Gastrointestinal stromal tumour: presenting as an ovarian cystadenoma

Sneha Jawalkar , Rachana Karajagi, Surekha Ulhas Arakeri

Pathology, BLDE(Deemed to be University) Shri B M Patil Medical college, Hospital and Research centre, Vijayapura, Karnataka, India

Correspondence to
Dr Sneha Jawalkar, Pathology, BLDE(Deemed to be University) Shri B M Patil Medical college, Hospital and Research centre, Vijayapura, Karnataka, India; sneha.jawalkar@bldedu.ac.in

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SUMMARY

A gastrointestinal stromal tumortumour (GIST) is an uncommon gastrointestinal neoplasm that can arise from any part of the gastrointestinal tract. They can rarely present as a pelvic mass, which might result in a gynaecological condition being misdiagnosed in a female patient. A woman in her early 70s presented with a huge pelvic mass. Abdomen-pelvis CT scan showed a significant cystic mass in the left-sided pelvis with a mass effect on adjacent structures, which suggested a possibility of an ovarian cystadenoma. Her CA-125 was normal. She underwent an exploratory laparotomy with pelvic mass excision. A diagnosis of a gastrointestinal stromal tumour (GIST) arising from the ileum was made on a histopathology study.

BACKGROUND

Gastrointestinal stromal tumours (GISTs) are frequent mesenchymal tumours of the gastrointestinal (GI) tract, which evolve from the so-called 'pacemaker' of the digestive system—the interstitial cells of Cajal.[1 2] Men in their middle years tend to have them the most.[2] In 40%–60% of cases, they are in the stomach, 25%–30% in the jejunum/ileum, 5% in the duodenum and 5%–15% in the colorectum.[2 3] Less than 5% of GIST develops outside the GI tract, including in the pharynx, mediastinum omentum, mesentery, liver, gall bladder, pancreas, retroperitoneum, abdominal wall and peri-vesical tissue.[4–6] Inadvertently discovered during surgery or radiological examinations, GISTs are frequently identified as isolated lesions that might vary from less than 1 cm to 30 cm in size.[2 3] Small-bowel GISTs are more frequently diagnosed when they are large and present with complications such as bowel obstruction, viscus perforation, bleeding and ulceration.[1 2 5] They usually present as an abdominal mass and less commonly present as a pelvic mass that might mimic an ovarian adnexal tumour radiologically, especially when they compress atrophic adnexal structures in an elderly woman. In this article, we present a case of small intestinal GIST extending to the pelvis, which was diagnosed as an ovarian cystadenoma clinically.

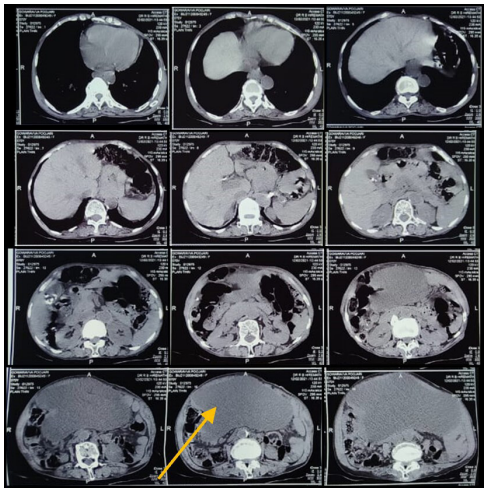

Figure 1 CT scan of the abdomen-pelvis showing a large cystic mass with multiple septations compressing the adjacent structures.

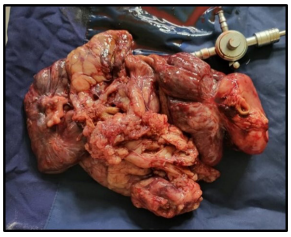

Figure 2 Postoperative gross appearance showing mass adhering to the ileum.

CASE PRESENTATION

A woman in her early 70s came to the hospital with reports of uniform abdominal distention, which was insidious in onset, and gradually progressive for over a year along with acute pain in the abdomen, constipation, and difficulty in micturition for 6 months.

A non-tender, slightly mobile, firm mass was noted on palpation in the left iliac fossa.

