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Short Report

A Case Report Anal Gastrointestinal Stromal Tumor Presenting with Obstipation

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ABSTRACT

Gastrointestinal stromal tumor (GIST) is an uncommon gastrointestinal neoplasm with high rate of malignant transformation. The tumor is found mainly in the stomach and the small intestine, and the anorectum is an extremely rare anatomical location for this pathology. GIST is often discovered incidentally by a computerized tomography scan, endoscopy or during a laparotomy for other reasons. We describe a case of 64-year-old female with a mass in the anal canal presenting with tenesmus and difficulty defecation. Endoscopic ultrasonography and magnetic resonance imaging showed a mass at the rectum, externally compressing the vaginal wall. The patient was managed by a local excision which was preceded by an upfront therapy with imatinib. Histological examination showed proliferation of spindle cells stained with CD34 compatible with GIST. On post-operative follow-up visit, the patient did well with continuing imatinib therapy.

Keywords: anal canal; endoscopic ultrasonography; gastrointestinal stromal tumor; imatinib; magnetic resonance imaging

INTRODUCTION

Gastrointestinal stromal tumors (GIST) originate from mesenchymal cells in the wall of the gastrointestinal (GI) tract and accounts for 1.0% to 2.0% of GI neoplasms. The tumor is most commonly located in the stomach (55.0%), followed by the small bowel (31.8%), colorectum (6%), and the esophagus (less than 1.0%)[1]. The incidence of GIST is similar in both genders, and it may come at any age groups.

GIST is often discovered incidentally during abdominal computed tomography scans (CT scan), endoscopy, or surgical procedures for other reasons. The clinical presentation in symptomatic patients is determined by the location of the mass; abdominal pain, GI bleeding and GI obstruction from a huge exophytic growth tumor[1,2]. Endoscopic characteristics of GIST include a smooth shaped tumor with normal overlying mucosa, firm consistency on

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compression. Mucosal ulceration may appear in a large GIST. In a CT scan, a GIST may also appear as a well circumscribed extraluminal mass with heterogeneous enhancement in the arterial phase. Furthermore, necrotic hemorrhagic areas or degenerative components can be found especially in large GISTs. On the other hand, a CT scan revealing hypervascular lesions on the liver, could indicate the existence of metastatic disease. Magnetic resonance imaging (MRI) is superior in detecting rectal GISTs, any liver metastasis, hemorrhage, and tumor necrosis. At the time of presentation, about a half of all GIST patients have metastatic disease, which provides deleterious effects on prognosis. Endoanal ultrasound is another excellent tool for the evaluation of subepithelial tumors in the anal canal. Anal GISTs can be visualized in the intersphincteric plane as a hypoechoic round mass with smooth margins and no area of lymphadenopathy[3]. Furthermore, when endoscopic ultrasonography (EUS) reveals a heterogeneous, irregular extraluminal margin or a cystic component, a malignant GIST should be considered. A preoperative biopsy is not necessary when a lesion is in the operable stage but taking a preoperative specimen would be appropriate when treating patients with disseminated disease or those in the locally advanced stage[4]. A final diagnosis is established via both its unique microscopic features and by utilizing an immunochemistry panel (CD117, DOG-1, CD34, actin, desmin, S-100 and ki-67).

SHORT REPORT

A 64-year-old Asian female was referred for evaluation of an anal mass discovered by herself for a year. The patient complained that she had tenesmus at-rest, difficulty defecating requiring digital evacuation but without pain, rectal bleeding, or fecal incontinence. There was no loss in appetite or significant weight loss. On physical examination, she appeared in good health and there was no conjunctival pallor. Neither sides of her inguinal lymph-nodes were palpable. The abdomen was soft in palpation, showed no point of tenderness and no palpable abdominal mass. Digital rectal examination found a smooth fixed mass occupying the rectovaginal space, involving the parametrium and the anterior rectal wall. The rectal mucosa was smooth and showed no ulceration.

