

AMSER Case of the Month

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35-year-old male with a one-month history of
progressive abdominal pain

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Patient Presentation

- **HPI:** A 35-year-old male presents to the ED complaining of a one-month history of progressive aching, poorly localized abdominal pain, with associated weight loss, diarrhea, and facial flushing. He reports severe pain, rating it as a 7/10 in severity with mild improvement with hydrocodone. He cannot identify any specific triggers. He endorses easy bruisability, dyspnea on exertion, and denies any other associated symptoms
- **Medications:** Hydrocodone-acetaminophen
- **Past Medical History:** Unremarkable
- **Social History:** No alcohol or tobacco use. No work-related hazardous exposures

Objective Data

- **Vitals**
 - Within normal limits
- **Pertinent Exam Findings:**
 - Abdomen non-distended, diffusely tender to palpation, slightly worse in epigastric area, non-peritonitic.
- **CBC**
 - Unremarkable
- **CMP**
 - Unremarkable

What Imaging Should We Order?

Select the applicable ACR Appropriateness Criteria

Variant 4: Acute nonlocalized abdominal pain. Not otherwise specified. Initial imaging.

Procedure	Appropriateness Category	Relative Radiation Level
CT abdomen and pelvis with IV contrast	Usually Appropriate	☼☼☼
CT abdomen and pelvis without IV contrast	Usually Appropriate	☼☼☼
MRI abdomen and pelvis without and with IV contrast	Usually Appropriate	○
US abdomen	May Be Appropriate	○
MRI abdomen and pelvis without IV contrast	May Be Appropriate	○
CT abdomen and pelvis without and with IV contrast	May Be Appropriate	☼☼☼☼
Radiography abdomen	May Be Appropriate	☼☼
FDG-PET/CT skull base to mid-thigh	Usually Not Appropriate	☼☼☼☼
WBC scan abdomen and pelvis	Usually Not Appropriate	☼☼☼☼
Nuclear medicine scan gallbladder	Usually Not Appropriate	☼☼
Fluoroscopy upper GI series with small bowel follow-through	Usually Not Appropriate	☼☼☼
Fluoroscopy contrast enema	Usually Not Appropriate	☼☼☼

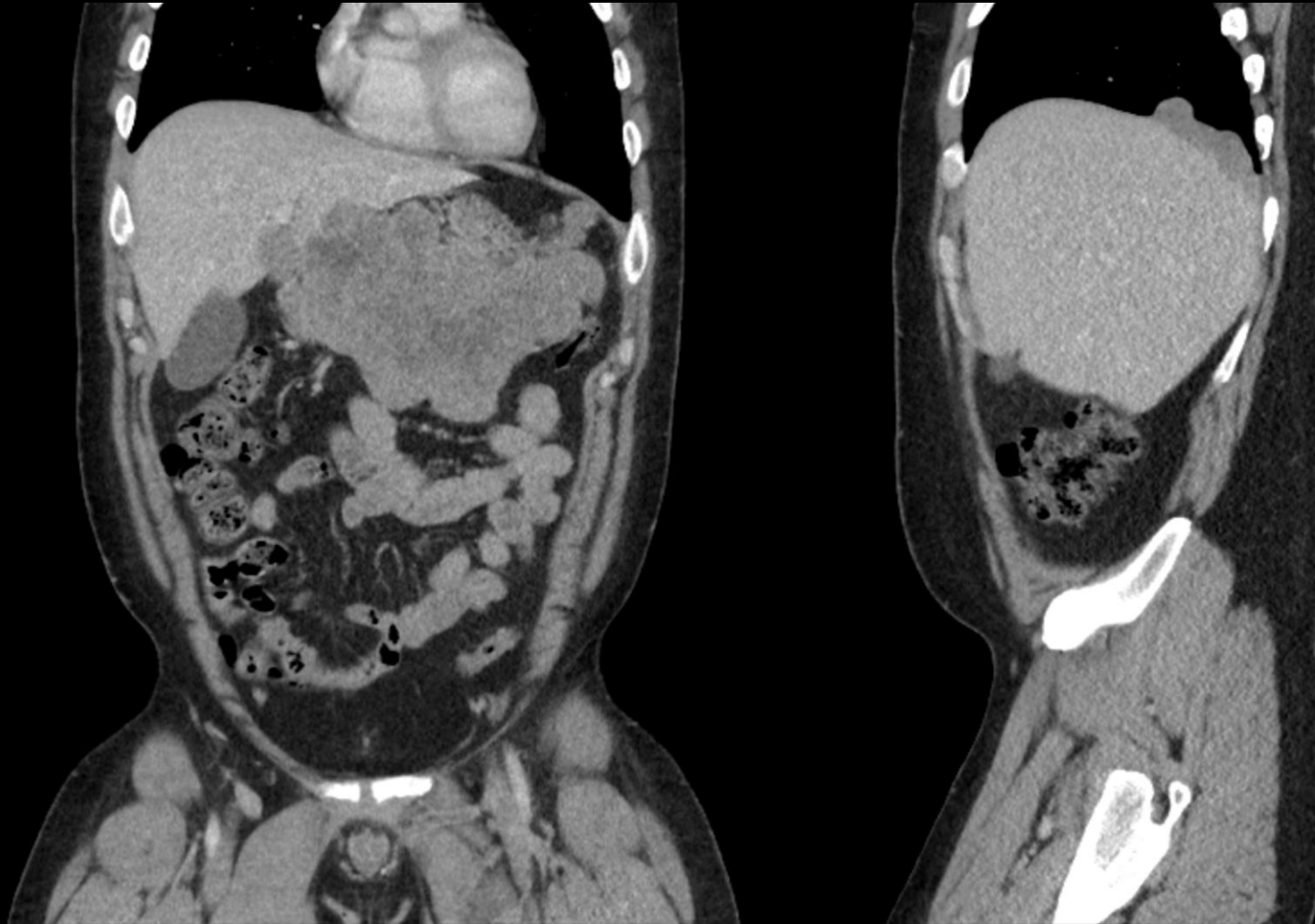
This imaging modality was ordered by the ER physician



