

# Peritoneal Carcinomatosis Arising from Primary Anorectal Melanoma

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## ABSTRACT

Anorectal melanoma is a rare and aggressive malignancy with a poor prognosis. Anorectal melanoma makes up approximately 1 to 3% of all anorectal malignancies. There are no known risk factors for anorectal melanoma. Patients frequently experience a delay in diagnosis due to multiple factors including nonspecific symptoms and misdiagnosis for other benign entities. Anorectal melanoma has a high potential for distant metastases and radiographic imaging plays a key role in evaluating for metastatic disease. Common sites for metastasis include pelvic lymph nodes, lungs, liver, skin, and brain. We present a case report of a 75 year old female with a history of transanal excision of primary anorectal melanoma who presented with increasing abdominal pain and distention. Computed tomography scan of the abdomen and pelvis showed metastatic disease to the peritoneum with findings of extensive peritoneal carcinomatosis, demonstrating the aggressive nature of anorectal melanoma.

## CASE REPORT

### CASE REPORT

A 75 year old female presented with a bleeding anal mass 5 years ago. She underwent colonoscopy and biopsy of the anal mass, which led to the diagnosis of anorectal melanoma. She was subsequently treated with transanal excision followed by postoperative radiotherapy and ipilimumab immunotherapy. She was in sustained remission until 1 year ago when she developed a local recurrence of tumor with biopsy results demonstrating malignant infiltration of the lamina propria with immunohistochemical staining positive for S100 and melan A, consistent with recurrent malignant melanoma (Figure 1 and 2). She underwent a second transanal excision of the recurrent disease at this time.

Nine months later the patient felt another enlarging anal mass and had a third transanal excision. A CT of the abdomen and pelvis performed approximately one month prior to the third excision demonstrates no evidence of metastatic disease (Figure 3). Approximately three weeks after the third excision, she presented to the hospital with worsening abdominal pain and distention and loss of appetite. Abdominal radiograph showed signs of ascites with bulging flanks and diffuse increased density of the abdomen (Figure 4). Following the third excision, the patient underwent a CT of the abdomen and pelvis, which showed peritoneal thickening with innumerable mesenteric and peritoneal nodules and ascites, compatible with new extensive peritoneal carcinomatosis (Figures 5-7). During the patient's hospital course she underwent abdominal ultrasound to evaluate for abdominal paracentesis, which showed peritoneal thickening and

nodularity with ascites (Figure 8). Due to the extent of disease spread and poor prognosis, the patient was referred to palliative medicine. She died three weeks after initial presentation to the hospital.

## DISCUSSION

### Etiology & Demographics:

Anorectal melanoma is a rare and aggressive disease with a reported incidence of 1 to 3% of all anal tumors [1]. The incidence rate of anorectal melanoma in the United States has been estimated as 2.7 cases per 10 million people per year [2]. Melanomas arise from melanocyte cells which are a derivative of neural crest cells. There are melanocytes located throughout the body including the skin, eyes, and mucosal surfaces. There are numerous melanocytes in the mucosa of the anal canal which explain why melanoma may develop in the anorectal region. Cutaneous melanoma develops when melanocytes are exposed to ultraviolet B light and undergo malignant transformation. The pathogenesis for anorectal carcinoma however is less understood [3]. There are no known risk factors for anorectal melanoma. Anorectal melanoma is the third most common type of melanoma after cutaneous and ocular melanomas. Anorectal melanoma represents less than 1% of all melanomas [4]. The onset of disease typically presents in the 6<sup>th</sup> decade of life with some studies reporting a higher prevalence among females [5, 6].

### Clinical & Imaging Findings:

Anorectal melanoma will present with similar clinical symptoms as other rectal disorders including bleeding, constipation, rectal pain, and change in bowel habits. It is often initially misdiagnosed for other benign entities such as hemorrhoids and polyps. Colonoscopy and direct visualization are essential in making the diagnosis [5]. Our patient initially presented with a bleeding anal mass which was biopsy-proven melanoma.

Anorectal melanoma is a highly aggressive neoplasm with approximately 32% of patients presenting with metastatic disease at the time of initial diagnosis. The most common sites of metastasis include the liver, lungs, and brain [7]. We present a case of primary anorectal melanoma with metastasis to the peritoneum resulting in peritoneal carcinomatosis. Peritoneal carcinomatosis represents widespread metastatic disease within the peritoneal cavity. Peritoneal carcinomatosis is typically seen with many organ based malignancies including ovarian tumors and gastrointestinal stromal tumors (GISTs) [8]. CT findings are nonspecific and many non-neoplastic conditions can present in a similar manner. CT imaging of peritoneal carcinomatosis includes multiple heterogeneously enhancing peritoneal and omental masses representing diffuse neoplastic infiltration. Peritoneal carcinomatosis can also present as large infiltrating masses throughout the omentum known as 'omental caking' [8]. Our patient presented with increasing abdominal pain and distension three weeks status post transanal excision of primary anorectal melanoma. CT scan of the abdomen and

pelvis demonstrated extensive peritoneal and omental nodularity with several large intraperitoneal masses, findings compatible with peritoneal carcinomatosis arising from anorectal melanoma. This case report demonstrates the high potential for metastatic disease in patients with primary anorectal melanoma.

