

Case Report

Aggressive systemic mastocytosis presenting as cryptogenic cirrhosis, refractory ascites, and cytopenias

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Abstract:

Aggressive systemic mastocytosis (ASM) is a rare clonal mast cell neoplasm characterized by extracutaneous mast cell infiltration leading to progressive organ dysfunction. Hepatic involvement is uncommon but clinically significant, as it may manifest with portal hypertension and refractory ascites, closely mimicking cryptogenic cirrhosis; thereby, contributing to diagnostic delay. In this report, we present a case of ASM with predominant hepatic involvement and associated cytopenias, underscoring the clinical, laboratory, and histopathologic features critical to establishing the diagnosis.

A 77-year-old Thai man presented with a 6-month history of progressive abdominal distension, early satiety, and significant unintentional weight loss. He initially received a diagnosis of cryptogenic cirrhosis with portal hypertension. Laboratory evaluation revealed normocytic anemia, thrombocytopenia, hyperglobulinemia, and isolated elevation of alkaline phosphatase with preserved hepatic synthetic function. Imaging studies demonstrated cirrhotic liver morphology, splenomegaly, generalized lymphadenopathy, and large-volume ascites. Bone marrow biopsy showed hypercellular marrow with spindle-shaped mast cell infiltration expressing CD117, mast cell tryptase, CD2, and CD30. Liver biopsy demonstrated sinusoidal infiltration by atypical mast cells with concordant immunophenotypic features. Serum tryptase was markedly elevated, and the KIT D816V mutation was detected, confirming the diagnosis of ASM. The patient developed refractory ascites requiring repeated therapeutic paracentesis and was subsequently treated with midostaurin, resulting in substantial clinical improvement, resolution of ascites, weight gain, and partial hematologic recovery.

Keywords : ● Systemic mastocytosis ● Aggressive systemic mastocytosis ● Ascites ● Portal hypertension
● Cytopenia

J Hematol Transfus Med. 2026;36:315-23.

Received 11 March 2026 Corrected 17 March 2026 Accepted 14 September 2026

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รายงานผู้ป่วย

โรคมาสโตไซโทซิสชนิดรุนแรง มาด้วยตับแข็งไม่ทราบสาเหตุ ร่วมกับภาวะน้ำในช่องท้องที่ติดต่อการรักษาและภาวะเม็ดเลือดต่ำ

ทัศน์ัย สัตยรักษา สุวรรณภรณ์ เยาว์พฤษชัย จุฑาวุฒิ ไทพานิชย์ และ มุทิตา สุรกีจวร

สาขาวิชาโลหิตวิทยา ภาควิชาอายุรศาสตร์ คณะแพทยศาสตร์ จุฬาลงกรณ์มหาวิทยาลัย และ โรงพยาบาลจุฬาลงกรณ์ สภากาชาดไทย

บทคัดย่อ

โรคมาสโตไซโทซิสชนิดรุนแรง (Aggressive systemic mastocytosis, ASM) เป็นเนื้องอกจากมาสต์เซลล์ (Mast cell) ที่เกิดจากการเพิ่มจำนวนแบบโคลนเดียวที่พบได้ไม่บ่อย มีลักษณะเด่นคือการแทรกซึมของมาสต์เซลล์ในอวัยวะนอกผิวหนัง ส่งผลให้เกิดความเสียหายของอวัยวะ การมีพยาธิสภาพที่ตับพบได้น้อย แต่มีความสำคัญทางคลินิก เนื่องจากอาจแสดงอาการด้วยภาวะความดันโลหิตสูงในหลอดเลือดพอร์ทัลและภาวะน้ำในช่องท้องที่ติดต่อการรักษา ซึ่งมีลักษณะคล้ายโรคตับแข็งไม่ทราบสาเหตุ ส่งผลให้การวินิจฉัยล่าช้า รายงานฉบับนี้นำเสนอผู้ป่วยโรคมาสโตไซโทซิสชนิดรุนแรงซึ่งมีอาการเด่นทางตับร่วมกับภาวะเม็ดเลือดต่ำ โดยแสดงรายละเอียดของอาการแสดงทางคลินิก ผลการตรวจทางห้องปฏิบัติการ และลักษณะทางพยาธิวิทยาที่นำไปสู่การวินิจฉัย

