

## Introduction

Porphyria is a term that refers to a group of genetic disorders. This group is sometimes called the porphyrias. Porphyria affects the nervous system or skin. Sometimes it can affect both.

Porphyria is caused by problems with how your body makes a substance called heme. Heme is found throughout the body, especially in your blood and bone marrow, where it carries oxygen.

This reference summary explains porphyria. It covers the symptoms, causes, diagnosis and treatment of the disorder.

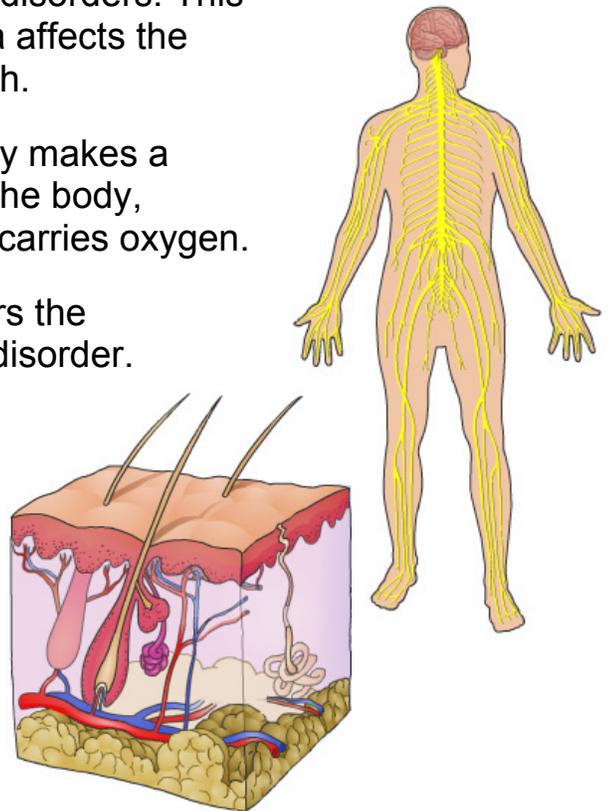
## What is Heme?

Heme is a chemical compound that contains iron. It is what gives blood its red color. The largest amounts of heme are in the blood and bone marrow. Heme is found within red blood cells in the form of hemoglobin. Hemoglobin is a protein that carries oxygen from the lungs to all parts of the body.

Heme can also be found in the liver. Heme is a part of proteins that have many functions. These functions include:

- Breaking down hormones, drugs and other chemicals
- Generating high-energy compounds that keep liver cells alive and working normally

The body makes heme mainly in the bone marrow and liver. Each step of making heme is controlled by one of eight different enzymes. Enzymes are proteins that help chemical reactions happen in the body.



If the body does not have enough of any one of the enzymes to make heme, the process is disrupted. As a result, chemicals called porphyrins can build up in the body tissues. This causes the illness porphyria.

## Symptoms

There are many different types of porphyria. The two main types are acute or cutaneous. The symptoms vary depending on the type.

Acute porphyria affects the nervous system. Common symptoms of acute porphyria include:

- Muscle numbness, tingling, paralysis or cramping
- Personality changes or mental disorders
- Pain in the abdomen, chest, limbs or back
- Constipation or diarrhea
- Vomiting



Acute porphyria can also cause:

- Fever
- Trouble urinating
- Urine that looks red or brown

Acute porphyria may also cause:

- Anxiety or paranoia
- Dehydration and excessive sweating
- Difficulty sleeping
- Hallucinations
- High blood pressure
- Seizures

Cutaneous porphyria affects the skin. People with this type of porphyria develop blisters, itching and swelling of the skin when it is exposed to sunlight.

Some types of porphyria can affect both the nervous system and the skin. People with these types of porphyria may have symptoms of both acute and cutaneous porphyria.

---

This document is for informational purposes and is not intended to be a substitute for the advice of a doctor or healthcare professional or a recommendation for any particular treatment plan. Like any printed material, it may become out of date over time. It is important that you rely on the advice of a doctor or a healthcare professional for your specific condition.

Symptoms can vary greatly in severity. Some people may have no signs or symptoms of the disorder. These people are said to have latent porphyria. People with porphyria do not have symptoms all the time. Certain triggers can cause an attack of porphyria. These include some medicines, smoking, drinking alcohol, infections, stress and sun exposure.

Other triggers of porphyria include:

- Certain diets and fasting
- Certain hormones like estrogen
- Too much iron

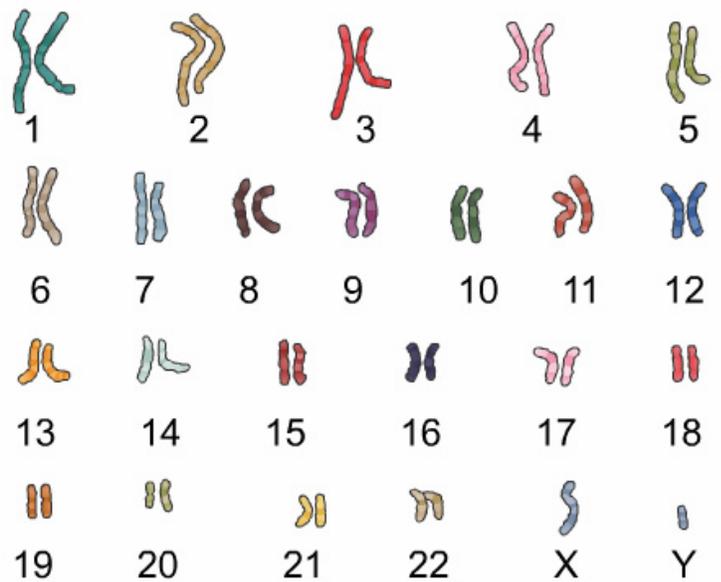
Attacks develop over hours or days. They can last for days or weeks.

