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MANAGEMENT OF RAPIDLY PROGRESSIVE SYMPTOMATIC SPLENOMEGALY WITH ASSOCIATED PANCYTOPENIA: A RARE CASE OF SPLENIC HAMARTOMA

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ABSTRACT

Introduction: Splenomegaly with focal splenic lesions represents a complex diagnostic entity, encompassing a broad spectrum of haematological, infectious, autoimmune, infiltrative and malignant disorders. Distinguishing benign from malignant pathology is particularly challenging when splenomegaly is accompanied by cytopenias and a rapidly progressive clinical course.

Case description: A 43-year-old woman presented with severe left upper quadrant abdominal pain and persistent vomiting. Physical examination revealed splenomegaly, and laboratory investigations showed pancytopenia. Abdominal ultrasonography, computed tomography scan and magnetic resonance imaging demonstrated a large heterogeneous splenic mass with progressive enlargement and mass effect on adjacent organs. ^{18}F-fluorodeoxyglucose positron emission tomography scan showed no abnormal metabolic activity. Extensive laboratory evaluation and image-guided biopsy failed to establish a definitive diagnosis. Despite conservative management, the patient's clinical condition progressively deteriorated, with persistent symptoms and worsening hypersplenism. Owing to the ongoing diagnostic uncertainty and clinical deterioration, splenectomy was performed for both diagnostic and therapeutic purposes. Histopathological examination confirmed the diagnosis of splenic hamartoma.

Conclusion: This case highlights the limitations of non-invasive diagnostic modalities in the evaluation of large splenic masses and underscores the importance of considering rare benign entities in the differential diagnosis of clinically aggressive splenic pathology. In carefully selected patients with progressive symptoms and inconclusive investigations, splenectomy remains a valuable strategy to achieve diagnostic clarity and symptom control.

KEYWORDS

Splenic hamartoma, splenomegaly, diagnostic dilemma, abdominal pain, hypersplenism

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LEARNING POINTS

- Splenomegaly associated with focal splenic lesions and pancytopenia poses a major diagnostic challenge due to substantial overlap between benign and malignant conditions.
- In symptomatic patients with progressive clinical deterioration and persistent diagnostic uncertainty, splenectomy can be both diagnostic and therapeutic.
- Splenic hamartomas are rare benign lesions that may present with non-specific symptoms and features of hypersplenism.

INTRODUCTION

Splenomegaly accompanied by focal splenic lesions represents a significant diagnostic challenge, as it may reflect a wide spectrum of haematological, infectious, autoimmune, infiltrative and malignant conditions with substantial clinical and radiological overlap[1,2]. In this context, distinguishing between benign and malignant aetiologies is often difficult, particularly when splenomegaly is associated with cytopenias, systemic manifestations or a rapid progressive clinical course[3]. Consequently, in selected patients with progressive symptoms, hypersplenism or persistent diagnostic uncertainty despite extensive investigation, splenectomy may be required to achieve both diagnostic clarification and effective symptom control[2].

We report a case of a rapidly progressive splenic mass associated with marked splenomegaly and pancytopenia, in which extensive non-invasive diagnostic evaluation failed to establish a definitive diagnosis. Splenectomy ultimately revealed a splenic hamartoma. This case illustrates the diagnostic complexity of splenomegaly with focal splenic lesions and underscores the importance of considering rare benign entities in the differential diagnosis of clinically aggressive-appearing splenic pathology.

CASE DESCRIPTION

A 43-year-old woman presented to the emergency department with a 2-day history of persistent vomiting and severe abdominal pain. The pain was predominantly located in the left upper quadrant, radiating to the left lower abdomen, and was rated as 8/10 in intensity. She denied fever, diarrhoea, changes in bowel habits, weight loss or night sweats.

Her medical history was unremarkable, with no known chronic illnesses, previous surgeries or relevant family history. Due to the absence of prior symptoms, she had not previously undergone haematological investigations or abdominal imaging. She was an active smoker with a 20 pack-year smoking history.

