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Importance of Screening Acute Porphyria in Severe Polyneuropathy reckoned as Guillain Barre Syndrome

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Introduction

Acute intermittent porphyria (AIP) is a rare, genetic disorder which induces severe disabilities. Development of symptoms is associated with various exacerbating factors, from medications to poor oral intake or stress in patients with genetic factors. Clinical manifestations include abdominal pain, neurological symptoms and sensorimotor polyneuropathy. Considerable recovery could be expected with early diagnosis and treatment.

Case presentation

A 23-year-old single woman presented to the hospital because of cramping abdominal pain in February 2018. An abdominal computed tomography (CT) scan showed two intussusception sites. She was discharged 10 days after underwent laparoscopic manual reduction and post operation care, with limited oral intake. Six hours after discharge, she had a sudden deterioration of consciousness level and four serial convulsions taking about 3-5 minutes long. Brain CT and CSF tapping were normal, but the patient's condition continued to aggravate with the need to admit to intensive care unit (ICU) requiring intubation and mechanical ventilation. Magnetic Resonant Imaging (MRI) was taken and posterior reversible encephalopathy syndrome was observed without definite etiology (Figure 1). On the 23th postoperative day (POD), both upper and lower limbs power was of Medical Research Council (MRC) grade 2/5, progressively deteriorated to grade 1/5 the day after, with all deep tendon reflex were absent in all limbs. Nerve conduction study (NCS) revealed severe sensorimotor axonal polyneuropathy with no response in routine NCS of bilateral upper and lower extremities. Patient was considered Guillain-Barre Syndrome (GBS) and treated with intravenous immunoglobulin for 5 days on POD 31, with no definite effect. Abdominal pain, neurological symptoms and unexplained severe polyneuropathy raised the suspicion of AIP. 10 days after suspicion, AIP diagnosis was confirmed by urine porphobilinogen (102.01 mg/day) on POD 40 (Table 1). Intravenous hemin was administered 2 times after diagnosed, for 4 days each. All extremities power showed no change after hemin injection. NCS was taken 2 months later after quadriplegia. There were no responses in all sensory and motor nerves tested. It is considered as a sequel of AIP.

Conclusion

AIP could cause severe peripheral axonal polyneuropathy. Porphyric neuropathy should be considered as one of the differential diagnoses in a patient with an acutely progressing severe polyneuropathy, especially as with GBS.

Table 1. Laboratory data of case patient at POD 31.

Laboratory tests	Values	Normal range
Porphobilinogen, 24hour Urine	102.01mg/day	0 - 2.5mg/day
Delta-Aminolevulinic acid, 24hour Urine	31.0mg/day	1.5– 7.5mg/day
Total porphyrin, blood	Negative	
Total porphyrin, urine	Positive	
Spot urine Coproporphyrin	Positive	
Urine Porphyrin	Positive	
Urine Uroporphyrin	Positive	
Anti GM1 Ab IgM	Negative	
Anti GD1b Ab IgM	Negative	
Anti GQ1b Ab IgM	Negative	
Anti GM1 Ab IgG	Negative	
Anti GD1b Ab IgG	Negative	
Anti GQ1b Ab IgG	Negative	
Anti-Mitochondrial Ab	Negative	
Anti-Smooth muscle Ab	Negative	
LKM1 Ab	Negative	
ANA titer, serum	< 1 : 40	< 1 : 40

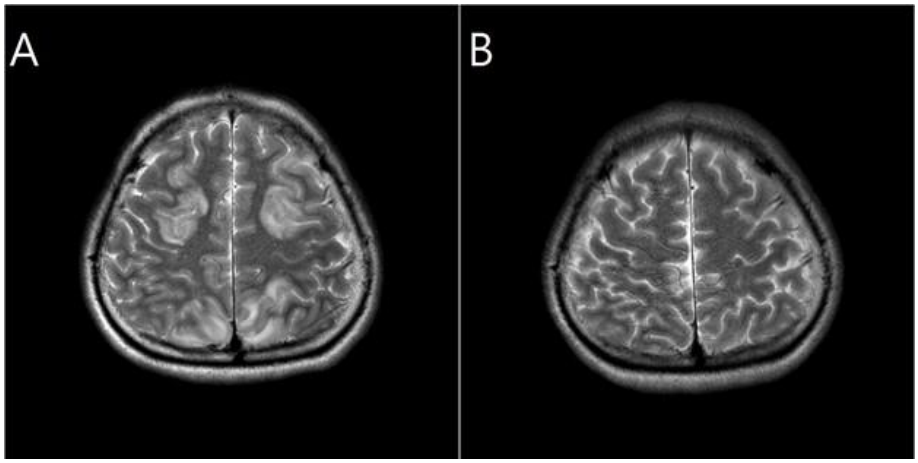

Figure 1. Brain magnetic resonance imaging performed on the POD 10(A), POD 21(B). Axial T2-weighted images of POD 10(A) revealed both frontal and parietal cortex and subcortical white matter signal intensity change. POD 21 images(B) revealed complete remission.