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Case report of diffusely metastatic rectal GIST

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ABSTRACT

This is a case report of an aggressive, diffusely disseminated Stage IV rectal gastrointestinal stromal tumor (GIST) in a 57-year-old male that presented for symptoms of malaise, constipation, and twenty pound weight loss in 2 months. Upon rectal examination, a hard 4 centimeter submucosal mass was found at the 9–12 o'clock position. Liver and lung metastases were visualized on computerized tomography (CT) of the chest, abdomen, and pelvis on metastatic work-up. He was deemed a poor surgical candidate due to diffuse metastatic disease and referred for palliative chemotherapy. The patient had suffered a perforation of his rectal wall two weeks after his initial presentation and passed away shortly thereafter. He never received palliative chemotherapy. We present a case report as a unique case of an extremely aggressive and quickly fatal GIST tumor.

1. Introduction

A gastrointestinal tumor (GIST) is a rare mesenchymal tumor of the gastrointestinal tract, with only approximately 6000 cases reported each year in the United States [1]. The stomach is the most common location of GIST presentation (60–70%), followed by small bowel (25%), rectum (2%), and the esophagus (2%), with other various locations accounting for the rest of the presentations [1]. GISTs arise from both smooth muscle and neural elements with the cell of origin believed to be the interstitial cells of Cajal, an intestinal pacemaker cell [1].

The diagnosis of GIST is made through immunohistochemistry and electron microscopy where the presence of staining for tyrosine kinase receptor KIT (CD 117) is confirmatory for the presence of the interstitial cells of Cajal [2]. It is estimated that over 95% of GIST tumors exhibit staining for CD 117 making it a reliable tumor marker in the diagnosis of GIST [2]. Approximately two-thirds of GISTs exhibit CD34 staining. Histologically, these tumors may exhibit a spindle pattern, an epithelioid pattern, or a mixed subtype [2].

Diagnosis of GIST is usually made on the basis of computerized tomography (CT) of the abdomen and pelvis or magnetic resonance imaging (MRI). Endoscopic ultrasound may also facilitate a biopsy if the tumor appears unresectable. If the tumor appears resectable, biopsy should not be performed, because of the risk of tumor rupture and possibility of upstaging the malignancy through intraabdominal spillage of tumor cells [1].

We present a case we encountered at our large academic institution of a highly aggressive rectal GIST tumor that was not amenable to resection, resulted in rectal perforation, and patient death approximately two weeks after the initial diagnosis.

2. Case Presentation

This is a 57-year-old male patient with past medical history of obesity and hypertension who was found to have a rectal mass on initial clinic visit. The patient has never had a colonoscopy, he was occult blood negative, and his hemoglobin and liver function tests were normal. He reported a twenty-pound weight loss over a period of two months duration along with generalized malaise, decreased appetite, and constipation. He also described abdominal pain located in the mid-epigastric region with radiation to the right upper and left upper quadrants of the abdomen, worse with food intake. He had family history significant for breast, lung, and bone cancer on his mother's side. His psychosocial history was significant for occasional drinking, never smoking, and he lived by himself.

Physical exam revealed a palpable mass on rectal exam located at the 9–12 o'clock position 6 centimeters (cm) from anal verge visualized on anoscopy (Fig. 4). He did not appear cachectic. CT of the chest, abdomen, and pelvis for staging revealed diffuse metastatic disease to the liver, poorly circumscribed mass involving the rectum measuring 3.6 cm by 6.0 cm with two adjacent left-sided pathologic lymph nodes (Fig. 1). He also had a small lung nodule.

Rectal mass biopsy of a partially obstructing lesion on colonoscopy revealed sections of inflamed mucosa with areas of spindle cells and focal epithelioid cell proliferation that lied adjacent to the mucosa. The immunohistophenotype revealed the

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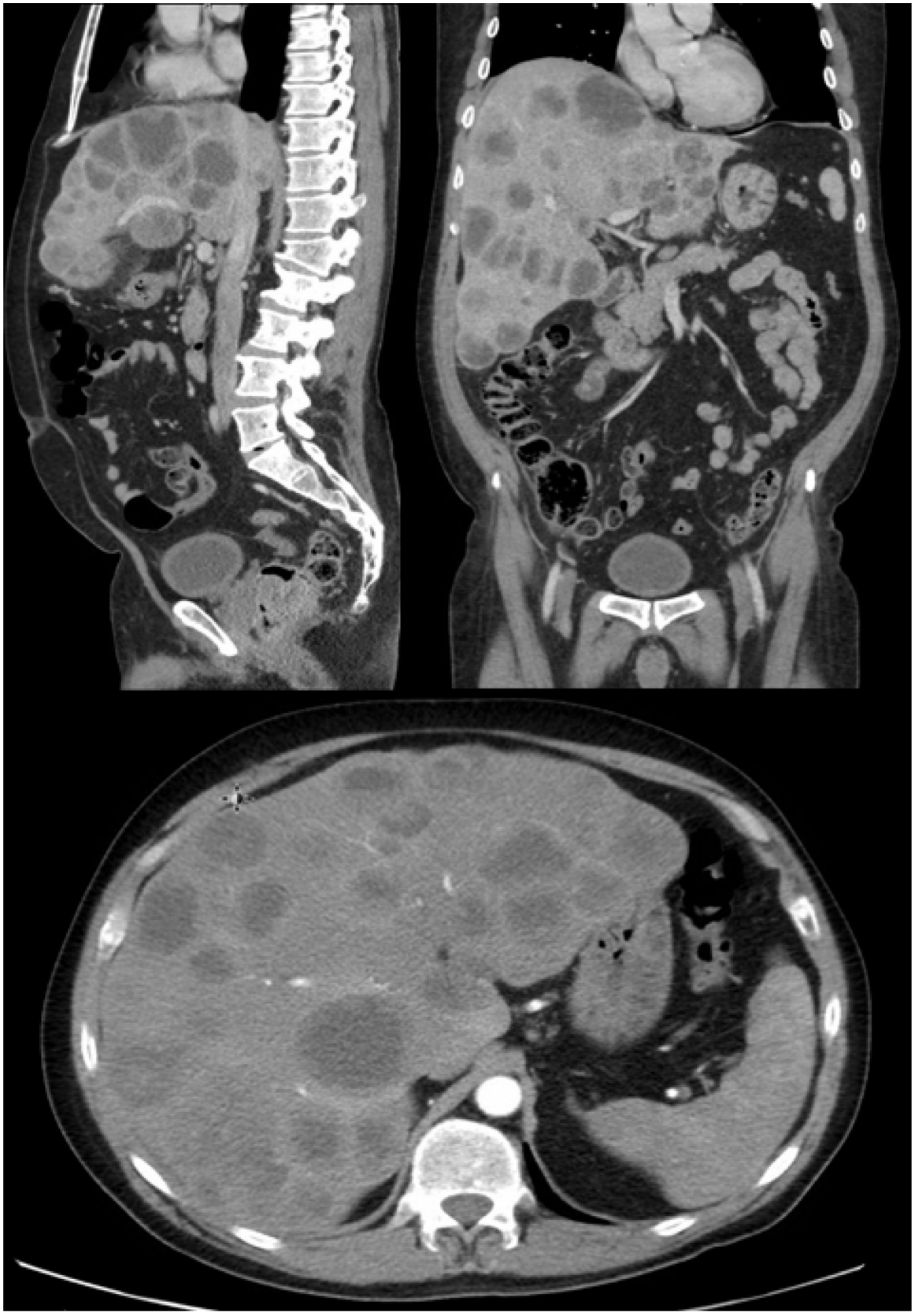

Fig. 1. Stage IV rectal gist found on CT of the Abdomen and Pelvis with diffuse liver metastases with asymmetric rectal mass measuring 3.6 cm × 6 cm in size.

