Gastrointestinal Tumors

Case Report

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# Mast Cell Sarcoma of Small Intestine, Early Diagnosis, and Good Prognosis: An Extremely Rare Case Report and Review of the Literature

Bita Geramizadeh[a, b] Sara Nabavizadeh[c, d] Alireza Rezvani[e, f]
Nadereh Shamsolvaezin[g] Alireza Zahedinassab[g] Neda Khodadadi[g]
Pouya Iranpour[h]

[a]Department of Pathology, Medical School of Shiraz University, Shiraz University of Medical Sciences, Shiraz, Iran; [b]Transplant Research Center, Shiraz University of Medical Sciences, Shiraz, Iran; [c]Student Research Committee, Shiraz University of Medical Sciences, Shiraz, Iran; [d]Otolaryngology Research Center, Shiraz University of Medical Sciences, Shiraz, Iran; [e]Department of Medicine, Medical School of Shiraz University, Shiraz University of Medical Sciences, Shiraz, Iran; [f]Hematology Research Center, Shiraz University of Medical Sciences, Shiraz, Iran; [g]Kowsar Hospital, Shiraz, Iran; [h]Department of Radiology, Medical School of Shiraz University, Shiraz University of Medical Sciences, Shiraz, Iran

**Keywords**
Mast cell sarcoma · Gastrointestinal neoplasm · Extracutaneous mastocytoma

**Abstract**
Gastrointestinal mast cell sarcoma is a rare variant of mastocytosis. It is a unifocal tumor with high destructive capacity and metastatic potential. Diagnosis of mast cell sarcoma can be challenging and might be so delayed that unfavorable prognosis may be expected. In this case report, we will describe our experience with a case of mast cell sarcoma in the small intestine of an elderly woman, which was diagnosed early on throughout the course of her disease and successfully treated. The patient was a 59-year-old woman who presented with abdominal pain, flushing, weight loss, and vomiting. Imaging studies supported the existence of an infiltrative neoplasm in the jejunum. Then, surgical removal of the tumor was performed. The presence of mast cells in the resected tumor was confirmed by immunohistochemistry, histopathology, and Giemsa staining. After almost a year of follow-up, the patient's overall condition was fine, and no signs of recurrence were found. This is the first reported case of successfully treated gastrointestinal mast cell sarcoma. All of the previously reported cases had been diagnosed after recurrence with no response to treatment. Our case shows the significance of early diagnosis and treatment in this condition and its impact on outcome and prognosis. That could be achieved only if the pathologist has a high suspicion for this rare disease and keeps it in the back of one's mind.



## Background

Mast cell sarcoma (MCS) is an unusual type of mastocytosis, which presents as a unifocal mass lesion and solid tumor containing high-grade neoplastic mast cells [1]. Clinical manifestation depends on the tumor location [2]. Because of diverse clinical manifestations

karger@karger.com
www.karger.com/gat



Correspondence to:
Bita Geramizadeh, geramib@gmail.com

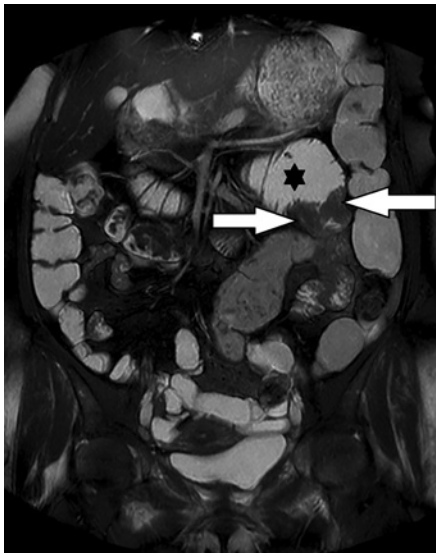

**Fig. 1.** Coronal T2-weighted image through the abdomen demonstrates focal segmental increased wall thickness of the jejunum (white arrows) with proximally distended loop (asterisks) as the result of partial bowel obstruction.