ผู้ป่วยชายชาวไทยอายุ 77 ปี มาพบแพทย์ด้วยอาการท้องโตเพิ่มขึ้นอย่างต่อเนื่องเป็นระยะเวลา 6 เดือน ร่วมกับอาการอัมเร็วและน้ำหนักลดโดยไม่ทราบสาเหตุ ระยะแรกได้รับการวินิจฉัยเป็นโรคตับแข็งไม่ทราบสาเหตุร่วมกับภาวะความดันโลหิตสูงในหลอดเลือดพอร์ทัล การตรวจทางห้องปฏิบัติการพบ ภาวะโลหิตจางชนิดเม็ดเลือดแดงขนาดปกติ ภาวะเกล็ดเลือดต่ำ ภาวะโกลบูลินในเลือดสูง และพบค่าเอนไซม์อัลคาไลน์ฟอสฟาเตสสูงขึ้นเพียงค่าเดียว โดยการทำงานของตับยังอยู่ในเกณฑ์ปกติ การตรวจทางภาพถ่ายรังสีพบลักษณะตับแข็ง ม้ามโต ภาวะต่อมน้ำเหลืองโตทั่วร่างกาย และภาวะน้ำในช่องท้องปริมาณมาก การตรวจชิ้นเนื้อในไขกระดูกมีเซลล์มากกว่าปกติ ร่วมกับการแทรกซึมของมาสต์เซลล์ที่มีลักษณะผิดปกติ ซึ่งมีการแสดงออกของ CD117, mast cell tryptase, CD2 และ CD30 การตรวจชิ้นเนื้อตับพบว่าบริเวณหลอดเลือดไขว้ชอยด์มีการแทรกซึมของมาสต์เซลล์ที่มีลักษณะผิดปกติ โดยมีลักษณะทางอิมมูโนโฟโนไทป์สอดคล้องกับเซลล์ที่ตรวจพบในไขกระดูก ตรวจพบระดับทริปเทส (tryptase) ในเลือดสูงผิดปกติ และพบการกลายพันธุ์ของยีน KIT D816V ซึ่งยืนยันการวินิจฉัยโรคมาสโตไซโทซิสชนิดรุนแรง ผู้ป่วยมีภาวะน้ำในช่องท้องที่ติดต่อการรักษาจำเป็นต้องได้รับการเจาะน้ำในช่องท้องซ้ำหลายครั้ง ต่อมาได้รับการรักษาด้วยยา midostaurin ส่งผลให้อาการทางคลินิกดีขึ้นอย่างชัดเจน ภาวะน้ำในช่องท้องลดลง น้ำหนักตัวเพิ่มขึ้น และมีการฟื้นตัวของความผิดปกติของเม็ดเลือดบางส่วน

คำสำคัญ : ● โรคมาสโตไซโทซิส ● โรคมาสโตไซโทซิสชนิดรุนแรง ● น้ำในช่องท้อง ● ความดันหลอดเลือดพอร์ทัลสูง
● ภาวะเม็ดเลือดต่ำ

วารสารโลหิตวิทยาและเวชศาสตร์บริการโลหิต. 2569;36:315-23.

Introduction

Systemic mastocytosis (SM) is a rare clonal hematologic neoplasm characterized by the abnormal accumulation of neoplastic mast cells in the bone marrow and one or more extracutaneous organs including liver, spleen, gastrointestinal tract, and lymph nodes.^{1,2} The clinical spectrum of SM is highly heterogeneous,³ ranging from indolent forms with near-normal life expectancy to advanced variants associated with significant organ dysfunction and poor survival.¹ Owing to its rarity and diverse clinical manifestations, SM is frequently underdiagnosed or misclassified, particularly when presenting with atypical features such as unexplained cytopenias or portal hypertension.

According to the National Comprehensive Cancer Network (NCCN) Guidelines (Version 1.2025), the classification of SM follows the 2022 World Health Organization (WHO) framework and includes indolent SM, smoldering SM, aggressive SM, and SM with an associated hematologic neoplasm and mast cell leukemia.^{1,4} This classification is clinically relevant, as treatment decisions are guided primarily by the presence of mast cell-related organ damage, termed C-findings,³ rather than mast cell burden alone. C-findings include cytopenias, hepatosplenomegaly with portal hypertension or ascites, malabsorption with weight loss and skeletal destruction.^{4,5}

The molecular pathogenesis of SM is driven predominantly by activating mutations in the *KIT* proto-oncogene, most commonly the *KIT* D816V mutation,^{1,4,6} resulting in constitutive mast cell activation and survival. Advances in high-sensitivity molecular techniques have significantly improved diagnostic accuracy and prognostic stratification.⁶ Given the availability of targeted therapies and the risk of irreversible organ damage in advanced disease, early recognition and accurate classification of SM are essential.