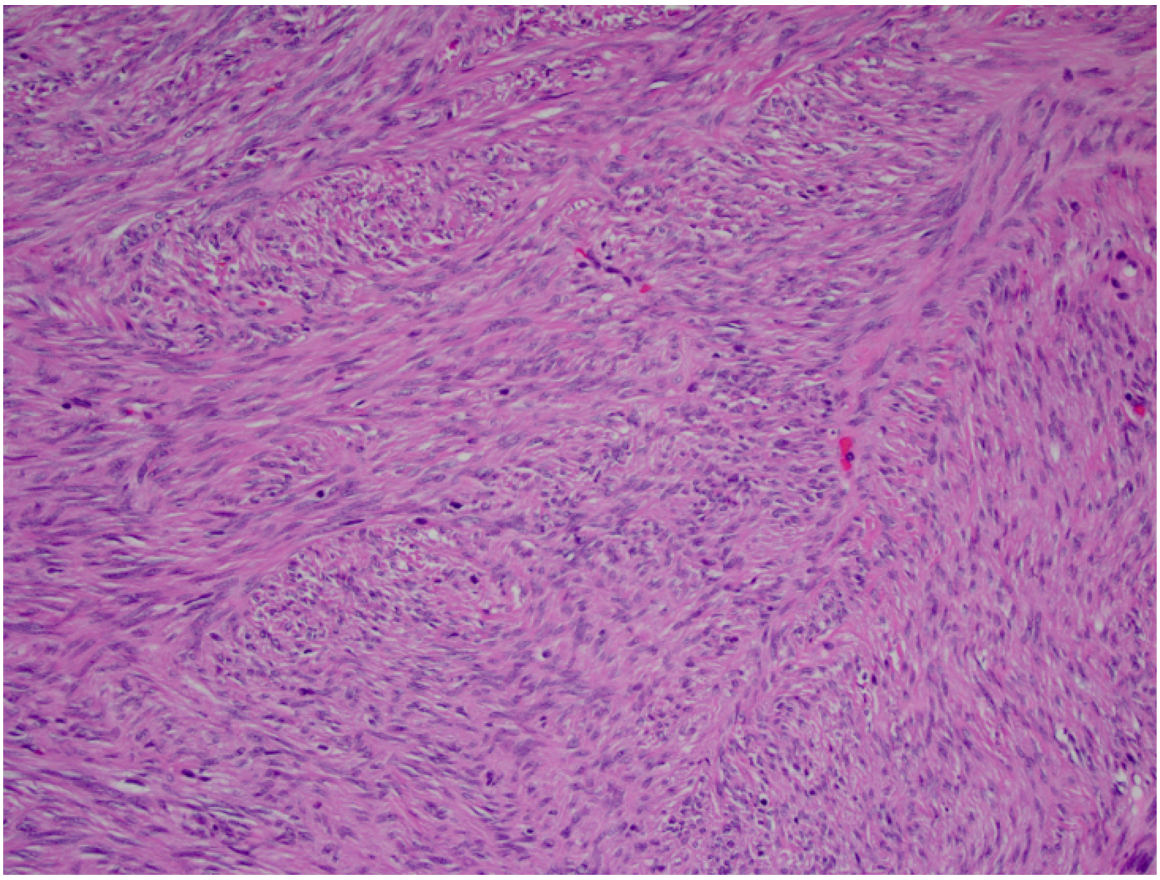

Fig. 2. Stage IV rectal gist histology revealed the tumor to be positive for CD117, CD34 and negative for CKAE1/AE3, CK7, CK20, synaptophysin, SM actin which was consistent with a GIST. The mitotic rate was 6 per 30 HPF or 8.3 per 50 HPF.

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He was determined to be a nonsurgical candidate due to diffuse metastatic disease and referred to oncology clinic for palliative chemotherapy (Fig. 3).

He presented to the emergency department two weeks later with abdominal pain and free air in his abdomen and imaging from outside hospital that revealed a rectal perforation (Fig. 6). The patient refused surgery as he did not want to undergo an invasive procedure with little chance for cure and wished to go home and be with family for comfort measures and passed away two weeks later. Due to the rapid nature of his tumor progression, he never received treatment with imatinib mesylate.

