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# Gastrointestinal Stromal Tumor (GIST): About 3 Cases

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***Authors' contributions***

*This work was carried out in collaboration among all authors. All authors read and approved the final manuscript.*

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***Case Report***

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## ABSTRACT

Gastrointestinal stromal tumors (GISTs) are mesenchymal tumors that typically occur in the gastrointestinal tract. About 70% of GISTs are benign, while approximately 30% are malignant. GISTs represent less than 0.1% to 3% of all gastrointestinal tumors. These tumors are thought to originate from intestinal cells of Cajal (ICCs) or stem cell-like precursors. ICCs, often referred to as the "pacemaker" cells of the GI autonomic nervous system, regulate GI motility and are found around the myenteric plexus and the muscularis propria throughout the GI tract.

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GISTs are composed of different types of cells: approximately 70% are spindle cells, 20% are epithelioid cells, and 10% are mixed cells. Around 85% of GISTs contain mutations in one of two receptor tyrosine kinases (RTKs), KIT or PDGFRA, which play a central role in the pathogenesis of these tumors. Additionally, about 95% of GISTs test positive for the CD117 antigen.
Clinically, GISTs are most commonly found in the stomach (60%), followed by the small intestine (30%), duodenum (5%), rectum (3%), colon (1%), and oesophagus (0.5%). Occurrences in the omentum and mesentery are rare. GISTs affect men and women equally and most commonly occur between the ages of 50 to 70 years. The annual incidence rate is between 6.5 and 14.5 per million people worldwide.
In our study, we report three cases of GISTs located in the duodenum, jejunum, and transverse colon. Diagnosis in these cases was confirmed via CT scan and all cases tested positive for the CD117 antigen.

*Keywords: Gastrointestinal stromal tumors; CD 117 C-kit; duodenal GIST; jejunal GIST; transverse colon GIST.*

## 1. INTRODUCTION

Gastrointestinal stromal tumors (GISTs) comprise less than 1% of all gastrointestinal (GI) tumors and are the most common mesenchymal tumors. Globally, GISTs are most prevalent in China, Taiwan, Korea, Japan, and Norway. [1,2,3]. These tumors can range in size from less than 1 cm to more than 40 cm, with an average size of 5 cm at the time of clinical diagnosis [4,5,6].

The vast majority of GISTs are sporadic, but 5-10% occur in association with the following conditions:

1. Carney triad (GIST, paraganglioma and pulmonary chondroma)
2. Carney-Stratakis syndrome (GIST and paraganglioma)
3. Neurofibromatosis type 1
4. Familial GISTs, which involve germline mutations in C-KIT or PDGFRA and often present withmultiple tumors [5,7].

**Types of GIST:**

1. Benign GISTs (70%): These are usually not life-threatening and are typically smaller than 2 to 5 cm.
2. Malignant GISTs (30%): These cancerous tumors can invade nearby tissues within the abdomen and spread to other parts of the body, such as the liver and lungs, through lymphatic or blood transmission [4,6].

**Symptoms of GISTs include:**

- Abdominal pain
- A lump or mass in the abdomen
- Blood in the stool or vomit
- Anaemia and fatigue due to chronic bleeding
- Loss of appetite or weight loss

**Diagnosis:** The following tests may be used to diagnose or rule out GISTs:

1. Blood tests, such as a complete blood count and blood chemistry tests.
2. Endoscopy, including upper GI endoscopy for oesophageal and stomach GISTs, and colonoscopy for colonic lesions.
3. CT scans, which typically show a soft tissue mass with central areas of necrosis and smoothmargins.
4. MRI and PET scans, which are procedures used to evaluate malignant tumors.
5. Endoscopic ultrasound (EUS), which is the most accurate method for diagnosing submucosal lesions in the oesophagus and stomach.
6. Pathological diagnosis through biopsy, identifying mesenchymal neoplasm with spindle cells or epithelioid cells, and positive for KIT (CD117). Approximately 97% of GISTs are immunohistochemically positive for KIT (CD117) [8,9,10].

