

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 tumour (GIST) is a common mesenchymal tumour of the gastrointestinal tract, yet it is relatively uncommon compared with epithelial

gastrointestinal malignancies.^{1,2,3} Specific national incidence data for GIST tumour in Malaysia are scarce, but it is increasingly recognised due to advances in diagnos-

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tic methods and increased public awareness of the healthcare system. GIST tumours are known to originate from a ‘pacemaker’ cell of the gastrointestinal system, known as interstitial cells of Cajal.^{1,2} As it is mesenchymal in origin, the GIST tumour is composed of spindle cells. If this tumour presents in unusual sites like the pelvic region, it may cause confusion and be misdiagnosed as a gynaecological disease such as uterine or ovarian cancer. We report a case of an unusual presentation in a postmenopausal woman who presented as adnexal mass consistent with ovarian cancer. However, intra-operatively, the mass was found to originate from the stomach and to have grown downward to the pelvic organs, masquerading as a pelvic organ mass.

CASE REPORT

A 56-year-old postmenopausal woman presented with abdominal swelling and discomfort of four months duration. She also experienced constitutional symptoms, including weight loss and loss of appetite. However, there were no changes in her bowel habits, nor did she have any symptoms of bowel obstruction. She had no significant past medical or surgical history and no family history of malignancy.

She was seen by the gynaecological team for further assessment. Upon examination, a firm-to-hard, mobile mass measuring 12 cm x 8 cm was palpated in the right lower abdomen. Pelvic examination revealed a firm and mobile right adnexal mass. A digital rectal examination detected an extraluminal mass 4 cm from the anal verge. Examination of the other system was unremarkable.

Initial abdominal ultrasound revealed a 12 cm x 8 cm right adnexal mass with a solid-cystic appearance and irregular borders. Her tumour markers, CA 125,

carcinoembryonic antigen (CEA), lactate dehydrogenase (LDH), and alpha-fetoprotein, were unremarkable. A subsequent computed tomography (CT) scan of the abdomen and pelvis showed a heterogeneous, large, lobulated solid-cystic pelvic mass with necrotic area and calcification, possibly ovarian in origin, with multiple liver lesions suggestive of metastasis. Superiorly, the mass also appeared to have poor fat plane delineation with the ileum, transverse colon, and greater curvature of the stomach, while posteriorly, poor fat plane delineation with the uterus and right broad ligament (**Figure 1a, b, and c**).

Both oesophagogastroduodenoscopy and colonoscopy revealed normal findings. Based on the imaging and clinical findings, a presumptive diagnosis of a pelvic malignancy, likely ovarian in origin was made. After a multidisciplinary discussion, a decision was made to proceed with exploratory laparotomy and tumour debulking.

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 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, consisting of 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, firm to hard in consistency, whitish in colour, with lobulated and cystic areas