3. Discussion

Gastrointestinal stromal tumors can arise anywhere in the gastrointestinal region with 2% occurring in the rectum [1]. While the majority of GISTs have a benign course, a GIST tumor can rarely exhibit an aggressive course. Such was the case in our patient. Prognosis on aggressiveness of a GIST tumor can usually be made on examination of tumor size and histologically [3]. If the tumor is less than 2 cm and mitotic count lower than five per fifty high-power field (HPF), the risk of aggressive disease is thought to be very low. Alternatively, if the tumor is larger than 10 cm, and if the mitotic count is higher than ten per fifty HPF, the risk of aggressive clinical behavior is considered to be high (Fig. 5). Of note, gastric GIST tumors are thought to have a better prognosis compared to small bowel and rectal GIST tumors [4].

GIST tumors are typically managed with surgical resection to negative margins and an intact pseudocapsule [1]. Lymph node involvement is rare with GISTs [1]. As such, there is no role for lymphadenectomy or sentinel lymph node biopsy, as is the case with most cancers of mesenchymal origin. If the tumor presents with metastases or local advancement to the point where surgical therapy would result in excessive morbidity, the patient is treated with tyrosine kinase inhibitor imatinib mesylate [5]. In the case of a small rectal GISTS, local excision can be attempted. In a GIST greater than 2 cm, either a low anterior resection or an abdominoperineal resection should be performed depending on the distance from the dentate line [1].

Rectal GISTs are extremely rare and most are amenable to surgical resection. Rectal GISTs account for approximately 2% of all GIST tumors, 0.1% of all colorectal tumors, and have an estimated incidence rate of 0.45 per million persons [6]. However, the clinical course and treatment of GISTs are similar regardless of site of origin.

The discovery of imatinib mesylate therapy in the 1970s revolutionized the treatment of GISTs and significantly improved the survival of patients. The overall survival rate for a patient with a rectal GIST varies based on its tumor grade [3]. Those with low-grade rectal GISTs have a significantly longer survival than those with a high-grade GIST (median 5–10 years versus 2–3 years respectively) [3,6]. The limited available data suggests that only 10–20% of patients with rectal GISTS are cured by resection and that recurrence is a potential risk for up to 15 years [6]. The most common cause of death after resection of a rectal GIST is distant metastasis, rather than local recurrence, and involvement of the liver is a frequent event [6].

