Beyond The Acute Abdomen: Diagnostic Challenges in Abdominal Pain From Acute Intermittent Porphyria and Lupus

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Introduction

Chronic abdominal pain can be a diagnostic challenge, particularly in the case of an acute abdomen. A high index of suspicion for atypical etiologies must be maintained to avoid unnecessary interventions. Acute intermittent porphyria (AIP), a porphyrin metabolism disorder, may present with severe abdominal pain, accompanied by central and peripheral neuropathy. Similarly, systemic lupus erythematosus (SLE) may manifest with diffuse abdominal pain. We present the case of a patient with AIP and SLE presenting with acute diffuse abdominal pain mimicking peritonitis

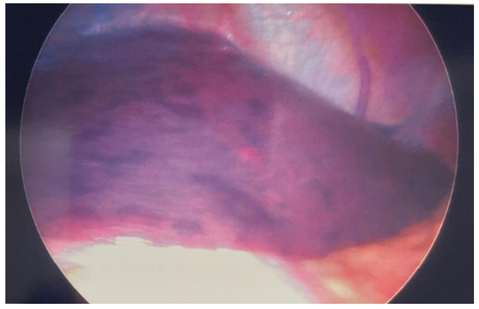
Case Report

A 24-year-old African American female presented with one week of anorexia, nausea, vomiting, and generalized abdominal pain. She had no prior medical or surgical history but had two recent emergency room visits for presumed urinary tract infection/cystitis. Laboratory studies revealed mild hyperbilirubinemia (1.5 mg/dL) and bilirubinuria without hematuria, pyuria, or bacteriuria. She was not anemic. CT imaging suggested possible minute pneumoperitoneum. On examination, she had diffuse tenderness with rebound but no guarding or rigidity.

Her history revealed recurrent abdominal pain episodes over three years, each requiring hospitalization and conservative management. Repeat CT with oral contrast during this admission showed no pneumoperitoneum or clear etiology. She was admitted for bowel rest, and further workup included hepatitis panel, autoimmune markers, and urine porphobilinogen.

Due to persistent pain without identified etiology, a diagnostic laparoscopy was performed. No bowel, colonic, or gynecologic abnormalities were identified; however, the liver appeared indurated and heterogeneous, and a biopsy was obtained. Postoperatively, her symptoms resolved, and she recovered uneventfully.

Pathology review demonstrated a well-differentiated hepatocellular lesion with glycogenotic hepatocytes, mild steatosis, and squamous epithelium associated with chronic inflammation. A postoperative MRI showed no focal hepatic lesion. Subsequent laboratory evaluation showed a positive ANA (1:320), elevated double-stranded DNA antibodies, and urine porphobilinogen (88.9 mg/g creatinine). Taken together, these findings were consistent with a dual diagnosis of AIP and SLE.



- Intraoperative Photo: Indurated and heterogenous appearing liver

Discussion/Conclusion

In the setting of acute abdomen with unknown etiology, diagnostic laparoscopy can be a useful tool to eliminate certain diagnoses. However, in patients with vague, intermittent, and recurrent symptoms, alternative diagnosis should be considered In this case, the diagnostic workup ultimately revealed findings consisted with both AIP and SLE, underscoring the importance of maintaining a broad differential and exercising clinical judgment before committing patients to the operating room.

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