**CT Imaging Classification:**

1. Small GIST: less than 5 cm
2. Intermediate GIST: 5 to 10 cm.
3. Large GIST: more than 10-20 cm[11].

## 2. CASE PRESENTATIONS

### 2.1 Case 1- Duodenal GIST

A 50 year-old male was admitted to our center on 20/10/2009, with complains of pain in abdomen

and palpable lump in the epigastric region of the abdomen. Patient was evaluated for intra abdomen lump. His CT abdomen revealed 8x6 cm, spherical mass, located at 2nd part of duodenum, it was heterogenic mass with central necrosis and having smooth margins.

On physical examination lump was having little mobility in transverse direction. There was no obstructive jaundice. All laboratory investigation was normal. Open laparotomy revealed as well-defined, smooth marinated tumor at 2nd part of duodenum measuring a size 8x6 cm. The growth was exophytic in nature, so we performed complete excision of the mass, without opening the lumber of duodenum. We performed total wide resection with primary closure.

On gross specimen of 8x6 cm, spherical in shape with smooth margin. Histopathologically tumor was constituted by spindle cell with low nuclear pleomorphism and tumor was positive for CD 117 antigen test. Post operatively patient was discharged on 10th postoperative day without any complication follow up of 5 years, patient was healthy and there was no recurrence. The patient oral imatinib 400 mg per day for 2 years achieves curative results (Figs. 1-8).

