

An unusual presentation of gastrointestinal stromal tumour arising from the Ileum and involving the urinary bladder

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ABSTRACT

Gastrointestinal stromal tumors (GISTs) are mesenchymal tumors that typically originate from the stomach and small intestines, though they can occasionally originate from unusual sites that do not involve the GI (gastrointestinal) tract. Men between the ages of 60 and 65 are more likely to develop GISTs. Histology and IHC (immunohistochemistry) are used to confirm the diagnosis of these tumors, which are rarely symptomatic and are typically found accidentally on radiography or during surgery. This case illustrates an uncommon presentation in a 65-year-old man who complained of fatigue, rectal bleeding, and dysuria as a result of an underlying infraumbilical tumor. The tumor was later identified as a GIST originating from the ileum and simultaneously affecting the bladder, and it was successfully treated with neoadjuvant imatinib therapy and surgical resection.

Keywords: Bladder gist, Extraintestinal gist, Ileal gist, Imatinib therapy, GIST

INTRODUCTION

The most prevalent mesenchymal tumors originating from the gastrointestinal tract are called gastrointestinal stromal tumors (GISTs). Rarely do they arise from the bowel mesentery, omentum, and retroperitoneal tissue (2–11%), and they are typically found in the stomach (40–70%) and small intestine (15–44%). 1 Extra GISTs (EGISTs) are tumors with similar IHC markers to GIST that develop in the abdomen without affecting the gastrointestinal tract, in the omentum (80%), mesentery, or retroperitoneal structures, and have no connection to the stomach or intestinal wall. Two The interstitial cells of Cajal are the source of GISTs. KIT and PDGFR (platelet derived growth factor receptor) protooncogenes are upregulated in approximately 85–95% of GIST cases. 3 GIST usually manifests around age 60. In children and young adults, GISTs are uncommon. Males were found to have a higher prevalence than females. 1 Although the majority of GISTs happen occasionally, there is ample evidence of GISTs in families with inherited KIT mutations. Furthermore, the development of GISTs has been associated with a number of genetic syndromes, including neurofibromatosis type I, Carney's triad (gastric GIST, paraganglioma, and pulmonary chondroma), and Carney's dyad (gastric GIST and paraganglioma). 3 According to their histological appearance, GIST subtypes include mixed (10%), epithelioid (20%), and spindle cell (70%). The spindle cell subtype usually appears as a large, diverse, enhancing mass that comes from the small intestine or stomach. It typically exhibits a favorable response to imatinib therapy and frequently carries distinctive KIT and/or PDGFRA mutations. The overall prognosis for this subtype is usually better. 3 The less common epithelioid subtype is more common in young females and usually starts in the stomach..

It frequently lacks the distinctive KIT and PDGFRA mutations, in contrast to the spindle cell type. This subtype is linked to a worse response to imatinib therapy and has a propensity to metastasize to lymph nodes, a phenomenon rarely seen in the spindle cell type. 3 IHC is used to diagnose GISTs based on CD117 positivity, which is present in 95% of cases. Size, site, and mitotic index are among the prognostic factors. One Abdominal pain, exhaustion, dysphagia, early satiety, and obstruction are among the symptoms that patients with GISTs may encounter. Additionally, they may exhibit acute gastrointestinal bleeding from erosion of the gastric or bowel mucosa or chronic gastrointestinal bleeding that results in anemia. A potentially fatal intraperitoneal hemorrhage can occur when GISTs burst into the abdominal cavity. 4 About half of GIST patients develop metastases. Metastases most frequently occur in the liver (65%), with the peritoneum coming in second (21%). Bones, lungs, and lymph nodes are thought to be uncommon locations for GIST metastases. 5 The main technique for precisely determining the site, size, location, and extent of GIST is computed tomography (CT). Secondary lesions can be located with the

