

Catching the GIST: Massive Gastrointestinal Stromal Tumor Presenting as Acute Dyspnea

Hytham Rashid¹, Johnathon Chung^{1*}, Mimi Phan Haghshenas¹, Michael Harrison Humphries¹,
Sivatej Sarva²

1. Department of Internal Medicine, University of Houston/HCA Houston Healthcare Kingwood, TX, USA

2. Department of Critical Care, University of Houston/HCA Houston Healthcare Kingwood, TX, USA

* Correspondence: Johnathon Chung DO, Department of Internal Medicine, University of Houston/HCA Houston Healthcare Kingwood,
TX, USA

(✉ chungjohnathon@gmail.com)

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ABSTRACT

We present a case of a 55-year-old woman presenting with worsening shortness of breath and constipation over the course of three days. Initial computed tomography scan showed a large, complex abdominal mass with a vascular pedicle and possible pedunculated origin along the inferior aspect of the greater curvature of the stomach. The mass was further evaluated on magnetic resonance imaging showing an active hemorrhage. The patient became hemodynamically unstable and general surgery was consulted for evaluation. Mass resection was performed, and biopsy revealed KIT/CD117+ and DOG1/ANO1+ gastrointestinal stromal tumor staged as T4. Although definitive diagnosis of a gastrointestinal stromal tumor requires biopsy, prompt clinical and radiological recognition is critical for patients to receive definitive treatment of mass resection.

CASE REPORT

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A 55-year-old Caucasian woman presented to the emergency department complaining of worsening shortness of breath and constipation over three days. The patient reported her abdomen seemed to be enlarging over the past year despite diet and exercise. She reported symptoms of constipation, anorexia, generalized headache and weakness. She believed these symptoms were due to her history of irritable bowel syndrome. The patient reported a 10 pack-year tobacco smoking history. She denied any family history of cancers.

On initial physical exam, the patient was hemodynamically stable with a markedly distended, but non-tender abdomen. Laboratory analysis showed normocytic anemia with a

hemoglobin of 8.3 g/dL (normal: 12-16g/dL). Additional laboratory analysis revealed an elevated cancer-antigen-125 of 138 U/mL (normal: <38.1 U/mL). Computed tomography (CT) angiography of the chest revealed no pulmonary embolus. However, CT scan of the abdomen with intravenous contrast revealed a large, complex mass that measured 30 x 29 x 18 centimeters (cm) with a vascular pedicle and possible pedunculated origin along the inferior aspect of the greater curvature of the stomach (Figure 1). Secondary mass effect on the abdominal organs, bowels, and lungs was noted. Magnetic resonance angiography of the abdomen showed much of the mass in its central portion appeared to demonstrate T1 hyperintensity suggesting internal hemorrhage (Figure 2). Blood pressure recordings in the emergency department

showed the patient becoming increasingly hypotensive and tachycardic. General surgery and oncology were consulted for further evaluation.

The patient was transported to the operating room, and the mass was resected from the posterior stomach (Figure 3A). During the operation, the mass was found to be supplied by multiple vascular beds, including a large branch of the left gastric artery. The pathology identified a KIT/CD117+ and DOG1/ANO1+ gastrointestinal stromal tumor (GIST) staged as T4 (Figure 3B, 3C). The patient's post-operative course was unremarkable, and she was discharged with outpatient oncology follow up for the initiation of imatinib therapy. On outpatient follow-up one month later, she reported resolution of her shortness of breath and gastrointestinal symptoms.

DISCUSSION

Etiology & Demographics:

GISTs arise from a mutation in the KIT protein or platelet derived growth factor receptor alpha (PDGFRA). These sporadic mutations cause activation of tyrosine kinase receptors, which lead to hyperplasia and ultimately neoplasia [1]. GISTs are the most common mesenchymal tumors of the gastrointestinal (GI) tract, accounting for 80% of all such tumors [1]. Of all GI malignancies, GISTs comprise of up to 3%. GISTs are usually diagnosed in patients over the age of 40. [1]. These tumors affect men and women equally, while some researchers report a possible male predilection [2].