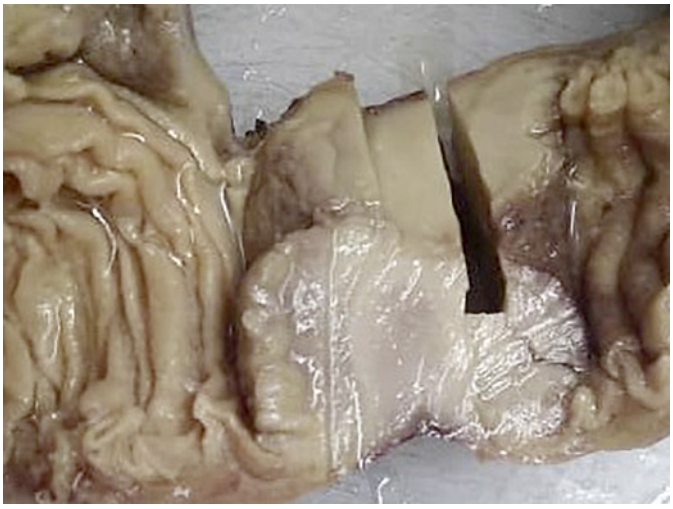

**Fig. 2.** An ill-defined mass in the jejunal wall measuring 4 × 3 × 2 cm.

and the lack of optimal therapeutic options, the mortality rate is high, and prognosis has been reported poor due to high metastatic potential, destructive infiltrations, and delayed diagnosis [3]. So far, there are very few case reports of MCS, and only limited knowledge exists on the characteristics and management of the disease. To the best of our knowledge, there have been only 5 case reports of MCS afflicting the gastrointestinal tract in the English literature so far. In all of the previous cases, diagnosis and treatment have been considerably delayed, and adverse outcomes have ensued; therefore, here, we share our experience with a case of MCS, which has been treated successfully, and after about a year of follow-up the patient was well and free of symptom.

## Case Presentation

Our patient was a 59-year-old woman presenting with a history of upper abdominal discomfort, weight loss, and vomiting for 2 months before being referred to our center. She was also complaining of hot flushes during the last 3 months. The patient's past medical history, family history, and physical examination were unremarkable. Laboratory findings demonstrated mild to moderate anemia (Hb = 10.8 g/dL, HCT = 35.4%, MCV = 81.5 fL, MCH = 24.9 pg) as well as a high lactate dehydrogenase level (378 IU/L, average level <280 IU/L). Abdominal ultrasonography failed to show any significant findings, so magnetic resonance enterography (MRE) was performed. MRE provided some evidence suggestive of a circumferential thickening pattern in the wall of the proximal part of the jejunum, accompanied by distention of the bowel loops. Moreover, shouldering appearance of the lumen's margins and mucosal destruction were also reported, which were highly suggestive of an infiltrative process. Additionally, an enlarged lymph node was seen on the posterior aspect of the greater curvature of the stomach (Fig. 1).

Since the symptoms were suggestive of small bowel obstruction and a more deeply located carcinoma could not be excluded, the patient underwent jejunal resection and anastomosis with a margin of 6.5 cm from the duodenal and 15 from the jejunal site. The intraoperative assessment demonstrated anastomotic integrity at the time of surgery; a feeding jejunostomy was placed 36 h postoperation. Six days after admission, the patient was commenced on a diet and discharged in good condition.

The received specimen in the pathology department consisted of a small segment of the jejunum, measuring 20 cm in length and 8 cm in greatest diameter. The serosal surface was smooth and gray with a palpable mass. The lumen showed a 4 × 3 × 2 cm exophytic tumor at the site of the ligamentum teres, with a circular growth pattern on the luminal surface. The tumor was invaded lamina propria, submucosa, muscularis propria, subserosa, serosa, and peri-intestinal fat, while regional lymph nodes were grossly free of tumor (Fig. 2).

Histologic investigation of the resected tumor demonstrated discohesive sheets of medium-sized immature cells with round to oval, lobulated, or indented nuclei. These cells mostly had clear to eosinophilic cytoplasm (Fig. 3). Furthermore, Giemsa staining showed a metachromatic tone in the tumor cells' cytoplasm, a finding that supports the mast cell origin of the tumor.

On immunohistochemistry assay, LCA, and C-kit (CD117) were positive in tumor cells while CD1a, CD20, CD3, MPO, Cytokeratin (AE1/AE3, CK7, CK19, and CK20), and Vimentin tests showed negative results; therefore, this neoplasm was confirmed to be a true MCS. Finally, the Ki-67 marker of proliferation was measured, and the results showed levels as high as 30%. As no bone marrow cytogenetic abnormality or involvement has been found in this patient, systemic mastocytosis was ruled out. KIT mutation analysis showed gain-of-function mutations in the KIT receptor tyrosine kinase, an aspartic acid to valine substitution (D816V).

