Case Report

Pain Abdomen: Journey of a Young Lady from a Gastroenterologist to a Neurologist

Sahoo Haramohan[1], Chandra Krishna[2], Verma Vikas[2], Sharma Gaurav[3], Goyal Bhupesh[4]

[1]Assistant Professor, Department of Neurology, Heritage Institute of Medical Sciences, Varanasi, Uttar Pradesh, India. [2]Junior Resident, Department of General Medicine, Heritage Institute of Medical Sciences, Varanasi, Uttar Pradesh, India. [3]Professor, Department of Radiology, Heritage Institute of Medical Sciences, Varanasi, Uttar Pradesh, India. [4]Junior Resident, Department of Radiology, Heritage Institute of Medical Sciences, Varanasi, Uttar Pradesh, India

Abstract

Background: A neurologist treating a case of pain abdomen is quite a surprise to many!! But scarcely, it may happen like that. It is because the disease porphyria can present to the Department of Neurology with neuropathy and can at times present with recurrent pain abdomen as well. In such cases, neurogenic inflammation leads to pain abdomen. This pain in the abdomen can be misdiagnosed initially by clinicians and treated symptomatically. However, it is of prime importance to diagnose this entity early because, with each bout of attack, neuropathy progresses. Again, certain drugs must be avoided as they can precipitate acute attacks. We present here a case of a young lady with recurrent pain abdomen who was treated by multiple gastroenterologists symptomatically and finally turned out to be a case of porphyria.

Keywords: Neurogenic Pain Abdomen, Porphyria, Recurrent Pain Abdomen.

Received: 28 July 2025 **Revised: 02 August 2025** **Accepted: 20 August 2025** **Published: 05 January 2026**

Introduction

Porphyrias are a group of inherited disorders where the enzymatic defects in heme biosynthesis lead to the accumulation of intermediate metabolites.[1] Acute intermittent porphyria (AIP) is the most severe form. Females are most affected. Symptoms are aggravated during ovulation, menstruation, and pregnancy due to changes in sex hormone levels.[2] Patients can present to gastroenterologists with abdominal symptoms like recurrent poorly localized pain abdomen, to gynecologists with symptoms of pain abdomen during the menstrual cycle, to general physicians with nonspecific fever with no source of infections, to psychiatrists with features of anxiety, and neurologists with features of neuropathy with proximal predominant weakness.[3] However, these patients can present with a multitude of symptoms in a single acute attack as well. We present here a case of a porphyric young lady who presented to the Department of Neurology with features mimicking Guillain-Barre syndrome and excruciating pain in the abdomen.

CASE REPORT

An 18-year-old young lady presented to our hospital with a history of weakness in all four limbs for 4days. Weakness was initially noticed by the patient in her lower limbs as she was unable to get up from the squatting position (in the washroom) and she needed help from the wall and floor to stand. It progressed rapidly and the next day she was unable to hold her slippers tightly and the slippers slipped out of her toes quite often while walking. The very same day she found that she was unable to raise her arm to comb her hair in the afternoon and she felt as if she was too tired to do that. The next morning while waking up from bed she noticed that she was unable to turn on her bed and could not change her posture from supine to sitting. She called for help for the same. At this point, she was taken to a local hospital for treatment. After some basic investigations and initial treatment, she was referred to our hospital.

At the presentation, the patient was conscious and cooperative. Her vital parameters were normal with a blood pressure of 114/76mm of Hg, pulse rate of 88/minute, and saturation of Oxygen of 96%. Her higher mental function and cranial nerve examination were within normal limits. On motor system examination, proximal power was more affected than distal. Hyporeflexia was present. The bilateral plantar reflex was mute. The sensory system was intact. So, a provisional diagnosis of Guillain-Barre syndrome was made. A nerve conduction study was advised which showed motor axonal polyneuropathy involving both upper and lower limbs. So, after confirmation, intravenous immunoglobulin (IVIG) was started at a dose of 2 grams/kg in a divided dose over 5 days. On the 8th day of her illness, we performed a cerebrospinal fluid analysis, but it did not

