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EXPECT THE UNEXPECTED: JEJUNAL GASTROINTESTINAL TUMOUR MIMICKING OVARIAN MALIGNANCY- A CASE REPORT WITH REVIEW OF LITERATURE

Pathology

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ABSTRACT

Gastrointestinal stromal tumours (GISTs) are mesenchymal tumours of alimentary tract comprising 0.2% of gastrointestinal tumors and only 0.04% of small intestinal tumours. Jejunal GISTs are one of the rarest subtypes. GISTs display various morphological forms like spindle and epitheloid cells in a variety of patterns and can be submucosal, intramuscular or subserosal in location. Grossly they are solid and cyst with variable hemorrhage and necrosis. Most of the gastrointestinal stromal tumors have mutations in either KIT (CD117) or PDGFRα gene. DOG 1 is a sensitive and specific marker of GIST independent of CD117 or PGDFRα expression. Here we present a case of malignant jejunal GIST with missed diagnosis on CECT abdomen.

KEYWORDS

GIST, jejunal, CD117

INTRODUCTION:

Gastrointestinal stromal tumours (GISTs) are mesenchymal tumours of alimentary tract arising as a result of activating mutations of proto-oncogenes c-KIT or platelet derived growth factor receptor alpha (PGDFRA) polypeptide.[1] This increases tyrosine kinase receptor activity resulting in uncontrolled proliferation of stem cells that differentiate into intestinal cells of Cajal.[2] GISTs comprise 0.2% of gastrointestinal tumors and only 0.04% of small intestinal tumours; jejunal GISTs being the rarest subtype. [3]

CASE HISTORY:

A 55 year old female patient presented with abdominal pain radiating to back along with vomiting. She had signs and symptoms of small bowel obstruction. Her USG abdomen showed a mass lesion in pelvis. CECT abdomen suggested right ovarian malignancy with regional extensions into multiple loops of terminal ileum, caecum and anterior abdominal wall with the right ovarian vein draining from a structure posterior to mass lesion likely suggestive of origin of mass lesion. Serum levels of β HCG was 6.12 mIU/ml, CA 125 was 18.86 U/ml, CEA was 0.734 ng/ml, CA 19.9 was 4.38 U/ml and AFP was 0.82 ng/ml. Patient was taken up for exploratory laparotomy and a 10×10 cms of jejunal mass was seen one feet away from the duodeno-jejunal junction and was resected with 4cms of jejunum on both sides. Tumour was large, bulky, nodular and intramural in location extending to serosa with foci of haemorrhage and necrosis. A nodule of 2×2 cm was present on the right side of pelvic peritoneal surface. The uterus and bilateral ovaries were found to be healthy.

GROSS: Received segment of jejunum measuring 19×9 cm, showing an ulcerated nodular growth measuring 10×4×3 cm, extending from anti-mesenteric border to anterior surface along with a 2× 2 cm peritoneal nodule. Cut section of tumour showed multiple areas of haemorrhage along with solid areas.

MICROSCOPIC EXAMINATION: Sections from the jejunal mass revealed spindle cell tumor with extensive necrosis and hemorrhage. Spindle cells were short, uniform with tapered nuclei and amphophillic slightly fibrillary cytoplasm. They formed cellular sheets and fasicles with whorled and storiform pattern. The tumor extended from muscle coat of jejunum to serosa with mitoses <5/10 HPF. Re-sected margins were free and no lympho-vascular invasion was seen. Peritoneal nodule also showed tumour of similar histology with extensive necrosis. Histological features were suggestive of malignant gastrointestinal stromal tumour with peritoneal metastasis. Immunohistochemistry showed high expression of CD117(intense and diffuse), CD34 and Vimentin and negative expression of S-100 and HMB-45.

