

Metastatic Rectal Gastrointestinal Stromal Tumor: A Case Report

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Abstract

Rectal gastrointestinal stromal tumors (GISTs) are rare mesenchymal neoplasms, accounting for only a small proportion of gastrointestinal stromal tumors. Their nonspecific clinical presentation and predominantly submucosal and exophytic growth pattern may make diagnosis challenging. We report the case of a 61-year-old man presenting with anal pain, pruritus, and chronic constipation. Pelvic magnetic resonance imaging revealed a large rectal mass with predominantly exoluminal growth, diffusion restriction, and enhancement after gadolinium administration. Thoracoabdominopelvic computed tomography demonstrated hepatic and pulmonary metastases associated with peritoneal carcinomatosis. CT-guided biopsy of the rectal mass confirmed the diagnosis of a gastrointestinal stromal tumor, with positive immunohistochemical staining for CD117 and DOG1. Given the metastatic stage at diagnosis, systemic treatment with imatinib was initiated. This case highlights the rare rectal location of GISTs and emphasizes the complementary role of pelvic MRI and CT in local tumor assessment and metastatic staging.

Keywords: *Gastrointestinal Stromal Tumor; GIST; Rectum; Magnetic Resonance Imaging; Computed Tomography; Metastasis; Imatinib*

Introduction

Gastrointestinal stromal tumors (GISTs) are the most common mesenchymal tumors of the gastrointestinal tract, although they remain relatively rare. They are believed to arise from the interstitial cells of Cajal or their precursors and are typically characterized by immunohistochemical expression of KIT (CD117) and DOG1. At the molecular level, activating mutations in the KIT or PDGFRA genes, which encode receptor tyrosine kinases, are identified in the majority of adult GISTs [1,2].

While the stomach and small intestine are the most common sites of GISTs, rectal involvement remains exceptional, accounting for approximately 5% of all GISTs and less than 0.1% of rectal tumors [3,4]. This unusual location represents a diagnostic challenge because of its often-nonspecific clinical presentation and the predominantly submucosal and exophytic growth pattern of these tumors.

In this context, imaging plays a central role in the diagnostic and therapeutic workup. Pelvic magnetic resonance imaging (MRI), owing to its excellent soft-tissue contrast resolution, enables accurate assessment of the tumor and its relationship with the mesorectum, mesorectal fascia, sphincter complex, and adjacent pelvic organs. Thoracoabdominopelvic computed tomography (CT) complements this evaluation by allowing the detection of distant metastases, particularly hepatic and peritoneal involvement [1,5].

We report the case of a 61-year-old man with a rectal GIST presenting at a metastatic stage, with hepatic and pulmonary metastases associated with peritoneal carcinomatosis. Through this case, we aim to highlight the imaging features of this rare tumor location and emphasize the complementary roles of MRI and CT in locoregional assessment and metastatic staging.

Case Report

A 61-year-old man with a history of active chronic smoking estimated at 45 pack-years and chronic alcohol consumption for approximately 30 years presented with anal pruritus, moderate-to-severe burning anal pain, and chronic constipation, with preserved general condition. His family history was notable for colorectal cancer in his mother.

Clinical examination revealed a soft abdomen with mild pelvic tenderness and low-volume rectal bleeding. Rectoscopy demonstrated an ulceroproliferative rectal lesion. The patient had never previously undergone colonoscopy. Histopathological examination of the initial endoscopic biopsy showed glandular hyperplasia associated with a moderately differentiated adenomatous polyp, without evidence of malignant cells.

Pelvic magnetic resonance imaging (MRI) subsequently revealed a large, poorly defined rectal mass with irregular margins and predominantly exoluminal growth, extending to the upper rectum. The lesion measured approximately 42 × 113 mm in transverse and craniocaudal dimensions and was located 4 cm from the anal verge. It demonstrated intermediate signal intensity on T1- and T2-weighted images, high signal intensity on diffusion-weighted imaging with low apparent diffusion coefficient (ADC) values, and enhancement following gadolinium administration (Figure 1).

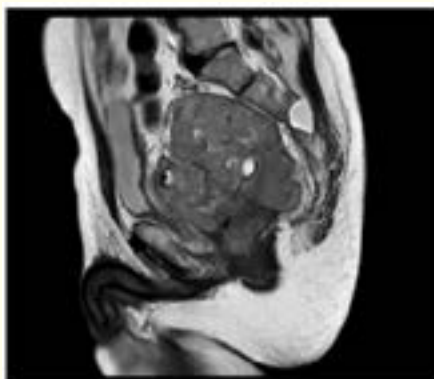


Figure 1: Rectal GIST on pelvic MRI.

