Case Report

Jejunal GIST — An Obscure Cause of Melaena

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Gastrointestinal Stromal Tumours (GISTs) are one of the rare causes of Alimentary Tract Neoplasm. They arise from the Interstitial Cells of Cajal (ICC) with overexpression of proto-oncogenes like KIT, PDGFRA and BRAF-Kinase, etc[1]. The typical location of these tumors is the stomach with Jejunal GIST being the rarest variant. The maximal incidence of the disease is reported in the sixth decade of life. GISTs are often asymptomatic and clinicians are misdirected towards a different diagnosis because of its variable nature of presentation. The clinical feature typically ranges from non-specific abdominal symptoms like nausea, vomiting, bloating, etc, to abdominal emergencies like hemorrhage, anemia, or obstruction. Although mostly benign, there is a high probability of progression to malignancy[2]. Thus, in patients with no appreciable cause of gastrointestinal bleed or chronic abdominal discomfort, GIST should have high suspicion index. This can help to limit the progression and thereby prevent further complications. Due to its location, it is difficult to identify by Endoscopy or Colonoscopy. Single Balloon Enteroscopy (SBE) and CT Scan of Abdomen are the primary investigational modalities. The treatment aims at resection of the mass with continued Postsurgical surveillance and targeted molecular therapy in some cases.

Herein, we report a 14-year-old boy with chronic paleness and gradual onset repeat episodes of Melaena. Despite repeated blood transfusions, the patient had a Hemoglobin <7g/dl. All his initial investigations, including Endoscopy and Colonoscopy, were normal. CT abdomen raised suspicion of a small mass (3-4cm) in the proximal small intestine and was confirmed by Single Balloon Enteroscopy. The lesion was tattooed during the procedure and subsequently, resection was carried out with wide margins. Due to its small size and low mitotic rate on HPE (<5-50/hpf), imatinib was not indicated and only continued postsurgical surveillance is advised for a period of 3 years.

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Gastrointestinal Stromal Tumour (GIST) is one of the scattered mesenchymal Tumour of the Gastrointestinal Tract (GIT). It accounts for only 0.2% of all GI Tumours. The most common location is the stomach and small intestine accounts for only 20% of the GIST. Among the small intestinal variant, Jejunal is the rarest subtype. The majority cases of the disease occur at 50-70 years of age.

Mazur and Clark first introduced GIST in 1983. They arise from the Interstitial Cells of Cajal (ICC), which are known to be the pacemaker cells of the small intestine. [3]The growth of the Tumours is primarily driven by the over-expression of proto-oncogenes C-KIT, Platelet-Derived Growth Factor (PDGF)-alpha polypeptide or BRAF kinase. These proto-oncogenes drive the activity of Tyrosine Kinase Receptors at ICC, thereby facilitating growth.

They present with vague symptoms that can be easily missed or ignored in the initial setting. Some individuals present with nausea, vomiting, and vague abdominal pain. In later stages or if the mass >5 cm repeated bouts of Melaena develops. Some patients also present with worsening Anemia (not responding to blood transfusion) and loss of appetite. In some instance, there can be a palpable mass that needs immediate intervention.

Editor's Comment :

- Jejunal GIST is a very easily missed diagnosis for Melaena that shall be kept into consideration. The reason being the atypical nature of presentation and difficulty in visualisation by rationale investigation techniques. Hence, attention should be paid while listing the differentials for a sudden onset Melaena without an otherwise evident condition.

The initial management should be to visualise the mass using imaging studies. The definitive line of diagnosis is postsurgical biopsy and immuno-histochemistry.

There is a 50% chance of recurrence in patients bearing the primary tumour[4]. Therefore, proper follow-up is required after surgery. Follow-up can be done using serial CT-Abdomen and Pelvis or Ultrasonography (preferred in young individuals) every three to six months. The uncovering of newer generation biologics like imatinib and sunitinib are effective against metastatic and unresectable variants.

So, we present over here an unusual case of Jejunal GIST in a 14-year-old boy.

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A 14-year-old boy presented to the Emergency Department with worsening pale complexion and tarry black stool for two weeks. He had one episode of dizziness that resolved spontaneously a few days before presentation. There was no associated abdominal pain or other symptoms.