Case report

A 77-year-old Thai man, a retired government officer, presented progressive abdominal distension for six months, accompanied by early satiety and unintentional weight loss of approximately 20 kg within one month. The weight loss was likely multifactorial, related to both reduced oral intake from early satiety and ongoing fluid removal from repeated paracentesis. He had been independent in activities of daily living. He denied fever, abdominal pain, hematemesis, melena, dyspnea, orthopnea, or foamy urine. He presented to a private hospital and received a diagnosis of cirrhosis of unknown etiology and treated with diuretics and therapeutic abdominal paracentesis.

His medical history was notable for dilated cardiomyopathy with recovered ejection fraction; a transthoracic echocardiogram (TTE) performed two months prior to presentation demonstrated a left ventricular ejection fraction (LVEF) of 56% with hypokinesia of the anteroseptal and inferior walls. Additional comorbidities included type 2 diabetes mellitus and hypertension. He denied alcohol consumption for more than four decades and had no history of viral hepatitis, blood transfusion, or herbal medication use.

On physical examination, he appeared chronically ill but alert. Vital signs were stable. Physical examination revealed moderate abdominal distension with shifting dullness, impalpable spleen and bilateral pitting leg edema. No stigmata of chronic liver disease, superficial lymphadenopathy, skin rash, flushing, or urticaria pigmentosa were observed.

Initial laboratory evaluation revealed normocytic anemia, monocytosis, and thrombocytopenia with hemoglobin (Hb) 8 g/dL, hematocrit (Hct) 26.8% and mean corpuscular volume (MCV) 90 fL. The white blood cell (WBC) count was $3.28 \times 10^3/\mu\text{L}$, with a differential count of 56.7% neutrophils, 23.2% lymphocytes, and 16.8% monocytes; the platelet count was $59 \times 10^3/\mu\text{L}$. Liver biochemistry demonstrated isolated elevation of

alkaline phosphatase (up to 340 U/L) with preserved bilirubin transaminases and renal function. Serum total protein was 8.1 g/dL with preserved albumin level of 3.1 g/dL, while hyperglobulinemia up to 5 g/dL was observed. Coagulation studies showed slight prolongation of prothrombin time (PT) and international normalized ratio (INR) of 18.9 and 1.71, respectively, with normal activated partial thromboplastin time (APTT). Viral hepatitis serologies were negative. Autoimmune profile revealed positive antinuclear antibody with homogeneous and fine speckled patterns (1:80 to 1:160), and low-titer anti-smooth muscle antibody positivity (1:80). Ceruloplasmin level was 0.430 g/L (within normal limits). His ascitic fluid analysis revealed straw colored fluid, red blood cells (RBC) 4,200 / μ L, white blood cells (WBC) 125 / μ L with differential of polymorphonuclear neutrophil (PMN) 10.4% and mononuclear cells 89.6%, protein 3.1 g/dL, albumin 1.19 g/dL, lactate dehydrogenase (LDH) 57 IU/L, and triglyceride 52 mg/dL. The profile showed high serum-ascites albumin gradient (SAAG), high protein ascites, mononuclear cell predominates, and negative bacterial, mycobacterial, and flow cytometry for clonal B cell detection. Cytology showed macrophage-rich fluid with reactive mesothelial cells. Immunohistochemical staining for mast cell markers was not performed; therefore, peritoneal involvement could not be excluded.

Contrast-enhanced computed tomography (CT) of the abdomen demonstrated cirrhotic liver morphology and splenomegaly (14.9 cm) without focal lesions, with multiple enlarged lymph nodes involving porta hepatis, portocaval, left gastric, both renal hila, paraaortic, aortocaval, anterior diaphragmatic and right external iliac regions, measuring up to 2.3 cm in the greatest dimension, along with large-volume ascites. Importantly, no evidence was found of portal vein thrombosis, Budd-Chiari syndrome or hepatocellular carcinoma (Figure 1).

