Case Series

Clinicopathological Spectrum of Gastrointestinal Stromal Tumours: A Case Series

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Abstract

Gastrointestinal stromal tumors (GISTs) are rare neoplasms of the gastrointestinal tract, accounting for less than 1% of all primary GIT tumors. In this small sized series, GISTs were identified in both the stomach and small intestine, highlighting the anatomical variability of these tumors. The primary objective of this case series was to characterize the clinicopathological features, histomorphology, immunohistochemical profile, and risk stratification of GISTs. Four patients diagnosed with GIST between January 2024 and December 2024 at a tertiary care hospital in Gujarat, India, were included in the study. The patients were adults aged 37–58 years. Spindle cell morphology was the predominant histological pattern observed, consistent with the typical morphology of GISTs. The novelty in this case series highlights the diagnostic utility of histomorphology and immunohistochemistry in resource-limited settings and underscores the importance of risk stratification in guiding management.

Keywords: gastrointestinal stromal tumor; GIST; tyrosine kinase inhibitor; KIT mutation; PDGFRA mutation

Introduction

Gastrointestinal stromal tumors (GISTs) are rare neoplasms of the gastrointestinal tract, accounting for less than 1% of all primary GIT neoplasms.[1] Contrary to some literature that suggests the stomach is the most common site for these tumors, our findings indicate that both the stomach and small intestine are equally involved.[2] GISTs are mesenchymal tumors originating from interstitial cells of Cajal or pacemaker cells, which are a precursor stem cell population located within the myenteric plexus.[3] The average age of diagnosis is around 60 to 65 years, with a nearly equal occurrence in both males and females.[4] These tumors can develop anywhere in the gastrointestinal tract, most frequently in the stomach, followed by the small intestine, colon, rectum, esophagus, and rarely in the appendix. Extra-intestinal GISTs may arise in the mesentery, omentum, or retroperitoneum. Clinically, they can present as indolent cases or with symptomatic courses, and they may be detected incidentally through radiology, endoscopy, or as abdominal masses.[5] GISTs typically harbor activating mutations in the KIT gene (c-KIT), which encodes a receptor tyrosine kinase, as well as mutations in the PDGFRA gene (platelet-derived growth factor receptor alpha). These mutations are a target for medical therapy with tyrosine kinase inhibitors (TKIs).[6] The primary treatment for GISTs is surgery, with medical management available as a complement. The prognosis for patients depends on factors such as tumor size, location, the presence of mitotic activity, and necrosis. Immunohistochemical analysis typically shows that GISTs express markers such as CD117, DOG1, and CD34.[7]

Table 1: Risk stratification according to modified Miettinen and Lasota's algorithm.

Mitotic Index	Size (cm)	Stomach	Small bowel
<5/5 mm^2	≤ 2	Very low	Very low
	>2 and $\leq$5	Very low	Low
	>5 and $\leq$10	Low	Moderate
	>10	Moderate	High
$\geq$5/5 mm^2	≤ 2	Very low	Moderate
	>2 and $\leq$5	Moderate	High
	>5 and $\leq$10	High	High
	>10	High	High

Case Series

Total 4 cases of GIST were studied for clinicopathological, histomorphology and IHC expression with their risk stratification. Surgical specimens were received in pathology department of hospital, routinely processed in paraffin and stained with H and E stain. They were reported by pathologists with reference to 8th edition AJCC (American Joint Committee on Cancer)[8]. Risk stratification was done according to modified Miettinen and Lasota's algorithm (table 1).[7] All demographic and clinical details were obtained from case records. Immunohistochemistry was carried out on an automated Ventana Benchmark XT platform using antibodies against CD117 (polyclonal), DOG-1 (SP31), CD34 (QBEnd/10), SMA (1A4), Desmin (D33), S-100 (polyclonal), Cytokeratin AE1/AE3, and Chromogranin A (LK2H10). Four patients diagnosed with GIST between January 2024 and December 2024 at a tertiary care hospital in Gujarat, India, were included in the study. Ethical approval from institute was not obtained as it was retrospective case analysis. This case series highlights the diagnostic utility of histomorphology and immunohistochemistry in resource-limited settings, underscores the importance of risk stratification in guiding management and thus exhibiting its novelty in guiding for small sized, resource limited laboratories.

