CONDITION

Porphyria

Porphyria is a group of conditions in which chemicals used to make heme build up and affect the nerves or skin.

WHAT IT IS
Group of conditions causing harmful porphyrin buildup

BODY SYSTEM
Liver, nervous system, skin, and blood production

MAIN SYMPTOMS
Severe abdominal pain, nerve symptoms, blistering, light sensitivity

WHAT CAUSES IT
Usually inherited enzyme changes; some forms acquired

HOW IT IS DIAGNOSED
Urine, blood, stool porphyrin tests; sometimes genetic testing

MAIN TREATMENT
Attack treatment, trigger control, and light protection

CAN IT BE CURED?
Sometimes - transplantation can cure selected severe forms

IS IT AN EMERGENCY?
Sometimes - severe acute attacks need hospital treatment

LONG-TERM OUTLOOK
Often manageable once the type and triggers are known

Overview

Porphyria is the name for a group of rare conditions involving heme production. Heme is part of hemoglobin, the substance in red blood cells that carries oxygen. It is also used by the liver and other organs.

The body makes heme through a series of chemical steps. Each step needs a specific enzyme, which is a protein that helps a chemical reaction happen. In porphyria, one of these steps does not work normally. Chemicals called porphyrins or their building blocks can then collect in the body.

There are several types of porphyria. They are often divided into two main groups:

Some types can cause both acute attacks and skin symptoms. The exact type matters

because symptoms, triggers, tests, and treatments are different.

Many people with an inherited gene change never develop symptoms. Others have occasional attacks or long-term light sensitivity. Correct diagnosis matters because an acute attack can require hospital treatment, while skin symptoms may improve with targeted care and protection from light.

- Acute porphyrias mainly affect the nervous system and can cause sudden attacks
- Cutaneous porphyrias mainly affect the skin, especially after light exposure

Symptoms

Acute porphyria attacks usually develop over hours or days. Severe abdominal pain is common. The pain may be widespread and may not have an obvious cause on examination or routine scans.

Other symptoms can include:

Severe attacks can cause seizures, dangerously low blood sodium, marked weakness, or problems with breathing. Symptoms such as abdominal pain and dark urine have many possible causes, so they do not confirm porphyria by themselves.

Skin symptoms differ by type. Porphyria cutanea tarda and some inherited forms can cause fragile skin, blisters, slow-healing sores, and small white bumps called milia. These changes most often affect the backs of the hands, forearms, face, or other light-exposed areas. Scarring or changes in skin color may follow.

Erythropoietic protoporphyria usually causes burning, stinging, swelling, and redness soon after exposure to sunlight or strong visible light. Blisters are less typical. The pain can be intense even when the skin looks only mildly affected.

Symptoms may begin in childhood or adulthood, depending on the type.

- Nausea, vomiting, constipation, or diarrhea
- Pain in the back, chest, or limbs
- A fast heartbeat or raised blood pressure
- Anxiety, restlessness, confusion, or difficulty sleeping
- Muscle weakness, numbness, or tingling
- Dark or reddish urine, especially after it stands in light

Causes and risk factors

Most porphyrias result from an inherited change in a gene involved in heme production. Inheritance patterns differ. Having a gene change does not always mean that symptoms will develop.

Porphyria cutanea tarda is often acquired rather than directly inherited. It develops when the activity of a liver enzyme becomes too low. Factors linked with this include excess iron, alcohol, smoking, estrogen medicines, hepatitis C, HIV, and certain inherited iron-handling changes. Not everyone with one of these factors develops porphyria.

In someone with an acute hepatic porphyria, an attack may be triggered by:

Triggers are not the same for everyone. A medicine that is unsafe for one form of porphyria may not be relevant to a skin-only form. Do not stop an important medicine without medical advice.

Sunlight and visible light trigger skin injury in cutaneous porphyrias. They do not usually cause the abdominal and nerve symptoms of an acute attack.

