Author's response to reviews

Title: Monophasic Synovial Sarcoma presenting as a primary ileal mass: Case Report and Literature Review.

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Primary Ileal Monophasic Synovial Sarcoma: Case Report and Literature Review.

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Abstract: Synovial sarcoma is a rare malignant mesenchymal tumor mainly arising in the periarticular tissue in young adults. There are few cases reported in other different areas. We report the first case of 29-year-old lady with synovial sarcoma arising in the ileum. The literature of this rare gastrointestinal tract sarcoma is reviewed.

Keywords: Synovial sarcoma, ileum.

Background: Synovial sarcoma is a malignant mesenchymal tumor of uncertain histogenesis. It may be biphasic, monophasic, or poorly differentiated [1]. Synovial Sarcoma is a rare entity representing 5-10% of all soft tissue sarcomas, typically occurring around the joints, mainly the knee [2, 3]. Occasionally, it arises in the head and neck [4, 5], lungs [6, 7, 8], heart [9], retroperitoneum [10], prostate [11] and intraneural [12]. There are few cases reported of synovial sarcoma arising in the gastrointestinal tract. We report a case of primary synovial sarcoma arising in the ileum of a 29 years old female. The patient's tumor displays morphologic features and immunohistochemical staining consistent with this disease. Our case is the first reported case arising in the ileum.

Case Report:

A 29-year-old female presented to Emergency Room complaining of 5-day history of epigastric and lower abdominal pain, intermittent, colicky in nature, associated with nausea and vomiting. No fever, change in the bowel habits or urinary symptoms. She is on regular follow up with the gynecology clinic because of lower abdominal heaviness and distension for several months as a case of uterine fibroids. She is divorced, nulliparous, with no PV bleeding. Her past history was significant for epilepsy on medications and she is 1 year post laparoscopic appendectomy with no
histopathology report. On examination; she was alert and conscious. Her vitals were stable. Her abdomen was distended with a hard pelvi-abdominal mass which is not tender. Her bowel sounds were positive.

CT abdomen and pelvis showed Pelvi-abdominal mass adjacent to the distal small bowel loops separate from the uterus and ovaries, heterogeneously enhancing, measuring roughly 65 x 99 mm with multiple areas of necrosis.[Fig. 1 A and B]

CT chest showed Indeterminate multiple bilateral sub-pleural 2-4 mm lung nodules, too small to biopsy.

The patient underwent diagnostic laparoscopy which showed ascitic fluid with a big mass at the lower abdomen. When biopsy was attempted, a cystic component of the mass ruptured and bleeding occurred. For this reason it was converted to open through a lower midline incision. A 10 X 10 cm mass arising from the terminal ileum, occupying the pelvis till the umbilical area was found. The uterus, ovaries and the urinary bladder were not involved. The tumour was excised with 10 cm free margins of small bowel.

The patient recovered well in the postoperative period and discharged home the 7th post operative day. She didn’t receive adjuvant treatment and she remained disease-free for 6 months with no clinical or radiological evidence of local recurrence.

Pathologic findings

Gross findings

The resected specimen composed of a segment of small bowel measuring 17 x 2 cm. It was adherent to a portion of ascending colon. An 8 x 7 x 3 cm white solid mass with homogenous cut surface was bulging from the serosal side of the small bowel and it is 4 cm away from the proximal resection margin. The tumor is adherent to the large bowel but not infiltrating it grossly. Few lymph nodes are identified within the mesenteric fat.
Histologic findings

Microscopically, the tumor was composed of cellular spindle cells with mild to moderate degree of pleomorphism arising from the small bowel wall and mainly involving submucosa, muscularis propria and serosa. Focal mucosal erosion is identified. The tumor cells show moderate amount of eosinophilic cytoplasm, evenly distributed nuclear chromatin and few conspicuous nuclei [figure 2A and B]. Mitotic count was 1-2 per 10 high-power fields. A metastatic focus was identified within a single mesenteric lymph node [figure. 2C].