### Causes

There are many types of porphyria. Most types of porphyria are inherited. This means that people with porphyria get it from their parents.

Genes tell the body to build certain substances. Every person has thousands of genes. Our genes make us look the way we do. They also have something to do with our health. Each person has 23 pairs of chromosomes. Genes are found on the chromosomes. There are many genes involved in heme production.

Genes can either work correctly or incorrectly. If a gene does not work correctly, it is called defective. If one of the genes involved in heme production is defective, the body will not have enough of one or more of the enzymes that help produce heme. The type of porphyria a person has depends on which enzyme the body is lacking.



Chromosomes

Less often, porphyria may be acquired rather than inherited. This means that it is not caused by defective genes. Instead, these types of porphyria are caused by the effects of certain diseases or exposure to chemicals.

This document is for informational purposes and is not intended to be a substitute for the advice of a doctor or healthcare professional or a recommendation for any particular treatment plan. Like any printed material, it may become out of date over time. It is important that you rely on the advice of a doctor or a healthcare professional for your specific condition.

## Diagnosis

Symptoms of porphyria are similar to symptoms of other disorders. This can make porphyria hard to diagnose. Your healthcare provider will ask you about your symptoms. He or she will also perform a physical exam.

If your healthcare provider thinks you may have porphyria, lab tests may be done to confirm it. Blood, urine and stools tests are used to diagnose porphyria. Since diagnosing the different types of porphyria can be complex, your healthcare provider may need to do several rounds of tests.



## Treatment

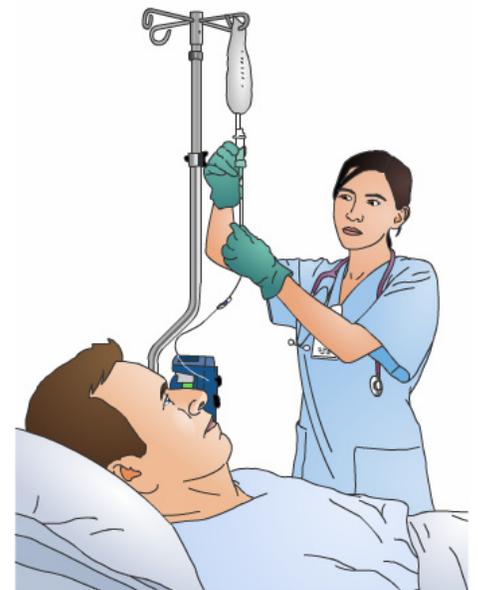
Each type of porphyria is treated differently. It usually involves medicine, treatment with heme or drawing blood.

Treatment for acute porphyria includes relieving symptoms. Medication may be prescribed to control pain. Treating any infections or other underlying illnesses that may have caused a porphyria attack is also important.

Hemin or hematin injections are often used. Hemin and hematin are forms of heme. Giving the body heme can help reduce porphyrin levels.

Severe attacks may require hospitalization for treatment. Intravenous fluids and other nutrients can help keep the body hydrated during an attack. Intravenous means giving medicine or other substances through a needle or tube inserted into a vein. It is also called IV.

Treatment for cutaneous porphyria focuses on reducing the amount of porphyrins in the body. This helps to relieve symptoms.



Limiting exposure to sunlight can help treat cutaneous porphyria. Avoid direct contact with sunlight when possible. Wearing long sleeves and a hat can help protect your skin. Use sunscreen when you go outside.

---

This document is for informational purposes and is not intended to be a substitute for the advice of a doctor or healthcare professional or a recommendation for any particular treatment plan. Like any printed material, it may become out of date over time. It is important that you rely on the advice of a doctor or a healthcare professional for your specific condition.

Phlebotomy can help reduce iron and porphyrin levels in the body for people with cutaneous porphyria. Phlebotomy is drawing blood, which contains a lot of heme, from one of the body's veins. It may need to be done several times before symptoms go away. Medication may be used to treat cutaneous porphyria in people who cannot have phlebotomy. Drugs used to treat malaria absorb extra porphyrins quickly and help your body get rid of them.

Daily doses of beta carotene or other carotenoids may be used as long-term treatment for cutaneous porphyria. Carotenoid is a substance that makes certain fruits and vegetables yellow, orange, or red. All carotenoids are antioxidants. The body uses beta carotene to make vitamin A. Vitamin A is needed for healthy skin and eyes. It may help skin become more tolerant of sunlight.

Avoiding triggers can also help prevent attacks of porphyria. Porphyria can be triggered by many different environmental factors that can be difficult to avoid. Talk to your healthcare provider for tips to prevent attacks.

## Summary

Porphyria is a term that refers to a group of genetic disorders. This group is sometimes called the porphyrias. Acute porphyria affects the nervous system. Cutaneous porphyria affects the skin. Some types of porphyria can affect both. Porphyria is caused by problems with how your body makes heme. Heme is found throughout the body, especially in your blood and bone marrow.

Eight different enzymes are needed to make heme. If the body does not have enough of one of the enzymes, a person has porphyria. The type of porphyria a person has depends on which enzyme the body is lacking. Most types of porphyria are inherited. Less often, porphyria may be caused by other disorders or exposure to chemicals.

Each type of porphyria is treated differently. Treatment usually involves medicine, injections of heme or drawing blood. Avoiding triggers can also prevent future attacks.



---

This document is for informational purposes and is not intended to be a substitute for the advice of a doctor or healthcare professional or a recommendation for any particular treatment plan. Like any printed material, it may become out of date over time. It is important that you rely on the advice of a doctor or a healthcare professional for your specific condition.