Findings (unlabeled)

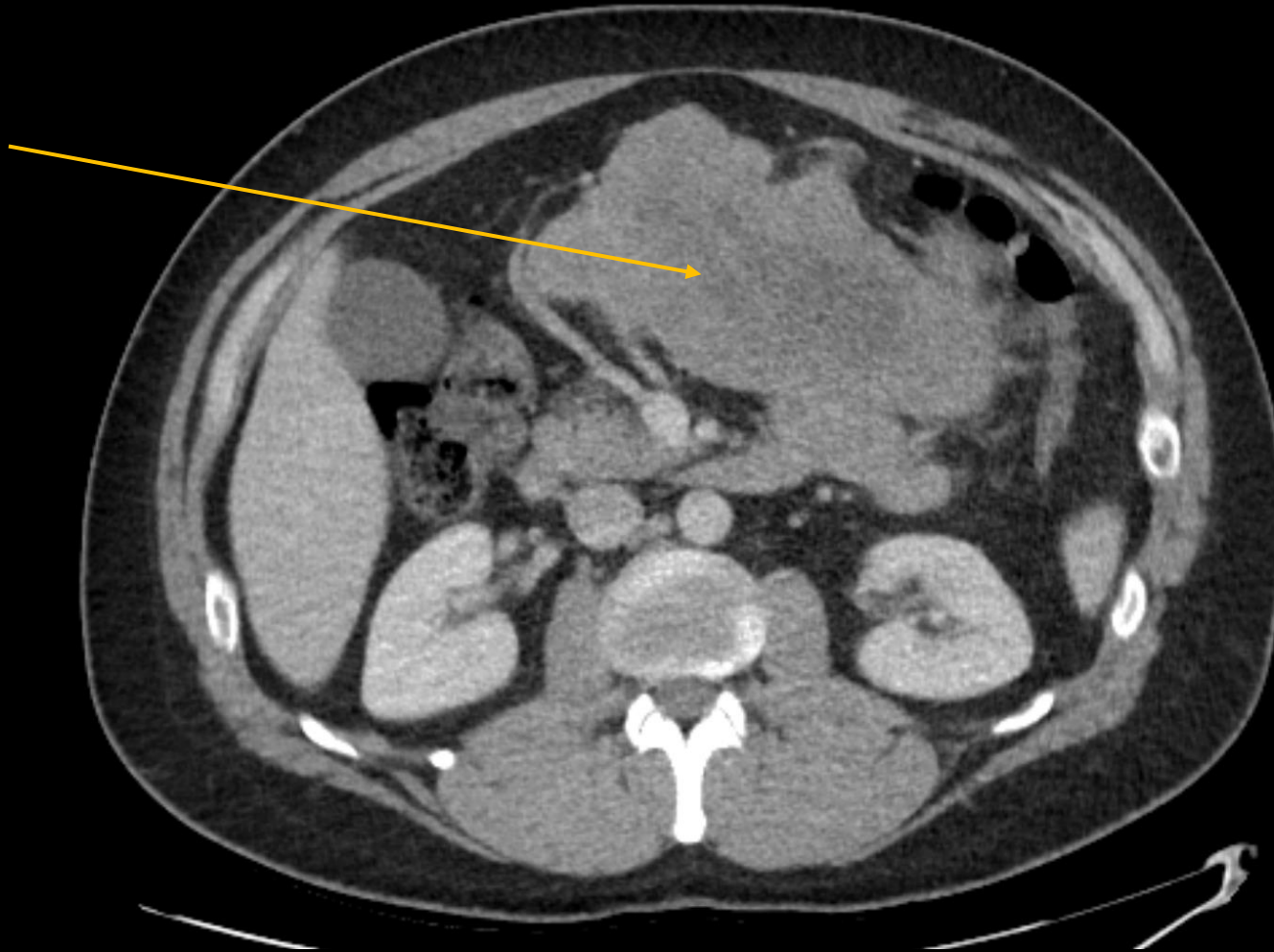


Findings (unlabeled)



Findings (labeled)

- There is an 8.5 x 16.8 x 12.2 cm peritoneal soft tissue mass, with minimal heterogeneity and central hypoattenuation
- Minimal adjacent fat stranding
- No ascites or focal fluid collections



Axial image, CT with contrast

Findings (labeled)

- Soft tissue mass appears to be a well-defined conglomerate of rounded peritoneal masses



Coronal image, CT with contrast

- Lobulated pleural-based soft tissue mass along the right posterior hemidiaphragm



Sagittal image, CT with contrast

Differential Diagnosis

Most Common

Less Common

Least Common

Metastatic Disease

- Lymphoma
- Peritoneal carcinomatosis

Other Secondary Peritoneal Neoplasms or Tumor-like Lesions

- Mesenteric desmoid
- Mesenchymal tumors
 - GIST

Primary Peritoneal Neoplasms

- Mesothelial tumors
 - Malignant peritoneal mesothelioma
- Epithelial tumors
- Uncertain origin
 - Desmoplastic small round cell tumor

