Case report

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Acute intermittent porphyria: cases reports and review of the literature and new treatments

Abstract

Background: Porphyrias are congenital disorders of heme synthesis, a molecule necessary for aerobic life. Its synthesis is catalyzed by eight enzymes that can be affected, resulting in seven different porphyrias. Mutations in these eight genes affect the following enzymes: aminolevulinate dehydratase (ALAD), porphobilinogen deaminase (PBGD), uroporphyrinogen synthase (UROS), uroporphyrinogen decarboxylase (UROD), coproporphyrinogen oxidase (CPOX), protoporphyrinogen oxidase (PPOX), and ferrochelatase (FECH).[1,2] There are four acute porphyrias and four non-acute porphyrias. The clinical presentation of the latter is primarily sensitivity to sunlight.[2] In porphyria, the different intermediates initially accumulate in the liver or bone marrow. Therefore, porphyrias are classified as hepatic or erythropoietic.

Method: We report three cases of patients in the productive stage, including high-performance athletes, whose main symptom was abdominal pain. In one case, surgery was performed multiple times, resulting in various abdominal and systemic complications. We therefore emphasize the importance of early diagnosis and avoiding fatal outcomes, as described in one of the patients.

Case presentation: Case 1: A 45-year-old female patient with multiple surgical history, recurrent cholangitis with stent placement on 4 occasions, presents abdominal pain predominantly in the hypochondrium and right flank of 16 hours of evolution, in her admission laboratories she presents anemia, mild hyponatremia. A CT scan and intestinal transit were performed with a report of intestinal occlusion with a site of origin in the sigmoid colon in which tachycardia, arterial hypertension, anemia and hyponatremia were reported. She underwent LAPE in which adhesions of the colon to the cystic fossa were found mainly, for which adherenciolysis was performed. In the postoperative period, the anemia worsened, so 2 red blood cell concentrates were transfused, later she was discharged due to improvement. Two months later, she was readmitted and transferred from another institution where LAPE + intestinal resection was performed, presenting data of respiratory distress, in relation to pulmonary thromboembolism, pleural effusion thoracentesis was performed, she was discharged due to improvement; Two months later, she was readmitted and transferred from another institution where LAPE + intestinal resection was performed, presenting data of respiratory distress, in relation to pulmonary thromboembolism, pleural effusion thoracentesis was performed, and treatment with anticoagulants was started.

Case 2 : Female patient of 33 years old who presents important history: practices endurance sport Triathlon, surgical, Appendectomy, initial clinical picture, abdominal pain predominantly in the hypochondrium and right flank of 24 hours of evolution, being evaluated in the emergency service where an abdominal CT scan was performed without showing alterations, evaluated by the surgery department who considered that the clinical and tomographic findings do not correspond to cholecystopathy, supportive management was started with Intravenous Glucose and a hypercaloric diet, her admission laboratories with slight hyponatremia, the evolution is towards normalization, ALA and CPB tests in urine are requested, resulting positive

Case 3: 29-year-old female with the following important history: Gynecological and obstetrical: Pregnancy 1, Cesarean section 1, begins with a picture of approximately 1 week of evolution characterized by generalized abdominal pain being evaluated by a doctor, who diagnoses functional colonopathy, being managed with antispasmodic drugs, without resolution of the picture, presenting with greater intensity, reason why it is decided to admit her for hospital management, opioid drugs are administered due to persistence of abdominal pain, abdominal distension and constipation, studies are performed Computed axial tomography of the abdomen without acute pathology, persisting with the picture without response to management, relevant laboratory tests with hyponatremia 125 mEq / l, with further deterioration, primarily in the neurological sphere, with the presence of restlessness and disorientation, without response to management with anxiolytics and subsequently antipsychotics, subsequently presenting symptoms of respiratory difficulty, with supplementary oxygen support, it is decided to admit him to the Intensive Care Unit, due to progression of ventilatory failure, it is decided to manage his airway with orotracheal intubation, with sedation and analgesia and airway hemorrhage occurs, a dark uresis is presented, for which ALA and CPB tests are requested in urine, resulting positive, his management begins with intravenous glucose, cimetidine, without response, due to prolonged intubation it is decided to perform a tracheostomy, evolving with relative stability, but presenting a new episode of airway hemorrhage and coagulation disorders, with manifestations of severe pulmonary hemorrhage, without response to the management of blood derivatives, she dies of multiple organ failure, hematological, renal, hepatic, neurological disorder mainly.