On physical examination, the patient appeared uncomfortable due to pain. Abdominal examination revealed tenderness over the left upper quadrant and palpable splenomegaly. Bowel sounds were normal, and there were no signs of peritoneal irritation or peripheral lymphadenopathy. During routine breast examination, a painless palpable mass approximately 1 cm in diameter with associated abnormal nipple discharge was noted in the right breast. The remainder of the clinical examination was unremarkable.

Initial laboratory investigations demonstrated pancytopenia and elevated lactate dehydrogenase levels, raising concern for an underlying haematological or infiltrative disorder. Liver and renal function tests were within normal limits, and tumour markers were negative. Breast imaging and diagnostic fine needle aspiration of the palpable breast mass revealed no evidence of malignancy.

Abdominal ultrasonography revealed moderate-to-severe splenomegaly, according to established radiological criteria[2], with the spleen measuring up to 18 cm in length, and a large heterogeneous lesion occupying the upper pole. A contrast-enhanced computed tomography (CT) scan confirmed splenomegaly (approximately 17.3 cm) and demonstrated a large heterogeneously enhancing nodular mass measuring 14.6 cm, with areas suggestive of splenic infarction or thrombosis *(Fig. 1)*. No enlarged abdominal or thoracic lymph nodes were identified. At this stage, both malignant causes, including primary splenic lymphoma or metastatic disease, and benign splenic tumours, were considered.

Given the combination of splenomegaly, pancytopenia and a large splenic mass, an extensive diagnostic work-up was initiated to exclude haematological malignancies, chronic infections, storage diseases, autoimmune and infiltrative disorders *(Table 1)*. Serological testing for viral hepatitis and



Figure 1. *Contrast-enhanced computed tomography scan demonstrating splenomegaly and a large heterogeneous enhancing nodular lesion involving the upper pole of the spleen.*

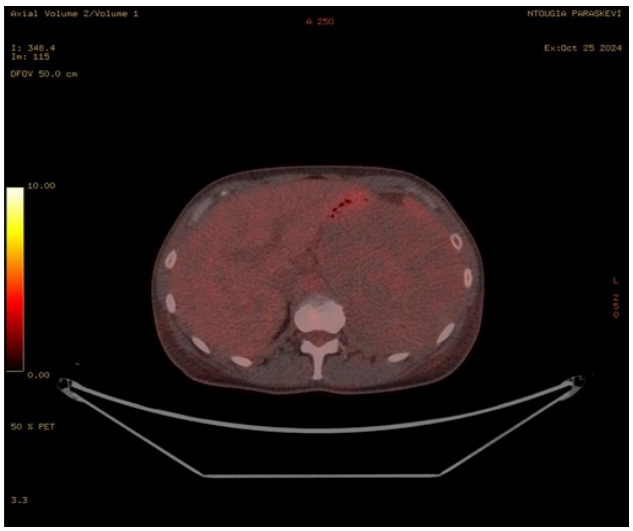


Figure 2. ^{18}*F-fluorodeoxyglucose positron emission tomography scan showing splenomegaly without pathological metabolic activity.*

bacterial infections, including *Leptospira spp.*, *Leishmania spp.* and *Borrelia spp.*, was negative. Autoimmune screening revealed positivity for anti-β2-glycoprotein antibodies; however, the diagnostic criteria for antiphospholipid syndrome were not met. An abdominal fat pad biopsy excluded amyloidosis, flow cytometry showed no evidence of lymphoproliferative disease, and enzymatic testing ruled out Gaucher disease.

Further evaluation with ^{18}F-fluorodeoxyglucose positron emission tomography (^{18}F-FDG PET) scan demonstrated splenomegaly without abnormal metabolic activity, including within the splenic lesion *(Fig. 2)*, reducing the likelihood of malignancy but not allowing definitive exclusion. Follow-up magnetic resonance imaging performed 2 weeks later showed further enlargement of the spleen (18 cm) and the nodular lesion (15 × 11.6 cm), with mass effect on the stomach and left kidney, without imaging features diagnostic of a specific benign or malignant entity. Due to persistent diagnostic uncertainty, an ultrasound-guided fine-needle aspiration of the splenic lesion was performed. Histopathological analysis demonstrated extramedullary haematopoiesis without evidence of malignancy. Despite this, the patient's clinical condition progressively deteriorated during hospitalization, with ongoing vomiting, inability to maintain adequate oral intake and persistent abdominal pain.