Sagittal T2-weighted pelvic MRI demonstrates a large rectal mass with predominantly exophytic growth, irregular margins, and heterogeneous T2 signal intensity, with small internal hyperintense areas suggestive of cystic or necrotic changes.

Thoracoabdominopelvic computed tomography (CT) confirmed the large rectal mass, which showed predominantly exoluminal growth, irregular margins, heterogeneous enhancement, and internal areas of necrosis (Figure 2). The staging CT additionally demonstrated hepatic lesions consistent with metastases, a subhepatic peritoneal carcinomatosis nodule (Figure 3), and a pulmonary nodule in the anterior segment of the right lower lobe (Figure 4).

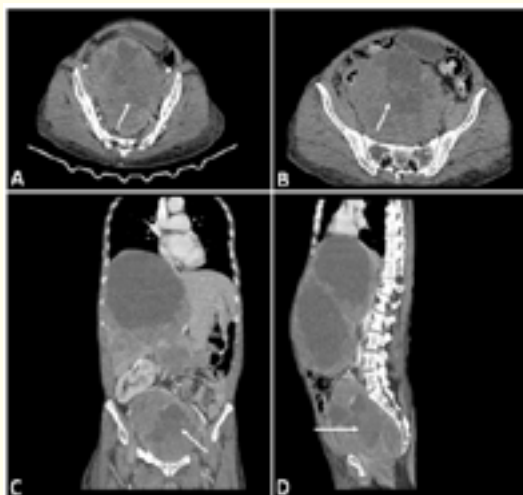


Figure 2: CT appearance of the rectal GIST.

Contrast-enhanced CT images demonstrate a large, heterogeneous rectal mass with predominantly exophytic growth and irregular margins (arrows). (A) Unenhanced axial CT image. (B) Axial CT image obtained after contrast administration. (C) Coronal and (D) sagittal portal venous phase reformations better demonstrate the large craniocaudal extent of the tumor and its heterogeneous appearance with internal areas of necrosis.

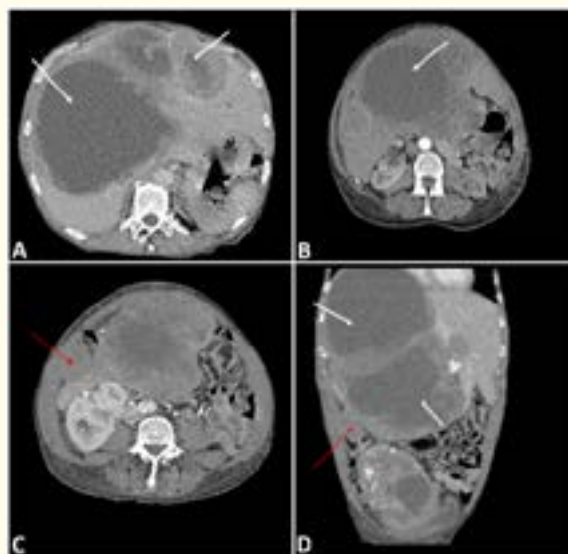


Figure 3: Hepatic and peritoneal metastases.

Contrast-enhanced abdominal CT demonstrates multiple large heterogeneous hepatic metastases with central areas of necrosis (white arrows), associated with a subhepatic peritoneal metastatic nodule (red arrows). (A) Unenhanced axial CT image. (B) Arterial-phase axial image. (C) Portal venous-phase axial image and (D) coronal portal venous-phase reformation.

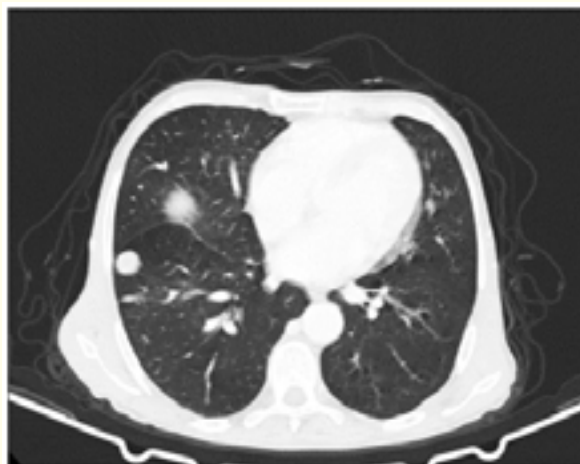


Figure 4: Pulmonary metastatic involvement.