Conclusion: Due to the complexity and diagnostic difficulty involved in identifying this disease, it is of vital importance and medical interest to share experience, established approaches, and management of the case identified in our hospital unit, in order to provide comprehensive support to the medical community.

Keywords: Acute hepatic porphyria, cases reports, review of literature

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Ervin Manzo Palacios MD,[1] García Miranda GM MD,[2] Alejandro Lastiri López MD,[3] Jorge Alfonso Perez Castro y Vazquez MD,[3] Alejandro Díaz Hernández MR[4]

[1]Head Department Critical Care Medicine Hospital Angeles Metropolitano, México
[2]Department Anestesiology (retired) Hospital de Ortopedia "Victorio de la Fuente Narvaez", Instituto Mexicano del Seguro Social, México
[3]Department Surgery, Hospital Angeles Metropolitano, México
[4]Department Surgery Resident Hospital Angeles Metropolitano, México

Correspondence: Ervin Manzo Palacios, Head Department Critical Care Medicine Hospital Angeles Metropolitano, Tlacotalpan 59, Roma Sur, Mexico City CP 06760, Tel +52 52651800 Extension 1307

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Abbreviations: ALAD, aminolevulinate dehydratase; PBGD, porphobilinogen deaminase; UROS, uroporphyrinogen synthase; UROD, uroporphyrinogen decarboxylase; CPOX, coproporphyrinogen oxidase; PPOX, protoporphyrinogen oxidase; FECH, Ferrochelatase; AIP, Acute intermittent porphyria; ALA, Aminolevulinic acid; PBG, Porphobilinogen

Introduction

Porphyrias (known as "Kings' disease or Vampire disease"), are congenital disorders of heme synthesis, a molecule necessary for aerobic life. Its synthesis is catalyzed by eight enzymes that can be affected, resulting in seven different porphyrias. Mutations in these

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eight genes affect the following enzymes: aminolevulinate dehydratase (ALAD), porphobilinogen deaminase (PBGD), uroporphyrinogen synthase (UROS), uroporphyrinogen decarboxylase (UROD), coproporphyrinogen oxidase (CPOX), protoporphyrinogen oxidase (PPOX), and ferrochelatase (FECH).[1,2] 4 acute porphyrias and 4 non-acute porphyrias. The clinical presentation of the latter primarily manifests with sensitivity to sunlight.[2] In porphyria, the various intermediates initially accumulate in the liver or bone marrow. Porphyrias are therefore classified as hepatic or erythropoietic. There are five hepatic porphyrias, four of which are classified as acute due to the symptoms they present, affecting the nervous system. The fifth hepatic porphyria is classified as chronic, presenting photosensitivity without neurological symptoms; it is called brown cutaneous porphyria.

There are three forms of acute porphyria: acute intermittent porphyria, variegate porphyria, and hereditary coproporphyria.[2] A fourth, extremely rare, recessive form of AHP called 5-aminolevulinic acid dehydratase (ALAD) deficiency porphyria (MIM 125270) is different from the other three in its clinical presentation and is not included in this review.[3,4] Hepatic porphyrias present with neurological symptoms due to the accumulation of d-aminolevulinic acid and porphybilinogen. Erythropoietic porphyrias: erythropoietic protoporphyria and congenital erythropoietic porphyria, primarily manifest as skin and hematopoietic system disorders.[1] Porphyrias are rare metabolic disorders, which means that lack of awareness or confusion about the clinical picture with other more common pathologies leads to a delay in diagnosis, resulting in increased morbidity, mortality, and psychological stress for patients. Therefore, it is important to consider the diagnosis of porphyria as an etiologic entity in patients with abdominal pain, skin lesions, or both, in order to achieve early diagnosis and provide timely treatment.[1,5]

Approximately 99 percent of carriers remain asymptomatic; the prevalence of symptomatic patients is 1:100,000 to 1:200,000. Patients with acute porphyria can suffer fatal attacks, with neurological, psychiatric, abdominal, and cardiovascular symptoms.[2] Summer porphyria and hereditary coproporphyria may also present with cutaneous symptoms such as blistering lesions on exposed areas. More than 75% of patients with an acute attack require hospitalization. Treatment consists of avoiding triggers, including surgical procedures, infection, fasting, smoking, alcoholism, sex hormones, xenobiotics, and p450-inducing medications; therapeutic approaches such as carbohydrate loading with oral and intravenous glucose and intravenous hemin when nervous symptoms are present, in addition to analgesics and antiemetics. Early and appropriate treatment can prevent consequences such as the need for mechanical ventilation, seizures, and tetraplegic symptoms.[2] The difficulty in diagnosis can lead to unnecessary surgical interventions, which can translate into a diagnostic delay of up to 15 years.[2] Acute intermittent porphyria is a rare disease in our setting. In the European population, it occurs with a frequency of 1:100,000. It should be noted that the true incidence and prevalence are uncertain because it is a difficult disease to diagnose and its spectrum ranges from asymptomatic to clinical manifestations that can easily be confused with other conditions.[2] Acute intermittent porphyria is a genetic disease, inherited with an autosomal dominant pattern. It is due to a partial deficiency in porphobilinogen deaminase (PBGD), also known as hydroxymethylbilane synthase (HMBS).[6]

The spectrum of manifestations of acute intermittent porphyria includes severe abdominal pain, often diffuse in 85% to 95% of cases, accompanied by nausea and vomiting, bloating, constipation, and sometimes diarrhea; neurological manifestations such as muscle weakness, tetraplegia, and seizures; and cutaneous manifestations, which are more common in prophyrias such as porphyria cutanea tarda. In addition, fluid and electrolyte imbalances such as hyponatremia may occur due to inadequate secretion of antidiuretic hormone or due to gastrointestinal or renal losses.[7]

Acute attacks occur in 10% of carriers and are triggered by various factors, including medications, weight loss, hormonal changes, and fasting. Cutaneous porphyrias are rarely fatal, but they can cause social isolation and a decrease in quality of life due to the need to avoid sunlight to reduce skin damage.[5,8]

This article presents the case of a patient with acute intermittent porphyria, presenting her most common symptoms and the medical challenges faced by both patients and physicians. We will explore advances in the diagnosis and treatment of this rare disease, as well as current research seeking to provide greater understanding and therapeutic options. Due to the complexity and diagnostic difficulty involved in identifying this disease, it is of vital importance and medical interest to share experience, established approaches, and management of the case identified in our hospital unit, in order to provide comprehensive support to the medical community.

Epidemiology

Prevalence is 1:100,000; however, genetic studies have shown that the prevalence of pathogenic variants is 1:1,300 at presentation.

Erythropoietic protoporphyria is a rare disorder, with prevalences ranging from 1/143,000 in the United Kingdom to 1/75,000 in the Netherlands. However, it is the most common form of porphyria in children.[1]

The prevalence and incidence of acute intermittent porphyria vary between regions and depending on whether or not latent cases are considered. It is the most common of the porphyrias; its deficiency has a prevalence of 60 cases per 100,000 people in northern European countries.[9]

In Europe, the prevalence of manifest acute intermittent porphyria ranges from 5 to 23 cases per million inhabitants, with a mean of 5.9 cases. The highest prevalence of the disease has been reported in Nordland (Norway) and Arjeplog (Northern Sweden), with 600 and 2,000 cases per 100,000 inhabitants, respectively.[10]

The most common clinical presentation includes severe, generalized abdominal pain without evidence of peritoneal irritation.[7] It occurs most frequently in women of childbearing age, so the diagnosis should be considered in women aged 15–50 years with recurrent abdominal pain in 90% of cases. Symptoms are uncommon before menarche or after menopause.