### Treatment & Prognosis:

The mainstay treatment of anorectal melanoma is surgical resection. There is continual controversy over the most effective method of surgical treatment with either wide local excision (WLE) or abdominal perineal resection (APR) [1]. Due to the rarity of the disease, there is limited data and research comparing the outcomes of the different surgical techniques. A retrospective review shows a shift toward local excision as the surgical treatment for anorectal melanoma [9]. Local excision has benefits of a quicker recovery time and minimal effect on bowel function compared to APR [1, 9]. However, WLE has not proven to show a significant change in overall outcome compared to APR. One retrospective review demonstrated 75% of patients having recurrent disease irrespective of the extent of surgical excision [9]. Another larger retrospective study demonstrated a survival of 21 months for patients who underwent WLE and 17 months for patients who underwent APR [1, 10]. Given the lack of significant difference in overall outcome and survival, wide local excision seems to be favored, as it is a less invasive technique. Other adjuvant therapies including chemotherapy and radiation have a less clear role in the treatment of anorectal melanoma.

Anorectal melanoma has a poor prognosis with most patients having metastatic disease at the time of initial diagnosis. Patients with anorectal melanoma who have undergone treatment have a mean survival of 20 months. [11]. Anorectal melanoma is highly aggressive with a 5-year survival rate of 6% to 22% [2].

### Differential Diagnosis:

There are many disease processes that can resemble peritoneal carcinomatosis. The main differential diagnoses to consider for peritoneal carcinomatosis include peritoneal lymphomatosis, peritoneal tuberculosis, pseudomyxoma peritonei, peritoneal sarcomatosis, and malignant peritoneal mesothelioma.

### Peritoneal Lymphomatosis

Peritoneal lymphomatosis represents the spread of aggressive lymphomas to the peritoneum. It is a rare manifestation of lymphoma and is most frequently seen with non-Hodgkin lymphomas. Peritoneal lymphomatosis often shows significant improvement with chemotherapy treatment [12]. CT findings of peritoneal lymphomatosis include bulky thickening of the omentum with peritoneal nodularity and a small amount of ascites. US findings include hypoechoic mesenteric infiltration. On MRI, the peritoneal nodules are characterized by hypointense signal on T1WI and intermediate signal on T2WI. FDG PET/CT findings include diffuse FDG uptake throughout the omental and mesenteric nodules. The

key distinguishing radiographic features are extensive lymphadenopathy and confluent mesenteric masses which encase the mesenteric vessels, creating the “sandwich” sign [13]. The masses are typically bulky and homogeneous with less vascularity compared to peritoneal carcinomatosis. Organomegaly is also a common manifestation seen in lymphoma cases, helping to make the diagnosis of peritoneal lymphomatosis.

#### Peritoneal Tuberculosis

Peritoneal tuberculosis comprises approximately 1-3% of tuberculosis cases and represents the sixth most common extra-pulmonary site. Tuberculosis involving the peritoneum is predominantly seen in patients less than 40 years old with an increased prevalence in females [14]. Clinical findings include abdominal pain, fever, gastrointestinal symptoms, and weight loss. Three types of peritoneal tuberculosis have been described: the ‘wet’ type, the ‘fixed fibrotic’ type, and the ‘plastic’ (‘dry’) type. The ‘wet’ type will show a large amount of abdominal ascites. The ‘fixed fibrotic’ pattern will demonstrate discrete omental masses that adhere to the adjacent bowel. The ‘plastic’ type appears as diffuse thickening of the omentum and mesentery with fibrous adhesions [13]. If there is significant ascites, radiographs can demonstrate medial displacement of the bowel. On US, the mesentery will appear echogenic and thickened with or without the presence of ascites. MRI findings include diffuse peritoneal thickening with low signal ascites on T1WI and high signal ascites on T2WI. FDG PET/CT images show diffuse increased FDG uptake of the peritoneal masses and nodules. The peritoneal thickening of tuberculosis will typically have a smoother and more organized appearance compared to that seen in peritoneal carcinomatosis. Calcified mesenteric macronodules are also a common finding associated with peritoneal tuberculosis.

#### Pseudomyxoma Peritonei

Pseudomyxoma peritonei is a rare entity of diffuse intraperitoneal mucinous implants secondary to a ruptured mucinous neoplasm. The estimated incidence is one to two per million per year [15]. Clinical findings include abdominal distention and discomfort. Imaging findings can be nonspecific and resemble a similar appearance to peritoneal carcinomatosis. However, there are several findings that are characteristic of pseudomyxoma peritonei. One key finding seen on CT imaging is scalloping of the visceral surfaces, commonly the liver surface. Other important findings on CT include loculated intraperitoneal fluid collections and scattered curvilinear calcifications [13, 15]. On US, the peritoneal masses will appear echogenic and scalloping of the visceral surfaces may be present. On MR, the loculated collections are characterized by low signal on T1WI and high signal on T2WI. FDG PET/CT findings include diffuse increased FDG uptake of the peritoneal masses. There will often be a soft tissue mass of the appendix representing the primary mucinous tumor.