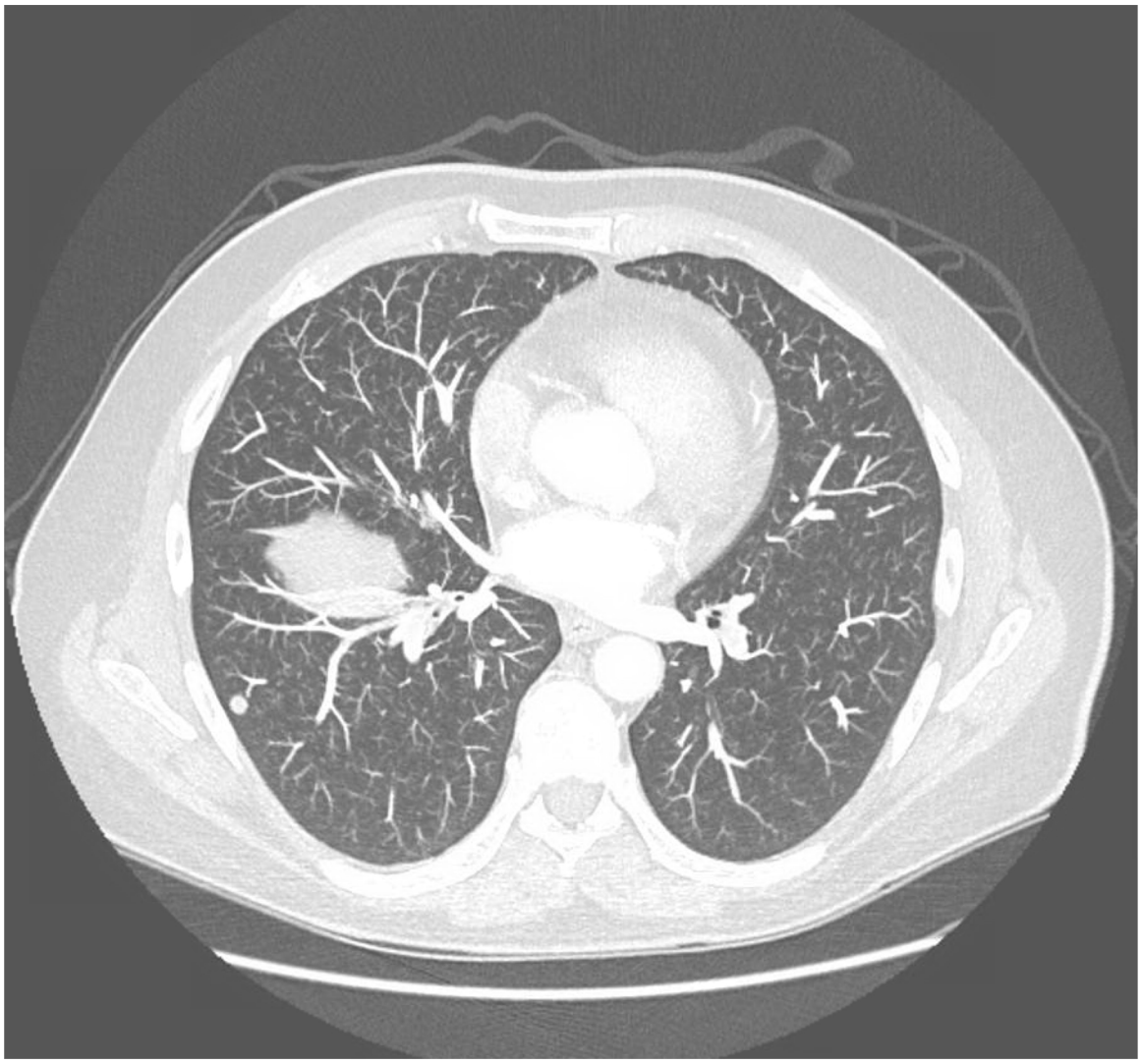

Fig. 3. Stage IV rectal gist found on CT of the Abdomen and Pelvis with a small right pulmonary nodule suggestive of metastases to the lung.

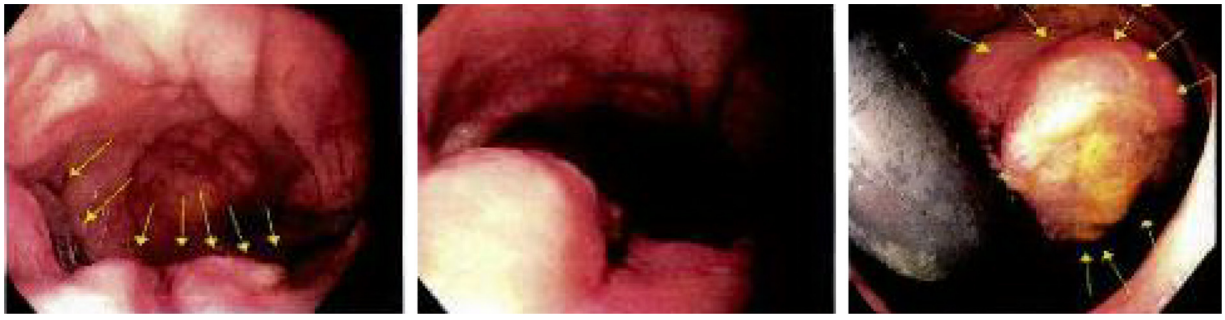

Fig. 4. Rectal mass found on colonoscopy which was biopsied and later found to be a Stage IV GIST rectal cancer.

