

GIST in a Middle-Aged Patient: Clinical Case and Review of Diagnostic and Therapeutic Approaches

Ana Itzel Vélez Pérez^{1*}, Eduardo Alberto Leyva Aguilera², María Guadalupe Nieblas Serrano³, Aneliz Alamilla Valenzuela⁴, Ana Denisse Aboytes Bucio⁵, Gustavo Alejandro Paniagua Frausto⁶, Uziel Manjarrez Gutiérrez⁷, Francisco Javier Delgado Dávalos⁸, Ernesto Fernando Montelongo Sedano⁹, Chávez Maldonado Jorge Alberto¹⁰, Escamilla Bastida Marco Antonio¹¹, María Dolores Ferrara Theriot¹², García Morales Mariana¹³

¹Universidad Autónoma del Estado de México.

²Universidad Popular Autónoma del Estado de Puebla.

³Hospital General Zona Numero 1, Ciudad Obregón, Sonora.

⁴Instituto Politécnico Nacional.

⁵Universidad Autónoma de Guadalajara.

⁶Universidad de Monterrey.

⁷Benemérita Universidad Autónoma de Puebla.

⁸Universidad Autónoma de Guadalajara.

⁹Universidad Autónoma de Aguascalientes.

¹⁰Universidad Nacional Autónoma de México

¹¹Universidad Autónoma del Estado de México

¹²Universidad de Monterrey.

¹³Universidad Autónoma del Estado de México.

ABSTRACT

ABSTRACT: Gastrointestinal stromal tumors (GISTs) are rare mesenchymal neoplasms representing 1-2% of gastrointestinal tract tumors. They arise from the interstitial cells of Cajal, which regulate intestinal motility, and are often associated with mutations in the KIT or PDGFRA genes, making them susceptible to targeted therapy with tyrosine kinase inhibitors, such as imatinib. GISTs can occur anywhere in the gastrointestinal tract, with the stomach (60-70%) and small intestine (20-30%) being the most common sites. Clinical presentation is often nonspecific, including abdominal pain, gastrointestinal bleeding, and intestinal obstruction, which can delay diagnosis. The standard treatment for localized GISTs is complete surgical resection without lymphadenectomy due to their low metastatic potential to lymph nodes. However, for advanced or unresectable cases, imatinib therapy significantly improves patient survival. This article presents a case of GIST, highlighting its diagnosis, therapeutic approach, and clinical evolution, along with a review of current literature. The case is characterized by a fibrous mass in the colon, with histopathological confirmation of GIST and high mitotic activity, treated with imatinib as adjuvant therapy. A favorable response was observed at six months post-surgery with no recurrence or metastasis. The article discusses the current strategies for diagnosis and treatment, including the importance of surgical resection, targeted therapy, and close follow-up for early detection of recurrence.

KEYWORDS: Gastrointestinal Stromal Tumors (GIST), Gastrointestinal Tumors, Surgical Treatment, Surgical Resection, KIT and PDGFRA Mutations, Metastatic Disease.

ARTICLE DETAILS

Published On:
30 April 2025

Available on:
<https://ijmscrs.com/>

INTRODUCTION

GIST in a Middle-Aged Patient: Clinical Case and Review of Diagnostic and Therapeutic Approaches

Gastrointestinal stromal tumors (GISTs) are rare mesenchymal neoplasms, accounting for approximately 1-2% of gastrointestinal tract tumors. They arise from the interstitial cells of Cajal, which regulate intestinal motility, and often present mutations in the KIT or PDGFRA genes, making them susceptible to targeted therapies with tyrosine kinase inhibitors such as imatinib.

GISTs can occur anywhere along the gastrointestinal tract, with the stomach (60-70%) and small intestine (20-30%) being the most common sites. Their clinical presentation is variable and nonspecific, including abdominal pain, gastrointestinal bleeding, and intestinal obstruction, which may delay diagnosis.

The standard treatment for localized GISTs is complete surgical resection without lymphadenectomy due to their low metastatic potential to lymph nodes. However, in advanced or unresectable cases, imatinib therapy has significantly improved patient survival.

