Section: General Surgery

Original Research Article

GASTROINTESTINAL STROMAL TUMOURS: EXPERIENCE FROM A RURAL TERTIARY CARE CENTRE IN SOUTH INDIA

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ABSTRACT

Background: Gastrointestinal stromal tumors (GISTs) are rare mesenchymal neoplasms of the gastrointestinal tract, arising from the interstitial cells of Cajal with characteristic immunomorphological expression by CD117 and DOG-1. Over the past decade, significant technological advancements and improved diagnostic modalities have enabled the detection of these tumors at much earlier stages with better survival outcomes. However in rural areas often these tumors remain under-recognized due to limited access to diagnostics with immunohistochemistry and imaging facilities, thereby resulting in delayed diagnosis and larger sized, high-grade tumors at initial presentation itself. The aim of this study is to analyze the clinicopathological spectrum of GISTs diagnosed at a rural tertiary care center with review of literature.

Materials and Methods: This retrospective observational study was conducted in the Departments of Pathology and General Surgery at our rural tertiary care centre from 2023 to 2025. All cases diagnosed histopathologically and confirmed by immunohistochemistry for CD117 and/or DOG-1 were included in the study. Demographic details, clinical presentation, tumor site, gross morphology, size, histopathological type, and risk stratification were analyzed using descriptive statistics.

Results: Sixteen patients were included in the study (male: female ratio 1.2:1; mean age of presentation 60 years). The most common symptom was abdominal pain (56.3%), and the stomach was the predominant site (37.5%), followed by jejunum (25%). Tumor sizes ranged from 2 cm to >10 cm, with 37.5% tumors exceeding 10 cm of size with high grade features and necrosis. Histologically, 81% were spindle-cell type, and 50% belonged to the high-risk category based on mitotic activity and necrosis. All tumors were positive for CD117 and/or DOG-1.

Conclusion: Our study highlights the diagnostic challenges of GISTs in rural India, where late presentation, large tumor size, and higher histological grade are seen by the time patients presents clinically probably to neglect and lack of awareness of screening programs. Thus, this study underscores the importance of early detection techniques and improved diagnostic access in rural India to mitigate delayed diagnosis of such rare tumors.

Keywords: Rural tertiary care center, GIST, gastrointestinal stromal tumors, mesenchymal neoplasms.

INTRODUCTION

Gastrointestinal (GI) tumors are most commonly epithelial in origin, whereas non-epithelial tumors—derived from mesenchymal, neuroendocrine, or lymphoid cells—are relatively rare and diversified. These tumors include GISTs, leiomyomas, leiomyosarcomas, schwannomas, neuroendocrine tumors (NETs), and lymphomas.[1] Gastrointestinal stromal tumors (GISTs) are the most common type of mesenchymal neoplasms of the gastrointestinal tract, comprising about 1–2% of all gastrointestinal malignancies. These tumors are known to arise from the interstitial cells of Cajal, and typically express CD117 (c-KIT) and DOG-1 on immunohistochemistry.[1,2] These tumors are likely to possess mutations in the KIT gene which enables uncontrolled activation of tyrosine kinase and is responsible for cell mitosis, thus leading to uncontrolled cellular proliferation.[2] With the advent of technology, these lesions can be identified through imaging and endoscopy as submucosal lesions however definitive classification requires histopathological and immunohistochemical confirmation. Despite advances in understanding individual tumor types and comprehensive evaluation techniques, due to limited access in rural healthcare settings, these lesions are often delayed in detection and confirmation thereby, resulting in poorer clinical outcomes.[3] Our study aims to evaluate the clinical presentation, and pathological profile of patients diagnosed with GIST in a rural tertiary care teaching hospital emphasizing diagnostic challenges and various patterns of presentation.

