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Small Bowel Gastrointestinal Stromal Tumor Presenting as an Abdominal Mass: A Case Report

Wiam Fellahi[1*], Zaynab Bellamlik[1], Imad El Azzaoui[1], Amine Maazouz[1], Abdelhak Bensal[1], Mohamed Bouzroud[1], Hakim El Kaoui[1], Sidi Mohammed Bouchentouf[1], Mountassir Moujahid[1]

[1]Department of Visceral Surgery 1, Mohammed V Military Hospital, RABAT, MOROCCO

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***Corresponding author:** Wiam Fellahi
Department of Visceral Surgery 1, Mohammed V Military Hospital, RABAT, MOROCCO

Abstract **Case Report**

Gastrointestinal stromal tumors (GISTs) are rare mesenchymal tumors of the digestive tract, accounting for approximately 1–2% of all gastrointestinal neoplasms. Their prognosis depends on several factors, including location, rupture status, tumor size and mitotic index, which together determine the risk classification and guide therapeutic management. We report the case of a 70-year-old male patient admitted for the management of a small bowel GIST at the Department of Visceral Surgery I at the Mohamed V Military Hospital in Rabat. The patient underwent complete surgical resection. Histopathological and immunohistochemical analysis confirmed a high-risk GIST measuring 10.5 cm, with a high mitotic index. Adjuvant therapy with targeted therapy (imatinib) was initiated due to the high risk of recurrence.

Keywords: GIST; Small bowel, Surgery, Imatinib.

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INTRODUCTION

Gastrointestinal stromal tumors (GISTs) are malignant mesenchymal tumors arising from the interstitial cells of Cajal, which function as pacemaker cells regulating gastrointestinal motility due to their location in the muscular layer[1]. They are often located in the stomach and small bowel, and less frequently in the rectum, colon, or esophagus [1,2]. The global incidence is estimated at between 10 and 20 cases per million people per year [3].

GISTs account for approximately 18% of all sarcomas and represent the most common subtype of sarcoma of the digestive tract in adults [4].

They constitute a heterogeneous group in terms of clinical presentation, molecular characteristics, and therapeutic response [5].

Diagnosis may be incidental in up to 20% of cases, often during CT scan, a gastrointestinal endoscopy, or during surgical procedure [6,7]. Symptomatic cases typically present with nonspecific signs such as abdominal pain, a palpable mass, or complications including hemorrhage, perforation, or obstruction. Asymptomatic forms have also been described [6,8,9].

Histopathological diagnosis is based on morphological analysis and confirmed by immunohistochemistry, which most tumors expressing KIT (CD117) and/or DOG1 (discovered on GIST) markers [10] expressed in approximately 95% of cases and are the most sensitive and specific ones. Otherwise, additional markers such as CD34, SMA, desmin, or S100 protein may assist in the diagnosis [11].

Biopsy (through echoendoscopy, percutaneous approach, or surgery) is considered on a case-by-case basis. The guidelines specify that it is not routinely performed, particularly in cases of typical resectable tumors, and that it is considered depending on the chosen treatment (suspected metastasis, necessary neoadjuvant therapy, diagnostic uncertainty) [12].

The prognosis and treatment currently depend on histology, mutation status (KIT, PDGFRA), and the risk of recurrence [9].

Although the identification of the c-KIT proto-oncogene and the introduction of imatinib mesylate have changed the therapeutic, complete surgical resection

remains the standard treatment for localized forms [13]. While targeted therapy with tyrosine kinase inhibitors is indicated in high-risk or metastatic cases [10].

We report the case of a 70-year-old patient with a high-risk small bowel GIST managed surgically in our department.

Case Presentation

We report the case of Mr OB, aged 70.

His medical history included type 2 diabetes, hypertension, ischemic heart disease, and deep vein thrombosis (DVT) under treatment.

Symptoms began in two months with intermittent abdominal pain localized to the epigastric and hypogastric regions, occurring outside mealtimes. This was associated with decline in general condition, including asthenia and unexplained weight loss. There was no history of vomiting, bowel obstruction, or gastrointestinal bleeding (no hematemesis, melena or rectal bleeding).

