Case Report

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A Case of Myelofibrosis Masquerading as Pericarditis, the Role of Hematologic Evaluation in Systemic Inflammation: A Case Report

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ABSTRACT

This case report describes a middle-aged female with untreated hypertension who was released from the hospital with signs and symptoms of sharp pleuritic chest pain, with a concern for a cardiac issue. Following the concern for a cardiac diagnosis, diagnostic testing determined profound ST elevations across the ECG, severe anemia (hemoglobin 9.6 g/dL), and inflammatory activity (ESR 93 mm/hr, CRP 77.7 mg/L), with high LDH (2185 U/L). There was evidence on imaging of left atrial enlargement, hepatosplenomegaly, liver lesions, and mineralized bone changes, with myelofibrosis (MF) confirmed through marrow biopsy showing marrow fibrosis and atypical megakaryocytes. This case illustrates the diagnostic considerations of MF, which can present with various differential diagnoses. Treatment of the patient consisted of managing symptoms with aspirin; the possible options of treatment included long-term therapy with a JAK2 inhibitor or stem cell transplant. This case underscores the importance of considering MF in patients with unexplained cytopenias, splenomegaly, and systemic inflammation.

Introduction

Myelofibrosis is characterized by the abnormal proliferation of hematopoietic stem cells, resulting in bone marrow fibrosis, cytopenias, and extramedullary hematopoiesis. Patients typically experience a range of nonspecific symptoms, including profound fatigue, early satiety, and other systemic signs, alongside more distinctive findings such as splenomegaly, leukoerythroblastosis, and elevated inflammatory markers [1,2]. The diagnostic process can be particularly challenging due to the heterogeneous presentation of the disease, which can easily be confused with various solid tumors, autoimmune conditions, and infections [3]. This case report highlights a patient whose initial symptoms were primarily acute cardiac issues that mimicked pericarditis, emphasizing the necessity for thorough hematologic evaluations in the context of systemic inflammation and unexplained cytopenias. This reinforces the importance of considering a broad differential diagnosis when faced with complex clinical presentations.

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Case presentation

A middle-aged woman with a history of untreated hypertension presented to the hospital with sharp, pleuritic chest pain. She appeared tired, and as she described the pain that worsened with every deep breath, it was clear she was very uncomfortable. The initial tests were alarming: her ECG showed diffuse ST elevations (Figure 1), and we immediately planned for expedited evaluation to rule out pericarditis or damage to her heart. Lab results revealed severe anemia (Hb 9.6 g/dL). Her inflammatory markers were elevated, with an ESR of 93 mm/hr and CRP of 77.7 mg/L, and notably, her lactate dehydrogenase (LDH) level was markedly elevated at 2185 U/L, suggesting multiorgan involvement beyond cardiac pathology.

Initial lab and ECG results were significant, but further evaluation was required to clarify the etiology. Imaging modalities provided more evidence of involvement beyond just the heart. Echocardiography demonstrated a preserved ejection fraction (65%) but revealed severe left atrial enlargement and mitral valve thickening.

Given the discordance between her cardiac symptoms and systemic laboratory abnormalities, we expanded our differential to include hematologic, inflammatory, or infiltrative disorders. Abdominal CT (Figure 2) showed massive hepatosplenomegaly with perihepatic fluid and sclerotic bone lesions, which could represent anything from lymphoma to metastatic disease or osteomyelitis. CT was prioritized over ultrasonography for follow-up due to its ability to concurrently assess hepatic lesions, splenic architecture, and osseous changes—critical for evaluating EMH progression in MF. While ultrasonography is safer and more cost-effective for initial screening, CT's superior spatial resolution aligns with NCCN recommendations for monitoring myeloproliferative neoplasms with suspected complications [1]. We consulted with a hematologist, who recommended a peripheral blood smear (PBS), bone marrow aspiration, and biopsy (BMA/B). The PBS showed only teardrop-shaped red cells (Figure 3); bone marrow aspiration yielded a hemodilute specimen, but biopsy revealed a grim reality—fibrosis had taken over much of her marrow space, and atypical megakaryocytes pointed squarely toward myelofibrosis (MF), a rare and serious bone marrow disorder.

