Gastrointestinal stromal tumor (GIST). Clinical surgical case presentation.

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Abstract

Introduction: Gastrointestinal stromal tumors (GISTs) are rare tumors. It originates in the interstitial cells of Cajal, and a mutation in the kit gene (growth factor receptor tyrosine kinase) appears to be the leading cause of tumor growth. The clinical manifestations depend on the location of the primary tumor. There was no difference between the sexes; the average reported age was approximately 60–70. A high proportion of GISTs are asymptomatic and are often discovered incidentally during an endoscopic study or on radiological images obtained for another purpose.

Clinical case: This is a 68-year-old man with a history of type 2 diabetes mellitus, ischemic heart disease, and high blood pressure. An ultrasound examination revealed a solid mass measuring approximately 10 × 9 cm at the head of the pancreas. The patient was asymptomatic.

Diagnostic workshop: The complete blood count, blood chemistry, liver, and pancreas function tests were normal. Abdominal CT revealed a highly vascular soft tissue mass measuring 11 × 10 × 10 cm, located in the posterior wall of the stomach, extending into the omental cavity and in contact with the head of the pancreas.

Treatment: The vascularized tumor involving the lesser curvature of the stomach at the level of the gastric antrum was removed via exploratory laparotomy. Pathology revealed a spindle cell and epithelioid mesenchymal neoplasm, which was consistent with an intermediate-risk gastrointestinal stromal tumor (GIST). Immunohistochemistry revealed CD117, CD34, SMA, S-100 protein, and Ki-67.

Evolution: A PET scan contrasted with 18F-FDG was requested, which was normal. The evaluation was favorable, with no tumor activity at the presentation time.

Conclusions: GISTs are potentially malignant tumors, and gastric tumors are usually positive for CD117 and DOG-1. Surgical resection is the standard of care.

Keywords:

Gastrointestinal stromal tumor, GIST, laparotomy, case report.

Abbreviations

DHL: Lactic dehydrogenase.
CT: computed axial tomography.

Additional information

No supplementary materials are declared.

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Authors' contributions

Jorge Jiménez Barahona: Conceptualization, research, writing—original draft, resources, software, supervision.

Pablo Jiménez Benavides: Conceptualization, research, writing—original draft, resources, software, supervision.

María Del Carmen Palacios del Campo: Methodology, Data curation, Formal analysis, Funding acquisition, Project management, Validation, Visualization, Writing – review and editing.
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Availability of data and materials

The datasets used and analyzed during the present study are available from the corresponding author upon reasonable request.

Introduction

Gastrointestinal stromal tumors (also known as GISTs) are rare diseases that originate from nerve cells of the digestive tract and are responsible for activating the contractions that propel food contents from the mouth to the anus [1].

Gastrointestinal stromal tumors or GISTs represent only 1–3% of all malignant neoplasms of the stomach and 15–20% of those of the small intestine. Its incidence is 0.72- 0.85 cases per 100,000 inhabitants. Most studies have reported an increase in incidence since 2000; however, this may result from improvements in diagnostic criteria rather than an actual increase in incidence. They originate in the interstitial cells of Cajal, whose mutation in the kit gene (receptor tyrosine kinase growth factor) appears to be the leading cause of the growth of these tumors [2- 5].

The most common locations are the stomach (50–60%) and the small intestine (30–35%). It rarely appears in the esophagus, colon, rectum, or abdominal cavity.

GISTs are usually associated with mutations in the KIT or PDGFRA genes. These mutations abnormally activate cell signaling pathways that promote tumor growth. Approximately 85% of GISTs have mutations in the KIT gene, whereas 5–10% have alterations in PDGFRA [3].

The clinical manifestations depend on the location of the primary tumor. There was no difference between the sexes; the reported average age was approximately 60–70.

These tumors are typically associated with nonspecific symptoms, such as early satiety and bloating, unless they ulcerate, bleed, or grow large enough to cause pain, obstruction, or other manifestations related to their disproportionate size. In the case of esophageal GIST, dysphagia is the first specific symptom in this location [5].