- (A) Axial noncontrast-enhanced phase
- (B) Axial arterial phase
- (C) Axial portal venous phase
- (D) Coronal portal venous phase

Bone marrow biopsy was performed due to unexplained cytopenias accompanied by monocytosis raising concern for an underlying clonal hematologic or infiltrative disorder. The examination revealed a hypercellular marrow (cellularity 90%) with preserved trilineage hematopoiesis and a proliferation of atypical spindle-shaped mast cells comprising approximately 30% of nucleated cells, associated with fibrosis. Immunohistochemistry demonstrated that the mast cells were positive for CD117, mast cell tryptase, CD2, and CD30 and negative for CD20 and CD3. CD25 immunostaining was not performed, (unavailable at our institution) and this has been acknowledged as a limitation. Overall, these findings fulfil the diagnostic criteria for SM (Figure 2).

Liver biopsy also demonstrated focal involvement by SM characterized by infiltration of atypical mast cells within the hepatic sinusoids. Immunohistochemistry demonstrated positivity for CD117, mast cell tryptase, CD2 (focal), and negativity for CD20, CD3, CD5, CD7, CD4, CD8, and AE1/AE3 (Figure 3). Serum tryptase level was markedly elevated up to 189 μ g /L (normal < 13.5 μ g/L). Molecular analysis, including next-generation sequencing (NGS), detected the *KIT* D816V mutation, confirming the diagnosis of SM.

The patient required weekly large-volume paracentesis with albumin replacement for refractory ascites. Despite optimized supportive care, his clinical condition progressively deteriorated with worsening cytopenias and functional decline. Delayed recognition of an underlying infiltrative systemic disorder precluded the timely initiation of disease-modifying therapy. After the diagnosis of ASM was established, treatment with midostaurin was initiated. His performance status improved significantly, with a 2-kg weight gain within one month. Symptoms of abdominal distension and early satiety secondary to ascites were markedly ameliorated, and he has not required abdominal paracentesis since initiation of therapy, with concurrent reduction in splenic size on ultrasound from 14.9 cm to 10 cm. Laboratory evaluation demonstrated gradual improvement in thrombocytopenia, an increase in serum albumin, and a decline in alkaline