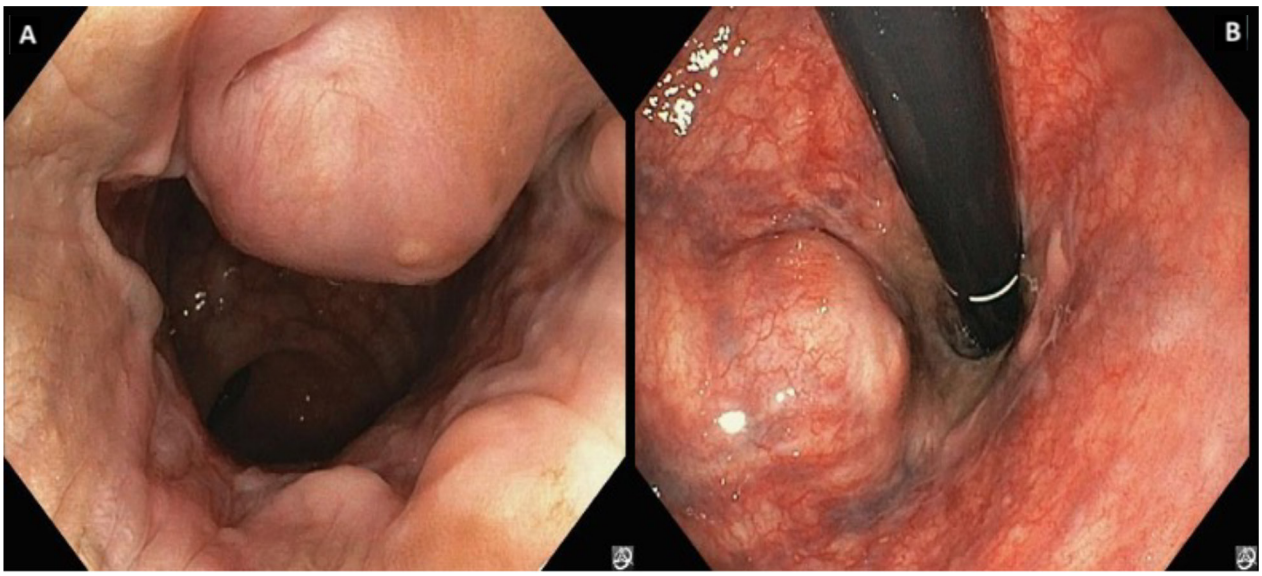

Figure 1 A: Oval shaped subepithelial mass, 3 cm in diameter, at dentate line without umbilication or ulcer, B: U-turn view of the subepithelial mass at the dentate line

On initial investigation, complete blood count showed: White blood cell 7470/mm^3, Neutrophil 55.7% Lymphocyte 35.2%, Hemoglobin 13.4 g/dL, Mean corpuscular volume 86.8 fL, Platelet count 281x10^3/µL. A colonoscopy showed a subepithelial mass, 3 cm in diameter, at the dentate line without umbilication or ulcer (Figure 1). Rectal MRI showed a 4.6x3.4x6.4 cm enhancing lobulated mass at the anterior wall of lower rectum extending to compress the posterior vaginal wall with no evidence of invasion or infiltration to the surrounding tissue. There was no local lymphadenopathy. An EUS found a hypoechoic mass of 4.3x6.2 cm in diameter arising from the second layer beneath the dentate line (Figure 2). A needle biopsy done by EZ shot 3 plus revealed a spindle cell tumor with the mitotic figure count at 0 mitoses/10 HPF. Immunohistochemistry results were positive for c-KIT protein, CD34, SMA and negative for desmin (Figure 3).

The patient was advised to start taking a preoperative targeted therapeutic agent, imatinib 400 mg per day for 4 months, in order to reduce the size of the tumor prior to a surgical resection. Later on, the patient was definitely treated by transanal local excision. A follow-up rectal MRI showed that the anal GIST significantly reduced in size. After 1 year, the patient was doing well under on-going molecular targeted therapy (imatinib). Fecal and urinary continence could be well maintained.