Address for correspondence: Dr. Sahoo Haramohan, Assistant Professor, Department of Neurology, Heritage Institute of Medical Sciences, Varanasi, Uttar Pradesh, India
E-mail: _subham.sahoo125@gmail.com

DOI:
10.21276/amit.2026.v13.i1.287

How to cite this article: Haramohan S, Krishna C, Vikas V, Gaurav S, Bhupesh G. Pain Abdomen: Journey of a Young Lady from a Gastroenterologist to a Neurologist. Acta Med Int. 2026;13(1):15-17.

show any albumin cytological dissociation. However, the patient did not deteriorate further and was put on physiotherapy.

On the day of her discharge, the patient complained of excruciating pain abdomen which was poorly localized. On examination, she had no specific site of tenderness. The same day she started menstruating as well. So, initially, she was given mefenamic acid with dicyclomine hydrochloride. But she did not respond. A gynecology reference was taken and an ultrasound abdomen with pelvis was performed. It was normal. She became quite anxious and irritable. So, we sought the help of a Gastroenterologist who advised serum amylase and lipase and upper gastrointestinal (UGI) endoscopy. All were within normal limits. Computed tomography angiography of the abdomen was advised. It was also within normal limits [Figure 1].

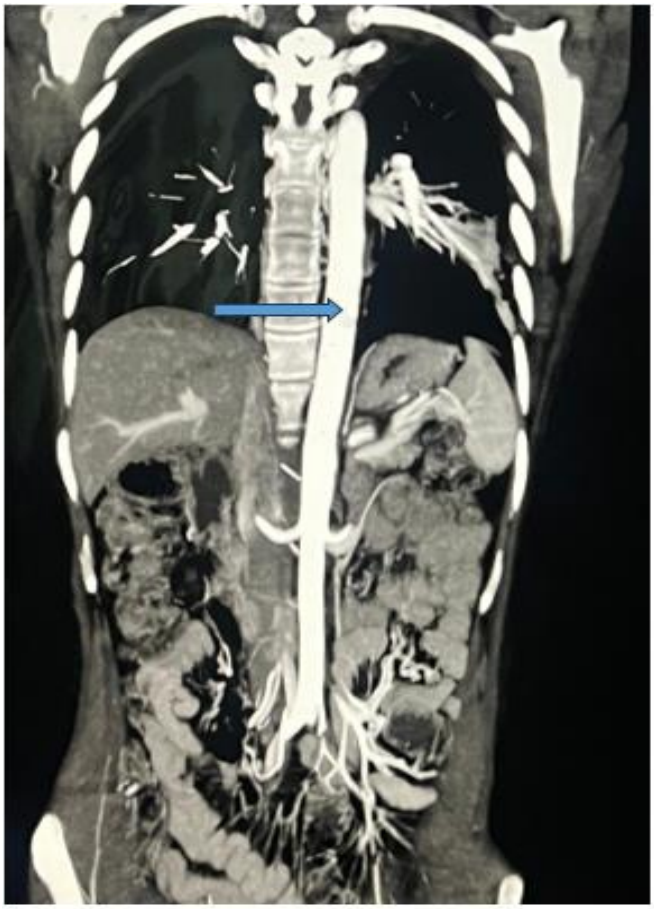

Figure 1: Computed tomography Angiography of the abdomen showing the descending aorta (arrow) and its branches which are within normal limits.

So, we decided to dig into her history of past illness. His attendants said that she had a recurrent history of self-limiting pain in the abdomen for the last 6 years (once or twice/year). However, the frequency and severity of symptoms have increased for the last 2 years (almost 8 episodes in 2years). She had been investigated for the same. She had undergone both upper and lower GI endoscopy twice before and every time it was within normal limits. With such a history of recurrent pain abdomen and motor axonal neuropathy with predominant proximal weakness, we decided to rule out porphyria. We sent urine porphobilinogen (PBG). It was positive. The 24-hour urinary PBG was 172mg/24 hours (normal value is 0-4mg/24 hours). So, a final diagnosis of acute intermittent porphyria (AIP) was made and she was treated for the same. With dextrose infusion along with narcotic analgesics, the pain in the abdomen subsided. At 3-month follow-up, the patient was doing fine, and her proximal weakness had improved.