#### Peritoneal Sarcomatosis

Peritoneal sarcomatosis is a rare manifestation of peritoneal metastasis seen with sarcoma neoplasms. The most common tumors associated with this peritoneal entity include gastrointestinal stromal tumors (GISTs), liposarcomas, and leiomyosarcomas. CT findings are similar to that of peritoneal carcinomatosis with disseminated soft tissue implants seen throughout the peritoneum and omentum. The peritoneal soft tissue nodules seen with sarcomatosis typically demonstrate a more spherical appearance with increased vascularity. There is also a minimal amount of abdominal ascites seen with sarcomatosis, helping to differentiate from peritoneal carcinomatosis [16, 17]. On US, it is often difficult to appreciate the peritoneal nodules due to a minimal amount of ascites. There are variable signal characteristics of the peritoneal nodules on both T1WI and T2WI. There will be increased FDG uptake of the peritoneal nodules and masses on FDG PET/CT imaging. Peritoneal involvement seen with GISTs will frequently show heterogeneously enhancing, large necrotic masses [16].

#### Malignant Peritoneal Mesothelioma

Malignant peritoneal mesothelioma is an aggressive disease with a rapid and fatal course. The most significant risk factor is long-term exposure to asbestosis. There is a higher prevalence of disease in male patients and commonly presents in the fifth and sixth decades. There are several patterns of peritoneal involvement seen on CT imaging. It can present as a dry type, which is characterized by large intraperitoneal masses, diffuse peritoneal nodularity, and minimal ascites. Another pattern termed the wet type will appear as thickening of the peritoneum, omental masses, and ascites. Peritoneal mesothelioma can also present as a combination of both the dry and wet patterns [18]. Abdominal radiographs may show medial displacement of bowel loops if there is a significant amount of ascites present. US findings are variable with peritoneal masses appearing as either hypoechoic or echogenic. On MRI, the peritoneal nodules and masses are characterized by low to intermediate signal on T1WI and high signal on T2WI. FDG PET/CT imaging will demonstrate mild increased FDG uptake of the intraperitoneal nodules and masses. Peritoneal calcifications are uncommonly seen with mesothelioma, however, pleural based calcifications are frequently present. [19]

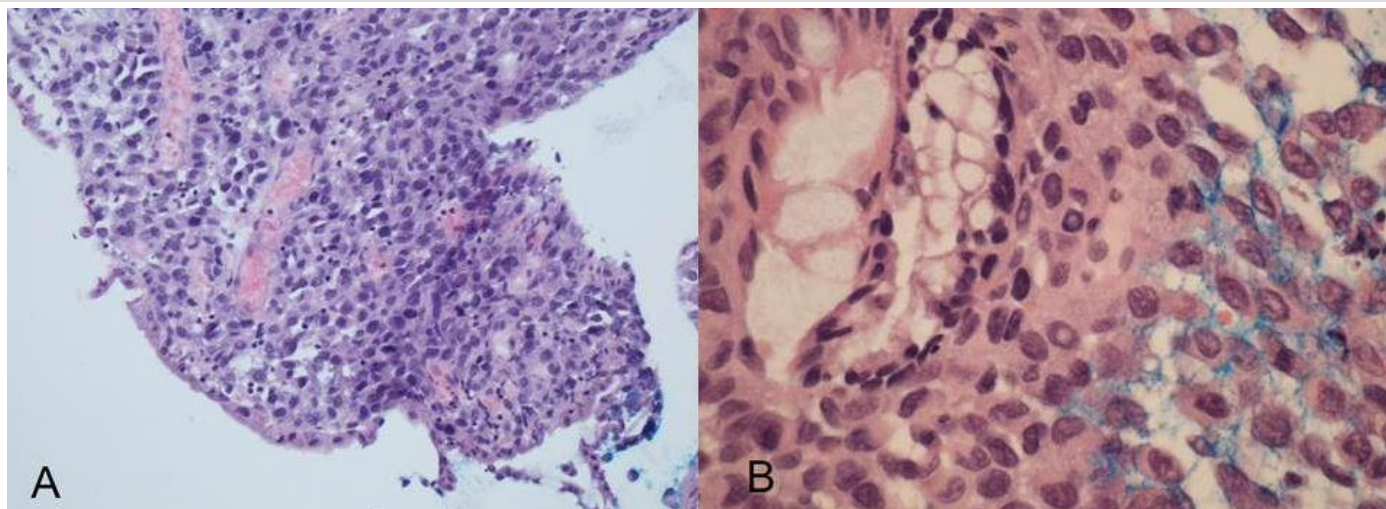
#### TEACHING POINT

Imaging is essential in evaluating metastatic disease in cases of primary anorectal melanoma with metastases commonly involving the lymphatic system, lungs, skin, and brain. Anorectal melanoma is an aggressive malignancy that can lead to peritoneal carcinomatosis with CT findings of multiple heterogeneously enhancing peritoneal and omental masses, representing diffuse neoplastic infiltration.