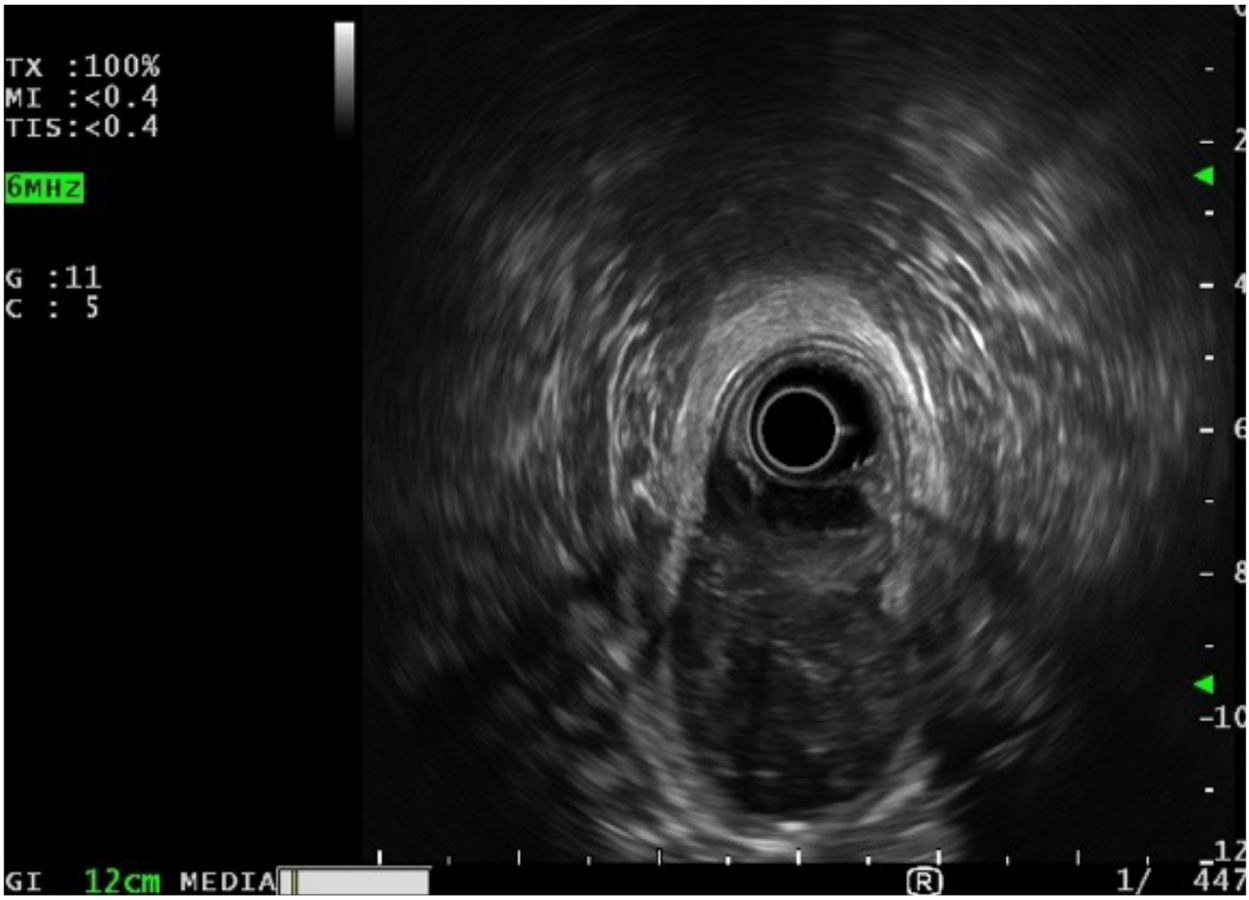

Figure 2 EUS showed an anal mass, a hypoechoic mass arising from the second layer beneath the dentate line

