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Gastrointestinal Stromal Tumour with Intratumoural Endometriosis

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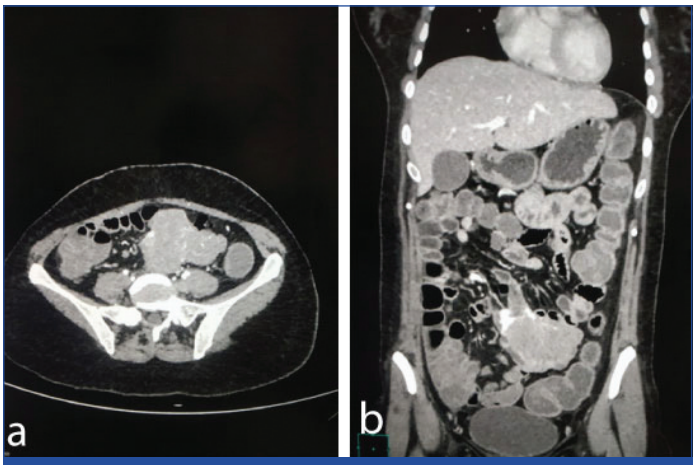
A 38-year-old female with no known co-morbidities, presented to the Department of Surgical Gastroenterology with complaints of pain in abdomen and per rectal bleeding for the past one day. She also complained of intermittent fever, generalised weakness and breathing difficulty for one week.

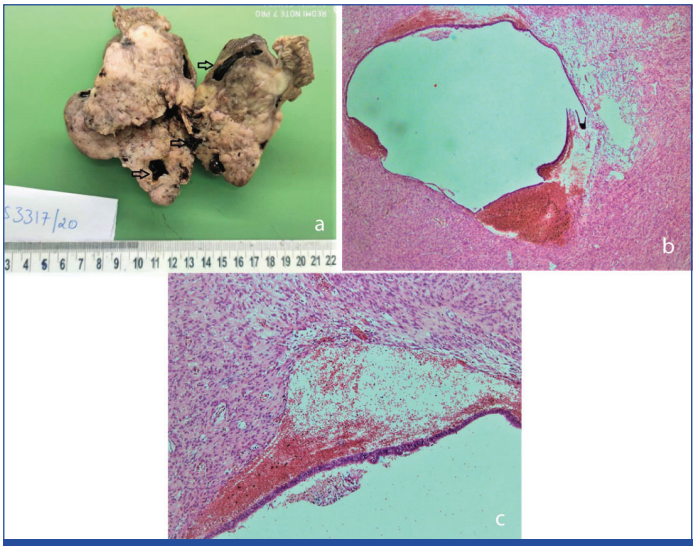
Laboratory examination revealed that the patient was anaemic (haemoglobin was 4.8 gm/dL) for which she was transfused with 7 units of packed red blood cells. Clinical examination of the abdomen revealed a well-defined intra-abdominal lump in the right iliac fossa of about 7×6 cm. Upper gastrointestinal endoscopy was normal. Colonoscopy showed presence of altered blood throughout the colon, however the mucosa was normal. Further radiological investigations were done. On ultrasound abdomen, a well-defined, heterogeneously hypoechoic solid mass lesion measuring 5.4×7.3 cm with lobulated outlines was visualised in the right iliac fossa. Computed Tomography (CT) abdomen [Table/Fig-1a] confirmed a serosa based bowel mass, which was likely involving the ileum. Further, on CT angiography, it was found that the soft tissue mass was present in mesentery and was inseparable from distal jejunal and proximal ileal loops [Table/Fig-1b]. There was prominent vascularity within and around the margins of this mass and had arterial supply from ileal branches. In view of the radiological findings, the possibility of Gastrointestinal Stromal Tumour (GIST) was suggested.

The patient underwent laparotomy exploration, where a whitish, well encapsulated, solid mass was found in the ileal segment. The mass was not invading nearby organs or structures and was separated completely. The specimen was submitted for histopathological evaluation. On gross examination, the segment of intestine measured 3.5 cm in length along with a large serosa based nodular tumour measuring 9.5×7×6 cm. On opening the intestinal segment, mucosa appeared unremarkable. Cut surface of tumour was grey white to grey brown, solid and lobulated with some cystic areas [Table/Fig-2a]. The cystic spaces varied in size from 0.5 to 3.4 cm in maximum diameter, and were filled with haemorrhagic material. The tumour was 1 cm from both resected ends of intestinal segment.