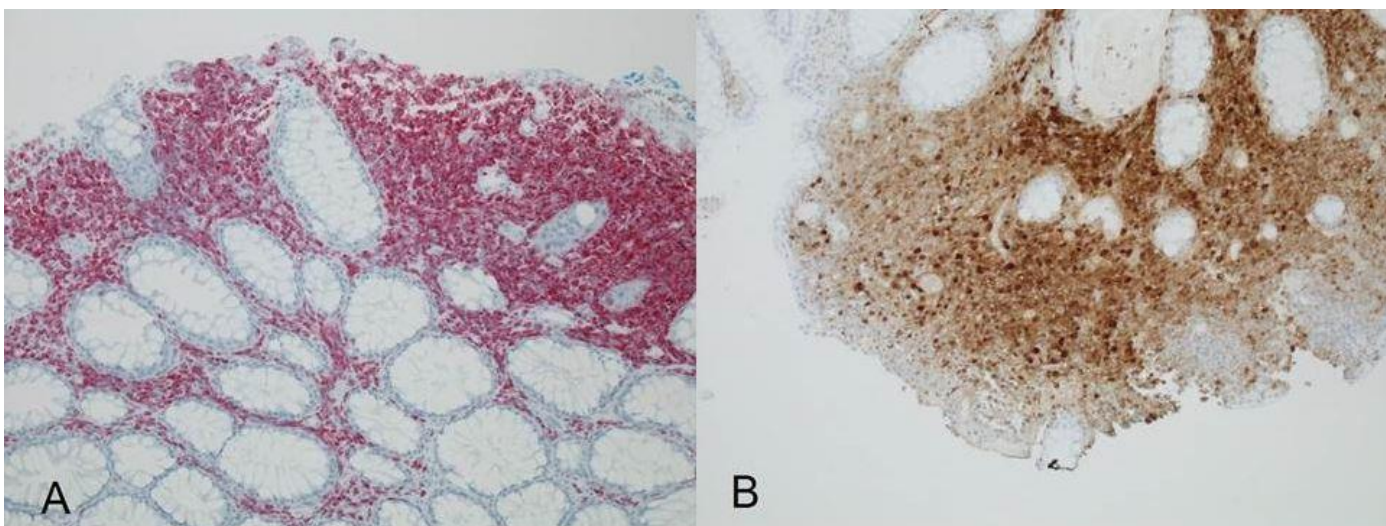
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FIGURES



**Figure 1:** 75-year-old female with peritoneal carcinomatosis from primary anorectal melanoma.  
Findings: A- Low magnification haematoxylin and eosin stained histologic slide demonstrates malignant cells within the lamina propria  
B- High magnification haematoxylin and eosin stained histologic slide shows malignant cells with nuclear inclusions.



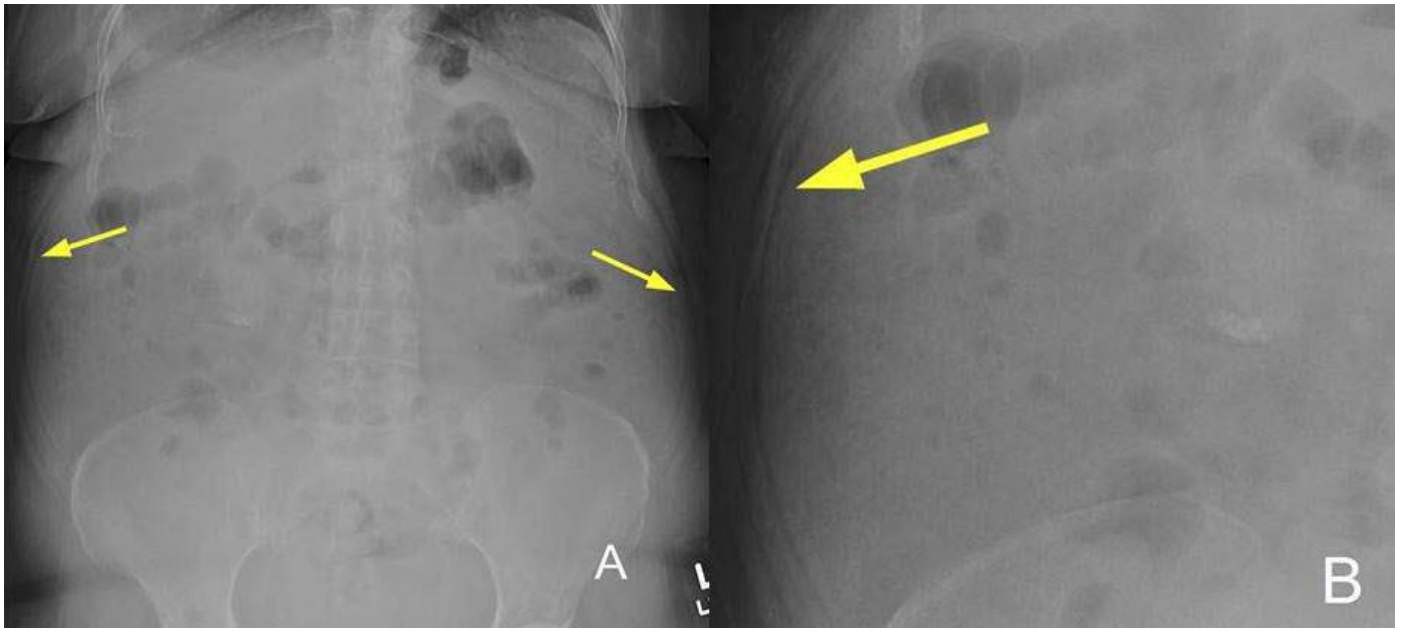
**Figure 2:** 75-year-old female with peritoneal carcinomatosis from primary anorectal melanoma.  
Findings: Figure 7A- Positive immunohistochemical stain with Melan-A; Figure 7B- Positive immunohistochemical stain with S-100



