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CASE REPORT | SMALL BOWEL

Unmasking Gastrointestinal Stromal Tumor: Gastrointestinal Stromal Tumor Presenting With Recurrent Gastrointestinal Bleeding

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ABSTRACT

A jejunal gastrointestinal stromal tumor (GIST) is a rare malignant mesenchymal neoplasm of the gastrointestinal tract that arises from the interstitial cells of Cajal. Massive and recurrent bleeding due to jejunal GIST is a diagnostic challenge which may lead to delays in treatment. GISTs as the source of bleeding are already rare. This case is further characterized by endoscopic visualization and clipping of a site of bleeding before eventual surgical resection for recurrent bleeding.

KEYWORDS: gastrointestinal stromal tumor; gastrointestinal bleeding; gastrointestinal tumors; subepithelial lesions; small intestinal masses; neoplasm

INTRODUCTION

A gastrointestinal stroma tumor (GIST) is a rare mesenchymal neoplasm of the gastrointestinal tract originating from the interstitial cells of Cajal or their precursors.[1,2] These cells act as a pacemaker for peristaltic motor activity within the wall of the intestine. These tumors may originate from anywhere in the GI tract with the most common locations including the stomach and small intestine and less common locations including the colon, rectum, or esophagus <5%.[3] GISTs reportedly account for between 0.1% and 3% of all gastrointestinal malignancies, with 1 study reporting an incidence rate of 1- 2:100,000 persons per year.[3] However, most GISTs present asymptomatically and are diagnosed incidentally in 18% of cases on imaging or surgically, so the true prevalence is

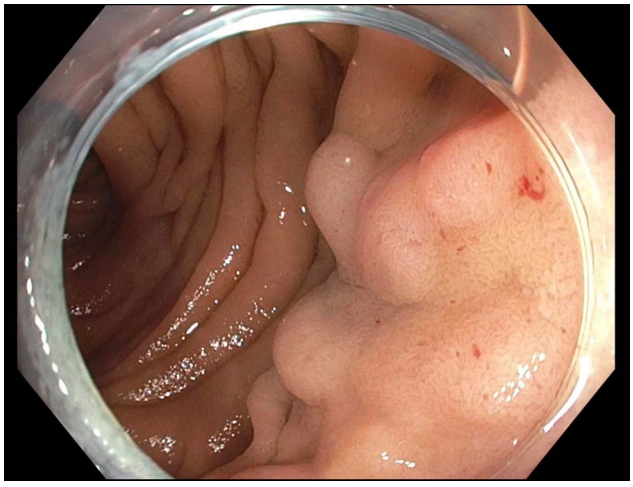

Figure 1. An endoscopic image of the subepithelial lesion in the jejunum.

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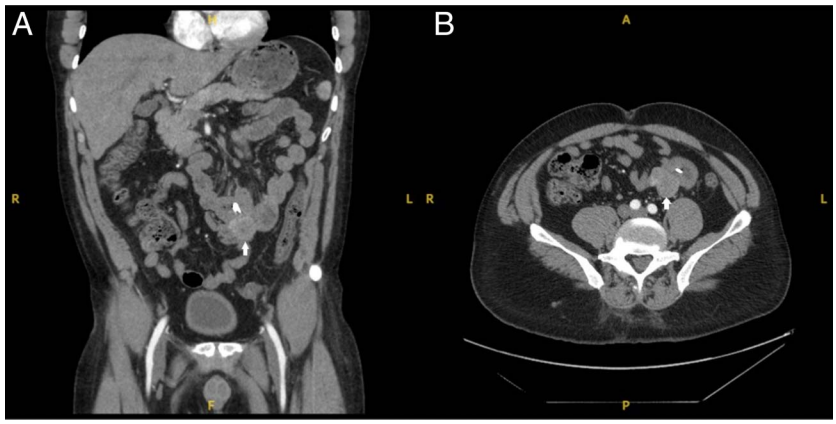

Figure 2. A computed tomography image of the abdomen and pelvis with angiography showing multiple surgical clips near the small bowel loops with an adjacent soft tissue mass measuring 3.2 cm. Figure A is a coronal view and B is a transverse view with white arrows pointing towards the mass.

