ORIGINAL RESEARCH PAPER

General Surgery

ANORECTAL GASTROINTESTINAL STROMAL TUMORS (GISTs) - CASE REPORT & SYSTEMATIC REVIEW

KEY WORDS: Anorectal GISTs, Case report, Systematic review

Dr Joseph Francis MBBS, MS, Associate Professor, Dept of General Surgery, Travancore Medical College, Kollam, Kerala.

Dr. Linta Maria David MBBS, Junior Resident, Dept of General Surgery, Travancore Medical College, Kollam, Kerala.

Dr. Anees U MBBS, Junior Resident, Dept of General Surgery, Travancore Medical College, Kollam, Kerala.

ABSTRACT

Anorectal gastro intestinal stromal tumors constitute about 5 % of all GISTs. Of these only 2-8% are from anal canal, hence anal canal GIST constitutes an extremely rare group of tumors. Here we are reporting a rare case of anorectal gastrointestinal tumor. A 54-year-old male presented with mild pain and bleeding per rectum of recent onset. The patient was evaluated and found to be having a small sized anorectal GIST for which local excision was done, histopathological examination showed low grade GIST. We are presenting the case with relevant illustrations of the radiological and histopathological images. Also, we are presenting a systematic review of anorectal gastrointestinal tumors conducted by us.

INTRODUCTION

Background And Significance

Gastrointestinal Stromal tumors (GISTs) constitutes the most common mesenchymal neoplasms of the gastro intestinal tract. They arise from the interstitial cells of Cajal (pacemaker cells) and associated with mutations in the KIT and PDGFRA genes. GISTs of anal canal and rectum are often grouped together and account for 5% of all GISTs. Of these only 2-8% are from anal canal, hence anal canal GIST constitutes an extremely rare group of tumor.

Case Report

A 54 year old male patient, presented with recurrent episodes of mild pain on the right side of anal canal and bleeding per rectum. The patient has no co-morbidities. He is a known smoker and known alcoholic.

Per Rectal examination revealed a hard mass between 7 and 10 o' clock position in the submucosal plane adherent to the deeper structures.

On investigation lower GI endoscopy showed submucosal swelling noted at 9 o' clock position involving anal canal and distal rectum.

Endoscopic ultrasound [EUS] showed well defined hypoechoic avascular lesion measuring 2.0 x 1.5 cm with central calcification seen in distal rectum.

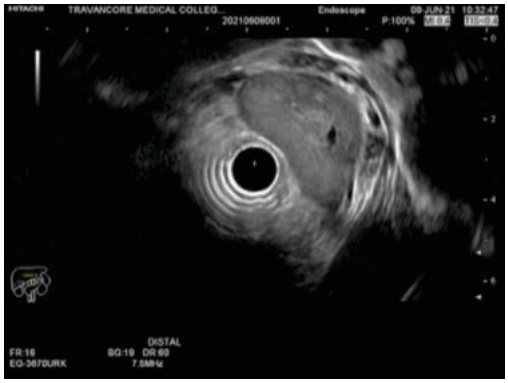

Fig 1: Endoscopic Ultrasound Image Showing The Tumor

MRI plain and contrast showed well defined enhancing lesion arising from the right lateral wall of anorectal junction measuring 2.2 x 1.5 x 2.2 cm.

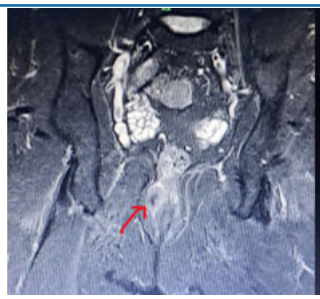

Fig 2 (a) - showing T2 STIR Coronal view of pelvis with arrow pointing at the tumor

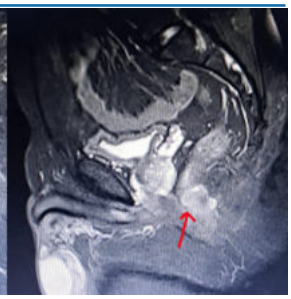

Fig 2 (b) – shows arrow pointing the tumor in T2 STIR Sagittal view of pelvis

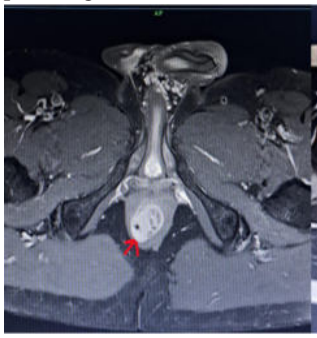

Fig 2 (C): Tumor visualized in T1WI- Axial cut

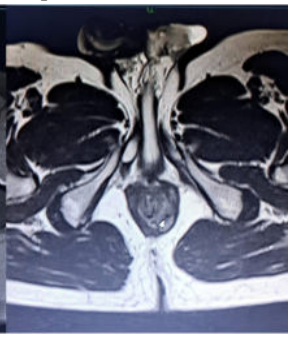

Fig 2 (d): Tumor seen in T2WI- Axial cut

In view of the small size of tumor and non-involvement of adjacent structures, the patient had undergone local resection through anal canal with the aid of self-retaining speculum preserving the anal sphincters.