On physical examination, the patient was in good general condition (WHO performance status 1, ASA II, nutritional grade 2). He was hemodynamically stable with normal vital signs (HR=93, BP=13/8, RR=12, SpO_2=99%), conjunctivae normally colored.

Abdominal examination revealed no abnormalities on inspection; a soft abdomen with a mobile, painless abdominal mass in the hypogastric region measuring 8x5cm. There were no signs of inflammation in the area, no hepatosplenomegaly. Digital rectal examination was normal. No lymphadenopathy was detected, including the left supraclavicular (Troisier's) node.

The patient also presented with signs of left lower limb DVT, including pain, redness, and edema extending to the calf.

An abdominal ultrasound revealed a well-defined heterogeneous mass located above the bladder containing small cystic areas. The mass was roughly oval in shape measuring approximately 110x65mm, suggestive of a GIST.

A follow-up CT scan (AP CT) showed a mesenteric mass adjacent to ileal loops, roughly oval, surrounded by an incomplete capsule of mixed fluid and tissue density with mild enhancement, measuring 87x64 mm initially suggestive of a ruptured GIST. There was infiltration of adjacent mesenteric fat but no evidence of bowel obstruction, lymphadenopathy, or distant metastasis (no pathologically suspicious contrast uptake in the liver) and no abdominal-pelvic fluid.

Further staging with thoraco-abdomino-pelvic CT scan confirmed a well-defined exophytic ileal mass measuring 88 × 69 × 63mm, with heterogeneous enhancement and areas of necrosis, without evidence of rupture or metastatic disease.

The tumor was resectable on CT scan: no invasion of adjacent organs, no vascular invasion, no peritoneal carcinomatosis or liver metastases.

Given its small bowel location and size greater than 5 cm, this lesion falls into a group at high risk of malignancy, according to the MIETTINEN classification, subject to histopathological confirmation.

Pre-operative evaluation showed stable clinical status. ECG revealed atrial fibrillation with a heart rate of 100 bpm. Echocardiography showed moderately reduced left ventricular function (ejection fraction 47%) and a normal left ventricular wall thickness.

Laboratory findings were within acceptable ranges:
WBC 8900x10^3/ µL, Hb 12.8 g/dl, platelets 320x10^3/ µL, PT 71%, APTT 1.1
Troponin 5 ng/l
CRP 49.7 mg/l, Na 136 mmol/l, K 4.84 mmol/l, bicarbonate 27 mmol/l, total protein 72 g/l, urea 0.31 g/l, creatinine 7 mg/l, Ca 88 mg/l, AST 21 UI/l, ALT 17 UI/l, PAL 154 U/l, GGT 71 U/l, lipase 31 UI/l.

Surgical exploration revealed a 15 cm tumor arising from an ileal loop located approximately 10 cm from the terminal ileum. There was no peritoneal carcinomatosis or liver metastasis.

The patient underwent segmental resection of approximately 20 cm of small bowel with primary end-to-end anastomosis. No lymphadenectomy was performed. Five nodules from the Douglas pouch were also excised, peritoneal closure and drainage by a 16 Fr Redon drain placed in the Douglas pouch.

Postoperative recovery was uneventful with return of bowel function on postoperative day 6. The patient was discharged on postoperative day 8 with therapeutic LMWH for at least 3 months, pending the histopathology results, and was subsequently referred to oncology.

Histopathological examination confirmed a gastrointestinal stromal tumor measuring 10.5 cm with a high risk of progression (small bowel location with extraluminal growth, spindle-shaped tumor cells and epithelioid cells, diffuse architecture with fibro-myxoid stroma, high mitotic index with mitoses estimated at 10 mitoses per 50 high-power fields). Surgical margins were clear, negative lymph node dissection 3N-/3N with 5 satellite tumor nodules.

A nodule in the Douglas pouch was histologically in favor of connective tissue infiltrated by the same tumor proliferation described above.

Immunohistochemistry showed diffuse positivity for CD117 and DOG1.
Anti-AML antibody focally positive; Anti-CD34, anti-H-caldesmone and anti-desmin antibodies negative.

The tumor was classified as high-risk. And the patient was referred for adjuvant therapy with imatinib.