When these symptoms are misdiagnosed and pathogenesis is promoted by inappropriate treatment (especially porphyrinogenic drugs, hypocaloric diet and inadequate pain management leading to increased stress), patients may gradually develop a peripheral motor neuropathy that first affects the proximal upper extremities and may progress to paralysis of the extensor muscles of the hands and arms.[11] Among symptomatic patients, >90% experience only or fewer than 4 attacks per year throughout their lifetime. Some attacks may be related to the luteal phase, being triggered by high progesterone levels.[12] Acute porphyrias are also associated with an increased risk of developing chronic diseases such as high blood pressure, chronic kidney disease, chronic hepatitis, and hepatocellular carcinoma.

Pathophysiology

Heme synthesis follows the following steps. It is first channeled through the mitochondrial enzyme d-aminolevulinic acid synthetase,

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which condenses glycine and succinyl coenzyme A to form 3,5-aminolevulinic acid, the exclusive amino acid for the formation of heme.[3,5] In porphyria, the various intermediates initially accumulate in the liver or bone marrow. Therefore, porphyrias are classified as hepatic or erythropoietic.

There are five hepatic porphyrias, four of which are classified as acute due to the symptoms they present, affecting the nervous system. The fifth hepatic porphyria is classified as chronic, presenting with photosensitivity without neurological symptoms; it is cutaneous brown porphyria. Acute intermittent porphyria (AIP) is the most common of the hepatic porphyrias. The enzyme deficiency becomes evident when heme production is stimulated by nutritional, hormonal, or environmental factors. This leads to the accumulation of ALA and PBG, which are capable of crossing the blood-brain barrier, causing neurotoxicity. Some triggers of acute porphyria include medications, steroids (progesterone), nutritional changes, alcohol, and smoking.[5]

Name	Age of onset	Cutaneous symptoms	Enzyme	Gene (locus)	Prevalence (cases per million)
AIP	>puberty	No	CMBS	CMBS (11q23.3)	5.4
VP	>puberty	Blistering	PPOX	PPOX(1q23.3)	3.2
HCP	>puberty	Blistering	CPOX	CPOX (3q11.2)	0.2
ADP	Childhood	No	ALAD	ALAD (9q32)	<0.0001

Characteristic of the four types of acute porphyria; all can present with attacks of abdominal pain. [3,5]

Clinical manifestations

The main manifestations of hepatic porphyrias are peripheral neuropathy and neuropathic abdominal pain. Abdominal pain occurs in 85–95% of acute attacks. It is a generalized abdominal pain accompanied by nausea, vomiting, bloating, constipation, and sometimes diarrhea. Rebound pain and abdominal rigidity are present in mild forms or may be absent. Other symptoms include insomnia, palpitations, seizures, and hallucinations. There may be fluid and electrolyte imbalances such as hyponatremia due to inappropriate secretion of antidiuretic hormone or excess renal and/or gastrointestinal sodium excretion. Reddish urine may occur with sun exposure due to porphobilin, which occurs in 75% of cases.

Acute porphyria attacks typically occur in three phases, which do not necessarily occur in sequence.

First phase: Patients may experience prodromal symptoms such as behavioral changes, anxiety, and drowsiness.

Second phase: Abdominal pain, accompanied by tachycardia, hypertension, hyponatremia, red or brown urine, nausea, vomiting, and constipation, with no evidence of peritoneal irritation.

