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International Foundation for Gastrointestinal Disorders (www.iffgd.org)

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What is Acute Hepatic Porphyrria?

Acute hepatic porphyrria (AHP) is a group of rare genetic conditions that can cause severe gastrointestinal (GI), neurologic, psychiatric, cardiovascular, and skin symptoms. There are four types of AHP which include:

- acute intermittent porphyrria,
- variegate porphyrria,
- hereditary coproporphyrria, and
- ALAD-deficient porphyrria.

It is estimated that 5 in every 100,000 persons have AHP. Most people with AHP start to develop symptoms when they are adults. While AHP can affect anyone, it is more common in women and Caucasians.

People with AHP have a defective gene that leads to problems making heme, which is an important part of hemoglobin. Hemoglobin is a protein in red blood cells that carries oxygen throughout the body. This defect leads to the buildup of toxic chemicals in the body that can damage nerve cells and cause severe symptoms. Sometimes the AHP attacks can be life-threatening. Below are the most common symptoms of AHP, many other symptoms may occur.

Signs and Symptoms of Acute Hepatic Porphyrria

Acute hepatic porphyrria (AHP) can cause a wide range of symptoms and it is different for each patient. Those with AHP can experience gastrointestinal, neurologic, psychiatric, cardiovascular, or skin symptoms. The most common symptom is abdominal pain, which can be very severe and felt throughout the belly. The pain can also last for many hours.

Patients with AHP can have sudden (acute) symptoms that can be life threatening. These symptoms can happen in many different parts of the body and often require a hospital visit. Those with AHP can also experience chronic symptoms. Chronic symptoms are ones that last a long time and may include:

- anxiety
- depression
- fatigue - feeling low energy, overly tired
- nausea - a feeling of sickness felt in the abdomen, stomach, chest, or head with feeling an urge to vomit
- pain - physical discomfort and/or suffering in the body

Gastrointestinal Symptoms of AHP

AHP often affects the GI tract and can cause the symptoms listed below.

- Pain and/or discomfort – physical discomfort and/or suffering in the belly area (stomach and intestines). 85-95% of people with AHP that experience this symptom.
- Nausea – a feeling of sickness felt in the abdomen, stomach, chest, or head with an urge to vomit. 43-88% of people with AHP that experience this symptom.
- Vomiting – bringing food back up from the stomach into the mouth. 43-88% of people with AHP that experience this symptom.
- Constipation – This can mean any or all of these: having bowel movements (BMs) less often, typically 3 or fewer times per week; the passing of hard pellet-like BMs, difficulty or straining when passing a BM, feeling unable to completely empty, or feeling the need to have a bowel movement, but are not able to. 48-84% of people with AHP that experience this symptom.
- Diarrhea – loose, watery, or frequent bowel movements. 5-12% of people with AHP that experience this symptom.

Neurological Symptoms of AHP

AHP also commonly leads to neurologic and psychiatric symptoms. The symptoms are related to the nervous system, including the brain, spinal cord, and nerves of the body. Symptoms that impact the nervous system include:

- Pain in arms, legs, chest, back, neck, or head. 50-70% of people with AHP that experience this symptom.
- Muscle weakness. 42-68% of people with AHP that experience this symptom.

- Mental symptoms like agitation, confusion, depression, hallucinations. 40-58% of people with AHP that experience this symptom.
- Seizures. 10-20% of people with AHP that experience this symptom.

Other symptoms that patients with AHP can experience are fast heart rate, high blood pressure, dark or reddish urine, and skin blisters on areas exposed to sun, among others

About IFFGD

The International Foundation for Gastrointestinal Disorders (IFFGD) is a 501(c)(3) nonprofit education and research organization. We work to promote awareness, scientific advancement, and improved care for people affected by chronic digestive conditions. Our mission is to inform, assist, and support people affected by gastrointestinal disorders. Founded in 1991, we rely on donors to carry out our mission. Visit our website at: www.iffgd.org.

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