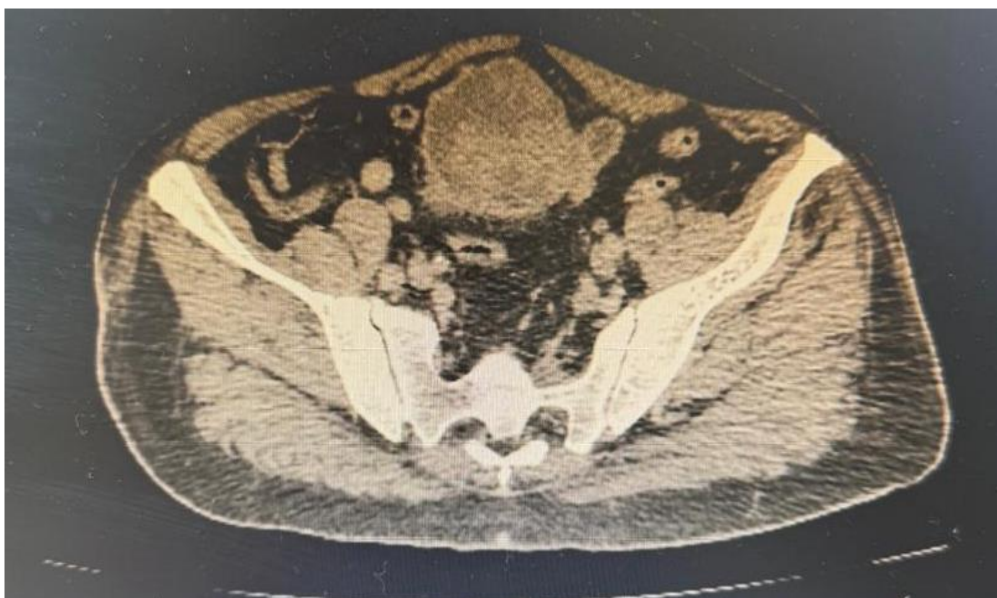

Fig. 1: **Contrast enhanced thoraco-abdomino-pelvic CT scan showing a well-defined mass arising from the ileal wall, oval in shape. The lesion demonstrates an exophytic growth pattern with heterogeneous enhancement and hypodense areas consistent with necrosis. The mass measures 88×69×63 mm (HxTxAP) and is associated with infiltration of the adjacent fat without evidence of rupture**
(Department of Visceral Surgery '1', HMIMV, Rabat, MOROCCO)