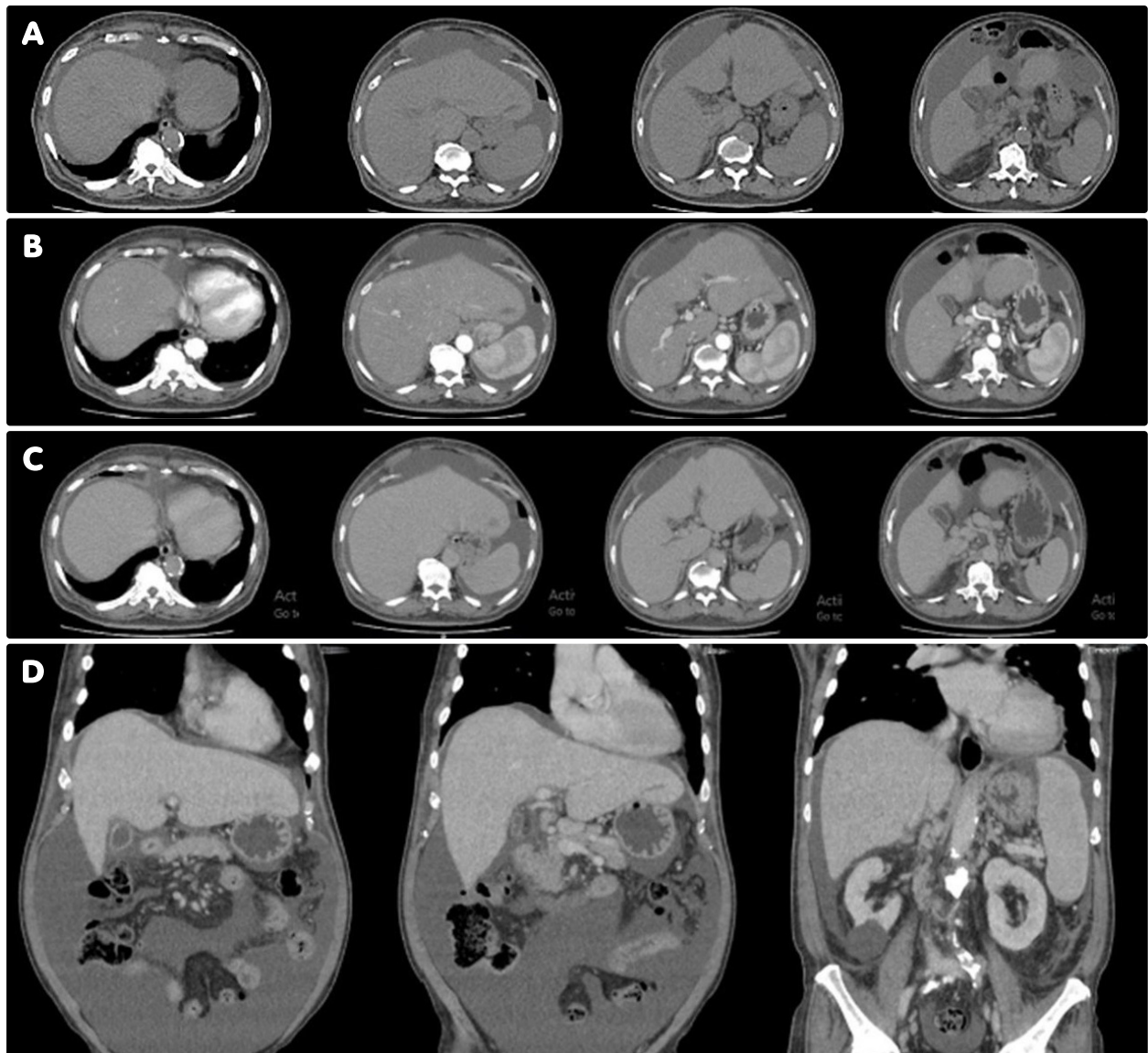


Figure 1 (A-D) Contrast-enhanced CT of the abdomen demonstrated cirrhotic liver morphology with large amount of ascites, splenomegaly without focal lesions and multiple enlarged abdominal lymph nodes measuring up to 2.3 cm in the greatest dimension.

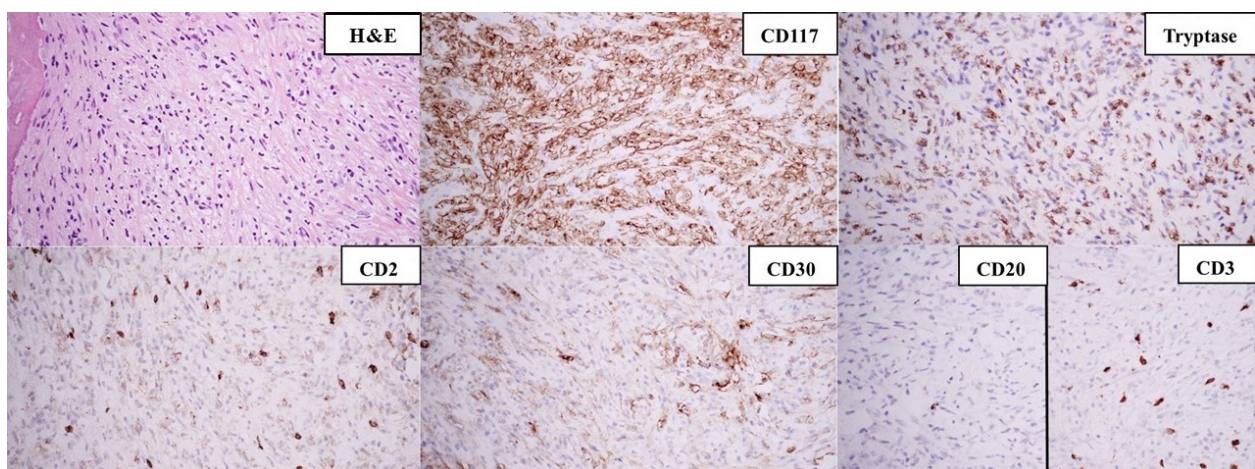


Figure 2 Histopathologic examination of the bone marrow revealed a hypercellular marrow with multifocal aggregates of atypical spindle-shaped mast cells and associated fibrosis. Immunohistochemistry demonstrated positivity for CD117, mast cell tryptase, CD2, CD30 and negativity for CD20 and CD3.