help of Positron Emission Tomography (PET). However, a histological examination is necessary for the final diagnosis of GIST, and CD117 positivity on IHC is usually required for confirmation. Surgical resection continues to be the mainstay of treatment. Neoadjuvant targeted therapy using tyrosine kinase inhibitors (like imatinib) may be used for patients with locally advanced tumors or metastases. One Imatinib is a selective small molecule inhibitor that targets three receptor tyrosine kinases: PDGFRA, the transmembrane receptor KIT, and the chimeric BCR-ABL fusion oncoprotein linked to chronic myeloid leukemia. Imatinib, formerly known as STI 571, functions by competitively blocking these tyrosine kinases' activity at the enzyme's adenosine triphosphate (ATP)-binding site. 6 Tyrosine phosphorylation in the proteins involved in signal transduction pathways is inhibited as a result of this action. 7 Imatinib neoadjuvant therapy is both practical and advised, with a minimum recommended duration of six months. It has been demonstrated to significantly reduce tumor mass, which in turn reduces operative morbidity. A three-year course of imatinib treatment is advised for patients who have a high risk of relapse in the adjuvant setting. 7 Other medications like sunitinib, regorafenib, ripretinib, and avapritinib are used in cases of imatinib resistance or refractoriness. In this case study, a 65-year-old man had symptoms of urinary discomfort, rectal bleeding, and general weakness that were later linked to an uncommon tumor. A later diagnosis identified the tumor as a GIST that simultaneously affected the fundus of the bladder and the ileum. The purpose of this case study is to persuade other surgeons to be more watchful when handling atypical small bowel GIST presentations that infiltrate other organ systems. In order to guaranty successful surgical outcomes in such unusual cases, it also seeks to emphasize the crucial role that genetic mutation studies play in managing GISTs.

CASE REPORT

A 65-year-old man who works as a farmer came to our tertiary care hospital's medicine department's outpatient ward complaining of generalised weakness, poor appetite, and exhaustion for the past two months. He was receiving treatment for anaemia. Upon presentation, the patient's vital signs included a heart rate of 92 beats per minute, blood pressure of 120/90 millimetres of mercury, oxygen saturation of 98% on room air, and a haemoglobin level of 7, which is indicative of microcytic hypochromic anaemia. His total leukocyte count (TLC) and platelet counts were normal. The remaining panel of pertinent urine and blood tests showed no obvious pathology and were within normal ranges. The patient was sent to the surgical department due to symptoms of minor dysuria, frequent urination, blood in stools, and abdominal discomfort. A large (14 x 10 x 10 cm) solid mass lesion in the lower infraumbilical region compressing the ileal loops with obscured fat planes between the lesion and bladder was discovered by additional radiological investigations using ultrasound sonography (USG) and contrast-enhanced computerised tomography (CECT). The preoperative CT guided biopsy revealed a mesenchymal tumour with myxoid stroma, and the IHC report suggested CD117, DOG1, CD34, and Desmin positivity, confirming the diagnosis of GIST. The patient received neoadjuvant Imatinib targeted therapy for six months, and was instructed to follow up after therapy was finished. The patient was scheduled for surgical resection since further imaging revealed that the tumor's infiltration of the bladder persisted after treatment.

Following the necessary preoperative optimisation, the patient was scheduled for an elective exploratory laparotomy. During the procedure, a 14 x 10 x 10 cm tumour that originated from the ileum's antimesenteric surface and involved the bladder's fundus was discovered.

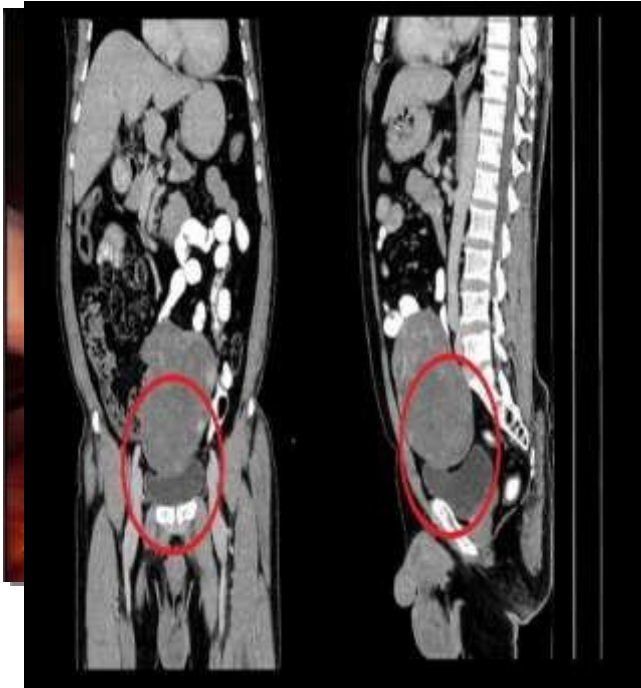


Figure 1: CECT abdomen + pelvis films showing involvement of bladder.