Given the progressive symptom burden, increasing splenic size with pressure effects on adjacent organs, and failure to establish a definitive diagnosis despite extensive investigations over 2 months of hospitalization, a multidisciplinary decision was made to proceed with splenectomy for both diagnostic and therapeutic purposes.

Histopathological examination of the resected spleen revealed a splenic hamartoma, characterized by disorganized proliferation of splenic red pulp elements, confirming a benign diagnosis. No evidence of malignancy was identified. The postoperative course was uneventful, with rapid resolution of abdominal pain and vomiting and gradual improvement in oral intake. Laboratory abnormalities related to hypersplenism resolved during follow-up, and the patient remained asymptomatic at subsequent outpatient review. Appropriate perioperative vaccinations and prophylactic measures were administered according to standard recommendations.

DISCUSSION

Splenomegaly is a diagnostically challenging clinical finding due to its broad differential diagnosis, overlapping clinical manifestations and frequently non-specific laboratory abnormalities[1]. It may reflect pathology involving multiple organ systems, including haematological, hepatic, infectious, autoimmune, infiltrative, metabolic and malignant disorders. In many cases, the spleen functions as a reactive organ rather than the primary site of disease,

Category	Potential diagnoses	Diagnostic test performed	Results
Liver disease/ congestion	Cirrhosis, portal hypertension, splenic or portal vein thrombosis	Liver function tests, Doppler ultrasonography, abdominal CT	No evidence of liver disease or vascular thrombosis
Haematological malignancy	Lymphoma, leukaemia, myeloproliferative disorders	Peripheral blood flow cytometry, ^{18}F-FDG PET, splenic biopsy	No evidence of malignancy
Infections	Viral (EBV, CMV, HIV, hepatitis), parasitic (*Leishmania spp.*), bacterial (*Leptospira spp.*)	Serology and antigen-based assays	Negative
Storage diseases	Gaucher disease, Niemann-Pick	β-glucosidase enzyme activity	Within normal range
Autoimmune	Antiphospholipid syndrome, systemic lupus erythematosus, Felty syndrome	Autoantibody testing	Diagnostic criteria not fulfilled
Infiltrative disorders	Amyloidosis, sarcoidosis	Chest CT, abdominal fat pad biopsy	Negative
Solid malignancy	Metastatic disease	Tumor markers, CT, ^{18}F-FDG PET, breast biopsy	No evidence of malignancy

Table 1. Differential diagnosis of acute or rapidly progressive splenomegaly with pancytopenia and diagnostic work-up.

Abbreviations: CT, computed tomography; EBV, Ebstein-Barr virus; CMV, cytomegalovirus; HIV, human immunodeficiency virus; ^{18}*F-FDG PET,* ^{18}*F-fluorodeoxyglucose positron emission tomography.*

Category	Representative entities	Typical clinical / imaging clues	Diagnostic limitations
Benign vascular lesions	Haemangioma, hamartoma, lymphangioma	Often incidental; may be associated with splenomegaly or hypersplenism	Imaging findings are often non-specific
Inflammatory/ reactive lesions	Sclerosing angiomatoid nodular transformation, extramedullary haematopoiesis	Variable enhancement patterns; may mimic neoplastic lesions	Considerable overlap with malignant entities
Haematologic malignancies	Lymphoma, multiple myeloma	Splenomegaly with or without lymphadenopathy; FDG-avid lesions	Focal lesions may lack diagnostic specificity
Primary splenic malignancies	Angiosarcoma	Large heterogeneous mass; areas of haemorrhage or necrosis	Rare but highly aggressive; imaging alone insufficient
Metastatic disease	Breast, lung, melanoma	Usually occurs in the setting of disseminated malignancy	Often indistinguishable from primary splenic tumours

Table 2. *Differential diagnosis of solid splenic masses and limitations of imaging in establishing a definitive diagnosis. Abbreviations: FDG, fluorodeoxyglucose.*

further complicating identification of the underlying aetiology.