Following the diagnosis, the patient's pericarditis-like symptoms improved with aspirin therapy. She chose not to undergo further hospitalization due to personal reasons. Before being discharged, a plan was set for her to follow up urgently with a hematology-oncology specialist to evaluate treatment options with a JAK2 inhibitor, such as ruxolitinib, and to assess her risk. Repeat imaging was also scheduled in 4–6 weeks to monitor any changes in her hepatosplenomegaly, and her genetic testing results were to be reviewed to inform targeted therapy. During her follow-up visit four weeks later, she reported mild symptomatic relief attributed to aspirin but continued to experience ongoing fatigue. Testing revealed the presence of the JAK2 V617F mutation, and treatment with ruxolitinib was initiated. Three months later, a CT scan showed that her hepatosplenomegaly remained stable, and her inflammatory markers, including CRP and ESR, had significantly improved.

Discussion

This case centers on a middle-aged woman grappling with untreated high blood pressure, whose clinical signs and laboratory results ultimately led to a diagnosis of myelofibrosis (MF). The complexities of diagnosing MF are underscored in this patient's experience, particularly considering that its symptoms can closely mimic those of other conditions, such as pericarditis, liver disease, or metastatic cancer.

The patient presented with sharp, pleuritic chest pain and diffuse ST elevations on ECG, raising concern for acute pericarditis or cardiac injury. However, markedly elevated inflammatory markers and LDH suggested systemic involvement beyond cardiac pathology. The extreme LDH elevation—a nonspecific marker of cell turnover seen in hematologic malignancies, hemolysis, or tissue damage—particularly indicated potential multiorgan involvement [3]. This suspicion was further supported by imaging findings of hepatosplenomegaly with unusual hepatic lesions, which, along with the laboratory abnormalities, pointed toward a hematologic etiology. The constellation of findings was most consistent with extramedullary hematopoiesis (EMH), a hallmark feature of advanced myelofibrosis (MF) [4].

Cardiac structural changes (e.g., left atrial enlargement) were attributed to chronic hypertension, likely resulting from long-standing high blood pressure, and diffuse ST elevations reflected pericardial inflammation. Hepatosplenomegaly and bone abnormalities (Figure 2) were consistent with EMH, a hallmark of advanced disease that pointed toward a myeloproliferative disorder like MF. The PBS and bone marrow biopsy findings confirmed the diagnosis of MF, showing features such as marrow fibrosis and atypical megakaryocytes that are characteristic of the disease [2]. MF arises from the uncontrolled growth of blood stem cells, leading to fibrosis in

