

Medical Image of the Week: Lynch Syndrome

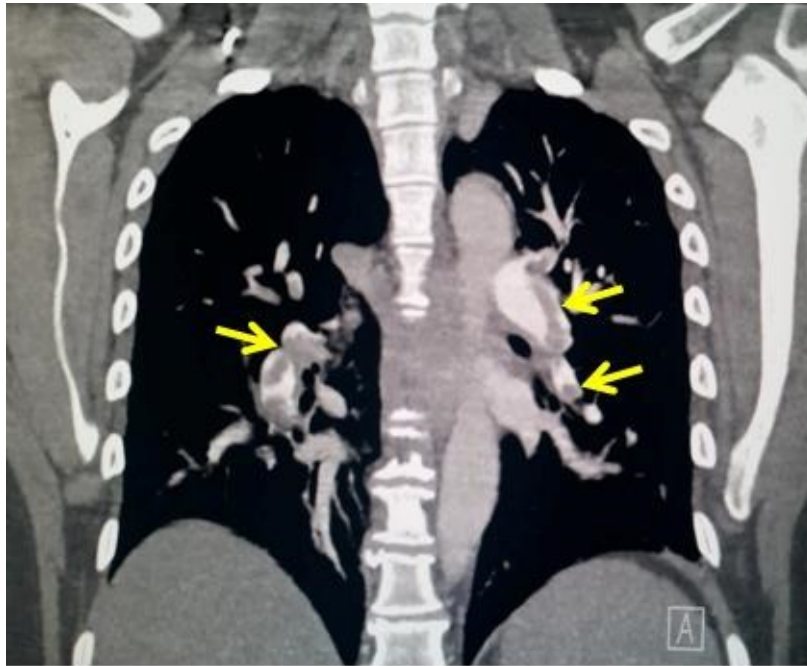


Figure 1. Coronal CT of the chest showing bilateral filling defects in pulmonary arteries representing pulmonary embolism.

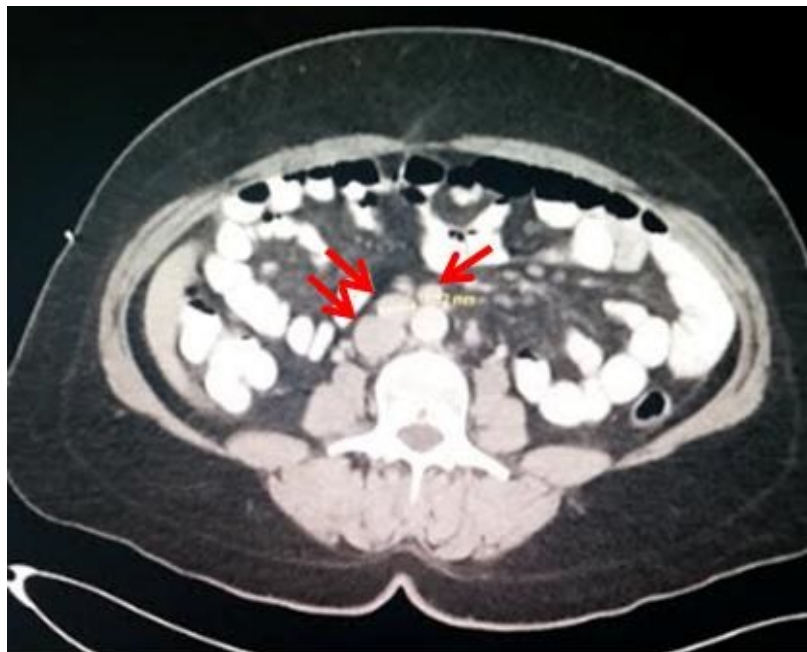


Figure 2. Axial CT of the abdomen/pelvis showing periaortic lymphadenopathy suggestive for metastatic disease.

