Acute intermittent porphyria (AIP) is an autosomal dominant hepatic porphyria caused by a deficiency of hydroxymethylbilane synthase, an enzyme involved in heme biosynthesis. The onset of symptoms usually occurs in adolescence, with females being more affected than males. Patients with AIP usually present with neurological, gastrointestinal, and autonomic features. Rare manifestations of AIP during presentation can result in diagnostic dilemmas.

A 22-year-old male presented with epigastric pain radiating to back and vomiting for 3 days. His medical history was unremarkable. He was a regular alcohol consumer for 3 years and had his last drink 1 week prior to the onset of symptoms. He also used to smoke cannabis regularly. He denied history of high risk behavior and had no sick contacts. Examination was unremarkable except for mild epigastric tenderness. Investigations showed high serum amylase and lipase. Contrast enhanced computed tomography of abdomen showed bulky pancreatic body and tail. He was diagnosed to have acute pancreatitis due to alcohol and was managed symptomatically. He improved symptomatically 5 days following admission and was discharged.

He was readmitted 6 weeks later with epigastric pain after a binge of alcohol 1 day prior to the onset of pain. Examination then showed high blood pressure (150/100 mmHg) and mild epigastric tenderness. Investigations showed high serum amylase, lipase with mild hyponatremia (serum sodium 124 meq/l). The patient was euvoletic, serum osmolality was low, urinary osmolality and sodium were high suggesting syndrome of inappropriate antidiuretic hormone secretion. On the third day of admission, patient developed one episode of generalized tonic clonic seizures. After the episode of seizure, he had persistent tachycardia and high blood pressure. Magnetic resonance imaging of brain showed multifocal lesions in the right frontal, left temporal, and bilateral occipital subcortical white matter [Figure 1]. The lesions were hypointense on T1-weighted images, hyperintense on T2-weighted and fluid-attenuated inversion recovery without any diffusion restriction and contrast enhancement and apparent diffusion coefficient (ADC) maps were hyperintense suggesting posterior reversible encephalopathy syndrome (PRES) [Figure 2]. Electroencephalography showed diffuse slow waves without epileptiform discharges and cerebrospinal fluid analysis was normal. Tests for connective tissue disorders, viral infections, heavy metal poisoning, drug overdose, thyroid function, cannabis abuse, and antithyroid antibodies were all negative. Repeat contrast enhanced computed tomography of abdomen showed edematous and bulky pancreas. Following the episode of seizure, the patient developed psychiatric manifestations including suicidal tendency, walking restlessly in the night, and visual hallucinations. The neuropsychiatric manifestations, PRES on imaging, autonomic dysfunction, and syndrome of inappropriate antidiuretic hormone secretion in a young patient made us consider porphyria. The porphyrin
precursors in the urine (porphobilinogen and δ-aminolevulinic acid) were high suggesting acute intermittent porphyria. The patient was managed with glucose infusion and a high carbohydrate diet. Few days later he developed respiratory distress and was mechanically ventilated. He later developed hypotension possibly secondary to dysautonomia. He was sent to another center for further management as heme was not available in our hospital.

A combination of neurological symptoms along with abdominal pain and autonomic dysfunction in a patient make us all think of AIP. Atypical presentations delay the diagnosis and treatment. Our patient had two episodes of acute pancreatitis and the first episode was attributed to alcohol. The second episode of pancreatitis was associated with other features like psychiatric manifestations, PRES on imaging, autonomic dysfunction, and syndrome of inappropriate antidiuretic hormone secretion which helped us in considering an alternate diagnosis. Our patient used to consume alcohol regularly and had history of cannabis abuse which could explain most of the manifestations which he had including psychiatric, autonomic dysfunction, pancreatitis, and hyponatremia.

Twenty-two cases of PRES with AIP were reported previously in the literature and most of them were females. AIP attacks in them usually started in the third and fourth decades of life, and the mean age of PRES at presentation was 44 years. Most of the patients with AIP-associated PRES presented with seizures. The precise mechanism of vasoconstriction in AIP-associated PRES remains unknown. Various mechanisms were postulated previously like hypertension during an acute attack of AIP exceeding the autoregulation limit and hyperperfusion followed by breakdown of the blood–brain barrier leading to vasogenic edema. Severe heme deficiency during an acute attack of AIP causes a lack of the heme protein nitric oxide synthase, which can cause vasoconstriction. Acute pancreatitis as an initial presentation of AIP was previously reported. Acute pancreatitis in AIP may be due to spastic obstruction of the sphincter of Oddi, because of autonomic neuropathy or porphyrin neurotoxicity (due to excessive levels of the porphyrin precursors, heme deficiency in nervous tissue, and depletion of essential substrates or cofactors arising from the heme biosynthetic pathway defect). Patients having acute pancreatitis with neuropsychiatric manifestations and autonomic dysfunction should be tested for porphyria.

To conclude, atypical manifestations of a disease make the diagnosis difficult. The knowledge of unusual presentations of rare diseases like AIP is necessary to the treating physicians in order to make a prompt diagnosis. We report a patient with AIP who had unusual manifestations including acute pancreatitis and PRES during presentation causing diagnostic dilemma.

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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