**Figure 3 (left):** 75-year-old female with peritoneal carcinomatosis from primary anorectal melanoma.  
Findings: Axial non-contrast CT images of the abdomen and pelvis demonstrate no evidence of metastatic disease prior to the third transanal excision.  
Technique: Axial CT; mA 120; kV 120; 5.0 mm slice thickness

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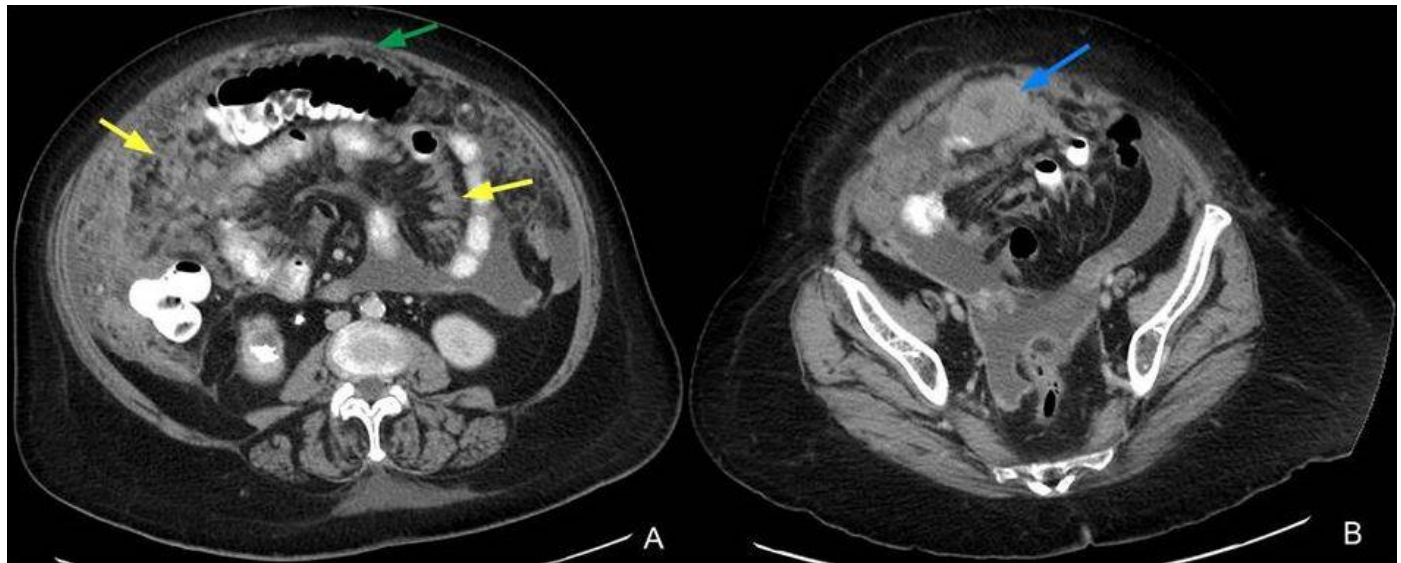
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**Figure 4:** 75-year-old female with peritoneal carcinomatosis from primary anorectal melanoma.

Findings: Supine images of the abdomen shows diffuse increased density and bulging of the flanks with lateral displacement of the properitoneal fat stripe (yellow arrows), representing underlying ascites.