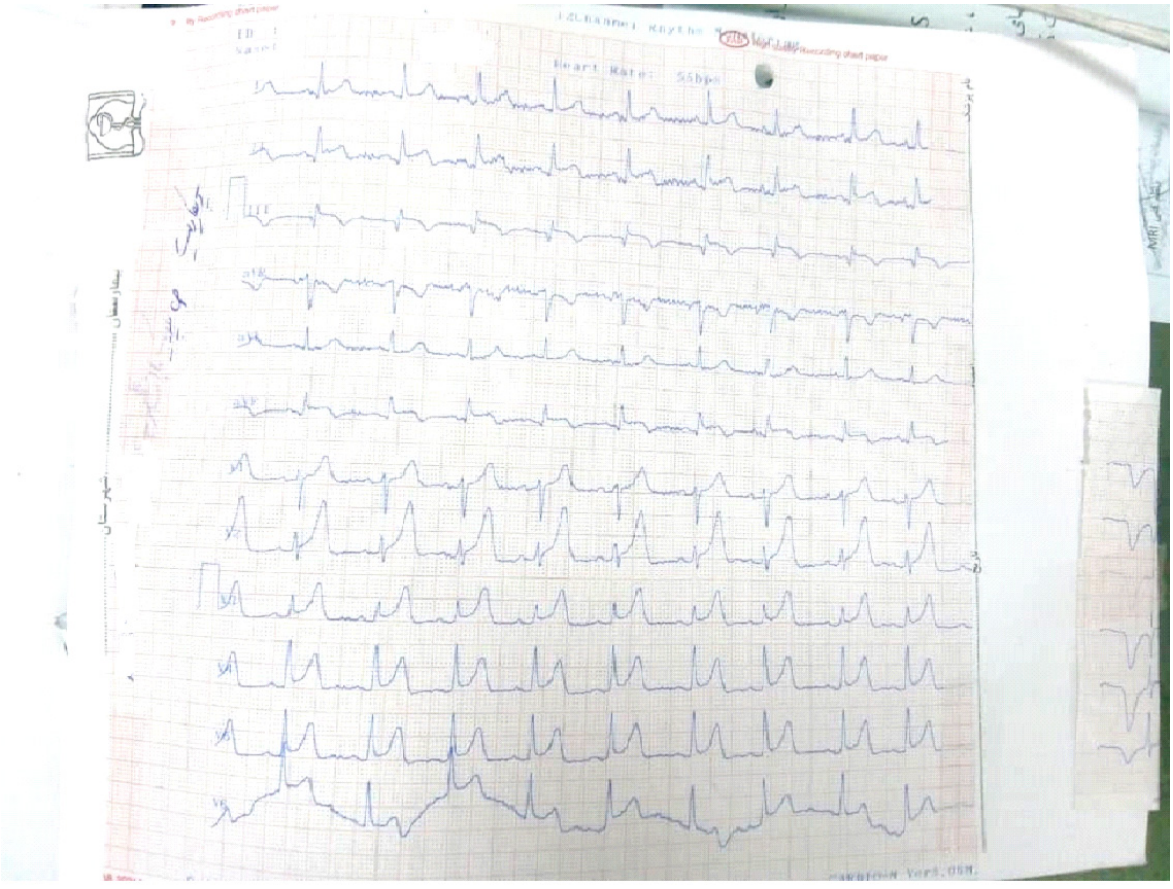

Fig. 1. ECG showing diffuse ST elevations

the bone marrow, reduced blood cell counts, and extramedullary hematopoiesis, which often results in splenomegaly and various systemic symptoms [5]. The high levels of LDH and inflammatory markers in the patient likely indicated a high disease burden and inflammation driven by the disease, which is common in myeloproliferative neoplasms [6].

MF is primarily caused by the clonal proliferation of hematopoietic stem cells, often linked to mutations in the JAK2, CALR, or MPL genes. In this case, the presence of the JAK2 V617F mutation led to the persistent activation of the JAK-STAT signaling pathway, which in turn resulted in dysregulated cytokine signaling, as evidenced by elevated levels of IL-6 and TNF-α [6]. This dysregulation helps explain the patient's pronounced systemic inflammation. These findings align with her constitutional symptoms, including significant fatigue. A key feature of MF is the progressive bone marrow fibrosis that occurs due to the increased deposition of collagen by stromal cells. This response is a result of abnormal megakaryocyte proliferation, which is supported by the biopsy findings in this patient. As the fibrosis replaces functional marrow, extramedullary hematopoiesis (EMH) begins to take place in the spleen and liver. This process accounts for her hepatosplenomegaly and the presence of perihepatic fluid (Figure 2) [4]. Additionally, EMH not only leads to anemia through the sequestration and destruction of red blood cells but also results in the formation of teardrop cells due to ineffective erythropoiesis (Figure 3).

The patient's remarkably high LDH indicates increased cell turnover associated with EMH and tissue ischemia, likely arising from splenic congestion. Furthermore, her symptoms resembling pericarditis may be attributed to cytokine-mediated serositis or the rare but documented possibility of direct EMH involvement in the pericardial tissue [5].

This case highlights the diverse ways MF can present, often leading to confusion with infections, cancers, or autoimmune diseases. The differences between the patient's cardiac symptoms—such as ST elevations and pericarditis-like chest pain—and systemic signs, including an enlarged liver and spleen, pointed to the need for a wide-ranging diagnostic approach. Ultimately, a bone marrow biopsy was crucial in confirming MF, emphasizing the importance of

