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A Case of Acute Intermittent Porphyria Mimicking Guillain-Barré Syndrome

Guiilain-Barre Sendromunu Taklit Eden Bir Akut İntermitant Porfiri Olgusu

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ABSTRACT

Patients with acute intermittent porphyria attacks present with severe abdominal pain, neuropathy and psychiatric disturbances. Porphyric neuropathy mostly causes confusion in clinical practice, and patients with porphyria are rarely correctly diagnosed early in the course of the illness. We report a patient with acute intermittent porphyria mimicking Guillain-Barré syndrome with acute onset weakness that rapidly progressed to severe quadriplegia.

Keywords: Guillain-Barré syndrome; porphyria; porphyric neuropathy

ÖZ

Akut aralıklı porfiri atakları olan hastalar şiddetli karın ağrısı, nöropati ve psikiyatrik rahatsızlıklar ile başvururlar. Porfirik nöropati, klinik uygulamada çoğunlukla kafa karışıklığına neden olur ve porfiri olan hastalar hastalığın seyrinde erken dönemde ender olarak doğru teşhis edilir. Bu yazıda, şiddetli kuadriplejiye hızla ilerleyen ve akut başlangıçlı zayıflık gösteren Guillain-Barré sendromunu taklit eden, akut aralıklı porfiri olan bir hastayı sunuyoruz.

Anahtar Kelimeler: Guillain-Barré sendromu; porfiri; porfirik nöropati

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INTRODUCTION

Porphyrias are an inherited group of disorders related to enzyme deficiencies in the heme biosynthesis. Autosomal dominant inherited forms are acute intermittent porphyria (AIP), variegate porphyria and hereditary coproporphyria, while d-aminolevulinic acid dehydratase deficiency is an autosomal recessive inherited condition (1, 2). Patients with AIP in addition to abdominal pain and neuropsychiatric disturbances, electrolyte imbalances, seizures, confusional states, autonomic changes may accompany to attacks, and worsen the clinical state (1, 3). Porphyric neuropathy may cause substantial confusion and diagnostic challenge in clinical practice (4-6). We present a case with acute onset abdominal pain accompanied by weakness that rapidly progresses to severe quadriplegia with facial diplegia and respiratory failure, and being misdiagnosed as Guillain-Barré syndrome.

CASE REPORT

A 17-year-old female patient presented with weakness of the arms and legs which was told to have rapidly progressed in four days. It was reported that she had been admitted to another hospital emergency room with acute onset abdominal pain which led up to a normal routine work-up a week ago. On admission to our hospital, she was alert, and cooperative with intact cranial nerves. Weakness did not differ in proximal and distal muscles (Medical research council grade 3/5). Deep tendon reflexes were absent except for bilateral triceps reflexes; plantar reflexes were normal; no sensory deficit or sphincter dysfunction was detected; no skin lesions were present. Her electrophysiological investigation was normal for motor and sensory nerve conduction studies, and presented only mild reduction in interference pattern during needle electromyography (EMG). An acute motor axonal neuropathy variant of Guillain-Barré syndrome was suspected, and immunoglobulin therapy was started at 0.4 gr/kg/day dose. However, at the following day of her admission respiratory distress evolved rapidly, and she was given thiopental and intubated. She had hypertension and tachycardia resistant to treatment. Biochemical work-up showed hyponatremia (127 mmol/L). Cerebrospinal fluid was clear, as the biochemistry screen and microscopy showed. All of the tests performed for vasculitis, and lead intoxication were normal. Her urine was reddish-brown in color, and was darkened by daylight (Fig. 1). Porphyria was suspected and thiopental was immediately switched to propofol. At the end of the first week of admission, facial diplegia, restriction of eye movements in four directions, and quadriplegia developed. A second EMG was performed on the second week of admission with findings of pronounced decrease in motor muscle action potentials, and preserved sensory nerve conduction studies, revealing an acute motor axonal neuropathy. Later on, family members recalled that she had been evaluated 10 months ago as inpatient for medication-resistant abdominal pains and a first-time epileptic seizure without any findings in routine biochemistry, abdominal ultrasonography, and abdominal and cranial CT. Her gynecologic examination was also normal. At that time, her colonoscopy findings were suspicious for Crohn disease, showing some ulcerative lesions at the terminal ileum and colon. Moreover, we learned that one of her cousins had been diagnosed porphyria ten years ago.