unknown.[4] In addition, GISTs were initially classified as a type of smooth muscle neoplasm (ie, a leiomyoma, leiomyosarcoma, or leiomyoblastoma) until 1983 when studies found it was a distinct entity characterized by immunohistochemical c-KIT positivity and pathologic features of spindle cells, epithelioid cells, or a mixture.[5] Clinical presentation of GISTs varies depending on the localization of disease with the spectrum of symptoms including gastrointestinal bleeding, hemoperitoneum, anemia, abdominal mass, or absence of symptoms.[3] GISTs can spread outwardly toward the lining of the intestinal wall or inwardly into the intestinal lumen.[3] GISTs are classified by risk potential, with risk stratification calculated based on tumor size, mitotic rate, location, and perforation.[4] Gastric GISTs behaved less aggressively than small bowel GISTs of similar size and mitotic activity.[6]

CASE REPORT

A 47-year-old African American man presented with 5 months of recurrent melena episodes. During a previous hospitalization, he had an esophagogastroduodenoscopy, colonoscopy, and pill camera endoscopy, which were negative for acute gastrointestinal bleeding. His medical history is significant for *Helicobacter pylori* gastritis. Remaining comorbid conditions include iron deficiency anemia and alcohol dependence. On initial physical examination, vital signs are stable and rectal examination shows melena. Laboratory testing showed a positive fecal occult blood test and anemia with hemoglobin 8.9 g/dL (reference range 11.1–15.9 g/dL).

Initial esophagogastroduodenoscopy with push enteroscopy and colonoscopy were negative for active bleeding. Small capsule endoscopy revealed an actively bleeding lesion in the mid-to-distal part of the duodenum. Repeat push enteroscopy found a subepithelial lesion in the jejunum which was resected and clipped (Figure 1). A follow-up computed tomography (CT) scan with intravenous contrast was negative for active bleeding 3 days postendoscopy though it noted a soft tissue mass measuring 3.2 cm in the small bowel adjacent to the endoscopy clips (Figure 2).

The patient had recurrent symptomatic rectal bleeding 5 days after jejunal polyp resection. The hemoglobin acutely decreased to 5.8 g/dL from 7.1 g/dL (reference range 11.1–15.9 g/dL), so he was transfused 1 unit packed red blood cells. Surgery was consulted and performed a laparoscopic small bowel resection with removal of 2 jejunal masses (Figure 3). Only one of these masses was seen on the initial CT report. The jejunal masses

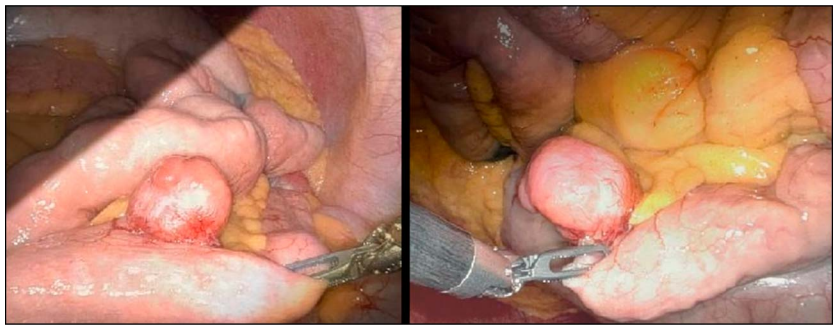

Figure 3. An intraoperative image from the small bowel resection surgery showing 2 jejunal masses each approximately 3 cm in diameter. One mass is about 10 cm distal to the ligament of Treitz and the other about 40 cm distal from the first mass. The more proximal mass appears to contain endoscopic clips.