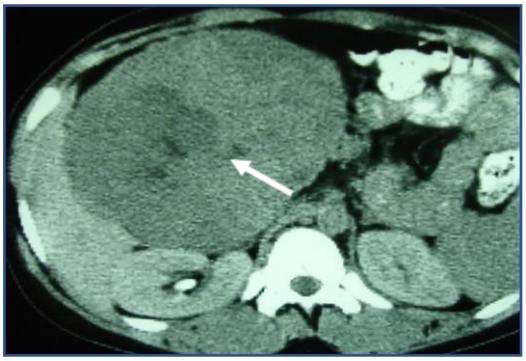

**Fig. 1. CT abdomen showing a 8x6 cm spherical mass, heterogenous and smooth margins**

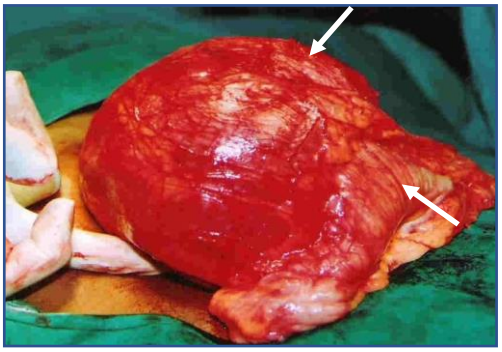

**Fig. 2. Intraoperative photograph showing exophytic growth at second part of duodenum**

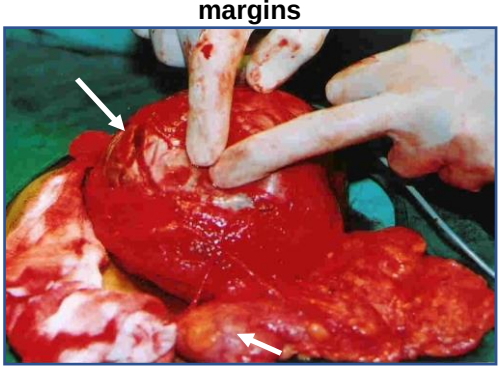

**Fig. 3. Intraoperative photographs showing spherical mass**

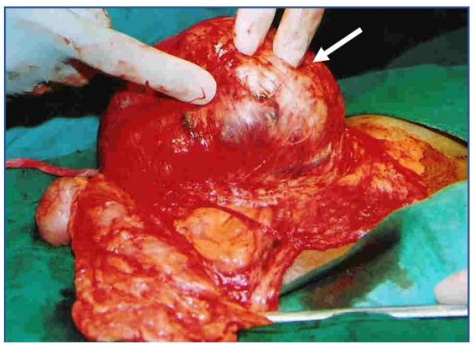

**Fig. 4. Intraoperative photographs showing excision of mass**

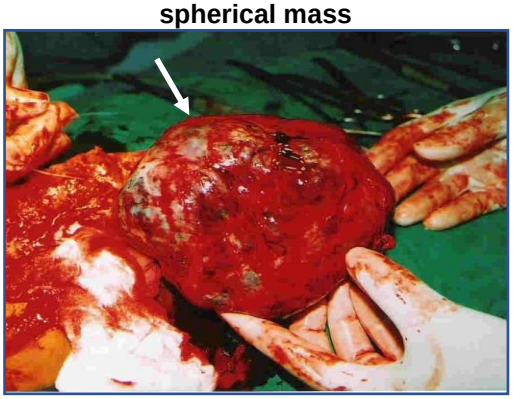

**Fig. 5. Intraoperative photographs showing total excision of cystic mass**

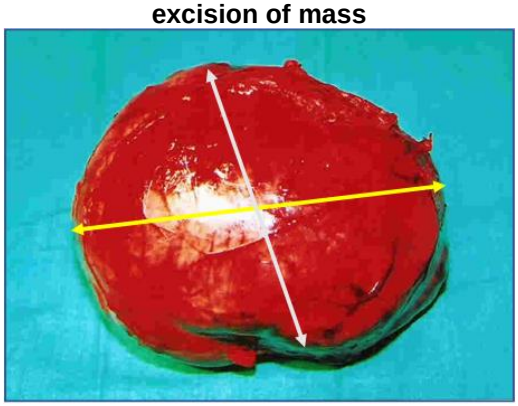

**Fig. 6. Photographs showing total excision of cystic mass of size 8x6 cm**

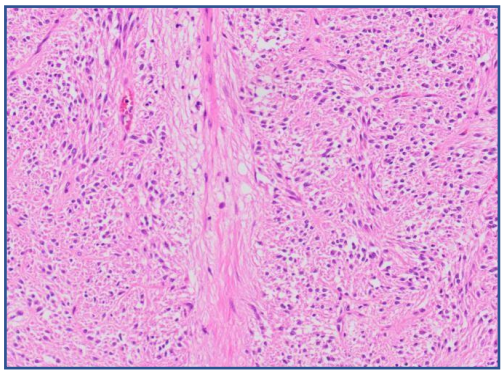

**Fig. 7. Tumor with spindle cells with low mitotic ratio**

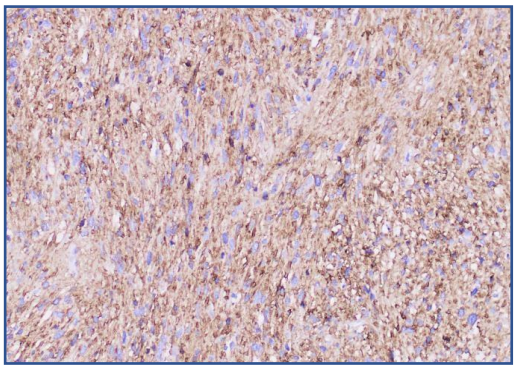

**Fig. 8. Tumor cells with diffuse CD 117 positivity**

## 2.2 Case 2 – Spontaneous Perforation of Jejunal GIST

We report a 55 years old male who presented to our center on 01/01/2012, with severe abdominal pain, distended and tender abdomen. Plain x-ray abdomen gas under diaphragm, abdominal ultra-sonography revealed duodenal perforation with free fluid in the abdomen.

On physical examination heart rate 110/mints, tachycardia, respiratory rate 22/minute and BP 130/90. Abdominal examination revealed abdominal distension tender and abdomen sings of perforation peritonitis. All laboratory investigation normal with raised leukocyte count.

We concluded the diagnosis of duodenal perforation and decided to do diagnostic laparoscopy first. To our surprise there was no duodenal perforation. Upon entrance to the peritoneal cavity, a gush of turbid, free intra peritoneal fluid mixed with enteric content come out. We decided to perform explorative laparotomy. Examination of the abdomen revealed the presence of a 3x2 cm polypoid mass with central perforation into the proximal jejuna 25-30 cm away from the duodeno-jejunal junction or ligamentum tares. The growth had a central perforation. All other organs were normal. The growth was resected with free 5 cm margin and end to end anastomosis was performed. Through peritoneal lavage was done with saline wash and abdomen closed after putting the drainage tube was placed in the right flank. Post-operative period was uneventful and the patient was discharged on the 10$^{th}$ post-operative day after stich removal.