MATERIALS AND METHODS

This retrospective observational study has been conducted in the Departments of Pathology and General Surgery, between 2023 to 2025 for a period of 30months at Sri Madhusudan Sai Institute of Medical sciences and Research, Muddenahalli, Chikkaballapura, Southern India. Cases diagnosed histopathologically as mesenchymal neoplasm with morphological features favoring GIST and confirmed with immunohistochemistry with CD117 and DOG1 have been included in the study. Uncertain biopsy report like inadequate tissue and IHC negative for CD117 and/or DOG1 were excluded from the study.

Relevant data were retrieved from laboratory databases and case records which include demographic details, clinical presentations, anatomical site, gross morphology, histopathology, and immunohistochemistry (CD117 and DOG-1). Data were entered into Microsoft Excel 2019 (Microsoft Corporation, USA) and analyzed using SPSS Statistics. Descriptive statistics such as mean, percentage, and frequency distribution were applied to summarize demographic and clinicopathological variables.

RESULTS

A total of 16 cases were analyzed which comprised 12 cases (75%) of surgical resections and 4 core biopsies (25%). There were 9 male patients (56.3%) and 7 female patients (43.7%), with male to female ratio being 1.2:1. The age range was between 38yr to 80yrs with average age of presentation being 60yrs [Table 1]. Most of the patients presented in the 7th decade (37.5%).

Table 1: Age distribution

Age group	Number	Percentage
20-30yrs	Nil	-
31-40yrs	2	12.5%
41-50yrs	2	12.5%
51-60yrs	3	18.75%
61-70yrs	3	18.75%
71-80yrs	6	37.5%

Most common presenting symptom was abdominal pain, seen in 9 patients (56.3%) with duration of symptoms varying from intermittent episodes spanning over a decade to acute severe presentation of 10 days duration in cases of intestinal obstruction [Table 2].

Table 2: Most common presenting symptoms

Presenting symptoms	Number	Percentage
Abdominal pain	9	56.25%
Weight loss	1	6.25%
Mass abdomen	3	18.7%
Altered bowel habits	1	6.25%
Intestinal obstruction	2	12.5%

Gastric GISTs were more commonly encountered, seen in 6patients (37.5%) comprising 3 cases from fundus, followed by jejunal GISTs (25%)[Table 3]. One female patient had a history of mastectomy for ductal carcinoma of breast, 1year prior to the presentation of acute intestinal obstruction diagnosed secondary to ileal GIST.

Table 3: Anatomical sites

Site	Number	Percentage
Ileal	2	12.5%
Jejunum	4	25 %
Duodenum	1	6.25%
Stomach – Fundus	3	18.75%
Stomach - Pylorus	1	6.25%
Stomach - Body	2	12.5%
Mesenteric mass	2	12.5%
Mesocolon	1	6.25%

Of the 16 cases, 14 were evaluated pre-operatively with CT and/or CECT with diagnosis of GIST [Fig 1]. Two cases, of which one mimicked as ovarian mass / mesenteric mass was suspected to be GIST intra-operatively arising from distal part of stomach measuring 14x12cm.

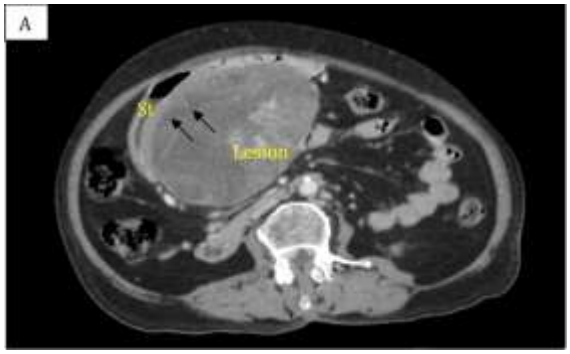

Figure 1A: Gastrointestinal stromal tumour of pylorus of stomach. Heterogeneously enhancing exophytic lesion arising from pylorus of stomach (arrows) and extending into lesser sac. (St- stomach).