This article presents a case of GIST, highlighting its diagnosis, therapeutic approach, and clinical evolution, along with a review of the current literature.

CLINICAL CASE

This is a 50-year-old woman, previously healthy, who presents with a clinical picture of vague abdominal symptoms that started approximately one year before her intervention. Symptoms include intermittent abdominal pain, fullness, nausea, and episodes of vomiting. The pain is located in the

upper abdomen and is diffuse in nature, not related to food intake but tends to worsen after meals. The patient has no history of significant gastrointestinal diseases or family history of digestive cancer.

Given the prolonged course and persistent symptoms, an abdominal computed tomography (CT) scan was performed. The imaging report shows a fibrous mass located in the colon, with inconclusive characteristics regarding its origin, prompting the decision to conduct further studies to determine its nature (**Figure 1**). The CT scan also shows signs of colon wall thickening, with no evidence of metastasis in other organs or lymph nodes.

Subsequently, an upper gastrointestinal endoscopy was performed, revealing signs of chronic active gastritis without findings suggesting a mass or obstructive lesion, reinforcing the need to evaluate the tumor identified on the CT scan with other diagnostic techniques. As the symptoms are nonspecific and the imaging suggests an ill-defined lesion, an exploratory laparotomy protocol was chosen.

During the exploratory laparotomy, the following findings were noted: a mass adhered to the transverse colon near the hepatic flexure, approximately 8 × 8 cm in size. The lesion was irregular in shape, with blood vessels approximately 3 mm in caliber attached to the mesentery surrounding the affected area. Despite the adhesion, the mass showed no signs of perforation or invasion of adjacent organs (**Figure 2**).



Figure 1. Abdominal CT scan: tumor of uncertain behavior in the colon.



Figure 2. Solitary fibrous tumor 8 x 8 cm.

To determine the definitive diagnosis, a surgical resection of the mass was performed (**Figure 3**).

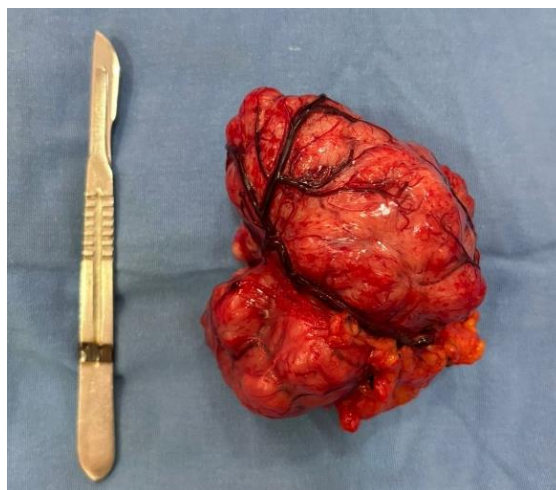


Figure 3. Fibrous tumor with blood vessels of 3mm caliber.

The sample was sent for histopathological analysis, which confirmed the diagnosis of gastrointestinal stromal tumor (GIST). In this case, the lesion is high-risk due to its size and microscopic features of high mitotic activity.

The histopathological report reveals spindle-shaped cells with strong positivity for the CD117 (c-KIT) marker in immunohistochemistry, confirming the nature of GIST.

Subsequently, staging evaluation was conducted to rule out possible metastasis. Additional studies, including a chest CT and abdominal ultrasound, showed no metastatic lesions.

Adjuvant therapy with imatinib was initiated due to the risk of recurrence based on the tumor's high mitotic activity and size. The patient was closely monitored with periodic imaging and monitoring for possible treatment side effects. Six months post-surgery, a follow-up abdominal CT showed a favorable response with no evidence of recurrence or metastasis.

DISCUSSION

Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal neoplasm of the digestive tract, though their incidence is relatively low. They originate from the interstitial cells of Cajal and, in most cases, present mutations in the KIT or PDGFRA genes, which allows for diagnosis and targeted treatment. Their clinical presentation is variable and nonspecific, including symptoms like abdominal pain, gastrointestinal bleeding, and intestinal obstruction, though in some cases they may be incidental findings on imaging studies.