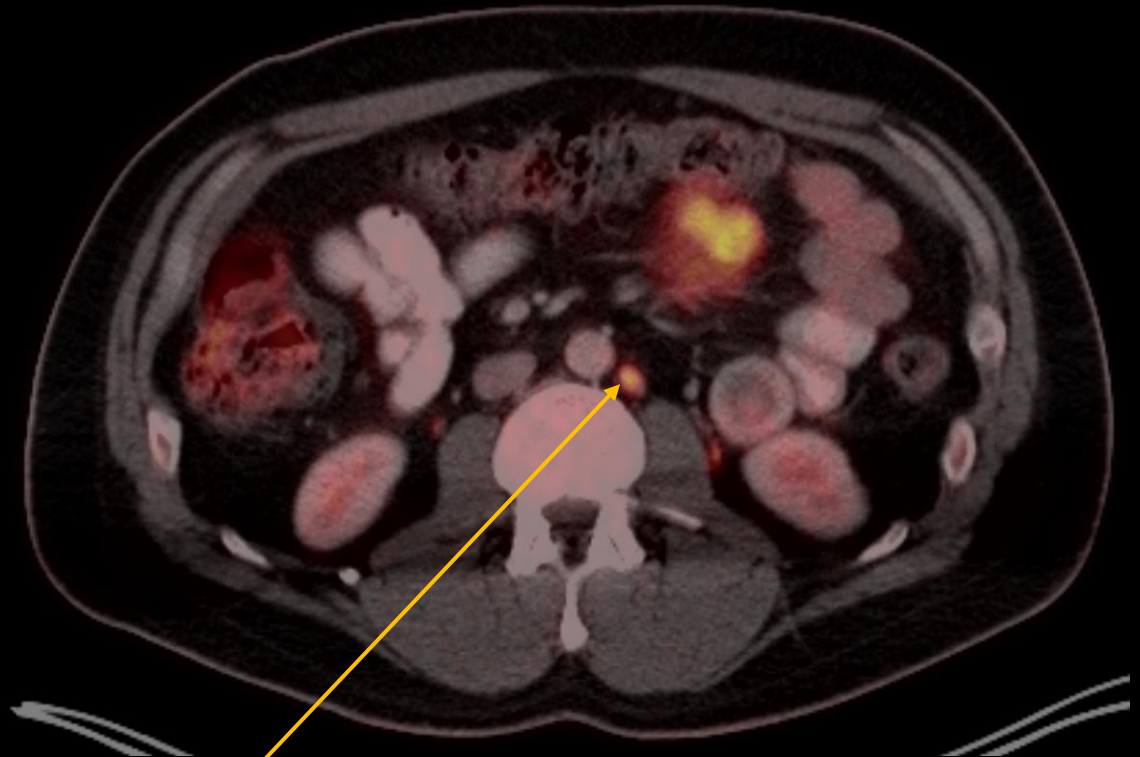
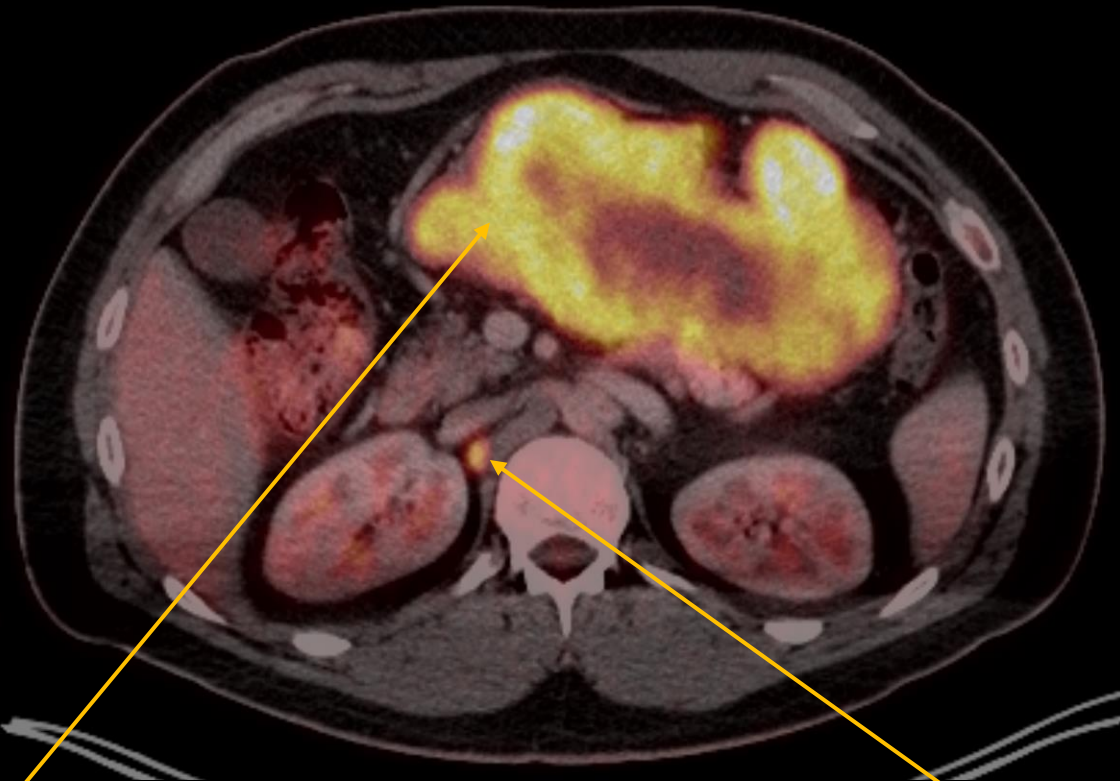
Biopsy and Pathological Analysis

