

Case Report

A rare case of rectal gastrointestinal stromal tumour: diagnosis and management

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ABSTRACT

Gastrointestinal stromal tumours are frequently defined as KIT-(CD117) or PDGFRA-positive mesenchymal spindle cell tumors. Gastrointestinal stromal tumors most commonly occur in the stomach and small intestine, with up to 5% of GISTs occurring in the colon and rectum. We presented a case of distal rectal GIST diagnosed by colonoscopy, colonoscopy with biopsy, computed tomography. The patient underwent Abdominoperineal resection (APR) and was confirmed on histopathology to have rectal GIST with tumor size more than 5 cm and mitotic rate more than 5/50 high power field (HPF). All GISTs are considered to have malignant potential, and, for that reason, all rectal GISTs should be considered for resection.

Keywords: Gastrointestinal stromal tumors, Imatinib, Rectal mass, KIT mutation

INTRODUCTION

Gastrointestinal stromal tumour or GIST was a name given in 1983 to a group of gastrointestinal tumours which were otherwise could not be classified as being smooth muscle or neurogenic origin.³ Gastrointestinal stromal tumors make up only 1% of primary GI cancers and only 0.1% of tumors arising in the rectum. They are frequently defined as KIT-(CD117) or PDGFRA-positive mesenchymal spindle cell tumors. KIT is a tyrosine kinase receptor and an important target in therapy. Gastrointestinal stromal tumors most commonly occur in the stomach (50-60%) and small intestine (30-40%), with up to 5% of GISTs occurring in the colon and rectum.^{1,2} Elderly men in their 70s appear to be at higher risk.

CASE REPORT

A 66 year old female patient was admitted to our institution with complaints of bleeding per rectum since 8 months, complaints of progressive constipation since 5 months, and history of dyspnoea on exertion since 3

months. She had no history of any comorbidities. She was pale with normal vitals and systemic examination. Digital rectal examination revealed hard irregular polypoidal growth 1 cm from anal verge, upper extent of the growth could not be reached, extending from 2 o'clock to 7 o'clock and the growth was not fixed. Her haemoglobin (5.2 g/dl). Peripheral smear showed hypochromic microcytic anaemia. Albumin was marginal. Colonoscopy revealed- large polypoidal ulcerated friable mass lesion seen starting 1 cm proximal anal verge extending up to middle 1/3rd rectum causing mild luminal narrowing. Rest of the colon and rectum was normal. Multiple biopsies were obtained. Which was suggestive of gist with high risk of malignancy.

Contrast enhanced computed tomography of abdomen showed asymmetric short segment heterogeneously enhancing wall thickening for approximate length of 7 cm with irregular polypoidal mass lesion of approximate size 7×4×7 cm arising from anterior wall of rectum. Mass was protruding into lumen causing partial luminal occlusion, approximately 1 cm from anal verge and extending up to

rectosigmoid junction with non-enhancing areas of necrosis.

After optimization of patient, patient was operated and abdominal perineal resection with permanent colostomy was done.

Post-operative days were uneventful. Patient was started on oral liquid on post-operative day 1 and was on full. Oral diet since day 3. Abdominal drain and foleys catheter were removed on day 3. Patient was discharged on day 5, after explaining stoma care. After consultation and referral to medical oncologist, patient has been started on imatinib therapy.

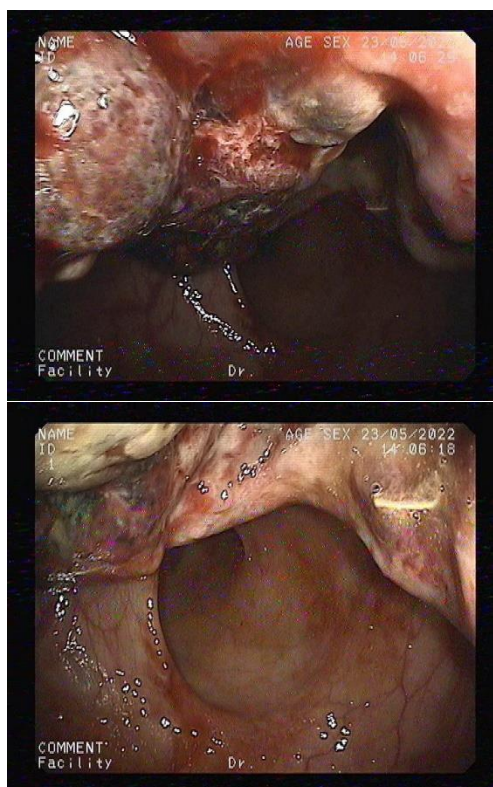


Figure 1: Colonoscopy image showing polypoidal growth in rectum.

