells. (D) CD3 immunostain highlighting admixed reactive T-lymphocytes (IHC, ×400).

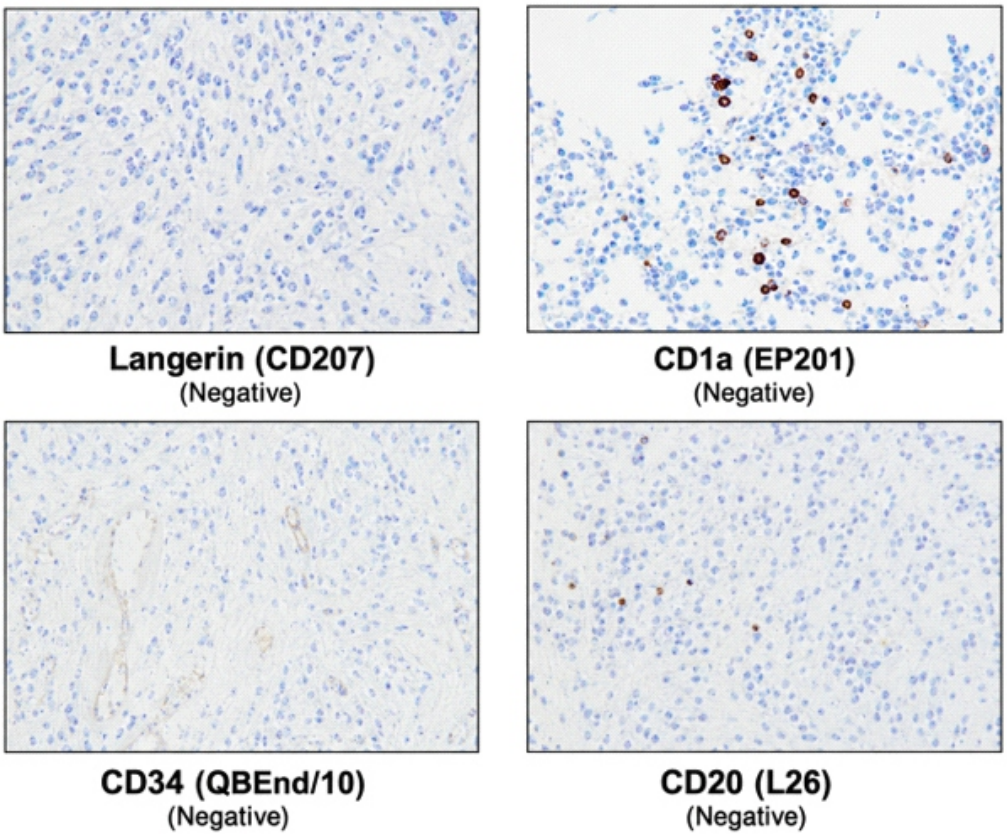

Figure 5: Immunohistochemical markers with negative findings in Erdheim–Chester disease. Langerin (CD207), CD1a, CD34, and CD20 showed negative staining in the lesional histiocytic cells, helping exclude Langerhans cell histiocytosis and lymphoproliferative disorders (IHC, ×400).

Table 1: Investigations Performed in the Present Case

Investigation	Findings	Interpretation
Complete Blood Count (CBC)	Persistent anemia	Suggestive of chronic underlying pathological process
Peripheral Blood Smear	Normocytic normochromic red blood cells	Consistent with refractory anemia without evidence of hemolysis
Total Leukocyte Count	Within normal limits	No significant leukocytic abnormality
Platelet Count	Within normal limits	No thrombocytopenia or thrombocytosis
Liver Function Tests (LFT)	Within normal limits	No hepatic dysfunction detected
Renal Function Tests (RFT)	Within normal limits	No renal impairment identified
Serum Electrolytes	Within normal limits	No significant metabolic imbalance
Inflammatory markers	Non-contributory	No definitive inflammatory or infective etiology established
Radiological Investigations	Inconclusive	No definitive malignant or infective pathology identified
Bone Marrow Aspiration	Inadequate aspirate due to fibrosis	Suggestive of marrow fibrosis/infiltrative pathology
Bone Marrow Biopsy	Normocellular marrow with diffuse histiocytic infiltration and fibrosis	Indicative of infiltrative histiocytic disorder
Histopathological Examination	Foamy histiocytes with pale eosinophilic cytoplasm	Characteristic morphology of Erdheim–Chester disease
Immunohistochemistry: CD68	Positive	Confirms histiocytic/macrophage lineage
Immunohistochemistry: CD163	Positive	Supports non -Langerhans histiocytic proliferation
Immunohistochemistry: CD3	Positive in reactive lymphocytes	Indicates admixed reactive T - lymphocytes
Immunohistochemistry: CD1a	Negative	Excludes Langerhans cell histiocytosis
Immunohistochemistry: Langerin (CD207)	Negative	Further excludes Langerhans cell histiocytosis
Final Diagnostic Impression	Erdheim–Chester disease	Diagnosis established based on histopathology and IHC profile