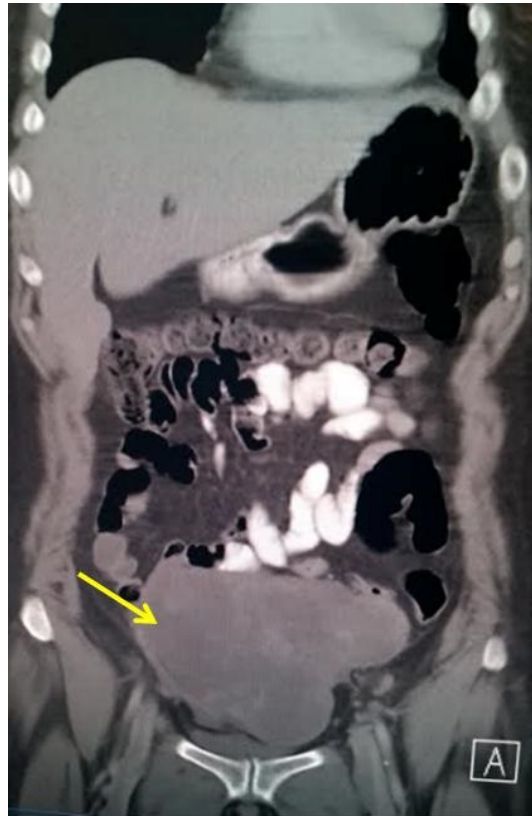


Figure 3. Coronal CT of the abdomen/ pelvis showing a 13 x 13.6 cm solid and cystic mass above and to the right of the uterus concerning for right ovarian neoplasm.

A 43-year-old woman with a history of anemia, thrombocytopenia, and recent treatment for pyelonephritis was transferred to our hospital for increasing shortness of breath. Four months prior to admission, she developed unprovoked bilateral deep vein thrombosis (DVT) and pulmonary emboli (PE) and was started on rivaroxaban at that time. At presentation, she was complaining of worsening shortness of breath, heavy menstrual bleeding and pain in her calves. CT angiography of chest showed multiple pulmonary emboli to the lower lobes and left upper lobe (Figure 1) and lower extremity venous Doppler showed extensive, acute deep vein thrombosis involving the femoral, popliteal and calf veins bilaterally.

Rivaroxaban was held due to anemia and thrombocytopenia and there was concern for respiratory failure since she developed new DVT and PE. She was transfused with 1 unit of packed red blood cells and started on a heparin drip. She continued to have significant menorrhagia, the heparin drip was discontinued, and subsequently, an inferior vena cava filter was placed.

On further questioning, the patient reported a 26 pound weight loss over the past three weeks. This combined with her menorrhagia requiring blood transfusion prompted

further imaging. CT of the abdomen and pelvis showed a 13 x 13.6 cm solid and cystic mass representing a right ovarian neoplasm that was contiguous with the uterus as well as periaortic adenopathy suggestive of metastasis (Figures 2 and 3). Further investigation into the patient's family history identified significant history for breast and ovarian cancers on her maternal side. Genetic testing of the patient showed a germline mutation in the MSH2 gene, consistent with Lynch syndrome.

Lynch syndrome, also known as hereditary non-polyposis colorectal cancer, is a hereditary cancer syndrome characterized by mutations in DNA mismatch repair genes. The majority of affected individuals will develop colorectal or endometrial malignancies; however these individuals are also at increased risk for developing ovarian neoplasms. The lifetime risk of developing ovarian cancer in women with Lynch syndrome is 7% (3-14%) compared to 1.4% in the general population (1). However, there is no survival difference between women with Lynch syndrome and the general population (1). If ovarian malignancy is present at diagnosis of Lynch syndrome, prophylactic hysterectomy and bilateral salpingo-oophorectomy is recommended (2). Otherwise, management can include prophylactic surgery or screening with annual pelvic exams and transvaginal ultrasounds. Persons with lynch syndrome should also receive surveillance for other associated malignancies such as colorectal or endometrial cancer (1,2).

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References

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2. Lindor NM, Petersen GM, Hadley DW, Kinney AY, Miesfeldt S, Lu KH, Lynch P, Burke W, Press N. Recommendations for the care of individuals with an inherited predisposition to Lynch syndrome: a systematic review. *JAMA.* 2006 Sep 27;296(12):1507-17. [\[CrossRef\]](#) [\[PubMed\]](#)