Journal Of Clinical Neurology, Neurosurgery And Spine

SCIENTIFIC LITERATURE

Special Issue Article "Convulsions Causes and Treatment" Case Report

Repeated Misdiagnosis of Acute Intermittent Porphyria as Neurological Disorders: Learn from One Chinese Case

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ARTICLE INFO

Received Date: July 22, 2022
Accepted Date: August 24, 2022
Published Date: August 25, 2022

KEYWORDS

Acute intermittent porphyria
Guillain-Barré syndrome
Abdominal pain
Neuropsychiatric manifestations
Non-convulsive epileptic seizures

Citation for this article: Nian Yu, Feng Li, Xing-jian Lin, and Qing Di. Repeated Misdiagnosis of Acute Intermittent Porphyria as Neurological Disorders: Learn from One Chinese Case. Journal Of Clinical Neurology, Neurosurgery And Spine. 2022; 4(2):129

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ABSTRACT

Acute Intermittent Porphyria (AIP) is an autosomal dominant disorder due to the gene mutations in Porphobilinogen Deaminase (PBG), also called hydroxymethylbilane synthase. This enzymatic deficiency leads to the marked accumulation of the porphyrin precursors, aminolevulinic acid and PBG in the body. The porphyrin precursors could act on the target tissues through the systemic circulation and consequently produce a series of clinical syndromes, including intermittent paroxysmal abdominal pain and neuropsychiatric manifestations. Reportedly, AIP could exhibit various nonspecific symptoms involving many systems. Hence it is easy to escape correct diagnosis in clinical practice. Here, we provided a case of AIP ever misdiagnosed as acute abdomen, depression, non-convulsive epileptic seizures and Guillain-Barré syndrome successively.

INTRODUCTION

Acute Intermittent Porphyria (AIP) may have various nonspecific symptoms, including intermittent paroxysmal abdominal pain and neuropsychiatric manifestations. It is easy to escape correct diagnosis or misdiagnose them in clinical practice. Here, we provided a case of AIP ever misdiagnosed as acute abdomen, depression, non-convulsive epileptic seizures and Guillain-Barré syndrome (GBS) in successive years.

CASE REPORT

A 25-year-old female was admitted with recurrent abdominal pain for 21 months and limbic weakness for 10 months on July 28th 2017. From the mid-Dec 2014, she initially manifested with paroxysmal upper abdominal pain without regular intervals. She had undergone a surgery due to the markedly gaseous bowel distention revealed by an abdominal computed tomography scan at that time. However, the surgical approach did not provide obvious remission for her. In the following three years, she had also been considered as partial intestinal obstruction, chronic gastritis, irritable bowel syndrome, non-convulsive epileptic seizures and psychogenic non-epileptic seizures separately in different hospitals. From Oct 16th 2016, she presented with rapidly progressive symmetric weakness affecting remote muscles of the legs and arms. Subsequently the nerve conduction studies showed acute motor axonal neuropathy on her bilateral upper limb. Although the result of her cerebrospinal fluid was normal at that time, she was diagnosed as GBS because of no other obvious reasons found for her symmetric motor paralysis. This episode caused her severe pneumonia, for which she had to be transferred to an Intensive Care Unit (ICU). After 5 months at ICU, the power of her limbs was gradually improved, but the numbness and weakness in her distal limbs remained in existence. She had no obstetric history

and denied any similar family history. Results of physical examination in our department was as following: her skin showed no rashes and also no yellowness; her muscle power was grade 4/5 in upper limbs and in lower limbs with predominantly extremity weakness; her tendon reflexes were absent symmetrically; the hyperalgesia of her lower limbs presented in stocking distribution. There were no abnormal found in her brain MRI/MRA and sphenoidal electroencephalogram monitoring. But we found that her urine turned burgundy after exposure to sunlight (Figure 1.1). So combined with all her previous symptoms, she was suspected to suffered from a rare disease of AIP. This diagnosis was confirmed by her genetic analysis which showed Hydroxymethylbilane Synthase (HMBS) gene was heterozygous (Figure 1.2). The written informed consent on this case report for study was obtained from this patient. This study was also approved by the medical ethics committee of the Nanjing Brain Hospital affiliated to Nanjing Medical University.

