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A 20 Years Old Lady with Quadriparesis and Convulsion: A Case Report

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Abstract

Acute intermittent porphyria (AIP) is a rare autosomal dominant metabolic disease that is caused by a partial deficiency of hydroxymethylbilane and deficiency results in the accumulation of delta-aminolaevulinic acid (ALA) and porphobilinogen (PBG). ALA is believed to be neurotoxic and affects the autonomic, peripheral and central nervous systems. We found a 20 years old Bangladeshi female complaining of severe debilitating weakness of all limbs. She was suffering from recurrent upper abdominal pain for last one year and underwent an emergency appendicectomy 4 months back. 2 months back she experienced several episodes of generalized tonic clonic seizure. Last 21 days she suffered from acute onset quadriparesis and tingling and numbness in distal part of limbs with high colored urine which especially found at morning after wakening but she denied any history of fever, dysuria. She had palpitation, extreme sleep disturbance, neck and back pain. She had a history of injectable contraceptive 1 month back. Her vitals were normal except there was tachycardia. Motor examination revealed flaccid quadriparesis. All modalities of sensation were impaired up to knee. The electrophysiological findings are consistent with sensory-motor axonal polyneuropathy. The result of quantitative urinary PBG was positive. The diagnosis of acute intermittent porphyria was made on the basis of available evidence. The patient showed improvement in symptoms with the initial treatment. The disease management was focused on oral anti-seizure medication, sleep disorder medication, a controlled diet (300gram glucose/day), physical therapy and psychiatric support. Though, Hematin was not given due to unavailability. *[Journal of National Institute of Neurosciences Bangladesh, July 2025;11(2):191-197]*

Keywords: Acute intermittent porphyria; delta-aminolaevulinic acid; porphobilinogen; Hematin;

Introduction

Porphyria is a group of inherited disorders caused by defects in heme biosynthesis. Three forms of porphyria are associated with peripheral neuropathy: acute intermittent porphyria (AIP), hereditary coproporphyria (HCP), and variegate porphyria (VP). The acute neurologic manifestations are similar in each, with the exception that a photosensitive rash is seen with HCP and VP but not in AIP.[1] Acute intermittent porphyria (AIP) is a rare autosomal dominant metabolic disease that is caused by a partial deficiency of hydroxymethylbilane (HMBS), the third enzyme in the heme biosynthesis pathway. This deficiency results in the accumulation of upstream metabolic products, such as delta-aminolevulinic acid (ALA) and porphobilinogen (PBG). ALA is believed to be neurotoxic and affects the autonomic, peripheral, and central nervous systems, thereby causing related symptoms, including severe abdominal pain, hypertension, respiratory failure, and quadriplegia. Because AIP has a broad spectrum of clinical manifestations, it can be easily confused with other diseases[2]. Attacks of porphyria can be precipitated by certain drugs (usually those metabolized by the P450 system), hormonal changes (e.g., pregnancy, menstrual

