

“Whether you just received a new porphyria diagnosis or you’ve been struggling with the condition for years, it can be hard to deal with. No matter how severe your case, the symptoms of porphyria can make simple activities feel like unconquerable tasks.”

What Is Porphyria?

Porphyria is a rare disorder that primarily affects the nervous system or skin and can also cause abdominal pain. Patients usually inherit the disorder because parents pass the gene abnormalities to their children. When an individual has the condition, their cells fail to change the chemicals porphyrin and porphyrins precursors in their body into heme — a substance that makes blood red.

The body creates heme mostly in the liver and bone marrow. Bone marrow is the sponge-like, soft tissue inside your bones which makes stem cells that turn into one of the three forms of blood cells: white blood cells, red blood cells and platelets.

The heme biosynthetic pathway is the process of making heme. Eight enzymes control each step in the process. If any of the enzymes are at a low level, the body will have issues making heme. This is known as a deficiency. Porphyrin and porphyrins heme precursors start building up in the body, causing illness.

Symptoms of Porphyria

Porphyria symptoms may vary in how severe they are, as well as by type and among people. Some individuals who have gene mutations causing porphyria don’t ever have any symptoms.

Acute Porphyrias

Acute porphyrias include types of the disorder that can cause nervous system symptoms that come on suddenly and can be severe. Symptoms can last a few days to several weeks and generally improve gradually after an attack. The common type of acute porphyria is intermittent porphyria.

Symptoms and signs of acute porphyria might include:

- Pain in your legs, chest or back
- Severe abdominal pain
- Nausea and vomiting
- Diarrhea or constipation
- Urination problems
- Breathing problems
- Muscle pain, numbness, tingling, weakness or paralysis
- High blood pressure
- Irregular or rapid heartbeats (palpitations)
- Brown or red urine
- Mental changes like confusion, anxiety, paranoia, disorientation or hallucinations
- Seizures

Causes of Porphyria

Each form of porphyria comes with the same cause — a heme production problem. Heme is a hemoglobin component, a protein in the red blood cells that transfer oxygen to the entire body from the lungs.

Heme provides blood with its red color, and it contains iron. Heme production occurs in the bone marrow and liver and involves various distinct enzymes. When there's a shortage of any of these enzymes, it creates an excess buildup of specific chemical compounds associated with the production of heme. The lack of enzyme will determine the specific type of porphyria.

You inherit most forms of porphyria. Most porphyrias occur when just one parent passes along an altered gene. The specific type will determine the risk of porphyria development or if you pass it to your children.

Acute porphyria episodes, which hardly ever occur before puberty, could become triggered by certain drugs, including:

- Seizure medications
- Barbiturates
- Birth control pills
- Sulfa antibiotics

Other possible triggers are:

- Drinking alcohol
- Smoking
- Fasting
- Sun exposure
- Menstrual hormones
- Stress
- Infections

Types of Porphyria

There are various types of porphyria, but you can classify them into two categories:

1. **Hepatic:** Liver problems can cause the hepatic forms of the disease. Symptoms of the hepatic forms involve central nervous system problems and abdominal pain.
2. **Erythropoietic:** Problems with red blood cells cause erythropoietic forms.

Hepatic porphyria has five types:

1. Acute intermittent (AIP)
2. Variegate porphyria
3. Hereditary coproporphyria (HCP)
4. Porphyria cutanea tarda (PCT)
5. Aminolevulinic acid dehydratase deficiency (AVADDP)

The most common type is PCT.

Physical Effects of Porphyria

The form of porphyria you have determines the potential complications you could experience. If not treated promptly, acute porphyrias can threaten your life. While you're having an attack, you could experience:

- Breathing problems
- Dehydration
- High blood pressure
- Seizures

Episodes of acute porphyrias often require treatment at a hospital. There could be long-term physical effects, as well, with recurrent acute attacks that include chronic kidney failure, chronic pain and chronic liver damage.

Mental Effects of Porphyria

One study evaluating self-rated psychosocial components in individuals with acute porphyria found anxiety (mostly) and depression is more common in individuals with porphyria than in the general population. Almost half (46 percent) of the study's subjects reported some issues with anxiety and/or depression, with 26 percent experiencing anxiety and 13 percent experiencing depression.

Current Treatments Available for Porphyria and Their Side Effects

The treatment for porphyria depends on what type you have and the severity of your symptoms. Treatment usually involves identifying the signs and avoiding their triggers and if symptoms do occur, relieving them.

Avoiding triggers could include:

- Not using recreational drugs or alcohol
- Not using medicines known to trigger an acute attack — your doctor can provide you with a list of unsafe and safe drugs
- Not smoking
- Avoiding dieting and fasting involving severe restriction of calories
- Minimizing sun exposure — wear protective clothing when outdoors, use an opaque blocking sunscreen like a sunscreen with zinc oxide and use window filters when indoors
- Taking specific hormones to avoid premenstrual attacks
- Taking steps for reducing emotional stress
- Treating illnesses and infections promptly

Acute Porphyrias

Acute porphyria treatment focuses on relieving symptoms quickly and preventing complications.

Treatments could include:

- **Hemin Injections:** A medicine that's a form of heme will limit the production of porphyrins in the body. You could experience side effects such as tenderness, pain or swelling along your veins or discoloration of your skin.
- **Intravenous Glucose, or Sugar You Take Orally:** To maintain a sufficient carbohydrate intake
- **Hospitalization:** For treating symptoms such as vomiting, severe pain, problems breathing and dehydration

Porphyria History

The "porphyrin" term, back in 1841, came from the Greek term "porphyrus," which meant reddish-purple. Scientists first believed the blood's reddish color was from iron. One scientist conducted an experiment to show this wasn't the case. He used concentrated sulfuric acid to wash the dried blood and free the iron. Then, he used alcohol to treat it, and the iron-free residue that resulted appeared with a reddish-purple color even though no iron compound was in it.

Gerardus Johannes Mulder, in 1844, determined the iron-free, purplish substance's chemical composition and named it "hematin."

Resources:

1. <https://www.mayoclinic.org/diseases-conditions/porphyria/symptoms-causes/syc-20356066>
2. <https://www.ncbi.nlm.nih.gov/pubmed/16435203>
3. <http://www.porphyrifoundation.com/about-porphyria>
4. <https://clinicaltrials.gov/ct2/results?cond=Porphyria+&term=&cntry=&state=&city=&dist=>