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A case of small bowel GIST mimicking an ovarian mass

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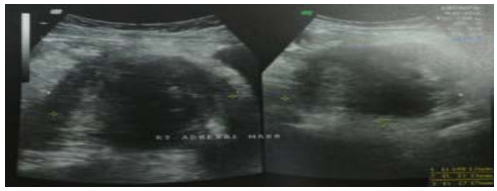
Abstract : 55 year old female came with complaints of abdominal pain and vomiting for 2 weeks. On examination she had a firm mass in the hypogastric region which was not mobile. Imaging studies suggested an adnexal mass and a diagnosis of ovarian mass was made provisionally. on laparotomy, a mass was found adherent to the uterus and bladder and connected to the ileal mesentery via a stalk. en bloc excision of the mass along with resection of the involved bowel segment done followed by anastamosis of the bowel ends. HPE report came as low grade GIST. she was put on Imatinib. The Gastrointestinal stromal tumours are the most common mesenchymal tumours of gastrointestinal tract. Majority are associated with mutations in the c-KIT gene. They grow as endophytic masses or exophytic excrescences. The most common site is the stomach followed by the small bowel. Most are asymptomatic. The rest present with vague symptoms or features due to complications like obstruction or perforation. Diagnosis is established by imaging and immunohistochemistry. The most important prognostic factors include the size, the site and mitotic count. Surgical excision is the main mode of treatment and it offers the only mode of cure. Adjuvant therapy is by using tyrosine kinase inhibitors. 5 year survival rate varies from 28-60 percent. Long term yearly follow up is necessary to detect recurrences.

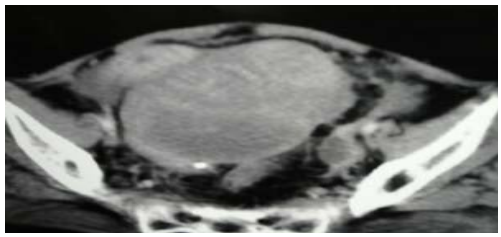
Keyword :GIST, ckit mutation, CD117, surgical excision, Imatinib

CASE REPORT:

55 year old female patient was admitted in our unit with complaints of abdominal distension for 2 weeks involving the lower abdomen which was associated with dull aching pain for the past 3 days which brought her to the hospital. She also had occasional episodes of vomiting. There was no history of vomiting or altered bowel habits. She had loss of appetite since the onset of pain. She was a known diabetic for two years and was on oral hypoglycaemic agents. She was dehydrated. Vitals were stable. On examination of the abdomen, it was soft. Tenderness was elicited in the right iliac fossa and hypogastric regions. A firm mass of size approximately 6 cm in diameter was felt occupying the hypogastrium. It had an irregular surface. Lower border was not palpable. It didn't move with respiration. It was intra abdominally located. There was no hepatomegaly/ free fluid. There was no palpable Supraclavicular node. Per rectal and per vaginal examinations were not contributory. Provisional diagnosis of a retroperitoneal soft tissue tumour was made and was evaluated. Routine blood investigations were within normal limits. X rays of the chest and abdomen didn't reveal any abnormality. Ultrasound of the abdomen revealed a hetero-echoic right adnexal mass.

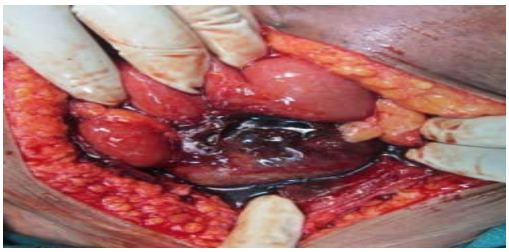


CECT of the abdomen showed evidence of about 10*8*7 cm heterogeneously enhancing mass lesion noted in the pelvis probably arising from right adnexa giving the impression of COMPLEX RIGHT ADNEXAL MASS LESION

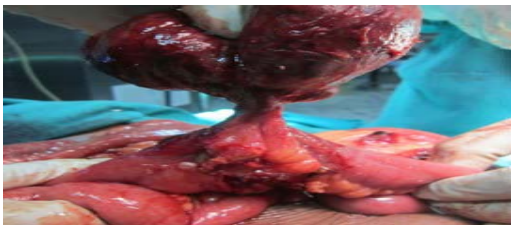


CA 125 level was slightly elevated (42.7 U/ml; normal reference < 35 U/ml). Provisional diagnosis of a right adnexal mass probably of ovarian malignancy was made and planned for laparotomy. At laparotomy, a mass was found adherent to the uterus and bladder below and the small bowel above. By sharp dissection, it was freed from the bladder and uterus. Both ovaries were found to be normal. The mass 10*10*8 cm in size was