Discussion

Porphyrias are a group of pan-ethnic metabolic disorders resulting from a defect in the activity of a specific enzyme in the heme biosynthesis.[4] They are of two types: Hepatic porphyrias and erythrocytic porphyrias. Inheritance can be autosomal dominant, autosomal recessive, or X-linked. AIP is the most severe form. Females are most affected and there is aggravation of symptoms during ovulation, menstruation, and pregnancy due to changes in sex hormone levels.[2] These patients can present to any department due to varied manifestations. The most common symptoms are abdominal pain (55%), tachycardia (80%), dark urine (75%), peripheral motor deficit (70%), and seizures (20%).[3] Porphyria can affect autonomic, central, and peripheral nervous tissue. Peripheral neuropathy affects motor neurons with degeneration of axons.[5] Patients present with proximal muscle weakness.

In our case patient had multiple episodes of abdominal pain but despite multiple visits to general physician and gastroenterologists, it could not be diagnosed. UGIE and LGIE were performed twice which were normal. So, she was given symptomatic treatment. Every time the pain in the abdomen resolved spontaneously. When she presented to our Department of Neurology she was having weakness of all four limbs. Initially, the weakness was in the lower limbs and rapidly spread to the upper limbs. Weakness was predominantly proximal. There was no sensory abnormality. The nerve conduction study was suggestive of axonal neuropathy. So, initially patient was treated in the line of GBS. However, after 7 days of disease onset when the CSF study was done, it did not show features of albumin-cytological dissociation. So, a hint of doubt was cast over the diagnosis.

When the patient was about to get discharged, she developed pain abdomen. This time pain abdomen was excruciating but not localized to any specific region. Every possible investigation was done to rule out differentials of pain abdomen. However, all the investigations were within normal limits. So, it was clear that the patient was not a conventional case. When we sum up the whole narrative of the patient from proximal weakness with axonal neuropathy to recurrent pain abdomen with aggravation during the menstrual cycle, we could think of porphyria as one of the differentials. That is why we sent urine for quantitative estimation of porphobilinogen, which came to be quite higher than the normal range. Thus, the patient was treated in the line of porphyria. Acute phase subsided.

After 3 months of follow up patient had recovered significantly. The power of the upper and lower limbs had improved. The patient had mild difficulty in getting up from sitting. However, she was able to perform the rest of her day-to-day activities.

Conclusion

Porphyria is a great masquerader. It can mimic any disease as its symptoms are so nonspecific. Patients with porphyria can present to any department with a variety of symptoms. So, treating physicians must be aware of the symptoms and always keep it as a possible differential diagnosis.

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

References

1. Mutluay B, Köksal A, Çelık RGG, Bülbül HH. A Case of Acute Intermittent Porphyria Mimicking Guillain-Barré Syndrome. Noro Psikiyatr Ars. 2019 Oct 15;56(4):311-312.
2. Vassiliou D, Lindén Hirschberg A, Sardh E. Treatment with assisted reproduction technologies in women with acute hepatic porphyria. Acta Obstet Gynecol Scand. 2021 Sep;100(9):1712-1721
3. Bissell DM, Anderson KE, Bonkovsky HL. Porphyria. N Engl J Med. 2017 Aug 31;377(9):862-872.
4. Phillips JD. Heme biosynthesis and the porphyrias. Mol Genet Metab. 2019 Nov;128(3):164-177.
5. Bonkowsky HL, Schady W. Neurologic manifestations of acute porphyria. Semin Liver Dis. 1982 May;2(2):108-24.