- Certain prescription, over-the-counter, or herbal medicines
- Fasting, crash dieting, or eating too few carbohydrates
- Alcohol use
- Infection, surgery, or other physical stress
- Hormonal changes, including changes during the menstrual cycle

Diagnosis

Testing should be matched to the symptoms and suspected type. A small rise in porphyrins on an unrelated test does not always mean that someone has porphyria.

During a possible acute attack, the key first test is usually a protected urine sample measuring porphobilinogen, often called PBG, and aminolevulinic acid, or ALA. These are heme-building chemicals that rise sharply in most acute hepatic attacks. Testing is most useful while symptoms are present.

Further urine, blood, and stool tests can show the pattern of porphyrins and help identify the specific type. Samples may need protection from light.

For blistering skin symptoms, urine and blood porphyrin measurements can help identify porphyria cutanea tarda and related conditions. Suspected erythropoietic protoporphyria requires a specialized blood test that measures and separates forms of protoporphyrin in red

blood cells.

Genetic testing may confirm an inherited type and help with family testing. However, a gene change alone does not prove that current symptoms are caused by porphyria. Many carriers never have an attack.

Other tests may check blood count, sodium levels, kidney and liver function, iron levels, and possible causes of liver injury. These results also help guide treatment safely.

Treatment and management

Treatment depends on the type of porphyria and whether symptoms are active.

A significant acute attack is usually treated in hospital. Intravenous hemin reduces the liver's production of the chemicals that cause nerve injury. Hospital care may also include treatment for pain and nausea, careful fluid management, and monitoring of sodium, blood pressure, muscle strength, and breathing.

Glucose given by mouth or vein may be used in selected mild situations under medical supervision. It is not a substitute for hemin when an attack is severe or progressing.

For people with frequent acute hepatic attacks, a specialist may consider preventive treatment such as givosiran. This medicine reduces production of an early heme-building enzyme. Regular blood tests are needed because treatment can affect the liver, kidneys, and other blood measurements.

Porphyria cutanea tarda is commonly treated by reducing excess iron through planned blood removal, called phlebotomy. Low-dose antimalarial medicine may be an alternative in selected people. Associated factors such as hepatitis C, alcohol exposure, estrogen use, or excess iron may also need attention.

Management of light-sensitive porphyrias includes protective clothing, hats, shade, window filters, and planning around light exposure. Ordinary sunscreen may not block all wavelengths that cause symptoms. Afamelanotide may increase tolerance to light in some adults with erythropoietic protoporphyria where it is available.

Liver or bone marrow transplantation can correct selected severe forms, but these are major procedures reserved for uncommon circumstances.

Complications

Repeated or severe acute attacks can damage nerves. This may lead to ongoing pain,

weakness, altered sensation, or reduced mobility. Recovery from nerve injury can take months and may be incomplete. Severe weakness can affect breathing.

Some people with acute hepatic porphyria develop long-term high blood pressure or kidney problems. There is also an increased risk of liver cancer in certain acute hepatic porphyrias, even without cirrhosis. A specialist can advise whether regular liver imaging is appropriate.

Porphyria cutanea tarda may occur alongside iron overload or chronic liver injury. Blistering forms can leave scars, changes in skin color, or small cysts.

In erythropoietic protoporphyria, protoporphyrin can contribute to gallstones. A small proportion of affected people develop serious liver injury. New jaundice, worsening upper abdominal pain, or very dark urine needs prompt assessment.

Treatments can also require monitoring. Repeated hemin may affect veins or contribute to iron buildup. Preventive medicines may require regular liver and kidney blood tests.

Prevention

Inherited porphyria cannot usually be prevented. A person is born with the relevant gene change, although symptoms may never appear.

If you have an acute hepatic porphyria, avoiding known triggers may reduce attacks. This can include eating regularly, avoiding crash diets, limiting alcohol, and checking medicines against a reliable porphyria drug-safety database. A medical alert card or bracelet can help during unexpected treatment.

Do not avoid all medicines. Untreated illness can itself trigger an attack, and many medicines are considered safe. A porphyria specialist or pharmacist can help check an appropriate database when a new medicine is needed.