- Histological analysis
 - High-grade neoplasm with small round blue cell morphology, numerous mitoses, and extensive necrosis
- Immunophenotypic staining
 - Positive: pankeratin, desmin, CD99, and P16
 - Negative: S100, WT1, chromogranin, synaptophysin
- FISH
 - Positive EWSR1 rearrangement
- Sarcoma NGS Fusion Panel
 - EWSR1-WT1 fusion detected

Final Dx:

Desmoplastic Small Round Cell Tumor

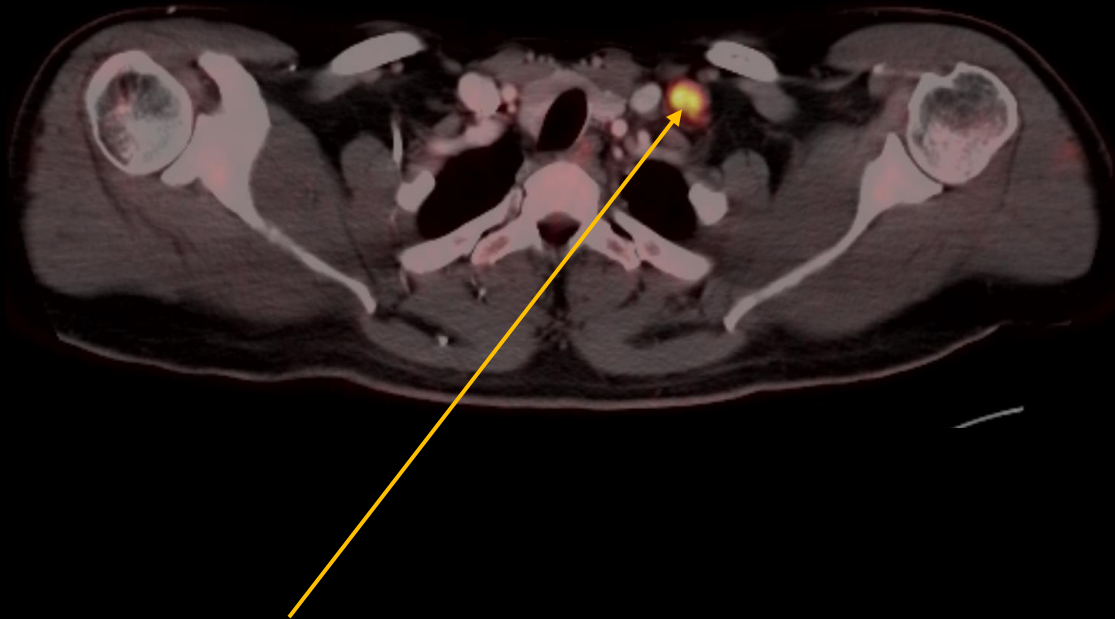
Follow-up Imaging (PET/CT Axial Images)



