

Case Presentation: 55 Year Old Man with Chronic Abdominal Pain

Rose Emlein, MS3
Michael Baldwin, MD

Clinical History

- 55 year old male who presented to the ED with 4 hours of acute increasing of pressure-like left upper quadrant abdominal pain that radiates to his left flank and is associated with bloating.
- He reports that this pain has been present for the past 3 weeks, and has been getting progressively more severe. He has previously seen PCP regarding this pain, but no further workup was pursued. He has not taken anything for the pain. He presented this evening due to new onset subjective shortness of breath and worsening of pain with deep inspiration
- Positive for increasing fatigue, hot flashes, shortness of breath, increasing hypertension, long-standing erectile dysfunction, and weight loss of 12 pounds over the past months.
- Denies dizziness, syncope, chest pain, headache, dysuria, hematuria, testicular pain, nausea, vomiting, constipation, diarrhea, hematochezia, melena, rash, focal numbness or weakness, fall or injury, new or unusual activities, recent travel, recent surgery, cough, congestion, sore throat.

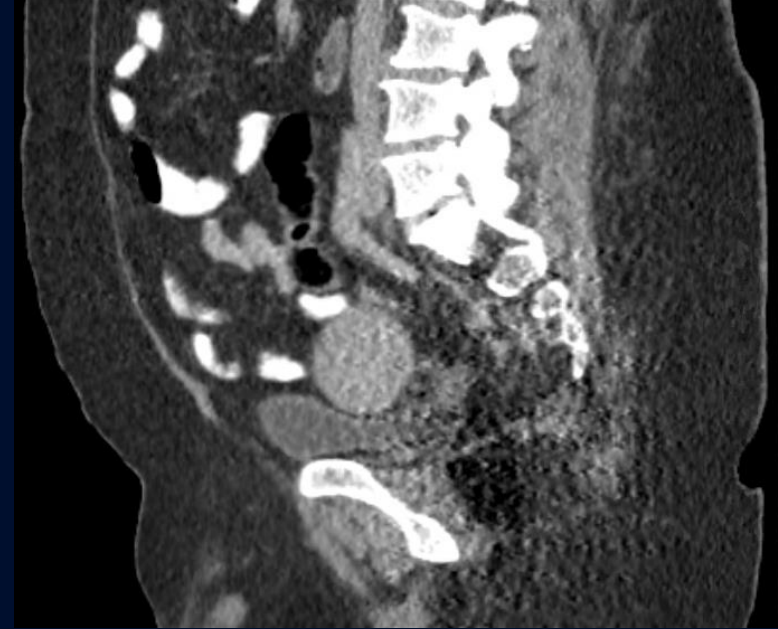
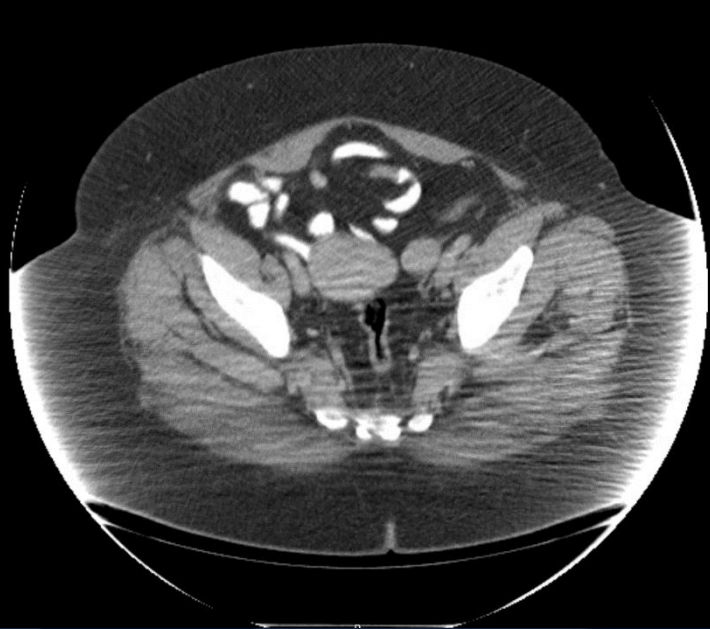
Additional History

