

Jejunal Gastrointestinal Stromal Tumor: Case Report and Literature Review

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ABSTRACT

Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal neoplasms of the gastrointestinal tract and originate from the interstitial cells of Cajal. Although most of these tumors are located in the stomach, a smaller proportion develop in the small intestine, with the jejunum being a relatively uncommon site. The clinical presentation of GISTs is variable and often nonspecific, which may hinder early diagnosis. The most frequent symptoms include abdominal pain, anemia, gastrointestinal bleeding, or the presence of an abdominal mass; however, in some cases, the tumor may be detected incidentally during imaging studies performed for other reasons. We present the case of an adult patient with a gastrointestinal stromal tumor located in the jejunum who presented with progressive abdominal pain and weight loss. Imaging studies revealed a tumor lesion arising from the small intestine, and segmental surgical resection with adequate margins was performed. Histopathological and immunohistochemical analysis confirmed the diagnosis of gastrointestinal stromal tumor, with positivity for CD117 and DOG1. The postoperative course was favorable, and the patient remained under clinical follow-up without evidence of tumor recurrence. This case highlights the importance of considering GISTs in the differential diagnosis of small bowel tumors, as well as the fundamental role of histopathological and immunohistochemical studies in confirming the diagnosis. Surgical treatment remains the main therapeutic option for localized tumors, while targeted therapies have significantly improved the prognosis of patients with advanced disease.

KEYWORDS: Gastrointestinal stromal tumor; GIST; jejunum; small intestine; gastrointestinal mesenchymal tumor; histopathological diagnosis; CD117; DOG1; surgical resection.

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INTRODUCTION

Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal neoplasms of the gastrointestinal tract. They originate from the interstitial cells of Cajal or their precursor cells, which are involved in the regulation of intestinal motility. These tumors are characterized by the expression of the tyrosine kinase receptor KIT (CD117) and, to a lesser extent, the marker DOG1, findings that are essential for their immunohistochemical diagnosis.

GISTs can arise in any segment of the gastrointestinal tract; however, their anatomical distribution is variable. Approximately 60–70% occur in the stomach, whereas 20–30% originate in the small intestine, with the jejunum being a less frequent location. Other locations include the colon,

rectum, and, exceptionally, the esophagus or extragastrointestinal sites.

The clinical presentation of GISTs is heterogeneous and depends mainly on tumor size, location, and growth pattern. In the early stages, they may be asymptomatic and detected incidentally during imaging studies or surgical procedures performed for other causes. When clinical manifestations are present, they may include abdominal pain, palpable mass, gastrointestinal bleeding, anemia, or obstructive symptoms. In the case of GISTs located in the jejunum, diagnosis may be particularly challenging due to the nonspecific nature of symptoms and the difficulty in detecting them by conventional endoscopic methods.

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The treatment of choice for localized GISTs is complete surgical resection with negative margins, avoiding intraoperative tumor rupture, as this is associated with a greater risk of recurrence. In recent years, the development of targeted therapies, such as tyrosine kinase inhibitors (for example, imatinib), has significantly improved the prognosis of patients with advanced or metastatic disease.

In this context, the presentation of clinical cases is relevant to expand knowledge regarding the different forms of presentation, diagnosis, and management of GISTs, especially in less frequent locations such as the jejunum. In the present article, we describe the case of a gastrointestinal stromal tumor located in the jejunum, highlighting its clinical characteristics, diagnostic findings, and therapeutic approach, accompanied by a review of the literature.

CASE REPORT

We present the case of a 58-year-old male patient with no relevant past medical history who presented to the emergency department with a clinical picture of approximately three months' duration characterized by intermittent abdominal pain localized to the mesogastrium, accompanied by a sensation of abdominal fullness, asthenia, and unintentional weight loss of approximately 6 kg during that period. In the two weeks prior to admission, the patient reported occasional episodes of nausea and progressive worsening of abdominal pain intensity.

His non-pathological personal history was notable for occasional smoking since youth and social alcohol consumption. He denied previous abdominal surgery, chronic degenerative diseases, or a family history of gastrointestinal neoplasms.

On physical examination, the patient was conscious, oriented, and hemodynamically stable. The abdomen was soft and depressible, with mild pain on deep palpation in the periumbilical region, without signs of peritoneal irritation. No palpable masses or visceromegaly were identified. Bowel sounds were present and without evident abnormalities.

Initial laboratory studies showed a hemoglobin level of 10.8 g/dL, with findings consistent with mild normocytic normochromic anemia. The remaining biochemical parameters were within normal limits.

Due to the persistence of symptoms, contrast-enhanced abdominal computed tomography was requested, which revealed a well-defined solid lesion arising from small bowel loops, located in the proximal jejunum, measuring approximately 5.2 × 4.8 cm, with heterogeneous enhancement after contrast administration and areas suggestive of central necrosis. No regional lymphadenopathy or liver lesions suggestive of metastases were observed.