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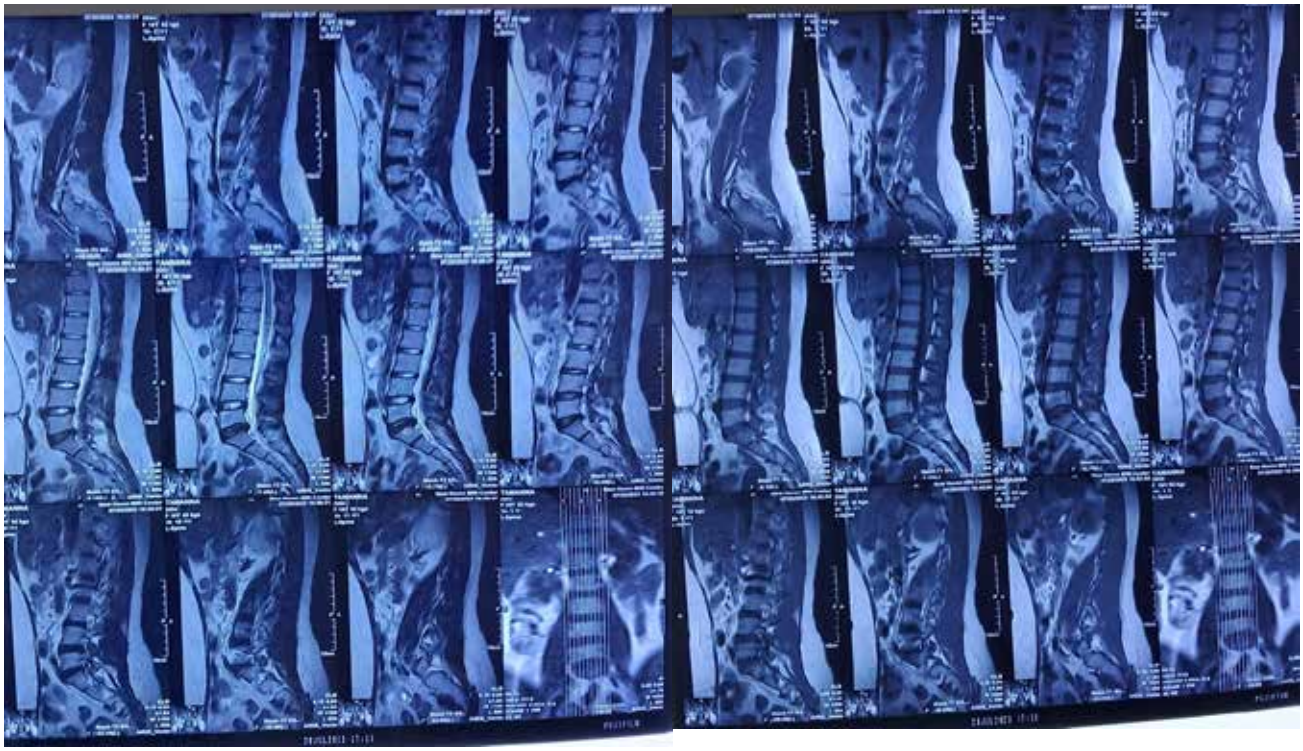
cycle), and dietary restrictions. An acute attack of porphyria may begin with sharp abdominal pain. Subsequently, patients may develop agitation, hallucinations, or seizures. Several days later, back and extremity pain followed by weakness ensues, mimicking GBS. Weakness can involve the arms or the legs and can be asymmetric, proximal, or distal in distribution, as well as affecting the face and bulbar musculature. Dysautonomia and signs of sympathetic overactivity are common (e.g., pupillary dilation, tachycardia, and hypertension). Constipation, urinary retention, and incontinence can also be seen1. Because many symptoms of the porphyria's are nonspecific, diagnosis is often delayed. Laboratory measurement of porphyrin precursors (5 aminolaevulinic acid (ALA) and porphobilinogen (PBG)) in the urine or porphyrins in the urine, plasma, erythrocytes, or feces is required to confirm or exclude the various types of porphyria. However, a definite diagnosis requires demonstration of the specific gene defect. The genes encoding all the heme biosynthetic enzymes have been characterized, permitting identification of the mutations causing each porphyria. Molecular genetic analyses now make it possible to provide precise heterozygote or homozygote identification and prenatal diagnoses in families with known mutations[3]. The reported prevalence of HMBS gene mutations is high (1/1,675); however, the clinical penetrance is comparatively low: in Europe, AIP is estimated to affect approximately 5.4 people per million (1/ 185,000). Moreover, 10.0% to 40.0% of patients may have symptoms of peripheral nerve damage during an AIP attack, but they are usually only mildly affected[2]. However, in the present case, the patient had neuropathy that presented intensely, with a relatively unusual clinical picture and severe, long-lasting sequelae.