- PMH of Class 3 obesity, hypertension, hyperlipidemia. Denies personal history of cancer
- The patient is a retired police officer who works as a security officer in marijuana facility. Lives with wife and has two grown children with no difficulty conceiving. Denies current or past tobacco use. Drinks alcohol 1-2 times per month, socially. Denies history of illicit drug use.
- Family history of prostate cancer in uncle and grandfather at advanced ages.
- Overdue for routine 10 year colonoscopy. Has never had colonoscopy before.

Initial Examination and Evaluation

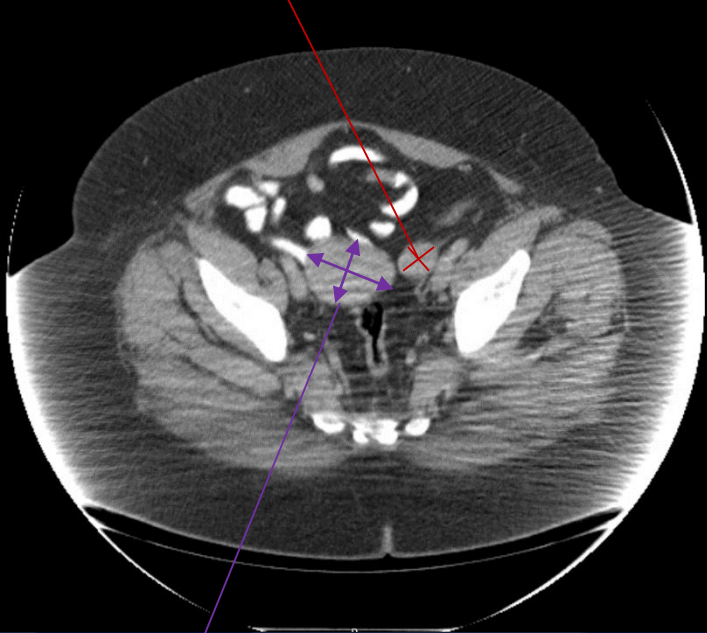
- **Physical Exam**
- BP 187/101 (BP Location: Right arm, Patient Position: Sitting) | Pulse 80 | Temp 97.9 °F (36.6 °C) (Temporal) | Resp 16 | SpO2 97%
- Patient is well appearing and in no acute distress. Abdomen is soft and non-distended with a normal contour. No tenderness to palpation, no rebound, no guarding. No CVA tenderness. Examination of all other systems was benign.
- Initial labs sent were unremarkable:
 - H/H: 14.5/44.7
 - WBC: 11
 - Platelets: 248
 - Chemistries: WNL
 - LFTs: WNL
 - Lipase 20
 - Troponin: negative
- The patient was sent for CT abdomen and pelvis with IV contrast