Case 1

A 39-year-old man presented with upper abdominal pain of two days' duration and a single episode of vomiting fifteen days earlier. Examination revealed abdominal distension with localized upper abdominal tenderness. Ultrasonography showed a distended stomach and small bowel loops, with a well-defined hypoechoic, vascular lesion measuring 56 $\times$ 34 mm in the right upper and mid-abdomen, associated with mild ascites. Exploratory laparotomy was performed, and a jejunal segment containing the lesion was resected.

Grossly, the tumor measured 4.8 $\times$ 4.5 $\times$ 3.0 cm, with a greyish-white cut surface showing areas of necrosis, haemorrhage, and mucosal ulceration. Histopathological examination revealed a spindle cell neoplasm arranged in fascicles and whorls, with minimal pleomorphism, low mitotic activity (2–3 per 5 mm^2), and approximately 30% necrosis. Immunohistochemistry showed strong positivity for CD117 and DOG-1, patchy SMA positivity, negativity for CD34, and retained SDHB expression. A diagnosis of low-risk, spindle cell–type gastrointestinal stromal tumor of the jejunum was made. The patient was advised routine follow-up, with no adjuvant therapy indicated.

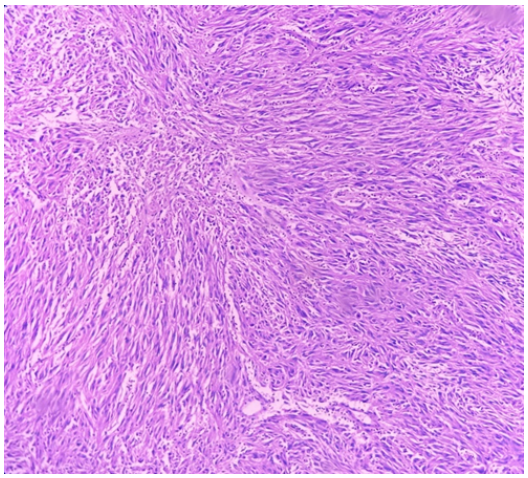

Figure 1: GIST showing spindle cells arranged in interlacing fascicles and whorls, low grade, jejunum. (H & E, 20x)

Case 2

A 58-year-old man presented with recurrent hematemesis for two months. Upper gastrointestinal endoscopy revealed a submucosal bulge along the lesser curvature of the stomach with intact mucosa. Contrast-enhanced CT showed a lobulated gastric mass in the gastrohepatic space measuring 7.2 × 4.8 cm with internal cystic areas, suggestive of a neoplastic lesion. The patient underwent wedge resection.

Grossly, the tumor measured 7.8 × 6.0 × 5.0 cm and was solid-cystic. Histopathology revealed an epithelioid-type gastrointestinal stromal tumor, Grade 2 (high grade), with a mitotic rate of 6–7 per 5 mm^2 and no necrosis. Immunohistochemistry showed positivity for CD117, DOG-1, CD34, and vimentin, with negativity for desmin and smooth muscle actin, confirming the diagnosis. Based on tumor size and mitotic activity, the tumor was classified as high risk, and the patient was advised adjuvant imatinib therapy with close follow-up.

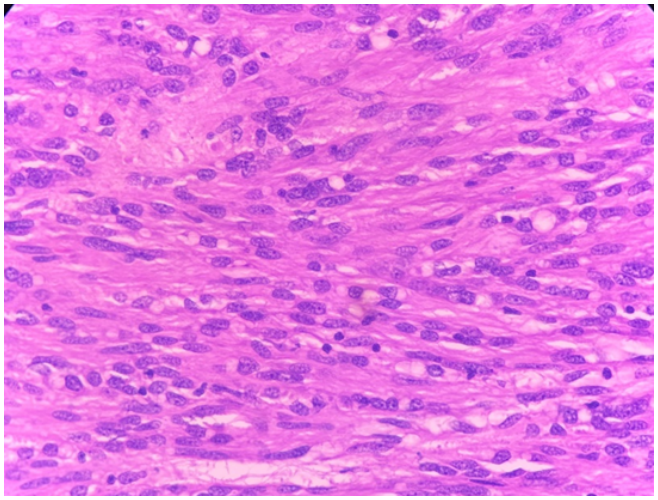

Figure 2: Epithelioid variant of GIST, high grade, stomach. (H & E, 40x view)

