Research and Reports in Gastroenterology

Case Report

A SCITECHNOL JOURNAL

Delayed Diagnosis of a GIST: A Case Report from a Resource-limited Setting

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Received date: March 1, 2020 ; **Accepted date:** March 15, 2020 ; **Published date:** March 22, 2020

Abstract

Gastrointestinal stromal tumours (GISTs) are rare but constitute the most common sub-epithelial tumours of the gastrointestinal (GI) tract. Small GISTs are mostly benign, asymptomatic, and discovered accidentally. However, larger tumours have been reported with a myriad of presentations. The diagnosis of a large malignant gastric GIST in a 35 years old male who presented with recurrent melaena was delayed.

This patient presented to various hospitals with recurrent melaena and later noticed a progressive, painless epigastric swelling. Endoscopy, unnecessarily delayed until he got to the fourth hospital, revealed a large sub-mucosal gastric mass with an overlying oozing ulcer. The patient had laparotomy and partial gastrectomy with complete resection of the tumour. Histology and immunohistochemistry confirmed a malignant GIST that was CD117 positive, S100 positive, and SMA negative. The patient did well postoperatively and was placed on Imatinib for one year.

Failure to do endoscopy in unexplained recurrent melaena can delay GIST diagnosis.

Keywords: GIST

Introduction

GIST is the most common sub-epithelial tumour of the GI tract. It is usually found in the stomach (60%), and small bowel (30%), but colonic (5%), rectal, Oesophageal, and even omental and ovarian locations have been described [1]. The interstitial cell of Cajal in the muscularis propria is the specific cell of origin of this neoplasia. Though GISTs have malignant potentials, only 10-30% is malignant, accounting for 1-3% of all GI cancers [2].

The classical endoscopic appearance of a sub-epithelial GI mass with a normal mucosa should raise an index of suspicion for a GIST. Final diagnosis rests on histology and immunohistochemistry. The finding of CD117 expression differentiates GISTs from other differentials with the same histological appearance, such as leiomyomas, schwannomas, and fibrous tumours [3].

The majority of GISTs are solid tumours with cystic presentations a rare occurrence [4]. GISTs are usually asymptomatic detected incidentally at surgery or by ultrasound or radiological studies. Large GISTs may be symptomatic and commonly present with nausea, abdominal pain, anorexia, and weight loss [5]. Some of the atypical presentations of GISTs reported include extra-gastrointestinal intra-abdominal locations, cystic, multicystic, and mixed tumours, pelvic and ovarian sites, pneumo-peritoneum, and GISTs co-existing with other tumours[6-11].

Melaena stool should raise a high suspicion of GI bleeding and warrant a timely endoscopic request [4]. This case report was a young man who had recurrent melaena and anaemia that necessitated multiple blood transfusions. The diagnosis of GIST was delayed for more than one-year duration until the onset of a progressive epigastric swelling.

Case Report

A 35 years old man presented with a year history of recurrent black stools. There was no history of haematemesis, epigastric pain, nausea, vomiting, or weight loss. No change in bowel habits. He was referred to a retainer hospital where he had a blood transfusion, but no endoscopy was done. On review later, an epigastric mass was noted. The mass appeared uniformly echogenic on ultrasound scan and was not attached to the liver, gall bladder, pancreas, or spleen. No retroperitoneal lymphadenopathy was noted. He was subsequently referred to another hospital for a review where CT abdomen was done, and the findings suggested a retroperitoneal tumour. No endoscopy was done at the second hospital. A second opinion was sought at a third hospital where endoscopy was requested but the hospital had no facilities for endoscopy.

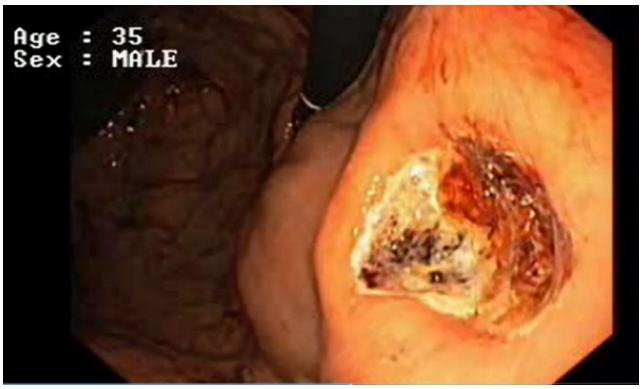

Figure 1: Subepithelial gastric mass with overlying oozing ulcer

The patient was prepared and had laparotomy with partial gastrectomy and complete resection of a large tumour measuring 16 cm by 12 cm by 11 cm arising from lesser curvature 4 cm below the GE junction and not adherent to the liver, gall bladder, or portal vessels. He made satisfactory recovery postoperatively.

Histopathology report confirmed a malignant GIST (mitosis rare) with solid cellular fusiform or spindle cells forming dense fascicles arranged in an interlacing fashion. The cells had carrot-shaped coarse hyperchromatic nuclei. Immunohistochemistry findings were

consistent with a GIST that was cytoplasmic CD117 positive, focal S100 positive and gastric muscle SMA negative.

The postoperative management was uneventful. The patient was discharged on Imatinib 400 mg daily for one year. A repeat CT and PET scan at six months postoperatively were normal.

Discussion

Previous studies have described many atypical presentations, chiefly having to do with locations and nature of the tumour. Extra-gastrointestinal sites are commonly omental or mesenteric with an ovarian location reported by only one reviewed study [7-10]. Most GISTs are solid tumours, but cystic ones have also been reported [1,4,6,7].

The cause of the melaena in GISTs is bleeding vessels from mucosal ulceration caused by pressure necrosis because of the large tumour size 1, 5 In addition to the risk of bleeding; tumour size of GIST is a predictor of symptoms as well as malignant potentials [2,12-14]. Small-sized GISTs are usually asymptomatic and benign, and the larger the tumour, the more likelihood of being symptomatic and malignant [1,13,14]. Interestingly, this large tumour measuring grossly 16 cm ×12 cm × 11 cm at pathological examination did not present with the usual symptoms and had already developed early malignant features (rare mitosis).

The diagnosis of a GIST in the case under review was delayed due to skill gaps and reduced access to endoscopic and other radiological services in developing countries. A timely upper GI endoscopy and accurate interpretation of CT images would have helped in the early diagnosis of this case before a substantial tumour growth and the onset of malignant changes. Prompt tumour resection and commencement of Imatinib would have prevented the development of the anaemia, the need for multiple blood transfusions, and perhaps the malignant progression of the GIST.

This case highlights the need for more training in both radiology and gastroenterology in resource-limited countries. It also demonstrates the beauty of teamwork, collaboration, and the need for prompt referral of cases not adequately managed to higher hospitals and health facilities.

Conclusion

In conclusion, delays in making an endoscopy request, non-availability of endoscopic services, and high cost of imaging are recurrent factors that negatively impact endoscopic access in developing countries. Failure to do an endoscopy (both esophagogastroduodenoscopy and colonoscopy) in unexplained melaena can delay the diagnosis of not only GIST but other GI-related pathologies and malignancies. Therefore, an index of suspicion of a GIST should be high in unexplained GI bleeding. Unexplained GI bleeding should be referred for urgent endoscopy. GISTs should be considered as a differential diagnosis of unexplained GI bleeding. Early diagnosis of GISTs will ensure early treatment and reduce the risk of recurrence and malignant progression.

Acknowledgements

The author wishes to thank all those who participated in the successful management of this case.

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