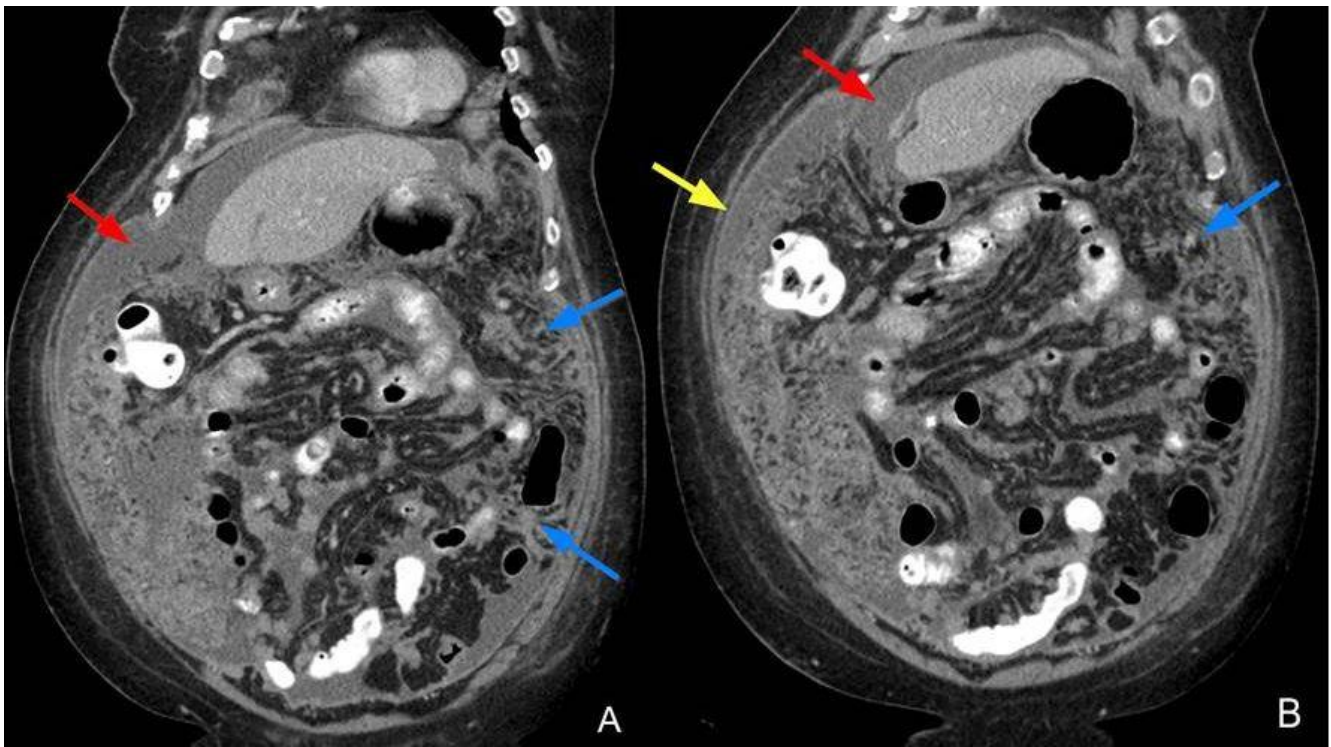
Technique: Supine radiograph of the abdomen; mA 500; kV 80



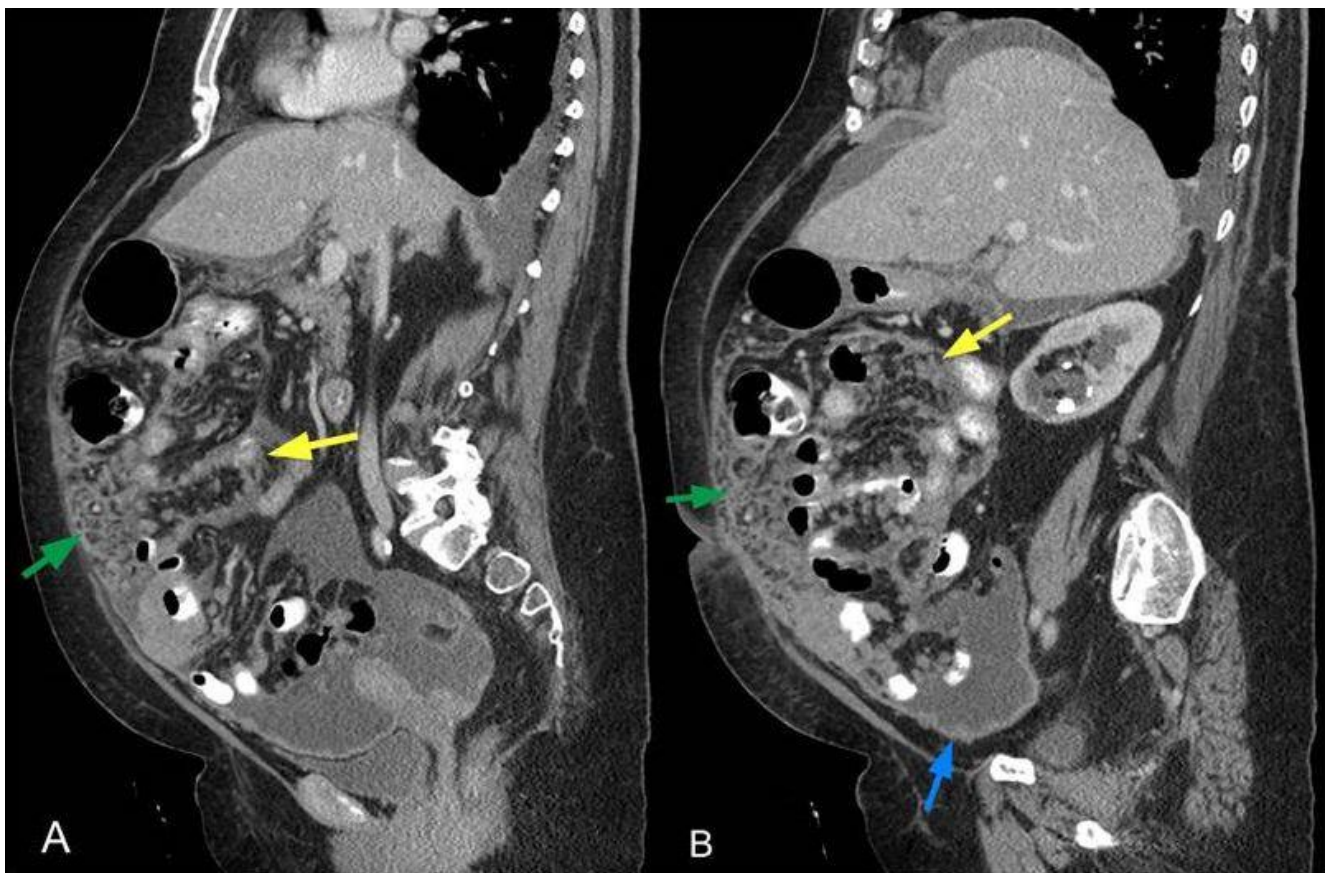
**Figure 5:** 75-year-old female with peritoneal carcinomatosis from primary anorectal melanoma.