A high proportion of GISTs are asymptomatic and are often discovered incidentally during an endoscopic examination or on radiological images obtained for another purpose. This incidental finding can lead to a significant diagnostic delay. Many patients present with metastases at diagnosis (up to 50% in some series). Physicians must include GISTs in the differential diagnosis because of the importance of early diagnosis in these patients [6].

Diagnosing a gastric GIST usually involves imaging studies such as ultrasound and contrast-enhanced computed tomography. Endoscopy may reveal a submucosal mass, but a biopsy is often necessary for confirmation. Simple endoscopy cannot accurately distinguish between intramural and extramural tumors. In this sense, endoscopic ultrasonography has proven to be a valuable technique for characterizing these masses by identifying the layer of origin and allowing tissue acquisition through guided puncture for diagnostic pathological studies, which is suitable for immunohistochemical testing [7, 8].

Biopsies for pathological examination and immunohistochemistry are key to definitive diagnosis, with most GISTs being positive for CD117 (KIT) and DOG1. However, preoperative biopsy is not recommended for a resectable lesion if there is high clinical and radiological suspicion of a GIST and if the lesion is completely resectable. A preoperative biopsy is preferred to confirm the diagnosis if metastatic disease is suspected, if neoadjuvant treatment with imatinib is being considered, or in cases where there is high operative morbidity or the diagnosis is unclear.

Surgery is the only potentially curative treatment for suspected resectable GIST. The primary goal of this procedure is to ensure clean resection margins during complete tumor resection and that the tumor can be removed without rupturing the tumor pseudocapsule; however, wide margins have no benefit for disease control. Lymphadenectomy is not necessary because lymphatic involvement is rare. Intraoperative liver and parietal peritoneum examination is essential for identifying possible metastases [6, 9].

Tyrosine kinase inhibitor therapy: Imatinib is the standard treatment for metastatic or unresectable GISTs and is also used as adjuvant therapy in certain high-risk patients.

The prognosis for patients with gastric GISTs depends on the tumor size, mitotic index, and presence of metastases. Small tumors with a low mitotic index tend to have a good prognosis, whereas larger and more aggressive tumors may have a greater risk of recurrence or metastatic spread.

Clinical case

Medical record

A 68-year-old male patient with a history of type 2 diabetes mellitus who was receiving oral hypoglycemic agents was diagnosed with ischemic heart disease and high blood pressure. During a cardiovascular checkup, a routine abdominal ultrasound was requested, and a solid mass measuring approximately 10 × 9 cm was found in the center of the abdomen at the level of the head of the pancreas. The patient was asymptomatic and referred to the oncologic surgery service.

Diagnostic workshop

Evaluation tests were requested. The complete blood count, blood chemistry, and liver and pancreas function test results were within normal ranges. Her LDH was normal. An abdominal and pelvic CT revealed a highly vascularized soft tissue mass measuring 11 × 10 × 10 cm in diameter, located at the posterior wall of the stomach and projecting toward the antrum. This mass extends into the omental bursa and is in contact with the head of the pancreas and the left lobe of the liver (Figure 1). Based on these findings, an endoscopic ultrasound was requested, which revealed a perigastric tumor lesion that preserved cleavage planes with neighboring structures (Figure 2).

Figure 1. Simple and contrast-enhanced abdominal tomography.