Figure 2: Intraoperative image showing origin of the tumour from antimesenteric border of small bowel loops (ileum).

Removal of the tumour with wedge resection of the involved wall of ileal loop and excision of the tumour surface involving fundus of bladder was done. The ileal defect was closed primarily and primary closure of the defect in urinary bladder was done in two layers. The patient was vitally stable throughout procedure and tolerated the resection and primary repair well.

Figure 3: Intraoperative picture post excision of specimen showing a defect in Urinary bladder with visible bulb of foleys catheter.

The histopathological examination of the excised specimen, measuring 14×10×10 cm and weighing 650 gm, revealed a circumscribed, solid, lobulated mass with a smooth, grey white pseudo capsule covered with blood vessels and adipose tissue interspersed at places. The specimen exhibited two raw areas at the sites of resection: 1) a 3×2.5 cm tubulonodular region, likely of intestinal origin, and 2) a 6.5×5.5 cm irregular roughened surface located over the inferior convexity of the tumor.



Figure 4: Excised specimen of the tumour showing ileal and bladder attachments.

The lymph node biopsy sent from the mesentery of involved bowel loop showed reactive sinus histiocytosis, no evidence of lymphoreticular spread of malignancy. The microscopic findings were consistent with GIST arising from small intestine probably ileal in origin with no mucosal invasion, no tumour necrosis, abnormal mitoses or vascular invasion.

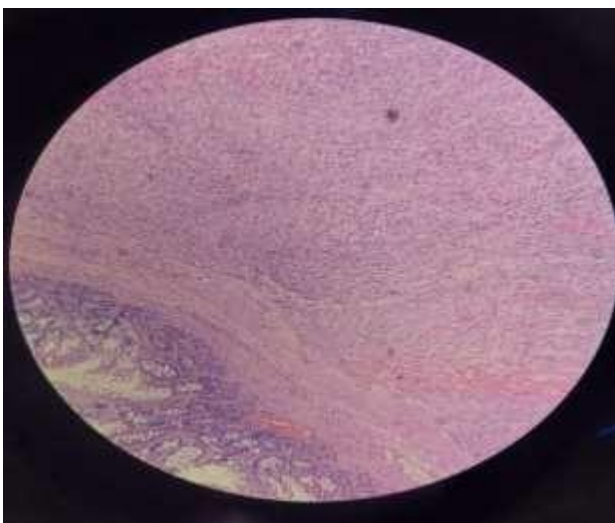


Figure 5: Histopathological slide of the tumour specimen showing normal small intestinal mucosa with a submucosal well circumscribed mass composed of spindle cells arranged in fascicles, no evidence of tumour invasion into the intestinal mucosa.

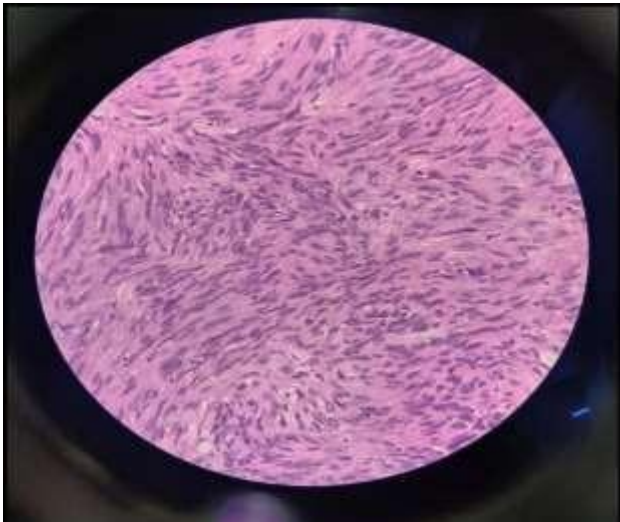


Figure 6: High magnification slide of the excised specimen showing no evidence of marked atypia, tumour necrosis or abnormal mitosis.