Surgical resection of hepatic metastases of GIST tumors has not been effective [7–9]. The local recurrence rate of rectal GIST is high at 67% [6]. As with other GIST tumors, adjuvant chemotherapy with imatinib mesylate has been found to be effective in the treatment of rectal GISTs [11,12]. Overall, this case demonstrates that "high-risk" rectal GISTs can be very aggressive in nature and may result in rectal perforation.

4. Conclusions

GIST tumors of the rectum, while rare and thought to have a good prognosis, need to be approached with caution due to their relatively unpredictable nature of aggressiveness. Imatinib mesylate chemotherapy must be started as soon as feasible. A large, high-risk rectal GIST based on mitotic power and size of tumor >5 cm is associated with aggressive metastatic dissemination to the liver and is strongly suggestive of poor survival.

	Size*	Mitotic Count†
Very low risk	<2 cm	<5/50 HPF
Low risk	2–5 cm	<5/50 HPF
Intermediate risk	<5 cm	6–10/50 HPF
	5–10 cm	<5/50 HPF
High risk	>5 cm	>5/50 HPF
	>10 cm	Any mitotic rate
	Any size	>10/50 HPF

Fig. 5. Risk stratification of GIST based on size and mitotic count from Fletcher et al. [3]. Abbreviations: HPF, high-power field.

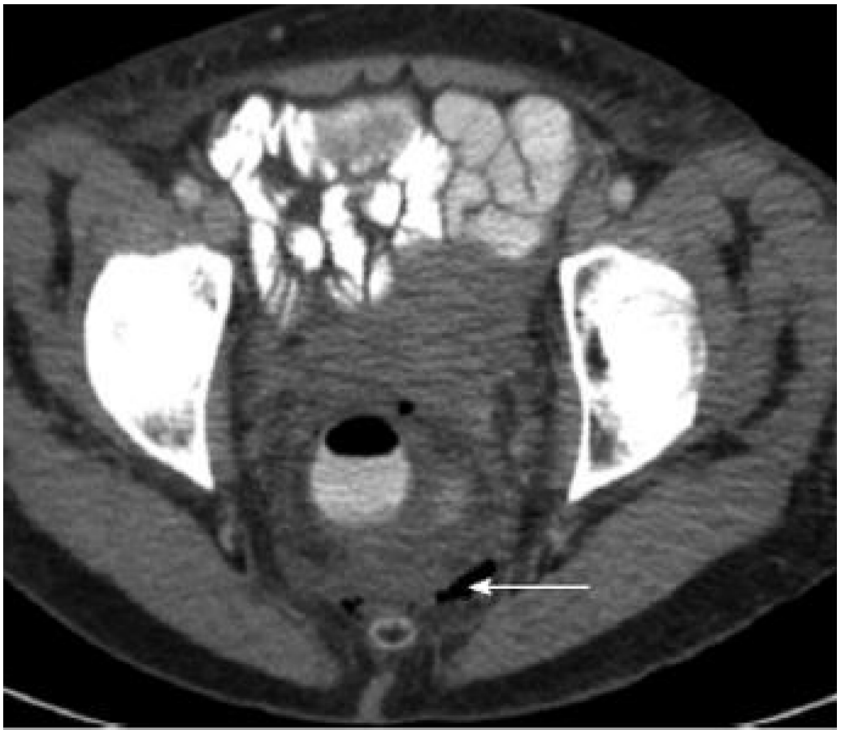

Fig. 6. Rectal perforation found on CT of the Abdomen and Pelvis with free air seen in the mesorectum.

Disclosure statement

The authors of this paper have nothing to disclose.

Conflicts of interest

The authors declare that they have no competing interests. We have no personal or financial conflicts of interest related to the preparation and publication of this manuscript.

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Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Author contribution

Yana Puckett, MD: Writing manuscript.
Amir Aryaie, MD, FACS: Writing manuscript, editing.

Aknowledgement

Our work has been reported in line with the SCARE criteria [10].

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