

Gastrointestinal Stromal Tumour Masquerading as a Gynaecological Neoplasm

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Abstract

Gastrointestinal stromal tumours (GISTs) are mesenchymal tumours commonly found in the gastrointestinal tract, particularly in the stomach, but they can mimic gynaecological tumours. We report the case of a 56-year-old postmenopausal woman who presented with a right adnexal pelvic mass. Computed tomography (CT) revealed a large, lobulated pelvic mass with liver lesions. Panendoscopy showed normal findings, and her tumour markers were within normal limits. Based on these findings, the gynaecological oncology team proceeded with a laparotomy and tumour debulking, diagnosing the pelvic mass as likely ovarian in origin. Intraoperatively, the mass was found to originate from the stomach and extend posteriorly to the transverse colon. An en-bloc resection with primary anastomosis was performed and histopathology confirmed a high-risk GIST. The patient made a good post-operative recovery and was scheduled for adjuvant chemotherapy at her follow-up visit. This case highlights the importance of considering GIST as a differential diagnosis in postmenopausal women presenting with an abdominopelvic mass that may mimic an ovarian tumour.

Keywords: Stomach neoplasm; Interstitial cells of Cajal; KIT mutation; CD34 antigen; Ovarian neoplasm

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INTRODUCTION

Gastrointestinal stromal tumours (GISTs) are the most common mesenchymal tumours of the gastrointestinal tract, although they are relatively uncommon compared

with epithelial gastrointestinal neoplasms.^{1,2,3} Specific national incidence data for GISTs in Malaysia are limited; however, the condition is increasingly recognised

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owing to advances in diagnostic modalities and greater public awareness of healthcare services. GISTs are believed to originate from the interstitial cells of Cajal, the ‘pacemaker’ cells of the gastrointestinal tract.^{1,2} As mesenchymal tumours, GISTs are typically composed of spindle cells. When arising in unusual locations such as the pelvis, these tumours may mimic gynaecological malignancies and lead to misdiagnosis as uterine or ovarian cancer. We report a rare presentation in a postmenopausal woman who initially presented with an adnexal mass suspicious for ovarian malignancy. Intraoperatively, however, the mass was found to originate from the stomach and extend inferiorly into the pelvis, thereby masquerading as a pelvic organ tumour.

CASE REPORT

A 56-year-old postmenopausal woman presented with a four-month history of abdominal swelling and discomfort. She also reported constitutional symptoms, including weight loss and loss of appetite. However, there was no change in bowel habits, and she did not experience symptoms suggestive of bowel obstruction. She had no significant past medical or surgical history, nor any family history of malignancy.

She was referred to the gynaecological team for further assessment. On examination, a firm-to-hard, mobile mass measuring 12 × 8 cm was palpable in the right lower abdomen. Pelvic examination revealed a firm, mobile right adnexal mass. Digital rectal examination identified an extraluminal mass located approximately 4 cm from the anal verge. Examination of the other systems was unremarkable.

Initial abdominal ultrasonography revealed a 12 × 8 cm right adnexal mass with a solid-cystic appearance and irregular borders. Tumour markers, including CA-

125, carcinoembryonic antigen (CEA), lactate dehydrogenase (LDH), and alpha-fetoprotein (AFP), were within normal limits. Subsequent computed tomography (CT) of the abdomen and pelvis demonstrated a large, heterogeneous, lobulated solid-cystic pelvic mass with areas of necrosis and calcification, likely ovarian in origin, as well as multiple hepatic lesions suggestive of metastases. Superiorly, the mass demonstrated poor fat-plane delineation with the ileum, transverse colon, and greater curvature of the stomach, while posteriorly, there was loss of the fat plane with the uterus and right broad ligament (**Figure 1a, b, and c**).

Both oesophagogastroduodenoscopy and colonoscopy revealed no abnormal findings. Based on the clinical and radiological findings, a presumptive diagnosis of pelvic malignancy, likely ovarian in origin, was made. Following multidisciplinary discussion, a decision was made to proceed with exploratory laparotomy and tumour debulking surgery.

