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Case Report

Anorectal gastrointestinal stromal tumor: a rare case report

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ABSTRACT

A gastrointestinal stromal tumor (GIST) is a mesenchymal tumor of gastrointestinal tract arising from the interstitial cell of Cajal with rare occurrence in anorectum. Mutations of c-KIT proto-oncogene characterized by the expression of the KIT (CD-117) tyrosine kinase or platelet-derived growth factor receptor alpha (PDGFRα) is frequently seen with these tumours. Elderly patients with age greater than 50 are at higher risk of GIST. Here, we present a case of a 51-year-old male who presented with vague dull aching pain in the right-side perianal area since last one month with a submucosal mass in the right posterolateral wall of the anal canal and rectum. The histopathological and immunohistochemistry study of the biopsy sample reported strongly positive for CD117 and less than 3% of tumor cells showed Ki67 positivity. The patient underwent laparoscopic abdomino-perinel resection. Post-operatively, the patient was started on adjuvant imatinib with regular follow-up.

Keywords: GIST, Mesenchymal tumor, Anorectal tumor, Imatinib

INTRODUCTION

A gastrointestinal stromal tumor (GIST) is a gastrointestinal tract neoplasm with mesenchymal origin arising from the interstitial cell of Cajal first described in 1983 by Mazur and Clark.[1] With their unique histological, immunophenotypic and molecular genetic features, these tumours are considered different from typical smooth muscle tumours and schwannomas; however, historically GIST was classified under GI-smooth muscle tumours.[2]

Mutations of c-KIT proto-oncogene characterized by the expression of the KIT (CD-117) tyrosine kinase or PDGFRα is frequently seen with these tumours.[3] Elderly patients with age greater than 50 are at higher risk of GIST with median diagnostic age of approximately 60 years.[4] GISTs are most common in the stomach (60-70%), followed by small intestine (20-25%), colon and rectum (5%), and esophagus (<5%), of these anorectal GIST remains a rare entity with a diagnostic challenge due to its rarity.[5]

Due to anatomical constraints of pelvis and higher propensity of local recurrence, anorectal GISTs provide a unique challenge compared with other locations within the GI tract. Due to its rarity, we present a case of anorectal GIST in a 51-year-old male and its management outcome with a focus on pathological identification, site of origin, prognosis, and the treatment.

CASE REPORT

A 51-year-old male presented with vague dull aching pain in the right side perianal area since last one month. The patient did not have any history of weakness, weight loss, or abdominal pain. There was no history of constipation or bleeding in stool. The patient underwent incisional biopsy in general surgery department, which turned out to be gastrointestinal tumor.

On perineal examination, an ulcer of size 2×3 cm extending from 7 to 10 o' clock position, 1 cm away from the anal verge was present (Figure 1). Digital rectal examination revealed a submucosal mass palpable in the

posterolateral wall of the anal canal and rectum present on the right side (5 to 11 o'clock position. Upper limit of the mass was 7-8 cm from anal verge. MRI pelvis showed a single well encapsulated smooth marginated with exophytic T2 heterointense lesion with areas of necrosis (Figure 2). The mass measured 5.2×4.6×9.2 cm and was situated along the right lateral wall of anorectum in intersphincteric plane extending into perianal, ischiorectal and perineal region. The lesion was abutting posterior surface of right side of prostrate. Capsule breach was seen along inferomedial aspect of the lesion. There was no meso-rectal infiltration or perirectal lymphadenopathy.

Contrast enhanced CT of abdomen and pelvis suggested same findings without any evidence of distant metastasis. CT chest showed no evidence of distant metastasis. Histopathological examination of incisional biopsy performed in general surgery department showed spindle cells with eosinophilic cytoplasm and hyalinized stroma with a mitotic activity of less than 5/50 HPF suggestive of GIST with low malignant potential (Figure 3). Immunochemistry analysis reported that the tumor cells were strongly positive for CD117 and less than 3% of tumor cells showed Ki67 positivity. Tumor cells were negative for SMA and S100.

