Abdominotransanal resection of a strangulated rectal carcinosarcoma

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SUMMARY

This is a case of a 75-year-old man who presented with a 7-month history of a reducible rectal mass. The patient came to the emergency department with a prolapsed necrotic bowel involving a strangulated segment with the rectal mass. He underwent an abdominotransanal resection through a combined abdominal and perineal approach. His postoperative course was unremarkable. Histopathological and immunohistochemical studies showed a rectal carcinosarcoma. Because of a state-mandated lockdown due to the COVID-19 pandemic, the patient failed to follow-up. He was later seen to have metastatic progression. Owing to the poor functional status of the patient, the shared decision of the multidisciplinary team, the patient and his family was to manage him with palliative intent.

BACKGROUND

Carcinosarcoma is a rare type of mixed mesodermal malignancy consisting of both of an epithelial and a mesenchymal component. 1 When encountered, it usually affects the head and neck region. Very rarely, these tumours may arise from the gastrointestinal tract—with an even lower reported incidence in the more distal segments. Prior to this report, only eight reported cases have fulfilled the criteria for rectal carcinosarcoma. This report documents a case of a 75-year-old man who underwent surgical resection for a strangulated rectal carcinosarcoma.

CASE PRESENTATION

A 75-year-old man came to the emergency department with a prolapsed rectal mass. He had no known comorbidities and had no previous surgery. He presented with a 7-month history of a reducible rectal mass with associated colicky abdominal pain, weight loss, hematochezia and incontinence. Prior to this consult, he was seen by a private physician, and was managed as a case of haemorrhoidal disease without the benefit of a rectal examination or a colonoscopy. Due to the persistence of symptoms, he consulted with another physician who was able to detect the presence of a fungating rectal mass. No biopsy of the mass was performed.

INVESTIGATION

A contrast-enhanced CT scan showed a multilobulated, heterogeneously-enhancing intraluminal mass with internal calcifications in the rectal area intimately related to the prostate. No liver lesions were noted. Note that this imaging was performed prior to the incarceration of the rectal mass.

Figure 1 Representative cuts of the abdominal CT scan with triple contrast enhancement showing the multilobulated, heterogeneously-enhancing intraluminal mass with internal calcifications in the rectal area intimately related to the prostate. No liver lesions were noted. (figure 1).

Persistent prolapse of the rectal mass prompted consult at our institution. The patient was haemodynamically stable with unremarkable abdominal findings. Rectal examination revealed a 15 × 10 cm necrotic, foul-smelling prolapsed mass (figure 2).

TREATMENT

The patient was prepared for emergent surgery, for which an abdominotransanal resection (ATAR) with no neorectal construction, using a combined abdominal and perineal approach, was performed. The abdominal phase was started with a midline infraumbilical laparotomy incision that extended to the symphysis pubis. This allowed better exposure for the dissection. A lateral-to-medial mobilisation of the sigmoid colon from the peritoneal reflection, and high ligation of inferior mesenteric...
artery and vein were performed, followed by total mesorectal excision (TME). TME involves the removal of the entire rectum, including the surrounding lymphovascular tissues, using sharp dissection. The perineal phase was performed starting with a circumferential incision at the mucocutaneous junction of the anal opening. Total intersphincteric resection then followed, which was performed by circumferentially dissecting between the external anal sphincter and the internal anal sphincter. This resulted in the inclusion of the internal anal sphincter in the resection of the tumour, while preserving the external anal sphincter. The mass was delivered through the perineal incision, and a permanent stoma was created.

Intraoperatively, an irreducible fungating rectal mass was seen prolapsing distally beyond the anal verge, measuring 23.5 cm in greatest diameter (figures 3 and 4). There were areas of necrosis and foul-smelling discharge. Further exploration showed enlarged mesenteric nodes along the inferior mesenteric vessels, dilated proximal bowel loops and serous pelvic ascites. No signs of distant spread were noted. The postoperative course was uneventful.

Histopathology revealed a high-grade malignancy for which the primary consideration was a carcinosarcoma, involving the submucosa and perirectal fat, with associated lymphovascular space invasion (figure 5). All 19 pericolic and pararectal lymph nodes; the proximal, distal and radial margins of resection; and the peritoneal fluid collected were negative for malignant cells. Immunohistochemical (IHC) testing confirmed the diagnosis of rectal carcinosarcoma (ie, cytokeratin (CK) AE1/AE3 positive in the epithelial cells, and vimentin positive in the mesenchymal cells) (figure 6).