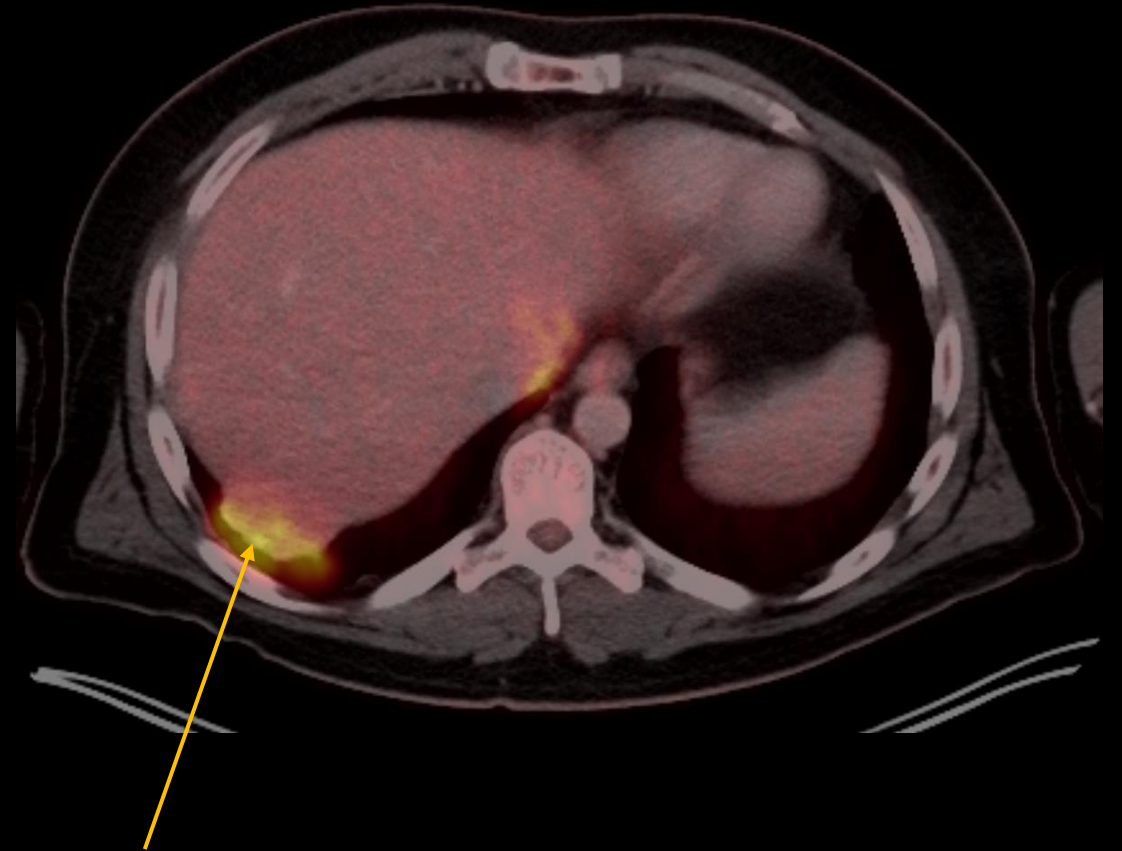
- Heterogenous soft tissue mass with a rim of metabolic activity and central necrosis

- Scattered enlarged FDG-avid retroperitoneal lymph nodes

Follow-up Imaging (PET/CT Axial Images)