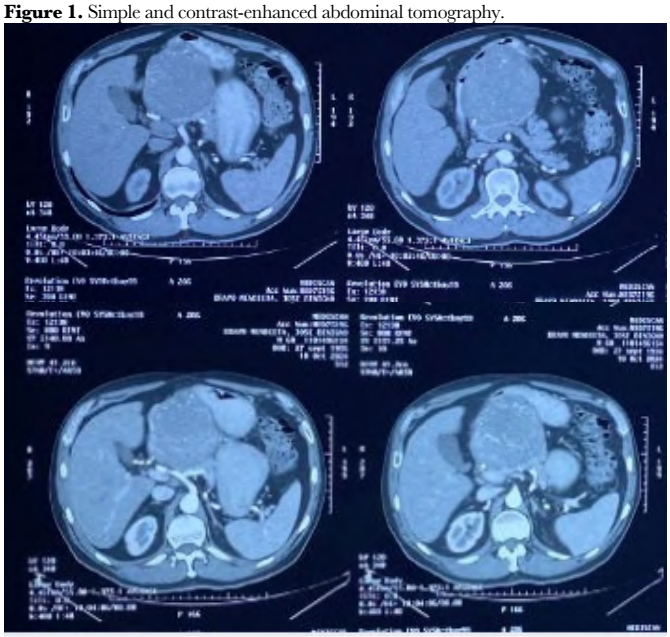
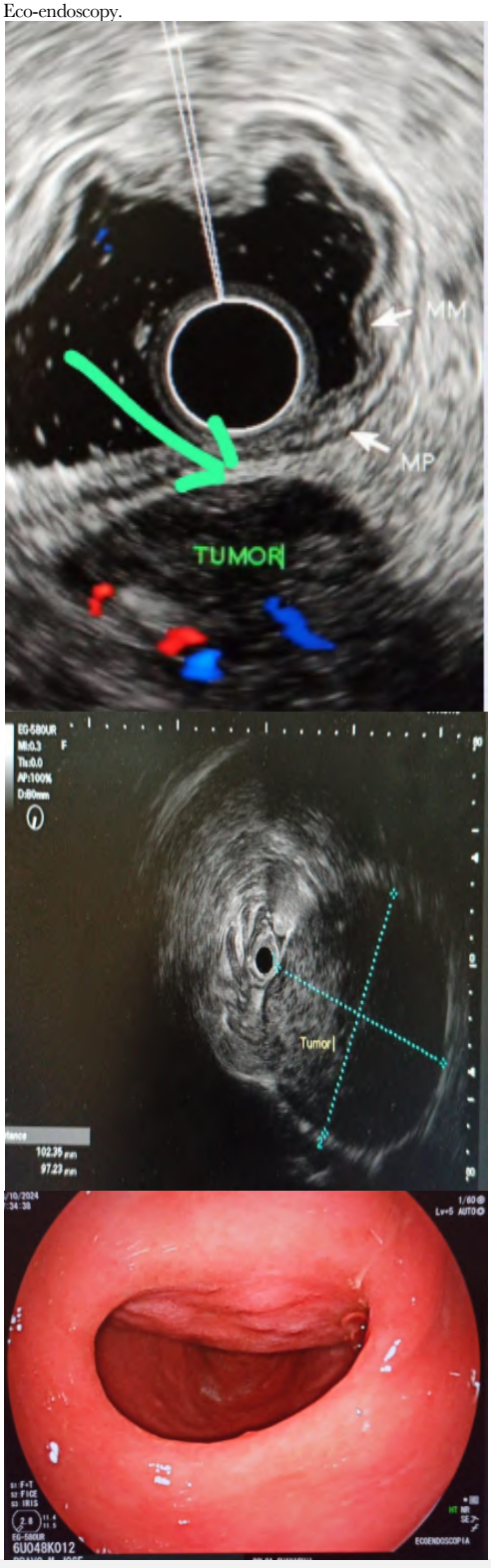


Figure 2. Eco-endoscopy.