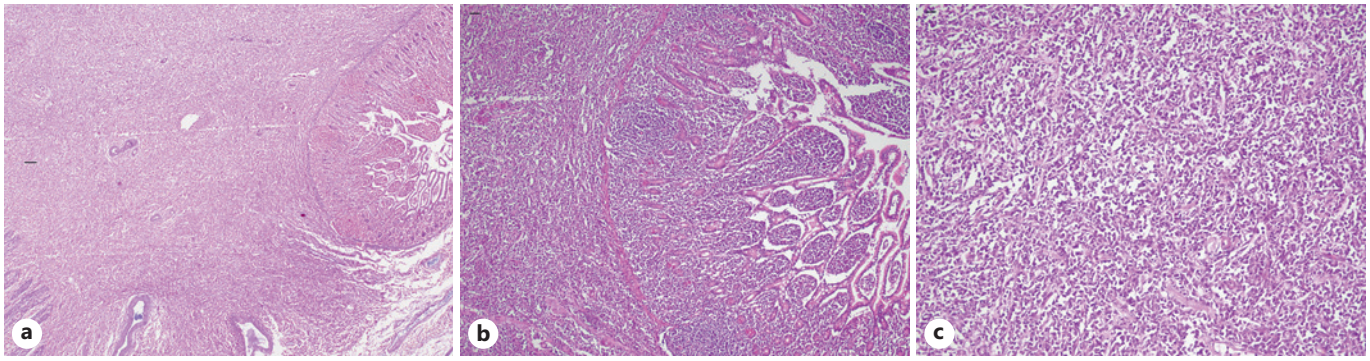


**Fig. 3. a–c** Sections from jejunal mass show diffuse infiltration of atypical cells with high N/C ratio and hyperchromatic nuclei (in three powers by H and E stain).

After the operation, the patient was put on a regimen of Imatinib, 400 mg once a day for 6 months. So far, the patient has been followed, with no evidence of metastasis or recurrence.

## Discussion

MCS is a rare, aggressive solid tumor with high metastatic potential. WHO classified the disease in 2016 as a variant of mastocytosis, presenting with high-grade unifocal tumors with no bone marrow or other extracutaneous organ infiltration [4].

Although MCS is common among animals, only a few cases have been reported in humans (less than 30). Among all, the entire published cases on MCS located in the gastrointestinal tract to date consist of five case reports in the English literature; therefore, the experience on the management of the condition is sparse. Moreover, lack of resemblance in clinical histological, and genetic features, to other mast cell neoplasms could make its diagnosis even more challenging [5]. Most MCS-reported cases had initially been misdiagnosed as lymphomas, mastocytosis, acute myeloid leukemia (AML), or Langerhans cell histiocytosis, further delaying proper treatment. These difficulties in diagnosis, progression to mast cell leukemia, and failure of conventional chemotherapy led to poor prognosis and a short life expectancy [5, 6].

Herein, we report a case of small intestinal MCS with an early diagnosis and successful treatment. Furthermore, the demographic, clinical, and histological characteristics of previous literature on GI tract MCS are summarized in Table 1 [2, 7–9].

There were three female patients and 2 reported male patients, all middle-aged (32–66 years old). The clinical manifestations of GI tract MCS are not different from other neoplasms in that region. Based on the review of available literature, almost all patients had initially presented with abdominal pain and anemia; this was also true in our case, who presented with abdominal pain, anemia, vomiting, and was hospitalized with signs of intestinal obstruction. Laboratory findings in these patients reported microcytic anemia with normal to high levels of lactate dehydrogenase. Our patient had a history of hot flushes since several months before surgery, which was completely relieved after tumor resection.

Reported imaging studies were ultrasonography, CT scan, magnetic resonance imaging, and MRE [10]. The most common radiological findings were dilated bowel loops along with circumferential thickening of the intestinal wall and enlarged lymph nodes. Nevertheless, no specific imaging hallmark was reported for MCS.

The gold standard for the diagnosis of MCS is a tissue diagnosis. However, due to the considerable morphological similarity of GI tract MCS with other GI tract neoplasm, broad immunohistochemical assessment is necessary.