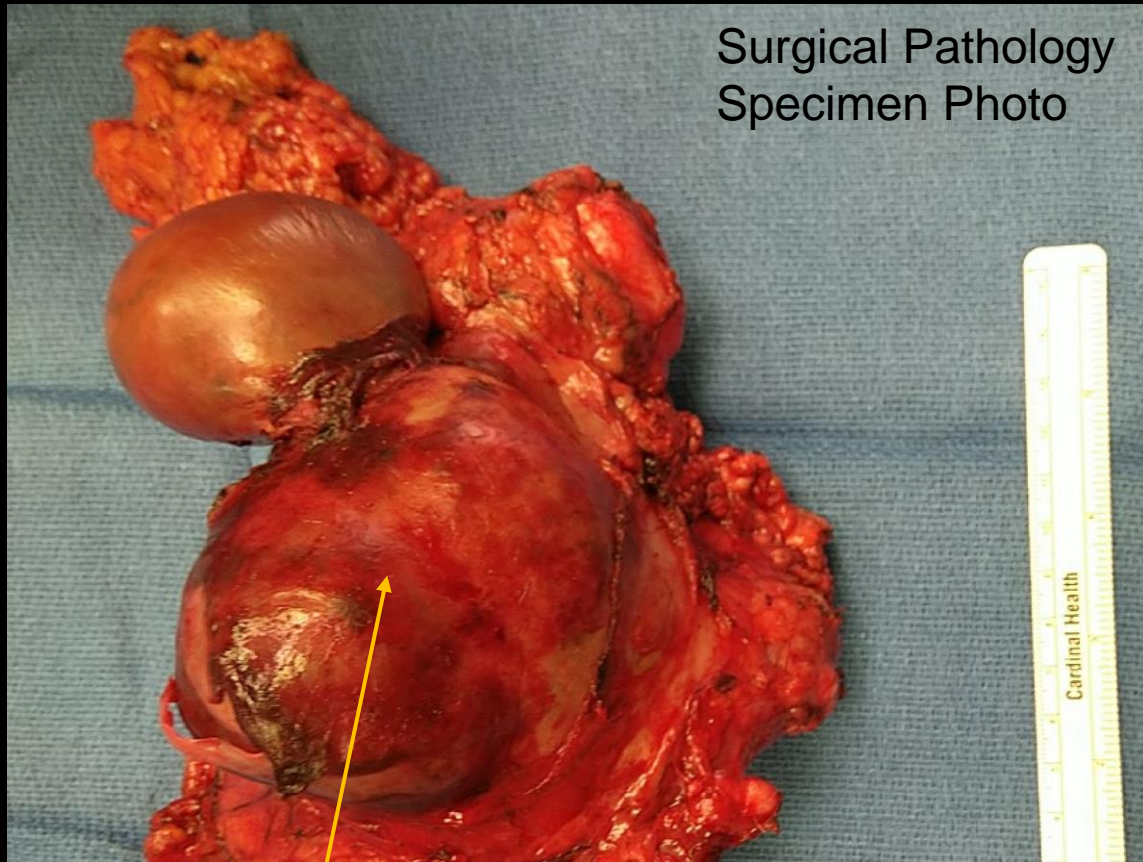


- Hypermetabolic left supraclavicular node



- Lobulated FDG-avid soft tissue mass arising from right posterior hemidiaphragm

Surgical Resection and Treatment



- s/p partial gastrectomy, omentectomy, right hemidiaphragm resection, and HIPEC with cisplatin

- Multiple residual enlarged retroperitoneal nodes s/p surgical resection and polychemotherapy

Case Discussion

- **Epidemiology**

- Desmoplastic small round cell tumor is a rare neoplasm, with an incidence of ~0.3 cases/million
- Most commonly occurs in the peritoneal cavity of adolescent and young adult males
- Mean age at diagnosis is 19 years

- **Clinical Presentation**

- Vague and non-specific. Typically presents with crampy non-localized abdominal pain, diarrhea, weight loss, and abdominal distention

- **Pathophysiology**

- Unknown histogenesis and tissue of origin. Related to the Ewing sarcoma family of tumors
- Caused by a de-novo translocation of t(1;22)(p13;q12), that leads to the fusion of the Ewing sarcoma RNA binding protein 1 (*EWSR1*) and Wilm's tumor suppressor (*WT1*) genes

Case Discussion

- **Imaging Features**

- Solitary bulky peritoneal mass, often >10 cm may be the only imaging finding seen on initial presentation
 - Less commonly presents with infiltrative appearance and diffuse peritoneal thickening ± malignant ascites
- Heterogenous appearance on CT, typically with central hypoattenuation due to intra-tumoral necrosis or hemorrhage ± scattered punctate calcifications
- T1 heterogenous hypointensity and T2 heterogenous hyperintensity on MR imaging