Histopathological of the resected specimen showed, as submucosal nodular tumor composedly of spindle shaped cells with mild nuclear pleomorphism and 5 mitotic figures 50 high power fluids. Surgical lines of resection were free. Immune histochemistry study revealed positive for CD 117 and diagnosis confirmed a jejunal GIST. The patient oral imatinib 400 mg per day for 2 years achieves curative results (Figs. 9-16).

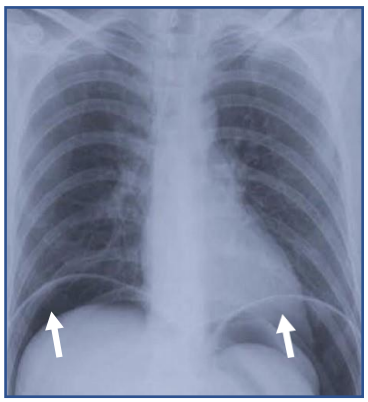

**Fig.9. Plain x-ray abdomen chest shows gas under diaphragm**

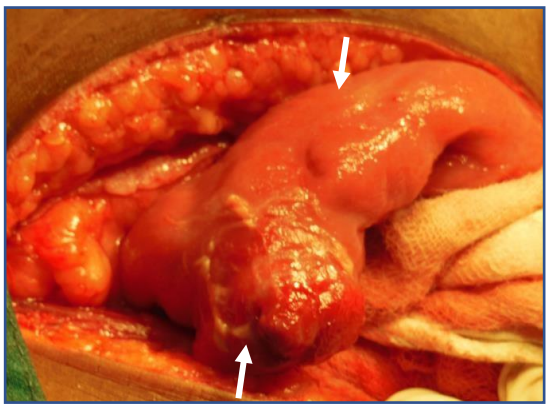

**Fig. 10. Intraoperative photograph showing polypoidal Mass at jejunum of size 3x2 cm**

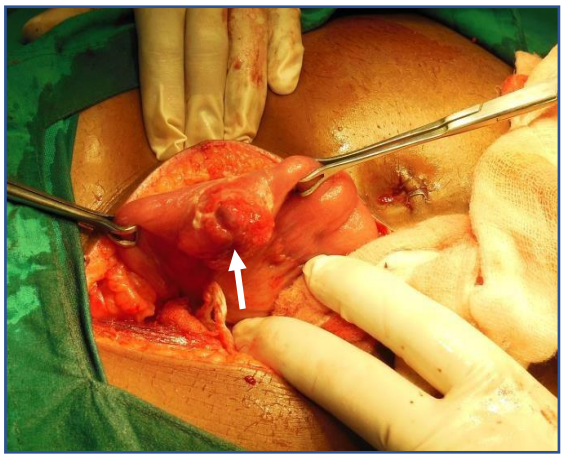

**Fig. 11. Intraoperative photograph showing polypoidal Mass at jejunum of size 3x2 cm**

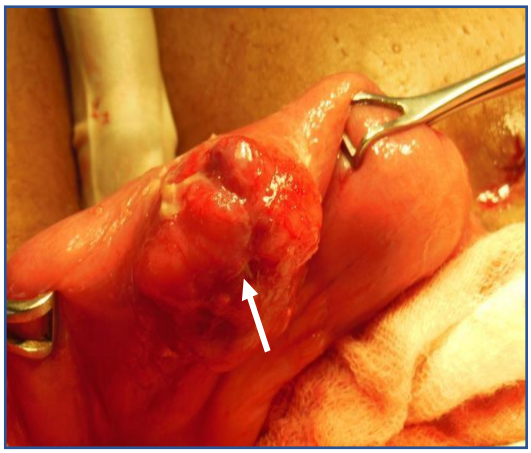

**Fig. 12. Intraoperative photograph showing polypoidal Mass with central perforation**

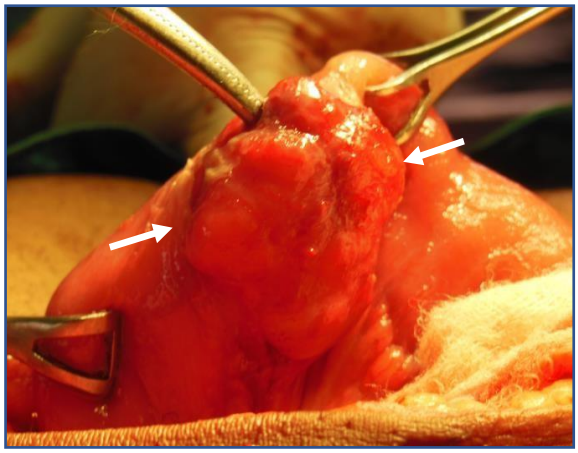

**Fig. 13. Intraoperative photograph showing polypoidal Mass at jejunum**

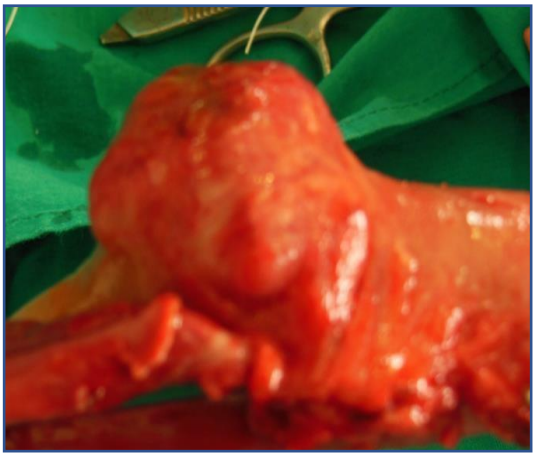

**Fig. 14. Intraoperative photograph showing localized excision of mass with 5 cm margin**

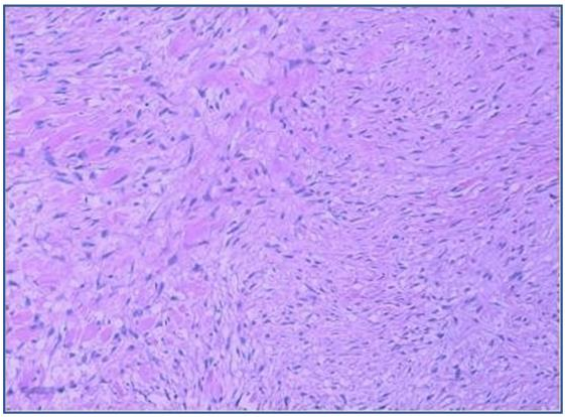

**Fig. 15. Tumor with spindle cells with low mitotic ratio**

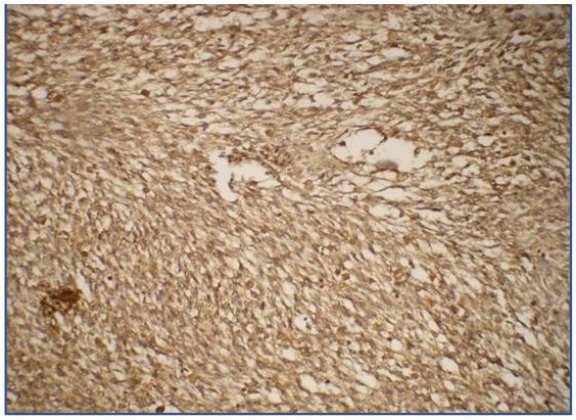

