



BASIC INFORMATION

DESCRIPTION

Any of a group of disorders in which the body has too much porphyrin. Porphyrin is a chemical that works with enzymes in the body to make heme. Heme is the part of blood that makes it red and carries the oxygen. Porphyrin can affect both sexes. Types include:

- Acute intermittent porphyria (AIP).
- Variegate porphyria (VP).
- Hereditary coproporphyria (HCP).
- ALAD dehydratase porphyria (ADP).
- Porphyria cutanea tarda (PCT).
- Erythropoietic porphyria (EPP).
- Congenital erythropoietic porphyria (CEP).
- Hepatoerythropoietic porphyria (HEP).

FREQUENT SIGNS AND SYMPTOMS

- Symptoms vary and can appear over hours or days. A variety of factors can trigger an attack. Symptoms usually affect the nervous system or the skin.
- Chest, abdominal, or leg pain.
- Muscle cramps and weakness.
- Numbness and tingling in the feet and hands.
- Emotional and mental changes including depression, mania, anxiety, agitation, confusion, and others.
- Skin changes, including itching, blistering, and sensitivity to the sun.
- Excessive hair growth.
- Urine turns color (dark-red, purplish, or brown).

CAUSES

Porphyrin builds up in the body due to enzyme deficiencies. Enzymes are required to complete the porphyrin-heme process. There are several different types of enzyme deficiencies. Most types are inherited. Others develop during a person's life.

RISK INCREASES WITH

Family history of porphyria.

PREVENTIVE MEASURES

- Cannot be prevented at present.
- Porphyria attacks may result from drugs, alcohol, smoking, infection, reduced calorie intake, stress (physical and emotional), excess iron, hepatitis C, pregnancy, and before a menstrual period. Talk to your health care provider about avoiding these factors where possible.

EXPECTED OUTCOMES

There is no cure. Some people with porphyria never have symptoms. In others, the symptoms may be mild to severe. Symptoms can usually be relieved with treatment. A few patients may have complications.

POSSIBLE COMPLICATIONS

A variety of complications may occur that cause physical problems as well as psychological problems.



DIAGNOSIS & TREATMENT

GENERAL MEASURES

- Your health care provider will do a physical exam and ask questions about your symptoms and activities. Medical tests are done to measure porphyrins in the urine, blood, and stool.
- Treatment involves medical care during attacks, preventing attacks, and counseling if needed. Specific treatment will depend on the type of porphyria.
- Don't take any drugs, herbs, or supplements without medical advice.
- Avoid bright sunlight. If you must be in bright sun, use a hat and protective clothing.
- Phlebotomy may be done. A unit of blood is removed on a regular basis to get rid of excess iron in the body.
- Hospital care may be required for severe symptoms.
- Wear a medical alert type bracelet or pendant identifying your medical problem.
- Genetic counseling before starting a family is advised.
- To learn more: American Porphyria Foundation, P.O. Box 22712, Houston, TX 77227, (713) 266-9617 (not toll free); website: www.porphyrifoundation.com.

MEDICATIONS

- Your health care provider may prescribe:
 - Intravenous (IV) glucose or heme to help prevent or treat acute attacks.
 - Drugs for depression or anxiety.
 - Drugs to reduce nausea and vomiting.
 - Drugs to reduce the excess porphyrin in the body.
 - Drugs to help reduce premenstrual attacks.
 - Drugs for anemia (if needed).
 - Beta carotene for skin symptoms.

ACTIVITY

No limits except for the sunlight restrictions.

DIET

Eat a normal or high carbohydrate diet. If weight loss is desired, ask your health care provider for diet advice.



NOTIFY OUR OFFICE IF

- You or a family member has symptoms of porphyria.
- Any symptoms of an attack occur.
- New, unexplained symptoms develop.

Special notes:

More notes on the back of this page