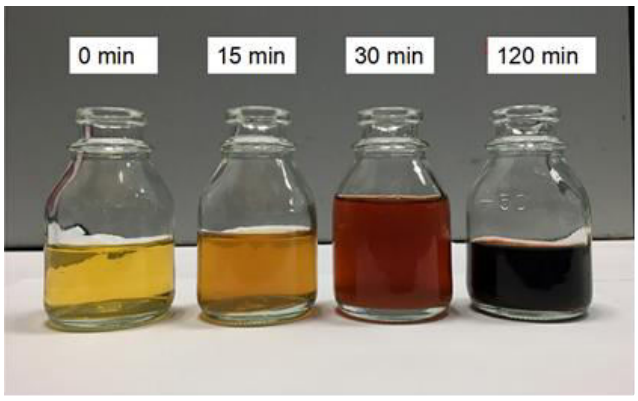

Figure 1.1: The color changes of her urine after exposure to sunshine. Initially, her urine color was pale yellow. With the extension of exposure time, her urine gradually turned dark red or burgundy.

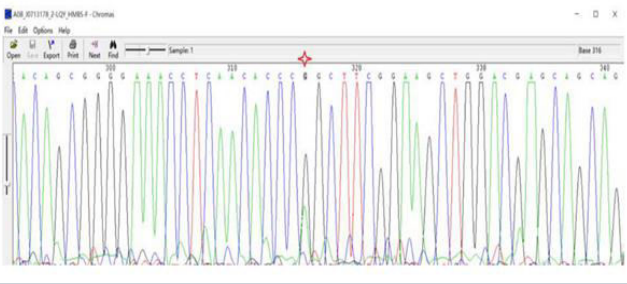

Figure 1.2: The results of her generation sequencing. Her gene test showed a heterozygous mutation in HMBS gene (G>GA), markedly by ✦. This genetic damage was considered as a cause of the AIP disease.

DISCUSSION

AIP is an autosomal dominant disorder due to the gene mutations in HMBS, which deficiency could lead to the accumulation of toxic PBG in the body [1]. The produced porphyrin precursors could do harm on various organs through the systematic circulation. According to the differences of acting sites, porphyrias are divided into erythropoietic form and hepatic form. Reportedly, AIP consisted of various nonspecific symptoms involving many systems, which was the most common type of hepatic porphyrias [2]. Here, the present case had ever been misdiagnosed as acute abdomen, depression and GBS, respectively. Indeed, it was difficult to make prompt correct diagnosis at the early stage of AIP. The predominant manifestation of AIP was acute intermittent attacks of gastrointestinal with or without neurological dysfunctions [3]. Reportedly, abdominal pain may occur in 85-95% of AIP patients [4]. Manifestations of AIP involving nervous system also showed various types, of which acute symmetric peripheral neuropathy was relatively a typical symptom and may occur in 80% of AIP patients [5]. Moreover, cranial nerve palsy, autonomic dysfunction, mental symptoms and seizures may also occur in AIP. Both PBG and uroporphyrins could be elevated in the urinary of AIP sensitively. Urinary PBG can turn into red fluorescent uroporphyrin and porphobilin after exposure to ultraviolet light, which may provide a specific clue for the diagnosis of AIP.

CONCLUSION

Therefore, AIP should be considered highly speculative among the patients exhibiting unexplained abdominal pain together with neuropathy. Further evaluation for its diagnosis should include the urinary PBG and its gene screening.

SUPPORT

This work has been supported by the two grants, one from the National Natural Science Foundation of China (Grant No. 81400981), and the other one from Nanjing Medical Science and Technology Development Foundation (QRX17179&YKK21116). The informed consent was obtained for experimentation with this patient.

CONFLICTING INTEREST

Nil

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