CT of the Abdomen and Pelvis with IV Contrast

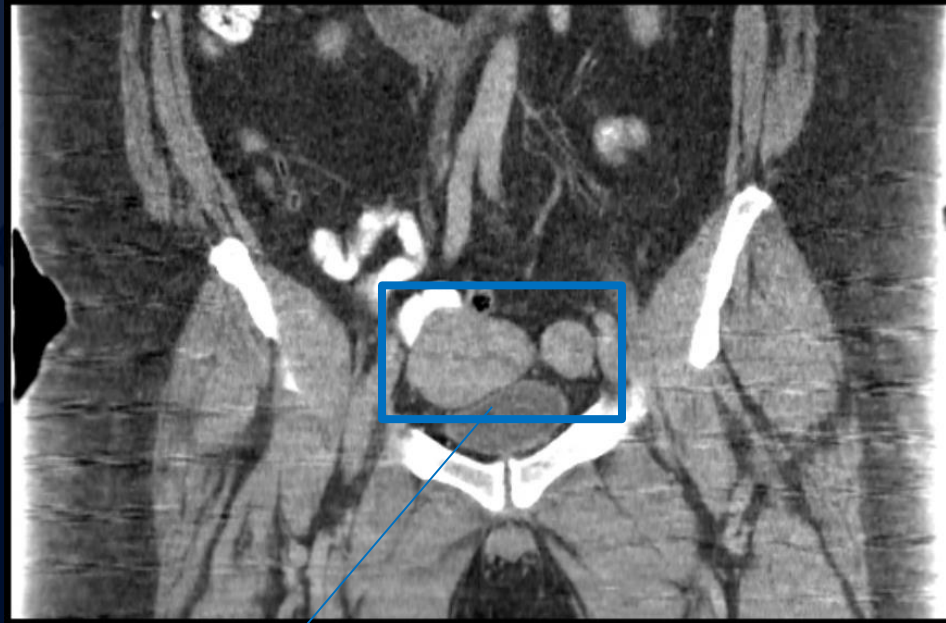


CT Abdomen and Pelvis with IV Contrast

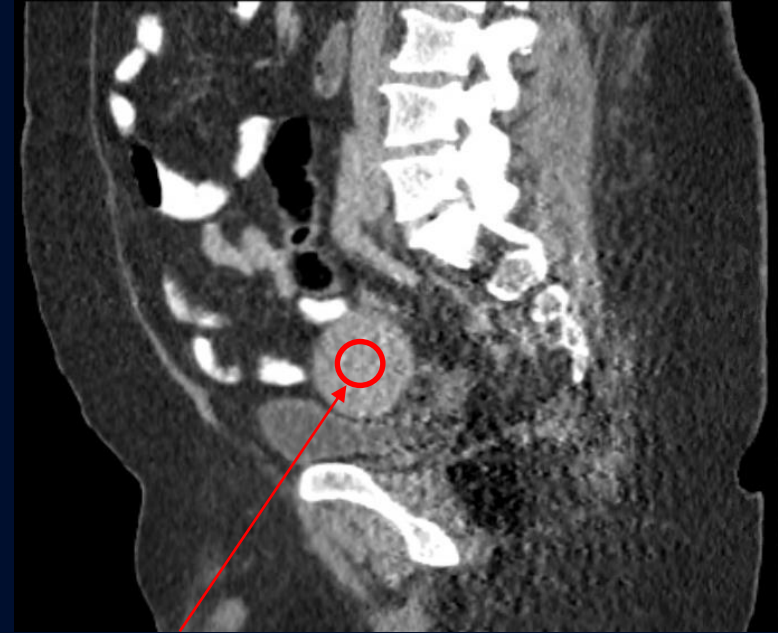
3.6 cm x 3.0 cm



8.3cm x 6.4cm



Two homogeneously enhancing oval lesions superior to the urinary bladder. No free fluid, free air, fat stranding



Mean: 61.22 Hounsfield Units

Differential Diagnosis for Presenting Symptoms

- At this time laboratory and CT evaluation unremarkable for causes of abdominal pain.
- Differential considered and ruled out: ischemic colitis, appendicitis, diverticulitis, testicular torsion, perforated viscous, and AAA, but history, exam, labs, and imaging do not support. Abdominal pain was thought to be most likely due to gastritis. Patient was discharged with PPI, Carafate, and PCP follow-up.
- 2 masses found on CT were considered to most likely be enlarged lymph nodes. He was referred to oncology for evaluation and further follow up. Initial differential from oncology included enlarged lymph node from prostate cancer or colon cancer, and lymphoma.
- Referred to IR for CT guided lymph node biopsy and GI for colonoscopy

Differential Diagnosis: Prostate Carcinoma

- Prostate cancer typical presentation:
 - Most patients are clinically asymptomatic at time of diagnosis
 - Uncommonly, patients may present with urinary symptoms, hematuria, and hematospermia
 - Bone pain is the most common presenting symptom in metastatic prostate cancer
- Our patient:
 - Family history of prostate cancer
 - Fatigue and weight loss over the past 3-4 months
 - Denies urinary symptoms
 - Denies bone pain

