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Case Report
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# Diagnosis of Neurovisceral Crisis of Acute Intermittent Porphyria in the Puerperium

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## Abstract

Acute porphyria's are rare inherited disorders due to deficiencies of heme synthesis enzymes. The clinical presentation is heterogeneous depending on the enzyme affected. In the case of acute intermittent porphyria, a deficiency of porphobilinogen deaminase is established.

The diagnosis is biochemical by the determination of accumulated precursors. Treatment is based on adequate metabolic input and supply of human hemin. Its association with pregnancy is rare. The patterns of symptoms in the pregnant woman varies considerably between individual patients. The time of maximum risk seems to be in the initial period of pregnancy and in the puerperium where changes in the balance of steroid hormones are more likely to occur.

**Keywords: Porphyria; Puerperium; Neurovisceral crisis**

## Abbreviations

PCT: Porphyria Cutanea Tarda; AIP: Intermittent Porphyria; EPP: Erythropoietic Protoporphyria; ED: Emergency Department; BP: Blood Pressure

## Introduction

Acute porphyria's are a group of hereditary disorders caused by genetic mutations in enzymes that are part of the biosynthetic cycle of the heme group, essential for the functioning of proteins such as hemoglobin and liver enzymes of cytochrome P450. Depending on the step of the synthetic cycle in which the enzymatic alteration takes place, the accumulation of one precursor or another occurs, giving rise to various types of porphyria's. They are classified as hepatic or erythropoietic depending on the system in which the precursors are accumulate, liver or bone marrow, respectively. Based on the clinical manifestations, there are three types: Porphyria Cutanea Tarda (PCT), Acute Intermittent Porphyria (AIP), and Erythropoietic Protoporphyria (EPP) [1-3].

## Case Presentation

We present the case of a 34-year-old postpartum woman with a history of allergy to penicillin and its derivatives. She was a victim of the 11th of March terrorist attacks in Madrid with traumatic enucleation of the left eye and since then in psychiatric treatment for post-traumatic stress syndrome, with a stable clinical situation.

After the birth, which she attended without incident, she was discharged home. Seven days after discharge, she presented pain in the genital and abdominal areas that forced her to go to the emergency department several times, finally being admitted to the gynecology department due to infection of the episiotomy wound with associated abdominal pain that did not respond to first step painkillers or opiates. She was assessed by the psychiatry and pain unit departments and was diagnosed with anxious-depressive syndrome and neuropathic pain related to the puerperium. When she was discharged, she continued to present episodes of abdominal pain, and she was again thoroughly evaluated without finding any responsible organic pathology. Three weeks later, she was attended by the ED for three episodes of generalized seizures that required orotracheal intubation. Transferred to the nearest center, she underwent a cranial CT scan, which was nonspecific, and a lumbar puncture for a temperature of 38ºC. The biochemical result ruled out meningitis. At that moment she is transferred to our center due to a lack of ICU beds. Since her arrival, she noted sinus tachycardia at 150 bpm and Blood Pressure (BP) of 210/120 mmHg. Treatment with labetalol is started. After nonspecific cranial CT findings, a cranial MRI was performed in which

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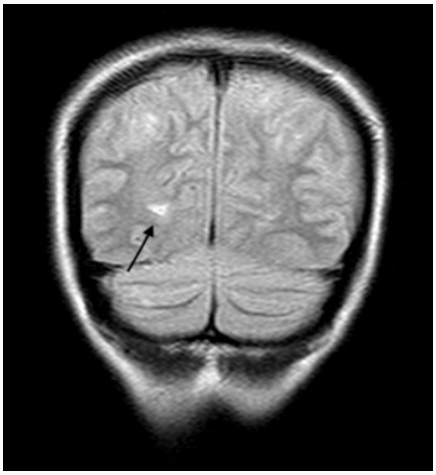

**Figure 1:** Cranial MRI imaging showing hyperintense lesions in the occipital subcortical white matter and in the cerebellar hemispheres that could be suggestive of reversible posterior leukoencephalopathy.

