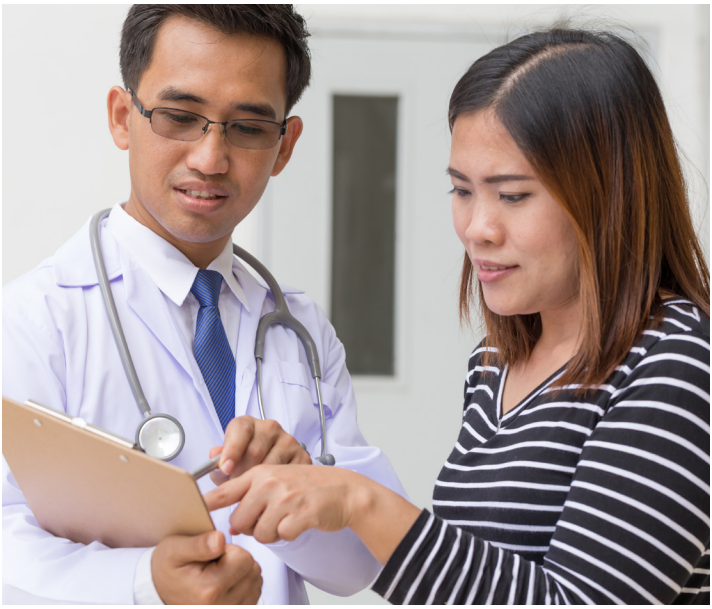
Acute Hepatic Porphyria



AMERICAN LIVER FOUNDATION®

Your Liver. Your Life.

Why is your liver important?

The liver is a vital organ responsible for many functions in your body. It is located under your rib cage on your right side. Some of the jobs the liver does include:

- Giving you energy
- Producing bile for digestion
- Processing what you eat and drink into nutrients your body can use
- Filtering out harmful substances from your blood

What is Acute Hepatic Porphyria?

Acute Hepatic Porphyria (AHP) is also known as Acute Porphyria. It refers to a family of rare genetic diseases characterized by potentially life-threatening attacks and, for some people, ongoing and sometimes lifelong pain and other symptoms that interfere with their ability to live normal lives.

There are four types of AHP. Acute Intermittent Porphyria (AIP) makes up about 80% of all cases. The remaining types are:

- Variegate Porphyria (VP)
- Hereditary Coproporphyia (HCP)
- ALAD Deficiency Porphyria (ADP)

AHP can be a hereditary disease, meaning that it can be passed from parents to children. This can occur if either one or both parents carry the defective gene, depending on the AHP type. Men and women inherit the disease equally as often; however, women tend to suffer symptoms more often than men.

What causes AHP and how does it affect the liver?

Porphyrins are compounds needed to produce heme. Heme is vital to the body and is responsible for breaking down medications and other substances.

AHP occurs when there is a problem with heme production in the liver. When heme is not produced properly, certain toxins called PBG (porphobilinogen) and ALA (aminolevulinic acid) accumulate in the liver and can further circulate throughout the body. ALA and PBG are associated with the painful attacks and other disease manifestations people with AHP experience.

What are some symptoms of AHP?

AHP can cause a wide range of symptoms that mimic those of other diseases, and some people with a defective gene associated with AHP may not have any symptoms whatsoever. People with AHP who experience symptoms can suffer from severe attacks that include very painful abdominal (belly) pain. Some people may also experience chronic symptoms such as pain in between attacks. Most people have at least one other symptom in addition to the belly pain. Some of these symptoms may include:

- Nausea and vomiting
- Diarrhea or constipation (watery stool or difficulty having bowel movements)
- Back or chest pain
- Muscle weakness
- Fatigue
- Fast heartbeat
- Seizures
- Anxiety and/or depression

- Confusion
- Skin blistering (in VP and HCP only)

The various symptoms of AHP can lead to physical and emotional suffering and exhaustion. This can affect every aspect of life, including overall physical comfort; the ability to work consistently; and maintaining a healthy level of social connectedness with others.

How is AHP diagnosed?

Diagnosis of AHP can be difficult because it is a rare disease, and the symptoms are so wide-ranging that they match those of other, more well-known illnesses. Symptoms can appear to be gastrointestinal (digestive system) in nature; they can seem to be associated with heart problems; they may seem to be muscle-related; they may be mistaken for gynecological problems; or they can present as neurological, psychological or emotional in nature.