Differential Diagnosis: Colon Carcinoma

- Colon cancer typical presentation:
 - Change in bowel habits – most common symptom
 - Rectal bleeding
 - Iron deficiency anemia
 - Rectal or abdominal mass
- Our patient:
 - Denies constipation, diarrhea, or change in bowel habits
 - Denies hematochezia or melena
 - H/H within normal limits
 - 2 unknown masses on CT, however are distinct from colon
 - Recent weight loss and fatigue

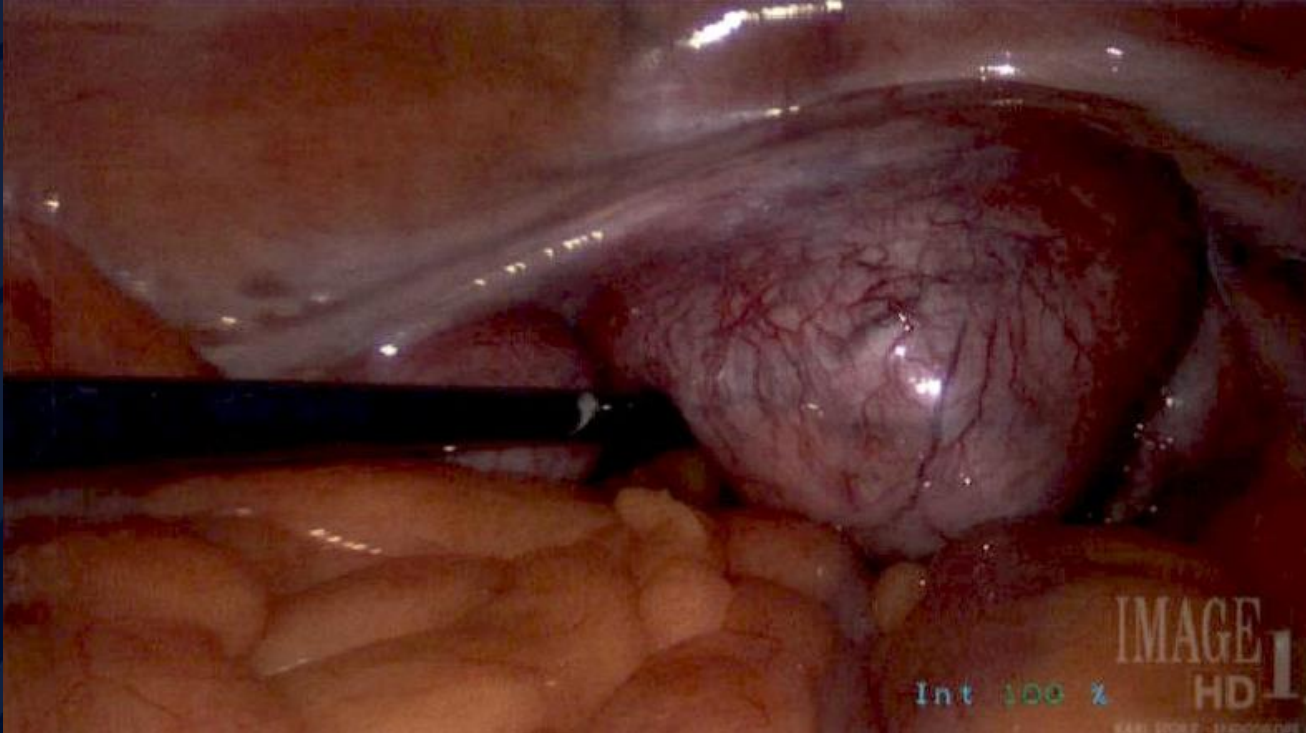
Differential Diagnosis: Lymphoma

- Lymphoma typical presentation:
 - Peripheral lymphadenopathy: typically firm and painless
 - Systemic B symptoms
 - Fever
 - Weight loss
 - Drenching night sweats
 - Fever of Unknown origin
 - Associated disorders
 - Autoimmune disease
 - Infections: HIV, HTLV-I, EBV, HBV, HCV, Borrelia burgdorferi, Chlamydia psittaci
 - IBD
- Our patient:
 - Weight loss without fevers or sweats
 - No disseminated lymphadenopathy
 - No history of above associated disorders