Case Presentation

A 20 years old Bangladeshi female admitted to the National Institute of Neurosciences and Hospital, Agargaon, Dhaka, Bangladesh through emergency department on 16th of October,2023 complaining of weakness of all limbs for 21 days. Initially it started from lower limbs simultaneously both proximal and distal group of muscles and within one day it involved proximal and distal group of muscles of upper limbs. The weakness was so severe that that within two days she became bed bound. She also mentioned about tingling and numbness in distal part of lower limb. On systemic query she said, she has been suffering from upper abdominal pain for last one year. The pain was mild to moderate in intensity associated with vomiting and burning sensation of abdomen. She visited a gastroenterologist but symptoms did not improve. 4 months back she felt lower abdominal pain and the pain



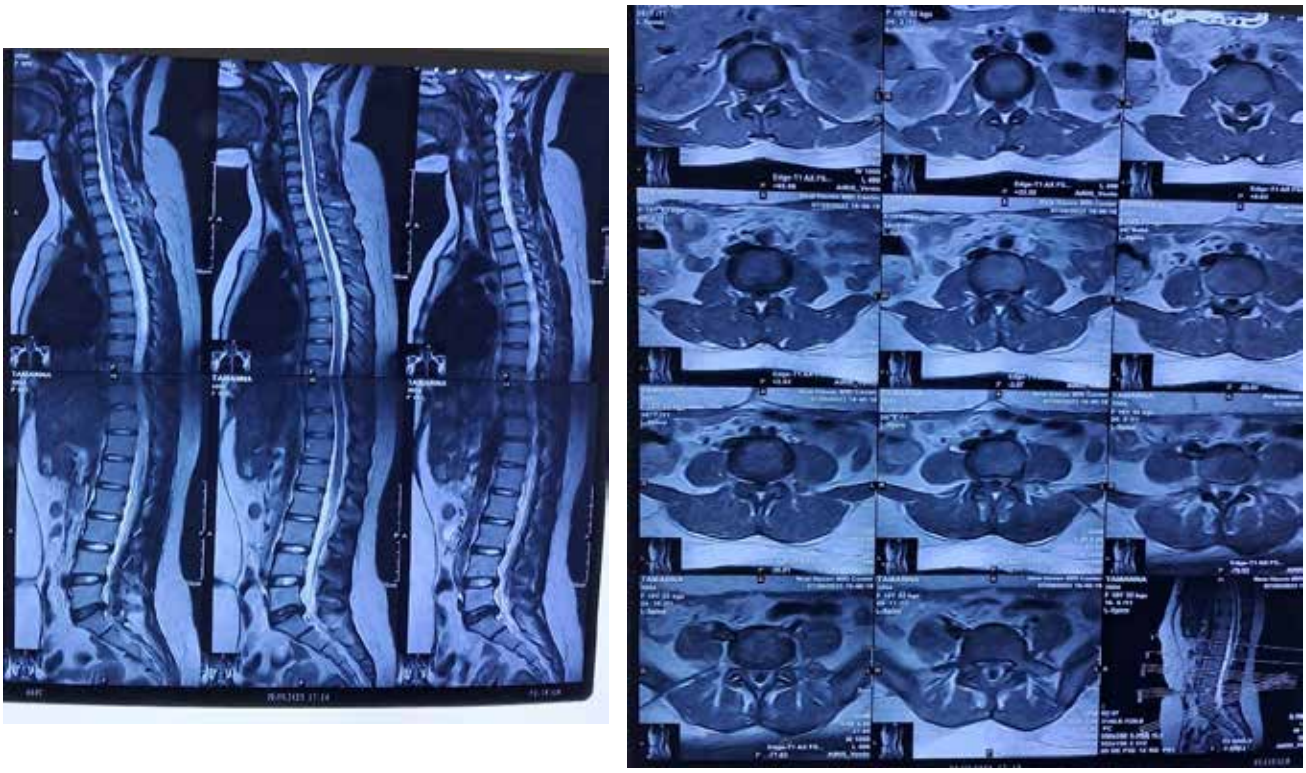
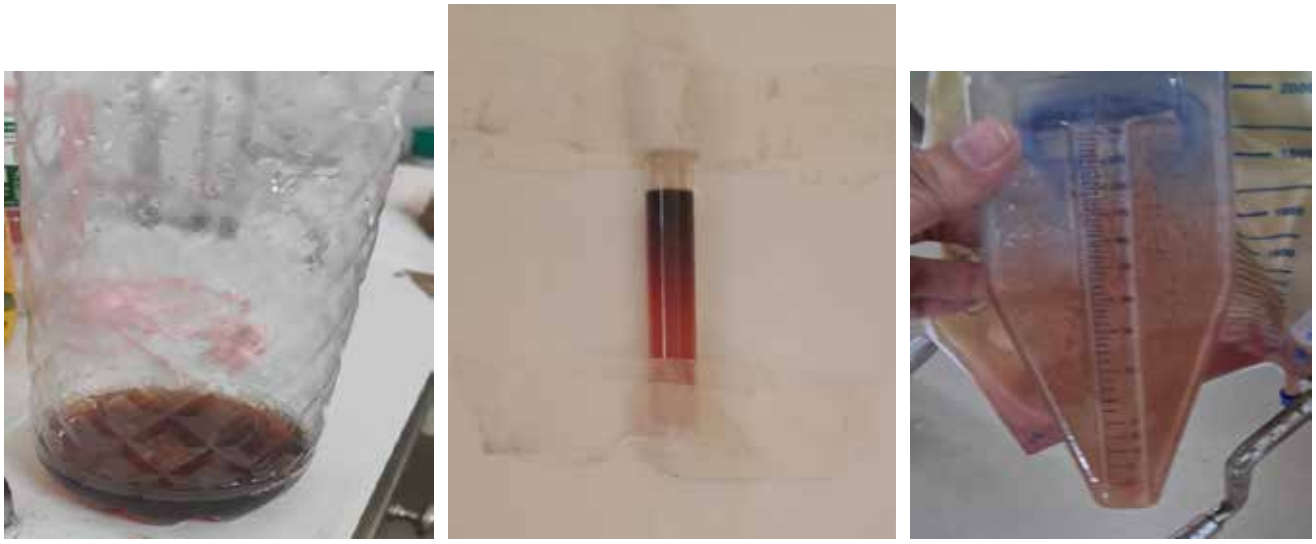