DISCUSSION

In the present case, a 47-year-old female presented with generalized weakness, refractory anemia, diarrhea, insomnia, and constitutional symptoms without any definitive diagnosis on prior investigations. Clinical examination revealed pallor with otherwise non-specific findings, while routine laboratory investigations were largely inconclusive except for persistent anemia. Similar demographic and clinical profiles have been described in previous studies. Cohen-Aubart et al. [5] and Estrada-Veras et al. [7] reported that Erdheim–Chester disease (ECD) commonly affects middle-aged adults and often presents with vague constitutional symptoms such as fatigue, weakness, weight loss, and multisystem complaints, resulting in delayed diagnosis. In our patient, radiological investigations failed to identify a definitive pathology, and diagnosis was ultimately established on bone marrow biopsy. Histopathological examination demonstrated diffuse infiltration by spindle-shaped and foamy histiocytes associated with stromal fibrosis. Ozkaya et al. [8] similarly described the characteristic histopathological features of ECD as xanthogranulomatous infiltration composed of foamy histiocytes with variable fibrosis and inflammatory infiltrates. Campochiaro et al. [6] further emphasized that fibrosis and histiocytic infiltration are among the most consistent pathological findings in ECD.

Immunohistochemical analysis in the present case revealed strong positivity for CD68 and CD163, while CD1a and Langerin (CD207) were negative. These findings were instrumental in differentiating ECD from Langerhans cell histiocytosis and other histiocytic disorders. Similar immunophenotypic profiles have been reported in the literature. Ozkaya et al. [8] demonstrated that ECD histiocytes consistently express macrophage markers such as CD68 and CD163 while lacking CD1a and Langerin expression. Haroche et al. [9] also highlighted the importance of negative CD1a and Langerin staining in establishing the diagnosis and excluding Langerhans cell histiocytosis.

The present patient demonstrated clinicopathological evidence suggestive of multisystem involvement, although characteristic skeletal findings were not documented. ECD is well known for its heterogeneous organ involvement affecting bones, cardiovascular system, retroperitoneum, lungs, skin, and central nervous system [6,13]. Estrada-Veras et al. [7] observed that extra-skeletal manifestations may predominate in many patients, contributing to atypical presentations and delayed recognition. Similarly, Cavalli et al. [13] described the highly variable clinical spectrum of ECD, ranging from isolated organ involvement to disseminated multisystem disease.

In the current case, molecular testing for BRAF V600E mutation was advised to guide targeted therapy. Recent studies have established ECD as a clonal disorder involving activation of the MAPK signaling pathway. Haroche et al. [3] demonstrated a high prevalence of BRAF V600E mutation in ECD patients, while Diamond et al. [4] reported significant clinical benefit with MEK inhibitor therapy in histiocytic neoplasms. These molecular discoveries have significantly transformed disease management. The patient in our study received supportive treatment and was referred to a tertiary care center for further evaluation and targeted management. Current consensus recommendations advocate molecular profiling & individualized targeted therapy for eligible patients [10,12]. Diamond et al. [11] demonstrated substantial therapeutic response with vemurafenib in BRAF-mutated ECD, whereas Goyal et al. [12] emphasized the importance of multidisciplinary evaluation and mutation-directed treatment strategies in improving outcomes.

Overall, the present case highlights the diagnostic challenges associated with Erdheim-Chester disease and underscores the importance of histopathological examination & immunohisto-chemistry in patients presenting with unexplained constitutional symptoms and infiltrative marrow pathology. Early recognition and timely molecular evaluation are essential for optimal management and improved prognosis.

CONCLUSION

Erdheim-Chester disease is a rare multisystem non-Langerhans cell histiocytic disorder with highly variable and non-specific clinical manifestations, often resulting in delayed diagnosis. The present case highlights the importance of considering ECD in patients presenting with unexplained constitutional symptoms, refractory anemia, and infiltrative bone marrow pathology. Histopathological evaluation combined with immunohisto-chemistry remains crucial for establishing the diagnosis, particularly the demonstration of CD68/CD163 positivity with absence of CD1a and Langerin expression. Early diagnosis and molecular evaluation for MAPK pathway mutations are essential for timely initiation of targeted therapy and improved patient outcomes.

Clinical Takeaway

- Erdheim–Chester disease should be considered in cases of unexplained anemia with histiocytic marrow infiltration and fibrosis.
- Bone marrow biopsy and immunohistochemistry play a pivotal role in differentiating ECD from other histiocytic disorders.
- CD68 and CD163 positivity with negative CD1a and Langerin strongly support the diagnosis of ECD.
- Early multidisciplinary evaluation and molecular testing for BRAF/MAPK pathway mutations are critical for guiding targeted therapy.
- Prompt recognition of ECD can facilitate appropriate treatment and potentially improve long-term prognosis.

LIMITATIONS & FUTURE PERSPECTIVES

The study's limitations include a single-centre setting, a relatively small sample size, and a short study duration, which may limit the broader applicability of the results. Future studies should incorporate multicentre designs with larger populations to enhance validity, assess long-term outcomes, and investigate advanced diagnostic & management approaches. Such efforts will improve overall patient care & help minimize complications.

CLINICAL SIGNIFICANCE

The clinical significance of this study lies in its potential to bridge the gap between research findings and practical healthcare applications. It emphasizes the importance of translating scientific observations into meaningful improvements in patient care, diagnosis, and treatment outcomes. By highlighting real-world relevance, the study contributes to evidence based medical practice and supports informed clinical decision making. Ultimately, the findings aim to enhance patient quality of life, optimize therapeutic strategies, and promote better disease management in clinical settings.

ABBREVIATIONS

BM: Bone Marrow
CD68: Cluster of Differentiation 68
CD163: Cluster of Differentiation 163
CD207: Cluster of Differentiation 207 (Langerin)
CT: Computed Tomography
ECD: Erdheim-Chester Disease
H&E: Hematoxylin and Eosin
IHC: Immunohistochemistry
MRI: Magnetic Resonance Imaging

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AUTHOR CONTRIBUTIONS

All authors significantly contributed to the study conception and design, data acquisition, or data analysis and interpretation. They participated in drafting the manuscript or critically revising it for important intellectual content, consented to its submission to the current journal, provided final approval for the version to be published, and accepted responsibility for all aspects of the work. Additionally, all authors meet the authorship criteria outlined by the International Committee of Medical Journal Editors (ICMJE) guidelines.

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CONFLICT OF INTEREST

Authors declared that there is no conflict of interest.

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All necessary consent & approval was obtained by authors.

CONSENT FOR PUBLICATION

All necessary consent for publication was obtained by authors.

DATA AVAILABILITY

All data generated and analyzed are included within this research article. The datasets utilized and/or analyzed in this study can be obtained from the corresponding author upon a re-asonable request.

USE OF ARTIFICIAL INTELLIGENCE (AI) & LARGE LANGUAGE MODEL (LLM)

The authors confirm that no AI & LLM tools were used in the writing or editing of the manuscript, and no images were altered or manipulated using AI & LLM.

AUTHOR'S NOTE

This article serves as an important educational tool for the scientific community, offering insights that may inspire future research diretions. However, they should not be relied upon independently when making treatment decisions or developing public health policies.

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