

CASE REPORT

PORPHYRIA: THE DISEASE THAT CALLS FOR AWARENESS

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ABSTRACT: Porphyria is a relatively uncommon condition and it should be considered in patients presenting with atypical medical, psychiatric, or surgical history. Paucity of clinical recognition of porphyria often delays diagnosis and consequently treatment. Also, signs and symptoms mimic other common medical and surgical conditions. So, awareness of porphyria along with proper and accurate diagnostic tests in conjunction could be of great boon to surpass the formidable challenges put forth by porphyric patients. Here, we present a case of 24 year old woman who suffers from porphyria and ultimately dies because of inappropriateness in diagnosis on the part of physicians which is due to lack of awareness and strong clinically suspicious eye.

KEYWORDS: Porphyria, Acute intermittent porphyria, porphobilinogen, Ehrlich.

INTRODUCTION: Porphyrias are a group of inherited disorders and are caused due to defective enzymes in heme biosynthetic pathway.⁽¹⁾ There are seven main types of porphyria which are broadly classified according to clinical features into neuropsychiatric, dermatological, and mixed forms.⁽²⁾ The most common of these is acute intermittent porphyria (AIP), which is dominantly inherited and characterized by the half-normal activity of porphobilinogen deaminase (PBGD) (EC 4.3.1.8).⁽³⁾

The usual clinical presentation involves abdominal pain, gastrointestinal symptoms and autonomic nervous system disturbances.⁽⁴⁾ These attacks are precipitated by endogenous factors such as drugs, hormones (i.e. estrogens), and emotional stress, pregnancy or starvation states.⁽⁵⁾

These symptoms of AIP mimic other medical and surgical illnesses, framing a diagnostic dilemma in physician's mind which consequently leads to misdiagnosis, wrong diagnosis or delay in diagnosis. This inappropriateness in patient care is preventable by high index of suspicion and awareness in physician's mind aided by biochemical diagnostics.

So we present here a case of AIP with usual symptoms but still the uneventful outcome of death was unpreventable due to delay in diagnosis.

CASE REPORT: A 26 year old G2P1 from a rural poor socio-economic class family arrived in the emergency department of hospital with complaints of fever, swelling of abdomen with severe diffuse colicky pain lasting for small duration. This was also associated with nausea and several episodes of vomiting. She also developed psychiatric symptoms in the form of suicidal thoughts, restlessness and sleeplessness. She gave a history of amenorrhea of 2 months. The patient was a nonsmoker and nonalcoholic.

On examination-patient was conscious and irritable, showed signs of anemia. Abdominal examination revealed tenderness in right lumbar and periumbilical area. Systemic examination was normal. On Mini-mental status examination, the patient was confirmed to be depressed with Hamilton Depressive Score 27.

On biochemical investigation- Hb was 9.6 gm%. WBC, RBC and platelet counts were normal. Electrolytes were normal except hyponatremia. Liver function test and kidney function tests showed no abnormalities. Anemia and hyponatremia were present and corrected by adequate treatment.

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Patient was operated for appendicectomy, which was followed by 3 episodes of generalized tonic-clonic convulsions on post op day 10. Then she developed pain in the back and limbs followed by quadriparesis on post op day 20. During the parietic attack, patient immediately became unconscious and was put on ventilatory support. At this stage, porphyria was suspected and urine was collected for biochemical investigation of porphyria where porphobilinogen was found to be positive in Ehrlich's test. Also, the urine turned dark brown on sunlight exposure. For confirmatory diagnosis, porphobilinogen deaminase (PBGD) test was done which was found to be positive.

Intervention began in the form of intravenous fluids and high dose dextrose along with general supportive measures, but in spite of meticulous treatment, there was no clinical outcome and patient succumbed to death.

DISCUSSION: Of all types of porphyria, acute intermittent porphyria is the most common type.⁽⁶⁾ Females usually have more severe symptoms and attacks may be precipitated by menstruation, pregnancy, and oral contraceptive use.⁽⁷⁾ Symptoms may vary considerably in the same patient during different episodes, as well as among patients with AIP. Because the clinical course can vary from acute, self-limiting attacks to attacks that result in chronic or progressive deficits, the attacks may mimic many other psychiatric or medical disorders, making the potential for misdiagnosis great.⁽⁸⁾

In our case, patient's porphyric attack was precipitated due to hormonal changes in the form of pregnancy but this fact which could have been an eye-catcher remained neglected.

Patient's psychiatric manifestations were treated by a psychiatrist. Patient also presented with the most common porphyric manifestation i.e. abdominal pain which again remained misdiagnosed. Infact, exploratory laparotomy was planned and appendicectomy was performed. This was followed by neurological manifestations in the form of neuropathy. Again patient was wrongly treated for neuropathic pain and porphyria was never suspected. However, Ehrlich's test was performed and AIP confirmation was done by test for PBGD.

CONCLUSION: AIP must be included in the differential diagnosis of psychiatric, neurological and/or gastric alterations whether family history is present or not. High clinical suspicion and performing simple tests in the form of Ehrlich's test can be of vital in suspecting/labeling the patient as AIP and thus preventing the fatal outcome of the disease due to lack of clinical recognition and delay in diagnosis.

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