The diagnosis of GIST is based on imaging studies such as contrast-enhanced CT, which allows evaluation of tumor extent, and MRI in specific cases. In accessible lesions, endoscopy and endoscopic ultrasound with biopsy can provide additional information. However, the definitive diagnosis is established through histopathological and immunohistochemical analysis, with CD117 (c-KIT) expression being the most characteristic marker, alongside DOG-1 and, in some cases, CD34.

The treatment of choice for localized tumors is complete surgical resection with negative margins, without the need for lymphadenectomy due to their low metastatic potential to lymph nodes. In smaller tumors without evidence of invasion, laparoscopic surgery may be an alternative. In high-risk cases, defined by size, location, and mitotic index, adjuvant therapy with imatinib has been shown to improve disease-free survival. In metastatic or unresectable disease, imatinib is the first-line treatment, with options such as sunitinib or regorafenib in cases of resistance or progression.

The evolution of patients with GIST depends on multiple factors. Gastric tumors have a better prognosis compared to those in the small intestine, while rectal tumors have a higher risk of recurrence. In patients undergoing complete resection, the five-year survival rate is high, but those with metastatic disease require prolonged treatment to improve their prognosis. With the use of targeted therapy, the median survival in advanced disease has significantly increased in recent years.

Clinical and radiological follow-up is crucial to detect recurrences or tumor progression, with periodic studies recommended based on the risk of each patient. Early diagnosis and a multidisciplinary approach have improved the management of GISTs, but challenges remain in optimizing therapeutic strategies and identifying new prognostic biomarkers.

CONCLUSION

This case illustrates the complexity in the diagnosis and management of gastrointestinal stromal tumors (GISTs), a rare but relevant entity among digestive neoplasms. The patient, with a clinical picture of vague abdominal symptoms, was diagnosed after a series of studies that included abdominal CT, endoscopy, and finally, exploratory laparotomy, which allowed for the resection of the mass. The definitive diagnosis of GIST was established through histopathological study, confirmed by immunohistochemistry positive for CD117, a characteristic marker of this neoplasm. Treatment with imatinib was

GIST in a Middle-Aged Patient: Clinical Case and Review of Diagnostic and Therapeutic Approaches

initiated due to the high risk of recurrence associated with the tumor's size and mitotic activity, consistent with current management of high-risk GISTs.

Globally, GISTs account for approximately 1-2% of all malignant gastrointestinal tract tumors, with the stomach and small intestine being the most common sites. Although they are rare tumors, their incidence has increased in recent decades due to advancements in imaging diagnostic techniques and greater awareness of the disease. GISTs are unique in their biology, as almost all present mutations in the KIT or PDGFRA genes, enabling the use of targeted therapies like imatinib, which has transformed the prognosis of patients with advanced or metastatic disease.

Surgical treatment remains the cornerstone in managing localized GISTs, with complete resection and negative margins being the intervention of choice. However, in high-risk GISTs or metastatic disease, the combination of surgery and tyrosine kinase inhibitors like imatinib has significantly improved survival, increasing long-term survival rates. In fact, patients with metastatic GISTs treated with imatinib have a significantly higher median survival compared to those without targeted treatment.

The prognosis of GISTs depends on multiple factors, such as size, location, mitotic index, and specific genetic mutations. Gastric tumors tend to have a more favorable prognosis compared to small intestine tumors, which have a higher risk of recurrence. Clinical and imaging follow-up is crucial to detect potential recurrences, as these tumors can reappear even years after initial resection.

This case underscores the importance of early diagnosis and a multidisciplinary approach in managing GISTs, highlighting the relevance of advances in personalized medicine, which allow for more effective and less toxic treatments for patients. As more research is conducted on the biology of these tumors and new therapeutic targets are identified, clinical outcomes are expected to continue improving worldwide.