Clinical examination revealed pallor, tachycardia and decreased breath sounds on the Left Lung Base. Mild grade bilateral pedal edema was also seen on examination, no Lymphadenopathy was noted. Abdomen was soft and no palpable mass was evident on examination. The Liver and spleen were not enlarged. Initial investigational reports showed Haemoglobin of 7.2g/dl. Despite repeated PRBC (Packed Red Blood Cells) transfusion, the patient was unable to keep up a Haemoglobin of >7g/dl.

Initial investigational report with Abdominal Ultrasonography, Upper Gastrointestinal Endoscopy and Colonoscopy revealed no significant finding. CECT Abdomen reported a suspicious mass in the jejunum (Fig 1).

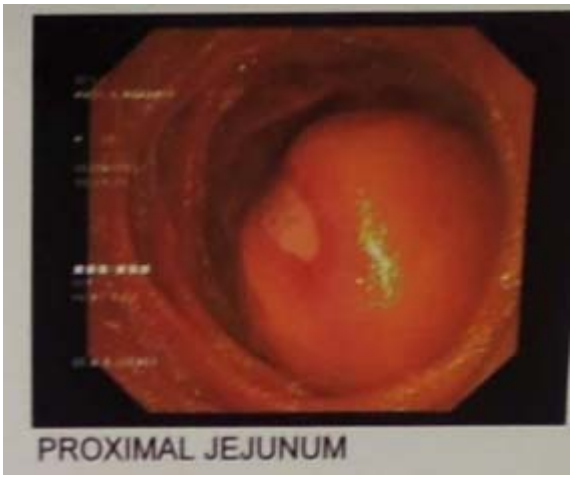

Fig 1 — CECT Abdomen showing a suspicious mass (as indicated by arrow)

This was followed by Single Balloon Enteroscopy, which visualised a 4-5cm sub-mucosal mass with central ulceration (Fig 2). The mass was inked and tattooed to aid identification during surgery.

Elective Exploratory Laparotomy was chosen as the line of management. It revealed an approximately 4cm tattooed extramural tumour at the proximal Jejunum, 25cm distal to the duodenojejunal flexure. There was no infiltration into nearby structures or any evidence of metastasis or Lymphadenopathy. The tumour along with adjacent lymph nodes was excised by resection of the Jejunum with wide mesentery and the gastrointestinal continuity was maintained by interrupted anastomosis.

Histopathological examination of the Tumour revealed a 3x2.2x2 cm sized mass protruding into the Jejunal lumen. It was composed of spindle-shaped cells arranged in fascicles and intersecting bundles. Mitotic figures were very sparse being <5/50 hpf. The lymph nodes did not show any Tumour deposits. Immunohistochemistry was positive for CD117, CD34, and desmin. These findings were consistent with benign Gastrointestinal Stromal Tumour (GIST). Staging was reported as T2N0M0. The risk stratification concerning the mitotic rate, size and location unveil a 4.3% chance of disease progression, which falls into low-risk category.

The patient was stable on discharge. He has been on follow-up for 24 months Postoperatively. Serial USG and Contrast-enhanced Computed Tomography (CECT) Abdomen were normal except for an incidental occurrence of Calculous Cholecystitis that was managed with Laparoscopic Cholecystectomy.

Discussion

GIST is one of the areas that is not explored in depth due to its low incidence and non-specific presentation. It is generally absent in the initial list of differentials in a patient presenting with Melaena. Thus, clinical diagnosis is based on the index of suspicion. Some intra-abdominal tumours can mimic GIST. Some of them are abdominal Leiomyoma, Leiomyosarcoma, Inflammatory Fibroid Polyp, Abdominal desmoid etc[5]. They are differentiated mainly based on histopathological examination.

As discussed about its variability in presentation, the diagnosis remains a challenge. Visualisation of the mass is the key to diagnosis. Although abdominal ultrasonogram is used as the initial investigation the Tumour can be easily missed due to its size, location, and the organ of origin. Therefore, CT abdomen is used to identify the lesion in its initial level of presentation. It tells us about the location, size, or calcification (if present). The degree of calcification also gives us a clue regarding the associated Necrosis, which may be a sign of malignancy.