The temporal pattern of disease evolution may assist in narrowing the differential diagnosis: acute presentations often suggest infectious or hypoxic causes, whereas chronic or progressively enlarging splenomegaly is more characteristic of storage disorders or myeloproliferative diseases[2]. In the present case, the absence of prior symptoms and the lack of previous haematological or imaging investigations, combined with a rapidly progressive clinical course and the coexistence of splenomegaly with pancytopenia, rendered inherited haematological conditions—such as sickle cell disease and congenital haemolytic anaemias—unlikely. These features prompted an extensive and systematic diagnostic work-up to exclude alternative causes of splenomegaly and hypersplenism *(Table 1)*.

The diagnostic evaluation was further complicated by the presence of a large splenic mass. Assessment of solid splenic lesions is inherently complex, as the spleen may harbour a wide spectrum of neoplastic entities – including haematological malignancies, primary splenic malignancies and metastatic disease – as well as pseudo-neoplastic processes such as benign vascular lesions and inflammatory or reactive conditions, all of which may exhibit overlapping clinical and imaging characteristics[3,4]. As summarized in *Table 2*, this broad differential diagnosis highlights the limitations of conventional imaging in establishing a definitive preoperative diagnosis[5].

In recent years, ^{18}F-FDG PET and image-guided splenic biopsy have been incorporated into the diagnostic evaluation of solid splenic lesions. ^{18}F-FDG PET is a valuable non-invasive tool owing to its high negative predictive value for malignancy[6]; however, FDG-negative findings do not fully exclude clinically relevant splenic pathology. Similarly, image-guided splenic biopsy may provide histological information, but its interpretation is limited by sampling error and the risk of false-negative results, particularly in rare primary splenic neoplasms[7,8]. In the present case, neither modality resolved diagnostic uncertainty: fine-needle aspiration demonstrated only extramedullary haematopoiesis, while the absence of abnormal FDG uptake lowered the suspicion of malignancy but failed to provide diagnostic certainty in the context of progressive splenomegaly, worsening symptoms and increasing mass effect.

Hypersplenism is generally considered a benign, secondary condition and is often managed conservatively. Splenectomy is therefore reserved for selected patients with severe, symptomatic cytopenias, splenic complications, or when hypersplenism precludes essential diagnostic or therapeutic decisions[2,9]. Contemporary series have demonstrated that diagnostic splenectomy is associated with acceptable morbidity and a high diagnostic yield, most commonly revealing lymphoid malignancies but also benign entities[9]. Under such circumstances, splenectomy is a valuable last-resort and evidence-supported strategy, providing both definitive diagnosis and therapeutic benefit. Nevertheless, splenectomy is not without risk. In addition to perioperative complications such as bleeding, infection or pancreatic injury, patients face lifelong risks related to asplenia, including thromboembolic events and overwhelming post-splenectomy infection[2]. These risks necessitate strict adherence to vaccination strategies, consideration of antibiotic prophylaxis and thorough patient education regarding the need for prompt medical evaluation in the event of febrile illness.

In our patient, histopathological examination following splenectomy ultimately revealed a splenic hamartoma. Splenic hamartomas are rare benign tumours, with an estimated incidence of approximately 0.001%, and are most commonly detected incidentally during imaging performed for unrelated indications[10]. Nevertheless, they may become symptomatic and typically present with non-specific manifestations such as abdominal pain, pressure-related symptoms or features of hypersplenism, rendering

pre-operative diagnosis particularly challenging[10]. In the present case, the clinically aggressive behaviour of the lesion – manifested by significant pressure effects on adjacent organs and severe pancytopenia due to hypersplenism – combined with the broad and heterogeneous differential diagnosis of splenomegaly, precluded a definitive diagnosis despite 2 months of extensive investigation. Under these circumstances, splenectomy became a necessary diagnostic and therapeutic intervention. Overall, long-term prognosis is excellent with respect to the underlying lesion, as splenectomy resulted in complete symptom resolution and normalization of haematological abnormalities, with no evidence of recurrence during follow-up. However, this therapeutic success was achieved at the cost of permanent loss of splenic function, inevitably exposing the patient to the lifelong risks associated with asplenia. This highlights the inherent clinical dilemma in such cases, underscoring the importance of careful patient selection, appropriate preventive strategies and long-term follow-up.

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