Axial chest CT image in lung window demonstrates a well-defined, rounded solid pulmonary nodule in the anterior segment of the right lower lobe, consistent with metastatic involvement in the context of metastatic rectal GIST.

Given the discrepancy between the initial endoscopic biopsy findings and the radiological appearance of the lesion, a CT-guided biopsy of the rectal mass was performed. Histopathological examination established the diagnosis of a gastrointestinal stromal tumor, and immunohistochemical analysis demonstrated positive staining for CD117 (KIT) and DOG1, confirming the diagnosis of rectal GIST.

Given the metastatic presentation, with hepatic and pulmonary metastases associated with peritoneal carcinomatosis, curative surgical management was not considered appropriate, and systemic treatment with imatinib 400 mg/day was initiated.

Discussion

Gastrointestinal stromal tumors (GISTs) are rare mesenchymal neoplasms that may arise throughout the gastrointestinal tract. The stomach is the most common primary site, accounting for approximately 50 - 70% of cases, followed by the small intestine in 20 - 30%. Colorectal involvement is considerably less frequent, and rectal GISTs represent approximately 5% of all GISTs and less than 0.1% of rectal tumors [3,4]. Their rarity, combined with the anatomical constraints of the pelvis and their potentially aggressive behavior, makes rectal GISTs a distinct diagnostic and therapeutic challenge [5].

GISTs are thought to originate from the interstitial cells of Cajal (ICCs) or from a common mesenchymal precursor. These cells, located within the muscular layers of the gastrointestinal tract, play a key role in gastrointestinal motility by generating and coordinating slow electrical waves. Morphological, ultrastructural, immunohistochemical, and molecular similarities between ICCs and GIST cells support this histogenetic relationship [6].

The clinical presentation of rectal GISTs is variable and often nonspecific, depending mainly on tumor size, growth pattern, and local extension. Reported manifestations include pelvic or anal pain, altered bowel habits, constipation, rectal bleeding, and tenesmus, while some tumors may be discovered incidentally [5,7]. In our patient, anal pruritus, burning anal pain, chronic constipation, and mild rectal bleeding were the presenting symptoms. This nonspecific clinical presentation illustrates the potential difficulty of recognizing rectal GISTs on clinical grounds alone.

Another important diagnostic feature is their predominantly submucosal and exophytic growth pattern. Because the overlying rectal mucosa may remain relatively preserved, conventional endoscopic biopsies can be inconclusive and may fail to obtain representative tumor tissue [1,5]. This diagnostic limitation was particularly illustrated in our case, in which the initial endoscopic biopsy showed glandular hyperplasia associated with an adenomatous polyp without malignant cells, despite the presence of a large rectal mass on imaging. The definitive diagnosis was subsequently established by CT-guided biopsy. This discrepancy highlights the importance of radiologic-pathologic correlation and appropriate image-guided tissue sampling when imaging suggests a submucosal mesenchymal tumor.

Imaging plays a central role in the evaluation of rectal GISTs, from initial characterization to locoregional assessment, metastatic staging, and therapeutic planning. On CT, rectal GISTs usually appear as soft-tissue masses with a predominantly exophytic growth pattern. Small tumors may be relatively homogeneous, whereas larger lesions frequently demonstrate heterogeneous enhancement related to necrotic, cystic, or hemorrhagic changes [1,7]. In our patient, CT demonstrated a large heterogeneous rectal mass with predominantly exophytic growth, irregular margins, and internal areas of necrosis. These features, together with the large tumor size and metastatic dissemination, reflected an advanced disease presentation.

Pelvic magnetic resonance imaging (MRI) is particularly valuable for locoregional assessment because of its excellent soft-tissue contrast resolution. It allows accurate evaluation of tumor size, local extension, and relationships with the mesorectum, mesorectal fascia, sphincter complex, and adjacent pelvic organs, which are essential parameters for therapeutic planning [1,5]. In our case, MRI demonstrated a large rectal mass with predominantly exophytic growth and heterogeneous T2 signal intensity. The lesion also showed diffusion restriction with low apparent diffusion coefficient values and enhancement following gadolinium administration. These findings contributed to tumor characterization and precise assessment of its local extent.