Third phase: Neurological signs, muscle weakness, paresthesias, paralysis, tetraplegia, and seizures.[5,6]

Diagnosis

Screening tests include porphobilinogen and d-aminolevulinic acid, and urine creatinine. The diagnosis of acute porphyrias can be confirmed after initial treatment with genetic testing for pathogenic variants in the HMBS, CPOX, PPOX, and ALAD genes. During attacks, ALA and PBG levels are elevated at least 5 times above normal. 24-hour collection is not recommended.[13] ALA and PBG levels may remain elevated for months to a year after an acute attack in Acute intermittent porphyria (AIP), but they decrease rapidly in VP and HCP. In asymptomatic patients with AIP, 15%–44% of patients may have normal urine 5-aminolevulinic acid (ALA) and porphobilinogen (PBG) levels. Therefore, repeat testing is recommended in these patients during an acute attack. In patients with frequent symptoms, normal urine ALA and PBG levels may rule out the diagnosis.[14]

When biochemical tests are positive, genetic testing should be performed to determine the specific type of hepatic porphyria. Sequencing of the four genes ALAD, HNBS, CPOX, and PPOX is performed to diagnose ALAD, AIP (Acute intermittent porphyria), HCP, and VP, respectively. Screening of first-degree relatives should be performed to identify patients at risk of experiencing an acute attack.[9,10] The triad of abdominal pain, peripheral neuropathy, and central nervous system involvement may occur. It is unclear how common this triad is.

First-line testing includes random urine PBG and total porphyrins.

When an acute attack of porphyria is suspected, measurement of urinary ALA and PBG should be performed in a random sample. A 24-hour urine collection is not necessary.[5] ALA levels are expected to increase by at least twofold. The urine sample should be protected from sunlight and kept at <4°C.

Once the porphyria causing the attack is diagnosed, treatment should be initiated, as initial treatment does not depend on the type of porphyria. Treatment can be initiated empirically in critically ill patients while awaiting urine ALA and PBG results.

Special considerations:

Anesthetic considerations

Anesthesia in patients with porphyria requires preventing an acute attack, especially in acute intermittent porphyria, as it increases the risk of death.

Factors that can increase morbidity

There are multiple factors that can trigger an attack, such as prolonged fasting. Maintaining a calorie intake before and after surgery is important, and glucose solutions are indicated if necessary. Patients should receive 2,000 calories per day enterally or 10% dextrose, or 300-500 g intravenously daily. Any fluid imbalances will be corrected before, during, and after surgery. Operating room lighting must be carefully monitored in patients with photosensitivity.[14,15]

Recommended anesthetics

The recommended anesthetics for safety are hypnotics such as propofol, and opioids such as morphine, fentanyl, alfentanil, and sufentanyl. Inhaled anesthetics such as sevoflurane and isoflurane are recommended. Regarding benzodiazepines, midazolam and lorezapine can be used. Vecuronium, rocuronium, cisatracurium, and atracurium are recommended as muscle relaxants. Barbiturates such as thiopental, phenobarbital, and benzodiazepines such as diazepam should be avoided due to their prolonged action; phenytoin and carbamazepine should not be used.[14,15]

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Side effects of anesthesia in patients with porphyria

The main side effect is the development of an acute attack, so it is important to use approved anesthetics at appropriate doses. However, given the complexity of the disease and all the factors that can provoke an acute attack, it is important to monitor the patient over the following days, as a late acute attack may occur. It is essential to monitor the patient's neurological and cardiovascular status; the treatment of choice for an attack is to determine delta-aminolevulinic acid (ALA) and porphobilinogen (PBG) levels. The elective treatment is to administer hemin at 3–4 mg per kg per day, with a maximum of 250 mg per day diluted in saline solution for 3–4 days.[14-16]

Anesthetic management in porphyria

Management will depend on the patient's age, the surgery to be performed, the classification of the porphyria, and the patient's condition. It is important to always administer the indicated anesthetics and avoid those that should not be used. The perioperative, intraoperative, and postoperative periods must be carefully monitored, as well as adequate pain management, using opioids at appropriate doses to avoid stressing the patient and causing an acute attack.[16]

Treatment

In patients without neurological symptoms, treatment can be started with 200 g of carbohydrates per day, for example, 2 liters of glucose solution per day, except in patients with hyponatremia, who should receive enteral carbohydrates, since fasting induces the expression of the D receptor (Peroxisome proliferation activated receptor, coactivator 1 A), which induces the expression of ALAS1. Sodium correction should be performed, taking into account that the main etiology of hyponatremia in porphyria is the inadequate secretion of antidiuretic hormone. Opioids may be required for pain control. In patients with severe hyponatremia that is difficult to control, hemin arginate dissolved in 20% albumin administered at a dose of 3 mg / kg should be started. Hypertension can be controlled with beta-blockers such as propranolol.