Due to the extreme range of symptoms, people with AHP are often incorrectly diagnosed. Many people see many doctors over the course of several years before they find out exactly what is wrong. These are just some of the diseases and conditions that someone with AHP could be incorrectly diagnosed with before finally receiving a correct identification of their disease:

- Fibromyalgia
- Endometriosis
- Irritable Bowel Syndrome
- Guillain-Barre Syndrome
- Hepatitis
- Psychosis
- Seizure disorders

"I was persistent in trying to improve my health. I wanted to know why I was sick...over the years, I saw many doctors.
I was treated like a drug seeker, like I had a mental illness, or like I was making it up. I talked to several experts around the country, but they had no answers as to why I was having frequent attacks. I just wanted my doctors to listen and try to understand. Honestly, that has been one of my biggest struggles. I share my story because people living with rare diseases have such a hard time getting their voices heard. But you are your best advocate. There is hope, and there is help. Do not give up."
Heather

People who suffer with extreme belly pain and have at least one of the other symptoms stated earlier should consider making an appointment with a doctor to begin discussing AHP. In addition to routine tests that may be done to diagnose illness, AHP testing can involve a urine test that measures (1) the levels of porphyrin precursors, PBG and ALA; and (2) porphyrins, all of which can be significantly higher than normal in AHP. The accuracy of these tests is better if they are done when someone is having an attack or shortly after they have had an attack.

Additionally, a genetic test can be done to help confirm an AHP diagnosis and help identify at-risk family members. The genetic test can be done using a saliva or blood sample. The benefits of having a genetic test include:

- Helping to confirm AHP as a diagnosis
- Pinpointing the specific type of AHP
- Determining the specific genetic mutation so that other family members can be tested

How is AHP treated?

There is no cure for AHP, but there are ways to help manage its symptoms. The U.S. Food and Drug Administration has approved some medications that doctors may prescribe to either reduce or treat AHP attacks. Disease management may also include pain medications and glucose supplementation.

Treatment can sometimes include hospital stays so that patients can be monitored and treated when they have attacks that produce serious medical issues such as dehydration, hallucinations, paralysis and breathing difficulties.

How is AHP managed?

It is beneficial for people with AHP to learn what kinds of situations or actions can trigger an attack. These situations can be as varied as the symptoms themselves. Here are some common triggers that can aggravate AHP and bring on an attack:

- Certain prescription and non-prescription ("over the counter") drugs

- Hormone replacement therapies
- Menstrual cycle/pregnancy and birth control
- Infections
- Stress
- Excess alcohol intake
- Extreme dieting/rapid weight loss
- Surgery/anesthesia

People cannot always control every aspect of life; however, even if it is a situation that cannot be controlled (such as menstrual cycles), communicating with your doctor and being as prepared as possible before an attack happens can help improve body, mind and spirit in the presence of AHP.

If someone has AHP, what kinds of questions should they ask their doctor?

It is important that people feel comfortable asking their care team questions to better understand the nature of their illness. Some conversation starters that you can use with your doctor are:

- Can you explain AHP to me in simple terms to help me understand the disease better?
- How did I get AHP?
- Can I pass AHP on to my children?
- Is it possible to have more than one kind of AHP at the same time?
- How can we find out if my AHP is damaging my liver?
- Are there any special tests I should have to find out the condition of my liver?
- How does AHP affect me if I have other diseases too?

- Can we try to figure out what triggers my AHP attacks?
- I can probably control some of my triggers, but what about the ones I cannot control such as getting my menstrual period?
- Is there any medication I can take that will help with pain or other symptoms?
- Are there any medications that could be harmful to me if I have AHP?
- Should I eat or avoid certain foods? What about taking vitamins?
- How do I know if I should call you or go to the hospital if I am having an attack?
- Will I be able to live a long life with AHP?

Where can people with AHP find more information or get assistance?

Even though AHP falls under the rare disease category, there are resources available to provide additional information and support.

American Liver Foundation:
www.liverfoundation.org
HelpLine 1-800-GO-LIVER

Alnylam Pharmaceuticals - Pinpoint AHP™
www.pinpointAHP.com/

American Porphyria Foundation:
www.porphyriafoundation.org

National Institutes of Health:
www.niddk.nih.gov/health-information/liver-disease/porphyria

National Organization for Rare Disorders:
www.rarediseases.org/rare-diseases/porphyria/

Clinical trials may be available to people with AHP who wish to be part of research studies that test new treatments before being approved for use by the general public. People with AHP can talk to their doctors about clinical trials, but here are some resources to search for clinical trials on your own:

www.liverfoundation.org/for-patients/resources/clinical-trials/

www.nih.gov/health-information/nih-clinical-research-trials-you

www.rarediseasesnetwork.org/cms/porphyrias/Studies

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