Based on the clinical and imaging findings, a diagnostic suspicion of a small bowel tumor was established, and surgical management was decided upon. Exploratory laparotomy was performed, revealing an exophytic tumor arising from the jejunal wall approximately 40 cm from the ligament of Treitz, measuring about 5 cm in diameter, well circumscribed, without evidence of infiltration into adjacent structures or peritoneal implants. No significant mesenteric lymphadenopathy was identified.

Segmental resection of the jejunum including the tumor was performed with adequate safety margins, followed by a two-layer hand-sewn end-to-end anastomosis. The surgical procedure was completed without complications.

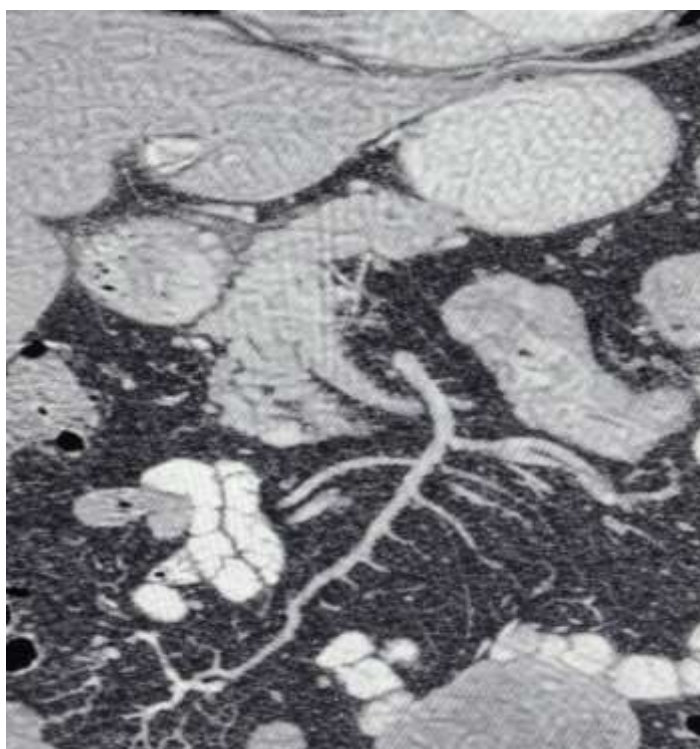


Figure 1. Computed tomography showing a tumor located at the jejunal level



Figure 2. Jejunal tumor.

Gross histopathological examination described a well-circumscribed tumor lesion measuring 5.1 cm in greatest diameter, with a solid appearance and focal areas of hemorrhage and necrosis. Microscopically, there was proliferation of spindle-shaped cells arranged in intersecting fascicles, with eosinophilic cytoplasm and elongated nuclei. A mitotic index of 3 mitoses per 50 high-power fields was identified.

Immunohistochemical analysis showed positivity for CD117 (KIT) and DOG1, with negativity for desmin and S-100, findings consistent with a gastrointestinal stromal tumor (GIST).

Based on tumor size and mitotic index, the tumor was classified as an intermediate-risk GIST according to prognostic classification criteria.

The patient had a favorable postoperative course, initiating oral intake on the third postoperative day and being discharged on the fifth day without complications. During clinical follow-up six months after surgery, the patient

remained asymptomatic, with no evidence of tumor recurrence or metastatic disease on follow-up imaging studies.

This case highlights the importance of considering gastrointestinal stromal tumors in the differential diagnosis of small bowel masses, particularly in patients with nonspecific abdominal symptoms, as well as the relevance of histopathological and immunohistochemical diagnosis for the definitive confirmation of this entity.

DISCUSSION

Gastrointestinal stromal tumors (GISTs) represent the most common mesenchymal neoplasms of the gastrointestinal tract. They originate from the interstitial cells of Cajal or related precursor cells, which function as pacemakers of intestinal motility. From a molecular standpoint, most GISTs harbor activating mutations in the KIT or PDGFRA genes, which lead to constitutive activation of the tyrosine kinase signaling pathway responsible for tumor growth.

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The estimated annual incidence of GISTs ranges from 10 to 15 cases per million inhabitants, being more frequent in adults older than 50 years. Although they may be located in any segment of the gastrointestinal tract, most develop in the stomach, whereas the small intestine represents the second most common site of occurrence. Within the small intestine, the jejunum is a less frequent location, but it is usually associated with more aggressive biological behavior compared with gastric tumors.

The clinical presentation of GISTs is variable and depends on tumor size, location, and growth pattern. In many cases, tumors remain asymptomatic during the early stages and are

detected incidentally. When clinical manifestations are present, they may include abdominal pain, gastrointestinal bleeding, anemia, palpable abdominal mass, or obstructive symptoms. Small bowel GISTs may also present with gastrointestinal hemorrhage or intestinal perforation.