Due to the fact that our hospital is only specialized in neurology, neurosurgery, and psychiatry; work-up of some rare laboratory tests, including porphyria screenings, had to be done in other facilities lacking

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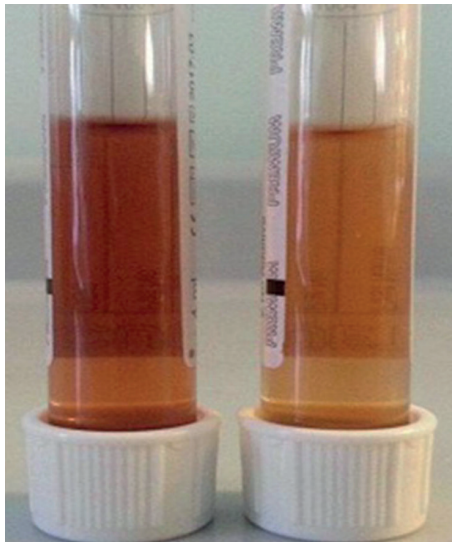

Figure 1. Urine sample showing reddish-brown color after exposure to daylight.

immediate results. In the meantime, treatment for suspicious porphyria was started with high glucose regime (400 gr/day). Investigations for porphyria revealed a urinary porphobilinogen (PBG) level of 145 µmol/L (reference value <µmol/L per hour), total urinary porphyrin of 3769 nmol/L (reference value<100 nmol/L). Haem arginate treatment (150 mg/day) could only be administered lately in the course, due to difficulties in obtaining this medication. However, her neurological status did not improve. She was quadriplegic and on ventilator support, and died from sepsis one month after admission.

DISCUSSION

Although AIP has been defined thoroughly, its prevalence is estimated as 1/75000, and 10–40% of the patients develop neuropathy (2, 3, 7). Porphyric neuropathy is manifested by symptoms and cerebrospinal fluid abnormalities resembling acute Guillain-Barré syndrome (3, 7). However, accompanying psychological features, a proximal predilection of asymmetric weakness, and electrodiagnostic findings indicative of an axonal polyradiculopathy and autonomic neuropathy suggest the diagnosis of porphyria (3). Absence of abdominal pain does not exclude the possibility of porphyria (7). Although our patient's weakness was not prominent in proximal muscles, she had hypertension and tachycardia due to autonomic impairment, hyponatremia, and axonal neuropathy which are all consistent with AIP. The mechanism for neurotoxicity has not been elucidated, but it has been suggested that toxic accumulation of heme synthase over-products in nerves start the neuropathy. Gene mutations leading to AIP are well known; however, not all family members with mutations may develop porphyria attacks (1, 3).

Diagnosis depends on detection of urinary PBG and its increase, as well as porphyrin, and aminolevulonic acid levels (1, 2, 4). Spot urine darkens to reddish or brown color when exposed to sunlight. First line in the immediate treatment of an acute attack is to remove precipitants like newly added medications (8). The trigger leading to an attack in most instances may be an inappropriate medication, low calorie diet, or infection. AIP attacks are also provoked by fasting, alcohol use, and hormonal changes (1). Our patient presented formerly to another hospital where she may have been treated for abdominal pain, although we do not know which medications were given. After this first induction, sedation with thiopental may have worsened the medical condition. Afterwards, haem arginate was not successful in reversing the attack due to delayed diagnosis and late onset due to difficulties in obtaining the medication. As in our hospital, Schutte et al. also stated that obtaining haem arginate in a state hospital setting was a challenge (7).

A patient with a confirmed diagnosis of porphyria presenting with acute quadriplegia accompanied by abdominal pain would strongly suggest porphyric neuropathy. But with an unknown porphyria diagnosis, all of the differentials including porphyria, lead and other intoxications and GBS are unequivocally possible especially in the acute phase. These entities may be indistinguishable clinically and electrophysiologically from each other, and the diagnosis may be quite challenging even for a highly experienced neurologist until the diagnostic work-up is completed as in our case report.

We conclude that patients with unconfirmed diagnosis of AIP may be misdiagnosed as Guillain-Barré syndrome, and treatment delay and inappropriate medications may worsen prognosis, and cause severe outcomes, for these reasons clinicians should have a high index of suspicion of AIP in the differential diagnosis of acute neuropathies.

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