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An atypical case of Guillain–Barré syndrome: acute intermittent porphyria

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Dear Editor,

We report here the case of a 26-year-old women hospitalized in our intensive care unit (ICU) for acute respiratory failure associated with an acute polyradiculoneuropathy considered initially as a Guillain–Barré syndrome (GBS). Her medical history began with acute abdominal pain mimicking appendicitis while on vacation in Africa, during which time she was taking antimalarial prophylaxis. An appendectomy was performed; the postoperative period was promptly complicated by peripheral neuropathy predominating in the lower limbs. Worsening neuropathy led to respiratory failure requiring mechanical ventilation and she was transferred to our ICU. Clinical findings on admission included proximal weakness with hypoesthesia, facial diplegia, and abolished tendon reflexes, except for the Achilles reflexes; additional central nervous system signs were present including visual blurring, hallucinations, and seizures. Hypertension, sphincter dysfunction, and severe gastroparesis impeding enteral feeding were initially ascribed to dysautonomia. The cerebrospinal fluid was normal and electromyography revealed acute motor axonal neuropathy. Blood chemistry revealed hyponatremia and persistent acute renal failure despite rehydration. Given the several atypical features for GBS, the diagnosis of acute intermittent porphyria (AIP) was finally suspected with significant delay. We confirmed AIP by the change in urine color when exposed to light and by high urinary porphobilinogen titration. Specific treatment with daily human hemin (Normosang®, Orphan Europe) was started as recommended [1], but she received a prolonged treatment as recurrent documented pulmonary infections worsened AIP (Fig. 1). AIP was controlled by administering high doses of human hemin, avoiding all contraindicated medications, controlling infections, and optimizing nutrition via a jejunostomy. The patient required over 6 months of mechanical ventilation via a tracheostomy, and is currently recovering in a rehabilitation center.

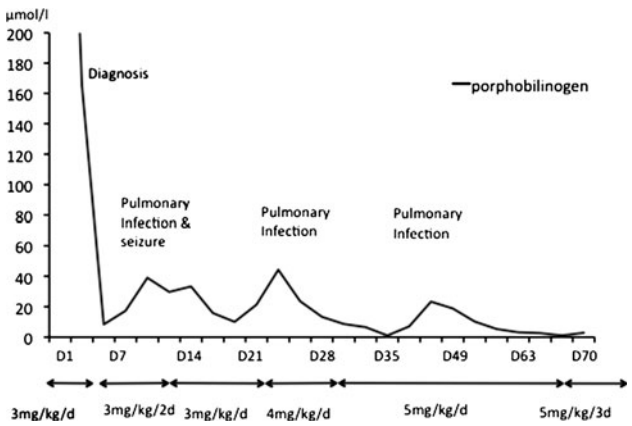

Fig. 1 Changes in porphobilinogen titration during the 5 months of ICU stay. Pulmonary infections were associated each time with an increasing titration of porphobilinogen; the first episode was also associated with seizures and hyponatremia. Recurrent infections worsened neurologic symptoms and delayed recovery and rehabilitation

AIP is an unusual autosomal dominant disorder resulting in neurovisceral attacks, occurring mostly in women. The most common manifestation is abdominal pain (often accompanied by constipation and signs of ileus), mimicking a surgical emergency. Tachycardia, hypertension, and sweating are often present. Neuropsychiatric manifestations include anxiety, agitation, and hallucinations. Seizures may be due to hyponatremia or represent a neurological manifestation of porphyria. Neuropathy, mostly muscle weakness, develops when contraindicated drugs are used. The differential diagnosis includes axonal forms of GBS and Miller–Fisher syndrome (opthalmoplegia and antibodies against GQ1b were absent in our case). Hyponatremia may be due to the syndrome of inappropriate antidiuretic hormone secretion, but gastrointestinal or renal sodium losses are sometimes important (until dehydration) [1].

We report here a severe case of AIP illustrating a diagnostic pitfall, and difficulties in management in an ICU.

Key points of management include:

1. Cautious selection of drugs used during supportive care (http://www.drugs-porphyria.org) and management with a reference center (European Porphyria Network, EPNET).
2. Administration of specific therapy based on human hemin [2],

although dosage and duration are unknown for chronic severe AIP. We empirically used high doses over several months as repeated tapering attempts were associated with rebounds of urinary porphobilinogen.
3. Early nutrition based on carbohydrate loading, possibly requiring a jejunostomy [3].
4. Tracking infections and providing prompt therapy is important because they are often the cause of AIP exacerbations.
5. Tracheostomy should be considered early in patients requiring mechanical ventilation, because of an expected prolonged duration of mechanical ventilation.
6. Replacement of deficient hepatic enzymes might restore excretion of porphyrin precursors to normal [4]. Liver transplant is not recommended during the acute phase as no outcome data are available in ventilator-dependent patients [5].

Finally, AIP is a rare differential diagnosis of acute motor axonal neuropathy and should be suspected when associated with abdominal pain in young women. The diagnosis is simple and cost-effective, so measurement of urinary porphobilinogen should be routinely obtained in patients with axonal GBS.

Conflicts of interest None.

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