Acute Hepatic Porphyria and Cutaneous Porphyria

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

What is Acute Hepatic Porphyria?

Acute hepatic porphyria (**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 porphyria,
- variegate porphyria,
- hereditary coproporphyria, and
- ALAD-deficient porphyria.

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.

What are Acute Hepatic Porphyria and Cutaneous Porphyria?

Porphyrias are a group of conditions where the body has problems making heme, which is an important part of hemoglobin. Hemoglobin is a protein in red blood cells that carries oxygen throughout the body. The two main classes of porphyrias are Acute Hepatic Porphyria (AHP) and cutaneous porphyria.

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Cutaneous porphyrias cause skin symptoms including sensitivity to sunlight, blisters on exposed skin (e.g., hands, arms, face), sudden painful redness and swelling, and itching, among others. The most common cutaneous porphyria is porphyria cutanea tarda (PCT). It is estimated that up to 20 in every 100,000 persons have PCT.

What Causes Acute Hepatic Porphyria and Cutaneous Porphyria?

Acute Hepatic Porphyria (AHP) runs in families and can be inherited. In this case, those with AHP have a defective gene for a protein involved in making heme. Those with porphyria cutanea tarda (PCT) often came in contact with an outside factor during their lives that made a protein in the heme pathway to not work as efficiently. Examples of these factors include alcohol use, smoking, infection with hepatitis C virus or HIV. For both AHP and PCT, these defective proteins lead to buildup of toxic chemicals in the body that cause symptoms. In order for hemoglobin to bind to toxic chemicals, such as carbon dioxide, and take them to the lungs to be filtered out of the body.

What Symptoms Do Those with Acute Hepatic Porphyria and Cutaneous Porphyria Experience?

People with AHP can experience severe attacks of gastrointestinal, neurologic, psychiatric, cardiovascular, and skin symptoms. These attacks can be life threatening and need urgent treatment in the hospital. In between episodes, those with AHP can also experience chronic pain and may later develop liver damage and kidney failure. On the other hand, those with PCT get skin blisters that can lead to permanent skin damage or become infected. Even after the skin heals, it can scar, become fragile, or become discolored.

How are Acute Hepatic Porphyria and Cutaneous Porphyria Diagnosed?

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AHP is generally diagnosed through a spot urine porphobilinogen (PBG) test during an acute attack. AHP can also be diagnosed with a genetic test, even when they are not having symptoms. PCT is typically diagnosed by measuring total porphyrins in the blood or urine.

How are Acute Hepatic Porphyria and Cutaneous Porphyria Treated?

For those with AHP, symptoms during an acute attack are usually treated with IV hemin in the hospital. To help prevent attacks, a once-monthly, self-injectable medicine called givosiran (Givalaari®) can be used. For patients with PCT and active skin lesions, either phlebotomy to reduce the body's iron levels or a drug, hydroxychloroquine, can be used. Phlebotomy involves safely removing blood from the body with an IV needle, typically placed inside the elbow or on the back of the hand. To help prevent future skin lesions, patients should avoid sun, alcohol, smoking, excess iron intake, and estrogens as well as treat hepatitis C virus or HIV when present.

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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