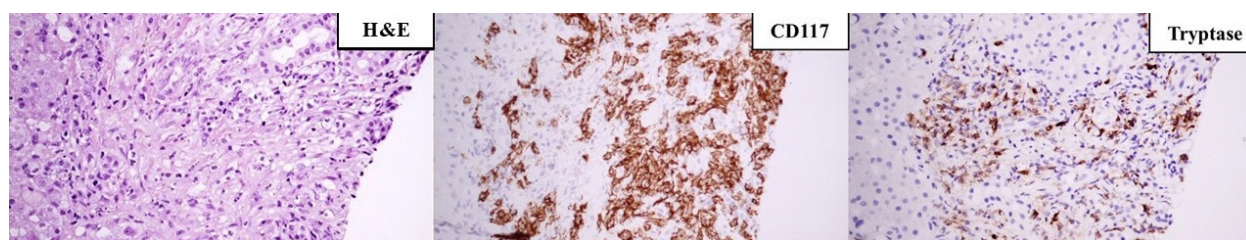


Figure 3 Histopathologic examination of liver biopsy revealed sinusoidal infiltration by atypical mast cells. Immunohistochemistry demonstrated positivity for CD117 and mast cell tryptase.

phosphatase level. However, the serum tryptase level remained elevated ($> 200 \mu\text{g/L}$; normal $< 13.5 \mu\text{g/L}$) at three to six months, and bone marrow mast cell infiltration remained 15% at approximately three months. During treatment with midostaurin, he developed hematologic adverse effects, including anemia and neutropenia, which were managed with supportive therapy consisting of weekly administration of granulocyte colony-stimulating factors (G-CSF) and erythropoietin-stimulating agent (ESA) (Figure 4, 5).

Discussion

In the present case, the patient exhibited several hallmark features of advanced SM as defined by the NCCN Guidelines including cytopenias, hepatosplenomegaly, refractory ascites, and lymphadenopathy; thereby, fulfilling multiple C-findings.³ The predominant hepatic manifestation initially led to a diagnosis of cryptogenic cirrhosis with portal hypertension. However, the clinical presentation and physical examination findings were not consistent with cardiac ascites. The patient denied dyspnea or orthopnea and was able to tolerate a supine position without discomfort. Physical examination revealed no evidence of heart failure. In addition, imaging demonstrated no evidence of hepatic venous outflow obstruction including Budd–Chiari syndrome or portal vein thrombosis. Combined with preserved hepatic synthetic function and disproportionate severity of portal hypertension, these findings raised strong suspicion for an infiltrative systemic process. This suggests a mechanism of sinusoidal and post-sinusoidal obstruction due to mast cell infiltration, resulting in a hemodynamic profile that closely mimics cardiac

ascites.^{4,7} After excluding viral hepatitis, alcohol exposure and metabolic liver disease, definitive diagnosis was established using bone marrow biopsy, revealing dense mast cell infiltration with aberrant immunophenotypic expression, and molecular confirmation of *KIT* D816V mutation established the diagnosis of aggressive ASM.⁸⁻¹⁰

SM is a biologically and clinically heterogeneous clonal mast cell neoplasm requiring an integrated diagnostic and therapeutic approach. Current NCCN Guidelines emphasize that diagnosis relies on a combination of clinical assessment, bone marrow histopathology, immunophenotypic demonstration of aberrant mast cell markers such as CD25, CD2, or CD30, and molecular detection of activating *KIT* mutations, most commonly *KIT* D816V. In patients without cutaneous involvement,⁴ SM may present with nonspecific manifestations including cytopenias, hepatosplenomegaly, lymphadenopathy, or portal hypertension, frequently resulting in diagnostic delay, as illustrated by this case.

Advanced forms of SM, including ASM and SM with an associated hematologic neoplasm (SM-AHN) and mast cell leukemia, are collectively categorized as advanced SM(AdvSM) and are defined by the presence of C-findings reflecting mast cell-mediated organ damage. Hepatic involvement represents a particularly challenging presentation, as mast cell infiltration may lead to portal hypertension, refractory ascites and cirrhosis-like morphology despite relatively preserved hepatocellular function, closely mimicking cryptogenic cirrhosis.¹¹ This phenomenon has been increasingly recognized in infiltrative and malignant conditions and underscores the importance of considering alternative etiologies among patients with apparent cirrhosis but preserved hepatic

