Background

Gastrointestinal stromal tumors (GIST) are rare neoplasms and account for less than 1% of all gastrointestinal tumors and about 5% of all sarcomas. GIST most commonly occur in the stomach (60%–70%), followed by the small intestine (20%–30%) and less commonly in the colorectum (5%–10%). Rarely, they present as metastatic lesions elsewhere, with the most common metastatic sites being liver (65%) and peritoneum (21%). Clinical presentation may include gastrointestinal bleeding, abdominal mass, abdominal pain, or bowel obstruction and acute abdomen. The diagnosis is established with biopsy and immunohistochemical staining with more than 95% of tumors showing positivity for c-KIT (CD117), GIST 1 (DOG1) and in some cases CD 34.

Case Presentation

A 74 year old female with a history of diabetes mellitus type 2 and hypothyroidism presented with a four day history of generalized abdominal pain with distension, constipation and projectile bilious emesis. She denied any fevers, chills, night sweats, anorexia or weight loss, prior history of hematochezia, melena or change in bowel habits. She denied personal or family history of malignancy. She was afebrile on presentation and had stable vital signs. Physical examination revealed a distended abdomen with mild generalized tenderness, no guarding or rigidity, and hypoactive bowel sounds. Laboratory data was only significant for mild azotemia. Commuted tomography (CT) scan revealed a high-grade small bowel obstruction with possible transition in the right pelvis (Figure 1) with multiple nodular masses within the mesentry and omentum consistent with peritoneal studding as well as a 9.4 cm mass in the right hemi-pelvis (Figure 2 & 3). Nasogastric decompression evacuated one liter of bilious content. Serum CA-125 was elevated at 67.2 U/mL (ref 5.0–35.0 U/mL) while serum CA 19-9 and CEA were normal. CT guided biopsy of the omental mass revealed gastrointestinal stromal tumor (GIST) with immunohistochemical staining showing positivity for CD 117, DOG1, Vimentin and CD34 while being negative for pan-keratin, CK7, CK20, desmin, Pax8 and S100 proteins. Mitotic activity was low (<5/50 per HPF). These findings confirmed the diagnosis of metastatic GIST. She was started on Imatinib therapy with significant reduction of tumor size and improvement in the patient's symptoms on six week follow up.

Discussion

GIST are rare neoplasms, comprising only <1% of gastrointestinal tumors and are even rarer to present as metastatic lesions. Early diagnosis and surgical resection of localized disease is the only curative intervention. It has been shown that large tumor size, high mitotic rate and tumor location in the small intestine are all associated with an increased recurrence rate. This patient presented with a pelvic tumor with high CA 125 concerning for a metastatic ovarian tumor, but was eventually diagnosed with metastatic GIST. Early histopathological diagnosis with mutational analysis enabled the patient to respond well to systemic tyrosine kinase inhibitor therapy and thus reinforces the importance of keeping a high index of clinical suspicion even if the tumor entity is rare.

Figure 1

Figure 2

Figure 3