DISCUSSION WITH REVIEW OF LITERATURE:

Gastrointestinal stromal tumors occur mostly in adults over 40 years old with peak incidence being between 60-65 years with slight male predominance.[2] GISTs display various morphological forms like spindle and epitheloid cells and rarely pleomorphic forms in a variety of patterns. They vary in diameter from 10 to >200mm and can be submucosal, intramuscular or subserosal in location. Grossly they are solid and cyst with variable hemorrhage and necrosis. The principle cell forms are spindled and epitheloid which can coexist in varying proportions. Various unusual forms and variants of gastrointestinal stromal tumors have been described.[4] Most of the gastrointestinal stromal tumors have mutations in either KIT (CD117) or PDGFRα gene.[5] However CD117 is not universally expressed in all GISTs and may be expressed in other tumours such as melanomas, adenoid cystic carcinomas, Merkel cell carcinomas, Kaposi sarcomas. DOG 1 is a sensitive and specific marker of GIST independent of CD117 or PGDFRα expression.[6] The clinical presentation depends on size of tumour and may include pain abdomen, occult gastrointestinal bleeding or intestinal obstruction.[7]

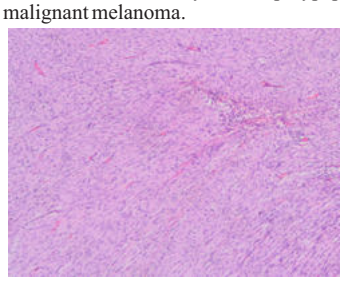
Sankey RE et al. documented a case of malignant jejunal GIST with tumour showing spindle and epitheloid cells with positive staining for CD117, DOG 1 and SMA on IHC along with exon 11 mutation on genetic analysis.[8]

In a case of malignant jejunal GIST presented by Dhull et al. a 15×10 cms jejunal growth was reported with unremarkable overlying mucosa and underlying submucosa, muscularis and serosa showing infiltration by plump spindle cells which showed widespread positivity for CD117.[3]

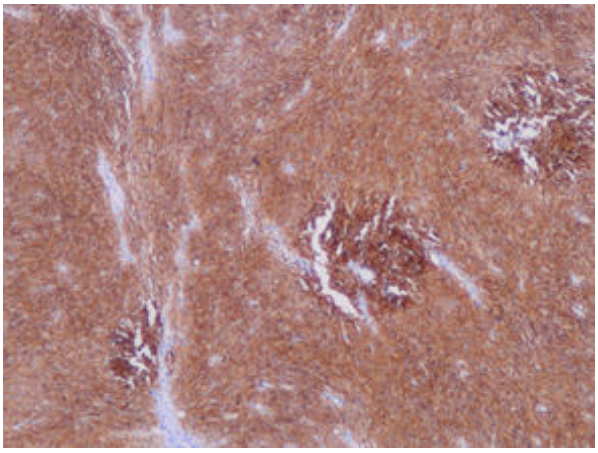
Khuri et al also reported a case of malignant jejunal GISTs with similar findings of a 10cms growth in proximal jejunum showing mixed spindled and epitheloid morphology on histopathology and invading the mucosa with mitoses >5/50 HPF. Tumor cells were positive for CD117 and DOG 1and negative for CD 34, Desmin, chromogranin, SOX 1, S100 and HMB 45.[9]

A similar case of malignant jejunal GIST was presented by Jaison J et al. where a 9×7×5 cms mass resected from the jejunum showed spindle cells in fascicular pattern with areas of haemorrhagic necrosis and mitoses >5/50 HPF. The tumor cells were positive for CD117.[10]

Histologically and with the support of immunohistochemistry gastrointestinal tumours have to be distinguished from true smooth muscle tumours and schwannomas of the gastrointestinal tract, intra-abdominal fibromatosis, inflammatory fibroid, polyp, paragangliomas and metastatic malignant melanoma.



H&E section shows dense cellularity with plump spindle cells



CD117 immunopositivity on IHC

CONCLUSION: The highlight of the case was the unusual presentation as jejunal GISTs are rare, comprising 0.04% of gastro-intestinal tumors and missed diagnosis on CECT. It was diagnosed only after laparotomy. Small intestinal gastrointestinal stromal tumors can present with varying degrees of symptoms which may not correlate to the size of the tumour leading to delayed diagnosis. Varied atypical presentations should be considered while evaluating small bowel pathologies to be able to diagnose GIST at an early stage. Surgery should be considered as primary treatment for localized and re-sectable tumours along with adjuvant treatment with Imatinib mesylate for successful outcomes.

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