Among the differential diagnoses of this condition are Langerhans cell histiocytosis and GI tract lymphoma. In resemblance to our case of MCS, Langerhans cell histiocytosis generally shows tumor cells with abundant, pale eosinophilic cytoplasm, and irregular nuclei. In addition, frequent admixed eosinophils are seen in both neoplasms. Nevertheless, longitudinal nuclear grooves, indistinct cytoplasmic borders, and immunohistochemical expression of S100, langerin, cyclin D1, and CD1a in Langerhans cell histiocytosis could be helpful for the diagnosis. Our case was negative for CD1a, and there have not been any eosinophil or Langerhans cells [11].

The main differential diagnosis of GI tract MCS is non-Hodgkin's lymphoma. Our case was negative for CD markers of lymphoma [12].

Although there is no definite treatment protocol for MCS due to the condition's rarity, the best reported therapeutic option is the surgical removal of the tumor with the combination of multiple modalities [3]. Some former studies have

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**Table 1.** Clinical and pathologic characteristics of gastrointestinal MCS

| Author/ Year | Age/ Sex | Tumor location | Symptom | Tumor size (cm) | KIT mutation | Immunohistochemical markers | Tryptase level | Bone marrow involvement | Mastocytosis | Bone marrow involvement | Treatment | Follow-up |
|---|---|---|---|---|---|---|---|---|---|---|---|---|
| Kojima, 1999 | 32/F | Ascending colon | Abdominal pain | NR | NR | Positive: CD45, CKIT Negative: CD30 | NR | NR | Yes | NR | Surgery, steroid, and radiation | Died after 2 years |
| Schwaab, 2013 | 62/F | Sigmoid | Abdominal pain | 4 | D816V | Positive: CD45, CKIT Negative: MPO | 350 μg/L | Yes | Yes | Yes | Surgery, cladribine | Died after 7 months |
| Bugalia, 2015 | NR*/M | Ileum | Abdominal pain | 4.5 | Asn-822-Lys (exon 17) | Positive: CD45, CKIT | NR | NR | No | No | Surgery, steroid, and imatinib | Alive after 6 months |
| Monnier, 2016 | 66/M | GI tract | NR | NR | Wild-type KIT (KIT WT) | Positive: CD45, CKIT Negative: MPO | NR | Yes | Yes | NR | NR | Alive after 4 months |
| Monnier, 2016 | 39/F | GI tract | NR | NR | Wild-type KIT (KIT WT) | Positive: CD45, CKIT Negative: MPO | NR | Yes | Yes | NR | NR | Died after 5 months |
| Current case | 59/F | Jejunum | Abdominal pain | 4 | D816V | Positive: CD45, CKIT Negative: MPO | NR | No | No | No | Surgery and imatinib | Alive after 12 months |

claimed that Kit sequencing could have a crucial role in the prognosis of the disease and its response to therapy. An acquired KIT D816V mutation is resistant to therapy with a tyrosine kinase inhibitor, while the tumors lacking this mutation show promising results [13]. However, our results showed good prognosis despite the presence of mutation.

In conclusion, our study describes a patient who suffered from a MCS of the small intestine. Our patient is the only one with a history of hot flushes (secondary to histamine release, which has been resolved after operation). The patient's early diagnosis and proper treatment resulted in the patient's life being saved, and her life expectancy increased. All the previously reported cases had been diagnosed after recurrence, when no response to treatment was obtained with a second pathologic opinion; therefore, early diagnosis and keeping this disease in mind will prevent delayed diagnosis and poor prognosis.

## Statement of Ethics

We have written informed consent to publish the details of their medical case and any accompanying images. This study protocol was reviewed and approved by Ethics Committee of Shiraz University of Medical Sciences (44–21).

## Conflict of Interest Statement

The authors have no conflicts of interest to declare.


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There is no sponsor and no funding source.


## Author Contributions

All the authors have contributed in the case report. Bita Geramizadeh: diagnosis of the pathology, writing and editing the paper, and searching the literature; Sara Nabavizadeh: helping to write the paper, searching the literature, and collecting patients' data; Alireza Rezvani: treatment of the patient and writing the paper and surgery of the patient; Nadereh Shamsolvaezin and Neda Khodadadi: helping to collect the patient's data; Pouya Iranpour: radiologic diagnosis of the case and writing the paper.

## Data Availability Statement

Further inquiries can be directed to the corresponding author. All the data are available in the patient's clinical chart.

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