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CASE REPORT

Gastrointestinal Stromal Tumor (GIST) Arising in the Setting of a Large Hiatal Hernia: A Case Report

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Background: The occurrence of a gastric neoplasm within a hiatal hernia is a rare clinical entity that poses significant diagnostic challenges. While imaging may initially suggest more common malignancies like adenocarcinoma, gastrointestinal stromal tumors (GISTs) should be considered as a critical differential diagnosis due to their distinct management implications.

Case Presentation: A 53-year-old Somali male presented with fatigue, dizziness, early satiety, and shortness of breath on exertion. CT revealed a large hiatal hernia with an 8.4×6.5 cm heterogeneous mass arising from both the herniated and non-herniated portions of the gastric wall, and extending into the esophageal lumen. Endoscopic biopsies were non-diagnostic. Surgical resection confirmed a high-risk GIST (pT4, 8.4 cm, mitotic index 8/50 HPF). The patient received adjuvant imatinib 400 mg daily. Follow-up CT on August 10, 2025, showed a 6.2×6.0 cm recurrent lesion.

Conclusion: A hiatal hernia with mass should prompt a strong suspicion for malignancy. Although gastric adenocarcinoma is more commonly suspected, GIST should remain an important differential. This case highlights the essential role of immunohistochemistry in achieving a definitive diagnosis and underscores the aggressive nature of high-risk GISTs. This necessitates complete surgical resection, consideration of adjuvant therapy, and rigorous radiological surveillance.

Keywords: hiatal hernia, gastrointestinal stromal tumor, gist, computed tomography, immunohistochemistry, neoplasm recurrence

Background

A hiatal hernia is a common condition where the stomach herniates through the esophageal hiatus of the diaphragm into the thoracic cavity. It is classified into four types, with Type I (sliding hernia) being the most prevalent, whereas Types II–IV (paraesophageal hernias) involve other parts of the stomach and sometimes adjacent organs.[1] While hiatal hernias are often incidental findings during imaging or endoscopic studies, their clinical significance increases when associated with complications such as incarceration or volvulus, which can result in organ ischemia, necessitating further evaluation and management.[2]

Gastrointestinal stromal tumors (GIST) are a rare type of mesenchymal tumor that predominantly occurs in the stomach and has only recently been identified as a distinct tumor category.[3] The interstitial cells of Cajal, derived from mesoderm, act as the pacemaker cells in the gastrointestinal tract and are thought to be responsible for the formation of GISTs.[4] Diagnosis is challenging due to variations in cytogenetics and immunohistochemistry among patients, and treatment of localized GIST typically involves complete surgical resection.[5,6] For high-risk tumors, adjuvant imatinib is standard. Neoadjuvant imatinib may be considered for borderline resectable or metastatic disease. Chemotherapy is not effective for GIST.[7–9] When such neoplasms arise within a herniated stomach, they may present with vague thoracoabdominal symptoms such as epigastric pain, nausea, vomiting, weight loss, heartburn, dyspnea, and palpitations.[10]

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This report details a rare case of a high-risk gastric GIST arising in the context of a large hiatal hernia. We aim to illustrate the pivotal role of cross-sectional imaging in identifying the neoplasm, discuss the radiological features that differentiate it from more common carcinomas, and emphasize the diagnostic challenge posed by this unusual presentation.

Case Presentation

We present the case of a 53-year-old Somali male with no chronic illnesses, a non-smoking status, and an unremarkable surgical and family history of cancer. He presented with fatigue, generalized weakness, and dizziness upon standing for 3 weeks. He then developed intermittent shortness of breath on exertion and early satiety. The patient also reported a chronic non-productive cough. He denied chest pain, hemoptysis, or hematemesis. On admission, his heart rate was 108 bpm (tachycardia), and blood pressure was 110/68 mmHg. Laboratory studies revealed a microcytic anemia with hemoglobin of 9.2 g/dL, hematocrit of 27.6%, and mean corpuscular volume of 72 fL. Platelet count was 450 × 10^3/μL (reactive thrombocytosis), and white blood cell count was within normal limits; random blood sugar was 122 mg/dl. ECG showed sinus rhythm with tachycardia.

