Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com/journal/sjmcr/home

OPEN ACCESS

Diagnosis and Management of Rectal Gastrointestinal Stromal Tumour (GIST), a Case Report

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| **Received:** 01.01.2019 | **Accepted:** 10.01.2019 | **Published:** 21.01.2019
DOI: 10.36347/sjmcr.2019.v07i01.009

Abstract **Case Report**

Gastrointestinal stromal tumour (GIST) are mesenchymal tumours containing spindle cells which originate from intestinal cell of Cajal, and showing CD117 positivity. Rectal GIST is rare, but the symptoms are the same with other rectal neoplasia. We report a case of a woman who presented with intermittent per rectal bleeding, mucous and faecal incontinence for two months. CT scan and MRI were done, and diagnosis was confirmed with tissue biopsy. She underwent Abdomino-Perineal Resection (APR) and recovered uneventfully. The diagnostic work-up, prognostic factors, choices of surgical treatment, and outcome of rectal GIST with imatinib therapy are discussed in this article.
Keyword: Rectal; GIST; Diagnosis; Surgery; Imatinib.

INTRODUCTION

Gastrointestinal stromal tumour (GIST) is defined as gastrointestinal mesenchymal tumours containing spindle cells and showing cluster of differentiation (CD) 117 or c-kit protein positivity [1-3]. The GIST cells are postulated to originate from the interstitial cell of Cajal, an intestinal pacemaker [4].

Stomach is the most common site for GIST (60%-70%), followed by small intestine (20%-25%) and less commonly rectum [1,5]. Rectal GISTs are rare entity. Only 0.1% of rectal tumours are found to be GISTs [5]. It has marked male predominance (71%), and occurs predominantly between 5th and 7th decades of life [1].

CASE REPORT

A 46-year-old woman presented with intermittent per rectal bleeding with mucous and faecal incontinence for two months. She was clinically pink, with no mass palpable per abdomen. Digital rectal examination (DRE) revealed fungating mass palpable at the anal verge. Colonoscopy showed a huge fungating mass above the dentate line, which was ulcerated and bleeding.

Histological examination confirmed the tumour as GIST, however tumour grade was not determined due to inadequate tissue size. Serum Carcinoembryonic Antigen (CEA) and Carbohydrate Antigen 19-9 (CA19-9) were within normal values. There was no distant metastasis demonstrated in computed tomography (CT) Thorax-Abdomen-Pelvis (TAP). CT Angiogram of the pelvis showed a pelvic mass with regional lymphadenopathy but no active bleeding. Magnetic resonance imaging (MRI) pelvis revealed a large lobulated exophytic rectal mass measuring 7.8cm x 7.2cm x 6.3cm, about 3cm from anal verge (Figure 1). The bulk of this mass was extraluminal, with smaller intraluminal component across the breached mucosa causing significant stenosis. The mass mostly encased by muscularis propria, with suspicious breach at three o'clock. There was clear plane with the peritoneal reflection, the uterus anteriorly, and the sacrum posteriorly (Figure 2).

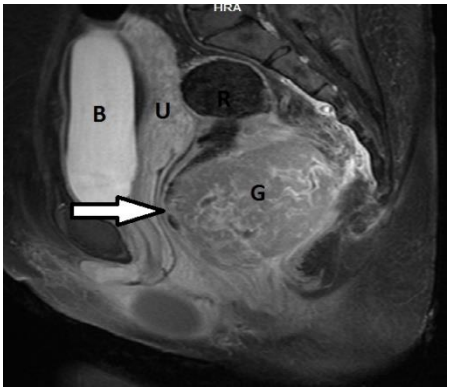

Fig-1: Sagittal T1-weighted MRI Pelvis shows a large lobulated exophytic extraluminal lesion with small intraluminal component with breached mucosa (arrowhead). There is clear plane with the peritoneal reflection, uterus, and sacrum. B, urinary bladder; U, uterus; R, rectal lumen; G, rectal GIST

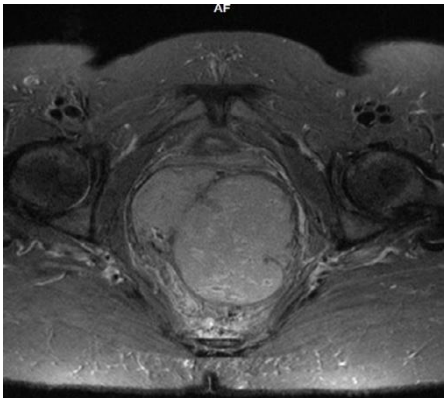

Fig-2: Axial T2-weighted MRI Pelvis

She underwent embolization of the rectal GIST, followed by Abdomino-Perineal Resection (APR) and recovered uneventfully from surgery. The tumour measured 12cm x 9cm x 7cm, located 5cm above the anal verge. All margins were clear and there were no lymph node metastasis. Histologically, the tumour showed spindle cell type with mitotic rate of 3/5mm^2. Immunohistochemistry tests were positive for CD117, CD34 and vimentin, but negative for S-100 protein and desmin.