Figure I: MRI of Lumbosacral Spine

was so severe that she was forced to get admitted in a hospital. With the available investigation and clinical basis, she was diagnosed as a case of appendicitis and appendicectomy was done immediately. 10 days after the operation, she developed weakness of both the lower limbs but it resolved within seven days. They visited a physician where MRI of Lumbosacral spine was done but no abnormality was found.

2 months back she again experienced abdominal pain and this time she developed several episodes of generalized tonic clonic seizure. 21 days before



Day-1 Day-3 During Discharg

Figure II: Urine Colour

admission she suffered from acute onset quadriparesis. Her sister also gave history of dark coloured urine which especially found at morning after wakening but she denied any history of fever, dysuria, frothy urine, or facial swelling. She had palpitation, extreme sleep disturbance, neck pain, back pain. The colour of the urine changes with sunlight. She had an injection of intramuscular contraceptive 1 month back.

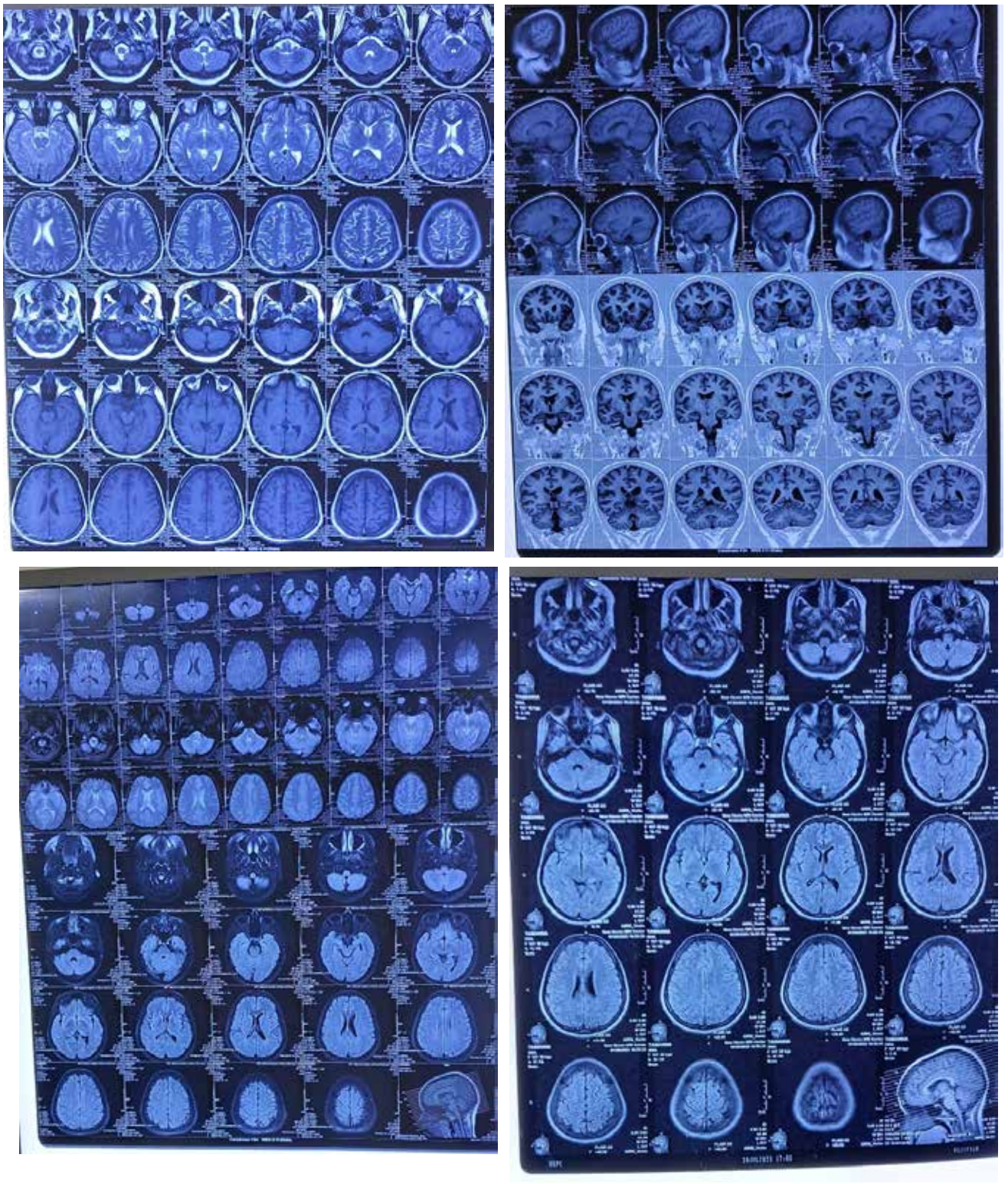


Figure III: MRI of Brain

NCS of limbs showed that CMAP amplitudes are reduced to absent in all studied nerves. F wave latencies are absent in right median or ulnar nerves.SNAP amplitudes are reduced to absent except right median nerve. The electrophysiological findings are consistent with sensory axonal polyneuroradiculopathy. Faced with the probable diagnosis of porphyria (while there was still no official result for urinary