Apart from indirect visualisation (as discussed), direct visualisation procedures are also used in identification. Upper GI Endoscopy, Colonoscopy and Single Balloon Enteroscopy are some examples. They can aid biopsy of

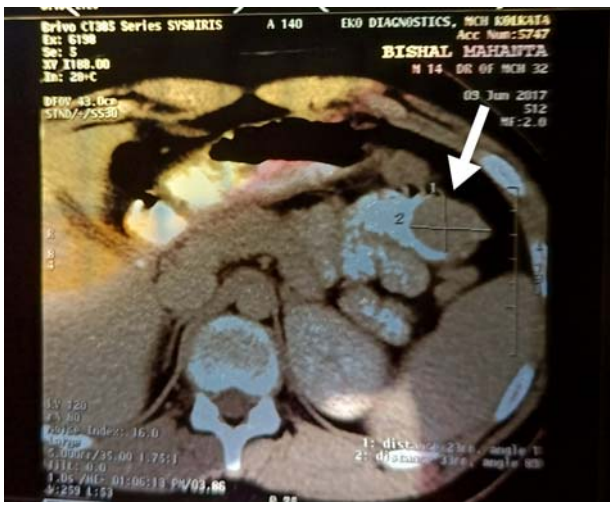

Fig 2 — Mass as visualised in SBE

Table 1 — *Risk of progression to GIST to malignant stage depending on its size and mitotic rate*

Tumour Parameters			Percentage of patients with progressive disease during long term follow-up			
Group	Tumour Size	Mitotic Rate	Gastric GISTs	Jejunal &Ileal GIST	Duodenal GIST	Rectal GIST
1	≤2cm	≤5-/-50-HPFs	0% none	0% none	0% none	0% none
2	>2cm ≤5cm	≤5-/-50-HPFs	1.9%very low	4.3%low	8.3%low	8.5%low
3a	>5cm ≤10cm	≤5-/-50-HPFs	3.6% low	24%moderate	34% high ‡	57% high ‡
3b	>10cm	≤5-/-50-HPFs	12%moderate	52%high	34% high ‡	57% high ‡
4	≤2cm	>5 / 50 HPFs	0% †	50% †	§	54%high
5	>2cm ≤5cm	>5 / 50 HPFs	16%moderate	73%high	50%high	52%high
6a	>5cm ≤10cm	>5 / 50 HPFs	55%high	85%high	86%high	71%high ‡
6b	>10cm	>5 / 50 HPFs	86%high	90%high	86%high	71%high ‡

the lesion or inking the site for identification during surgery. Recent advances like Capsule Endoscopy, Narrow-Band Imaging (NBI) are also important cornerstones for outlining the Tumour.

The curative intent for the disease treatment is surgical excision of the tumour with a clear narrow margin. The prognosis is dependent upon the size of the Tumour and its mitotic activity. If the size is >5 cm and mitotic activity is >5-50/hpf, there are chances of malignant potential. Miettinen and Lasota in 2006 have developed the risk table on follow- up information of thousands of patient having GIST over time[6]. The Table is hereby shared at the end of the discussion (Table 1).

With the advent of newer generation biologic drugs like imatinib, it is used as a neoadjuvant to achieve cytoreduction in cases where clear surgical excision cannot be made. This drug along with newly discovered sunitinib is used for several years with encouraging results as adjuvant and neoadjuvant therapy[7]. Thus, they have proved as successful molecular therapy for the management of recurrent or unresectable GIST.

Conclusion

GIST can easily evade detection. The pre-operative diagnosis is often missed due to its non-specific presentation and lower incidence. Thus, it should have high suspicion index in patients with no apparent cause of Melaena or unexplained cause of abdominal discomfort. Hence, an increased degree of awareness of GIST is of paramount significance to manage this rare but aggressive tumour. A multi-disciplinary approach with direct and indirect imaging, surgery, continued surveillance and targeted Molecular Therapy is the mainstay of management for successful outcomes.

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