Surgery was performed one week later. Intraoperatively, a large exophytic tumour arising from the greater curvature of the stomach was identified, with dense adherence to the transverse colon and its mesentery, but without involvement of the gynaecological organs. Owing to its size and exophytic growth pattern, the tumour had extended inferiorly into the pelvis, resulting in gravitational descent along the mesocolon. The general surgical team was consulted intraoperatively, and an en bloc resection was subsequently performed, comprising of a wedge resection of the gastric wall and segmental resection of the transverse colon, followed by primary colonic anastomosis.

Histopathological examination revealed multiple solid areas within the tumour that were firm to hard in consistency and whitish in colour, with lobulated and

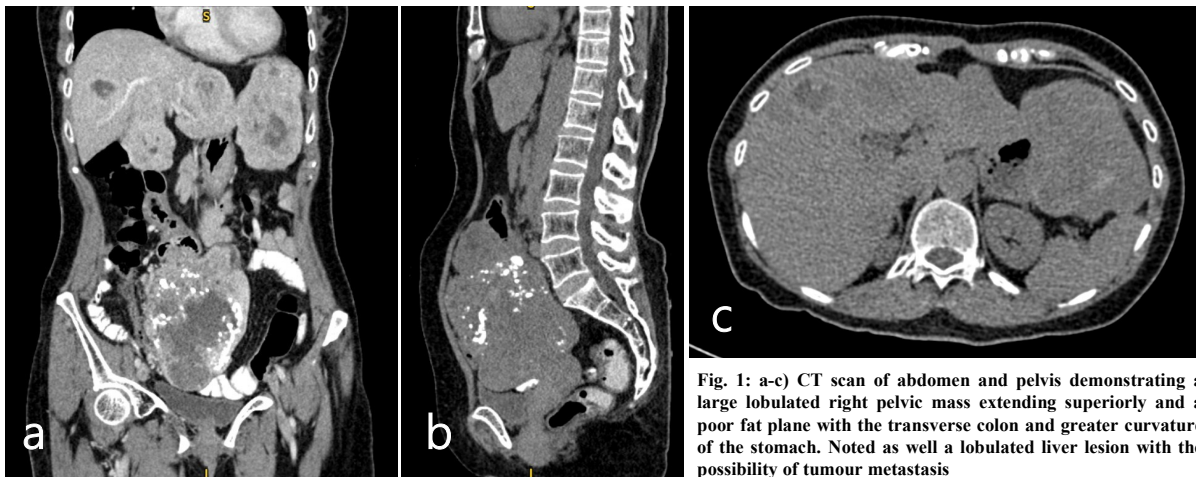


Fig. 1: a-c) CT scan of abdomen and pelvis demonstrating a large lobulated right pelvic mass extending superiorly and a poor fat plane with the transverse colon and greater curvature of the stomach. Noted as well a lobulated liver lesion with the possibility of tumour metastasis

cystic components containing haemorrhagic material. The tumour originated from the gastric wall, demonstrating outward growth with firm adherence to the outer surface of the transverse colon and part of the omentum (**Figure 2**). The mucosa of the stomach and transverse colon, as well as the omentum, appeared intact, with no evidence of mucosal infiltration.

Microscopically, the tumour arose from the muscularis propria of the stomach and extended beyond the serosa into the muscularis propria of the transverse colon. It was composed of spindle-shaped cells, with mitotic activity identified in 2 of 50 high-power fields (HPF) (**Figure 3**). Immunohistochemical staining demonstrated diffuse positivity for CD117 (c-kit) and CD34, while the tumour cells were negative for cytokeratin AE1/AE3 (CKAE1/3), smooth muscle actin (SMA), S-100 protein, desmin, Melan A, and HMB45 (**Figure 3**).

Postoperatively, the patient was discharged on postoperative day 7 and reviewed in the surgical clinic three weeks later. She was counselled regarding the histopathological diagnosis and the need for oncological referral and consideration of adjuvant therapy. However, she was subsequently lost to follow-up, and no adjuvant treatment was initiated. Consequently, interval imaging surveillance could not be performed.