3 (a)

3 (b)

Fig 3 (a) & (b) - Shows Gross Specimen After Local Resection

Histopathological examination- macroscopy showed – grey white lesion measuring 2.5 x 2 x 1.8 cm with focal area of calcifications.

Microscopy- showed spindle cells with moderate amount of eosinophilic cytoplasm and ovoid/ elongated vesicular nucleus. Mitotic figures [<5 mitoses/50 HPF] IHC- CD117- strong diffuse membranous and cytoplasmic positivity

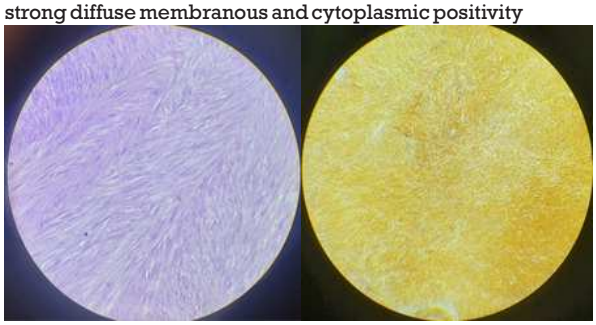

Fig 4: Microscopic View Of The Cut Section

Fig 5: IHC showing strong positivity

Postoperative period was uneventful. No symptoms of incontinence were present. No adjuvant therapy was advised due to low grade of neoplasm. Patient is on regular follow up for last 3 years and is asymptomatic with no evidence of recurrence.

Objectives Of The Review

The aims of this review are to analyze current knowledge on anorectal GISTs focusing on their epidemiology, clinical presentation, diagnostic approaches, treatment options and outcomes.

METHODS

Search Strategy

A comprehensive search of electronic databases including PUBMED, MEDLINE, and EMBASE was conducted. The search terms included "anorectal GIST, "gastrointestinal Stromal tumors" and related keywords. Studies published in English language up to 2023 were considered.

Inclusion And Exclusion Criteria

- Inclusion Criteria:

Original Research articles, case series and clinical trials.
Studies reporting on anorectal GISTs.
Studies with clearly defined diagnostic criteria.

- Exclusion Criteria:

Reviews, editorials and non-original articles.
Studies not specifically considering anorectal GISTS.
Case reports with insufficient data.

Data Extraction And Quality Assessment.

Data were extracted independently by two reviewers. The quality of these was assessed using the New Castle-Ottawa scale for observational studies and the Cochrane Risk of bias tool for clinical trials.

RESULTS

Study Selection And Characteristics

Out of 150 screened articles, 20 studies met the inclusion criteria comprising a total of 300 patients with anorectal GISTs. Studies assessed in design mostly included retrospective reviews and a few prospective Cohort studies

Epidemiology And Clinical Presentation

Anorectal GISTs primarily affect adults aged between 50-70 years, with a slight male predominance. Common symptoms include rectal bleeding, pain and obstructive symptoms. Some of the cases were asymptomatic and discovered incidentally.

Diagnostic Modalities

Endoscopic ultrasound (EUS), MRI and CT scans are pivotal in diagnosing anorectal GISTs. Biopsy and immunohistochemistry are necessary for confirmation, KIT (CD 117) and DOG 1 being the key markers.

Treatment Modalities

For localized anorectal GIST, the primary treatment is surgical resection. Depending upon the Location, tumor size and involvement of adjacent structures, the option of abdominoperineal resection may be chosen. Imatinib, a tyrosine kinase inhibitor is the mainstay in adjuvant therapy for high risk of metastatic disease.

Outcome And Prognostic Factors

The prognosis of anorectal GIST's is influenced by tumor size, mitotic index, and mutation type. The five-year survival rate ranges from 50% to 70%. Patients with small, low mitotic tumors have a better outcome. Recurrence is common, and requires long term follow up.

DISCUSSION

Comparison With Existing Literature

Anorectal GISTs are less common and generally have a poorer prognosis compared to GISTs in other locations possibly due to delayed diagnosis and the technical challenges of surgery in the anorectal region.

Clinical Implications

Early diagnosis and appropriate management are crucial for improving the outcomes in anorectal GISTs patients. The role of neoadjuvant therapy with Imatinib to downsize tumors and facilitate surgical resection is an area of active consideration.

Limitations

The rarity of anorectal GISTs causes limitation of the availability of large-scale studies, and most data comes from retrospective analysis. Additionally, there is variability in treatment protocols across studies.

Future Research Directions:

Further research is needed to establish standardized treatment guidelines, explore the role of novel targeted therapies and better understand the molecular biology of anorectal GISTs.

CONCLUSION

Anorectal GISTs are a rare and challenging subset of gastrointestinal stromal tumors. While surgical resection remains the mainstay of the treatment, adjuvant therapies have improved outcomes. Continued research and collaboration are essential to enhance the understanding and management of these tumors.

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