

RECURRENT ANAL CANAL GASTROINTESTINAL STROMAL TUMOR

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PJR October - December 2015; 25(4): 175-178

ABSTRACT

Gastrointestinal stromal tumors (GIST) are the most common mesenchymal neoplasms of the gastrointestinal tract; however these account for <1% of gastrointestinal tumors. Most common site is the stomach (50–60%), followed by small intestine (30–40%), colon (7%) and esophagus (1%), they rarely have extra-gastrointestinal involvement.¹ GIST of the anal canal are extremely rare. They along with GIST of the rectum represent 5% of GIST.² Of the anorectal GIST, GIST arising from the anal canal account for only 2%–8%. We present a case of recurrent anal GIST which was earlier treated by local excision.

Key words: Gastrointestinal Stromal tumor, MRI, Fistula

Introduction

GIST are defined as mesenchymal neoplasm expressing KIT protein, driven by *KIT* or *PDGFR* α (platelet derived growth factor alpha) mutations.³ They are regarded as derived from interstitial cells of Cajal (ICC). ICC serves as pacemaker cells that regulates motility, they are part of autonomic nervous system. Activation of *KIT* by mutations causes Cajal cell proliferation and GIST.⁴

Presentation of the patient is variable and depends on size, region involved by tumor and metastasis. Common symptoms are abdominal pain and GIT hemorrhage.

Radiological studies aid in diagnosis however histopathology and immunohistochemistry of the biopsy is confirmatory. The ultrasonographic images of GISTs are best acquired during endoscopic ultrasonography however, as GISTs are associated with air-filled viscera, image quality is often degraded

by intervening bowel gas. CT scan is modality of choice in the diagnosis and staging. CT scan can also be used to detect the presence of multiple tumors and can provide evidence of metastatic spread. MRI appears to be just as sensitive as CT scan. GIST appears hypo-intense on T2-weighted images. In localized GISTs, surgery is the primary treatment of choice. Surgery can be potentially curative, but watchful waiting may be considered in small tumors in carefully selected situations.⁵ In addition the GIST susceptibility to metastasize via bloodstream and to relapse as local recurrence, makes the surgical treatment controversial regarding the extent of resection. Authors who treated rectal GIST by an abdomino-perineal resection reported a low local recurrence, with no improvement in incidence of distant metastases and overall survival rate.⁶

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Submitted 11 June 2015, Accepted 22 June 2015

Case Report

A 65-year-old male presented with pain during defecation, constipation and tenesmus together with perianal swelling. Pain was nonradiating, dull aching, and persistent. There was no associated bleeding per rectum or weight loss. There was no significant relief with medication. He also complained of occasional fecal incontinence as well.

There had been past history of fistula in ano for 7-8 yrs for which he was operated 4 times. He also had history of a multilobulated anal canal mass at 7'o clock for which he was operated 3 yrs back and histopathology revealed it to be anal canal GIST with positive CD117 in tumor cells. Sections further revealed a spindle cell neoplasm composed of intersecting fascicles of spindle cells showing uniform nuclei. There were 14-15 mitosis/50HPF, making it fall in high risk category. Colonoscopy performed 6 months after the surgery revealed well healed peri anal scar, no residual disease was identified.

The patient was not a known case of piles/diabetes mellitus/hypertension/tuberculosis/or any other chronic ailment. The patient was a known smoker. He had no urinary complaints. There were no other complaints referable to chest and cardiac or nervous system.

His general physical examination and systemic review was unremarkable. On digital rectal examination, there was a firm to hard nontender mass at around 7'o clock to 9'o clock, starting 5cm from the anal verge. The mass had a bosselated surface, and the overlying mucosa was intact. The upper border of the mass could not be reached. No inguinal, iliac, para aortic, or supraclavicular lymph nodes were palpable. His hematological and biochemical parameters were all within normal limits, and he was HIV seronegative.

CT scan of the abdomen and pelvis showed a well-defined smoothly outlined mass in anorectal region causing luminal narrowing. MRI pelvis revealed a well marginated 52 x 52 mm mass in submucosal location of anal canal along its right posterolateral margin. The lesion was 3 cm from anal verge. The lesion was causing compressin and narrowing of anal canal lumen (Fig. 1 and 2). An associated fistulous tract was identified at 6'o clock position traversing internal and external sphincters to reach Right Ischio-

rectal fossa before exiting through skin. His colonoscopy and biopsy suggested recurrent GIST. The patient was sent for re do surgery and possible chemotherapy.