CT remains essential for staging and for detecting distant disease. The liver and peritoneum are the predominant sites of metastatic spread in GISTs [1,7]. In our patient, thoracoabdominopelvic CT demonstrated extensive hepatic metastatic involvement and a subhepatic peritoneal carcinomatosis nodule, in addition to pulmonary involvement. The presence of hepatic, peritoneal, and pulmonary disease at initial diagnosis underlined the advanced metastatic presentation of this rectal GIST. This case therefore illustrates the complementary roles of cross-sectional imaging: pelvic MRI provides detailed characterization of the primary rectal tumor and its locoregional relationships, whereas CT provides a comprehensive assessment of metastatic dissemination.

Although imaging findings may strongly suggest a GIST, definitive diagnosis relies on histopathological and immunohistochemical examination. Histopathology also contributes to risk assessment through parameters such as tumor size, mitotic activity, cellularity, and the presence of necrosis [8]. Immunohistochemically, KIT (CD117) is expressed in approximately 90-95% of GISTs, while CD34 is also frequently positive [8]. DOG1 has further improved diagnostic accuracy and is particularly valuable in tumors with weak or absent KIT expression [9,10]. In our patient, immunohistochemical positivity for both CD117 and DOG1 supported the diagnosis of rectal GIST.

At the molecular level, GISTs are predominantly characterized by activating mutations involving the KIT and PDGFRA genes, both of which encode receptor tyrosine kinases [11,12]. Identification of these molecular alterations has important diagnostic, prognostic, and therapeutic implications, particularly because tumor genotype may influence sensitivity or resistance to tyrosine kinase inhibitors [12,13]. Thus, the contemporary assessment of GISTs relies on the integration of imaging, histopathological, immunohistochemical, and molecular findings.

Complete surgical resection with negative microscopic margins (R0) remains the standard treatment for localized and resectable GISTs [5,13]. Rectal localization, however, may complicate surgical management because of the confined pelvic anatomy and the close relationship of the tumor with the sphincter complex and adjacent pelvic structures. In selected patients with small localized rectal tumors, less invasive local approaches may be considered [5].

Management differs substantially in advanced or metastatic disease. Imatinib, a tyrosine kinase inhibitor targeting KIT and PDGFRA, represents the standard first-line systemic treatment for advanced GISTs [14,16]. In patients with disease progression or acquired resistance, other tyrosine kinase inhibitors, including sunitinib and subsequently regorafenib, may be considered [14-16]. Cytoreductive surgery may have a role in carefully selected patients with metastatic disease controlled by targeted therapy, although its indication should be individualized [14]. In our patient, the presence of hepatic and pulmonary metastases associated with peritoneal carcinomatosis precluded curative surgical management, and systemic treatment with imatinib at a dose of 400 mg/day was initiated.

Our observation particularly emphasizes the diagnostic value of correlating imaging and pathological findings in rectal GISTs. The large predominantly exophytic rectal mass contrasted with the initially non-diagnostic endoscopic biopsy, leading to CT-guided tissue sampling and definitive diagnosis. Moreover, the complementary use of pelvic MRI for detailed locoregional assessment and thoracoabdominopelvic CT for metastatic staging provided a comprehensive evaluation of the disease and directly contributed to therapeutic decision-making.

Conclusion

Rectal gastrointestinal stromal tumors (GISTs) are rare mesenchymal neoplasms that may represent a significant diagnostic and therapeutic challenge because of their uncommon location, nonspecific clinical presentation, and predominantly submucosal and exophytic growth pattern. Imaging plays a central role in their assessment. Pelvic MRI provides accurate characterization of the primary tumor and its locoregional extension, whereas thoracoabdominopelvic CT is essential for evaluating distant metastatic spread and determining the overall extent of disease.

Definitive diagnosis relies on histopathological examination supported by immunohistochemical analysis, particularly the expression of CD117 and DOG1. The predominantly submucosal nature of these tumors should be considered when conventional endoscopic biopsies are inconclusive. This diagnostic challenge was illustrated in our patient, in whom the initial endoscopic biopsy was non-diagnostic and CT-guided biopsy ultimately established the diagnosis.

Our case illustrates a rare rectal GIST presenting at an advanced stage with hepatic and pulmonary metastases associated with peritoneal carcinomatosis. It highlights the importance of integrating clinical, radiological, histopathological, and immunohistochemical findings to achieve an accurate diagnosis and comprehensive staging. The complementary roles of pelvic MRI for locoregional assessment and CT for metastatic staging are particularly important in guiding therapeutic decision-making, especially in advanced disease requiring systemic targeted therapy.

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Conflict of Interest

The authors declare no conflict of interest.

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