Understanding Acute Hepatic Porphyria



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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

Symptoms of AHP

- Severe abdominal pain Pain that occurs inside the belly area (stomach and intestines)
- Seizures uncontrolled electrical surges in the brain that can cause sudden and temporary changes in muscles, behaviors, and ability to sense the environment around you
- Hallucinations experiencing something that is not there, typically through seeing or hearing
- Confusion being unable to think as quickly or clearly as your normal
- Skin blisters raised, fluid filled bumps on the skin
- Constipation 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 go but are not able to
- Diarrhea loose, watery, or frequent bowel movements
- A change in urine color

Patients with AHP can go years without being correctly diagnosed. This delay in diagnosis can lead to unnecessary treatments, surgeries, and stays in the hospital. This is due to AHP being such a rare condition and that symptoms are similar with other more common conditions, such as: 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 lifethreatening. Below are the most common symptoms of AHP, many other symptoms may occur.

- 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.

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