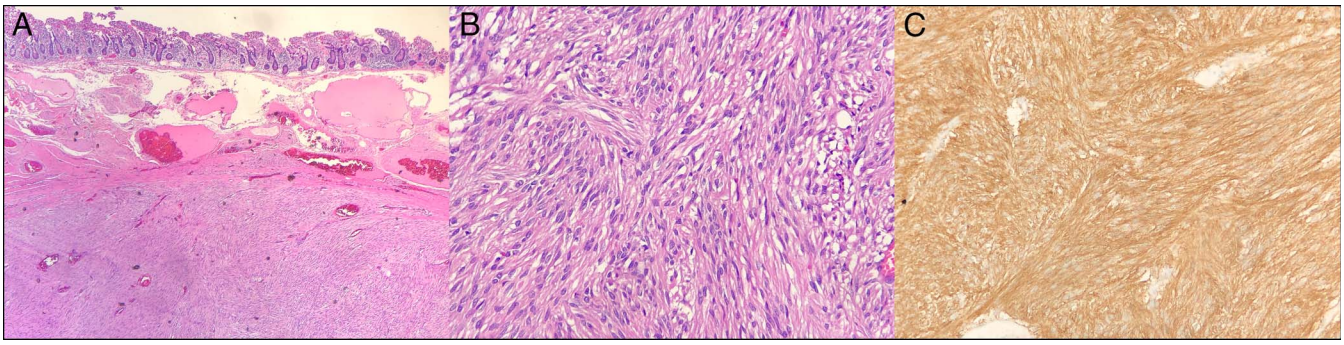

Figure 4. The pathology images under H&E staining at low power (40×) (A) and high power (100×) (B), and immunohistochemical staining with DOG1 (C). The H&E stains show spindle cell lesions arising in the muscularis propria deep to mucosa and submucosa. The DOG1 staining is positive in image (C). DOG1, discovered on GIST1; GIST, gastrointestinal stromal tumor; H&E, hematoxylin and eosin.

were approximately 3 cm in diameter and both distal to the ligament of the Treitz (10 and 50 cm distal, respectively). One of the masses was tagged by endoscopic clips. Immunohistochemical staining of the small bowel masses was positive for CD 117 and discovered on GIST1 markers, consistent with a diagnosis of low-risk spindle cell-type GIST (Figure 4). Pathology staging was mpT2 pN0 pMn/a, and margins were negative.

At 1-month postoperative follow-up, the patient is doing well from his surgery and anemia. His hemoglobin improved to 11.1 g/dL. The patient had a follow-up positron emission tomography and CT for tumor staging, which was negative for abnormal uptake and without evidence of residual disease. The patient established with an oncologist and continues to follow gastroenterology.

DISCUSSION

The events of this case occurred over a span of 5 months. The endoscopic workup on his initial presentation did not localize the bleeding. This is frequently the case in small intestinal GI bleeds which are found in 5%–10% of cases.[7] In addition, the presenting symptoms for GISTs vary depending on the location of the tumor and progression of the disease. If a source of bleeding is not found on endoscopy, the next recommended step is for CT imaging.

During the second hospital stay, a source of bleeding was found, clipped, and biopsied. Despite these measures, the patient had a recurrence of gastrointestinal bleeding. Imaging revealed a mass in the small bowel near the site of the endoscopic clip. For traditional GI bleeding, the options for management of recurrent bleeding include medical management, arterial embolization, repeat endoscopic clipping, or surgery. In this case, surgical resection is the gold standard for localized GIST.[1]

The resected tissue was examined to determine appropriate staging. GISTs are risk stratified based on tumor size, mitotic rate, location, and perforation.[4] The tumor size has been associated with a 5-year survival of 20% in tumors >10 cm and 60% in tumors less than 5 cm.[5] Primary gastric tumors behave less aggressively than small bowel and rectal primary tumors.[5]

In this case, the tumor was consistent with a low-risk spindle cell type on pathology. Low-risk GISTs are reported to recur up to 20 years after surgical resection. In more severe cases of GIST, imatinib is added as adjuvant treatment.[8] The postoperative positron emission tomography and CT was negative for residual or recurrent disease, so imatinib was not initiated for this patient. Yet, previous case studies have proposed the presence of GI bleeding in GISTs should be treated as a type of tumor perforation, thereby leading to a higher risk assessment.[9]

Key points:

- Providers should consider GISTs as a source of obscure GI bleeding.
- When investigating recurrent GI bleeding, consider CT imaging if a source is not visualized on endoscopy.

DISCLOSURES

Author contributions: Co-first authors T. Khan and S. Afaq were involved in the initial draft, writing, and editing of the case report. Coauthors N. Mishal and CL Miao were involved in the writing and editing process for the paper. C.L. Miao is the article guarantor.

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Previous presentation: This case was presented at the American College of Gastroenterology Meeting 2024 as a poster presentation titled "Gastrointestinal Stromal Tumors Presenting as Recurrent Gastrointestinal Bleeding" on Sunday October 27, 2024, at the Pennsylvania Convention Center in Philadelphia, PA.

Informed consent was obtained for this case report.

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