A comprehensive evaluation was performed using contrast-enhanced computed tomography (CT) of the chest and abdomen on a 128-slice multi-detector CT scanner on April 6, 2025. Images were acquired in both the arterial and portal venous phases following intravenous administration of iodinated contrast material, with axial, coronal, and sagittal reconstructions.

Reviewing the results, two board-certified radiologists with expertise in abdominal imaging identified Large hiatal hernia with a significant portion of the stomach herniated into the left thoracic cavity through the esophageal hiatus, consistent with Type III. Associated with a large, heterogeneously enhancing mass arising from the gastric wall and involving both herniated and non-herniated gastric segments. The mass extended into the distal esophageal lumen, measured approximately 8.4×6.5 cm, and displayed areas of necrosis. It abutted thoracic structures, including the left lower lobe of the lung, where adjacent atelectasis and a mild left pleural effusion were noted. Mildly enlarged perigastric lymph nodes were also noted; however, there was no evidence of liver or peritoneal metastasis, and no suspicious pulmonary nodules were seen at the time of imaging. Additionally, a round soft tissue lesion measuring approximately 3.8×3.2 cm was seen posterior to the stomach, abutting the posterior gastric wall (Figure 1). These findings were highly suggestive of gastric malignancy, most suspicious for adenocarcinoma. The differential diagnosis for the posterior gastric lesion included an exophytic gastric mass or metastatic lymph node. An endoscopic evaluation and histopathological correlation were therefore advised.

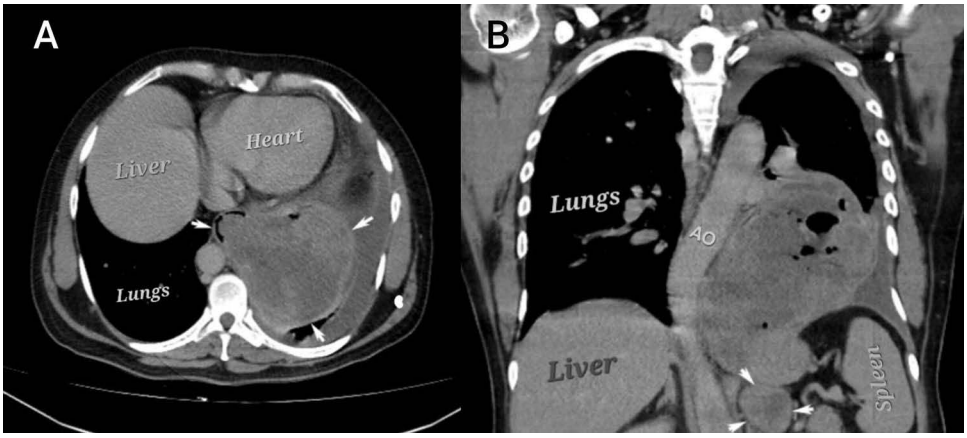

Figure 1 Preoperative contrast-enhanced CT scan (April 6, 2025). (**A**) Axial CT showing a large, heterogeneously enhancing mass (8.4 × 6.5 cm) arising from the herniated gastric wall (arrows). (**B**) Coronal CT revealing a round soft-tissue lesion measuring approximately 3.8 × 3.2 cm located posterior to the stomach and abutting the posterior gastric wall (arrows).
Abbreviation: AO, Aorta.

Endoscopy was performed following the CT scan and revealed a large gastric mass with normal overlying mucosa. Endoscopic biopsies were taken but were non-diagnostic, showing no malignant cells. Subsequently, an endoscopic ultrasound-guided core biopsy of the gastric lesion was performed preoperatively, which confirmed gastrointestinal stromal tumor (GIST). Given this diagnosis and the decision to proceed directly to surgery, the exophytic posterior gastric mass was presumed to be a GIST. Routine preoperative screening was performed according to institutional protocol for all surgical patients, including serological tests for Hepatitis B Virus (HBV), Hepatitis C Virus (HCV), and Human Immunodeficiency Virus (HIV). All results were negative.