PBG), symptoms were managed with medication adjustment according to the lists of non harmful drugs found on the website of Up To Date and potentially safe medication list from the UK Porphyria Medicines Information Service(UKPMIS). The list of allowed drugs was attached to the patients discharge paper and the patient as well as her attendant were instructed. The patient showed improvement in symptoms with the initial treatment,given the difficulty in acquiring Hematin. The disease management was focused on oral anti seizure medication (levetirecetum), sleep disorder medication (first chloral hydrate later Chlorpromazine), a controlled diet after dicussing with our Transfusion Medicine Unit (300 gram glucose/day), physical therapy,psychiatric support. She was discharged for OPD follow up. The result of urinary PBG was released on 31/10/23. Given the diagnostic confirmation, a report was made so that the patient could acquire the Hematin.

Table 1: Investigation summary

Name of investigation		Values
Complete Blood Count	Hb%	13.5 gm/dl
	ESR	7 mm in 1st hour
	MCV	76 fl
	MCH	25 pg
	MCHC	33 g/dl
	Platelet	263 X 109/L
Peripheral Blood Film	RBC shows normocytic normochromic, WBC was matured, platelets were adequate in number	
HBsAg	Negative	
Anti HCV	Negative	
Serum Creatinine	0.59 mg/dL	
	Serum Electrolytes	
	Na+	134 mmol/L
	K+	4 mmol/L
	Cl-	98 mmol/L
RBS	5.44 mmol/L	
SGPT	276 U/L	
Urine R/E	Normal	
Urinary PBG (Column chromatography) Qualitative measurement	Positive	
ECG	Sinus tachycardia	
Awake EEG	Consistent with normal findings	