- **Diagnosis**

- Requires biopsy and histopathologic analysis
- Histologic features: sharply demarcated nests of small round cells embedded in a hypervascular desmoplastic stroma
 - Immunohistochemistry: Typically positive for desmin, EMA, and cytokeratin
- RNA sequencing/fusion panel: Diagnostic confirmation with evidence of EWSR1-WT1 fusion

- **Treatment**

- Surgical resection + radiation therapy and polychemotherapy
- Prognosis is poor, even with treatment
 - 3-year survival rate less than 30%

References:

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2. Levy AD, Arnáiz J, Shaw JC, Sobin LH. Primary Peritoneal Tumors: Imaging Features with Pathologic Correlation. *RadioGraphics* 2008;28(2):583-607. doi: 10.1148/rg.282075175.
3. Levy AD, Manning MA, Miettinen MM. Soft-Tissue Sarcomas of the Abdomen and Pelvis: Radiologic-Pathologic Features, Part 2—Uncommon Sarcomas. *RadioGraphics*. *RadioGraphics*; 2017;37:797–812<https://dx.doi.org/10.1148/rg.2017160201>.
4. Pickhardt PJ, Bhalla S. Primary Neoplasms of Peritoneal and Sub-peritoneal Origin: CT Findings. *RadioGraphics* 2005;25(4):983-995. doi: 10.1148/rg.254045140
5. Rana V, Sharma S, Kamala R, et al.. Round cell tumors: Classification and immunohistochemistry. *Indian Journal of Medical and Paediatric Oncology*. *Indian Journal of Medical and Paediatric Oncology*; 2017;38:349.
6. Sheth S, Horton KM, Garland MR, Fishman EK. Mesenteric Neoplasms: CT Appearances of Primary and Secondary Tumors and Differential Diagnosis. *RadioGraphics*. *RadioGraphics*; 2003;23:457–473<https://dx.doi.org/10.1148/rg.232025081>.