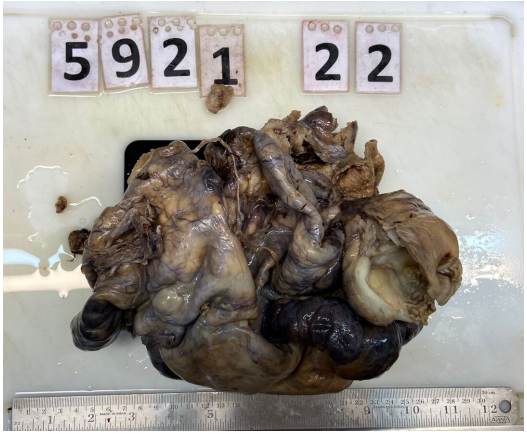

Figure 3 Gross appearance after the surgery. The tumour originated from the ileum.

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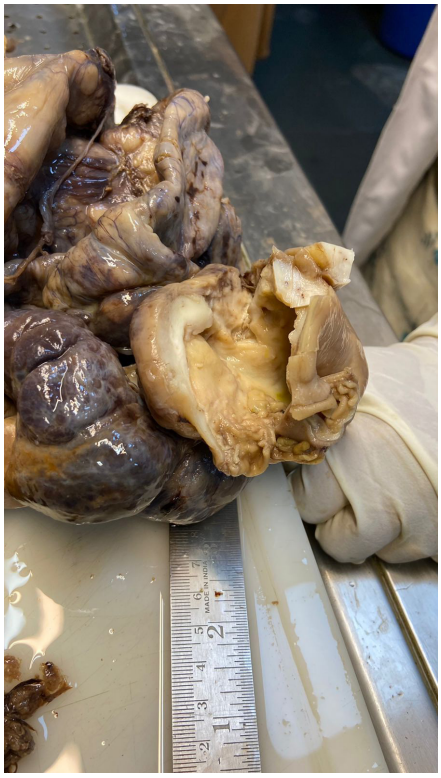

Figure 4 Part of the small intestine showing thick wall and loss of mucosal fold.

INVESTIGATIONS

An abdominopelvic ultrasound and CT scan showed a massive cystic mass measuring 23.8×21×20 cm in the pelvis extending into the abdomen (figure 1). The cyst showed an irregular wall with multiple septations with significant mass effect on the uterus and urinary bladder, displacing the bowel loops laterally. The uterus was seen in postmenopausal status. Left adnexal structures were not visualised and an impression of left ovarian cystadenoma was given. The patient's preoperative serum CA 125 was within normal limits done for three times in 6 months. Only the plain CT and USG have been considered to check the location and size of the mass.

TREATMENT

The patient underwent subsequent exploratory laparotomy with a dose of presurgical antimicrobial prophylaxis using cephalosporins and metronidazole. Intraoperatively, a ruptured

Figure 5 Cut open of cystic mass: inner surface showed multiple friable tissues, grey-white to grey-black and few solid areas noted.

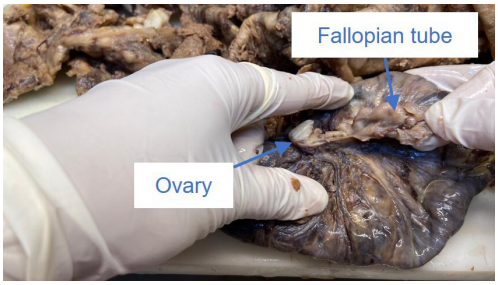

Figure 6 Left side adnexa was adherent to the mass. The ovary measured 1×1×0.5 cm and the fallopian tube measured 3 cm in length.

cystic and lobulated tumour (figure 2) which was adherent to the uterus and ileum was noted. The lesion was shaved from the uterus and excised along with resection and anastomosis of about 10 cm ileum.

The left adnexa was not visualised. The uterus and right adnexa did not exhibit any abnormalities and hence were retained. The mass was excised with resection of a small adherent ileal segment and sent for histopathology study.

