



CASE REPORT

Reversible Posterior Encephalopathy Syndrome in Patient with Acute Intermittent Porphyrin: A Case Report

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Abstract

Acute Intermittent Porphyria (AIP) is a metabolic disease caused by an inherited deficiency of heme biosynthesis. Acute attacks of AIP during pregnancy are very rare. However, it presents the obstetrician with challenging problems. Prompt diagnosis and early detection of the disease are crucial for the appropriate treatment and favorable prognosis. All pregnant patients who complained of unexplained abdominal pain should be considered the diagnosis of acute porphyria. This paper presents a patient with pregnancy complicated by AIP and Posterior Reversible Encephalopathy Syndrome (PRES), who had a good perinatal outcome.

Keywords

Acute intermittent porphyrin, Reversible posterior encephalopathy syndrome, Pregnant

Introduction

Acute Intermittent Porphyria (AIP) is a metabolic disease caused by an inherited deficiency of heme biosynthesis. Typical symptoms of the disease present as unpredictable attacks of acute pain, gastrointestinal symptoms, and neurological system disturbances. Acute attack of AIP can be triggered by pregnancy. However, acute attacks during pregnancy are very rare, if an AIP asymptomatic carrier had already diagnosed before the pregnancy, compared to an initial diagnosis of AIP in a "de novo" patient [1]. At present, AIP is considered a new cause of PRES. Posterior Reversible Encephalopathy Syndrome (PRES) is a clinico-radiological disorder characterized by headache, conscious disturbance, seizure and visual loss [2]. Here, we describe a woman who suffered the first attack of AIP during pregnancy and presented with PRES after cesarean section.

Case Report

A 24-year-old woman had been in good physical condition previously (with no symptoms of porphyria). However, she had a family history of acute porphyria. The woman presented with acute abdominal pain at the 35th day of pregnancy without obvious inducement (she didn't know conception at that time) and admitted to the Digestive Department of our Hospital. No nausea, vomiting, diarrhea or vaginal bleeding was reported. Physical examination showed generalized abdominal pain without rebound tenderness and muscle guarding. Endoscopic examination revealed fungal esophagitis and Chronic Non-Atrophic Gastritis. Treated with Omeprazole, carbohydrate support and fluid balance, the abdominal pain regressed. Her urine color changed to red when exposed to light. Based on her family history and the change of urine color, the diagnosis of acute porphyria was considered and then confirmed by urine analysis, which showed very high levels of porphobilinogen (urinary porphobilinogen: positive). She had several attacks of AIP in the later period of pregnancy and was cured by Normosang (haem arginate; 3 mg/kg bodyweight/day on four consecutive days), high-dose dextrose and saline infusion.

In the 38 + 3 weeks of gestation, she admitted to our Department due to uterine contractions and labor pain. An emergency cesarean section was performed for cephalo-pelvic disproportion. A live male infant weighing 2100 g was delivered. Apgar score was 10. Sudden-onset bilateral visual disturbance and seizure accompanied by headache and dizzy occurred on the

second postoperative day. Her blood pressure was 170/110 mmHg, and heart rate was 110 beats/min with regular heart sounds at that time. Magnetic Resonance Imaging (MRI) of the brain demonstrated hyperintense gyriform lesions on T₂-weighted images and hypointense to isointense lesions on T₁-weighted images in both parieto-occipital lobes with mild contrast enhancement. The diagnosis of PRES was made according to the clinical features and MRI findings. The patient was treated with mannitol and normosang, and strict blood pressure control therapy immediately. Two days after treatment, her blood pressure returned to normal, headache and dizzy disappeared. Her level of visual acuity began to improve, and fully recovered at the 6th day of treatment. A repeat MRI performed 2 weeks after the treatment confirmed complete disappearance of the lesions.

Discussion

There are seven different types of porphyrias, all defined by the enzyme affected or the symptoms produced by the disease [3]. AIP is the most common type of acute porphyria, due to a partial deficiency of the third enzyme in the haem biosynthesis pathway. Activation of the disease is related to various factors such as diet, stress, alcohol, starvation, drugs, surgery, menstruation, and steroid hormones. Pregnancy may act as a precipitating factor for acute attack of porphyria, because of the increased steroid hormones levels. Starvation due to hyperemesis gravidarum, fasting pre and post-operative, cesarean section, anesthesia and drugs can also lead to an acute attack. Thus, both early pregnancy and puerperium are periods of maximal risk for developing an attack of acute porphyria.

AIP is particularly difficult to identify during pregnancy, because abdominal pain is more likely to be of obstetric origin. The diagnosis of this disease is multidisciplinary, where gastroenterologist, neurologist, psychiatrist, and obstetrician are available. Additional findings that may help to establish the diagnosis include the change of urine color, a positive PBG urine analysis, and the use of drugs known to precipitate an acute attack of porphyria. If a patient with AIP presents with headache, conscious disturbance, seizure or visual loss, the diagnosis of PRES should be considered. The brain MRI findings can provide an important basis for the diagnosis of PRES.

If an acute attack of AIP was not diagnosed timely and treated appropriately, it was associated with dismal maternal and perinatal outcome. Neilson, *et al.* [4] reported that maternal mortality was 42.5% due to attacks of porphyria during pregnancy. Brodie, *et al.* [5] showed that 54% (27/50) of women with acute porphyria had an acute attack during pregnancy and that the babies born were significantly smaller than those who did not have a porphyria crisis. Andersson, *et al.* [6] demonstrated that miscarriage was more common amongst women with manifest AIP. Due to a better understanding of the disorder

and proper management, both the maternal and foetal outcome was remarkably improved over the years [7].

An acute attack of AIP is a potentially life-threatening event that requires rapid therapeutic intervention. Key principles of management should comprise removing all potential precipitating factors, symptomatic treatments and starting a hemin or high-dose glucose infusion. Porphyrinogenic drugs administered during anesthesia and labor must be identified and should be ceased. The administration of morphine or pethidine is recommended for pain relief. Nausea and vomiting can be treated with administration of prochlorperazine or promazine. Hypertension should be controlled by β -adrenergic blocking agents. The most important therapeutic step for AIP is the early intravenous administration of normosang. The administration of concentrated glucose and a high carbohydrate diet should also be applied as an adjuvant. The side effects of normosang are markedly little. Although the risks of using Normosang during pregnancy are not defined up to date, no after-effects have been observed in new-born babies whose mothers were treated with Normosang during their pregnancy. In addition, it is also essential to monitor the vitals, intake output, fluid and electrolyte balance.

In conclusion, the association of AIP and pregnancy is rare. However, it is a great challenging problem for obstetricians. Prompt diagnosis and early detection of the disease are crucial for the appropriate treatment and favorable prognosis. All pregnant patients who complained of unexplained abdominal pain should be considered the diagnosis of acute porphyria. If proper managements are taken, the patient with AIP may have an uneventful pregnancy and puerperium outcome.

Conflict of Interest

All authors declare that they have no conflict of interest.

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