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

<table>
<thead>
<tr>
<th>Procedure</th>
<th>Appropriateness Category</th>
<th>Relative Radiation Level</th>
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<tbody>
<tr>
<td>CT abdomen and pelvis with IV contrast</td>
<td>Usually Appropriate</td>
<td>🍀 العليا</td>
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<tr>
<td>CT abdomen and pelvis without IV contrast</td>
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<td>MRI abdomen and pelvis without and with IV contrast</td>
<td>Usually Appropriate</td>
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<td>US abdomen</td>
<td>May Be Appropriate</td>
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<tr>
<td>Radiography abdomen</td>
<td>May Be Appropriate</td>
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<tr>
<td>FDG-PET/CT skull base to mid-thigh</td>
<td>Usually Not Appropriate</td>
<td>🍀 العليا</td>
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<td>WBC scan abdomen and pelvis</td>
<td>Usually Not Appropriate</td>
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<td>Nuclear medicine scan gallbladder</td>
<td>Usually Not Appropriate</td>
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<td>Fluoroscopy upper GI series with small bowel follow-through</td>
<td>Usually Not Appropriate</td>
<td>🍀 العليا</td>
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<td>Fluoroscopy contrast enema</td>
<td>Usually Not Appropriate</td>
<td>🍀 العليا</td>
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This imaging modality was ordered by the ER physician.
Findings (unlabeled)
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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.

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

Coronal image, CT with contrast  
Sagittal image, CT with contrast
Differential Diagnosis

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

CT with contrast, Coronal image
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:


