Jejunal Gastrointestinal Stromal Tumor Presenting as Recurrent Lower GI Bleeding and Anemia: A Case Report

Abstract

Gastrointestinal stromal tumor (GIST) is the most common type of soft tissue sarcoma found in the gastrointestinal (GI) tract. These tumors originate from the interstitial cells of Cajal (ICC), which regulate GI motility. GISTs account for approximately 1% of all gastrointestinal neoplasms, with the stomach being the most common site (60%). The small intestine is the second most frequent location (20–30%), while the jejunum accounts for only 10%. Rarely, GISTs arise in the colon, rectum, or esophagus.

Typically, GISTs affect individuals aged 50–60 years and are often asymptomatic, making diagnosis challenging. In rare cases, jejunal GISTs can present as recurrent lower GI bleeding, posing additional diagnostic difficulties preoperatively. While computed tomography (CT) is the first-line imaging tool for diagnosis, confirmation requires pathological evaluation and immunohistochemistry. The hallmark feature of GIST is the expression of the CD117 antigen, which is 95% specific for these tumors.

We report a rare case of jejunal GIST in a 63-year-old male presenting with recurrent lower GI bleeding and anemia for five years. CT imaging revealed a 4.6 × 3.5 cm mass in the proximal jejunum. Initial diagnostic laparoscopy was challenging due to the mass's proximity to the duodenojejunal (D-J) junction. Subsequently, an open exploratory laparotomy was performed, allowing successful excision of the tumor with a 2.5 cm margin on each side. Reconstruction was achieved through a jejunojejunal end-to-end anastomosis. This case highlights the diagnostic and surgical challenges of jejunal GISTs and underscores the importance of a multidisciplinary approach to management.

Key words

Gastrointestinal stromal tumors (GIST), Jejunal GIST, Lower GI bleeding, CD 117, surgical management, immunohistochemistry

Introduction

In 1941, Golden and Stout described mesenchymal tumors originating from smooth muscle cells, categorizing them as leiomyomas, leiomyosarcomas, or leiomyoblastomas. The term *GIST* was first introduced by Mazur and Clark in 1983. Following the discovery of the KIT protein (CD117) in 1999, these mesenchymal tumors were reclassified as GISTs. [1,2,4]

Characteristics and Pathological Features

Approximately 70% of GIST patients develop symptoms, while 30% remain asymptomatic. Pathological analysis identifies three main cell types: [5,6]

- Spindle cell GIST (70%), Epithelioid cell GIST (20%), Mixed cell type GIST (10%)

The size of GISTs can vary widely, from less than 1 cm to over 40 cm, with an average size of 5 cm at clinical diagnosis.[13]

While most GISTs are sporadic, 5–10% are associated with specific conditions:

1. Carney Triad: GIST, paraganglioma, and pulmonary chondroma
2. Carney–Stratakis Syndrome: GIST and paraganglioma
3. Neurofibromatosis Type 1
4. Familial GIST: Associated with mutations in KIT (CD117) and PDGFRA genes. [1,2,7]

Types of GISTs

1. Benign GISTs (70%): Typically, smaller (2–5 cm) and curable.
2. Malignant GISTs (30%): Cancerous tumors capable of invading abdominal tissues, the peritoneal cavity, liver, and lungs. Notably, GISTs rarely metastasize via the lymphatic system. [2,3]

Common Locations of GISTs

- Stomach: 60%, Small intestine: 30%, Duodenum: 5%, Rectum: 3%, Colon: 1%, Esophagus: <1%
- Extra-gastrointestinal sites: Mesentery, omentum, retroperitoneum, appendix, gallbladder, pancreas, and pelvic organs. [1,2,5]

Symptoms of GISTs

- Bleeding (30–40%): Hematemesis, melena, or black stools, Nausea and vomiting
- Abdominal pain: Severe in cases of rupture or perforation
- Loss of appetite
- Early satiety
- Anemia: Due to obscure bleeding
- Weight loss and fatigue
- Obstruction: Small bowel obstruction or intussusception [8,9]

Diagnosis of GISTs

1. Blood Tests: Assess for anemia [13].
2. Imaging:
    - CT scan: Reveals a soft tissue mass with central areas of necrosis; calcifications are uncommon.
    - MRI: Differentiates solid and necrotic components.
    - PET scan: Useful for staging. [10,12]
3. **Endoscopic Procedures:**
    - Upper GI endoscopy or colonoscopy.
    - Endoscopic ultrasound with fine-needle biopsy.
    - Capsule endoscopy for lesions in the jejunum or small intestine.
    - Double balloon enteroscopy [2,4,6]

Pathological and Molecular Analysis

Biopsy is crucial for determining the mitotic rate (low, intermediate, or high grade). Immunohistochemistry and molecular analysis confirm diagnosis:

- KIT mutations: Present in 90% of GISTs.
- PDGFRA mutations: Found in 10% of GISTs.
- Immunohistochemical staining: 95% of GISTs are positive for CD117, and 80% stain positive for CD34 or DOG1. [1,2,4]

Case Report

We present the case of a 63-year-old man who arrived at our hospital with complaints of chronic lower gastrointestinal (GI) bleeding and severe anemia. He had experienced lower GI bleeding for the

past five years. One year ago, his hemoglobin level had dropped to 5 g/dL, necessitating the transfusion of five units of blood.

Initial evaluations included an upper GI endoscopy, which showed normal findings, and a colonoscopy, which revealed black tarry stools in the transverse and descending colon without evidence of intraluminal colonic growth. A contrast-enhanced computed tomography (CT) scan of the abdomen identified a large mass in the proximal jejunum measuring 4.7 × 4.2x3.6 cm. The diagnosis of jejunal GIST causing lower GI bleeding was confirmed.

On physical examination, the patient was alert and oriented but appeared pale. His abdomen was soft and non-tender. Laboratory investigations revealed anemia (hemoglobin: 8.5 g/dL) with normal renal and liver function tests. Preoperatively, the patient received two units of packed red blood cells.

The preoperative diagnosis posed significant challenges due to the tumor's location near the duodenojejunal junction. Under general anesthesia, a diagnostic laparoscopy was performed. However, the tumor's location near the duodenojejunal (D-J) junction made it difficult to mobilize laparoscopically, prompting the decision to proceed with a midline exploratory laparotomy.

During surgery, a tumor was located in the proximal jejunum, 5 cm distal to the D-J junction. The mass was lobulated, smooth-surfaced, and measured 4.7 × 4.2x3.6 cm. Segmental resection of the jejunum with a 2.5 cm margin on either side of the tumor was performed. Reconstruction was achieved through an end-to-end hand-sewn jejunojejunal anastomosis. A drainage tube was placed at the anastomosis site on the left side of the abdomen.

Gross examination revealed a lobulated jejunal mass with central ulceration, measuring 4.7 × 4.2x3.6 cm. microscopic findings are Sections reveal a spindle cell neoplasm centered in submucosal location with surface mucosal ulceration. It comprises intersecting fascicles and whorl like arrangement of spindle cells with collagen-like substance in background. The cells show bland stout nuclei with abundant cytoplasm. Mitoses are almost not seen. No necrosis seen. Resection margins are unremarkable Immunohistochemistry revealed strong positivity for CD117, confirming the diagnosis of GIST.

Conclude with: "Postoperative recovery was uneventful, and the patient was discharged with no recurrence reported on follow-up. No postoperative complications or recurrent lower GI bleeding episodes have been reported. Adjuvant therapy with imatinib was initiated, and the patient was advised to follow up every 3–6 months for ongoing monitoring and evaluation. **(Fig 1-9)**

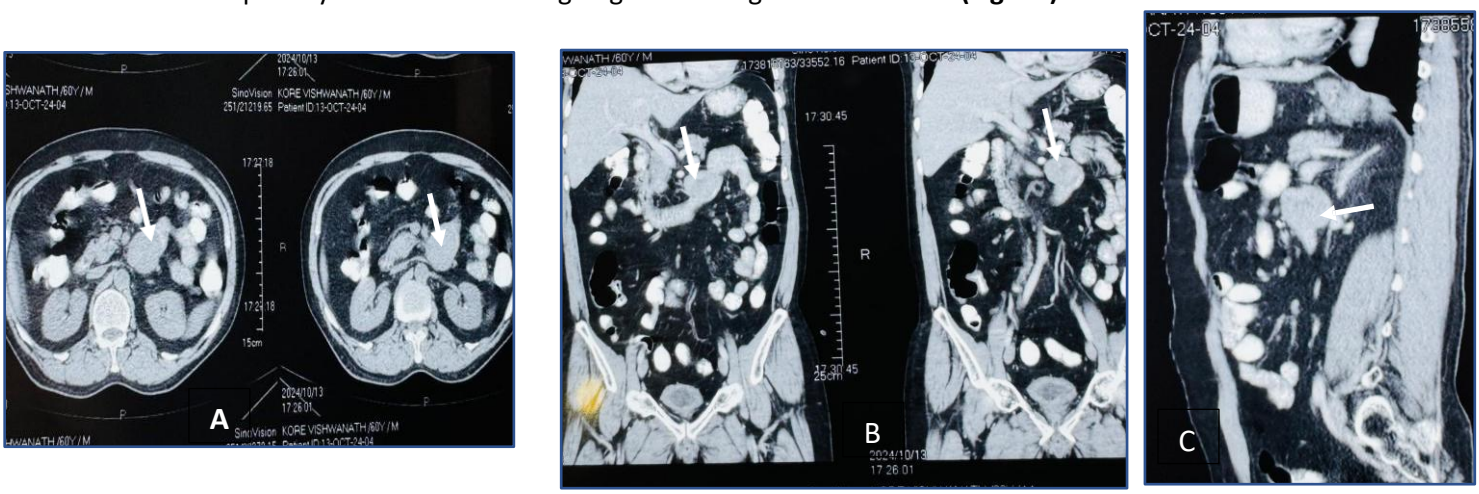


Fig 1 A – axis view, B- Coronal view, C- Sagittal view CT abdomen showing a proximal jejunal soft tissue mass, measuring 4.6x3.5cm.

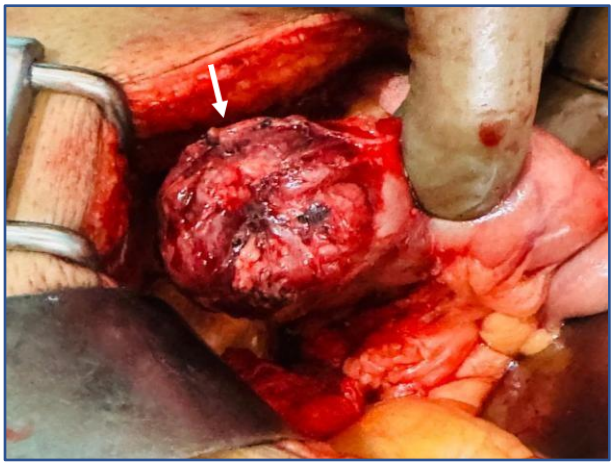


Fig-2 Intra operative photograph showing an extra luminal lobulated, smooth surface mass at proximal jejunum

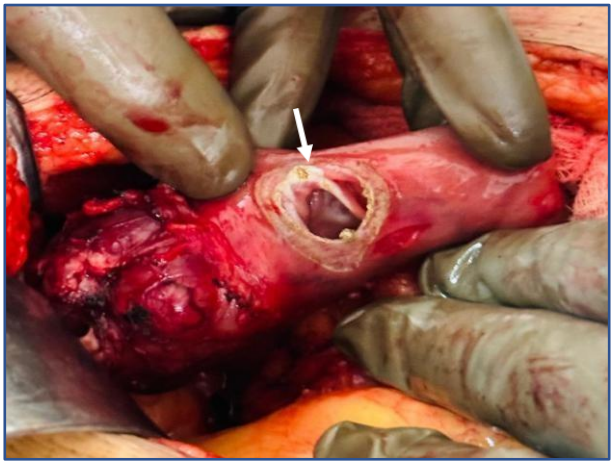
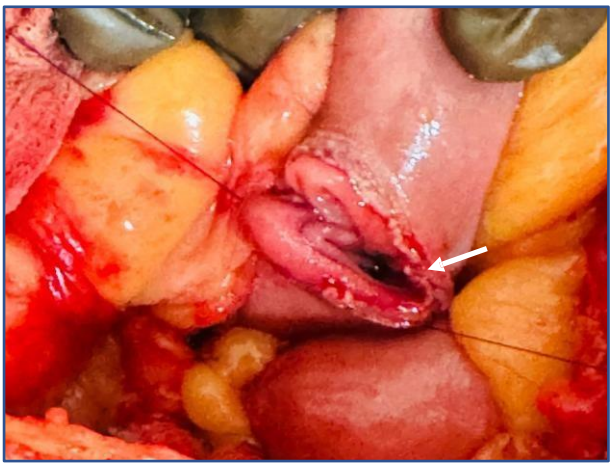


Fig-3 Intra operative photograph showing excision of jejunal mass on either side with 2.5 cm margin



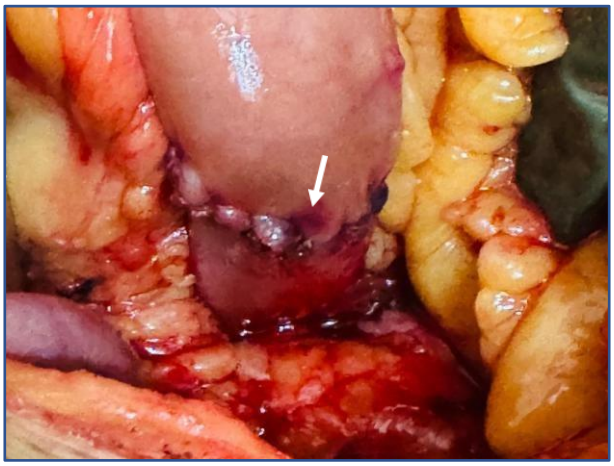


Fig-4,5 Intra operative photograph showing end to end Jeju-jejunal anastomosis

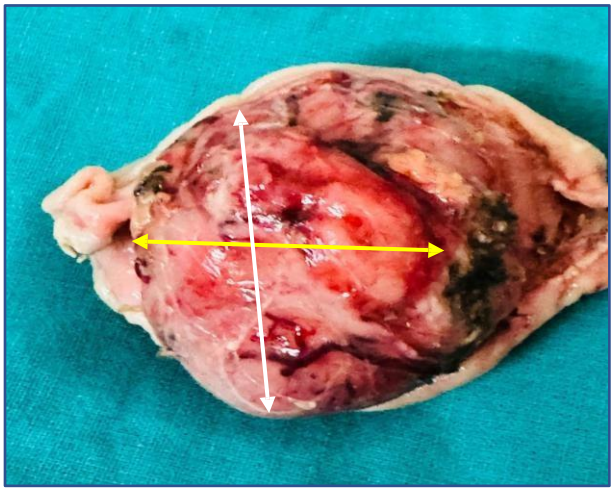


Fig-6 Photograph showing on gross extra-luminal mass measuring 4.7 × 4.2x3.6 cm

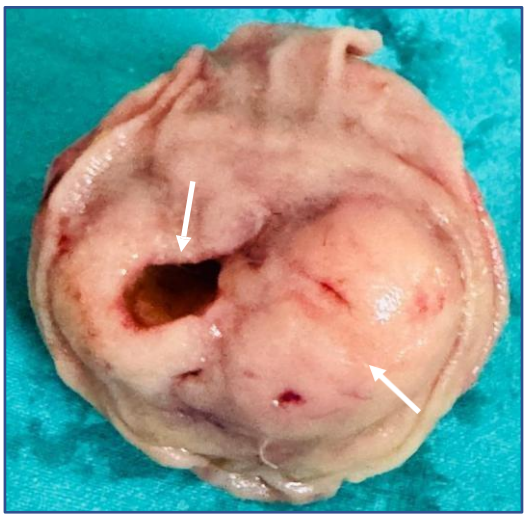


Fig-7 Intra-luminal cut surface well-circumscribed, smooth lobulated mass with ulcerated area

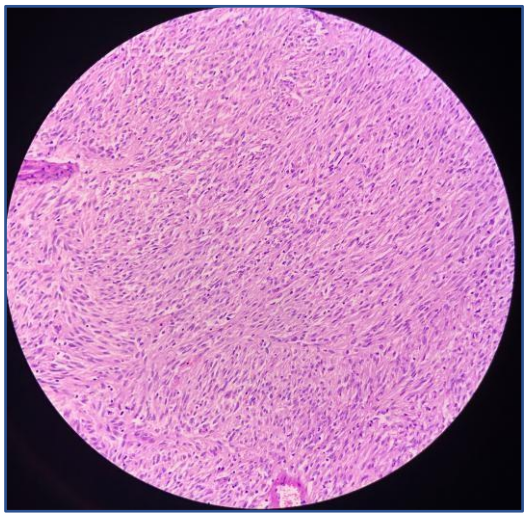


Fig-8 Histopathology photograph showing spindle cell neoplasm. Mitoses are almost not seen