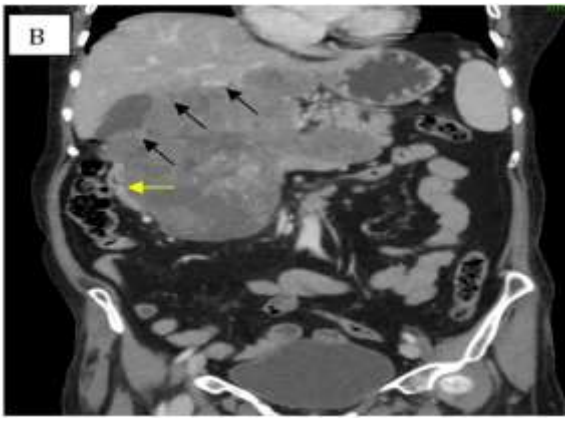

Figure 1B: Extent of gastrointestinal stromal tumour of pylorus of stomach. The lesion shows loss of fat planes with inferior margin of liver and gall bladder (arrow). It is abutting medial wall of second part of duodenum (yellow arrow).

Surgical interventions for GISTs in the present study included resection and anastomosis in 5 patients and subtotal gastrectomy in 5 patients. [Table 4]

Table 4: Types of Surgery.

Types of Surgery	Number	Percentage
Resection anastomosis	5	31.25%
Subtotal Gastrectomy	5	31.25%
Mass resection	1	6.25%
Whipple's resection	1	6.25%
Core biopsy	4	25%

In our study no significant post operative complications were noticed in the cases which underwent resection despite the large tumor size.

4 patients underwent core biopsies for evaluation of mesenteric masses, of which 2 patients had metastatic GISTs and were in-operable whereas the other two patients were lost for follow-up after diagnosis.

All the specimen/ core biopsies were submitted to histopathological examination with routine hematoxylin and eosin stain followed by immunohistochemistry with a panel of markers and all cases were positive for CD117 and /or DOG1.

Regarding the size of the tumor in the resected specimen, majority had a size ranging between 2-5cm (50%) followed by tumors ranging in size more than 10cm (37.5%) with a largest tumor measuring 20x16cm arising from jejunum. [Table 5 & Figure 2]. The mean size of the tumor in our study is 7.9cm.

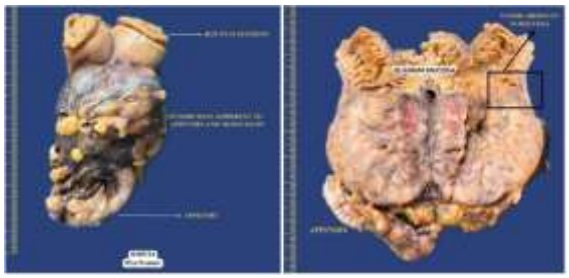

Figure 2: Gross picture of Jejunal GIST in a 65 yr female.

Table 5: Size of tumor

Size	Number	Percentage
<2cm	Nil	
2-5cm	08	50%
5-10cm	02	12.5%
>10cm	06	37.5%

Most common histopathological type was spindle cell type seen in 13 cases (81.25%) including core biopsies, where as one case (6.25%) was diagnosed as epithelioid GIST and two cases of GIST (12.5%) showed mixed morphological features, comprising both spindle and epitheliod type [Figure 3]