Skin injury can be reduced by limiting exposure to the wavelengths that trigger your type. Clothing and physical barriers are often more useful than sunscreen alone.

Relatives of someone with an inherited porphyria may be offered genetic counseling and targeted testing. This cannot prevent the gene change, but it may identify people who should know about medicines or other potential triggers.

Living with it and what to expect

The long-term course depends strongly on the type. Many people who carry a gene change for acute porphyria never have symptoms. Others have one attack, widely separated attacks, or

recurrent episodes. Hormonal patterns and other triggers may change over time.

After a severe acute attack, abdominal symptoms may settle within days, but weakness or nerve pain can take much longer to improve. Rehabilitation, pain support, and follow-up may be needed during recovery.

Porphyria cutanea tarda often enters remission after treatment, although it can return if iron builds up again or contributing factors remain. Follow-up testing can detect a return before extensive skin damage develops.

Light sensitivity in erythropoietic protoporphyria often begins in childhood and continues through adult life. Planning clothing, travel, workspaces, and lighting can reduce exposure. Treatment may improve light tolerance, but it does not remove the inherited tendency.

Many people continue work, education, exercise, travel, and family life. Having a written plan for acute symptoms, medicine checks, and specialist follow-up can make the condition easier to manage.

When to get medical help

Seek emergency care for:

Tell the emergency team if you have diagnosed or suspected acute porphyria. If available, bring your emergency plan or medical alert information.

Arrange prompt medical assessment for new blistering or painful light sensitivity, jaundice, persistent dark urine, or worsening upper abdominal pain. These symptoms are not always caused by porphyria, but they should be evaluated.

Routine follow-up is used to review symptoms, check liver and kidney health, monitor treatment, and discuss safe medicines. Contact your usual care team sooner if attacks become more frequent or your established symptom pattern changes.

- Trouble breathing or rapidly worsening muscle weakness
- A seizure, fainting, severe confusion, or unusual drowsiness
- Severe abdominal pain with weakness, vomiting, or neurological symptoms
- Symptoms of an acute attack that are quickly getting worse

Common questions

Is porphyria a type of cancer?

No. Porphyria is a problem in the pathway that produces heme. Some acute hepatic porphyrias increase the long-term risk of liver cancer, but porphyria itself is not cancer.

What does a porphyria attack feel like?

A typical acute attack may involve severe, persistent abdominal pain with nausea, constipation, a fast heartbeat, anxiety, dark urine, or weakness. Symptoms differ, and testing during the episode is needed because many other conditions can feel similar.

Can porphyria go away?

Inherited gene changes remain for life, but symptoms may be absent for long periods. Porphyria cutanea tarda can enter remission after treatment. Transplantation can cure selected severe forms, but it is rarely needed.

Does sunlight trigger every type of porphyria?

No. Light causes symptoms in cutaneous porphyrias. It does not usually trigger the abdominal and nerve attacks of acute intermittent porphyria, which does not cause skin symptoms.

Can I take regular medicines if I have porphyria?

Usually, yes, but some medicines can trigger acute hepatic porphyria. Check new prescription, nonprescription, and herbal products using a specialist drug-safety resource. Do not stop prescribed treatment suddenly without advice.

Can my children inherit porphyria?

Some types can be passed to children, but the chance and likely effects depend on the exact type and inheritance pattern. In several acute porphyrias, inheriting the gene change does not mean that symptoms will develop.

Sources

- National Institute of Diabetes and Digestive and Kidney Diseases. Porphyria: Symptoms, causes, diagnosis, and treatment .
- European Association for the Study of the Liver. EASL Clinical Practice Guidelines on the management of porphyrias . Journal of Hepatology, 2023.
- Wang B, Bonkovsky HL, Lim JK, Balwani M. AGA Clinical Practice Update on Diagnosis and Management of Acute Hepatic Porphyrias: Expert Review . Gastroenterology, 2023.
- GeneReviews. Acute Intermittent Porphyria . National Center for Biotechnology Information.
- US Food and Drug Administration. Givlaari prescribing information .



- US Food and Drug Administration. Scenesse prescribing information .