Preoperatively, despite a hemoglobin of 9.2 g/dL, no red blood cell transfusion was administered as the patient was hemodynamically stable and the Association for the Advancement of Blood and Biotherapies (AABB) threshold of 7 g/dL was not met. Intraoperatively, tranexamic acid (1 g IV) was given to reduce bleeding, and transfusion was withheld (hemoglobin remained above 7 g/dL).

The patient underwent a single combined surgery on April 18, 2025, via upper midline laparotomy. The procedure included reduction of the hiatal hernia, wedge resection of the gastric wall bearing the GIST (partial gastrectomy) with enucleation of the tumor from the distal esophagus, preserving as much functional stomach and esophageal sphincter as possible, Wedge excision of the posterior exophytic GIST, during which intraoperative inspection revealed a separate posterior mass, hiatal hernia repair with crural approximation. No formal lymph node dissection was performed. The surgical specimen was sent for histopathological and immunohistochemical evaluation. After the operation, hemoglobin declined to 8.3 g/dL on day 2, but transfusion was again withheld per the restrictive threshold, and intravenous ferric carboxymaltose 1000 mg single dose was administered instead.

Histopathological examination of both surgical specimens revealed mostly identical findings. Grossly, the tumors were well-circumscribed, tan-white masses with central necrosis, hemorrhagic foci, and areas of cystic degeneration. Microscopically, a gastric spindle cell neoplasm was identified, composed of elongated spindle cells with fibrillary cytoplasm, cigar-shaped nuclei, and focal nuclear palisading.

Immunohistochemical (IHC) staining demonstrated strong positivity for DOG1 and CD117 (c-KIT), while the tumor cells were negative for S100 and desmin. This immunoprofile—specifically the co-expression of DOG1 and CD117—is diagnostic of a gastrointestinal stromal tumor (GIST). The tumor was classified as high-risk for recurrence according to the modified NIH consensus criteria based on the following features: tumor size >8 cm (8.4 cm), mitotic index >5/50 HPF (8/50 HPF), tumor location (gastric), and presence of tumor rupture/necrosis (central necrosis present). The pathological stage was pT4 (invasion through serosa). The estimated 5-year recurrence-free survival for this risk category is approximately 30–40% without adjuvant therapy.

Following surgery, the patient was started on adjuvant imatinib 400 mg daily, initiated 4 weeks postoperatively to allow adequate wound healing. He was planned to remain on imatinib for 3 years unless recurrence or intolerance developed.

A follow-up contrast-enhanced CT scan of the chest and abdomen was performed on August 10, 2025 (3 months and 23 days post-surgery) to monitor disease status. Compared with the preoperative scan from April 6, 2025, the study revealed a new soft tissue lesion located posterior to the stomach with similar enhancement pattern, measuring 6.2 × 6.0 cm, abutting and indenting the posterior gastric wall. Associated focal wall thickening at the gastric cardia was also noted. The esophagus is distended with fluid without wall thickening. No evidence of hiatal hernia is noted (Figure 2). These findings raised high suspicion for early local recurrence or residual disease, consistent with an aggressive, high-risk GIST. Given disease progression while on adjuvant imatinib, the patient was switched to second-line therapy with Sunitinib 50 mg daily, administered on a 4-week-on, 2-weeks-off schedule, for long-term management. The patient was counseled on the high-risk nature of the GIST and the need for close surveillance. A follow-up schedule of clinical evaluation every 3 months with contrast-enhanced CT was recommended. However, no further imaging was obtained after August 10, 2025, as the patient moved to a rural area and was lost to follow-up. At the time of manuscript submission, his clinical status beyond that date remains unknown.