Case 3

37-year-old female presented with a one-year history of intermittent abdominal pain. On examination, the abdomen was soft and non-tender. Ultrasound revealed a 8.0 × 8.0 cm hypoechoic lesion in the left mid-abdomen near the umbilicus. CT abdomen showed a 9.5 × 7.2 × 5.5 cm heterogeneously enhancing exophytic mass with internal necrosis in the central mesentery and left hypochondrium, abutting jejunal loops and the inferior aspect of the pancreatic tail. Differentials included GIST and SPEN (Solid pseudopapillary Epithelial Neoplasm-Pancreas). The mass was excised along with part of the jejunum. Grossly, it measured 8.0 × 6.0 × 3.9 cm, with a solid greyish-brown cut surface, haemorrhage, and a small cystic area. Histopathology confirmed GIST, mixed spindle and epithelioid type, Grade 2 (high grade), with a mitotic rate of 7–8/5 mm^2 and focal necrosis, categorizing it as high risk. IHC was positive for CD117, DOG-1, S-100 weak positive and negative for CD34, confirming GIST. The patient was advised adjuvant Imatinib therapy and regular follow-up.

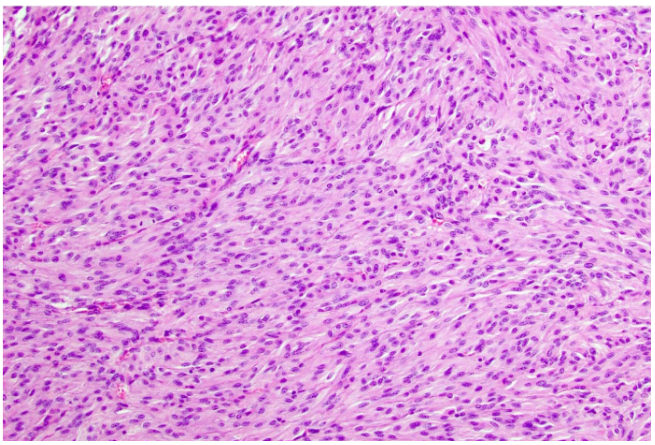

Figure 3: GIST with spindle and epithelioid cells morphology, high grade, jejunum. (H & E, 20x view)

Case 4

A 47-year-old male presented to the surgery outpatient department with a 12-day history of weakness, vertigo, and black stools. Contrast-enhanced CT of the abdomen revealed a well-defined, heterogeneously enhancing lobulated lesion measuring 3.1 × 2.9 × 2.8 cm in the submucosal region of the greater curvature of the distal stomach, suggestive of a benign neoplasm, likely a gastrointestinal stromal tumor (GIST). The patient underwent surgical resection of the affected gastric segment. Gross examination showed a tumor measuring 1.3 × 0.5 × 0.4 cm. The difference between radiological and gross tumour size is likely due to post-excision collapse and fixation-related tissue shrinkage. Histopathology confirmed a spindle cell type GIST, Grade 1 (low grade), with a low mitotic rate (0–1 mitoses/5 mm^2) and no necrosis. Immunohistochemistry was positive for CD117, DOG-1 and S-100 weak positive and negative for cytokeratin and chromogranin A, ruling out carcinoma and neuroendocrine tumors. The tumor was classified as very low risk, and the patient was advised routine follow-up without the need for adjuvant therapy.

