Diagnosing and Treating Acute Hepatic 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.

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Acute hepatic porphyria (AHP) is a rare condition with symptoms that are similar with other more common conditions (e.g., irritable bowel syndrome, fibromyalgia, endometriosis). This causes some patients to go many years before the correct diagnosis is made. Irritable bowel syndrome (IBS) is a chronic and recurrent disorder of the GI tract usually accompanied by diarrhea, constipation or both. Fibromyalgia (FM) is a condition marked by muscle pain all over the body, sleep problems, and fatigue. Endometriosis is a painful condition in women where tissue like the lining of the uterus or womb grows on the outside of the organ.

Diagnosing Acute Hepatic Porphyria

Lab tests are needed to properly diagnose AHP. The type of test that should be done depends on whether the person is having symptoms at the time. When a person having symptoms from AHP, a spot urine test for porphobilinogen (PBG), a porphyrin, may be used. This test simply involves urinating in a specimen container, often a small cup. The sample is then tested in the lab for PBG levels. If PBG levels are high, a healthcare provider can tell that a person has AHP. Other tests that may be done include a urine test for aminolevulinic acid (ALA) as well as testing for total porphyrins in the urine, blood, or stool. If someone has severe kidney disease, a blood test for PBG may also be done. This is due to the kidney being unable to properly filter urine.

The Food and Drug Administration (FDA) is one of the U.S. government's regulatory agencies. This agency oversees a broad range of topics that pertain to food, drugs and other products used daily.

The FDA works to protect public health by assuring that foods and drugs for humans and animals are safe and properly labeled. The FDA also ensures that vaccines, other biological products, and medical devices intended for human use are safe and effective.

Products approved by the FDA have been deemed safe, with benefits that are worth the possible risks. This is done after reviewing studies and tests that have been done on a product.

As AHP is caused by a gene defect, genetic testing can also be used to diagnose the condition. Unlike the urine tests mentioned above, the genetic test can be done when a patient is not experiencing symptoms. Genetic testing is also helpful for diagnosing the condition in family members of those with known AHP. The genetic test looks for problems in 4 different genes and can diagnose the specific type of AHP that one may have (acute intermittent porphyria, variegate porphyria, hereditary coproporphyria, and ALAD-deficient porphyria).

Treating Acute Hepatic Porphyria

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When someone with AHP is experiencing an acute attack, treatment with intravenous (IV) hemin in the hospital can help with symptoms. At that time, it is also important to try to identify what may have caused the attack (e.g., infection, medicine, diet changes) so that they can be avoided in the future.

A drug called Givosiran (Givalaari®) was approved by the U.S. Food and Drug Administration in 2019 for the treatment of adults with AHP. It is a medicine that is injected under the skin once a month. In clinical trials, patients on givosiran had fewer acute attacks from AHP when compared to those given a placebo. A placebo is a pill or treatment with no active ingredients. While taking givosiran, patients should have their liver and kidney tests monitored by their healthcare team.

For patients with AHP who suffer from repeated severe attacks, do not respond to IV hemin or givosiran, or have a very poor quality of life, liver transplantation can be considered as a last resort.

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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About the Publication

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