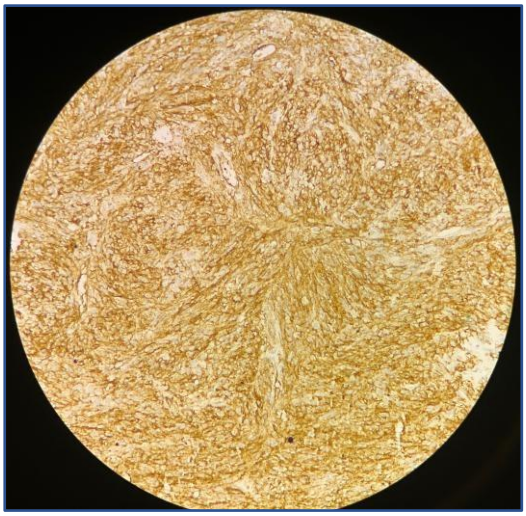


Fig-9 Immunohistochemistry Report Consistent with gastro-intestinal stromal tumor (GIST) - cKit positive

Discussion and management

Staging of GISTs

GISTs are classified into stages I through IV to guide treatment decisions. Staging is based on tumor size, location, and mitotic index per high-power field (HPF):

- **Stage I**: Tumor size between 2–5 cm, with a low mitotic rate (<5 mitoses per 50 HPF).
- **Stage II**: Tumor size between 5–10 cm, with a low mitotic rate (<5 mitoses per 50 HPF).
- **Stage III**: Tumor size >10 cm or with a high mitotic rate (>5 mitoses per 50 HPF).
- **Stage IV**: Tumor has spread to the peritoneal cavity, liver, or lungs, with a high mitotic rate. [1,2,3]

Management of GISTs

The management of GISTs incorporates four standard approaches:

1. **Surgery**
2. **Targeted Therapy**
3. **Watchful Waiting**
4. **Supportive Care**

The choice of treatment depends on factors such as tumor size, mitotic rate, location, and whether the tumor has ruptured or perforated. [10,11]

Surgical Management

Open Surgery

Localized, resectable tumors (2–5 cm in size) with low mitotic rates, unresectable tumors that have responded to targeted therapy and palliative or debulking surgery to control complications like bleeding.

The primary goal is complete resection of the tumor with an intact pseudocapsule and negative surgical margins. Lymph node removal is generally unnecessary, as GISTs rarely metastasize to lymph nodes. The surgeon must handle the tumor carefully to avoid disrupting the pseudocapsule, which could cause bleeding or tumor spread. [1,2,9]

Laparoscopic Surgery

Minimally invasive laparoscopy offers several advantages, including shorter hospital stays, less bleeding, and reduced postoperative pain. Oncological principles of laparoscopic GIST surgery include:

Complete resection with negative margins, preservation of the pseudocapsule to prevent tumor spillage and removal of the tumor in a specimen bag to avoid dissemination.

Segmental bowel resection is often sufficient for jejunal, small bowel, or colonic GISTs, with anastomosis performed extracorporeally or intracorporeally. Laparoscopy is comparable to open surgery in achieving safe and adequate tumor resection. [2,3,6]

Targeted Therapy

Targeted therapy is often combined with surgery and may be administered before or after the procedure:

- **Adjuvant Therapy**: Used post-surgery to reduce recurrence risk. Recommended for high-risk patients, typically for at least three years. Imatinib: First-line drug for KIT or PDGFRA mutation-positive tumors. Sunitinib: Second-line drug when resistance to imatinib develops.
- **Neoadjuvant Therapy**: Administered preoperatively to shrink tumors, potentially making previously inoperable tumors resectable.
- **Radiofrequency Ablation**: An alternative for patients who are poor surgical candidates due to advanced age or other health risks. [1,2,4]

Watchful Waiting

In select cases, where the tumor is asymptomatic or grows very slowly, watchful waiting may be employed if tumor less than 1 cm. This involves close monitoring without immediate treatment. [1,3]

Supportive Care

Supportive care focuses on symptom management and treating side effects. It is often used for advanced cases where the tumor progresses despite treatment. Pain Management radiotherapy may be used for pain relief in patients with large, metastatic tumors. [8,,9]

Prognosis

Localized GISTs five-year relative survival rate is approximately 93% and Metastatic GISTs five-year survival rate drops to **55%**. [11]

Conclusion

jejunal GISTs and Small bowel are rare and obscure causes of lower gastrointestinal bleeding and anemia. Computed tomography (CT) serves as the first-line imaging modality for diagnosis. Capsule endoscopy and balloon-assisted enteroscopy are valuable diagnostic tools for identifying the source of obscure GI bleeding. Surgical resection remains the primary treatment for small bowel GISTs, aiming for complete tumor removal with negative margins.

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