DISCUSSION

GISTs are the most common mesenchymal tumours of

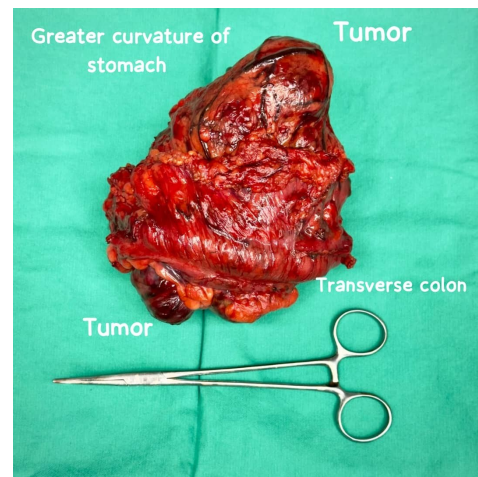


Fig. 2: En-bloc resection of the specimen; a hypervascular lobulated mass 15cm arising from the greater curvature of stomach and attached firmly posterior to the transverse colon and part of the omentum.

the gastrointestinal tract, although they remain relatively uncommon compared with epithelial gastrointestinal malignancies.¹⁻³ GISTs most frequently arise in the stomach compared with other sites in the gastrointestinal tract and originate from the interstitial cells of Cajal.^{1,2} In Malaysia, published data on GISTs are limited, with most available literature consisting of case reports and retrospective series. GIST is often regarded as a “silent” malignancy, typically diagnosed at a median age of around 60 years, with a reported predominance in males and in the Chinese ethnic group.^{1,4-6}

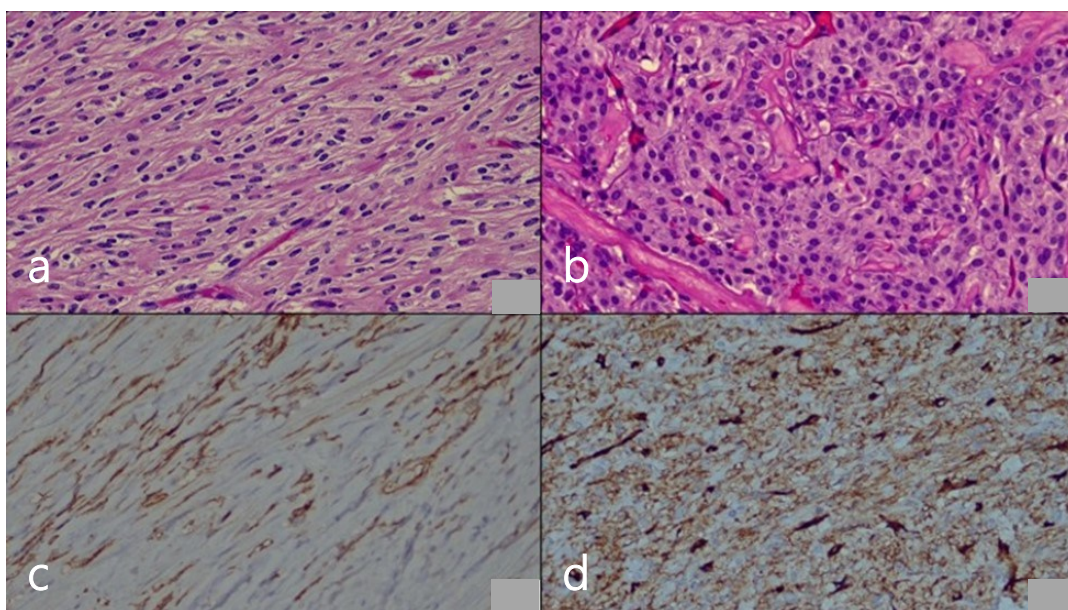


Figure 3: The microscopic features show the tumour composed of diffuse spindle cells (A) and other areas; the tumour cells are epithelioid with abundant cytoplasm (B). The nuclei are mildly pleomorphic and hyperchromatic. The tumour cells are positive to CD117 (C) and CD34 (D) (Magnification x400).