Clinical & Imaging Features:

The clinical presentation of patients with GISTs vary depending on the size and location of the tumor. Most commonly, patients present with gastrointestinal bleeding which may be acute or chronic. Patients with acute GI bleeding may present with melena or hematochezia. Chronic GI bleeding may present as anemia. In addition to GI bleeding, GISTs may also present with signs and symptoms of mass effect and nonspecific abdominal pain or discomfort. 15% to 30% of GISTs are found incidentally on surgery, imaging, or autopsy [1].

While not recommended as the initial imaging modality of choice, abdominal radiographs in patients with a GIST may show a nonspecific soft-tissue mass indenting or displacing the gastric air shadow with peripheral enhancement [2]. In barium studies, GISTs have the classic features of submucosal masses, like those of leiomyomas or leiomyosarcomas. Central areas of low attenuation correspond to hemorrhage, necrosis, or cyst formation within the tumor. On CT, GISTs are typically non-enhancing masses with areas of low attenuation from hemorrhage, necrosis, or cyst formation. Magnetic resonance imaging (MRI) features of GISTs vary depending on the degree of necrosis and hemorrhage. Solid portions of the tumor are typically low signal intensity on T1-weighted images and high signal intensity on T2-weighted images. Signals will vary at areas of hemorrhage within the tumor on both T1- and T2-weighted images [2]. In the case of our patient, the MRI showed

high-signal intensity on T1-weighted images, likely corresponding to the hemorrhage associated with the tumor.

Treatment & Prognosis:

The definitive treatment of GISTs is surgical resection [3]. The current guidelines for all GISTs from the National Comprehensive Cancer Network recommend a tyrosine kinase inhibitor, such as imatinib, as initial treatment whether the tumor is unresectable, metastatic, or post-operation [1]. Prognosis for local, noninvasive tumors is positive. However, as is the case for many tumors and malignancies, prognosis widely varies depending on the size, location, and mitotic count [1]. Other factors affecting prognosis include the presence of negative margins and avoidance of tumor rupture at the time of resection [4].

Differential Diagnoses:

Gastrointestinal leiomyoma

Like GISTs, gastrointestinal leiomyomas are prone to cause gastrointestinal bleeding [5]. Clinical presentation depends on tumor size, location, and the presence or absence of ulcerations. Larger leiomyomas are more prone to ulcerations and bleeding. However, most leiomyomas are found incidentally in asymptomatic patients. On CT, leiomyomas are smooth, solid masses with homogenous contrast enhancement [6].

Gastric lymphoma

Gastric lymphomas represent the most common site of extranodal lymphomas, accounting for 25% of all such lymphomas. However, gastric lymphomas comprise of only 1-5% of all gastric malignancies [7]. Gastric lymphomas also present later in life with no gender predilection. Gastric lymphomas can be associated with *Helicobacter pylori* infections (mucosa-associated lymphoid tissue lymphoma) or sporadic mutations (non-Hodgkins lymphoma) [8]. While it may be difficult to differentiate gastric lymphomas from GISTs on imaging, gastric lymphomas often present with retroperitoneal and local lymph node enlargement [9]. On CT, gastric lymphomas present as a homogenous mass with focal areas of low-density representing necrosis.

Gastrointestinal schwannoma

Gastrointestinal schwannomas typically present at about the 3rd to 5th decade of life [9]. Patients may present with vague abdominal discomfort or gastrointestinal bleeding due to the ulceration potential of these tumors. Pathologic evaluation can differentiate schwannomas from GISTs, as schwannomas are positive for S100 protein, which can be targeted and stained by immunohistochemistry [10]. On CT, gastrointestinal schwannomas are well-defined, rounded masses with homogenous attenuation [11]. On MRI, these tumors will present as low to intermediate intensity on T1-weighted images and high intensity on T2-weighted images [9].

Gastrointestinal carcinoid

Carcinoid tumors are neuroendocrine tumors that can arise in a number of locations. Most commonly, these tumors originate at the distal ileum and have the ability to secrete serotonin [12]. Due to these tumor's ability to produce serotonin, patients may present with symptoms of carcinoid

syndrome. These patients may present with diarrhea, flushing, right heart strain, bronchospasm, or vague abdominal pain [13]. On CT, carcinoid tumors are hyper-enhancing and hyper-vascular. On positron emission tomography (PET) scan, there is increased tracer uptake seen at the site of the carcinoid tumors [14].