OUTCOME AND FOLLOW-UP

Due to the delay in adjuvant treatment brought about by the COVID-19 pandemic, the patient developed metastatic spread to the lungs, retroperitoneal nodes and bones. Owing to the poor functional status of the patient, the shared decision of the multidisciplinary team (MDT), the patient and his family was to
Table 1  Summary of the documented cases of rectal carcinosarcoma

| Study          | Age/sex | Chief complaint   | Onset of symptoms | Distance from anal verge | Size of the tumour | Metastasis | Surgery                                | Lymph node involvement | Neoadjuvant or adjuvant treatment | Immunohistochemical studies                          | Recurrence | Survival |
|----------------|---------|-------------------|-------------------|--------------------------|--------------------|------------|----------------------------------------|------------------------|----------------------------------|-------------------------------------|------------|----------|
| Roncarioli et al  | 71/F    | Abdominal pain    | 2 months          | 2 cm                     | 8 cm               | N          | Anorectal resection with colostomy    | Y                      | Preoperative RT                   | Epithelial: cytokeratin and chromogranin, Mesenchymal: vimentin, desmin and actin | Pelvis after 2 months | 6 months |
| Takeyoshi et al  | 82/M    | Rectal bleeding   | Not reported       | 2 cm                     | Not reported        | N          | LAR                                    | N                      | N                                | Epithelial: CEA, Mesenchymal: vimentin | Skin after 1 month | 6 months |
| Ishida et al     | 80/F    | Abdominal pain    | Not reported       | 18 cm                    | Not reported        | N          | Hartmann procedure en bloc ileal resection-anastomosis | Y                      | N                                | Epithelial: cytokeratin and CEA, Mesenchymal: desmin, myoglobin, HHF35 and S-100 | Pelvis after 5 months | 6 months |
| Ozturk et al     | 65/F    | Rectal bleeding   | 3 months          | 2.2 cm                   | 2 cm               | N          | APR                                    | N                      | N                                | Epithelial: cytokeratin, Mesenchymal: vimentin and S-100 | N          | 5 years  |
| Tsekouras et al  | 60/M    | Constipation      | Few weeks          | 7 cm                     | 7 cm               | N          | APR                                    | Y                      | Postoperative chemoRT             | Epithelial: CAM, MNF and p53, Mesenchymal: vimentin and actin | Lungs, liver, peritoneum, pelvis and inguinal node after 4 months | 6 months  |
| Jeong et al      | 13/F    | Abdominal pain    | 3 weeks            | Not reported             | 13 cm              | N          | LAR                                    | Y                      | Postoperative chemotherapy        | Epithelial: cytokeratin, epithelial membrane antigen and CEA, Mesenchymal: vimentin | --         | --       |
| Kolodziejczak et al | 83/M    | Rectal bleeding   | Not reported       | 5.3 cm                   | Not reported        | N          | R2 resection of tumour with sigmoid loop colostomy | N                      | N                                | Epithelial: cytokeratin, Mesenchymal: vimentin and SMA | R2 resection only | 5 weeks  |
| Sudolov et al    | 80/F    | Rectal bleeding   | 4 weeks            | 4.2 cm                   | Not reported        | N          | Laparoscopic-assisted APR | Y                      | Preoperative RT                   | Epithelial: cytokeratin and CEA, Mesenchymal: vimentin, SMA and desmin | Pelvis, lungs after 16 months | 25 months |
| Manlubatan et al (2021) | 75/M    | Rectal mass       | 7 months           | At the verge            | 23.5 cm            | N          | Abdominotransanal resection           | Y                      | N                                | Epithelial: cytokeratin, Mesenchymal: vimentin | Lung, nodes and bone after 9 months | --       |

APR, Abdominoperineal resection; CEA, carcinoembryonic antigen; chemoRT, chemoradiotherapy; LAR, Low anterior resection; N, no; RT, radiotherapy; SMA, smooth muscle actin; Y, yes.
DISCUSSION
First described by Virchow in 1864, carcinosarcoma is a rare mixed mesodermal malignancy that consists of both glandular epithelial (carcinoma) and mesenchymal or connective tissue (sarcoma) components. In literature, the most common sites of occurrence are the head and neck regions and the female urogenital system. When seen in the gastrointestinal tract, it is most commonly seen in the oesophagus, and very rarely in the colorectum. It does appear that the more distal its location is in the gastrointestinal tract, the less frequently is it encountered. Among published case reports prior to 2021, a total of eight cases of rectal carcinosarcoma were described (table 1).

In a review of these cases, carcinosarcoma within the rectum was found mostly among the older population (mean age 67). Only one paediatric case has been reported to this date.

The malignant behaviour of carcinosarcoma is similar to adenocarcinoma with its tendency to metastasise to lymph nodes and distant sites. The sites of metastasis identified among the previous studies include the liver, lymph nodes, peritoneum, lungs and skin. Note that in the previous case reports, none of the patients had distant metastases on detection of the primary tumour. Similarly, chest CT scan performed for this patient did not show any pulmonary metastasis.

Clinical signs and symptoms of colorectal carcinosarcoma are related to obstruction and bleeding depending on the localisation, similar to other colorectal malignancies. These signs and symptoms manifest late, and approximately 50% of patients with colorectal carcinosarcoma present with obstructive symptoms.