Microscopic examination showed a thinly encapsulated tumour of moderate cellularity. The tumour was composed of spindle cells arranged in interlacing fascicles and sweeping bundles [Table/Fig-2b]. The cells had abundant eosinophilic cytoplasm and minimally pleomorphic elongated nuclei having fine chromatin and inconspicuous nucleoli. No significant cellular atypia, mitotic activity (<5/50 high power fields) or tumour necrosis were seen. In addition, tumour showed focal presence of variably sized cystic spaces. The cysts were lined by bland looking ciliated columnar epithelium and were filled with haemorrhagic fluid [Table/Fig-2c]. Few of these cysts also showed presence of a thin attenuated band of stromal cells beneath the epithelium along with some chronic inflammatory cells and hemosiderin laden macrophages. The tumour was present as a large serosal mass and was also involving the muscle coat and submucosa of attached ileal segment. Overlying mucosa was however free. On Immunohistochemistry (IHC), tumour cells were

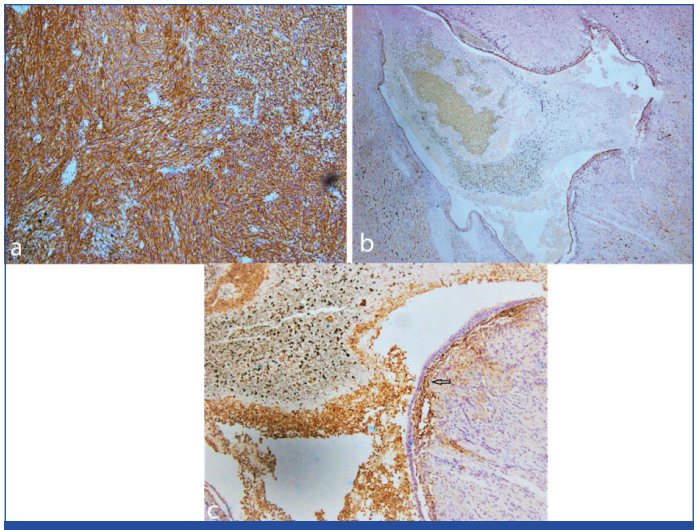


[Table/Fig-1]: a) Non contrast abdominal CT scan, Axial image showing heterogeneous mass present in intraperitoneal compartment; b) Intravenous contrast abdominal CT scan. Coronal image showing heterogeneous mass present in mesentery which was inseparable from distal jejunal and proximal ileal loop.



[Table/Fig-2]: a) Gross examination of showed a grey white nodular tumor showing variably sized cystic spaces filled with haemorrhagic material (marked by arrows); b) Tumor composed of spindle cells arranged in interlacing fascicles with focal presence of cystic areas (H&E 100X); c) Cysts lined by bland looking ciliated columnar epithelium showing hemorrhagic material in the lumen as well as the wall (H&E 200X).

positive for Cluster of Differentiation 117 (CD117) and Discovered on GIST-1 (DOG1) [Table/Fig-3a]. Smooth Muscle Actin (SMA) showed only focal and weak positivity. Ki-67 proliferation index was 3-5%. The columnar epithelial lining of cystic spaces showed Estrogen Receptor (ER) positivity [Table/Fig-3b]. CD10 was positive in stromal cells present underneath the columnar epithelium [Table/Fig-3c]. Based on these morphological and IHC features, final diagnosis of Benign GIST with co-existent Intratumoural Endometriosis was given. The patient is doing well in a two-year follow-up and has showed no signs of recurrence or any malignant transformation. No further treatment was given to the patient.



[Table/Fig-3]: a) Immunohistochemistry (IHC) showing strong DOG-1 positivity in the spindle cells (200X); b) IHC with ER antibody, showing nuclear positivity in the columnar epithelial lining of cystic spaces (100X); c) IHC showing CD10 antibody positivity in stromal cells present underneath the columnar epithelium (marked by arrow); non-specific expression in fibrinous exudate (200X).

Endometriosis is associated with increased risk of several malignancies with the best evidence for ovarian cancer [1]. Although, genital and extragenital endometriosis is known to undergo malignant transformation; but occurrence of endometriotic foci within unrelated soft tissue tumour is extremely uncommon [2,3]. On extensive literature search, there were only two case reports where intramural endometriosis was reported, one was a case of renal angiolipoma and other was a composite tumour of vulva consisting of dermatosarcoma protuberans and giant cell fibroblastoma [4,5]. Kholová I et al., suggested a possible link to the occurrence of cutaneous endometriosis at previous surgery sites as the patient had seven tumour recurrences and had underwent frequent surgical excisions [5].

No case of GIST with intratumoural endometriosis is ever reported making this present case as the first one. While the pathogenesis was not commented upon in angiolipoma case, authors of composite vulval tumour suggested possibility of scar endometriosis as tumour was recurrent. Present case represents an incidental pathological finding and defies a definite comment on pathogenesis. Rarity of this entity can confuse an inexperienced pathologist and precludes an accurate diagnosis, especially in small biopsy specimen. The presence of intestinal endometriosis could have been one of the possible reason of chronic and acute haemorrhage in this case. However, no such foci were detected on colonoscopy.

Occurrence of endometriosis within GIST is a unique, hitherto undescribed phenomenon. Awareness of this entity is important for an accurate diagnosis and management.

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