**Fig. 16. Tumor cells with diffuse CD 117 positivity**

### 2.3 Case 3 – Extra Luminal Transverse Colon GIST

A 50-year-old female was admitted to our center on August 14, 2010, with complaints of an epigastric intra-abdominal lump and abdominal pain of six months duration. She had no symptoms of luminal obstruction. Physical examination revealed an abdominal lump that moved with respiration and showed no visible

peristalsis. The swelling measured 8x6 cm in the epigastric region, was globular, and had smooth margins. Her kidney and liver function tests were normal. A CT scan of the abdomen revealed an 8x7 cm mass lesion associated with the transverse colon, with normal liver and spleen. Colonoscopy showed compression in the transverse colon area, but no intraluminal growth.

We decided to perform an exploratory laparotomy through a midline incision. The growth was observed arising from the mid-transverse colon along its serosal aspect, adherent to the stomach and posteriorly to the pancreas. The extraluminal transverse colon mass measured 8x7 cm, was spherical in shape, and had smooth margins. Two-thirds of the transverse colon, along with the growth, the middle colic artery, and the right colic artery, were ligated and removed en-bloc and End-to-end anastomosis was then performed. Perioperative palpation of all viscera was normal. The abdomen was closed after a thorough saline wash, and a drainage tube was placed in the right flank. The patient had an uneventful postoperative recovery and was discharged on the tenth postoperative day.