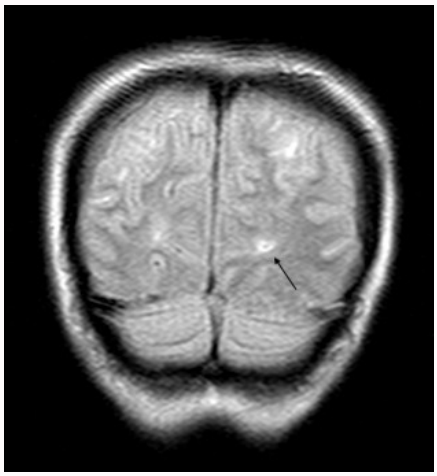

**Figure 2:** Cranial MRI imaging showing hyperintense lesions in the occipital subcortical white matter and in the cerebellar hemispheres that could be suggestive of reversible posterior leukoencephalopathy.

hyperintense lesions were observed in the occipital subcortical white matter and in the cerebellar hemispheres that could be suggestive of reversible posterior leukoencephalopathy (Figure 1, 2). The patient presented good clinical evolution and could be extubated 12 hours after admission. Since extubation, she had persistently referred colic-type abdominal pain, for which an abdominal CT scan was performed, which revealed marked distension of the large intestine with a cecum of 8 cm in maximum diameter. At the sigma level, an abrupt transition to a normal caliber rectum is observed without any cause that justifies it. After evaluation by surgery department and a colonoscopy without findings, hypotonia of the anal sphincter stands out, which could suggest the existence of aganglionic megacolon (adult Hirsprung). Associated with this, she had a history of acute urine retention after epidural puncture, which led to thinking of conus medullaris syndrome within the differential diagnosis. Lumbar and sacral MRI were requested, which were normal. After 4 days in the ICU, she was discharged with a diagnosis of seizures pending ruling out posterior reversible encephalopathy, newly diagnosed hypertension of unknown cause, and constipation with megacolon pending filiation. Four days after being on the hospital ward, the patient was re-admitted to the ICU due to a low level of consciousness. Analytical control revealed severe hyponatremia with sodium 109 mEq/L, plasma osmolarity 236 mOsm/Kg and urine 435 mOsm/Kg, compatible with SIADH. During admission, she remains spontaneously ventilated and hemodynamically stable with antihypertensive treatment. She currently associates paresthesia's and dysesthesias in lower limbs. The clinical history is reviewed again and after putting together the entire spectrum of symptoms and signs from the onset of the symptoms, coproporphyrin levels of 3920 mcg/24 h (normal value is <250 mcg/24 h) are obtained, when neurovisceral crisis of acute porphyria is suspected. Uroporphyrins value was 504 mcg/24 h (normal value is <50 mcg/24 h), total porphyrins was 4,420 mcg/24 h (normal value is between 15-300 mcg/24 h), PBG was 30.8 mg/24 h (normal value is <2 mg/24 h) and ALA was 40.8 mg/24 h (<7 mg/24 h)). Treatment with heme arginate was started at a dose of 150 mg/day for 4 days and the deficiency of the aminase PBG enzyme was confirmed in blood tests.

## Discussion/Conclusion

AIP is the most common type of acute porphyria, the European study by Elder et al. estimates its prevalence of symptomatic cases at 5.4 cases per million. The pathophysiology of the attacks is still unknown. The influence of female hormones would explain why acute attacks mainly affect women of childbearing age, although there are other possible triggers. Recognition of an acute attack in a patient not previously diagnosed with porphyria is difficult and is often delayed in time, since it occurs only when the patient has been treated on multiple occasions, as in the case we present. The main and most frequent symptom is severe and diffuse abdominal pain accompanied by severe hypertension and tachycardia. It should be noted that 40% to 58% of patients have psychiatric disorders that often present as the only initial symptom. The patient in the clinical case that we present was in psychiatric treatment for a post-traumatic stress syndrome. As diagnosis is delayed and exposure to heme precursor accumulation is prolonged, neurological complications such as seizures, reversible posterior leukoencephalopathy syndrome or neuropathy are more likely. Analytical controls are usually within normal limits except for hyponatremia that appears in 40% of attacks. Acute attacks are always accompanied by increased urinary excretion of porphyrins. The treatment of choice is the administration of hemin, which has the ability to suppress the activity of the enzyme delta-amino-levulinic acid synthetase by feedback, reducing the production of porphyrins and heme precursors. The interest of this case lies in how the diagnostic delay in time causes the patient to experience the full spectrum of symptoms of an acute porphyria neurovisceral crisis. Despite the fact that it is a rare entity, it should be considered mainly in those patients with multiple evaluations for non-specific symptoms, as well as psychiatric evaluations, since the diagnostic test is affordable and minimally invasive.

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