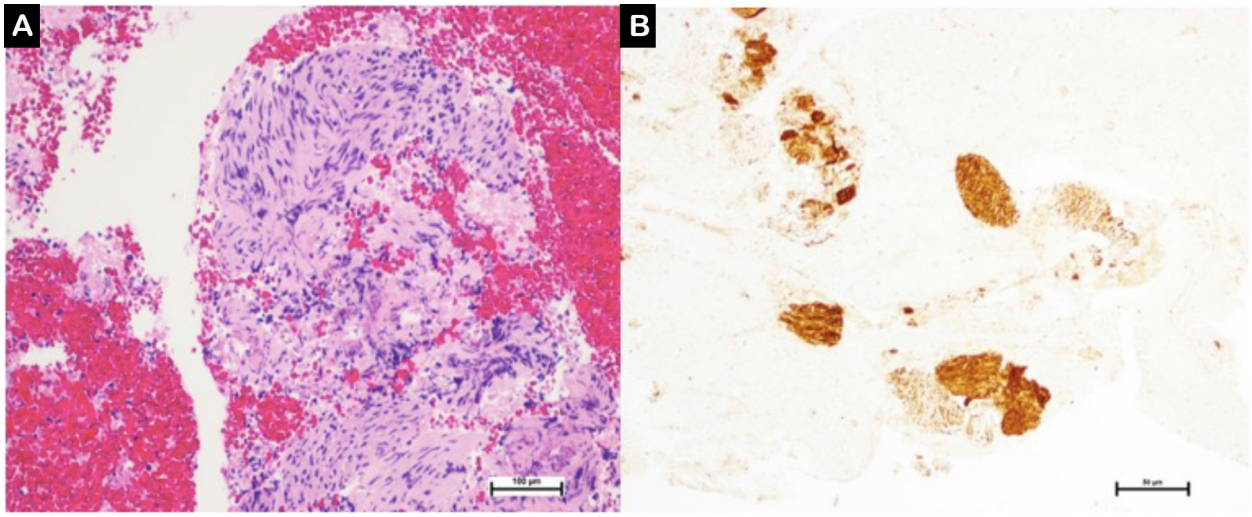

Figure 3 A: Hematoxylin and Eosin (×400) showing an eosinophlic, fusiform spindle cells with elongated nuclei, B: Positive immunohistochemistry stain for CD117

DISCUSSION

Anorectal GIST is a rare tumor, comprising approximately 5.0% of all GISTs in the GI tract and has malignant potential[5]. About a half of GISTs detected by a colonoscopy or a barium enema study are incidental findings when symptomatic patients present with either rectal bleeding, rectal pain, bowel habit change, signs of obstipation, or urinary symptoms[6]. Less than 10% of GISTs present as emergency conditions such as upper gastrointestinal bleeding and acute abdominal pain[7].

In our case, the patient had tenesmus, difficulty defecating and a mass found on digital rectal examination which involved the parametrium and anterior rectal wall with smooth rectal mucosa. Anatomical evaluation was done via colonoscopy, rectal MRI, and EUS which also allowed tissue acquisition for definitive histopathological diagnosis. The pathogenesis of GISTs is related to KIT gene and PDGFRA gene mutation.

Surgical resection without lymph nodes resection is the standard management for any GIST. Complete resection, as the key procedure in the treatment of rectal GIST, can ensure total tumor resection and avoid tumor rupture. Low rectal GISTs are a particular challenge because radical resection is associated with significant morbidity, and when the sphincter or pelvic floor muscles become involved, abdominal perineal resection and permanent colostomy are both required[8]. However, controversy exists in regards to whether an abdominoperineal resection or conservative surgery would be the best surgical procedure for anal canal GISTs. Several studies have also compared local resection, including traditional transanal excision, transanal minimally invasive endoscopy, transacral, the perineal resection, and the transvaginal approach with residual section and found that LR is superior and with a shorter operative time, less bleeding, and a rapid recovery, while oncology outcomes are similar[9]. Transanal resection has a minimally invasive effect, fewer postoperative complications, a high anal sphincter preservation rate, a R0 resection rate, and a better prognosis. In recent decades, imatinib has been an efficient preoperative therapeutic agent in reducing tumor size and mitotic activity; making *En bloc* surgical resection more feasible and preserving the anal sphincter[10,11]. Several

studies have recently confirmed the superiority and benefit of combining surgery with preoperative adjuvant imatinib for anorectal GIST. On the other hand chemotherapy was not found to improve the overall survival and cancer specific survival for patients with anorectal GIST.

In our case, we decided to utilize imatinib as a preoperative therapeutic agent for reducing the tumor size and preserving the anal sphincter before resection due to mass involvement with the levator ani muscle and in order to avoid abdominoperineal resection. The patient was taking imatinib due to an incomplete resection. A clinical evaluation, with rectal MRI at 1 year after the resection indicated that tumor dramatically reduced in size and there was no loss of fecal continence.

CONCLUSION

We presented a case of anorectal GIST, a rare but better clinical outcome when compared to GIST in other locations of the GI tract. This combination of preoperative targeted therapy and surgical resection can achieve good oncologic outcomes whilst preserving the anal sphincter.

CONFLICT OF INTEREST

The authors declare that they have no conflicts of interest.

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