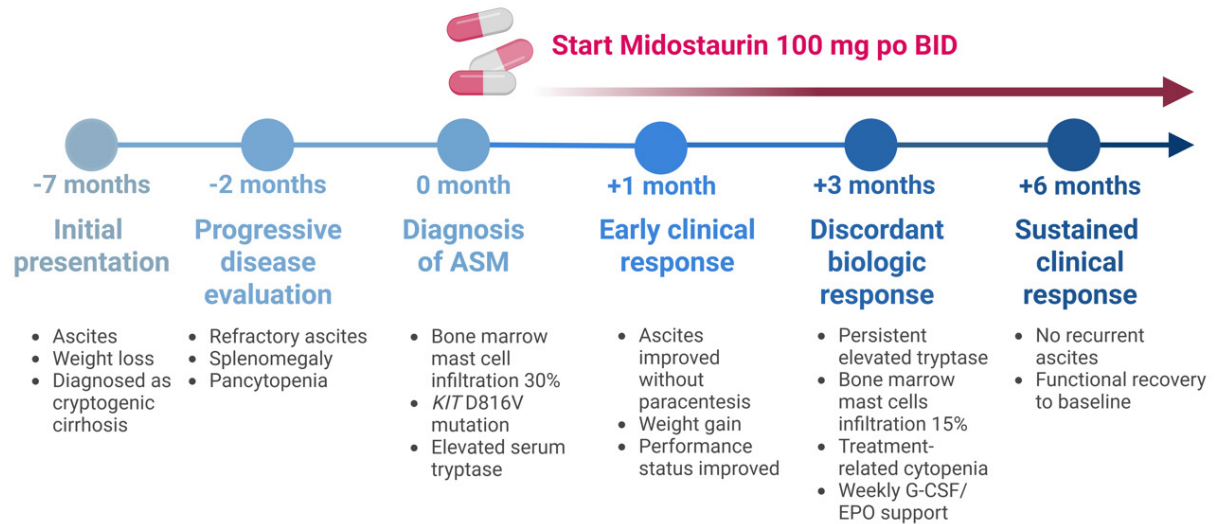


Figure 4 Clinical timeline of disease course, diagnosis, and response to midostaurin therapy.

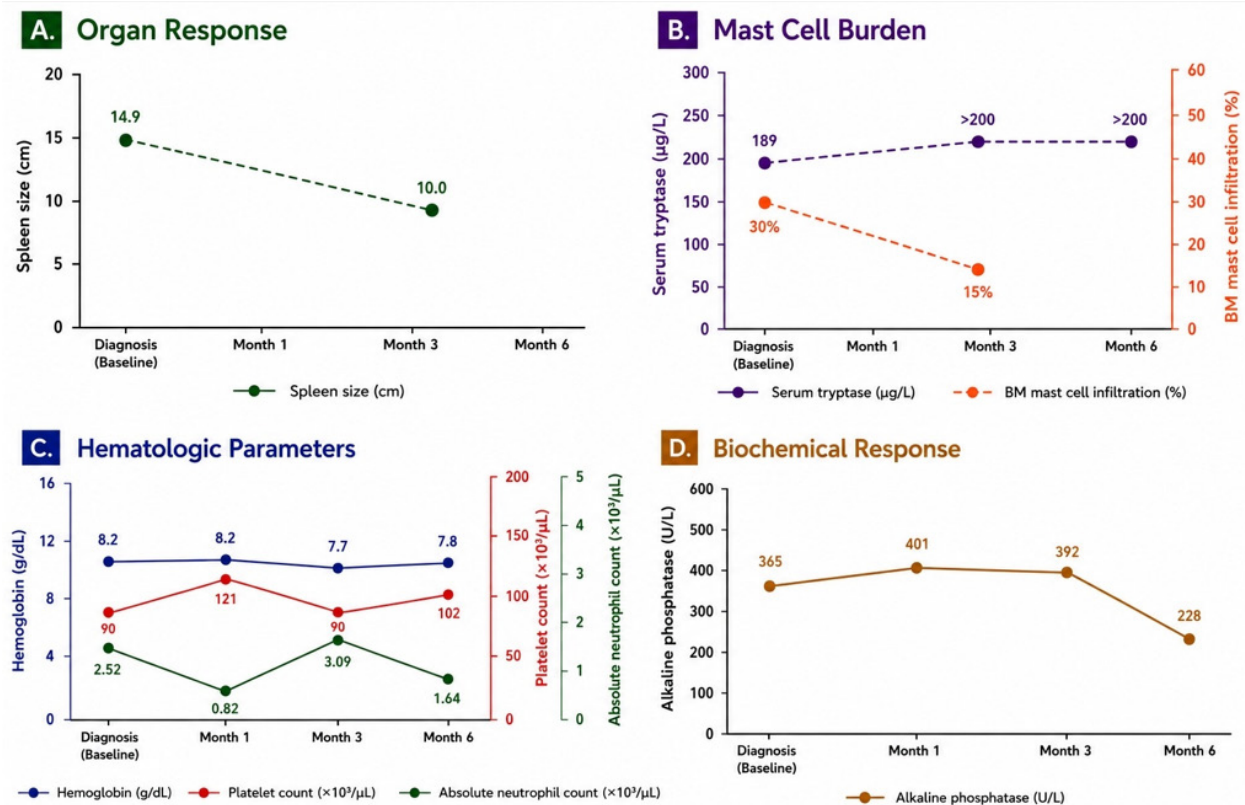


Figure 5 (A-D) Longitudinal treatment response during midostaurin therapy.