A histopathological study was done, and a large ruptured cystic mass attached to a segment of the ileum was noted macroscopically. The cystic mass measured 20×19×7.5 cm (figure 3) and the ileal segment measured 7.5×7.5×2 cm (figure 4). An irregular, nodular gray-white to the grey-black area was noted on the external surface. On cut open, the cystic mass showed multiple friable tissues and few solid areas (figure 5). On cutting open the intestinal loop, there was the loss of mucosal folds, and the walls appeared thick and were in continuity with the cystic mass. Also, a tortuous tubular structure measuring 3 cm in length and a small white nodular structure measuring 1×1×0.5 cm adhered to the mass (Figure 6)

The microscopic examination showed tumour tissue arising from the submucosa/muscularis propria of the intestine, proliferating internally and pushing the mucosa towards the luminal surface and externally towards the serosal surface (figure 7). Tumour tissue was arranged in interlacing fascicles, sheets, whorled, storiform and palisading patterns. Two types of tumour cells—spindle and epithelioid types were noted (figure 8). Many clear and vacuolated cells were also seen (figure 9).

An occasional mitotic figure of 01per 50 high power field was seen. The tubular structure was identified as a fallopian tube and

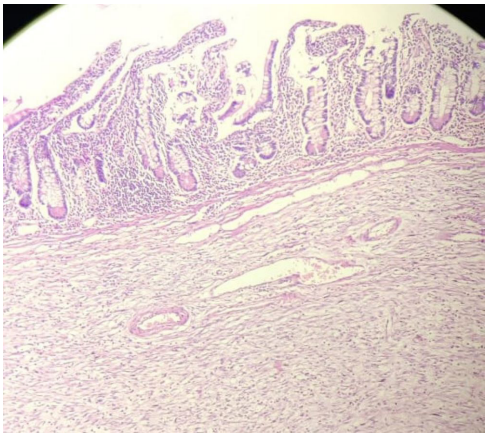

Figure 7 H&E stain, with the magnification of 40×, shows tumour arises from the submucosa/muscularis propria of the intestine and proliferates internally up to mucosa.

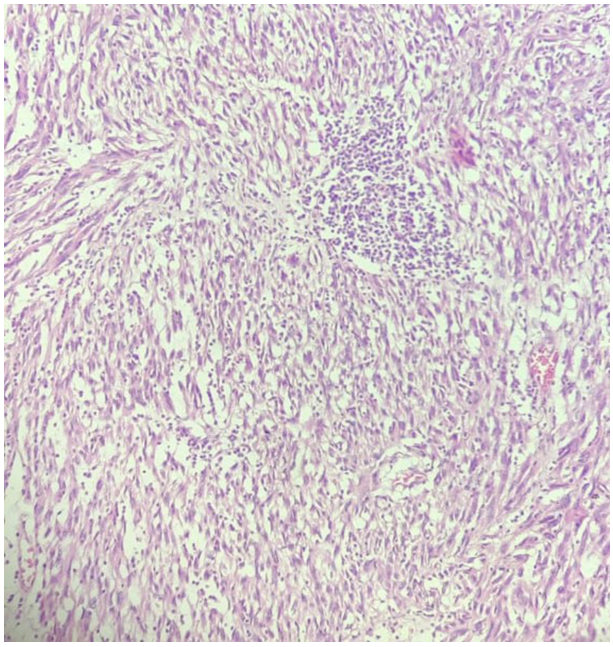

Figure 8 H&E stain, with the magnification of 200×, shows a tumour composed of spindle cells with pale eosinophilic cytoplasm with focal paranuclear vacuolisation.

a small nodular structure was identified as an atrophic ovary on the left side.

A diagnosis of GIST arising from the ileum was rendered on histopathology.

Immunohistochemistry (IHC) confirmed the diagnosis by showing diffuse intense positivity for CD117 (figure 10) and DOG-1 (figure 11).

Ki-67 IHC showed a proliferation index of 5%–6%.

So considered, the site (Ileum), size of more than 10 cm and proliferation index of 5%, the tumour was categorised into category 3b with a 50% chance of the disease progression to malignancy according to the prognostic criteria of resected GISTs by Armed Forces Institute of Pathology (AFIP).

Postoperatively, the patient is doing well, without any reports until. She is on adjuvant therapy with imatinib 400 mg/day once for up to 3 years.

