From Suspected Primary Myelofibrosis to T-Cell Lymphoma–Associated Hemophagocytic Lymphohistiocytosis: A Diagnostic Challenge

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Introduction

Hemophagocytic lymphohistiocytosis (HLH) is a life-threatening hyperinflammatory syndrome frequently associated with hematologic malignancies, particularly T-cell lymphomas. Diagnosis may be challenging when an occult malignancy presents with misleading initial findings.

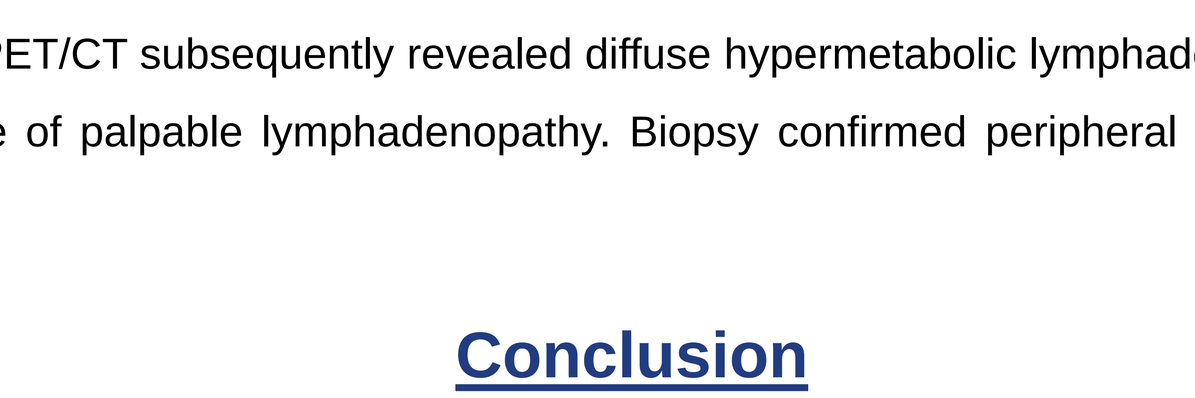
Results

A 56-year-old woman with chronic hepatitis C infection, without cirrhosis or portal hypertension, presented with a lumbar vertebral fracture. Preoperative evaluation revealed normocytic normochromic anemia. Laboratory investigations showed elevated ferritin (800 ng/mL), lactate dehydrogenase (1250 U/L), and ESR (>100 mm/h). Bone marrow examination was initially interpreted as suggestive of primary myelofibrosis (PMF). However, the diagnosis was questioned because of absent splenomegaly and negative JAK2 and other PMF-associated molecular testing.Over the following three weeks, she developed rapidly progressive pancytopenia and persistent high-grade fever reaching 41°C. Extensive infectious investigations were negative. Ferritin increased to >10,000 ng/mL, with hypertriglyceridemia (900 mg/dL) and hypofibrinogenemia (80 mg/dL). New massive hepatosplenomegaly developed, raising strong suspicion of HLH. Repeat bone marrow examination demonstrated hemophagocytosis. Dexamethasone and etoposide resulted in rapid clinical and hematologic improvement. PET/CT subsequently revealed diffuse hypermetabolic lymphadenopathy and multiple osteolytic lesions despite the absence of palpable lymphadenopathy. Biopsy confirmed peripheral T-cell lymphoma, not otherwise specified (PTCL-NOS).

Conclusion

This case highlights the diagnostic challenge of lymphoma-associated HLH and the potential for occult T-cell lymphoma to mimic PMF. Persistent clinicopathologic discordance, rapid clinical deterioration, and evolving HLH features should prompt timely reassessment and repeat diagnostic evaluation. Early recognition of the underlying malignancy is essential to avoid diagnostic delay and potentially improve outcomes.

Figure 1



PET/CT scan demonstrating diffuse metabolically active lymphadenopathy and multiple osteolytic lesions, revealing underlying peripheral T-cell lymphoma in a patient initially suspected to have primary myelofibrosis.

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