(A) Reduction in spleen size over time; (B) Serial assessment of mast cell burden using serum tryptase levels and bone marrow (BM) mast cell infiltration; (C) Serial hematologic parameters including hemoglobin (Hb), platelet count and absolute neutrophil count (ANC) and (D) Changes in alkaline phosphatase (ALP) levels during treatment follow-up

Reference ranges: Hb 13-17 g/dL; ANC $1.8-7.8 \times 10^3/\mu\text{L}$; platelet count $150-450 \times 10^3/\mu\text{L}$; ALP 40-120 U/L; serum tryptase $< 13.5 \mu\text{g/L}$

synthetic function. In the present case, although monocytosis was observed, no dysplastic features or clonal proliferation suggestive of an associated myeloid neoplasm was identified on morphologic assessment. Furthermore, NGS did not reveal additional pathogenic mutations associated with myeloid neoplasms. Taken together, these findings strongly argue against the presence of SM-AHN and support ASM as the dominant disease process driving the clinical phenotype.

Given the central role of activating *KIT* mutations in the pathogenesis of SM, particularly *KIT* D816V, targeted inhibition of *KIT* has become the cornerstone of therapy for advanced disease. Midostaurin is an orally administered multikinase inhibitor with activity against both wild-type and mutant *KIT*, including the *KIT* D816V variant.¹² In a pivotal open-label phase II trial, midostaurin demonstrated meaningful clinical activity among patients with advanced SM, including ASM, SM-AHN, and mast cell leukemia, achieving an overall response rate of approximately 60% using modified Valent and IWG-MRT response criteria. Treatment led to reductions in mast cell burden, serum tryptase levels, and organomegaly, establishing its role as a standard-of-care therapy and leading to US FDA approval at a dose of 100 mg orally twice daily in 2017.¹⁰⁻¹³

More recently, avapritinib,¹⁴ a highly selective and potent inhibitor of *KIT* D816V, has demonstrated deeper clinical and molecular responses in AdvSM. In the phase I EXPLORER and phase II PATHFINDER studies, avapritinib achieved high overall response rates of approximately 75%, with significant reductions in serum tryptase concentrations and bone marrow mast cell burden.^{2,12,14-16} The recommended starting dose is 200 mg orally once daily with dose modifications based on tolerability. These studies supported FDA approval of avapritinib for adults with AdvSM in 2021.

Emerging comparative data, although derived primarily from indirect and nonrandomized trials, suggest that avapritinib may be associated with improved overall survival and longer duration of treatment compared with midostaurin or cladribine in real-world and observational cohorts, with a trend toward greater reductions in serum tryptase and sustained treatment benefit.¹⁵ Although randomized controlled trials comparing these agents are lacking, current evidence suggests that avapritinib may offer enhanced potency and clinical impact among selected patients with AdvSM.¹⁶

In the context of the present case, midostaurin represented a rational and accessible *KIT*-targeted disease-modifying therapy, particularly in a real-world setting where treatment decisions are influenced by drug availability and reimbursement constraints. The patient experienced significant clinical improvement after one month of treatment including better performance status, weight gain, and symptom relief. However, serum tryptase levels and bone marrow mast cell burden remained unchanged at early assessment. Longitudinal follow-up will be required to clarify the trajectory of treatment response over time.

Conclusion

SM is a rare but clinically significant hematologic neoplasm with a broad spectrum of clinical manifestations. Advanced disease may cause severe organ dysfunction and may mimic more common conditions including cryptogenic cirrhosis and portal hypertension, leading to diagnostic delay. Therefore, a structured diagnostic workup incorporating histopathology, immunophenotyping and molecular testing is essential, together with risk-adapted management strategies. Increased clinical awareness and early recognition are critical to prevent irreversible organ damage and to optimize patient outcomes in the era of targeted therapy.

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