Although GISTs are exceedingly rare in children, paediatric cases are clinically and biologically distinct from adult disease. These tumours are frequently associated with syndromic conditions such as Carney–Stratakis syndrome or Carney triad and are commonly characterised by succinate dehydrogenase (SDH) deficiency rather than KIT or platelet-derived growth factor receptor alpha (PDGFRA) mutations. Clinically, paediatric and syndromic GISTs often show gastric predominance, multifocality, a female preponderance, and a variable response to tyrosine kinase inhibitors. These differences highlight the importance of recognising age- and syndrome-specific contexts when evaluating atypical presentations of GISTs.^{1,7}

Pathologically, GISTs commonly harbour mutations in the KIT or PDGFRA genes, resulting in constitutive activation of tyrosine kinase receptors and subsequent uncontrolled cellular proliferation. This molecular understanding has led to the development of targeted therapies, including tyrosine kinase inhibitors directed against KIT, which are used in the treatment of metastatic or unresectable GISTs.^{7,8} Immunohistochemically, gastric GISTs are typically positive for KIT, anoctamin-1 (DOG1), and CD34, with a minority expressing smooth muscle actin.^{7,9} In the present case, the tumour showed diffuse positivity for CD117 (c-KIT) and CD34, findings consistent with GIST.

The clinical presentation of GISTs varies according to tumour location, which can make diagnosis challenging. Although misdiagnosis as a pelvic gynaecological malignancy has been described predominantly in small bowel GISTs, large exophytic gastric tumours may similarly mimic adnexal pathology by extending inferiorly into the pelvis.¹⁰⁻¹⁴ Common clinical manifestations include gastrointestinal bleeding, abdominal pain, bowel obstruction, and/or a palpable abdominal mass. In some cases, GISTs are incidentally detected during routine investigations such as endoscopy or CT imaging.

According to the National Institutes of Health (NIH) consensus criteria for GIST prognosis, risk stratification is primarily based on tumour size and mitotic count. Joensuu et al. further emphasised that accurate risk assessment is essential in the era of adjuvant therapies targeting KIT and PDGFRA. The proposed modified classification incorporates additional prognostic factors, including tumour size, mitotic activity, and tumour site, to better guide selection of patients for adjuvant therapy.¹⁵ Based on these criteria, our patient would be classified as high risk, given the tumour size (>10 cm), mitotic index (<5 per 50 high-power fields),

and gastric origin.

The adnexal presentation in this case was misleading due to the tumour's exophytic growth pattern and inferior extension into the pelvis, resulting in close anatomical proximity to the right adnexa. On cross-sectional imaging, the mass appeared predominantly pelvic, with loss of fat planes involving adjacent structures, thereby mimicking an ovarian malignancy and obscuring its true gastric origin. Such exophytic GISTs tend to displace rather than directly invade surrounding organs, further contributing to diagnostic ambiguity, as reported in previous case series.^{12,14,16}

In addition, the absence of elevation in ovarian tumour markers, including CA-125, CEA, and AFP, should prompt consideration of alternative non-epithelial malignancies. GISTs characteristically do not produce serological tumour marker elevations, and normal tumour markers in the presence of a large solid pelvic mass should raise suspicion for mesenchymal or extragonadal pathology.^{13,15,17}

CONCLUSION

This case demonstrates how a large exophytic gastric GIST can descend into the pelvis and mimic an adnexal malignancy, resulting in diagnostic uncertainty. Recognition of atypical tumour growth patterns, normal tumour marker profiles, and limitations of cross-sectional imaging is essential to prompt consideration of non-epithelial malignancies and to facilitate timely multidisciplinary management.

Take Home Message

- Large exophytic gastric GISTs may extend inferiorly into the pelvis and mimic adnexal or ovarian malignancy.
- Displacement rather than true invasion of adjacent organs can obscure the primary site of origin on cross-sectional imaging.
- Normal ovarian tumour markers (CA-125, CEA, and alpha-fetoprotein) in the presence of a large solid pelvic mass should prompt consideration of non-epithelial or mesenchymal pathology.
- Multidisciplinary evaluation is essential to avoid misdiagnosis and to guide appropriate surgical and oncological management.

Abbreviations

GIST	Gastrointestinal stromal tumour
CEA	Carcinoembryonic antigen
LDH	Lactate dehydrogenase
CT	Computed tomography

SDH	Succinate dehydrogenase
PDGFRA	Platelet-derived growth factor receptor A
NIH	National Institutes of Health

Declarations

The authors declare no conflicts of interest, and no external funding was received for the research, authorship, and/or publication of this article.

Ethical Consideration

Written consent was obtained from all patients for publications of the clinical details and accompanying images.

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