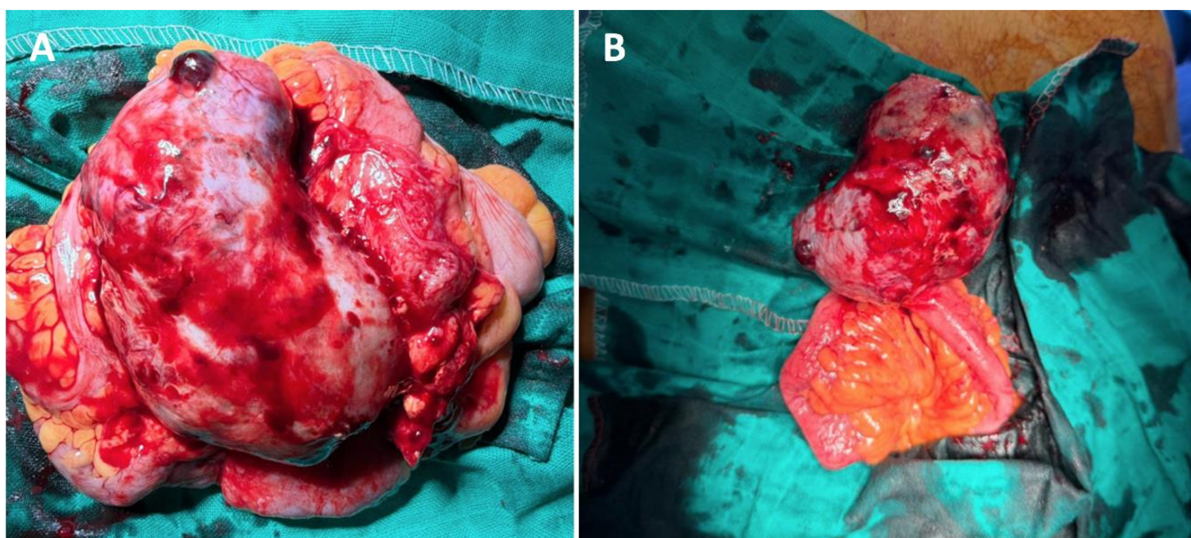

Fig. 2: Intraoperative findings showing a large exophytic tumor arising from the ileal loop.
(A) Anterior view of the abdominal mass.
(B) Close-up view demonstrating the tumor protruding from the small bowel wall.
(Department of Visceral Surgery '1', HMIMV, Rabat, MOROCCO)

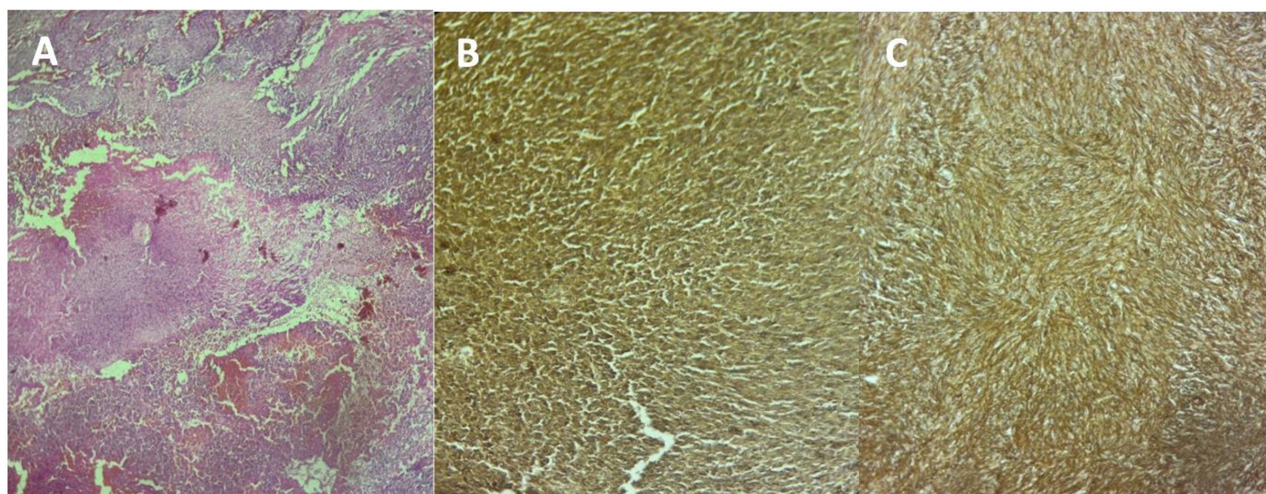

Fig.3: Histopathological and immunohistochemical features of the tumor
(A) Hematoxylin and eosin staining showing diffuse spindle cell proliferation with extensive areas of necrosis (magnification ×10).
(B) Immunohistochemical staining showing diffuse positivity for DOG1.
(C) Immunohistochemical staining showing diffuse positivity for CD117 (KIT).
(Department of Pathology, HMIMV Rabat, MOROCCO)

DISCUSSION

GISTs are rare neoplasms accounting for 1–2% of all gastrointestinal malignancies [14].

In the present case, the patient was 70 years old, which is consistent with previously published data such as a large systematic review by Soreide and al. reporting a median age at diagnosis of approximately 65 years across 29 studies including more than 13,550 patients from 19 countries [3]. Similarly, Khan and al. found that the majority of patients (63%) were older than 60 years [15] . Sex distribution appears to be generally balanced, although some series report a slight male predominance [16,17].

Most GISTs occur sporadically, and no well-established environmental risk factors have been identified. However, a subset of cases is associated with hereditary syndromes or specific genetic alterations [6] .

GISTs arise most commonly in the stomach (60–70%) and small intestine (25–35%). Less frequent locations include the colon, rectum, and appendix (approximately 5%), as well as the esophagus (2–3%). Rarely, primary tumors may originate in the omentum, mesentery, or retroperitoneum; however, most stromal tumors identified at these sites represent metastases from an undetected gastrointestinal primary [18].