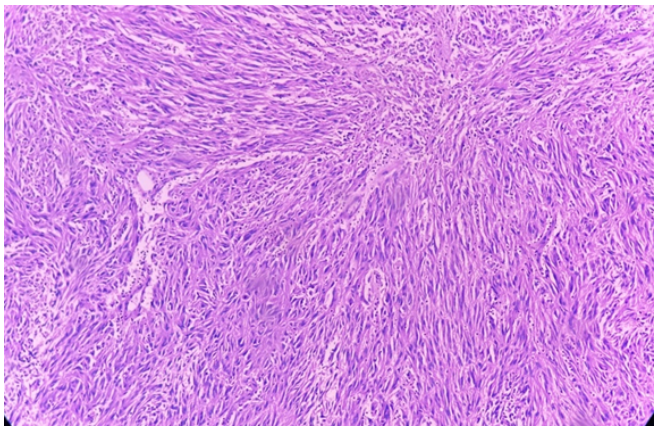

Figure 4: GIST showing spindle cells arranged in interlacing fascicles and bundles with eosinophilic cytoplasm, low-grade, stomach. (H & E, 10x view)

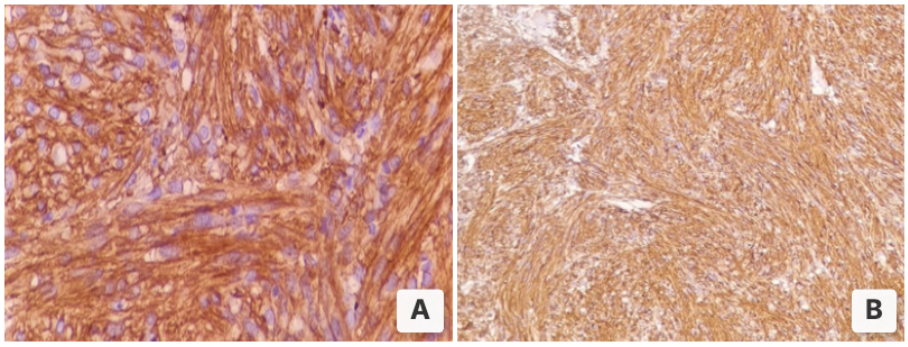

Figure 5: CD117 (c-KIT) positivity – epithelioid type GIST (Case 2, left) and spindle cell type GIST (Case 1, right) showing strong cytoplasmic positivity.

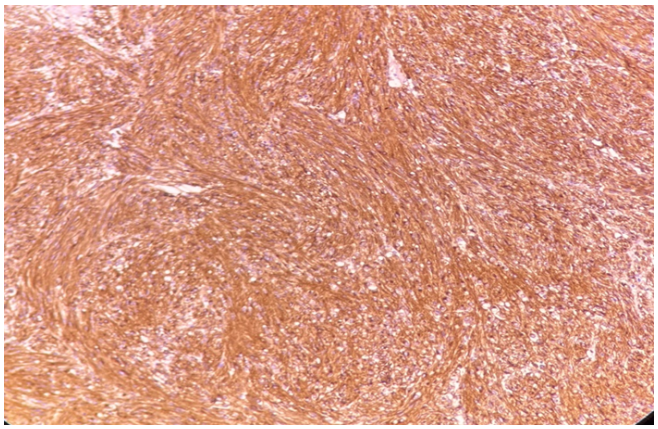

Figure 6: DOG-1 positivity showing strong cytoplasmic staining in tumour cells. (Case 3)

Discussion

GISTs are uncommon mesenchymal neoplasms of the gastrointestinal tract, representing less than 1% of all primary gastrointestinal tumours.[9] They demonstrate wide heterogeneity in clinical presentation, anatomical location, histomorphology,

Table 2: Clinicopathological spectrum with modified Miettinen and Lasota risk stratification algorithm for gastrointestinal stromal tumors based on tumor size, anatomical site, and mitotic index. (total cases=4)

Case	Age (years)	Sex	Presenting Symptoms	Location	Tumor Size (cm)	Histopathology	Mitotic Rate (/5 mm^2)	Grade
1	39	Male	Upper abdominal pain, vomiting	Jejunum	4.8 × 4.5 × 3.0	Spindle cell	2–3	Grade 1 (Low)
2	58	Male	Hematemesis	Stomach (lesser curvature)	7.8 × 6.0 × 5.0	Epithelioid	6–7	Grade 2 (High)
3	37	Female	Intermittent abdominal pain (1 year)	Jejunum/ mesentery	8.0 × 6.0 × 3.9	Mixed spindle & epithelioid	7–8	Grade 2 (High)
4	47	Male	Weakness, vertigo, black stools	Stomach (greater curvature)	1.3 × 0.5 × 0.4	Spindle cell	0–1	Grade 1 (Very low)

Table 3: Case wise analysis of histopathological features (n=4).

