

Gastrointestinal Stromal Tumour of the Small Intestine – Case Report

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ABSTRACT

Gastrointestinal stromal tumours (GISTs) are mesenchymal tumours that can be located in any part of the gastrointestinal system. Most often they occur in the stomach and in 20-30% of cases they occur in the small intestine. About 30% of all GISTs are malignant. We report a case of a 54-year-old patient with malignant GIST located in the small intestine and liver. The patient complained of discomfort and heaviness in the abdomen and parts of the abdomen were hard on palpation. Family physician performed an ultrasound of the abdomen which showed a large mass in the right hemiabdomen. CT scan of the abdomen showed a tumour mass of approximately 13x19x20 cm and GIST was suspected. Biopsy was performed and histopathological finding spoke in favour of GIST. GIST resection and Meckel diverticulum resection were performed. Histopathological finding confirmed it was a malignant GIST. After nine months, a control ultrasound showed an oval formation of the liver and excision was performed. The patient was discharged home and histopathological finding confirmed that liver formations were also GIST. GIST should be considered as a possible cause of abdominal difficulties and the treatment of choice is surgery.

Keywords: gastrointestinal stromal tumours, open resection, case report

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INTRODUCTION

Gastrointestinal stromal tumours (GISTs) are the most common mesenchymal tumours of the gastrointestinal tract [1]. The incidence is 10-15 cases per million. In 60% of cases, GISTs occur in the stomach, and in 20-30% of cases in the small intestine. They can rarely be found outside the gastrointestinal system - on the omentum, mesentery and retroperitoneally [2]. About 30% of GIST cases are malignant [3]. They can occur at any age, but most commonly are diagnosed later in life and equally among men and women [4]. The most common metastatic sites are the liver and peritoneum [5]. GISTs are usually asymptomatic but can present as gastrointestinal bleeding; acute as melena and haematochezia or chronic as anaemia. In addition, they can be presented as an abdominal mass that causes abdominal pain, bloating, discomfort, early satiety [6]. GISTs are diagnosed with CT or MR and the final diagnosis is based on a histopathological finding [7]. For resectable tumours larger than 2 cm, the first choice of

treatment is surgery. Imatinib is given preoperatively for tumour reduction and in cases of unresectable tumours [1].

CASE

In the beginning of 2020, a 54-year-old patient came to a family physician for a request to change antihypertensive therapy because he had been noticing erectile dysfunction since taking the medication. In 2018, the patient underwent total replacement of left hip, and a year later he had a pulmonary embolism. He was then diagnosed with genetic thrombophilia (heterozygous for PAI-1 4G / 5G) and was on anticoagulant therapy since. The patient was given a medication for hypertension and was ordered for a check-up in about 20 days. Upon control, he stated that he still had erectile dysfunction, but also a feeling of bloating, discomfort and heaviness in the abdomen. On palpation, part of the umbilical region, hypogastrica and the entire right lateral and right inguinal regions of the abdomen were hard. An ultrasound of the abdomen was performed immediately in the

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Figure 1. Ultrasound of the abdomen in a 54-year-old patient

family medicine office, where a large mass was observed in the right hemiabdomen (**Figure 1**). The patient was urgently referred to a gastroenterologist with suspected expansive tumour formation.

After examination by a gastroenterologist, a CT scan of the abdomen was performed and a tumour mass of approximately 13x19x20 cm was described. According to radiological characteristics GIST was possible, and a puncture of the formation was recommended. The patient was afebrile all the time, without nausea and vomiting. Appetite was good, body weight stationary. Mild anaemia (Hb 115) and elevated CRP (54) had been reported in the laboratory.

Over the next few days, under ultrasound control, a biopsy of the tumour formation in the right iliac region was performed and a histopathological finding was obtained that spoke in favour of GIST. The patient was hospitalized in March and a GIST resection and adjacent small bowel curvature were performed. The histopathological finding of the resected part of the small intestine confirmed that it was a gastrointestinal stromal tumour, malignant. During the stay, the patient received thromboprophylaxis, antibiotic prophylaxis, analgesic and his previous therapy, as well as

parenteral fluid and electrolyte replacement. After discharge, the patient was always in good general condition, afebrile, cardiocirculatory compensated, the abdomen was soft, elastic, palpably painless, regular peristalsis, normal functions and habits.

The findings showed various large, partly necrotic round formations in the IV, V, V / VI, VIII and VIII / VII segments of the liver, and the radiogram of the thoracic organs was orderly. Hospitalization and microwave ablation of formations had been agreed upon. The patient was hospitalized at the end of January and the excision was performed. The early postoperative course was orderly.