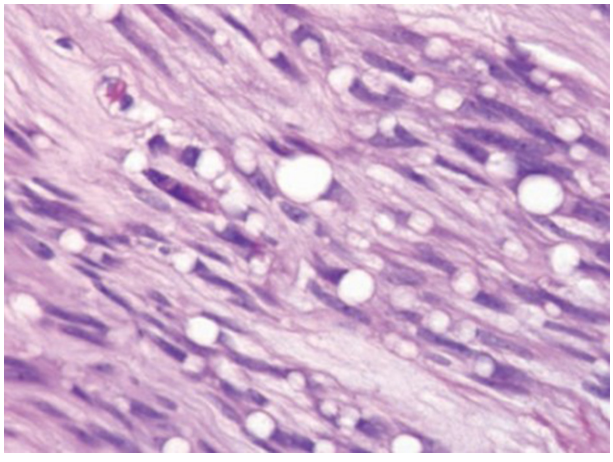

Figure 9 H&E stain, with the magnification of 400×, shows a tumour predominantly composed of spindle cells with intracytoplasmic vacuoles and moderate nuclear pleomorphism.

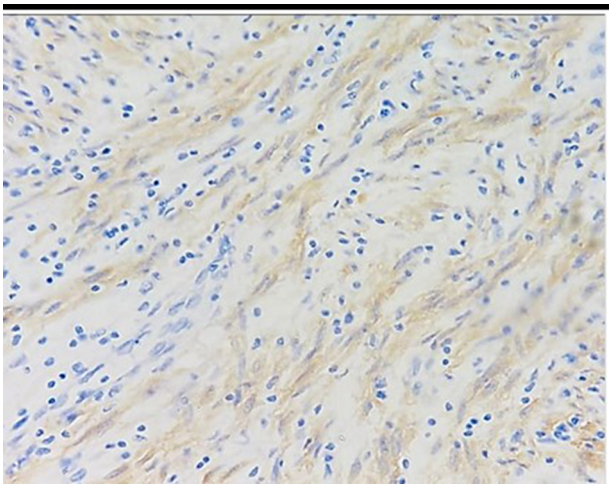

Figure 10 Section with the magnification of 200× shows CD117 strong cytoplasmic and membranous positivity.

DIFFERENTIAL DIAGNOSIS

The histopathological differential diagnosis includes tumours with neural differentiation, such as gastric schwannomas, tumours with fibrous differentiation, such as fibromatosis, tumours with smooth muscle differentiation such as leiomyoma and leiomyosarcoma and inflammatory pseudotumor.[3 7–9]

OUTCOME AND FOLLOW-UP

The patient is taking imatinib therapy and is followed up for every 3 months. She is doing well with no additional reports so far.

High-risk GIST requires a long-term follow-up because there is a considerable chance that the disease will return over the first 3 to 5 years, a follow-up programme with regular CT scans is recommended.[5 6 9]

DISCUSSION

The most frequent mesenchymal tumour of the GIT is the GIST, an uncommon neoplasm. Leiomyoma, granular cell tumours and inflammatory fibroid polyps are the other mesenchymal tumours of GIT.[3 7 8]

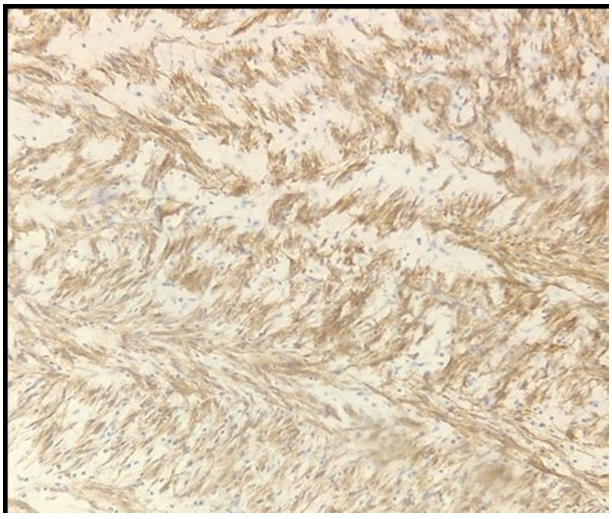

Figure 11 Section with a magnification of 200× shows DOG-1 strong cytoplasmic and membranous positivity.