Biopsy Results and Next Steps

- Colonoscopy: significant for 1 benign serrated adenoma
- Pelvic mass biopsy:
 - Benign epithelial lining with prominent smooth muscle without atypia
 - Stromal cells stain positive for androgen receptor and estrogen receptor
 - Findings consistent with “Müllerian duct remnants.”
- Patient was presented at tumor board, with consensus for suspicion for vestigial female organs and that it was reasonable to remove the masses for determination of final pathology and evaluation of risk
- Further lab work significant for:
 - Elevated estrogen of 237.2
 - Low testosterone of 239
 - Normal FSH of 3.6 and LH of 4.8
 - Karyotype: 46 XY
 - Normal Aldosterone/Plasma renin level of 2.1

Surgery and Final Pathology



- Intra Operative Findings: Large pelvic mass adherent to the anterior abdominal wall immediately superior to the urinary bladder. There was a small satellite lesion just to the left side of this dominant mass as well. There were two vascular structures on either side of the mass that appeared to be consistent with the round ligaments.
- Final pathology diagnosis: “presumed uterus – benign müllerian type epithelium and smooth muscle with cystic degeneration. Negative for malignancy.”



Diagnosis?

Persistent Mullerian Duct Syndrome

Final Diagnosis

- Persistent Müllerian Duct Syndrome
 - Caused by mutations in AMH gene with resulting low serum levels of anti-müllerian hormone. Fetal müllerian ducts are sensitive to action of AMH in 7th or 8th week of gestation, and AMH leads to regression of müllerian ducts in XY fetus.
 - In PMDS müllerian ducts persist due to errors with AMH production (Type I) or AMH receptor (Type II)
- This patient's persistent elevated estrogen levels likely caused enlargement of vestigial uterus from persistent müllerian ducts.
- Origin of elevation in estrogen remains unknown and is currently being worked up. Differential includes:
 - Liver – no evidence of cirrhosis and no significant alcohol intake.
 - Adrenal mass – no evidence on CT scan
 - Testicular tumors – no palpable masses
 - Increased adiposity – BMI of 50. This is the most likely cause.

References

- Chan, Y., & Levitsky, L. L. (2021, January 31). *Causes of differences of sex development*. UpToDate. https://www.uptodate.com/contents/causes-of-differences-of-sex-development?search=persistent%20mullerian%20duct%20syndrome&source=search_result&selectedTitle=1~3&usage_type=default&display_rank=1
- Freedman, A. S., Friedberg, J. W., & Aster, J. C. (2020, July 8). *Clinical presentation and initial evaluation of non-Hodgkin lymphoma*. UpToDate. https://www.uptodate.com/contents/clinical-presentation-and-initial-evaluation-of-non-hodgkin-lymphoma?search=lymphoma%20presentation&source=search_result&selectedTitle=1~150&usage_type=default&display_rank=1#H8
- Kantoff, P. W. K., Taplin, M. T., & Smith, J. S. (2020, June 19). *Clinical presentation and diagnosis of prostate cancer*. UpToDate. https://www.uptodate.com/contents/clinical-presentation-and-diagnosis-of-prostate-cancer?search=prostate%20cancer%20clinical%20presentation&source=search_result&selectedTitle=1~150&usage_type=default&display_rank=1#H3590481039
- Macrae, F. A., & Bendell, J. (2021, January 22). *Clinical presentation, diagnosis, and staging of colorectal cancer*. UpToDate. https://www.uptodate.com/contents/clinical-presentation-diagnosis-and-staging-of-colorectal-cancer?search=colon%20cancer%20presentation&source=search_result&selectedTitle=1~150&usage_type=default&display_rank=1#H2