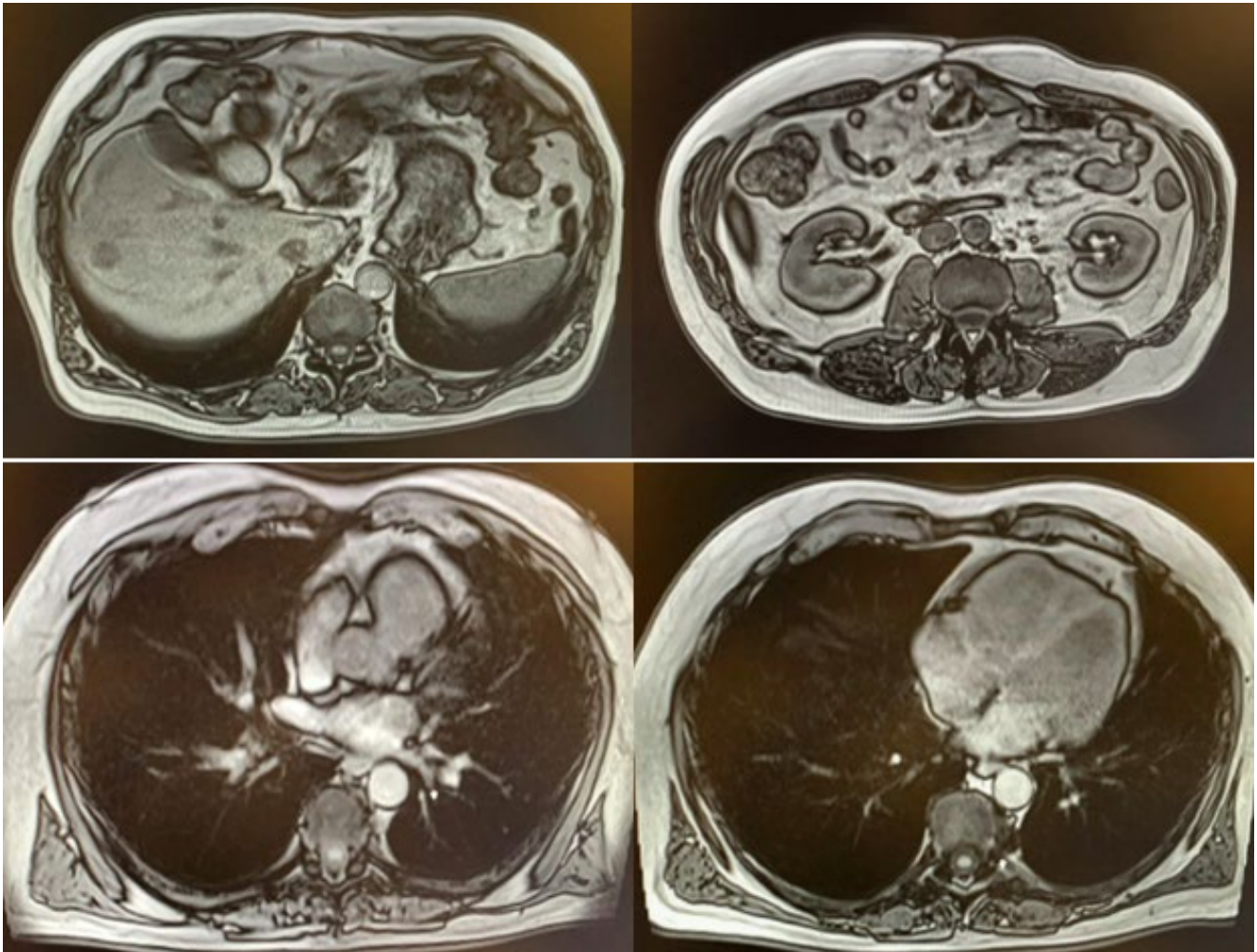


Figure 2. MR of the abdomen and pelvis in a 54-year-old patient

During the stay, the patient received thromboprophylaxis, antibiotic prophylaxis, analgesic, his previous therapy, parenteral fluid and electrolyte replacement. In further course of the stay, fever occurred and antibiotic therapy was changed after the observation. After that, the inflammatory parameters normalized and further course of the stay was without any problems. The patient was discharged home with a recommendation of relative rest and a controlled diet. The pathohistological finding corresponds to a gastrointestinal stromal tumour of malignant behaviour. The patient will be further monitored in the oncology clinic.