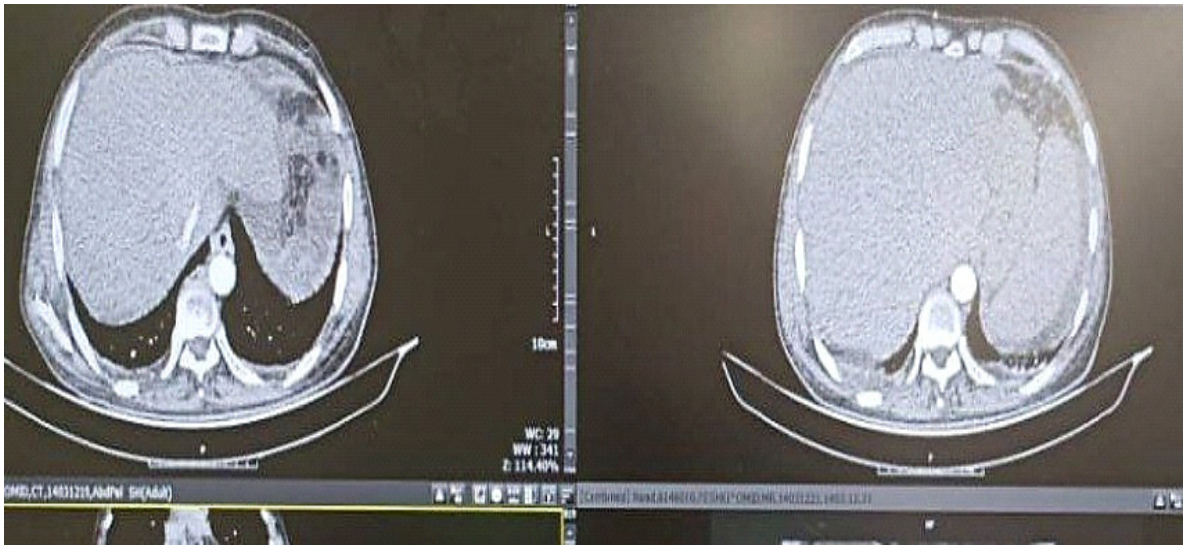

Fig. 2. Abdominal CT scan shows hepatosplenomegaly with perihepatic fluid

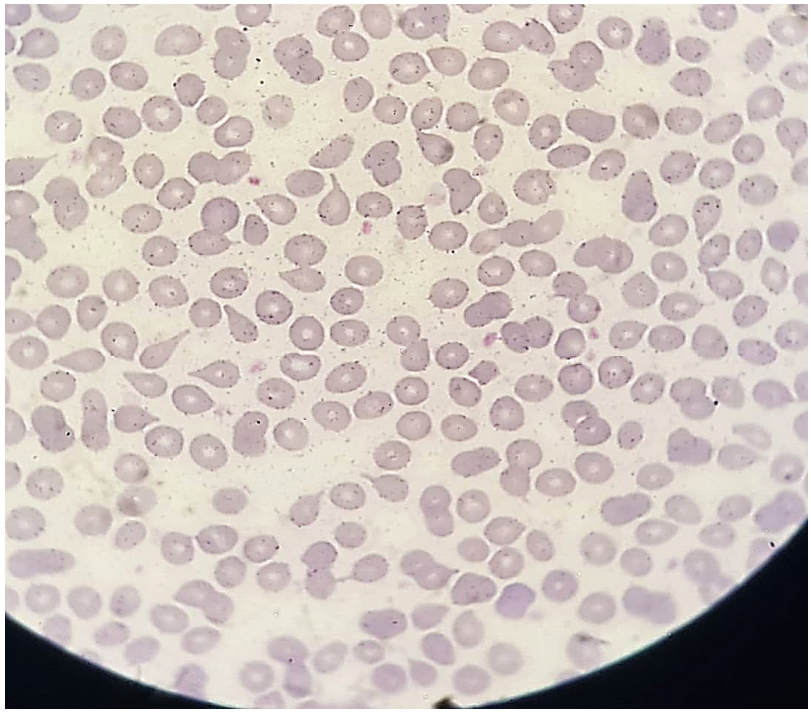

Fig. 3. PBS with teardrop-shaped red cells

conducting thorough hematologic tests when facing unexplained blood count abnormalities alongside systemic inflammation.

