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Case Report

Common tumor with uncommon presentation- Immunohistochemistry, Answered the bell

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ABSTRACT

Gastrointestinal stromal tumors (GISTs), though the most common mesenchymal tumors of the gastrointestinal tract, are rare accounting approximately 1% to 3% of all gastrointestinal tumors. We report a case of a female with GIST in the ileum presenting as acute abdomen. The present case highlights on surgery, histopathological examination and immunohistochemistry as a triad that helps in answering the bell for confirmed diagnosis.

1. Introduction

Gastrointestinal stromal tumors (GISTs), though the most common mesenchymal tumors of the gastrointestinal tract, are rare accounting approximately 1% to 3% of all gastrointestinal tumors.[1] Histopathological examination together with immunohistochemistry (IHC) plays an important role in confirming the diagnosis.[2] Here, we are presenting a GIST arising from ileum presenting as acute abdomen.

2. Case Report

A 44 year old woman presented to casuality with severe abdominal pain for a short duration of few hours before admission to hospital. The patient had normal bowel habits and no nausea or vomiting complaints. The patient's vital signs like heart rate, blood pressure, respiratory rate, and body temperature were stable. Physical examination revealed abdominal distension, generalized tenderness, and guarding. Clinical diagnosis of intestinal perforation was made. Further ultrasound reported as mixed echogenic lesion measuring 10x6 cms with increased vascularity in left iliac fossa suggestive of neoplastic origin. A computed tomography (CT) scan of the abdomen revealed a thick walled heterogeneously enhancing lesion with predominant central necrosis in left iliac fossa measuring 10.2x6.5x6.2 cms. There was partial encasement of inferior mesenteric vessels suggestive of ileal diverticulum tumor or GIST of small bowel. The mass was not adherent to any of the intraabdominal structures.

Emergency laparotomy done showed a well encapsulated, large mass measuring 10 x 6 cm was found arising from the small bowel. The tumour along with part of ileum was completely resected and end to end anastomosis of bowel loops was performed. The post operative period was uneventful. Grossly the specimen was a grey white lobulated mass measuring 12 x 6.5 x 5cms arising from the collapsed loop of part of the ileum measuring 5cms. The external surface of the mass was congested. Cut section was solid grey-white lobulated with hemorrhagic areas. Mucosa and wall of the ileum was grossly unremarkable without any perforation. Attached mesentery showed nine lymphnodes.(Figure 1a,b) On histopathological examination showed encapsulated lesion composed of diffuse sheets, intersecting fascicles and

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bundles of spindle cells. These spindle cells have central oval to blunt elongated nuclei with bland nuclear features and moderate cytoplasm. Admixed are less than10% of areas of tumor necrosis and cells with less than 5 mitoses per 20 high power fields. Adjacent ileal mucosa showed focal ulceration with congested blood vessels and wall was unremarkable. Surgical margins of the ileum and nine lymphnodes identified in mesentery were free from tumor.(Figure 2a-d) Immunohistochemical studies showed CD117 and DOG1 – strong, diffuse cytoplasmic positivity and CD34 focal positivity. Tumour cells were negative for S100, desmin, smooth muscle actin and cytokeratin.(Figure 3a-d) A diagnosis of low grade gastrointestinal stromal tumor –Spindle cell type - ileum was made.

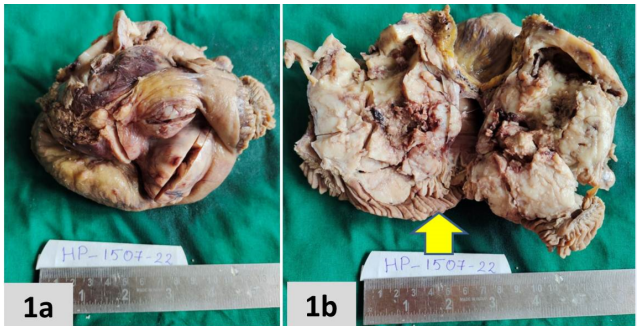

Fig. 1: a: Gross image showing globular mass external surface; **b:** Gross image showing globular mass cut surface which is grey-white solid, with collapsed mucosa and wall of ileum (yellow arrow).

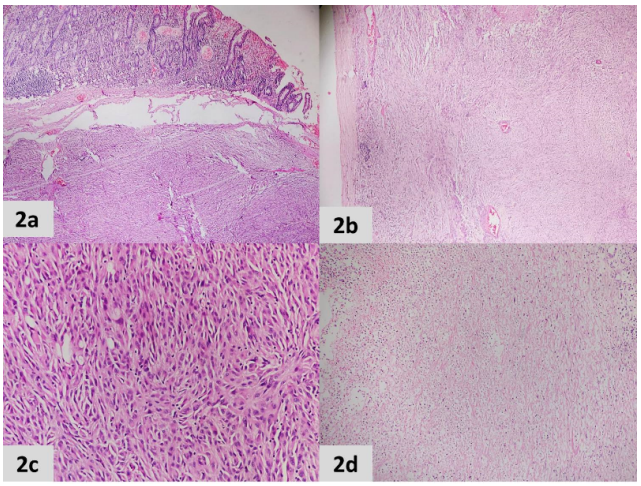

Fig. 2: a: Microscopy showing the ileal mucosa with focal ulceration and congestion and serosa showing tumor. (H&E, 10x); **b:** Microscopy showing well encapsulated tumor. (H&E, 10x); **c:** Microscopy showing tumor composed of spindle cells arranged in intersecting fascicles, bundles and sheets (H&E, 10x); **d:** Microscopy showing coagulative tumor necrosis. (H&E, 10x).