TEACHING POINT

Gastrointestinal stromal tumors may present with a constellation of vague, nonspecific symptoms. At the same time, their radiographic findings may be nonspecific for a wide array of differential diagnoses. However, recognition of key clinical, radiographic, and pathologic features can be essential in obtaining definitive treatment.

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FIGURES

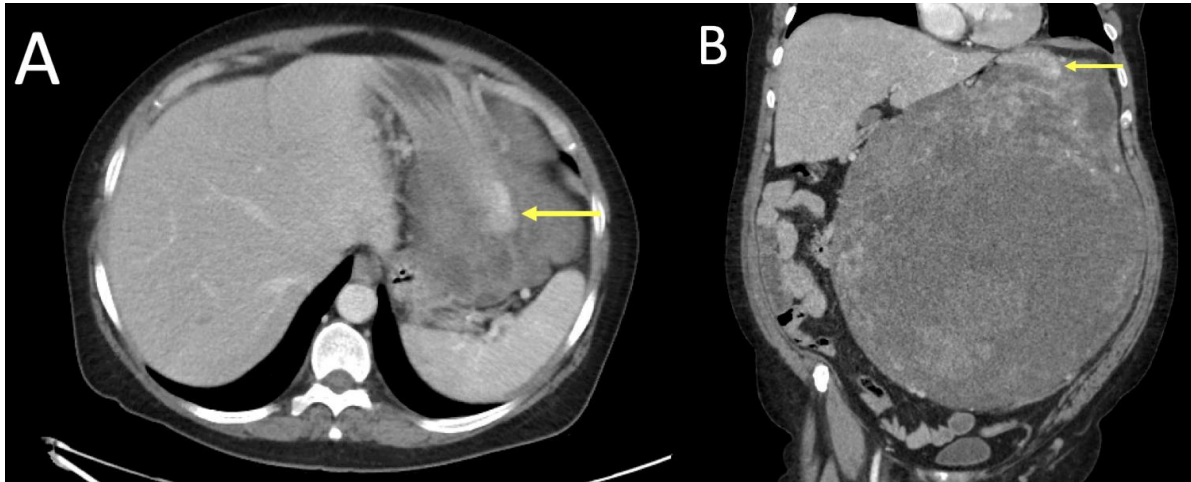


Figure 1: 55-year-old female with a gastrointestinal stromal tumor

Findings: Large, complex abdominal mass measuring 30 x 29 x 18 cm, with a vascular pedicle (yellow arrow) and possible pedunculated origin along the inferior aspect of the greater curvature of the stomach.

Technique: (A) Axial CT images were acquired at 178-551 mAs, 120 kV, and 5 mm slice thickness. (B) Coronal CT images were acquired at 140 mAs, 120 kV, and 2.5 mm slice thickness. Images obtained with GE Revolution EVO CT scanner using 100 mL iopamidol contrast solution.