Immunohistochemical findings

The tumor was positive for S100, EMA, BCL2, Vimentin and CD 99 [fig. 3A, B, C, D and F]. In addition to calretinin (focal), and synaptophysin (focal), CD56 (few cells). And it was negative for C-kit [fig.3E], CD34, CAM5.2, cytokeratin 7,20, 5/6 [fig. 3H], AE1/AE3, NSE, CD31, desmin [fig. 3G], Chromogranin, SMA, HMB45, Factor 8, Ber/EB4, and Melan-A.

Cytogenetic findings

Cytogenetic was conducted on formalin fixed paraffin embedded tumor sample. RT-PCR for synovial sarcoma-associated fusion transcript (SYT-SSX 1 and SYT-SSX 2) was negative.

Figure 2 A. a spindle cell neoplasm arising from the wall of the small bowel and pushing the mucosa toward the lumen, B. spindle cells with mild to moderate degree of pleomorphism and few conspicuous nuclei, C. a spindle cells metastatic focus in a mesenteric lymph node (hematoxylin- eosin, A. x2. B. x20. Cx4).
Discussion:

Gastrointestinal sarcoma accounts 0.1 to 3% of all GI malignancies and approximately 10% of all sarcomas [13, 14]. In the ileum, the majority of malignant sarcomas are GIST [15, 27]. To our knowledge there have been only 27 cases reported as synovial sarcoma arising in the GI tract, none of them in the ileum [16-32]. All of them shared the diagnostic histology and immunohistochemical features of synovial sarcoma. In 64% of the cases, the characteristic (X; 18) translocation was identified. We diagnosed our case depending on the diagnostic histology of monophasic synovial sarcoma and the IHC features. Although the molecular assay was negative for SSX-SYT1 and 2, the combined histological and immunohistochemical profiles were highly characteristic of monophasic synovial sarcoma.
The median age at diagnosis was 41.5 years (range, 14 to 75 years). There is no significant sex predilection with male to female ratio of 1:1.15. There are 11 and 12 cases of the GI tract synovial sarcoma aroused from the oesophagus (including one from the gastroesophageal junction) and stomach, respectively, representing the most common sites. The presenting symptom was pain, obstructive symptoms or bleeding. The histological subtypes were monophasic in 15 patients, biphasic in 11 patients including 2 patients with a poorly differentiated component. The remaining patients’ histological subtypes have not been reported. All of the reported cases treated surgically. The number of patients received adjuvant chemotherapy or radiotherapy or both are 5, 4 and 3, respectively. Only one patient had metastasis at first presentation. S. Billings [25] has reported a case of gastric SS with multiple liver metastases who died of the disease after 6 months. The survival period after diagnosis of all reported cases ranged from 1 to 224 months.

In soft tissue synovial sarcoma, numerous studies over the years have reported that it is a high grade malignancy with a high rate of metastasis leading to death; 5- and 10-year survival rates between 24- 68% and 11- 56%, respectively have been reported [33]. In the article by Bergh et al [33], the investigators have divided the synovial sarcoma patients into low and high risk patients depending on the patient’s age, tumour size and grade. A low risk group (patient age< 25years, tumour size< 5cm, and no histologic evidence of poorly differentiated tumour) and a high risk group (patient age ≥25 years, tumour size ≥ 5 cm, and poorly differentiated tumour). The question is: can we apply the same risk factors to the GI synovial sarcoma patients? I think we need more cases and more data about the published cases to study the behaviour of the same disease in a different part of the body. For the mean time, evidence says adequate primary surgery is essential to both local control and outcome.
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<td>intramural</td>
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<td>S</td>
<td>AWOD, 6</td>
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*The second case reported by Billings[25] could represent a metastatic focus of a primary neck tumor showed the same histopathological features.

Abbreviations: M, male; F, female; DOD, died of disease; AWOD, alive without evidence of disease; AWD, alive with residual disease; DOC, died of other cause; NR, not reported; S, surgery; Rad, radiotherapy; Chemo, chemotherapy.
References:


2. Kumar: Robbins Basic Pathology, 8th edition. 2007


