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Acute Intermittent Porphyria: Understanding the Disease, Its Management, and Real-Life Cases

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Introduction

Acute intermittent porphyria (AIP) is a rare inherited metabolic disorder caused by reduced activity of the enzyme hydroxymethylbilane synthase (HMBS), also known as porphobilinogen deaminase. It is the most common type of acute hepatic porphyria and follows an autosomal dominant inheritance pattern, although many people who carry the mutation never develop symptoms. [1,2]

When symptoms do appear, however, they can be severe, affecting multiple organ systems and sometimes becoming life-threatening if diagnosis and treatment are delayed. Because the symptoms often mimic more common medical conditions, AIP is frequently overlooked in the early stages. [3]

Pathophysiology and Causes

AIP results from a defect in the third step of the heme synthesis pathway. This defect causes toxic precursors—delta-aminolevulinic acid (ALA) and porphobilinogen (PBG)—to accumulate, particularly in the liver. [3]

Acute attacks are usually triggered by factors that increase the activity of the enzyme ALAS1, including: [2,4]

- Certain medications such as sulfonamides and barbiturates
- Hormonal fluctuations, especially during the menstrual cycle
- Fasting or restrictive dieting
- Alcohol intake
- Infections and physical stress

The buildup of ALA and PBG is believed to have neurotoxic effects, explaining the neurological and psychiatric symptoms commonly seen during attacks. [4]

Clinical Manifestations

AIP most commonly presents after puberty and is seen more frequently in women. [1] Patients typically experience a combination of abdominal, neurological, and psychiatric symptoms. [5]

Common Features

1. Abdominal Pain

The hallmark symptom is severe abdominal pain that is often diffuse and disproportionate to physical examination findings. Despite intense pain, abdominal examination may appear relatively normal, sometimes described as a "silent abdomen." [5]

2. Neurological Symptoms

Neurological involvement may include: [4,8]

- Peripheral neuropathy
- Progressive limb weakness
- Seizures
- Autonomic dysfunction such as tachycardia and hypertension

3. Psychiatric Manifestations

Patients may also develop: [4]

- Anxiety
- Agitation
- Depression
- Hallucinations or psychosis

Other important findings include hyponatremia, often related to syndrome of inappropriate antidiuretic hormone secretion (SIADH), and dark or "port-wine" colored urine caused by oxidation of porphyrin precursors on exposure to light and air.[2]

Diagnosis and Management

The gold standard for diagnosing an acute attack is the detection of elevated urinary porphobilinogen (PBG) during symptomatic periods. Markedly increased urinary PBG levels strongly support the diagnosis. [5] Genetic testing for HMBS mutations can confirm AIP and is valuable for family screening. [2]

Management Strategies

Avoidance of Triggers

Potentially porphyrogenic drugs should be stopped immediately, and adequate nutritional intake should be maintained. [8]

Intravenous Heme Therapy

Intravenous heme preparations, such as hematin or heme arginate, are considered the treatment of choice for moderate to severe attacks. These therapies suppress the overproduction of toxic heme precursors. [8]

Carbohydrate Loading

High-carbohydrate intake or intravenous glucose may help control mild attacks, especially when heme therapy is unavailable. [6]

Givosiran

Givosiran is a newer RNA interference (RNAi)-based therapy that reduces hepatic ALAS1 expression and is used to prevent recurrent attacks in selected patients. [1,9]

Case Study 1: Recurrent Abdominal Pain in a 27-Year-Old Woman

A 27-year-old woman presented with her third episode of severe periumbilical abdominal pain radiating to her back, accompanied by nausea and vomiting. During previous episodes, she had also experienced anxiety and panic attacks. [6]

On this admission, she reported dark red urine and developed rapidly progressive weakness affecting all four limbs. Laboratory investigations showed normal inflammatory markers, but urinary screening tests for porphobilinogen were strongly positive. [6]

Further history revealed that her 17-year-old brother had died two years earlier after experiencing similar unexplained symptoms. Based on the clinical presentation and laboratory findings, she was diagnosed with acute intermittent porphyria.

The patient received intravenous fluids, high-carbohydrate therapy, and counselling regarding trigger avoidance. Her condition gradually stabilized, and she was referred for specialist haematology follow-up. [6]

Case Study 2: Neuropsychiatric Crisis in a 23-Year-Old Man

A 23-year-old man arrived at the emergency department with abdominal pain, vomiting, and constipation. Initial investigations were inconclusive, and his symptoms were initially thought to be related to a gastrointestinal condition. [7]

As his condition worsened, he developed hypertension, renal impairment, and severe psychiatric symptoms, including hallucinations and suicidal thoughts. A detailed family history later revealed that his sister had died at a young age from a similar undiagnosed illness. [7]

Clinicians noted brownish discoloration of his urine, prompting further testing. A 24-hour urine analysis demonstrated markedly elevated levels of ALA and PBG, confirming an acute porphyria attack. [7]

The patient was treated with supportive care and intravenous hematin therapy, leading to gradual improvement in both his physical and psychiatric symptoms. [7]

Conclusion

Acute intermittent porphyria is a rare but potentially life-threatening disorder that often presents with nonspecific symptoms, making diagnosis challenging. Early recognition is essential, particularly in patients with recurrent unexplained abdominal pain accompanied by neurological or psychiatric symptoms. [8]

Prompt treatment, careful trigger avoidance, and newer targeted therapies such as givosiran have significantly improved outcomes and quality of life for patients with AIP. [9,10]

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