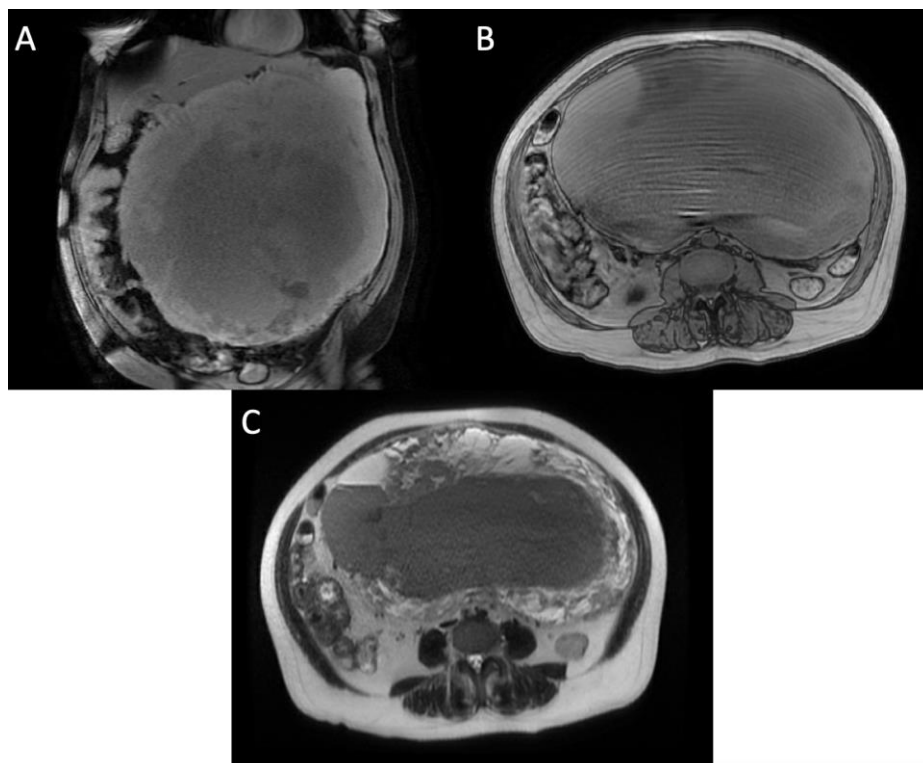


Figure 2: 55-year-old female with a gastrointestinal stromal tumor

Findings: A large, heterogeneous mass is visualized within the abdomen and pelvis, measuring approximately 30.1 cm in transverse dimension, 17.2 cm in AP dimension, and 30 cm in cranial caudal dimension. The majority of the mass appears to demonstrate T1 hyperintense signal suggesting internal hemorrhage. Peripherally within the mass, there is thick rind of heterogeneous enhancement seen.

Technique: (A) Coronal and (B,C) axial views obtained using a 1.5T GE Optima MRI scanner. The T1-weighted, fat saturated coronal image was performed using 16 mL gadolinium (Dotarem) contrast solution with a TE: 2.98, TR: 350. The T1-weighted pre-contrast axial image was performed with a TE: 2.012, TR: 335. (C) The pre-contrast T2-weighted image was performed with a TE: 82.784, TR: 509.492

