AMERICAN PORPHYRIA FOUNDATION

What is Acute Porphyria ?

This is the most common of the acute hepatic porphyrias. Its inheritance is autosomal dominant. The deficient enzyme is porphobilinigen deaminase (PBGD), also known as hydroxymethylbilane synthase (HMBS). A deficiency of PBGD is not sufficient itself to cause symptoms of acute porphyria, and other activating factors must also be present. These include hormones, drugs, and dietary changes. Sometimes, activating factors cannot be identified. Researchers are trying to identify unknown additional genetic traits that activate the disease in people with the inherited deficiency of PBGD.

Living with Acute Porphyria

Because the symptoms of acute porphyria mimic other common conditions, the diagnostic process can be long suffering for years before receiving a proper diagnosis.

This educational material is provided by the American Porphyria Foundation.

For more information, visit **porphyriafoundation.org**

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References

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2. Bonkovsky HL, et al. Acute porphyrias in the USA: features of 108 subjects from porphyrias consortium. The Amer. Jrnl of Med. 2014;127: 1233-1241.

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Effective Management of Acute Porphyria:

PATIENTS DESERVE ACCESS TO APPROPRIATE CARE

Patients Diagnosed with Acute Porphyria Deserve Access to Treatment

For people living with acute porphyria, there are treatments available, but many still encounter barriers to receiving accessible care at their preferred hospitals.

The American Porphyria Foundation (APF) urges all healthcare institutions to provide timely access to medically necessary care. We encourage everyone involved in the care of patients with acute porphyria to demand timely access to FDA-approved treatments. We believe:

Regulators should **advocate for patient access** to approved treatments

Providers need to understand acute porphyria and **provide prompt, appropriate treatment**

Patients **deserve access and coverage** for available, life-saving treatment options

Signs and Symptoms

Gastrointestinal

- Severe, intense abdominal pain
- Nausea and vomiting
- Constipation
- Diarrhea

Neurological

- Pain in the extremities, back, chest, neck, or head
- Muscle weakness
- Convulsions
- Mental symptoms
- Respiratory paralysis

Cardiovascular

- Fast heart beat
- High blood pressure

Urinary

- Dark reddish or purple urine

Managing Acute Porphyria

Acute porphyria is a genetic disease and there is no cure. However, there are strategies that can be used to prevent the onset or severity of attacks:

- Eating a balanced diet, including sufficient carbohydrates
- Avoiding dieting or fasting, even for short periods
- Avoiding certain medications that can trigger an attack
- Limiting physical and emotional stress
- Avoiding alcohol

People with frequent attacks should also consider wearing a medical alert bracelet.

Prompt Treatment is Critical

Even with proper diet and control of environmental factors, some patients still have recurring, and often severe attacks. Acute attacks typically require hospitalization and delays in treatment can be life-threatening and result in serious complications such as irreversible nerve damage.

Treatments can include:

- Medication to manage pain and other symptoms
- Glucose and carbohydrates given orally or intravenously
- Intravenous heme if the patient's symptoms fail to improve within 36 hours of glucose treatment