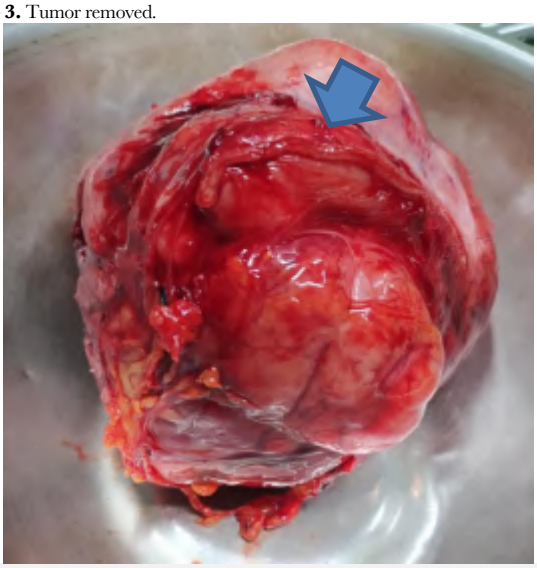


Treatment

An exploratory laparotomy was planned to assess tumor extension and the possibility of removal. Preoperative examinations and evaluations by cardiology and pulmonology services were requested. The patient was informed of the risks and benefits of surgery through informed consent. Exploratory laparotomy was performed under general anesthesia; the findings indicated the presence of a solid tumor approximately 15 cm in diameter, a vascularized tumor involving the lesser curvature of the stomach at the level of the gastric antrum, in contact with the left lobe of the liver and the head of the pancreas. The tumor was removed, including the margin of the gastric wall at the level of the lesser curvature (Figure 3). The surgical procedure was reported to be uneventful. The postoperative period was favorable.

The final pathology report indicated that the tumor was a spindle cell and epithelioid mesenchymal neoplasm, which is consistent with an intermediate-risk gastrointestinal stromal tumor (GIST) (Figure 4). Immunohistochemistry revealed positive CD117, CD34, SMA, S-100 protein, and Ki-67 expression, confirming the diagnosis of a GIST.

Figure 3. Tumor removed.



Tumor removed. The arrow shows the area of the gastric wall.

Evolution

A PET scan study was requested for disease staging, revealing no hypermetabolic macroscopic lesions indicative of tumor or metastatic disease that took up the 18F-FDG contrast agent. The patient's evaluation was favorable, showing no tumor activity at presentation. He remains under follow-up and surveillance.

Discussion

Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal tumors specific to the gastrointestinal tract and are usually defined as KIT (CD117)-positive tumors with a characteristic set of histological features. These tumors, derived from Cajal cells or their precursors, occur most commonly in patients over 50 years of age in terms of frequency: in the stomach (60%), jejunum and ileum (30%), duodenum (4–5%), rectum (4%), colon and appendix (1–2%) and esophagus (<1%), and rarely as primary extragastrointestinal tumors [10].

GISTs are potentially malignant tumors and tend to behave aggressively. GISTs affect men (55%) more than women, with an average age of 55-60 years [11].

These tumors are generally associated with nonspecific symptoms (early satiety, abdominal distension) unless they ulcerate, bleed, or grow large enough to cause pain, obstruction, or other manifestations related to their disproportionate size [5].

Many patients with gastric GIST are asymptomatic and are often discovered incidentally during an endoscopic study or in radiological images obtained for another purpose, as in the present case.

Computed tomography with oral and intravenous contrast is the standard for characterizing any abdominal mass, evaluating its extent and absence or presence of distant disease, and defining the intestinal margins [7].

Endoscopy may reveal a submucosal mass, but a biopsy is often necessary for confirmation. Simple endoscopy does not distinguish between intramural and extramural tumors. In this context, endoscopic ultrasonography has proven to be a valuable technique for characterizing these masses by identifying the layer of origin and allowing the acquisition of tissue by guided puncture for diagnostic pathological studies, which is suitable for immunohistochemical tests [8]. In the present case, imaging studies and endoscopy were performed to obtain a diagnostic impression of this pathology.

For many years, the mainstay of treatment for GIST has been surgical resection, which involves wide resections with clear surgical margins. Lymph node metastases are rare, making routine removal usually unnecessary. Postoperative chemotherapy with conventional agents and radiation therapy has also proven ineffective. In the present case, segmental gastrectomy of the tumor site, including the gastric wall margin at the lesser curvature, was performed. A definitive study confirmed the diagnosis of a spindle cell and epithelioid mesenchymal neoplasm, consistent with an intermediate-risk gastrointestinal stromal tumor (GIST), with the gastric wall free of lesions. The typical mitotic count does not exceed 5 × 50

HPF. In general, patients with similar parameters have a more favorable prognosis for gastric tumors than for intestinal tumors. Gastric GISTs ≤10 cm and ≤5 mitoses per 50 HPFs have a low risk of metastasis, whereas those >5 per 50 HPFs and >5 cm in diameter present a high risk of metastasis. In contrast, all intestinal GISTs >5 cm, regardless of the mitotic rate, have at least a moderate risk of metastasis, and all those > five mitoses per 50 HPFs have a high risk of metastasis [11].