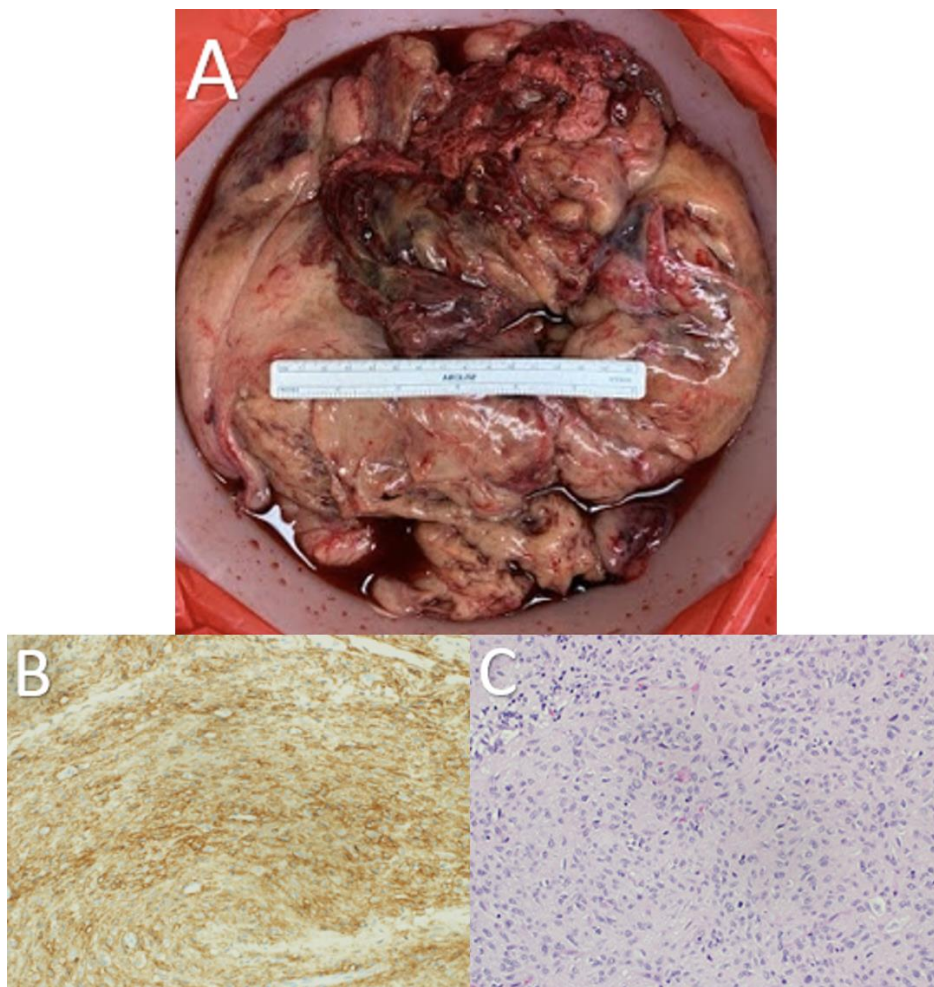


Figure 3: 55-year-old female with a gastrointestinal stromal tumor

Findings: A 27 x 26 x 8 cm yellow, tan to red, lobulated, disrupted, fragmented soft tissue mass was removed from the posterior gastric wall (A). The gastrointestinal stromal tumor was found to be of mixed spindle and epithelioid type (B, C)

Technique: Pathologic evaluation of the locally excised abdominal mass

Etiology	Mutations of the KIT protein or PDGFRA are seen in approximately 85% of sporadic cases
Incidence	<ul style="list-style-type: none"> • 80 % of mesenchymal tumors of the GI tract • 0.1-3% of all GI malignancies
Gender Ratio	GISTs occur equally among both males and females
Age predilection	Most commonly diagnosed later in life, with a median age in the 60s
Risk Factors	No known
Treatment	<ul style="list-style-type: none"> • Localized tumor: resection • Locally advanced: pre-operative imatinib before resection • Unresectable or metastatic disease: tyrosine kinase inhibitors – imatinib is first line
Prognosis	Dependent primarily on the tumor site, mitotic count, and tumor size. Other factors include presence of negative margins and avoidance of tumor rupture at the time of resection.
Findings on imaging	<ul style="list-style-type: none"> • Computed tomography (CT): soft tissue density with central areas of lower density when necrosis is present that occasionally appear as fluid-fluid levels. Enhancement is typically peripheral. • Magnetic resonance imaging (MRI): <ul style="list-style-type: none"> - T1: low signal intensity solid component. Peripheral enhancement - T2: high signal intensity solid component

Table 1: Summary table of gastrointestinal stromal tumors.

	X-Ray	Ultrasound	CT	MRI	PET
Gastrointestinal stromal tumor	If large, soft tissue density displacing bowel loops or other organs	Hypoechoic, solid mass	Soft tissue density with central areas of lower density	T1: low signal, solid; peripheral enhancement T2: high signal, solid	Areas of necrosis can have low or absent tracer uptake
Gastrointestinal leiomyoma	Not generally utilized	Utilized for uterine leiomyomas Usually hypoechoic compared to myometrium	Well-defined, solid mass with smooth contours and low, homogenous contrast enhancement	Low signals on T1 and T2 weighted imaging	Not generally utilized
Gastric lymphoma	Appearance on barium swallow studies may include bull's eye appearance, filling defects, or thickened gastric rugae	Hypoechoic, homogenous, and solid mass	Marked thickening of the stomach wall. Homogenous in attenuation, with focal areas of low-density representing necrosis.	T1: homogenous, and intermediate in signal intensity T2: heterogeneously increased signal intensity	Increased tracer uptake
Gastrointestinal schwannoma	Not generally utilized	Hypoechoic, heterogenous echogenicity	Well-define, round masses with homogenous attenuation	T1: low or intermediate signal T2: high signal	Increased tracer uptake
Gastrointestinal carcinoid	Not generally utilized	Not generally utilized	Hypervascular mass, associated with gastric wall thickening	T1: low signal T2: high signal	Increased tracer uptake

Table 2: Differential diagnosis table for gastrointestinal stromal tumors.

ABBREVIATIONS

CM = centimeter
 CT = computed tomography
 GI = gastrointestinal
 GIST = gastrointestinal stromal tumor
 MRI = magnetic resonance imaging
 PDGFRA = platelet derived growth factor receptor alpha

KEYWORDS

gastrointestinal stromal tumor; magnetic resonance imaging, magnetic resonance angiography; computed tomography, surgical resection; kit protein

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