Abstract

Acute Intermittent Porphyria (AIP) is a rare inherited disorder of heam synthesis which characteristically manifests episodes of abdominal pain, neuropsychiatric symptoms and autonomic symptoms. Due to its rarity and symptomatology the diagnosis is often delayed.

Case presentation

A 20 year previously healthy student investigated inward for 2 episodes of acute abdominal pain associated with nausea, constipation and treated symptomatically and discharged due to normal initial clinical assessment and investigations, presented with generalized tonic clonic seizure with tachycardia, high blood pressure, rhabdomyolysis with hyponatremia which needed intensive care unit admission with ventilator support. She had a positive Hoesch Test (Urine for porphobilinogen) confirming the diagnosis of AIP with characteristic history.

Conclusion

Acute intermittent Porphyria is a rare inherited disease where the diagnosis is often delayed or misdiagnosed. This case emphasizes the importance of early evaluation for possibility of AIP in a patient with recurrent abdominal pain.

Key words

Acute intermittent Porphyria, Recurrent abdominal pain, urine porphobilinogen