Acute Intermittent Porphyria: A Frequently Misdiagnosed Chameleon!

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ABSTRACT

Acute intermittent porphyria (AIP) is an inborn disorder of heme biosynthesis, autosomal dominant in inheritance. It is a frequent occurrence in young females of reproductive age group. While abdominal pain is the most frequent presentation of this disorder, it can present with a myriad of clinical and biochemical features, frequently leading to misdiagnosis of this condition. We present a case of a 17-year-old young female who presented with an acute onset weakness in all four limbs along with absent deep tendon reflexes but characteristically preserved ankle jerks, who was initially diagnosed as Guillain-Barré syndrome (GBS), treated with intravenous immunoglobulin (IVIg), succumbed to a chronic progressive course of weakness and put on oral steroids. Lack of improvement and subsequent development of abdominal pain led us to investigate her for urine for porphobilinogen which came out to be positive, thus leading to a final diagnosis of acute intermittent porphyria.

Keywords: Acute intermittent porphyria, abdominal pain, Guillain-Barré syndrome

Porphyrias are a group of relatively uncommon metabolic disorders produced by defective biosynthesis of heme. There are broadly two categories, i.e., hepatic and erythroid and clinically they can be classified as neurovisceral, cutaneous or mixed. Acute intermittent porphyria (AIP) is the most common of all and results from partial deficiency of porphobilinogen deaminase enzyme. Being an easily missed entity, it should be looked for with high index of suspicion in any patient presenting with acute onset weakness and abdominal pain. There have been case reports on misdiagnosis of AIP mostly as Guillain-Barré syndrome (GBS) due to acute presentation of the disease. We report a case here with acute presentation of weakness of all four limbs, subsequently attaining a progressive form of weakness and wasting, mimicking chronic inflammatory demyelinating polyneuropathy (CIDP). Based on our literature search, this transition from acute to chronic phase in AIP has not been described before.

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CASE REPORT

A 17-year-old young female presented with history of subacute onset weakness of all four limbs in the form of difficulty in carrying out overhead activities and performing fine activities, along with difficulty in rising from sitting position, for the last 4 months. There was no associated sensory complaint, difficulty swallowing, bowel or bladder involvement. She was treated 4 months back as a case of acute motor axonal neuropathy (AMAN) variant of GBS with intravenous immunoglobulin (IVIg) on the basis of her neurophysiological study, which revealed pure motor axonal affection of the tested nerves. She developed acute abdominal pain during hospital stay along with vomiting and was treated as a case of acute cholecystitis. After 15 days, as no significant improvement was found, she was subjected to nerve biopsy and started on oral corticosteroid treatment thinking of CIDP. She had minimal improvement with steroids; her nerve biopsy report was inconclusive and after 4 months, she presented to our institute with residual and static weakness. On asking about her family, she revealed that her younger sister suffered with fever, abdominal pain and seizures last year, which lasted for a month, followed by her sad demise.

On examination, the patient was tachypneic and had resting tachycardia. On neurological examination, there was wasting of posterior fibers of deltoid; both anterior and posterior compartments of arms and

Figure 1. Bilateral wrist drop.

Figure 2. Urine sample turned cola-colored on exposure to sunlight.

forearms bilaterally; interossei, chiefly the first dorsal interosseous; anteromedial compartment of thighs and calf muscles. Generalized hypotonia was present along with bilateral wrist drop (Fig. 1). Power was 4/5 in upper limbs at shoulder and elbow joints, 0/5 at dorsiflexors of wrists, 4/5 in lower limbs at hip and knee joints, 5/5 at ankle bilaterally. Deep tendon reflexes were absent, except ankle jerk which was 2+ bilaterally. Sensory and cerebellar examination was unremarkable. Her urine sample was sent for porphobilinogen and a sample was also kept in sunlight to see for change in its color (Fig. 2) considering the past history of acute abdominal pain, vomiting, neuropathy along with suspected positive family history. The report came out to be positive and patient was advised high carbohydrate diet and avoidance of all the drugs that precipitate porphyria. Thus, after a great diagnostic odyssey, the patient was finally labeled as AIP and advised high carbohydrate diet. On follow-up after 2 months, the patient has shown marked improvement in her functional status.

DISCUSSION

Porphyrias are heme biosynthetic disorders leading to accumulation of toxic porphyrin precursors and porphyrin itself, the excess of which accumulates in various tissues giving rise to a myriad of clinical features. There are eight main varieties of hepatic and erythroid porphyrias, amongst which AIP is the most common. It is caused by the deficiency of porphobilinogen deaminase leading to excessive accumulation and urinary excretion of porphobilinogen. AIP is most prevalent in young females of reproductive age group and crises mostly occur after puberty. This disease is manifested by acute gastrointestinal manifestations like abdominal pain, nausea, vomiting, constipation; neurological manifestations like neuropathy involving both motor and sensory nerves, psychiatric symptoms, seizures; cardiovascular manifestations like arrhythmias and autonomic disturbances.

The symptoms can range from acute crisis to chronic progressive neurological weakness, thus making it difficult to be diagnosed timely. AIP can mimic many other illnesses like in our case, the patient was initially thought to have GBS with co-existent cholecystitis. Subsequently, when she attained a chronic progressive course of weakness, she was treated as CIDP but all in vain. Misdiagnosis of GBS in a case of porphyria has been reported previously, highlighting the fact that muscular weakness progressing to quadriparesis can mimic GBS in a case of porphyria.

CONCLUSION

This case establishes the fact that AIP can be a great masquerader and thus easily misdiagnosed in clinical settings. Thus, a high index of suspicion is required when confronted with a blend of gastrointestinal and neurological manifestations in a patient in order to prevent a delayed diagnosis and grave outcomes.

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