Porphyria

National Digestive Diseases Information Clearinghouse

U.S. Department of Health and Human Services

NATIONAL INSTITUTES OF HEALTH

NIDDK
NATIONAL INSTITUTE OF DIABETES AND DIGESTIVE AND KIDNEY DISEASES

What is porphyria?

Porphyria is a term that refers to a group of disorders—the porphyrias—that affect the nervous system or skin, or both. Each type of porphyria is due to the deficiency of one of the enzymes needed to make a substance in the body called heme. Enzymes are proteins that help chemical reactions happen in the body. Making heme involves a series of eight different enzymes, each acting in turn.

Heme is a red pigment composed of iron linked to a chemical called protoporphyrin. Heme has important functions in the body. The largest amounts of heme are in the blood and bone marrow in the form of hemoglobin within red blood cells. Hemoglobin gives blood its red color and carries oxygen from the lungs to all parts of the body. In the liver, heme is a component of proteins that have many functions, including breaking down hormones, drugs, and other chemicals and generating high-energy compounds that keep liver cells alive and functioning normally.

The body makes heme mainly in the bone marrow and the liver. The process of making heme is called the heme biosynthetic pathway. Each step of the process is controlled by one of eight enzymes. If any one of the enzymes is deficient, the process is disrupted. As a result, porphyrin or its precursors—chemicals formed at earlier steps of the process—may build up in body tissues and cause illness.

What are the types of porphyria?

The table below lists each type of porphyria and the deficient enzyme responsible for the disorder. Porphyrias are often classified as

Types of Porphyria

Type of Porphyria	Deficient Enzyme
Acute Porphyrias	
ALAD porphyria	delta-aminolevulinic acid dehydratase
acute intermittent porphyria	porphobilinogen deaminase
hereditary coproporphyria	coproporphyrinogen oxidase
variegate porphyria	protoporphyrinogen oxidase
Cutaneous Porphyrias	
congenital erythropoietic porphyria	uroporphyrinogen III cosynthase
porphyria cutanea tarda	uroporphyrinogen decarboxylase (~50% deficiency)
hepatoerythropoietic porphyria	uroporphyrinogen decarboxylase (~90% deficiency)
hereditary coproporphyria	coproporphyrinogen oxidase
variegate porphyria	protoporphyrinogen oxidase
erythropoietic protoporphyria	ferrochelatase

acute or cutaneous. Acute types of porphyria affect the nervous system, whereas cutaneous types mainly affect the skin. Two forms of porphyria—hereditary coproporphyria and variegate porphyria—may be either acute or cutaneous, or both.

The most common type of porphyria overall is porphyria cutanea tarda. In the United States, acute intermittent porphyria is the most common acute porphyria.

What causes porphyria?

Most porphyrias are inherited disorders, meaning they are caused by abnormalities in genes passed from parents to children. Scientists have identified the genes for all eight enzymes in the heme pathway. Some forms of porphyria result from inheriting an abnormal gene from one parent. Other forms are due to inheriting two abnormal genes—one from each parent. The risk that members of an affected family will have the disease or transmit it to their children depends on the type of porphyria.

One type of porphyria—porphyria cutanea tarda—is most often an acquired disorder. It occurs when factors other than genes cause an enzyme deficiency in the liver.

Porphyria can be triggered by

- drugs such as barbiturates, tranquilizers, birth control pills, and sedatives
- chemicals
- fasting
- smoking
- drinking alcohol, especially heavy drinking
- infections
- excess iron in the body
- emotional and physical stress
- menstrual hormones
- exposure to the sun

What are the symptoms of porphyria?

People with cutaneous forms of porphyria develop blisters, itching, and swelling of their skin when it is exposed to sunlight. Symptoms of acute forms of porphyria include pain in the abdomen, chest, limbs, or back; numbness, tingling, paralysis, or cramping; vomiting; constipation; and personality changes or mental disorders. Acute attacks of porphyria can develop over hours or days and last for days or weeks.

Symptoms can vary widely in severity. Some people with gene mutations that can cause porphyria have no signs or symptoms of the disorder. These people are said to have latent porphyria.

How is porphyria diagnosed?

Doctors diagnose porphyria using blood, urine, and stool tests. Interpreting test results can be complex, and initial tests may be followed by further testing to confirm the diagnosis. Diagnosis may be delayed because the symptoms of porphyria are similar to symptoms of other disorders.

How is porphyria treated?