Clinically, GISTs may remain asymptomatic in up to 18% of cases, particularly when small, and are often detected incidentally during imaging (CT scan), endoscopy or surgery performed for unrelated conditions. When symptomatic, they typically present with nonspecific manifestations such as abdominal pain (as observed in our patient), nausea, vomiting, abdominal distension, or early satiety. Larger tumors may cause compressive or obstructive symptoms depending on their location. Less commonly, they may present with gastrointestinal hemorrhage or perforation leading to peritonitis [14].

Atypical clinical presentations have also been described, occasionally mimicking other intra-abdominal conditions such as hepatic lesions or pancreatic pseudocysts. These unusual presentations have been reported in several clinical cases, highlighting the broad clinical spectrum and diagnostic challenges associated with GISTs [19–21].

Several imaging modalities are used in the evaluation of GISTs, including computed tomography (CT), magnetic resonance imaging (MRI), FDG-PET, ultrasound, angiography and endoscopy. Contrast-enhanced CT is considered the gold standard imaging modality for diagnosis, staging, treatment planning and response assessment.

However, radiological interpretation can be challenging due to the heterogeneity of tumor appearance, influenced by location, growth pattern, and secondary changes such as necrosis, calcifications or metastasis. In addition, post-treatment changes may be subtle, and recurrence patterns differ from those of other gastrointestinal malignancies [22].

The CT scan findings in our case are consistent with those reported in the literature. According to the review by Joensuu and al. published in *The Lancet*, GISTs most commonly appear on CT scan as well-defined masses, exophytic growth with heterogeneous enhancement due to necrotic or cystic components. In our patient, CT scan revealed a large, exophytic ileal mass with heterogeneous enhancement and hypodense

areas suggestive of necrosis , in keeping with classical descriptions [6].

Contrast-enhanced thoraco-abdomino-pelvic CT remains essential for staging, allowing evaluation of the primary tumor as well as the search for metastases, particularly in the liver and lungs.

In this case, imaging demonstrated a locally advanced ileal tumor with mesenteric fat infiltration but no evidence of distant metastasis, supporting a diagnosis of localized disease. TAP CT scan therefore proves essential for diagnosing and critical for therapeutic decision-making [22].

The role of preoperative biopsy in GIST remains debated. A systematic review by Jakob and al. demonstrated that biopsy provides valuable histological and molecular information, particularly in cases of diagnostic uncertainty or when neoadjuvant therapy is considered.

The main concern remains the risk of tumor dissemination along the needle tract.

Nevertheless, analysis of the included studies showed that the risk is extremely low, with only rare cases reported, suggesting that biopsy is generally a safe procedure when performed under appropriate conditions [23].

In the present case, biopsy was not performed because imaging strongly suggested a resectable localized GIST, in line with current recommendations that do not mandate preoperative histological confirmation in clearly resectable cases [24].

Surgical resection remains the cornerstone of treatment for localized GISTs. International guidelines state that the gold standard involves complete surgical resection (R0) of the tumor, preserving the pseudocapsule and without routine lymphadenectomy, as lymph node metastases are rare. Surgery remains the only potentially curative treatment for localized disease [25].

In our case, complete R0 resection of the ileal tumor was achieved, consistent with guidelines from the European Society for Medical Oncology (ESMO) and the National Comprehensive Cancer Network (NCCN), which advocate segmental resection for localized small bowel GISTs with favorable oncologic outcomes [9,26].

Prognosis in GISTs depends on several factors, including tumor size, mitotic index, anatomical location, tumor rupture and the presence of necrotic changes.

Tumors larger than 5 cm and those arising in the small bowel are associated with a higher risk of recurrence compared with gastric GISTs. A high mitotic index and the presence of tumor necrosis are also adverse prognostic factors, whereas the absence of tumor rupture, as in our case, is associated with a more favorable outcome [27–30].

Several risk stratification systems have been developed, including the NIH classification developed by Fletcher and al., which categorizes risk into four levels based on tumor size and mitotic activity. The Armed Forces Institute of Pathology (AFIP) classification proposed by Miettinen and al., which incorporates anatomical location in risk determination [14].