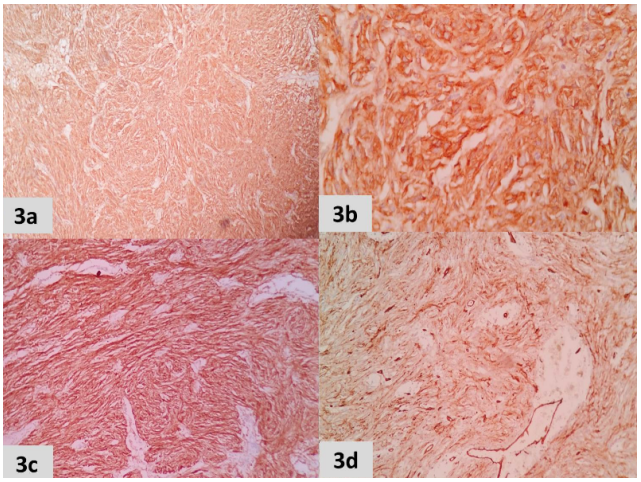

Fig. 3: a: Microscopy showing immunoreactivity for CD117 strong, diffuse cytoplasmic positivity. (IHC, 10x); **b:** Microscopy showing immunoreactivity for CD117 strong, diffuse cytoplasmic positivity. (IHC, 100x); **c:** Microscopy showing immunoreactivity for DOG1 strong, diffuse cytoplasmic positivity. (IHC, 100x); **d:** Microscopy showing immunoreactivity for CD 34 focal cytoplasmic positivity. (IHC, 100x)

3. Discussion

Mazur and Clarke coined the term GIST in 1983 for a discrete set of mesenchymal tumors of the gastrointestinal tract (GIT) having no ultrastructural or immunohistochemical features characteristic of smooth muscle differentiation.[1–3] Kindblom and associates in 1998 demonstrated that the actual cell of origin of these tumors is a pluripotent mesenchymal stem cell programmed to differentiate into interstitial cells of Cajal, the GI tract "pacemaker cells" - the cells responsible for initiating and coordinating gastrointestinal motility.[1–3] In 1998 Hirota and colleagues discovered that c- kit /CD 117 proto-oncogene gain-of-function mutations in these tumors that distinguished GIST as a unique clinical entity. 60-70 % of GISTs have been reported to arise in the stomach, whereas 20% to 30% originate in the small intestine and fewer than 10% from esophagus, colon, and rectum. GISTs can also occur in extraintestinal abdominopelvic sites such as the omentum, mesentery or retroperitoneum.[1–4]

GIST mostly affects individuals aged more than 50 years with male to female ratio of 1.5.[4] Our index was much younger age group lady. The GISTs clinical manifestations are diverse according to the tumor's anatomic site, size and extension into the adjacent structures. Approximately one-third of patients with GIST are asymptomatic for ages. Most common symptoms of small intestine GISTs are chronic anemia due to pressure necrosis by the tumor or ulceration of the overlying mucosa leading onto mucosal bleeding and intestinal obstruction.[1–4] Our case presented with acute pain abdomen of very short duration. Acute abdominal

symptoms were more frequent in patients with jejunal and ileal than in gastric GISTs.[5,6] Literature search reveals, case with acute abdomen are usually due to perforation caused by obstruction with increasing intraluminal pressure or tumor erosion that leads to wall necrosis. Other described mechanisms of perforation are tumor embolization of intestinal blood vessels, ischemia and replacement of the bowel wall by tumor cells followed by necrosis.[5–8] Our case had no perforation but presented with acute abdominal pain possibly due to intestinal obstruction.

Studies says that radiological imaging examination like ultrasonography, barium examination of the gastrointestinal system, CT, angiography or magnetic resonance imaging can establish the 100% correct diagnosis of GIST priorly.[5–8] In our case also possibility of small bowel tumor was made possibly of GIST on ultrasonogram and CT scan. Histopathological diagnosis of GIST depends on the morphological type of tumor cells and IHC. According to morphology, there are three types of tumor cells: spindle cell type (70%), epithelioid cell type (20%), and mixed cell type (10%).[2] In the present case it was spindle cell type with least necrosis and mitotic figures. On IHC all of the GISTs are positive for CD117 and DOG1 and 60–70% is positive for CD34. 30–40% of GISTs are positive for SMA, 10% positive for vimentin and 5% positive for S100 protein.[2] All of these markers were done in the present case. Hence a final diagnosis of low grade GIST arising from ileum was made. Prognosis is related to the size of the tumor and to the mitotic rate: tumors > 10 cm or with a mitotic rate of >5 per 50 HPF have a higher risk of recurrence, metastatic spread and are associated with a poor prognosis which is seen in 20-30% of cases.[7–9]

The treatment of GIST includes surgical resection with adjuvant chemotherapy with imatinib, the inhibitor of tyrosine kinase which helps to increase the survival and reduces recurrence after surgery.[9] Literature reveals that 85% of tumors can be completely resected and the incidence of recurrence and metastasis after radical surgery is 50%. Negative surgical margins are important to prevent the local recurrence of the tumor and lymph node involvement is rare.[8,9] In the present case, emergency laparotomy was done with tumor and ileum resection with end to end anastomosis. The surgical margins and lymphnodes from mesentery were free from tumor. Patient recovered well following the procedure and was referred to higher center for chemotherapy.

In summary, we report a case of a female with GIST in the ileum presenting as acute abdomen. GIST is common mesenchymal tumor but its presentation as acute abdomen is rare, hence to be included in the possible diagnosis of acute abdomen in emergency. The present case highlights on surgery, histopathological examination and immunohistochemistry as a triad that helps in answering the bell of confirmed diagnosis.

4. Source of Funding

None.

5. Conflict of Interest

None.

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