Present study	Tumour type			Tumour grade		Mitotic count (per 5 mm^2)	
	Spindle	Epithelioid	Mixed	Low	High	≤5	≥5
No. of cases	02	01	01	02	02	02	02

Table 4: Case wise risk assessment, IHC features and surgical management.

Case	IHC Findings	SDHB	Risk Category (Miettinen & Lasota)	Management
1	CD117+, DOG-1+, CD34−, SMA patchy +	Retained	Low risk	Surgical resection, follow-up
2	CD117+, DOG-1+, CD34+ Vimentin+, Desmin−, Actin−	Not done	High risk	Surgery + adjuvant Imatinib
3	CD117+, DOG-1+, S-100 Weak +, CD34−	Not done	High risk	Surgery + adjuvant Imatinib
4	CD117+, DOG-1+, Vimentin+, S-100 Weak +, Cytokeratin−, Chromogranin−	Not done	Very low risk	Surgical resection, follow-up

and biological behavior, as illustrated in the present case series.

Symptoms may vary from asymptomatic to acute abdomen presentation depending on tumour location and size.[10] Consistent with published literature, the stomach and small intestine were the most frequently involved sites in our series.[7] It commonly occurs in 50–60 years age group with rarity in paediatrics. Grossly tumour are well circumscribed with tan-white cut surface, haemorrhage, necrosis and cysts.[11] Histomorphologically, most important cell of origin in GIST is spindle cells (70%), epithelioid cells (20%) and remaining 10% are mixed cell type.[12] Differential diagnosis of submucosal GIT masses are leiomyoma, GIST, schwannoma, solitary fibrous tumour, neuroendocrine tumour.[7] Immunohistochemistry played a pivotal diagnostic role, with universal expression of CD117 and DOG-1, reaffirming their high sensitivity and specificity.[13] Tumor size and mitotic index emerged as the most critical prognostic factors, guiding risk stratification and therapeutic decisions. Low- and very low-risk tumors were effectively managed with complete surgical resection alone, whereas high-risk tumors warranted adjuvant imatinib therapy, in line with current consensus guidelines.[14, 15]

In the present study, the age of patients ranged from 37 to 58 years, with a median age of 43 years, which is slightly younger than that reported in the literature (commonly 55–65 years). This difference may be attributable to the small sample size of the study. A male predominance was observed, consistent with several Indian studies reporting a slight male preponderance.[16]

Interestingly, unlike most published series in which the stomach accounts for approximately 60–70% of cases, the stomach and small intestine were frequently involved in the present study. This finding suggests that intestinal GISTs may not be uncommon in the study population and highlights the importance of thorough evaluation of submucosal gastrointestinal masses irrespective of location. Histomorphologically, spindle cell type was the most common pattern (two cases), followed by epithelioid (one case) and mixed spindle–epithelioid morphology (one case). This predominance of spindle cell morphology is in agreement with published data indicating that nearly 70% of GISTs exhibit spindle cell features.

Table 5: Comparison of demographic and clinicopathological features of GIST with various studies.

Author	Country	Median Age (years)	Sex (M:F)	Predominant Symptom	Most Common Location	Mean/Median Tumour Size (cm)
Alqusous ST et al. (2016)	Jordan	56.8	Male (56.8%)	Abdominal pain	Stomach	8.2
Ud Din N et al. (2015)	Pakistan	53	Male (59.2%)	Abdominal pain / GI bleeding	Stomach	10.0
Gupta A et al. (2021)	India	52	Female (57.1%)	Abdominal pain	Stomach and small bowel	6.5
Sengupta R et al. (2020)	India	56	Male (57.4%)	Abdominal mass	Small bowel	7.2
Minhas S et al. (2016)	India	54	Not specified	Abdominal pain	Stomach	Not specified
Jumniensuk C & Charoenpitakchai M (2018)	Thailand	55	Male (50.0%)	GI bleeding	Stomach	8.0
Gaopande VL et al. (2016)	India	51	Female (58.1%)	Abdominal pain	Stomach	7.0
Present Study	India	–	Male (75%)	Abdominal pain, hematemesis, black stools	Stomach (2), Small intestine (2)	6.3