Findings: Axial contrast-enhanced CT images of the abdomen and pelvis demonstrate diffuse peritoneal thickening and nodularity (yellow arrows) with a large intraperitoneal mass seen in the lower abdomen (blue arrow). There is omental infiltration (green arrow) with diffuse ascites throughout the abdomen and pelvis.

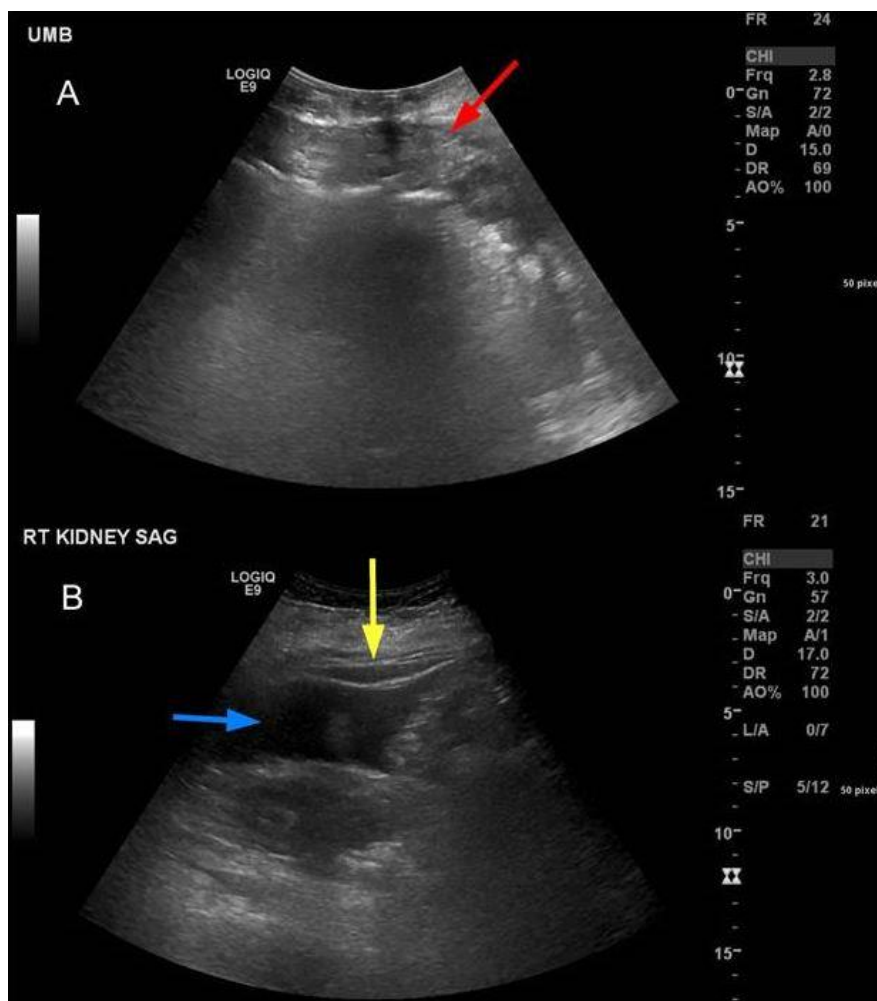
Technique: Axial CT; venous phase; mA 120; kV 120; 5.0 mm slice thickness; 100ml Omnipaque 300



**Figure 6:** 75-year-old female with peritoneal carcinomatosis from primary anorectal melanoma. Findings: Coronal contrast-enhanced CT images of the abdomen and pelvis show peritoneal thickening (yellow arrow) with diffuse omental infiltration (blue arrows) and ascites (red arrows), compatible with peritoneal carcinomatosis. Technique: Coronal CT; venous phase; mA 120; kV 120; 5.0 mm slice thickness; 100ml Omnipaque 300.



**Figure 7:** 75-year-old female with peritoneal carcinomatosis from primary anorectal melanoma. Findings: Sagittal contrast-enhanced CT of the abdomen and pelvis shows abdominal distention with diffuse infiltration of the omentum (green arrows) with multiple intraperitoneal nodules (yellow arrows) and peritoneal thickening (blue arrow). Technique: Sagittal CT; venous phase; mA 120; kV 120; 5.0 mm slice thickness; 100ml Omnipaque 300.



**Figure 8:** 75-year-old female with peritoneal carcinomatosis from primary anorectal melanoma. Findings: Ultrasound images of the umbilicus and right kidney demonstrate irregular hypoechoic nodules (red arrow), hypoechoic thickening of the peritoneum (yellow arrow), and anechoic ascites (blue arrow). Technique: Figure 8A and 8B: B-mode sonograms of the umbilicus and right kidney. Ultrasonography was performed utilizing a 3.5 MHz curvilinear transducer.