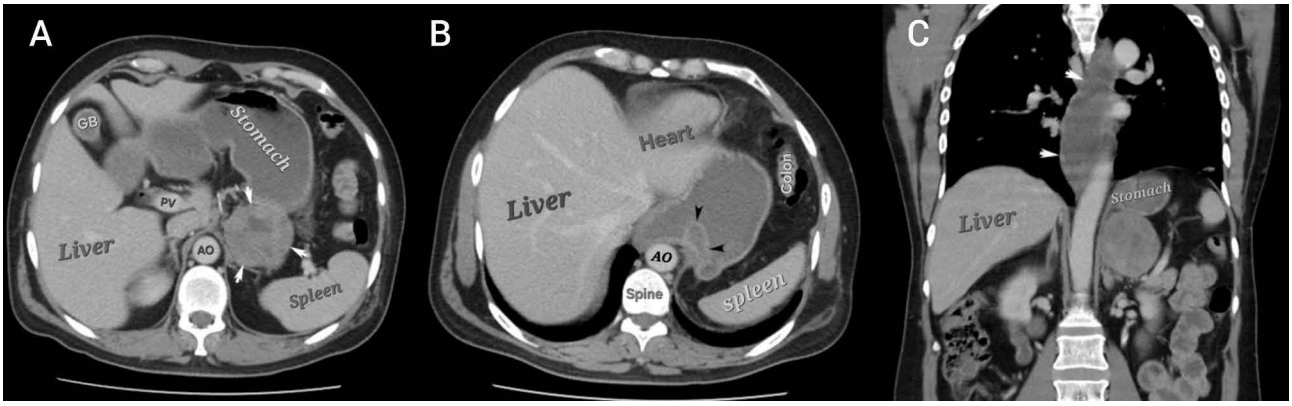

Figure 2 Postoperative follow-up contrast-enhanced CT scan (August 10, 2025). (**A**) Axial CT demonstrating a soft-tissue lesion posterior to the stomach (6.2 × 6.0 cm) abutting and indenting the posterior gastric wall (arrows). (**B**) Axial CT showing focal wall thickening at the cardia of the stomach (arrowheads). (**C**) Coronal CT showing a fluid-distended esophagus without wall thickening (arrows); no evidence of hiatal hernia is noted.
Abbreviations: GB, Gallbladder; PV, Portal Vein; AO, Aorta.

Discussion

The occurrence of a malignant gastric tumor within a hiatal hernia is a rare clinical finding, with only a few case reports documented in the literature.[10,11] This anatomical situation presents significant diagnostic challenges. The tumor's location in the thoracic cavity can lead to atypical symptoms, and routine endoscopy may not adequately visualize the herniated gastric segment, especially in cases of large paraesophageal hernias. This can result in a delayed diagnosis.[12,13]

Our case illustrates a critical diagnostic pitfall: the radiographic mimicry of a gastrointestinal stromal tumor (GIST). Initial contrast-enhanced CT findings revealed a large, heterogeneously enhancing mass with necrosis, transmural involvement, and extension into the esophagus and thoracic cavity. These features raised a strong suspicion of malignancy, most commonly adenocarcinoma.[14] However, the definitive histopathological and immunohistochemical profile (DOG1+, CD117+) confirmed the diagnosis of GIST.[15] This discrepancy highlights a fundamental oncologic principle: while imaging is effective for detecting and staging aggressive tumors, it cannot reliably determine the histologic type. The final diagnosis, which is crucial for guiding treatment, depends on tissue acquisition and immunohistochemical analysis.[16]

Follow-up imaging showed rapid growth of a soft tissue lesion posterior to the stomach, along with focal wall thickening, which raises concerns for recurrent or residual disease. This suggests an aggressive tumor biology, consistent with the resected tumor being classified as a high-risk (pT4) GIST.[17] The differential diagnosis for the posterior lesion includes a recurrent exophytic tumor nodule or, less likely, metastatic lymphadenopathy, which is a less common spread pattern for GIST compared to carcinomas.[18]