Prior to consultation at our institution, this patient was seen months ago by a primary care physician, and was managed as a case of haemorrhoidal disease due to the aforementioned signs and symptoms without the benefit of a rectal examination and colonoscopy. For any patient presenting with abdominal or colorectal symptoms, proper physical examination is essential; supported by appropriate diagnostic modalities, as indicated, such as colonoscopy. Screening colonoscopy should be started as early as 50 years of age for the general population.

Histopathological evaluation of the tumour is crucial in establishing the diagnosis. This shows a biphasic population composed of both malignant stromal and epithelial nature. The mesenchymal stromal component is composed of high grade pleomorphic spindle to epithelioid cells with high grade nuclear atypia and bizarre giant cells. There are no heterologous components identified. Malignant glands with nuclear atypia are found randomly dispersed into the sarcomatous component. There is no transitional area appreciated between the two populations.

Immunohistochemistry remains the gold standard for diagnosing carcinosarcoma. The two key features are differential staining of the distinct components (ie, positive IHC staining for both CKs and vimentin), and the lack of staining for epithelial markers in the sarcomatous component. These signs and symptoms manifest late, and approximately 50% of patients with colorectal carcinosarcoma present with obstructive symptoms.

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Immunohistochemistry remains the gold standard for diagnosing carcinosarcoma. The two key features are differential staining of the distinct components (ie, positive IHC staining for both CKs and vimentin), and the lack of staining for epithelial markers in the sarcomatous component. The carcinosarcoma component commonly shows reactivity to CK20 and carcinoembryonic antigen, whereas the sarcomatous component stains positively for vimentin, desmin, and smooth muscle actin. The biphasic component mentioned was further emphasised by the IHC studies, with CK AE1/AE3 highlighting only the carcinosarcoma cell population. Further IHC studies with mesenchymal markers, including H-caldesmon, DOG1, S100, SALL4 and CD31 do not impart a specific lineage differentiation for the sarcomatous component (figure 7).

With the following features, a diagnosis of carcinosarcoma was rendered.

The nomenclature for this diagnosis has been constantly changing, with ‘carcinoma with sarcomatoid component’ being the closest term in the latest edition of the WHO Classification of Tumours. However, with the prominence of the sarcomatous component in this tumour, it is the pathologist’s decision to retain the title of ‘carcinosarcoma’ as the more appropriate diagnosis.

No treatment guidelines have been established due to the rarity of this condition, but case reports have suggested that it should be treated in a similar way to the more common rectal adenocarcinoma. However, long-term benefits of neoadjuvant and adjuvant chemoradiotherapy have not yet been proven.

Of the eight reported rectal carcinosarcoma cases, two underwent neoadjuvant radiotherapy, one of which had recurrence, the other one had none. Two other patients underwent adjuvant treatment, one of which had documented recurrence.

For this patient, since there were signs and symptoms of bowel necrosis, he was brought to the operating room for an emergency procedure. An ATAR with no neorectal construction was performed.

Learning points
- Rectal carcinosarcoma is a rare entity that consists of both epithelial (carcinoma) and mesenchymal (sarcoma) components. Immunohistochemical study is the gold standard to diagnose this disease.
- Meticulous history-taking and thorough physical examination are imperative to avoid misdiagnosis of this rare and aggressive tumour.
- For the general population, screening colonoscopy should be started as early as 50 years of age. For any patient presenting with gastrointestinal symptoms, endoscopy should be performed, as indicated, to allow for early detection and timely management of any pathology.
- No standardised treatment guidelines have been established, but reports have suggested that it should be managed in a similar way to a rectal adenocarcinoma. Further investigation is needed to effectively manage this disease.
- Due to the aggressive nature of this disease, strict follow-up and close monitoring is highly encouraged. Likewise, a multidisciplinary team approach is important to render the best care for the patient.

Figure 7 Further immunohistochemical evaluation shows no specific lineage differentiation for the sarcomatous component of the tumour. (A) Caldesmon, (B) DOG1, (C) S100, (D) SALL4, and (E) CD31.
This technique has better outcomes in terms of healing and function compared with an abdominoperineal resection, where both the external and internal anal sphincters are removed. Had this been detected and diagnosed earlier, neoadjuvant therapy could have been offered as part of an investigational treatment option to decrease the size of the tumour prior to surgery. The collaborative efforts of a MDT, including colorectal surgeons, medical oncologists and radiation oncologists, is vital in planning the best strategy for this kind of patients.

In the limited case reports available, survival of patients with rectal carcinosarcoma ranged from 5 weeks to 5 years. Prognostic factors have not been clearly identified, but some of the indicators that were used include the size of the mass, lymphatic or vascular invasion, stage and the histology of the carcinomatous component, whether low, mid or high grade.\(^5\) Due to the aggressive nature of this disease, strict follow-up and close monitoring is recommended.\(^10\)\(^12\)

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