Each type of porphyria is treated differently. Treatment may involve avoiding triggers, receiving heme through a vein, taking medicines to relieve symptoms, or having blood drawn to reduce iron in the body. People who have severe attacks may need to be hospitalized.

Points to Remember

- Porphyria is a group of disorders—the porphyrias—that affect the nervous system or skin, or both.
- Each type of porphyria results from a deficiency of one of the enzymes needed to make heme.
- Most porphyrias are inherited disorders, but porphyria cutanea tarda is usually an acquired disorder.
- Cutaneous types of porphyria affect the skin, causing symptoms such as blistering, itching, and swelling.
- Acute types of porphyria affect the nervous system, causing symptoms such as abdominal pain, vomiting, numbness, and mental disorders.
- Each type of porphyria is treated differently.

Hope through Research

The National Institute of Diabetes and Digestive and Kidney Diseases conducts and supports research to help people with digestive diseases. A complete listing of clinical research studies, including those related to porphyria, can be found at *www.ClinicalTrials.gov*.

For More Information

For more information about porphyria, see the following online publications:

- *Learning about Porphyria*, available from the National Human Genome Research Institute at *www.genome.gov/19016728*
- *Porphyria*, available from the National Library of Medicine's Genetics Home Reference at *www.ghr.nlm.nih.gov/condition=porphyria*

Information about porphyria is also available from the following organizations:

American Porphyria Foundation
4900 Woodway, Suite 780
Houston, TX 77056–1837
Phone: 1–866–APF–3635 (273–3635)
or 713–266–9617
Fax: 713–840–9552
Email: porphyrus@aol.com
Internet: www.porphyriafoundation.com

National Organization for Rare Disorders
55 Kenosia Avenue
P.O. Box 1968
Danbury, CT 06813–1968
Phone: 1–800–999–6673
or 203–744–0100
Fax: 203–798–2291
Email: orphan@rarediseases.org
Internet: www.rarediseases.org

Iron Disorders Institute
2722 Wade Hampton Boulevard, Suite A
Greenville, SC 29615
Phone: 1–888–565–IRON (4766)
or 864–292–1175
Fax: 864–292–1878
Email: PatientServices@irondisorders.org
Internet: www.irondisorders.org

You may also find additional information about this topic using the following databases:

The NIDDK Reference Collection is a collection of thousands of materials produced for patients and health care professionals, including fact sheets, brochures, and audiovisual materials. Visit *www.catalog.niddk.nih.gov/resources*.

MedlinePlus brings together a wealth of information from the National Library of Medicine, the National Institutes of Health, and other government agencies and health-related organizations. MedlinePlus offers easy access to medical journal articles, a medical dictionary and medical encyclopedia, health information in Spanish, hospital and physician directories, drug and supplement lists, interactive patient tutorials, links to hundreds of clinical trials, and the latest health news. Visit *www.medlineplus.gov*.

This publication may contain information about medications used to treat a health condition. When this publication was prepared, the NIDDK included the most current information available. Occasionally, new information about medication is released. For updates or for questions about any medications, please contact the U.S. Food and Drug Administration at 1–888–INFO–FDA (463–6332), a toll-free call, or visit their website at *www.fda.gov*. Consult your doctor for more information.

This fact sheet is also available at *www.digestive.niddk.nih.gov*.

National Digestive Diseases Information Clearinghouse

2 Information Way
Bethesda, MD 20892–3570
Phone: 1–800–891–5389
Fax: 703–738–4929
Email: nddic@info.niddk.nih.gov
Internet: www.digestive.niddk.nih.gov

The National Digestive Diseases Information Clearinghouse (NDDIC) is a service of the National Institute of Diabetes and Digestive and Kidney Diseases (NIDDK). The NIDDK is part of the National Institutes of Health of the U.S. Department of Health and Human Services. Established in 1980, the Clearinghouse provides information about digestive diseases to people with digestive disorders and to their families, health care professionals, and the public. The NDDIC answers inquiries, develops and distributes publications, and works closely with professional and patient organizations and Government agencies to coordinate resources about digestive diseases.

Publications produced by the Clearinghouse are carefully reviewed by both NIDDK scientists and outside experts. This fact sheet was reviewed by Herbert L. Bonkovsky, M.D., Carolinas Health Care System.

U.S. DEPARTMENT OF HEALTH AND HUMAN SERVICES
National Institutes of Health

NIH Publication No. 08–4632
July 2008