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SCIENTIFIC LETTER

Discovery of Porphyria in a Postoperative Surgical Patient

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To the Editor: A 17-y-old boy was referred to us for abdominal pain for the past 3 mo. He had undergone appendicectomy 10 d back in a private hospital. He was the only child of a nonconsanguineous marriage with no similar complaints in the family. Within next 24 h, he developed bilateral upper limb weakness, diaphragmatic palsy, aphonia, and respiratory distress. At admission in PICU, he was conscious, responsive, aphonic with hypertension and tachycardia. Lower limbs were spared initially but progressively flaccid quadriparesis ensued. Differentials included pharyngeal-cervical-brachial variant of Guillain–Barre syndrome and brachial plexus neuritis. Investigations revealed hyponatremia (serum sodium 117 mmol/L). Rest of the haematological and biochemical investigations were normal. MRI head and spine and cerebrospinal fluid analysis was normal. Nerve conduction studies (NCV) revealed acute motor axonal polyradiculopathy (C5-C7). Key clinical findings of constipation, hypertension, hyponatremia, proximal symmetric motor weakness, and high colored urine made acute intermittent porphyria (AIP) a viable suspect. Quantitative urine porphobilinogen (PBG) was 63.49 mg/d (normal value < 3.40 mg/d) confirming the diagnosis of AIP. Although, intravenous hematin is the drug of choice, it was unavailable in our region so we resorted to treat with intravenous dextrose infusion followed by carbohydrate rich enteral feeds later. He received mechanical ventilation for 2 wk with gradual recovery and was discharged after 3 wk of hospitalization. Genetic analysis was declined by the family due to financial constraints.

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Porphyrias are rare genetic disorders involving hemebiosynthetic pathway. Acute porphyrias with neurological presentations include AIP, variegate porphyria (VP), and hereditary coproporphyria (HC) [1]. Lack of dermatological findings, made AIP the most likely diagnosis. It results from deficiency of PBG deaminase and can present with abdominal pain, altered sensorium and autonomic dysfunction [2]. Peripheral neuropathy can be presenting feature in 10%–40% cases. Predilection for upper limbs, proximal symmetric motor weakness with NCV findings of axonal polyradiculopathy are characteristic of porphyria neuropathy [3].

Urine for porphyrins is the most widely used screening tool [4]. Further confirmation requires genetic testing which is expensive, and often unavailable. Treatment involves avoidance of precipitating factors, administration of intravenous hematin or alternatively, high dextrose solutions enterally or parenterally [5]. Thus, porphyria should be considered as a differential diagnosis in any patient with unexplained neurological symptoms.

Compliance with Ethical Standards

Conflict of Interest None.

References

1. Puy H, Gouya L, Deybach JC. Porphyrias. Lancet. 2010;375(9718):924–37.
2. Balwani M, Singh P, Seth A, et al. Acute intermittent porphyria in children: a case report and review of the literature. Mol Genet Metab. 2016;119(4):295–9.
3. Simon NG, Herkes GK. The neurologic manifestations of the acute porphyrias. J Clin Neurosci. 2011;18(9):1147–53.
4. Kenny JM, Chipolombwe MJ, Molyneux E. A case of repeated coma in an 8-year-old boy. Trop Dr. 2007;37(4):264–5.
5. Patell R, Dosi R, Joshi H, Shah P, Tripathi R. Challenges in the successful management of a case of acute intermittent porphyria in India. Trop Dr. 2016;46(3):150–3.