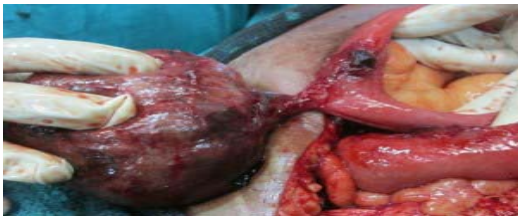
found to arise from the ileum about 100 cm proximal to the ileo-caecal junction. It was attached to the anti-mesenteric border and mesentery of the ileum via a stalk over a length of 3 cm. it was encapsulated containing haemorrhagic material.



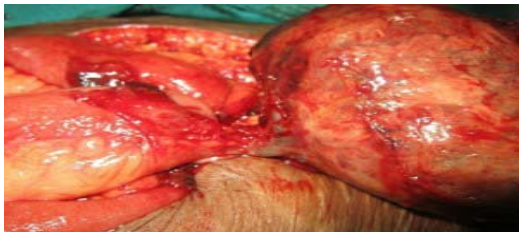
Mass adherent to the uterus and bladder and attached to the ileum



Mass attached to the ileum via a stalk

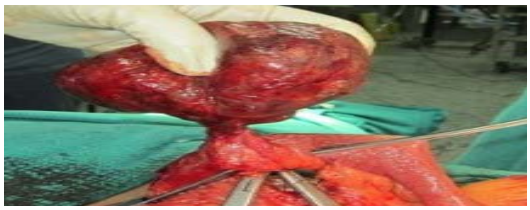


Attachment to the anti mesenteric border

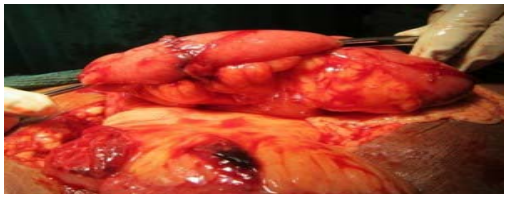


Attachment to the ileal mesentery

En bloc excision of the mass along with resection of the segment of ileum was done followed by end to end anastomosis.

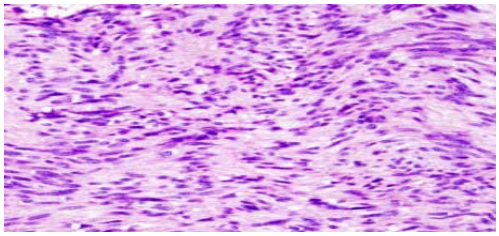


En bloc resection of the mass along with the ileal segment



End to end anastamosis

Histopathological examination showed circumscribed neoplasm with large central areas of cystic and hemorrhagic degeneration. The Mass showed fascicles and whorls of spindle cells with some of them showing clear cell change. IMPRESSION- Histology is consistent with LOW GRADE GIST. Resected ends free of infiltration.



HPE - low graqde GIST

IHC marker CD 117 was found to be strongly positive. Patient was put on Imatinib therapy.

DISCUSSION

Until the late 1960's, stromal tumors arising in the GI tract were referred to as smooth muscle neoplasms of the gastrointestinal tract. Immuno-histochemistry in the 1980's demonstrated that some of these tumors lacked features of smooth muscle differentiation, some had markers of neuronal differentiation and some had neither of the above markers. This led *Mazur and Clark* to coin the general term "*Gastrointestinal stromal tumors*" to collectively refer a group of mesenchymal tumors of neurogenic or myogenic differentiation.

INCIDENCE:

They are the most common mesenchymal tumours of the GIT constituting only about 1% of all gastrointestinal malignancies. The most common site of origin is the stomach (60-70%); followed by the small bowel (20-30%), duodenum (5%) and colon (<5%). Omentum, Mesentery and oesophagus constitute less than 1% of cases. The usual age at presentation is 50 to 70 years. They present as submucosal masses. They can be endophytic masses or grow out as exophytic excrescences. They are believed to originate from the interstitial cells of cajal.

ETIOPATHOGENESIS:

Gain of function mutation in exon 11 of c-KIT proto-oncogene located on chromosome 4 are associated with most GISTs. This causes intracellular signalling cascade over activity resulting in uncontrolled cellular proliferation. This was discovered by Hirota et al in 1998. 95% of GISTs are c- KIT positive. In c-KIT negative GISTs, mutations in PDGFRA gene can be seen in a subset of cases. BRAF mutations and PKC-theta gene mutations are occasionally found to cause GIST. Less than 5% of cases are associate with hereditary syndromes.