REFERENCES

- I. Agaimy, A., & Köhler, H. (2019). Gastrointestinal stromal tumors: A review of the pathologic features, diagnosis, and treatment. *Journal of Clinical Pathology*, 72(2), 102-109. <https://doi.org/10.1136/jclinpath-2018-2059>
- II. Miettinen, M., & Lasota, J. (2006). Gastrointestinal stromal tumors: Pathology and prognosis at different sites. *Seminars in Diagnostic Pathology*, 23(2), 70-83. <https://doi.org/10.1053/j.semdp.2006.02.006>
- III. Demetri, G. D., von Mehren, M., Blanke, C. D., van Oosterom, A. T., Garrett, C. R., & Verweij, J. (2002). Efficacy and safety of imatinib mesylate in advanced gastrointestinal stromal tumors. *The New England Journal of Medicine*, 347(7), 472-480. <https://doi.org/10.1056/NEJMoa020461>
- IV. Joensuu, H., & Hohenberger, P. (2009). Gastrointestinal stromal tumor. *The Lancet Oncology*, 10(2), 157-164. [https://doi.org/10.1016/S1470-2045\(08\)70360-8](https://doi.org/10.1016/S1470-2045(08)70360-8)
- V. Van der Zwan, J. M., van Oosterom, A. T., & O'Toole, D. (2012). The role of imatinib in the treatment of advanced gastrointestinal stromal tumors: A comprehensive review. *Cancer*, 118(17), 4190-4197. <https://doi.org/10.1002/cncr.26778>
- VI. Cony, M., Salazar, M., & Vera, R. (2017). Update on gastrointestinal stromal tumors: Diagnosis, treatment, and prognosis. *Revista Médica de Chile*, 145(9), 1234-1244. <https://doi.org/10.4067/s0034-98872017000900013>
- VII. Heinrich, M. C., & Corless, C. L. (2005). Gastrointestinal stromal tumors: Current concepts and management. *Archives of Pathology & Laboratory Medicine*, 129(3), 358-365. [https://doi.org/10.1043/1543-2165\(2005\)129<358:GSTCCM>2.0.CO;2](https://doi.org/10.1043/1543-2165(2005)129<358:GSTCCM>2.0.CO;2)
- VIII. Casali, P. G., Abecassis, N., & Whelan, J. (2018). Gastrointestinal stromal tumors: ESMO-EURACAN Clinical Practice Guidelines for diagnosis, treatment, and follow-up. *Annals of Oncology*, 29(4), 1025-1033. <https://doi.org/10.1093/annonc/mdy065>
- IX. Thway, K., & Fisher, C. (2017). Gastrointestinal stromal tumor: Pathology and prognostic factors. *Surgical Oncology Clinics of North America*, 26(4), 547-557. <https://doi.org/10.1016/j.soc.2017.05.005>
- X. Corless, C. L., Barnett, C. M., & Heinrich, M. C. (2011). Gastrointestinal stromal tumors: Analysis of prognostic factors and management. *Hematology/Oncology Clinics of North America*, 25(5), 1013-1037. <https://doi.org/10.1016/j.hoc.2011.07.001>
- XI. Kang, J. H., & Lee, S. H. (2015). Gastrointestinal stromal tumors of the stomach: A review of pathogenesis and treatment. *Journal of Korean Medical Science*, 30(6), 765-772. <https://doi.org/10.3346/jkms.2015.30.6.765>
- XII. Zeng, L., & Zhang, Z. (2014). The role of surgery in the treatment of gastrointestinal stromal tumors. *OncoTargets and Therapy*, 7, 1333-1342. <https://doi.org/10.2147/OTT.S64710>
- XIII. Verweij, J., & Casali, P. G. (2004). Imatinib mesylate: Antitumor activity and pharmacokinetics in gastrointestinal stromal tumors. *Journal of Clinical Oncology*, 22(10), 1856-1863. <https://doi.org/10.1200/JCO.2004.02.043>
- XIV. Hirota, S., & Isozaki, K. (1998). Gain-of-function mutations of c-kit in human gastrointestinal stromal tumors. *Science*, 279(5350), 577-580. <https://doi.org/10.1126/science.279.5350.577>

- XV. Koo, J. S., & Lee, J. K. (2017). Gastrointestinal stromal tumors in non-Asian populations: A review of prognostic factors and treatment outcomes. *World Journal of Gastroenterology*, 23(3), 558-567. <https://doi.org/10.3748/wjg.v23.i3.558>