Discussion

AIP is a rare metabolic disease caused by HMBS mutations. Although AIP occurs in all races, it is thought to be most common in northern Europe, with a calculated prevalence of 5.4 per million[4,5]. Between 10.0% and 40% of patients with porphyria may exhibit symptoms of peripheral nerve damage during an AIP attack, and such patients are usually mildly affected6. It is rare for a patient to experience severe peripheral nerve damage, such as that which caused the quadriplegia described in the present case report. There were several indications that could have led to the diagnosis of AIP in the present case because the patient had recurrent abdominal pain and peripheral nerve damage, but an accurate diagnosis was still delayed. In practice, 11.0% of patients diagnosed with AIP have previously undiagnosed acute porphyria; according to Pischik et al.[7], this is because physicians commonly misdiagnose this cause of acute polyneuropathy or encephalopathy. Neuropathic symptoms in most patients reach their maximal intensity at 2 weeks after onset[8]. In the present case, severe neuropathy, including quadriplegia, developed in 24 hours. Moreover, when the patient admitted in the ward, the symptoms of abdominal pain had already disappeared but the urine color was dark. Furthermore, both the analysis of precursors and porphyrins and the genetic analysis were not available in the local hospital. The relatively long duration between suspecting AIP and obtaining the results of the urinary PBG, which was outsourced to LalPath, India, delayed the patient's diagnosis by 15 days. The mechanisms of porphyric neuropathy remain unclear. The two major proposed mechanisms are related to a lack of heme, leading to dysfunction of the energy-dependent Naþ/Kþ ATPase[9], as well as the direct neurotoxicity of accumulated porphyrin precursors, including ALA and PBG[10,11]. The early administration of heme arginate may prevent the onset of neuropathy by reducing the expression of the rate-limiting liver enzyme ALA synthase 1 (ALAS1), thus impeding further progression[12]. Although hematin is the standard of treatment, especially when

neurological symptoms occur, our patient was treated with 10.0% intravenous glucose infusion during the AIP attack because heme arginate is not available in Bangladesh. Delayed onset of therapy and irreversible neurological damage may contribute to the poor effects of heme treatment[13,14]. The present patient's neuropathy deteriorated to the extent of quadriplegia which may be correlated with a poor outcome[15]. Similarly, O'Malley et al.[16] reported that, in a woman with complete flaccid paralysis, most muscle groups had returned to full strength after 3 years. Accordingly, the recovery period of porphyric neuropathy is calculated to be at least several months, and each patient's prognosis needs continuous attention[17]. We reported rare clinical symptoms of AIP: rapidly progressive neuropathy attacks with severe manifestations that are life-threatening and often difficult to diagnose. Through careful neurophysiological examinations, laboratory examinations, imaging examinations (MRI of the Brain), we finally confirmed the diagnosis of our patient and effectively contained her disease progression. This report may provide some reference for similar cases in the future. The main limitation of this case is that the patient was unable to receive heme treatment, which may have affected her prognosis.

Conclusion

The clinical presentations of AIP are nonspecific and may mimic many common surgical and medical conditions, such as appendicitis and GBS, which can result in diagnostic dilemma of this disease. When a patient present with severe progressive neuropathy, AIP should be taken into consideration, despite the appearance of symptoms of common diseases in the Neurology wards, such as hypokalemic periodic paralysis, myasthenia gravis, and severe GBS. Early diagnosis can avoid serious peripheral nerve damage, prevent recurrent episodes, and even reduce mortality.

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Conflict of interest: All authors declare that they have no competing interest.

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Authors' contributions
Conception and design: Amin MAA, Hossain S, Uddin MN; Acquisition, analysis, and interpretation of data: Uddin MN, Sarker M, Ahmed KMA; Manuscript drafting and revising it critically: Amin MAA, Hossain S; All authors read and approved the final manuscript.

Data Availability
The datasets generated and analyzed during this study are not publicly available due to patient confidentiality and institutional data protection policies. However, anonymized data may be made available from the corresponding author upon reasonable request, subject to approval by the institutional ethics committee.

Ethics Approval and Consent to Participate
Written consent was taken.

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