Acute porphyria: anatomically unexplained abdominal pain

Asako Hoshi, Hiro Takefuji, Tetsuro Hayashi, Mitsuya Katayama, Shinji Yoshida

Introduction:
Acute porphyrias are disorders of heme synthesis that could cause acute neurovisceral symptoms including abdominal pain. The diagnosis is challenging due to their rarity in Japan and nonspecific symptoms. We report a case of acute porphyria presenting with abdominal pain and diagnosed only after laparoscopic surgery, then discuss when to suspect this rare entity.

Case Presentation:
A 19-year-old woman of Ghanaian and Japanese ancestry presented to our hospital with 2 week history of severe abdominal pain. Computed tomography of the abdomen and pelvis revealed an adnexal cyst. On suspicion of ovarian torsion, she underwent laparoscopic cystectomy in the gynecology service. Because the abdominal pain did not improve after surgery, she was referred to our department for further evaluation. She was suffering from urinary retention and numbness of both thighs postoperatively. On examination, she appeared distressed by the abdominal pain. The blood pressure was 164/122 mmHg, and the pulse 103 beats per minute; the other vital signs were normal. The abdomen was soft, with diffuse tenderness. No neurological deficit was present. Laboratory testing revealed the serum sodium of 124 mEq/l (fractional excretion of sodium 0.7%), C-reactive protein 0.3 mg/dl, negative antinuclear antibody, and normal levels of thyrotropin and cortisol. The 24-hour urinary porphobilinogen was markedly elevated to 176.2 mg, which confirmed the diagnosis of acute porphyria. Although her abdominal pain improved spontaneously, she repeated episodes of abdominal pain even after.

Discussion:
The differential diagnosis of abdominal pain is broad, including rare diseases. The first step in narrowing the differential diagnosis is to identify the involved organ. If it cannot be identified, humoral factors should be considered as the cause. Acute porphyrias should be suspected especially in patients with dysautonomia, neurological symptoms, or the syndrome of inappropriate antidiuresis.