The patient was transferred to the intensive care unit (ICU) for postoperative hemodynamic stabilization and later moved to the general ward. He showed a rapid recovery in the general ward and was discharged thereafter. Currently, the patient is undergoing adjuvant Imatinib targeted therapy for one year, as recommended by oncologists. There has been no recurrence of symptoms postoperatively, and the patient's condition remains stable.

DISCUSSION

We found relatively few examples of GISTs infiltrating the bladder while reviewing the literature for our research. Adhesion to nearby organs may be often seen, even though GISTs seldom infiltrate tissues other than the gastrointestinal system. Although uncommon, instances of small intestinal GISTs involving the bladder in the form of compression, isolated involvement, or delayed invasion have been reported in cases of massive GISTs, recurrent GISTs, syndromic GISTs, and metastatic GISTs.^{5,8,9} A secondary cancer that affects the bladder may either directly extend from an advanced tumour of the distal gastrointestinal tract or adjacent pelvic organs, or it can metastasise. Two Rare instances of metastatic GISTs entering the bladder have also been reported, according to literature. The bladder is known to include metastases of primaries originating in the breast, stomach, and lung.⁹ Urinary symptoms may not always be evident when tiny intestine GISTs invade the bladder, making diagnosis difficult.

Tumours originating from intraabdominal tissues other than the stomach or intestinal wall that imitate the GISTs' IHC positivity are known as EGISTs (extra intestinal GISTs).^{Two} We refer to our case as invasion of the bladder by a small intestinal (ileal) GIST since the tumour behaviour, genetics, and prognosis of EGISTs itself are yet unknown. It would be contentious to classify our case as extra intestinal GIST. A solid lesion originating from ileal loops adhering to the bladder was identified in our instance by preoperative imaging and assessment. Following biopsy confirmation that the cancer was GIST, a trial of neoadjuvant targeted treatment was administered with the goal of achieving tumour reduction in order to minimise organ mutilation following removal. The gold standard for early tumour response to molecular targeted treatment or neoadjuvant therapy, according to guidelines, is FDG-PET-CT.⁵ A post-therapy CECT imaging was carried out since FDG-PET scans were not available at our facility, and it revealed no decrease in the tumour involvement of the bladder. The tumour material was not available at our facility, therefore mutational research could not be carried out.

Because mutations in KIT and PDGFRA might confirm the diagnosis when it is unclear, mutational studies can be essential in the diagnosis of GIST. KIT exon 9 mutant GISTs of the small intestine and colon seem to be more aggressive than tumours with a KIT exon 11 mutation, according to mutational investigations of GISTs.^{4,5} To forecast the likelihood of aggressive clinical behaviour in primary GISTs, a number of risk stratification indicators have been put forward, such as the National Institutes of Health (NIH) consensus criteria (Fletcher criteria), the

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modified .The Armed Forces Institute of Pathology criteria (Miettinen criteria) and the NIH consensus criteria (Joensuu criteria). These criteria include things like tumour rupture, mitotic count per high-power fields (HPFs), and tumour size and location.⁵

CONCLUSION

We chose elective laparotomy surgery as the next best course of action for the patient in order to overcome our center's lack of sophisticated analytical equipment. The cancer was removed by wedge resection of the affected ileal loop wall and excision of the tumour surface contacting the bladder fundus. This was followed by primary closure of the ileal loop defect and double-layered repair of the urinary bladder defect. Unfavourable genetic changes that may have been found by mutational tests may be the cause of the tumor's severe adhesions to the bladder wall and the lack of shrinkage during neoadjuvant treatment. We are happy with this case's successful surgical result since our patient tolerated the treatment well and is now leading a symptom-free, healthy life after the procedure.

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