When specifically examining the heart-related symptoms in MF, there's an interesting pathophysiological angle to consider. Initially, the pericarditis-like symptoms might suggest a direct cardiac issue, but they likely stemmed from broader systemic inflammation brought on by MF. In patients with the JAK2 mutation, elevated IL-6 levels can trigger inflammation that mimics pericarditis [6]. It's also worth noting that while rare cases of pericardial extramedullary hematopoiesis have been reported, imaging in this case did not support that diagnosis. The fact that symptoms improved with aspirin points toward an inflammatory cause rather than an infiltrative one.

This is consistent with previous studies showing that MF can often masquerade as autoimmune or

infectious conditions. For instance, researchers like Barbui et al. have noted that inflammation related to the JAK2 mutation can irritate serosal membranes, leading to symptoms that can confuse both patients and doctors [3]. Mesa et al. have described cases where MF presented with pleural and pericardial effusions, further reinforcing this point [4]. In cases where someone has persistent "pericarditis" without a clear cause, it's essential to consider investigating myeloproliferative neoplasms, especially if accompanied by:

- Unexplained low blood cell counts
- Enlarged spleen
- Significantly high lactate dehydrogenase (LDH) and inflammation markers

Recognizing these connections can help in making a timely and accurate diagnosis for conditions like MF. There are several long-term treatment options, such as JAK2 inhibitors (e.g., ruxolitinib), and for a minority of patients, allogeneic stem cell transplantation may be considered to treat MF [3].

Conclusion

This case illustrates the complex diagnostic challenges associated with myelofibrosis (MF), particularly when accompanied by symptoms reminiscent of pericarditis. It serves as a reminder of the importance of considering hematologic malignancies in patients who present with unexplained cytopenias, splenomegaly, and signs of systemic inflammation. The observed improvement in symptoms with aspirin suggests a likely inflammatory etiology linked to cytokine activity, potentially tied to JAK2 dysregulation, rather than indicating a primary cardiac issue.

This report highlights two key takeaways: first, myelofibrosis should be included in the differential diagnosis when evaluating cases of "idiopathic" pericarditis that also exhibit hematologic abnormalities. Second, a multidisciplinary approach is crucial for achieving an accurate diagnosis. Early evaluation of the bone marrow is essential in these situations, as it can lead to timely diagnosis and the initiation of targeted therapies, such as JAK inhibitors. These treatments can address both the underlying hematologic disorder and its related systemic complications.

Ethical Considerations

Compliance with ethical guidelines

There were no ethical considerations to be considered in this article.

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Conflict of Interests

The authors have no conflict of interest to declare.

References

[1] National Comprehensive Cancer Network. Myeloproliferative neoplasms (Version 2.2024) [Internet]. Plymouth Meeting (PA): National Comprehensive Cancer Network; 2024 [cited 2024 Jul 10]. Available from: https://www.nccn.org/professionals/physician_gls/pdf/mpn.pdf

[2] Arber DA, Orazi A, Hasserjian R, Thiele J, Borowitz MJ, Le Beau MM, et al. The 2016 revision to the World Health Organization classification of myeloid neoplasms and acute leukemia. Blood. 2016 May 19;127(20):2391-405. https://doi.org/10.1182/blood-2016-03-643544

[3] Barbui T, Tefferi A, Vannucchi AM, Passamonti F, Silver RT, Hoffman R, et al. Philadelphia chromosome-negative classical myeloproliferative neoplasms: revised management recommendations from European LeukemiaNet. Leukemia. 2018 May;32(5):1057-69. https://doi.org/10.1038/s41375-018-0077-1

[4] Mughal TI, Vaddi K, Sarlis NJ, Verstovsek S. Myelofibrosis-associated complications: pathogenesis, clinical manifestations, and effects on outcomes. Int J Gen Med. 2014 Jan 29;7:89-101. https://doi.org/10.2147/ijgm.s51800

[5] Tefferi A. Primary myelofibrosis: 2017 update on diagnosis, risk-stratification, and management. Am J Hematol. 2016 Dec;91(12):1262-71. https://doi.org/10.1002/ajh.24592

[6] Vainchenker W, Kralovics R. Genetic basis and molecular pathophysiology of classical myeloproliferative neoplasms. Blood. 2017 Feb 9;129(6):667-79. https://doi.org/10.1182/blood-2016-10-695940