CLINICAL PRESENTATION:

Most patients are often asymptomatic, especially in the early stages of tumour development and discovered incidentally by CT or endoscopy. Signs and symptoms are related to location of tumor. They may be in the form of

vague pain or discomfort (most common), early satiety, Anorexia, weight loss, nausea, anaemia. They may rarely present as a palpable mass. They may present with features of peritonitis due to perforation. They can cause significant hemorrhage. Obstructive features can occur and are site specific (dysphagia, constipation, jaundice)

DIFFERENTIAL DIAGNOSIS:

It includes benign and malignant neoplasms of small bowel, adenocarcinoma, soft tissue tumours, leiomyoma, schwannoma, solitary fibrous tumour, metastatic melanoma.

WORK UP:

It includes Routine blood investigations; CXR (for evidence of perforation), X-ray Abdomen (for evidence of obstruction or perforation), USG, CECT (investigation of choice for diagnosis and staging). Endoscopy can help in taking biopsy. Barium studies, MRI and EUS are also useful. PET CT is useful in detection of metastasis. Routine preoperative biopsy is not necessary in resectable tumours. In unresectable or metastatic tumours, biopsy can be done with open or endoscopic method and not percutaneously. Histology varies from well differentiated to poorly differentiated tumours. Immuno histo chemical markers for confirming the diagnosis include c-KIT/CD 117, CD 34, DOG-1.

BENIGN Vs MALIGNANT GIST:

The features suggestive of a benign tumour are

- Size less than 5 cm
- Low number of mitosis per HPF
- No mucosal invasion
- Low cellularity
- Low markers of cell proliferation

High-risk features for malignancy include any of the following:

- Tumor at least 10 cm in greatest dimension
- Presence of tumor rupture before or during surgery
- Intraperitoneal hemorrhage
- Multifocal intraperitoneal tumors
- Positive microscopic margins

TREATMENT:

Surgery is the mainstay of treatment in GISTs. Radical and complete surgical removal offers the only chance of cure. Even in locally advanced cases, debulking is helpful if adjuvant therapy is planned. Routine Lymphadenectomy is not necessary as chance for lymph nodal involvement is very rare. Laparoscopic surgery can be done for smaller GISTs. Adjuvant therapy consists of tyrosine kinase inhibitors Imatinib and Sunitinib. They can prolong the recurrence free period and overall survival following complete resection. They can also be used for palliatively in locally advanced and metastatic tumours. Imatinib can be used preoperatively to down size the tumour as well. Regorafenib is a recently approved drug for locally advanced unresectable GISTs not responding to the above two drugs.

PROGNOSIS:

The main prognostic factors include tumour size, site of tumour and mitotic count.

RISK STRATIFICATION:

Four risk groups are defined based on the size of the tumour and the mitotic count

RISK GROUP	SIZE	MITOTIC COUNT (per 50 hpf)
Very low risk	<2 cm	<5
Low risk	2-5 cm	<5
Intermediate risk	2-5 cm	6-10
	5-10 cm	<5
High risk	5-10 cm	6-10
	>10 cm	Any rate
	Any size	>10

Small intestine GISTs have a worse prognosis than gastric GISTs. The median survival for resectable tumours is 5 years and for unresectable/ metastatic tumours it is 10-12 months. The overall 5 year survival rate varies from 28 to 60%.

FOLLOW UP:

With prolonged follow up, any GIST has the potential to behave in a malignant fashion. 50% of primary localized tumors that are resected relapse after 5 years of follow up. Recurrences have been recorded even after 20 years. Yearly clinical examination and if necessary a CT / PET scan can be used for regular follow up.

CONCLUSION:

The Gastrointestinal stromal tumours are the most common mesenchymal tumours of gastrointestinal tract. Majority are associated with mutations in the c-KIT gene. Most are asymptomatic and the rest present with vague symptoms. Few present with features due to complications. Diagnosis is established by imaging and immunohistochemistry when biopsy is available. The most important prognostic factors include the size, the site and mitotic count. Surgical excision is the main mode of treat ment and it offers the only mode of cure. Adjuvant therapy with tyrosine kinase inhibitors helps in improving the recurrence free survival and overall survival. 5 year survival rate varies from 28-60%. Long term yearly follow up is necessary to detect recurrences.

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