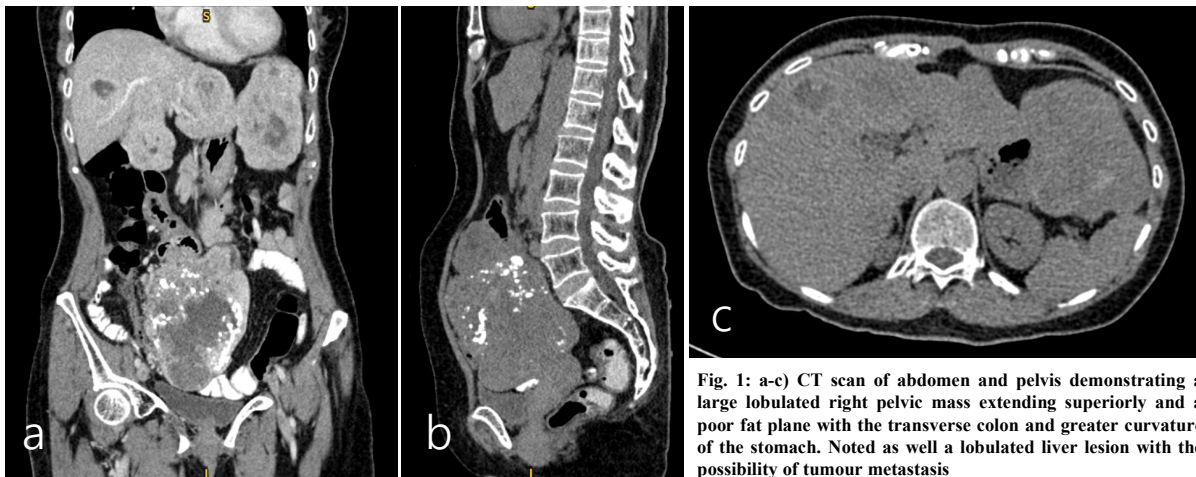


Fig. 1: a-c) CT scan of abdomen and pelvis demonstrating a large lobulated right pelvic mass extending superiorly and a poor fat plane with the transverse colon and greater curvature of the stomach. Noted as well a lobulated liver lesion with the possibility of tumour metastasis

containing haemorrhagic material. The tumour had originated from the gastric wall, growing outward, and attaching firmly to the outer surface of the transverse colon and part of the omentum (**Figure 2**). The mucosa of the stomach, transverse colon, and omentum appeared intact, with no infiltration observed.

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 consisted of spindle-shaped cells, with mitosis observed in 2 out of 50 high-power fields (HPF) (**Figure 3**). Immunohistochemical staining showed the tumour cells were diffusely positive for CD117 (c-kit) and CD34, but negative for Cytokeratin AE1 & AE3 (CKAE1/3), Smooth Muscle Actin (SMA), S-100 Protein (S100), Desmin, Melan A and HMB45 (**Figure 3**).

Postoperatively, the patient was discharged on post-operative 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 adjuvant therapy. However, she subsequently failed to follow up, and no adjuvant treatment was initiated. Consequently, interval imaging surveillance could not be performed.

DISCUSSION

GIST is a common mesenchymal tumour of the gastrointestinal tract, yet it is relatively uncommon compared with epithelial gastrointestinal malignancies.^{1,2,3} GISTs

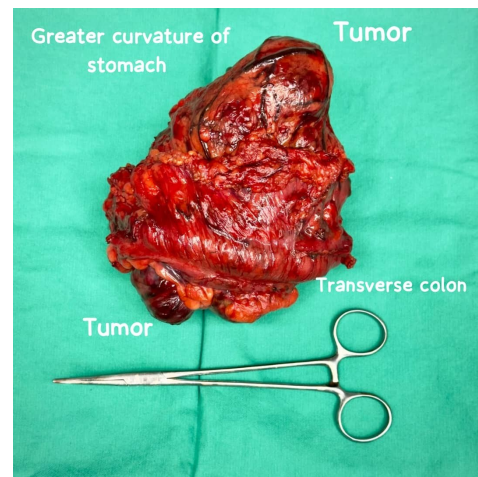


Fig. 2: En-bloc resection of the specimen; a hypervascular lobulated mass 15cm arising from the greater curvature of stomach and attached firmly posterior to the transverse colon and part of the omentum.

most frequently occur in the stomach compared to other parts of the gastrointestinal tract and originate from interstitial cells of Cajal.^{1,2} In Malaysia, studies on GIST tumours are not many, and most are case reports or retrospective studies. It is considered a silent cancer, with peak diagnosis typically around the age of 60, predominantly in males and commonly in the Chinese race.^{1,4-6} Although GISTs are exceedingly rare in children, their significance lies in the fact that paediatric GISTs constitute a biologically and clinically distinct entity compared with adult GISTs. These tumours are