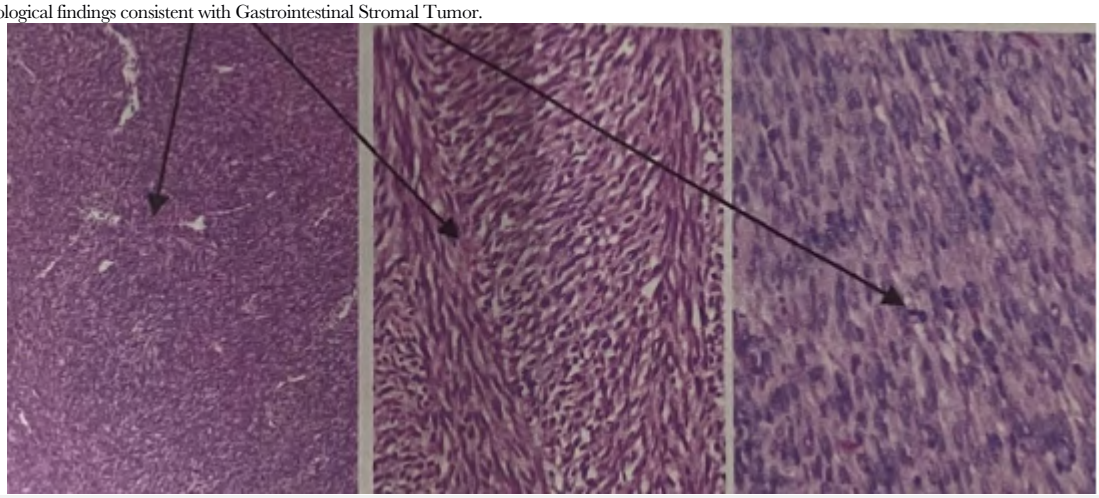
Approximately 60% of patients are cured with surgery. Adjuvant treatment with imatinib is recommended for patients with a substantial risk of recurrence if the tumor has an imatinib-sensitive mutation. Tyrosine kinase inhibitors substantially improve survival in advanced disease, but secondary resistance to these drugs is common [12]. In cases where resistance to imatinib has been confirmed, other TKIs, such as sunitinib or regorafenib, are used as second-line agents [13].

In the present case, owing to the number of mitoses, the patient had a low risk of metastasis, which was corroborated by a PET CT study, where no macroscopic hypermetabolic lesions suggestive of tumors or metastatic disease with affinity for 18F-FDG were evident.

The Clinical Oncology Service was consulted about the possibility of adjuvant treatment, but this was not indicated.

Recent studies in treating GIST have explored novel targeted therapies and biomarkers that may better predict the response to tyrosine kinase inhibitor therapy. Furthermore, the role of immunotherapy and gene therapies in managing GIST [14] and their impact on renal function [15 – 17] are being investigated.

Figure 4. Histological findings consistent with Gastrointestinal Stromal Tumor.



The number of typical mitoses does not exceed 5 x 50 HPF.

Conclusions

GISTs are potentially malignant tumors that are located primarily in the gastrointestinal tract. These rare tumors account for a small percentage of gastrointestinal neoplasms and are typically positive for CD117 and DOG-1. Surgical resection remains the standard of care. Therapy with tyrosine kinase inhibitors (TKIs), such as imatinib, is used for the treatment of metastatic or unresectable GISTs, and it is also used as adjuvant therapy in certain high-risk patients. Monitoring is advised due to the risk of recurrence.

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Statements

Ethics committee approval and consent to participate

This method is not required for clinical cases.

Consent to publication

The authors have written permission from the patient to publish.

Conflicts of interest

The research has no financial interests or conflicts of interest.

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