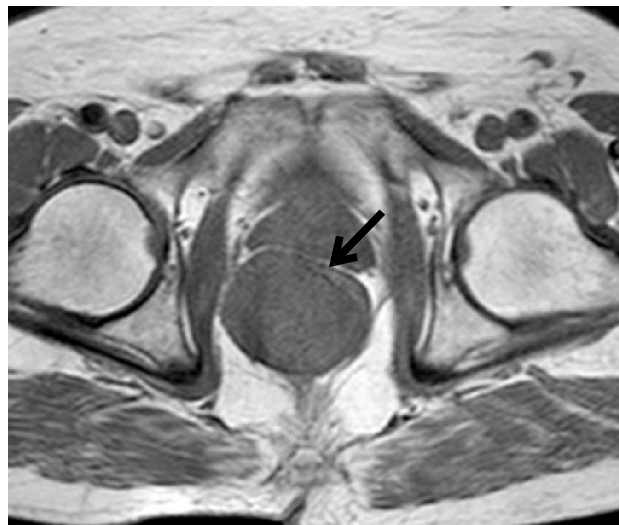


Figure 1: T1 weighted MR image shows a well circumscribed homogenous hypointense mass compressing the anal canal lumen from outside. Black arrow shows the compressed anal canal



Figure 2: T2W Coronal image again shows a lobulated anal canal submucosal mass having high T2 signal. Black arrow shows compressed anal canal lumen. Grey arrow shows Trans sphincteric perianal fistula at 6'o clock position

Discussion

Gastrointestinal stromal tumors (GISTs) account for less than 1% of gastrointestinal tumors, but they are the most common mesenchymal neoplasms of the gastrointestinal tract. GISTs are usually found in the

stomach or small intestine but can occur anywhere along the GI tract and rarely have extra-GI involvement. The term was first used in 1983 to describe an unusual type of nonepithelial tumor of the gastrointestinal tract that lacked the traditional features of smooth muscle or Schwann cells.⁷

The anal canal extends from the perianal skin (anal verge) to the rectal mucosa. An important landmark within the canal is the dentate or pectinate line, which represents the end of the squamous mucosa and the beginning of a zone of transition from squamous to nonsquamous (either transitional or rectal glandular) mucosa. Thus, tumors arising in the anal canal can be either keratinizing or nonkeratinizing depending on their location in relation to the dentate line. Importantly, both keratinizing and nonkeratinizing tumors appear to have similar biology and prognosis.⁸ Adenocarcinomas, on the other hand, behaves quite differently and should be treated like rectal cancers. Since there is no easily identifiable landmark between the rectum and anal canal, one has to rely on the pathologic classification of tumors in this area rather than the surgical or endoscopic classification.⁹

GIST are currently thought to originate from interstitial cells of Cajal. Mutational statuses of c-KIT and PDGFR α genes are the basis for the diagnosis of this neoplasia and represent the criteria for surgical therapy, expected chemotherapy response, and clinical outcomes.⁹

MRI is the imaging modality of choice for anal canal Gists due to its high soft tissue contrast. Generally these tumors present as well circumscribed, smoothly outlined solid masses causing narrowing of the luminal narrowing. There submucosal location makes endoscopy less favorable for diagnosis. Some lesions might have central necrosis. On MRI, they tend to have diffuse low signal on T1 and high signal on T2. Post contrast enhancement is homogenous in solid variety and ring pattern in case of necrotic center. Signs of malignancy on imaging include large size, lobulated contours, increase in size on follow up imaging and presence of hematogenous metastasis. Up to 50% of all GISTs will have evidence of metastatic disease at the time of presentation,³ which significantly impacts prognosis.¹⁰

Historically, prior to ST-571 (Imatinib; Glivec) up to 85% of tumours would locally recur or develop subsequent distal metastases despite treatment¹⁰ and had

proven to be resistant to standard chemotherapy.¹¹ Adjuvant chemotherapy with ST-571 (Imatinib; Glivec) is effective in the majority of cases and has had a dramatic impact of prognosis even with only one year of therapy, reducing recurrence at one year from 17% to 3%.¹¹


In our case, the tumor was initially removed surgically. There was no record available with the patient regarding intake of Imatinib.

Conclusion

Anal canal GISTs, though very rare but can be encountered in clinical practice. Recurrent GIST, an even rarer entity can be expected after complete resection of the original tumor. Proper knowledge of anal canal anatomy and differential possibilities of lesions in this region are crucial for diagnosis.

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