In this complex scenario, computed tomography (CT) is essential. It provides a comprehensive preoperative assessment by defining the relationship of the mass to the hernia, evaluating local invasion, and identifying any distant metastases.[19,20] While endoscopy and biopsy are critical for making a definitive diagnosis, the findings from CT dictate the urgency and extent of further intervention.[21] Traditional endoscopic biopsies are frequently false-negative for Gastrointestinal Stromal Tumors (GISTs) because these lesions are subepithelial, typically originating in the deep muscularis propria.[22] Since standard forceps only sample the superficial mucosa, they often fail to reach the tumor, necessitating the use of Endoscopic Ultrasound (EUS). EUS serves as the definitive modality by providing a cross-sectional view of the gastric wall layers and facilitating EUS-guided biopsy.[23] This allows for the acquisition of deep tissue samples required for essential immunohistochemical markers, such as CD117 and DOG1, to confirm the diagnosis.[24] This sequence underscores the necessity of a multidisciplinary approach in managing complex gastrointestinal cases.

From a management perspective, a gastrointestinal stromal tumor (GIST) within a hiatal hernia presents a unique surgical challenge, requiring a dual procedure that combines oncological resection with hernia reduction and repair.[25] This complexity emphasizes the need for a multidisciplinary approach, involving collaboration between gastroenterology, radiology, oncology, and surgical oncology to develop an optimal treatment strategy.[26] This strategy may include surgery and adjuvant therapy with imatinib, especially in high-risk cases like this one, to enhance patient outcomes.[27]

Our management approach incorporated risk stratification based on the modified NIH criteria, which evaluates tumor size, mitotic rate, and primary site to estimate recurrence risk. In accordance with the European Society for Medical Oncology (ESMO) clinical practice guidelines, multidisciplinary evaluation and genotyping for KIT and PDGFRA mutations were prioritized to guide prognosis and surgical planning.[28,29] Furthermore, the role of adjuvant imatinib in high-risk GIST was central to the postoperative strategy; adhering to current evidence, 400 mg daily for a duration of 3 years—or potentially extended up to 6 years—was considered to optimize recurrence-free and overall survival.[30,31]

A primary limitation of this case report is the lack of long-term follow-up beyond three months post-recurrence due to the patient's relocation to a rural area. We planned to continue sunitinib to the 11-month mark, the long-term durability of the response to second-line therapy remains unknown. Despite this constraint, the clinical record successfully captured the early recurrence and the subsequent transition to secondary treatment.

Conclusion

The presence of a large hiatal hernia with a heterogeneous mass is a red flag for malignancy. While imaging may initially suggest more common entities like gastric adenocarcinoma, this case underscores that gastrointestinal stromal tumors (GISTs) are a critical differential diagnosis. Key radiological clues, such as a large, necrotic mass with transmural involvement and extension into adjacent structures, should prompt this consideration. Ultimately, this report highlights that imaging characterizes morphology, not histology; the definitive diagnosis rests on histopathological and immunohistochemical confirmation (eg., DOG1, CD117), which is essential for guiding correct management. Furthermore, the early recurrence in this high-risk GIST highlights the aggressive potential of these tumors and the necessity of a multidisciplinary approach—including surgery, consideration of adjuvant therapy (imatinib for high-risk cases), and vigilant long-term surveillance—to optimize patient outcomes.

Ethics and Consent

Institutional ethical approval was not required for the publication of this case report, as it involves the retrospective description of a single clinical case and does not constitute human subjects research under local institutional policies. The authors obtained written informed consent from the patient for the publication of this report and any accompanying images. Patient anonymity has been strictly maintained. All procedures were conducted in accordance with the Declaration of Helsinki.

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Author Contributions

All authors made a significant contribution to the work reported, whether that is in the conception, study design, execution, acquisition of data, analysis and interpretation, or in all these areas; took part in drafting, revising or critically reviewing the article; gave final approval of the version to be published; have agreed on the journal to which the article has been submitted; and agree to be accountable for all aspects of the work.

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Disclosure

The authors report no competing interests in this work.

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