

## Acute Intermittent Porphyria Presented with Acute Intestinal Pseudo-obstruction

Sir,

An 18-year-old female presented to our hospital because of the history of abdominal pain, with distension, constipation, and vomiting of 2 days' duration. On detailed evaluation, she told for frequent episodes of pain abdomen during menstrual cycle for the last 3 months, for which she did not take treatment. There was no history of chronic illness. At admission, her vital parameters were stable except a tachycardia of 108/min. The abdomen was tender and distended. Bowel sounds were absent. Rest of the physical examinations were normal.

The hematology revealed hemoglobin of 11.2 g/dL, total leukocyte count 6900/mm<sup>3</sup> with normal differential count and 245,000/mm<sup>3</sup> platelets. The blood sugar was 90 mg/dL, blood urea 36 mg/dL, creatinine 1.03 mg/dL, aspartate transaminase 64 IU/L, alanine transaminase 62 IU/L, lactose dehydrogenase 265 IU/L, total protein 6.26 g/dL, amylase 90 U/L, and lipase was 131 U/L. The electrolytes including sodium, potassium, calcium, phosphorus, and thyroid profile were within normal limit. He tested negative for HIV, hepatitis B virus, and hepatitis C virus. Her urine turned dark red on exposure to light. Porphyrins were detected on urine spot examination. A 24-h urinary 5 aminolevulinic acid (ALA) was 20.85 mg/L (normal range 1–7 mg/L) and porphobilinogen was 5.76 mg/L (0–3.40 mg/L). The flat plate X-ray of the abdomen in erect posture showed distension of bowel loops with multiple air-fluid levels suggestive of intestinal obstruction. The ultrasonography of abdomen and pelvis also showed distended bowel loops. The liver, spleen, kidneys, and pancreas were normal. She was managed with continuous Ryles tube suction and intravenous fluids. Glucose infusion was started in a dose of 400 g/day and she got relieved.

Acute intermittent porphyria (AIP) is common in middle-aged females with an autosomal dominant pattern of inheritance. It is an inherited metabolic disorder of heme synthesis and caused by a partial deficiency of porphobilinogen deaminase. The

diagnosis of AIP is based on the detection of urinary excretion of abnormal quantities of uroporphyrin, coproporphyrin, and porphobilinogen. The common presenting symptoms are neurovisceral and neuropsychiatric. These are intermittent attacks of acute abdominal pain, constipation, tachycardia, hypertension, and neuropsychiatric symptoms. The pain abdomen can be associated with nausea, vomiting, diarrhea, cholecystitis, bladder paresis, urinary retention, and intestinal obstruction. These symptoms are precipitated by various triggers, that is, porphyrinogenic drugs, alcohol, infections, calorie-restricted diet, stress, and hormonal (pregnancy and menstrual cycle).<sup>[1,2]</sup>

The mechanism of these symptoms is primarily related to the autonomic dysfunctions. The pain abdomen, constipation, and hypertension are explained by the autonomic neuropathy in AIP. The critical deficiency of heme and heme protein in the liver and/or in neuronal tissue and overproduction of porphyrin precursor ALA from the liver which is a direct neurotoxin is responsible for disruption of the blood–brain barrier and blood–nerve barrier. The ALA initiates neuronal damage by inhibiting Na-K ATPase, modifies glutamatergic, and formation of free radicals and reactive oxygen species.<sup>[3]</sup>

The autonomic dysfunction, enteric ganglionitis/ganglionopathy,<sup>[4]</sup> local vasoconstriction, and intestinal ischemia are responsible for gastrointestinal symptoms of the AIP. The involvements of autonomic fibers are supported by vagus nerve demyelination, axonal loss, and chromatolysis of sympathetic ganglion cell in autopsies.<sup>[5]</sup>

To conclude, in young patients presented with intestinal pseudo-obstruction, after excluding the surgical causes, should be evaluated for a rare metabolic cause – AIP, which is a medical emergency.

### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have

given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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### Conflicts of interest

There are no conflicts of interest.

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