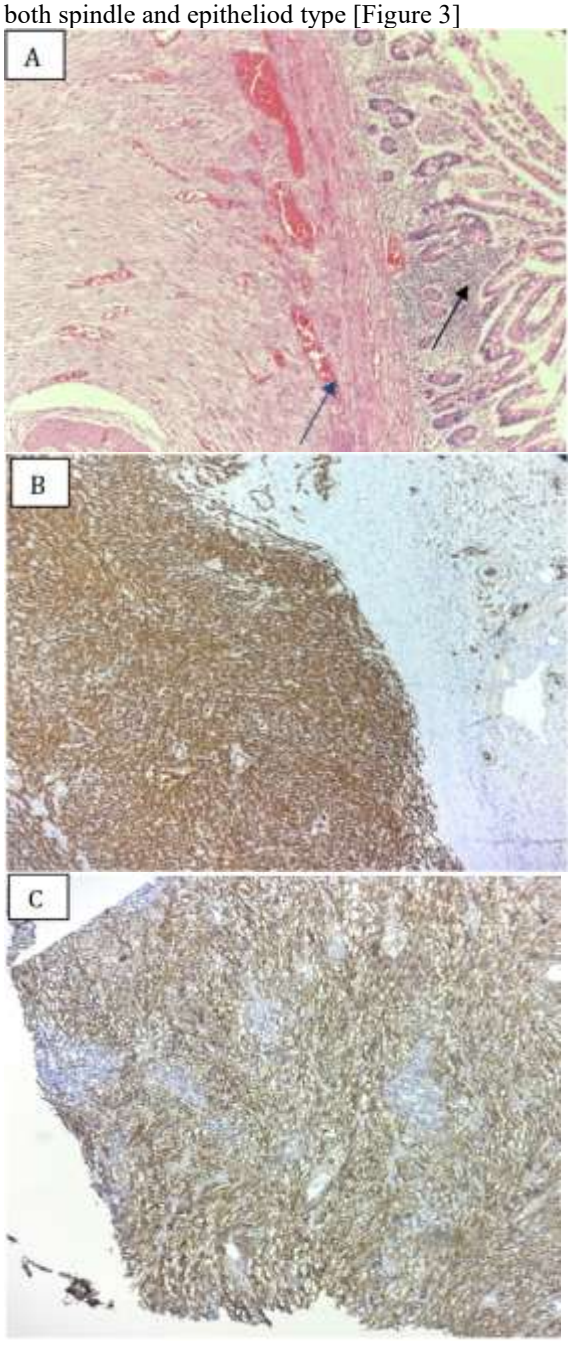

Figure 3: [A] Jejunal GIST, Jejunal mucosa(arrow) with submucosal spindle cell tumor (blue arrow) (H&E 10x). [B] Diffuse cytoplasmic to membranous positivity for CD117 (10x). [C] Diffuse membranous positivity in tumor cells for DOG1 (10x)

From the histopathological examination based on mitotic activity, site and size of tumors risk grading for surgically resected specimen was done (12 cases), excluding the core biopsies, high risk group were more prevalent i.e. 6 patients (50%) followed by 3 cases for intermediate group (25%) and 3 cases for low risk group (25%).

DISCUSSION

Gastrointestinal stromal tumors (GISTs) are rare mesenchymal neoplasms of the gastrointestinal tract(GI) arising from the interstitial cells of Cajal representing 0.52% of all GI malignancies with an incidence of 20,000 yearly worldwide and a prevalence of 10–20/1,000,000.[1,2] The term 'GIST' was coined by Mazur et al.; however, it was the pioneering work of Hirota et al. that recognized these tumors as a distinct pathological entity inview of their specific immunomorphological characteristics such as KIT (CD117), DOG1 expression and PDGFR mutations.[3,4] The past decade has witnessed significant advances in diagnostic modalities for GIST with better treatment regime, notably with tyrosine kinase inhibitors like imatinib, leading to improved survival outcomes. Despite this progress, these tumors remain undetected in early stages due to their diverse presentation in biological behavior and low incidence, there by lacking a universally accepted staging system. Very few limited studies are available in Asian literature reflecting the demographic and clinical features of these rare tumors especially from rural areas. In addition, we have noted that in our study there were 16 cases diagnosed and treated within a span of 2years highlighting their increased prevalence associated with unique challenges in rural settings. These include delayed diagnosis due to remote healthcare access and limited diagnostic infrastructure as many of our patients presented in late stages of disease, and few presenting with distant metastasis at the time of initial diagnosis itself.