<b>Etiology</b>	Unclear
<b>Incidence</b>	Approximately 2.7 cases per 10 million people per year in the United States; Reported incidence of 1 to 3% of all anal tumors
<b>Gender Ratio</b>	Female predilection
<b>Age Predilection</b>	Onset of disease most common in the 6 <sup>th</sup> decade of life
<b>Risk Factors</b>	Unknown
<b>Treatment</b>	Surgical excision is the mainstay treatment with either wide local excision or abdominal perineal resection
<b>Prognosis</b>	Highly lethal disease with an overall poor prognosis; 5-year survival rate of 6% to 22%
<b>Image Findings</b>	Anorectal melanoma has a high potential for metastasis. We describe a case with metastasis to the peritoneum. <u>Peritoneal Carcinomatosis:</u> <ul style="list-style-type: none"> <li>• CT: Multiple heterogeneously enhancing peritoneal and omental masses or large infiltrating omental masses referred to as ‘omental caking’ with diffuse ascites</li> <li>• MRI: On T1WI low signal ascites with intermediate signal omental nodularity. On T2WI high signal ascites with intermediate signal peritoneal and omental nodularity. DWI shows variable diffusion restriction</li> <li>• US: Hypoechoic thickening of the omentum and peritoneum, complex ascites containing internal echoes and septations</li> <li>• FDG PET/CT: diffuse mild increased FDG uptake throughout the peritoneum</li> </ul>

**Table 1:** Summary table for Anorectal Melanoma.



Differential Diagnosis	X-Ray	US	CT	MRI	FDG PET/CT
<b>Peritoneal Carcinomatosis</b>	Lateral displacement of the properitoneal fat stripe if significant ascites	Hypoechoic thickening of the omentum and peritoneum, complex ascites containing internal echoes and septations	Multiple heterogeneously enhancing peritoneal and omental masses; Large infiltrating omental masses referred to as 'omental caking'; diffuse ascites	T1WI: Low signal ascites with intermediate signal omental nodularity T2WI: high signal ascites with intermediate signal of the peritoneal/omental nodularity	Diffuse mild increased FDG uptake throughout the peritoneum
<b>Peritoneal Lymphomatosis</b>	N/A	Hypoechoic mesenteric infiltration	Bulky thickening of the omentum with peritoneal nodularity; Small ascites; Organomegaly	T1WI: typically hypointense signal of peritoneal nodules T2WI: intermediate signal of peritoneal nodules	Diffuse increased FDG uptake throughout omental and mesenteric nodules
<b>Peritoneal Tuberculosis</b>	Medial displacement of bowel if there is significant ascites	Echogenic thickened mesentery with or without ascites	Wet type – large amount of ascites; Fixed fibrotic type – discrete omental masses adhering to the adjacent bowel; Plastic type – diffuse thickening of the omentum and mesentery with fibrous adhesions	T1WI: low signal ascites; diffuse peritoneal thickening  T2WI: high signal ascites; diffuse peritoneal thickening	Diffuse increased FDG uptake of peritoneal masses/nodules
<b>Pseudomyxoma Peritonei</b>	Medial displacement of bowel loops due to ascites; may demonstrate curvilinear calcifications	Echogenic peritoneal masses; may see scalloping of visceral surfaces	Scalloping of the visceral surfaces, commonly involving the liver; loculated intraperitoneal fluid and curvilinear calcifications	TIWI: Low signal of loculated collections T2WI: High signal of loculated collections	Increased FDG uptake of peritoneal masses
<b>Peritoneal Sarcomatosis</b>	N/A	Often difficult to appreciate peritoneal nodules due to minimal ascites	Hypervascular and spherical appearing soft tissues peritoneal nodules; minimal ascites	Variable signal characteristics on T1WI and T2WI	Increased FDG uptake of peritoneal masses/nodules
<b>Malignant Peritoneal Mesothelioma</b>	Medial displacement of bowel loops if significant amount of ascites present	Variable – hypoechoic or echogenic masses with ascites	Dry type – large intraperitoneal masses with diffuse peritoneal nodularity and minimal ascites; Wet type – peritoneal thickening, omental masses and ascites Mixed – combination of dry and wet findings	T1WI: Low to intermediate signal of peritoneal masses/nodules T2WI: High signal of peritoneal masses/nodules	Mild increased FDG uptake of intraperitoneal masses and nodules

**Table 2:** Differential diagnoses table for Peritoneal Carcinomatosis.

#### ABBREVIATIONS

APR = Abdominal perineal resection  
CT = Computed Tomography  
DWI = Diffusion-Weighted Imaging  
FDG PET/CT = Fluorodeoxyglucose-18 Positron Emission Tomography  
GISTs = Gastrointestinal Stromal Tumors  
kV = Kilovoltage  
mA = Milliampere  
MHz = Megahertz  
mm = Millimeters  
MRI = Magnetic Resonance Imaging  
N/A = Not Applicable  
T1WI = T1-Weighted Imaging  
T2WI = T2-Weighted Imaging  
US = Ultrasonography  
WLE = Wide local excision

#### KEYWORDS

Anorectal melanoma; anal melanoma; peritoneal carcinomatosis; ascites; peritoneum; omentum; Computed Tomography; Magnetic Resonance Imaging

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