After confirming the diagnosis, patient underwent laparoscopic abdominoperineal resection (APR) with end sigmoid colostomy (Figure 4). Post operative period was uneventful. Patient was started on oral diet on post-operative day 2. Patient was discharged on postoperative day 5 after removal of urinary catheter and stoma therapist opinion for stoma care. No complications were reported at 1 and 3 month follow up.

Postoperative histopathology revealed circumscribed submucosal lesion measuring 7.4×4.5×5 cm with external surface of skin showing ulcer of size 2.5×1 cm. The margins were negative. Patient was started on adjuvant imatinib.

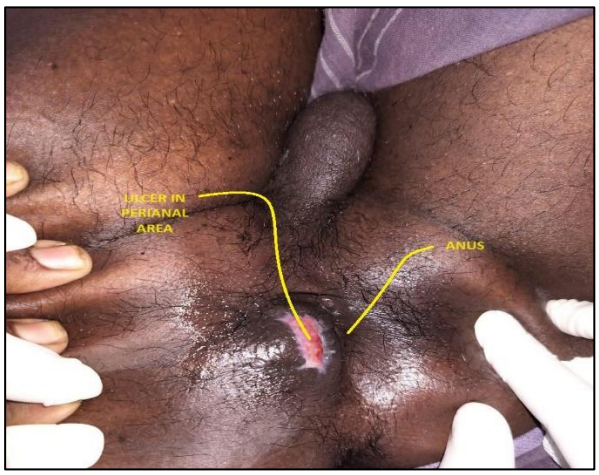

Figure 1: Clinical picture showing ulcer in perianal area.

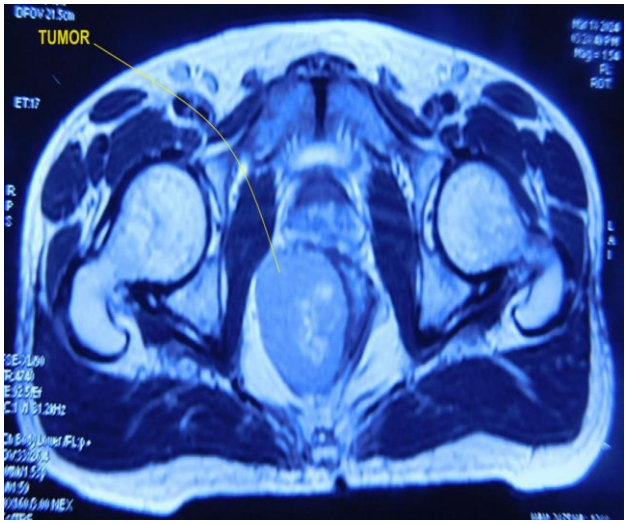

Figure 2: T2W axial image showing tumor in distal rectum.

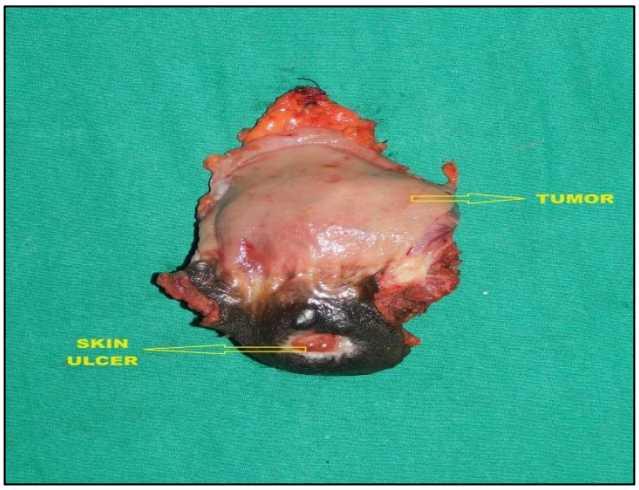

Figure 3: APR specimen.