She was planned for adjuvant Imatinib mesylate (Gleevec®, Novartis, Switzerland) therapy, however, due to financial constraint; she was not able to undergo the therapy, thus planned for yearly surveillance MRI pelvis and chest x-ray. So far, she has been disease free for 16 months.

DISCUSSION

The symptoms of rectal GIST are similar to other rectal tumours [5]. Small tumours are usually incidental, while large tumours commonly manifested by rectal bleeding, signs of obstruction, pain or rectal fullness [1, 6]. Rarely, urinary symptoms in male are due to mass effect or local prostatic infiltration [6].

The liver is the most common site of metastasis (65%) followed by the peritoneum (21%) Lymph nodes, bone and lung metastases are rare [1].

The diagnostic work-up for rectal GIST includes colonoscopy, CT scan, MRI pelvis, and tissue biopsy. However, the hallmark of GIST diagnosis is the immunohistochemistry.

The diagnosis of GIST is often histological. The majority of GIST cases have uniform appearances of either one out of three types: spindle cell (70%), epithelioid (20%), or mixed [2]. CD117, a c-kit proto-oncogene protein, is seen in almost all GISTs regardless of the site of origin, histologic appearance and biologic behaviour, hence its expression is considered to be best defining feature of GISTs [7]. Aside from consistent positivity for CD117, about 60% to 70% of GISTs show immunopositivity for CD34, 30% to 40% for smoothmuscle actin (SMA), and around 5% for S-100 protein [2].

Several biomarkers such as CEA and CA19-9 are integral in detection of colorectal cancers, however there is no evidence of their relation to detection of GISTs [8].

The role of CT scan is to characterize and delineate the full extension of large exophytic masses,

to detect local invasion and distant metastases, as well as tissue guided biopsy [6]. MRI helps in tumour localization, delineation of the relationships of the tumour to adjacent organs, and surgical planning, since it provides better soft-tissue contrast resolution and direct multiplanar imaging [6].

GISTs are hypervascular. CT angiography may be used to localize its organ of origin by identification of the tumoral blood supply [6], as well as for embolization of bleeding GIST.

Important prognostic indicators for GIST are tumour size and mitotic count [1,2,7]. The majority of rectal GISTs that were >5 cm with any number of mitoses or had >5 mitoses/50 HPF regardless of size showed malignant behaviour [1].

Surgery is the mainstay in the treatment for primary resectable GISTs. For rectal GIST, local excision, anterior resection of the rectum (sphincter preserving) and abdomino-perineal resection (APR) are among the few options of procedure. The choice depends on tumour size and location [1].

APR should be reserved only for loco-regional advanced lesions, recurrent tumours of the lower rectum, or large tumours of the lower rectum primarily resistant to tyrosine kinase inhibitors (TKI) treatment or severe drug intolerance [9].

Imatinib mesylate, a TKI that specifically inhibits most KIT, has been widely used and became the standard therapy for advanced un-resectable/metastatic GISTs [9]. It is also recommended for tumours larger than 5 cm, in order to get a negative resection margin, which is an important factor for survival in rectal GISTs [9,10]. It may downsize the tumour, to ensure more conservative, much easier, and safer surgical procedure, with higher rate of microscopically negative-margin (R0) resection, hence improving disease free survival (DFS) and overall survival (OS) [9].

CONCLUSION

Rectal GIST is rare entity. The diagnostic work-up of rectal GIST is exactly the same as for other rectal neoplasia. Immunohistochemistry is hallmark in the diagnosis, namely characterization of CD117 and CD34. MRI is helpful in localizing the tumour, delineating the tumour extension and surgical planning. The main prognostic indicators are tumour size and mitotic count. Surgery is the mainstay of treatment in non-metastatic rectal GIST. Imatinib therapy has markedly changed outcome as it is associated with higher R0 resections rate, improved DFS and OS.

Acknowledgment

We would like to thank the Director General of Health Malaysia for his permission to publish this article (KKM.NIHSEC.800-4/4/1 Jld. 60(29)).

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