Hemin: It decreases ALAS1 expression in the liver, resulting in a decrease in the accumulation and overproduction of ALA and PBG. Urine ALA and PBG levels should be measured before using hemin. P450-inducing medications should be discontinued, as they can directly increase ALAS1 messenger RNA.

In patients with epilepsy, magnesium sulfate, benzodiazepines, and levetiracetam are safe options for porphyria.

Liver transplantation is curative and is reserved for patients who have failed other therapeutic alternatives.

Treatment for patients with an acute attack requiring hospitalization is with hemin, preferably via CVC because it can cause thrombophlebitis.

Patients with acute porphyria should be monitored annually to identify liver disease.[12]

Acute porphyria attacks: treatment

The identification and elimination of triggering factors, such as porphyrinogenic medications, are the first steps in treating acute attacks.

Hospitalization is usually necessary, as is the administration of symptomatic treatment with adequate analgesia, laxatives, hypnotics, and anxiolytics when necessary.

Specimens should be collected as soon as possible for measuring heme precursor excretion. Carbohydrate administration, aimed at increasing intracellular energy availability, can be initiated early in acute attacks. If intravenous carbohydrate solutions are administered, sodium levels must be carefully monitored, as glucose infusion may aggravate hyponatremia. Carbohydrate loading can ameliorate symptoms and may suffice as a treatment for mild attacks.[19,20]

Intravenous administration of hemin should be considered for severe acute porphyria attacks.

Hemin restores heme deficiency and downregulates heme biosynthesis in hepatocytes.[4,17]

There are two human hemin therapeutics: one is a lyophilized human hemin (Panhematin), mainly used in the USA, [20,21] and the other is a stabilized complex with arginine (Normosang),[22] which is used in Europe and several other continents.[23] Consensus favors doses of 3–4 mg/kg body weight/day (a maximum of 250 mg heme arginate or 313 mg Panhematin), administered as a single dose into a large vein. The treatment is given for 2–4 consecutive days depending on the clinical response and severity of the attack. The use of hemin has not shown any negative effect on pregnancy and no adverse effects on the fetus and/or neonate.[24]

Small interfering ribonucleic acid: A new therapeutic strategy

Prior to the approval of givosiran, treatment options were limited, and disease management focused on avoidance of attack triggers and use of intravenous (IV) glucose or hemin for attacks.[25] For patients experiencing recurrent attacks, the impact of the disease can be severe[26,27]management may include prophylactic hemin, and, rarely, liver transplantation has been used as the treatment of last resort.[28,29] Hemin treatment carries the risk of adverse events (AEs), both acute (eg, headache, phlebitis) and chronic (eg, iron overload, venous thrombosis, venous obliteration, and central venous catheter complications).[30-33]

Givosiran is a subcutaneously administered RNA interference therapeutic approved for the treatment of AHP in adults (USA, Brazil, Canada),[34-36] and in adults and adolescents aged 12 years and older (European Economic Area, United Kingdom, Switzerland, Japan).[37] Targeting messenger RNA (mRNA) encoding A LAS1, givosiran lowers induced ALAS1, thereby preventing accumulation of ALA and PBG.[38-41]

Clinical studies have demonstrated that givosiran treatment leads to sustained lowering of urinary ALAS1 mRNA, ALA and PBG levels, and, in patients experiencing recurrent attacks, reduces the annualized attack rate (AAR) compared with placebo.[40,41]

Conclusion

This review and cases reports includes a description of the conditions of acute intermittent porphyria, its clinical manifestations, treatment, disease burden, and the impact of givosiran, a new treatment modality. Due to the complexity and diagnostic difficulty involved in identifying this disease, it is of vital importance and medical interest to share experience, established approaches, and management of the case identified in our hospital unit, in order to provide comprehensive support to the medical community.

Declarations

There are no conflicts of interest or sources of support.

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