The number of cases per million people varies between 11 and 15.[6 9] GISTs can develop sporadically at any age, with slightly more occurrence in men than in women.[2] In children and adolescents, the succinate dehydrogenase (SDH) mutant type of GIST occurs mostly in the stomach with a female preponderance.[3 4 6]

It is quite difficult to diagnose GISTs. Imaging examinations include ultrasound, CT scans, MRI, endoscopies with or without endoscopic ultrasonography and biopsies.[6 8] It is possible to misdiagnose GISTs as ovarian tumours since they rarely can present as pelvic masses. The only reliable diagnostic methods for GISTs are biopsy and IHC labelling.[6–8]

Microscopically they are of moderate to high cellularity with the presence of mainly spindle cells. Epithelioid cells, signet ring cells, oncocytic cells and pleomorphic cells with or without myxoid stroma can be seen.[6] The nuclear features of all these cell types are bland and oval with vesicular chromatin. The cytoplasm is pale eosinophilic.[4 6 9]

IHC is an important diagnostic tool for identifying GISTs and determining the appropriate treatment.[6]

CD 117(c-kit) and DOG 1 are the most used markers for GIST diagnosis.[1 6] However, in some cases, these markers may not be sufficient for a definitive diagnosis. In this situation, additional markers such as SDHB, CD 34, smooth muscle actin and S100A4 may be used to help confirm the diagnosis.[1 3 4]

Recently, a study published in the American Journal of Surgical Pathology suggested that the use of IHC for SDHB may help identify patients with a higher risk of recurrence or metastasis.[3 4]

A distinguishing feature of GISTs is positive CD117 IHC staining.[1 3 4 8] A CD117-activating somatic mutation occurs in approximately 95% of GISTs (c-kit).[1 2 6 9] The chromosome 4 proto-oncogene c-kit encodes the transmembrane receptor tyrosine kinase CD117.[3 4 7] Five per cent of GISTs are CD117-negative.[3 5 7 8]

They are essentially benign tumours with a malignant potential based on various prognostic factors.

The AFIP prognostic model is influenced by anatomical location, tumour size, mitotic rate, primary, and the completeness of resection.[6 8]

Poor prognosis indications include a mitotic rate greater than five mitoses per 50 high-power fields and a size greater than 10 cm with moderate to high malignant potential.[3 6 8]

GISTs are typically more aggressive in the small intestine. They metastasise to the liver and peritoneum with the least chance of spreading to lymph nodes.[3 6]

Surgery is the primary treatment option for localised GIST, and it represents the only chance for a cure.[3 8 9] The goal of surgery is to remove the tumour completely, along with any surrounding tissue that may be affected. This can be achieved through various surgical techniques, such as a partial or total resection of the involved intestine, depending on the location of the tumour.[1 6 9]

Antimicrobial prophylaxis becomes important to prevent complications due to surgical site infection. As suggested by Marano *et al*, in our case, we preferred a preoperative single dose of third-generation cephalosporins combined with metronidazole, which was continued for 5 days post-operatively.[10]

In addition to surgery, some patients with localised GIST may also benefit from targeted therapy with drugs such as imatinib mesylate or sunitinib malate.[1 3 4 6] These medications work by targeting specific molecules involved in GIST cells' growth and spread. Either before or after surgery, GIST patients are treated with imatinib (tyrosine-kinase inhibitor).[3 6]

Learning points

- Gastrointestinal stromal tumour (GIST) is often considered a tumour of low malignant potential due to the asymptomatic course of the disease.
- A clinical differential diagnosis of GIST must be considered in abdominopelvic tumours that are adherent to various organs, including the intestine and mimic ovarian tumours on imaging.
- The Armed Forces Institute of Pathology offers knowledge in GIST diagnosis and characterisation, thus enabling clinicians to make therapeutic decisions and determine its prognosis.

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ORCID iDs
Sneha Jawalkar http://orcid.org/0000-0001-7023-3000
Surekha Ulhas Arakeri http://orcid.org/0000-0002-1185-7021

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