In our patient, the ileal mass measured approximately 10.5 cm with heterogeneous enhancement and areas of necrosis without signs of rupture, but a mitotic index of 10 mitoses per 50 high-power fields, classifying it as high risk according to these criteria.

An additional noteworthy feature in this case is the presence of deep vein thrombosis (DVT), which led to the initial diagnosis. Although uncommon, thromboembolic event such as a DVT or pulmonary embolism have been reported in few cases of GIST (four cases illustrating the association between these tumors and venous thrombosis have been reported in the literature), Proposed mechanisms include tumor-related hypercoagulability and mechanical vascular compression contributing to the development of thrombosis in patients with GIST. Current guidelines recommend low-molecular-weight heparin (LMWH) as first-line treatment for the acute phase of cancer-associated thrombosis; with extended treatment considered for an indefinite period or until achievement of curative treatment [31].

Adjuvant therapy with tyrosine kinase inhibitors, particularly imatinib is currently the standard treatment for patients with resectable GISTs at high risk of recurrence.

Randomized trials have demonstrated that imatinib administered at 400 mg/day for three years significantly improves both recurrence-free and overall survival compared with shorter durations or no adjuvant therapy [12].

The benefit of imatinib is based on the inhibition of the activating tyrosine kinases KIT and PDGFRA, which are present in the majority of GISTs, thereby slowing malignant tumor progression according to von Mehren and al.[26].

International guidelines from ESMO and GEIS-SEOM emphasize the importance of molecular genotyping prior to treatment initiation to confirm sensitivity to imatinib and guide therapeutic management decisions [12].

In cases of resistance or intolerance, sunitinib is the standard second-line therapy, administered at the recommended dose of 50 mg daily for 4 weeks every 6 weeks or at 37.5 mg daily on a continuous basis, followed by regorafenib as third-line treatment [32].

In the present case, the large tumor size and high-risk classification according justified the use of adjuvant imatinib following complete surgical resection to reduce recurrence risk and improve long-term prognosis [12,26].

Follow-up after complete resection of GISTs and adjuvant treatment with imatinib, involves medical imaging for early detection of recurrence, most commonly in the liver and peritoneum. International guidelines recommend a follow-up strategy based on recurrence risk [33].

Both ESMO and NCCN guidelines recommend intensive monitoring during the first years after surgery since recurrence risk is highest [26].

Thus, according to ESMO recommendations, in cases of high-risk GISTs, monitoring through abdomino-pelvic CT or MRI is recommended every 3 to 6 months during the three years of adjuvant treatment with imatinib, then every 3 to 4 months for the two years following the end of treatment and every 6 months for the following three years and annually for up to 10 years [12].

For GISTs with an intermediate risk of recurrence, CT scan every 6 months for 5 years, then once a year is recommended. For low-risk GISTs, CT every 6–12 months for 5 years is recommended, and for very low-risk GISTs, no routine follow-up is necessary after complete resection [32].

In our case, given the high-risk features of the tumor, close radiological surveillance with CT every 3–6 months during adjuvant therapy with imatinib, followed by continued monitoring after treatment completion, represents the most appropriate strategy to ensure early detection of recurrence.

Ethical approval: Not required for case reports.

Consent: Written informed consent was obtained from the patient for publication of this case report and accompanying images.

Conclusion

Gastrointestinal stromal tumors are rare but represent the most common mesenchymal tumors of the digestive tract with an estimated global incidence of 10–20 cases per million people per year.

Diagnosis relies on imaging (CT scan), which is the gold standard to characterize the tumor and assess its extent, particularly with regard to peritoneal and hepatic spread, as well as histological and immunohistochemical analysis (expression of KIT (CD117) and DOG1 markers). Complete surgical resection with clear margins remains the standard treatment for localized disease.

In high-risk cases, adjuvant therapy with targeted therapies using tyrosine kinase inhibitors (imatinib) significantly improves outcomes.

Close radiological follow up through abdominal CT scans following treatment is essential for early detection of recurrence and improving patients' quality of life.

This case illustrates the importance of radiological diagnosis, an appropriate surgical strategy and the integration of targeted therapies in the management of GISTs to improve prognosis and long-term survival.

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