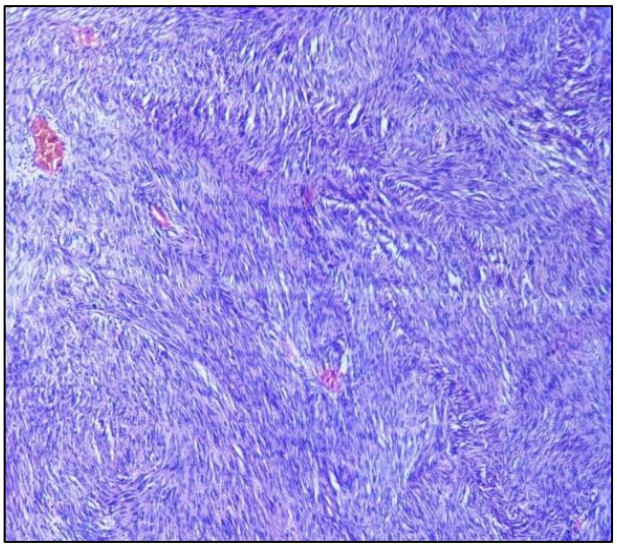

Figure 4: Spindle cells arranged in fascicles showing nuclear pleomorphism (100x).

DISCUSSION

GI stromal tumors of rectum are the rare entity and account for 3% of anorectal mesenchymal tumors.[6,7] GIST are currently believed to arise from interstitial cells of Cajal which are responsible for regulating the intestinal motility and other autonomic functions. Cajal cells are considered as subset of multipotential stem cell-like cells that differentiates into smooth muscle cells if KIT signaling is disrupted.

Histologically, most of the GISTs are characterized by the presence of spindle cells, with a small percentage of tumors possessing epithelioid pattern.

Mutations in the KIT proto-oncogene or PDGFRα are seen in GIST development and forms the basis of diagnosis. CD117 is the antigen corresponding to the KIT protein, which is considered to be a specific marker for the diagnosis of GIST and forms the basis of surgical therapy, expected chemotherapy response, and clinical outcomes. Most of the GIST were found to be CD117 positive followed by CD34 in an earlier reported case study.[8]

Expression of smooth muscle markers such as smooth muscle actin (SMA), Desmin, and h-caldesmon (HSD) are seen commonly in GISTs of the small bowel and rarely expressed in the rectum.[8] Mutation of exon 11 of KIT is often a ubiquitous feature of GISTs.[9]

Large tumour size (>5 cm) and high mitotic activity (>50 HPF) points towards high malignant potential of GIST, but the biological behavior of anorectal GIST is less clear. Patients may present with nonspecific symptoms, including abdominal pain, anemia, or weight loss. Anorectal GISTs may be felt as a smooth firm mass on per rectal examination.

Contrast-enhanced CT scans and MRIs are the best modalities for both detection and staging of tumour. GISTs usually appears as well circumscribed, eccentric masses with areas of ulceration or necrosis. Local imagining also helps determine the involvement of adjacent organs, the tumor's distance from the anal verge and helps to plan for surgical management.

Tissue diagnosis and immunohistochemistry with CD117 and CD34 positivity is required to confirm the diagnosis of GIST.[11]

Surgery remains treatment of choice for GIST. Anorectal GIST, due to associated complex anatomy, controversy remains whether APR or conservative surgery is the best alternative. Local recurrence is lower with APR, but there is no significant impact on distant metastasis and overall survival.

In a similar case, reported used combined treatment modality, including surgical resection and perioperative treatment with tyrosine kinase inhibitors-based systemic chemotherapy using imatinib mesylate and sunitinib malate, facilitating anus-preserving surgery. However, in present case report, there was skin ulceration because of previous incisional biopsy which made anus preserving surgery a difficult option and APR was a better option to achieve better oncological control.

Final histopathology and immunohistochemistry are helpful in guiding adjuvant therapy based on important prognostic factors like size, grade, mitotic figures and CD-117 score.

CONCLUSION

GIST anorectum is a rare and difficult-to-diagnose tumor. It is associated with mutations of c-KIT, characterized by the expression of the KIT (CD-117) tyrosine kinase. In view of perineal ulcer due to previous incisional biopsy, neoadjuvant therapy and organ preserving approach was disregarded and patient underwent laparoscopic APR. Post operatively patient was started on imatinib with regular follow-up.

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