GASTROINTESTINAL STROMAL TUMOR PRESENTING AS SMALL BOWEL OBSTRUCTION IN THE SETTING OF NEUROFIBROMATOSIS TYPE 1.

Abhishek Ravinuthala, MD and Brian R Boulay, MD MPH. University of Illinois at Chicago, Chicago, Illinois.

Introduction

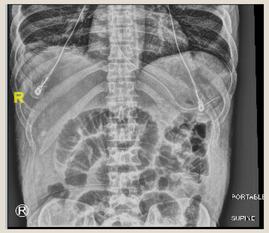
Gastrointestinal stromal tumors (GISTs) are mesenchymal masses that occur throughout the GI tract, predominantly in the stomach. GISTs typically present with symptoms of early satiety, vomiting, and signs of GI bleeding. They are normally sporadic but can be associated with certain hereditary conditions including Neurofibromatosis type 1 (NF-1). Here we describe the case of a patient presenting with small bowel obstruction and perforation secondary to GIST related inflammation.

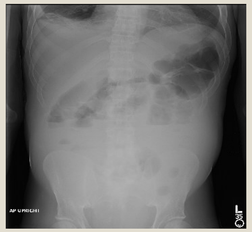
History of Present Illness:

A 46 year old man with a history of NF-1 and chronic constipation presented with progressive constant abdominal pain and distention for several days. He was previously evaluated in ER for similar symptoms 1 week prior and had not had a bowel movement since then. He endorsed nausea, abdominal pain, and the inability to pass gas or stool, as well as generalized weakness. On the other hand, he denied any hematochezia or melena. He denied any previous history of similar symptoms.

His gastrointestinal history is unremarkable aside from chronic GERD managed with Omeprazole.

In the ER he was afebrile and hemodynamically stable. Physical examination showed diffuse abdominal distention and tenderness without guarding or rebound tenderness as well as diffuse cutaneous neurofibromas. Serologic studies included normal white blood cell count and lactic acid level.





Evaluation and Intervention

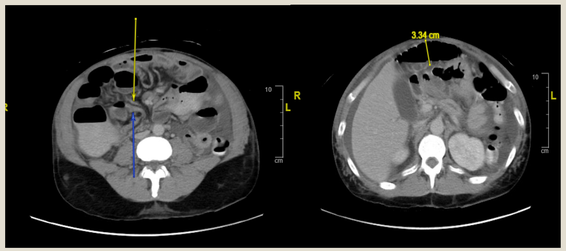
Initial abdominal radiograph revealed no intraperitoneal free air or fluid. Several mildly dilated loops of bowel without definite air present in colon.

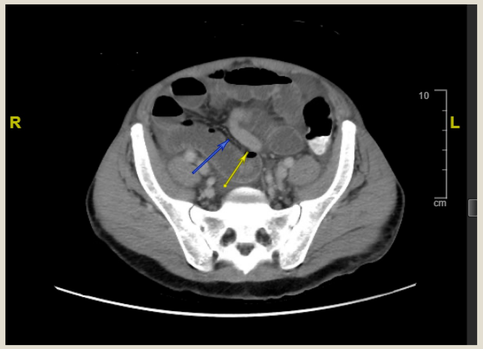
Point of Care Ultrasound revealed distended bowel loops without peristaltic forward flow.

CT abdomen/pelvis with IV contrast revealed small bowel obstruction with a transition point in the ileum and diffuse lymphoid tissue thickening along the mesenteric roots.

Patient initially managed with nasogastric tube placement. Symptoms continued to worsen, and patient failed conservative treatment.

CT Imaging Findings:

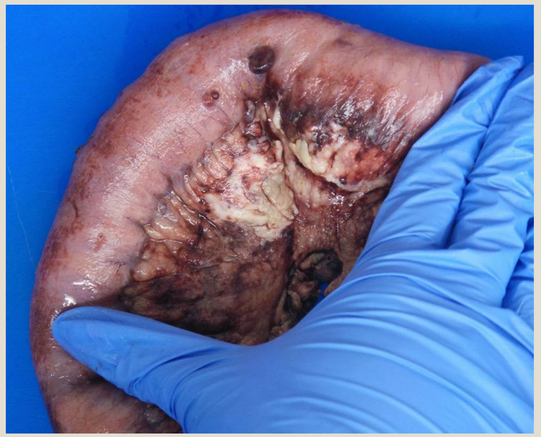




Surgical and Pathology Findings

Exploratory laparotomy was performed on hospital day 2. Intraoperatively the patient was found to have a thickened hemorrhagic mesentery in the central portions of the small bowel and a large adhesive band leading to an internal hernia. Segmental small bowel resection was performed.

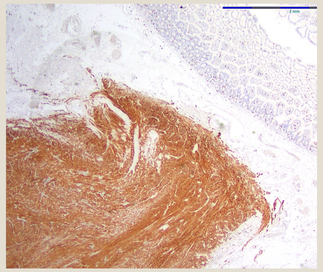
Pathology report confirmed the presence of two tumors measuring 0.8 cm and 0.2 cm within the jejunal wall at the junction of the mesentery, associated with serositis, a mural abscess and focal necrosis, and fibrinous adhesions. Tumor cells were positive for S100 and SOX10, as well as CD117 and DOG1, consistent with GIST.



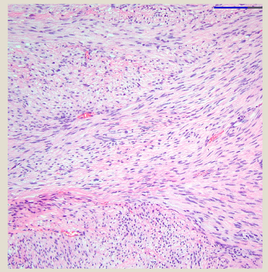
Gross specimen of small bowel resection showing bi-valved larger of two gastrointestinal stromal tumors (smaller nodule described as 2 mm is not shown here) viewed from the mesentery of jejunum.

Discussion/Conclusion

GISTs are a heterogenous group of masses that certain histological markers, and have an equally heterogenous pattern of presentation, varying widely based on tumor size and location. Most commonly found in the stomach, GISTs usually present with hemorrhage and anemia. Small bowel GISTs are next most common, and often associated with small bowel obstruction (SBO). With sporadic GISTs the obstruction is typically associated with a large tumor causing mechanical obstruction or intussusception. However, NF-1 patients may have GIST tumors which are small and multifocal, causing intense localized inflammatory changes which can result in bowel obstruction. It is particularly remarkable in our patient who had two nodular masses that were each less than 1cm wide, and yet produced such profound adhesions. Thus is an important diagnosis to consider in patients with new onset small bowel obstruction, especially in patients with predisposing conditions to GIST such as NF-1.



Serosal tumor shows diffusely and strongly positive immunohistochemical expression by CD117, shown here, and DOG1 (not represented here) confirming diagnosis of gastrointestinal stromal tumor. Positive expression is represented by brown immunostaining in contrast to negative expression within overlying unremarkable colonic mucosa.



Medium power view (!0 x objective) showing right angle fascicles of spindle cells consistent with gastrointestinal stromal tumor.

Contact

Abhishek Ravinuthala, MD
University of Illinois at Chicago
Email: Aravin8@uic.edu