The definitive diagnosis is established by histopathological and immunohistochemical study. Histologically, GISTs may show three main patterns: spindle cell, epithelioid, or mixed. Immunohistochemistry plays a fundamental role in diagnosis, with positivity for CD117 (KIT) and DOG1 being characteristic and highly sensitive markers for this entity.

TABLE 1. EPIDEMIOLOGY OF GASTROINTESTINAL STROMAL TUMORS

Characteristic	Epidemiological data
Annual incidence	10–15 cases per million inhabitants
Mean age at diagnosis	55–65 years
Sex predominance	Slight male predominance
Most frequent location	Stomach (60–70%)
Second most frequent location	Small intestine (20–30%)
Jejunal location	Approximately 10% of GISTs

TABLE 2. ANATOMICAL DISTRIBUTION OF GISTS

Location	Approximate frequency
Stomach	60–70%
Small intestine	20–30%
Colon and rectum	5%
Esophagus	<1%
Extragastrintestinal locations	<5%

TABLE 3. HISTOLOGICAL CLASSIFICATION OF GISTS

Histological type	Microscopic characteristics
Spindle cell	Elongated cells arranged in fascicles
Epithelioid	Rounded cells with abundant cytoplasm
Mixed	Combination of spindle and epithelioid patterns

TABLE 4. FLETCHER (NIH) PROGNOSTIC CLASSIFICATION

Tumor size	Mitotic index	Risk
<2 cm	<5 mitoses / 50 HPF	Very low
2–5 cm	<5 mitoses / 50 HPF	Low
<5 cm	6–10 mitoses / 50 HPF	Intermediate
5–10 cm	<5 mitoses / 50 HPF	Intermediate
>5 cm	>5 mitoses / 50 HPF	High
>10 cm	Any mitotic index	High

TABLE 5. MODIFIED MIETTINEN PROGNOSTIC CLASSIFICATION

Location	Tumor size	Mitotic index	Risk of progression
Stomach	<5 cm	<5 mitoses	Very low
Stomach	5–10 cm	<5 mitoses	Low
Small intestine	<5 cm	<5 mitoses	Low
Small intestine	>5 cm	<5 mitoses	Intermediate
Small intestine	>5 cm	>5 mitoses	High

From the therapeutic standpoint, complete surgical resection with negative margins is the treatment of choice for localized GISTs. Unlike other gastrointestinal tumors, systematic lymphadenectomy is generally not necessary, as lymphatic spread is uncommon.

In recent years, the development of targeted therapies, particularly tyrosine kinase inhibitors such as imatinib, has transformed the management of advanced or metastatic GISTs. These agents act by blocking the activity of KIT and PDGFRA, thereby controlling tumor growth and significantly improving patient survival.

The present case illustrates the importance of considering GISTs in the differential diagnosis of small bowel tumors, especially in patients with nonspecific abdominal symptoms or anemia of unknown origin. Early diagnosis, together with appropriate surgical management and proper prognostic stratification, is essential to optimize prognosis and reduce the risk of tumor recurrence.

CONCLUSION

Gastrointestinal stromal tumors (GISTs) represent the most common mesenchymal neoplasms of the gastrointestinal tract, although their location in the small intestine, particularly in the jejunum, is relatively uncommon. Their clinical presentation may be variable and nonspecific, which can delay diagnosis and hinder identification at early stages. In many cases, patients present with vague symptoms such as abdominal pain, anemia, or gastrointestinal bleeding, whereas in others the tumor may be detected incidentally during imaging studies or surgical procedures performed for other reasons.

The definitive diagnosis of GISTs is established by histopathological analysis and immunohistochemical confirmation, with positivity for markers such as CD117 (KIT) and DOG1 being characteristic. Likewise, the assessment of tumor size, mitotic index, and anatomical location allows prognostic stratification of the tumor through widely used classifications such as those proposed by Fletcher and Miettinen, which are essential for estimating the risk of recurrence and guiding clinical follow-up.

The treatment of choice for localized GISTs consists of complete surgical resection with negative margins, avoiding intraoperative tumor rupture, since this is associated with a higher risk of dissemination and recurrence. Unlike other gastrointestinal neoplasms, systematic lymphadenectomy is usually not necessary due to the low incidence of nodal metastases in these tumors. In selected cases with a high risk of recurrence or advanced disease, the use of targeted therapy with tyrosine kinase inhibitors such as imatinib has been shown to significantly improve survival and disease control. In conclusion, jejunal GISTs constitute an uncommon entity that may represent a diagnostic challenge because of their nonspecific clinical presentation. The combination of imaging studies, histopathological evaluation, and

immunohistochemistry is essential to establish an accurate diagnosis. Timely surgical resection, together with appropriate prognostic stratification and clinical follow-up, is essential to optimize patient management and improve long-term outcomes. Finally, the presentation of clinical cases contributes to expanding knowledge of this entity and strengthening early recognition of its clinical manifestations in medical practice.

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