The most common age group affected in our study was 71-80yrs (37.5%) which is slightly higher than the reported literature; however the mean age of presentation is 60yrs which is similar to other studies reported from India.[5,6] The youngest patient in our study was a 38 year old male, and the oldest patient was an 80 year old female. There were 9 male patients (56.3%) and 7 female patients (43.7%), with male to female ratio being 1.2:1 which is similar to the findings reported by Pandya.N et al.[5]

GISTs are often asymptomatic and can be discovered most of the times as incidental findings on imaging studies or endoscopies performed for other indications, as it is sub mucosal in location.[7] They are highly vascular, soft, and friable, therefore GI bleeding can be a common presenting symptom. Other symptoms may include abdominal pain, palpable lump in abdomen, and distention due to obstruction. However in our study 9 patients (56.3%) presented with chronic pain abdomen with duration of symptoms varying from intermittent episodes spanning over a decade to acute severe presentation of 3-7days in cases of intestinal obstruction. This is similar to the findings reported by Sethi SK et al.[8] However Li J et.al in his study of 112 cases reported an equal incidence of abdominal pain and upper gastrointestinal bleeding in 31.3% of cases.[9]

The most common site for GIST in our study was stomach, particularly in the fundus, followed by jejunal GISTs, which is in concordance with the literature especially seen in Asian populations.[10] A notable aspect in our study is the high rate of preoperative diagnostic accuracy achieved using advanced imaging modalities such as contrast-enhanced computed tomography (CECT) which plays a pivotal role in diagnosis, staging, and surgical planning of these tumors. GISTs typically appear as well-defined, enhancing soft-tissue masses, where larger tumors can be heterogeneous, associated with areas of necrosis or hemorrhage due to their vascular nature. These findings are consistent with the recent European and Indian data emphasizing the role of imaging in resource limited rural settings where it can serve as a primary non-invasive diagnostic tool, especially when endoscopic ultrasound or MRI is unavailable.[11] Surgical management for all the cases included in the study was resection and anastomosis of the involved segment of small bowel, Whipple procedure for duodenal GIST and subtotal gastrectomy for tumors arising from the stomach.

There were no significant postoperative complications in the patients who underwent surgical intervention, which is comparable to other studies in Indian literature reporting favorable outcomes with timely, focused surgical intervention.[12,13] Most of the tumors in our study ranged between 2-5cm (50%) followed by tumors ranging in size exceeding 10 cm, with areas of necrosis and high grade features. Similar observations were reported by Singh P et al., attributing to late clinical presentation, advanced tumor biology, and limited access to adjunctive therapies.[11]

Also in the present study majority of tumors were high or intermediate risk based on mitotic activity and size, necessitating aggressive surgical intervention and close postoperative surveillance.[11-13]

By documenting the clinicopathological spectrum and emphasizing the predominance of large, high-grade tumors in our rural population, our study emphasizes the urgent need to enhance community awareness, strengthen diagnostic capacity in peripheral laboratories, and establish integrated referral networks to mitigate diagnostic delays and improve clinical outcomes.

The limitations of our study include smaller sample size (n = 16) limiting the broader relevance of its findings with a single centre experience. The details of follow-up, survival outcomes, and response to adjuvant therapy (such as imatinib) could not be assessed as the patients were referred to higher oncology centers after surgery. Another drawback was the two-year study duration, which is insufficient to assess long-term presentation patterns, recurrence rates, and prognostic outcomes. Future multicentric studies with larger cohorts and extended follow-up are recommended to strengthen the conclusions.

CONCLUSION

In summary, the findings in our study likely reflect diagnostic delays and advanced disease stage at initial presentation possibly due to lack of awareness of early cancer symptoms and limited access to comprehensively equipped healthcare facilities. Early imaging with CECT can contribute significantly to timely diagnosis and appropriate surgical or medical management of GISTs. Despite these limitations, favorable surgical outcomes in operable cases indicate that timely surgical management remains achievable when sufficient diagnostic support is available. To conclude our study contributes valuable insights into the rural oncology burden and sets the foundation for future multicentric research aimed at early detection and equitable access.

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