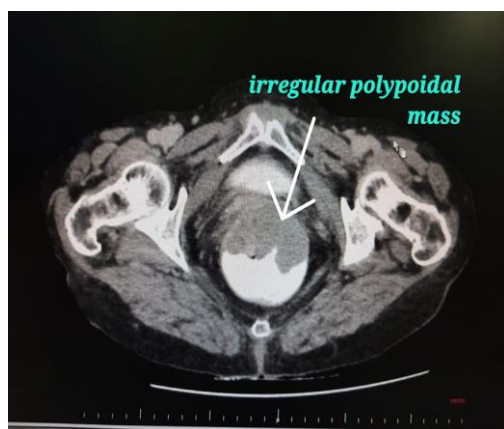


Figure 2: Pre-operative CECT scan of rectal GIST.



Figure 3: Resected specimen of sigmoid colon with anal canal.

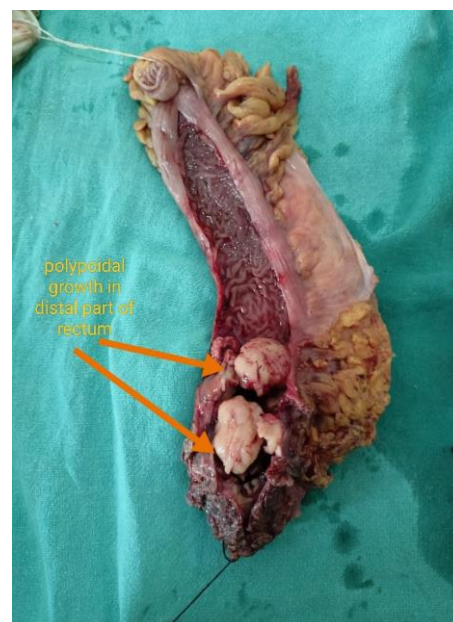


Figure 4: Cut open section of specimen showing growth in rectum just proximal to anal canal.

DISCUSSION

Gastrointestinal stromal tumours make up only 1% of primary GI cancers. Patients may present with nonspecific symptoms, including abdominal pain, GI bleeding, anaemia, or weight loss. Rectal GISTs are frequently found incidentally, either on cross-sectional imaging, screening colonoscopy, or clinical examination. Low-lying GISTs may be felt as a smooth, firm mass on digital rectal examination. Gastrointestinal stromal tumours usually appear as large, well-circumscribed, eccentric masses that enhance with intravenous contrast. Larger (>10 cm), heterogeneous tumours with areas of ulceration

or necrosis are associated with higher rates of malignancy in rectal GISTs.

Magnetic resonance imaging is beneficial in cases where CT cannot adequately identify the tumour or organ of origin, or assists in delineating invasion into surrounding pelvic structures. Endoscopic ultrasound is useful to further characterize the lesion. Most GISTs originate from the muscularis propria and occasionally from the muscularis mucosa, which can be distinguished on EUS as a hypoechoic lesion with well-defined margins. EUS-guided biopsy is preferred.⁴ Malignant GISTs have a high risk for metastasis, most commonly to the liver and intraperitoneally, whereas other sites such as lymph nodes, lungs, or bone are very rare.⁵ Positron emission tomography-computed tomography is useful in detecting metastases as well as evaluating tumour response to targeted molecular therapy.⁶

All GISTs are considered to have malignant potential, and, for that reason, all rectal GISTs should be considered for resection. American Joint Committee on Cancer recommendations on staging of rectal GIST: include -both tumour size (≤ 2 cm, 2-5 cm, 5-10 cm, and >10 cm) and mitotic rate (≤ 5 mitoses or >5 mitoses per 50 high-power field) to help determine rates of disease progression.⁷ Rectal GISTs provide a unique challenge compared with other locations within the GI tract, not only because of their worse prognosis and high local recurrence rate, but also because of the anatomical constraints of the pelvis. Consideration of Neoadjuvant Imatinib is important in all cases where a reduction in tumour size would substantially reduce the morbidity of the operation (e.g., large tumour size, borderline resectability, local organ invasion, or allow sphincter salvage). Imatinib should also be considered in those with intermediate- or high-risk tumour status. Neoadjuvant imatinib has been associated with improved surgical margins, and peri-operative imatinib has been shown to improve disease-free and overall survival.^{8,9} Trans anal endoscopic microsurgery (TEM) or trans anal minimally invasive surgery (TAMIS) may also be pursued in the case of similar GISTs within the distal to mid rectum. Proximal rectal GISTs often require a transabdominal approach such as an anterior resection. Larger and lower-lying tumours with local invasion or close proximity to the anal sphincters frequently demand an abdominoperineal resection to achieve oncologic clearance.

CONCLUSION

Surgical resection of gastrointestinal tumour forms the mainstay of treatment in rectal GIST, whenever and wherever possible. Neoadjuvant and peri-operative imatinib therapy decreases over all morbidity of rectal GIST patients with improved surgical margins, and preoperative imatinib has been shown to improve disease-

free and overall survival. Digital rectal examination and sigmoidoscopy followed by colonoscopy is the standard of care in evaluation and diagnosis in any cause of bleeding per rectum.

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