Tumor size ranged from 1.3 cm to 9.5 cm, with the largest tumor arising from the mesenteric/jejunal region. Areas of necrosis and hemorrhage were identified in two cases and were associated with higher-grade tumors. Mitotic activity varied from 0–1 per 5 mm^2 in low-grade tumors to 7–8 per 5 mm^2 in high-grade tumors, underscoring the mitotic index as a critical prognostic parameter.[7]

Immunohistochemically, all cases demonstrated strong positivity for CD117 (c-KIT), confirming the diagnosis of GIST, with DOG-1 also showing strong expression, supporting its diagnostic specificity. CD34 expression was variable, being positive in one case and negative in two, comparable to reports in the literature where CD34 positivity ranges from 60–70%. Vimentin positivity in tested cases further supported the mesenchymal origin of these tumors. Weak or focal S-100 positivity, as observed in Cases 3 and 4, has been reported in a small subset of GISTs and does not preclude the diagnosis. In such cases, strong CD117 and DOG-1 expression is considered more specific and diagnostically decisive, while S-100 was included primarily to exclude schwannoma and other neural tumors in the differential diagnosis. Retained SDHB expression in one case excluded SDH-deficient GIST, a distinct subset typically seen in younger patients with unique clinical behavior.

Using the modified risk stratification criteria proposed by Miettinen and Lasota, two cases were classified as high risk, one as low risk, and one as very low risk. Risk categorization correlated well with tumor size and mitotic activity.[7] Both high-risk tumors were larger than 5 cm with elevated mitotic rates and arose from the stomach and jejunum, respectively; these patients were initiated on adjuvant imatinib therapy. In contrast, low- and very low-risk tumors were managed surgically with close follow-up and no adjuvant therapy. In all cases, molecular testing for KIT and PDGFRA mutations is recommended for prognostication and therapeutic guidance but it could not be performed due to financial constraints of patients, lack of in-house molecular diagnostic facilities, and limited access to referral laboratories. Overall, this series emphasizes that even in resource-limited settings where molecular testing for KIT or PDGFRA mutations may not be routinely available, careful histomorphological assessment combined with immunohistochemistry remains pivotal for accurate diagnosis, risk stratification, and therapeutic decision-making. The integration of morphological features, mitotic index, and immunophenotype is essential in identifying patients who may benefit from targeted tyrosine kinase inhibitor therapy.

Limitations include the study sample is very small and retrospective in nature. Although molecular testing for KIT and PDGFRA mutations is recommended for prognostication and therapeutic guidance, it could not be performed in the present study due to financial constraints, lack of in-house molecular diagnostic facilities, and limited access to referral laboratories. In high-risk cases, treatment decisions were therefore guided by established clinicopathological risk stratification criteria.

Conclusion

This four-case series illustrates the heterogeneity of gastrointestinal stromal tumours with respect to site, morphology, and risk profile. The stomach was the most frequent location, and spindle cell morphology predominated, while the small

intestinal GISTs in this series showed epithelioid morphology and higher risk features. Immunohistochemistry using CD117 and DOG-1 proved essential for diagnosis in the absence of advanced molecular testing. All patients were managed surgically, with adjuvant targeted therapy reserved for high-risk disease. These cases emphasize the importance of correlating clinical, histopathological, and immunohistochemical findings for accurate diagnosis, risk stratification, and appropriate management of GISTs.

Abbreviations: GIST: Gastrointestinal stromal tumor; GIT: Gastrointestinal tract; IHC: Immunohistochemistry; TKI: Tyrosine kinase inhibitor; AJCC: American Joint Committee on Cancer; SDHB: Succinate dehydrogenase subunit B; SMA: Smooth muscle actin; SPEN: Solid pseudopapillary epithelial neoplasm; CT: Computed tomography; DOG-1: Discovered on GIST-1; CD: Cluster of differentiation; PDGFRA: Platelet-derived growth factor receptor alpha.

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