The gross specimen of the transverse colon mass measured 8x7 cm, had smooth margins, and was spherical in shape. Histopathological examination showed spindle cells with low mitotic activity and was positive for the CD117 antigen, indicating a GIST tumor. She did not receive postoperative chemotherapy and remained healthy during the five-year follow-up (Fig 17-24).

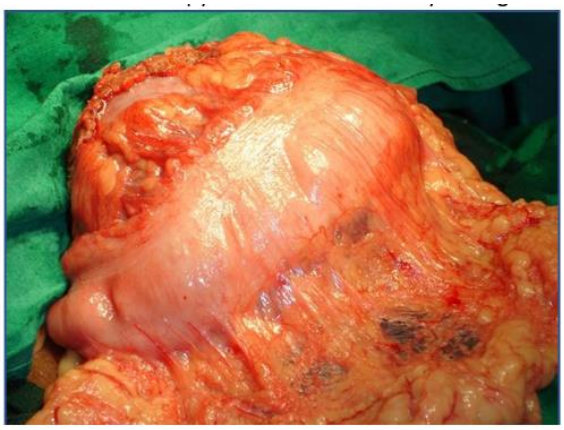

**Fig. 17. Intra operative photographs showing mass behind the mid-transverse colon**

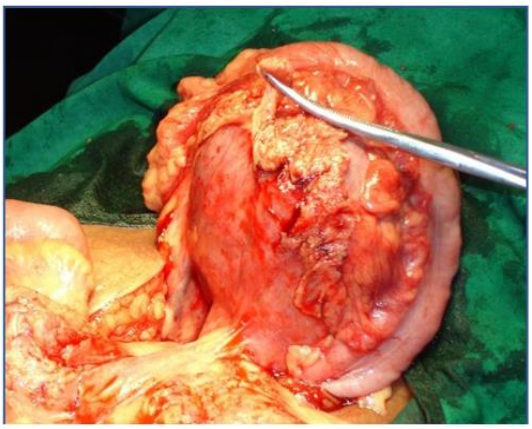

**Fig. 18. Intra operative photographs showing exophytic mass measuring 8x7 cm**

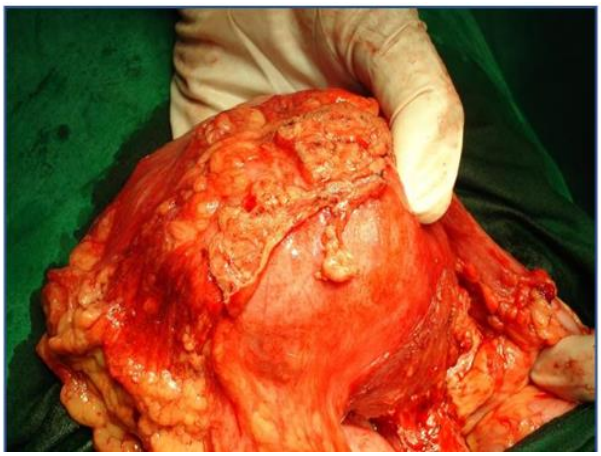

**Fig. 19. Intra operative photographs showing exophytic mass measuring 8x7 cm**

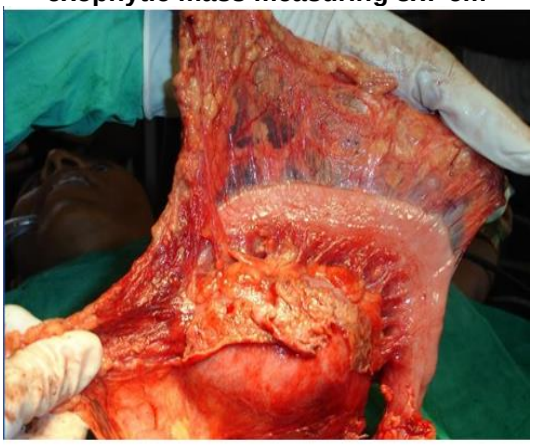

**Fig. 20. Intra operative photographs showing excision of exophytic mass along with transverse colon**

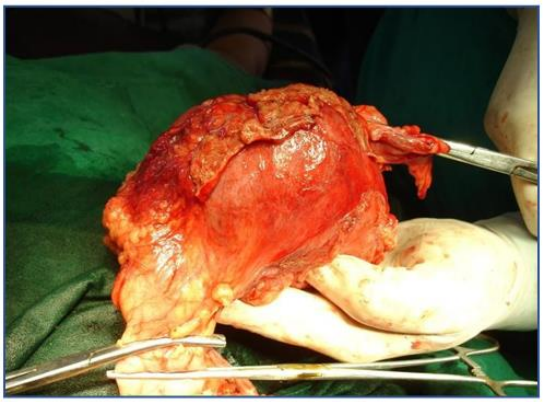

**Fig. 21. Intra operative photographs showing total excision of transverse colonic mass**

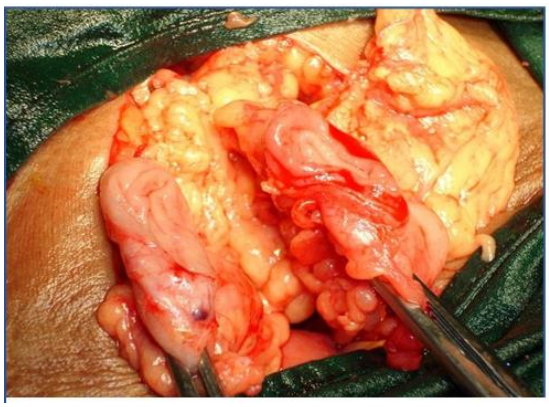