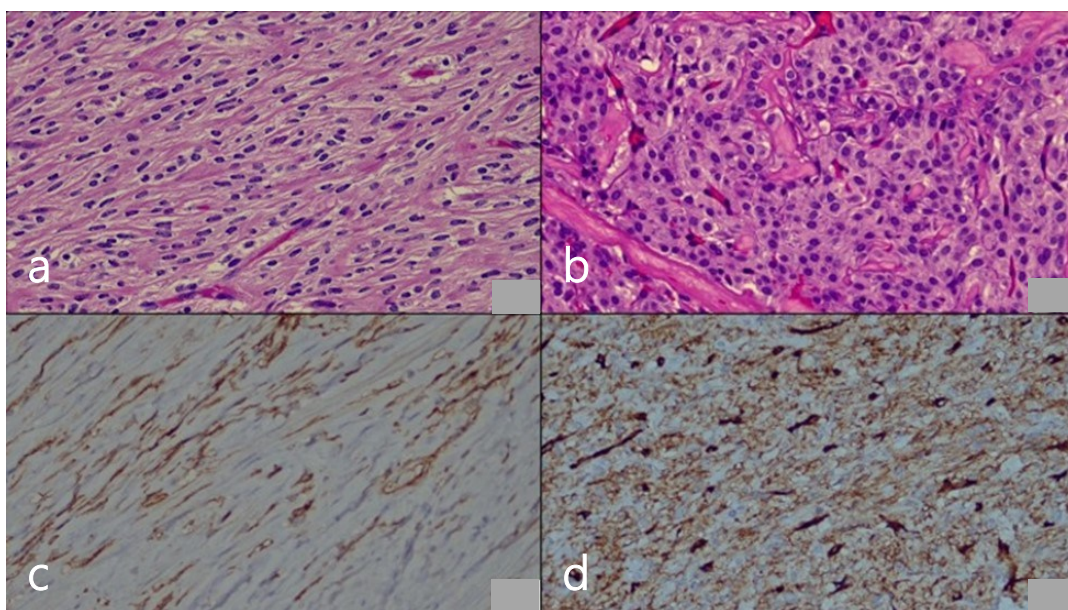


Figure 3: The microscopic features show the tumour composed of diffuse spindle cells (A) and other areas; the tumour cells are epithelioid with abundant cytoplasm (B). The nuclei are mildly pleomorphic and hyperchromatic. The tumour cells are positive to CD117 (C) and CD34 (D) (Magnification x400).

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 A (PDGFRA) mutations. Clinically, paediatric and syndromic GISTs tend to demonstrate gastric predominance, multifocality, female preponderance, and a variable response to tyrosine kinase inhibitors, underscoring the importance of recognising age and syndrome-specific contexts when evaluating atypical GIST presentations.^{1,7}

Pathologically, GISTs commonly have mutations in the KIT or PDGFRA genes, leading to activation of tyrosine kinase receptors and subsequent uncontrolled cell growth. This discovery has led to the development of targeted therapies, such as KIT tyrosine kinase inhibitors, which are used to treat metastatic or unresectable GISTs.^{7,8} Immunohistochemically, gastric GISTs are typically positive for KIT, anoctamin 1, and CD34, with a minority expressing smooth muscle actin.^{7,9} In our patient's case, the tumour was diffusely positive for CD117 (c-KIT) and CD34, consistent with the diagnosis of GIST.

The clinical presentation of GIST varies depending on its location, which can make diagnosis challenging. While misdiagnosis as a pelvic gynaecological malignancy has been described predominantly in small bowel GIST, gastric tumours may similarly mimic adnexal pathology when large and exophytic, allowing inferior extension into the pelvis.¹⁰⁻¹⁴ Common symptoms include gastrointestinal bleeding, abdominal pain, bowel obstruction, or the presence of an abdominal mass. In some instances, GISTs may be discovered incidentally during routine procedures such as endoscopy or CT scans.

According to the National Institutes of Health (NIH) consensus on GIST prognosis, risk stratification is based on tumour size and mitotic count. Joensuu *et al.* emphasised that accurate risk stratification is crucial with the advent of adjuvant therapies targeting KIT and PDGFRA. The study proposed modifying the classification to include factors such as tumour size, mitotic count, and site of origin to select patients for adjuvant therapy.¹⁵ Based on this classification, our patient would be considered high risk, given the tumour size (>10 cm), mitotic index (<5 per high-power field), 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 gastric GIST tend to displace rather than directly invade surrounding organs, further contributing to diagnostic ambiguity as reported in some case reports.^{12,14,16} In addition, the absence of elevation in ovarian tumour markers, including CA-125, CEA, and alpha-fetoprotein, should prompt consideration of alternative non-epithelial malignancies. Gastrointestinal stromal tumours characteristically lack serological tumour marker elevation, 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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