DISCUSSION

Stromal or mesenchymal neoplasms affecting the gastrointestinal tract typically present as subepithelial neoplasms. GISTs are neoplasms most often located in the stomach and proximal small intestine, but they can occur in any portion of the alimentary tract and occasionally in the omentum, mesentery and peritoneum [8,9]. Furthermore, in this area of the gastrointestinal tract we can find lipomas, liposarcomas, leiomyomas, true leiomyosarcomas, desmoid tumours, schwannomas, and peripheral nerve sheath tumours but they are less common [10]. All GISTs have the potential for malignant behaviour, even those 2 cm or less

with bland histologic features and consensus classifications focus on stratifying lesions according to the relative risk of recurrence and metastasis [8,11,12,13].

In general, tumours that are larger than 5 cm are lobulated, enhance heterogeneously, and have mesenteric fat infiltration, ulceration, regional lymphadenopathy, or an exophytic growth pattern on CT, are more likely to metastasize [14-16]. In contrast, GISTs with less metastatic potential tend to enhance in a homogeneous pattern and often show an endoluminal growth pattern.

Nodal involvement is rare in GIST and routine lymph node dissection is not indicated unless nodes are clinically enlarged.

GISTs occur throughout the gastrointestinal tract from the oesophagus to the anus. Within the gastrointestinal tract, GISTs are most common in the stomach (40-60%) and jejunum/ileum (25-30%) [17,18]. The duodenum (5%), colorectum (5-15%), and oesophagus ($\leq 1\%$) are less common sites. Tumours lacking any association with the bowel wall have been referred to as extragastrointestinal stromal tumours (EGISTs) and occur in the retroperitoneum, mesentery and omentum [18].

The presentation varies depending on the primary tumour location. In general, the distribution of clinical presentation is as follows: overt or occult gastrointestinal bleeding – 28% (small intestine) and 50% (gastric), incidental finding (asymptomatic) – 13-18%, abdominal pain/discomfort – 8-17%, acute abdomen – 2-14%, asymptomatic abdominal mass – 5% [1].

Paraneoplastic syndromes are rare in GIST however, potential paraneoplastic syndromes have been reported in a few patients, including consumptive hypothyroidism and non-islet cell tumour hypoglycaemia.

GISTs frequently metastasize to the liver and peritoneum, and rarely to regional lymph nodes. They uncommonly metastasize to the lungs, the most common site of metastasis for most soft tissue sarcomas.

About 20-30% of all cases are located in small intestine and 30% of GIST cases are malignant so it can be said that this presented case of malignant GIST located in small intestine is rare [3]. Contrast-enhanced computed tomography (CT) is the imaging method of choice to characterize an abdominal mass, evaluate its extent, and assess the presence or absence of metastatic disease, which most commonly involves the liver, omentum and peritoneal cavity [11].

Although magnetic resonance imaging (MRI) has a comparable diagnostic yield and lacks radiation exposure, CT is a preferred initial imaging study for screening and staging, except perhaps in patients who cannot receive intravenous contrast [19].

MRI may occasionally be preferred for GISTs at specific sites, such as the rectum, especially for evaluating anatomic extent for surgery or to evaluate suspected liver metastases.

The usual CT appearance of a GIST is that of a solid, smoothly contoured mass that enhances brightly with intravenous contrast. Very large tumours (>15 cm) may appear more complex due to necrosis, haemorrhage or degenerating components [11].

Endoscopy may be useful to further characterize the lesion if a gastric mass is identified. Both GISTs and leiomyomas may appear as a submucosal mass with smooth margins, with a normal overlying mucosa, and bulging into the gastric lumen. Central ulceration is occasionally seen. Endoscopy alone cannot accurately distinguish between intramural and extramural tumours [15].

Endosonographically, GISTs are typically hypoechoic, homogeneous lesions with well-defined margins, although they can rarely have irregular margins and ulcerations. Most GISTs originate from within the muscularis propria; small lesions may originate from the muscularis mucosa. Infrequently, the tumours are inhomogeneous, which has been attributed to liquefaction necrosis, connective tissue, and cystic and hyaline degeneration [15].

Preoperative biopsy is not generally recommended for a resectable lesion in which there is a high suspicion for GIST and the patient is otherwise operable. However, a biopsy is preferred to confirm the diagnosis if metastatic disease is suspected or if preoperative imatinib is considered prior to attempted resection in a patient who has a large locally advanced lesion thought to represent a GIST [8].

The differential diagnosis of a subepithelial tumour arising in the gastrointestinal tract is broad and can include GIST, leiomyosarcoma, leiomyoma, malignant melanoma, schwannoma, malignant peripheral nerve sheath tumour, fibromatosis (desmoid tumour), inflammatory myofibroblastic tumour, or even metaplastic (“sarcomatoid”) carcinoma [8].

CONCLUSION

Gastrointestinal stromal tumours (GISTs) should be considered as a possible cause of bloating, discomfort and heaviness in the abdomen. It is necessary to do the diagnostic method as soon as possible. The treatment of choice is surgery.

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