**Fig. 22. Intra operative photographs showing resection and colo-colic anastomosis**

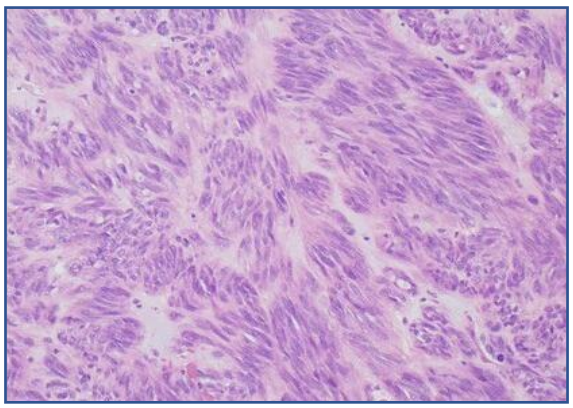

**Fig. 23. Tumor with spindle cells with low mitotic ratio**

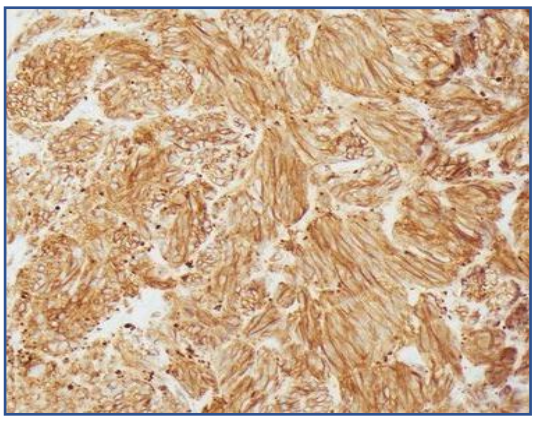

**Fig. 24. Tumor cells with diffuse CD 117 positivity**

## 3. DISCUSSION

Gastrointestinal stromal tumours were first defined by Mazur and Clark 6 after they detected a subgroup of gastrointestinal mesenchymal tumours that did not originate from smooth muscle or have neurogenic basis. In 1998, Kindblom and colleagues1 showed that these tumours originated from Cajal interstitial cells (pacemaker cells of myenteric plexus) or multipotent mesenchymal stem cells [4,7,10].

We classified tumours into 4 groups based on Fletcher classification:

1. very low-risk tumour (diameter < 2 cm and mitosis count < 5/50 high-power field [HPF]),
2. low-risk tumour (diameter 2–5 cm and mitosis count < 5/50 HPF),
3. intermediate-risk tumour (diameter < 5 cm and mitosis count 10/50 HPF or diameter 5–10 cm and mitosis count < 5 /50 HPF) and
4. high-risk tumour (diameter > 5 cm and mitosis count > 5/50 HPF or diameter > 10 cm andany mitotic rate) [5,9,11].

The management of patients with gastrointestinal stromal tumors (GISTs) involves a multidisciplinary team that includes pathologists, medical oncologists, surgeons, anaesthesiologists, and imaging experts. Four standard treatment approaches are utilized: [5,10,12].

- Surgery
- Targeted therapy
- Watchful waiting
- Supportive care.

The choice of treatment depends on various factors including tumor size, mitotic division rate, genetic makeup, primary location, whether it has metastasized, and if the tumor has

ruptured either spontaneously or during surgery [13,14].

### 3.1 Surgery

1. Open Surgery: For localized, resectable disease greater than 2 cm, surgical resection remains the cornerstone of treatment. The goal of surgery is complete resection with an intact pseudo- capsule and negative microscopic margins. The type of surgery depends on the size and location of the tumor in the GI tract: [4,6,12].

    - Wedge or segmental resection: For small GISTs.
    - Gastrectomy: Partial or total removal of the stomach for GISTs located in the stomach.
    - Bowel resection: Removal of part of the small or large intestine, or both.
    - En-bloc resection: Removal of the tumor and surrounding tissues as one piece, used for GISTs attached to nearby organs.

2. Laparoscopic Surgery- resection is feasible and safe for gastric GISTs and small bowel GISTs of 2 to 5 cm in size. Laparoscopic segmental or wedge resection with a linear stapling device is recommended for small gastric GISTs. Tumors in the small and large bowel typically require bowel anastomosis after resection, which can be performed using intracorporal or extracorporeal suturing or stapling. [5,6,9,12].

### 3.2 Targeted therapy

Targeted Therapy: Tyrosine kinase inhibitors (TKIs) are drugs that block signals needed for tumor growth. TKIs are used to treat GISTs that cannot be removed by surgery or to shrink tumors.

Commonly used TKIs include imatinib and sunitinib, which are administered as long as the tumordoes not grow [4,5,9].

### 3.3 Watchful Waiting

This involves closely monitoring a patient's condition without active treatment until signs or symptoms change.

### 3.4 Supportive care

Supportive care is provided if the GIST worsens during treatment or if side effects occur. The goal is to prevent or treat symptoms, side effects, and psychological, social, and spiritual problems, thereby improving the quality of life for patients. Radiation therapy may be used for pain management as part of supportive care [5,7,9].

### 3.5 Prognostic Factors for Localized GIST

Mitotic index, Tumor size, Tumor location (gastric, small bowel, rectal), Tumor rupture and Imaging characteristics are the prognostic factors for GISTs. Approximately 20-25% of gastric GISTs and 40- 50% of small intestinal GISTs are clinically aggressive. Tumor rupture significantly increases the risk of recurrence. Tumors larger than 10 cm, as well as those with lobulated or heterogeneous appearance, ulceration, regional lymphadenopathy, and exophytic growth on CT imaging, are more likely to recur. Survival Rates: The five-year relative survival rate for localized GISTs is 95%. For GISTs that have spread to distant parts of the body, the five-year survival rate is 40-50% [12,14,15,11].

## 4. CONCLUSIONS

GIST tumors appear to be a well-defined group of neoplasm to resulting from mutation in the C-kit gene and arising from the mesenchymal cells of the GI tract, specifically the intestinal cells of cajal called pacemakers cells of autonomus nervous system. They require multimodality therapy as well as long term follow up to provide the best chance for cure. Surgical resection is currently the “gold standard” in the management of GISTs. Complete resection with negative margins is the main goal of surgery.

## DISCLAIMER (ARTIFICIAL INTELLIGENCE)

Author(s) hereby declare that NO generative AI technologies such as Large Language Models (ChatGPT, COPILOT, etc) and text-to-image generators have been used during writing or editing of manuscripts.

## CONSENT

As per international standards or university standards, patient(s) written consent has been collected and preserved by the author(s).

## ETHICAL APPROVAL

It is not applicable.

## COMPETING INTERESTS

Authors have declared that no competing interests exist.

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