Porphyria

Porphyria is a group of disorders. It is caused by problems with how your body produces heme. Heme is a substance found in all body tissues. The largest amounts of heme are in the red blood cells, bone marrow, and liver. Your body uses special proteins (called *enzymes*) to convert chemicals called porphyrins into heme. If any one of the enzymes is abnormal, the process cannot continue. These porphyrins may build up and cause symptoms. The skin and nervous system are most often affected.

Porphyria is usually broken down into 2 categories:

- **Acute porphyrias** - include forms of the disease with mostly nervous system symptoms (neuron porphyrias). In some cases, there are both nervous system and skin symptoms (neurocutaneous porphyrias). Your body can be damaged in several ways from the acute porphyrias. High blood pressure, kidney failure, and liver cancer can result over time.
- **Cutaneous porphyrias** - include forms of the disease that cause skin symptoms without affecting your nervous system.

SYMPTOMS

Specific signs and symptoms depend on the type of porphyria.

People with **cutaneous porphyrias** can develop:

- Blisters, itching, and swelling of their skin when it is exposed to sunlight.

People with **Acute porphyrias** can develop:

- Pain in the chest, abdomen, limbs, or back.
- Other symptoms include muscle numbness, tingling, paralysis, cramping, vomiting, constipation, and personality changes.
- Different psychiatric symptoms can include paranoia, hallucinations, confusion, anxiety, depression, and violent behavior.

These **acute porphyrias** symptoms may appear intermittently. Other possible symptoms include:

- Unstable vital signs.
- Excessive sweating.
- Painful urination and other bladder problems. In some cases, urine may turn red or dark when exposed to light.
- Fever.
- Restlessness.
- Tremors.
- Vision problems (including blindness).
- Seizures.

In cases of neurocutaneous porphyrias, both skin and nervous system symptoms can occur at the same time. Attacks of porphyria can develop over hours or days. The attacks can last for days or weeks.

CAUSES

The porphyrias are passed from your parents to you (*inherited*).

- Some forms of porphyria result from inheriting an abnormal gene from one parent (*autosomal dominant*).
- Other forms are from inheriting an abnormal gene from each parent (*autosomal recessive*).
- The risk that individuals in an affected family will have the disease or transmit it to their children is different depending on the type. Porphyria can be triggered by:
  - Emotional and physical stress.
  - Chemicals.
  - Drinking alcohol.
  - Menstrual hormones.
  - Birth control pills.
  - Fasting.
  - Infections.
  - Exposure to the sun.
  - Sedatives.
  - Smoking.
  - Emotional and physical stress.
  - Drugs (barbiturates, tranquilizers).

DIAGNOSIS

Diagnosis may be difficult because the range of symptoms is common to many disorders. Porphyria is diagnosed through blood, urine, and stool tests. Interpretation of the tests may be complex. If tests are positive

for porphyria, specific results can narrow down the exact type that exists.

TREATMENT

Treatment must be specific to the person. Initial treatment can include:

- Stopping medicines that may have started the attack.
- Medicines to help relieve pain, nausea, and vomiting.
- A high carbohydrate diet and good hydration is essential.

Hospitalization is required if there is a severe attack. Medicines to help pain and nausea may be given through an intravenous line. Other important medicines that may be needed include:

- Hematin (a form on heme).
- Anti-seizure medicines, if needed.
- Medicines to control psychiatric symptoms.

HOME CARE INSTRUCTIONS

- Eat a high carbohydrate diet.
- Eat a low salt and fat diet if there are problems of high blood pressure.
- Avoid alcoholic beverages.
- Avoid smoking.
- For women, there may be a benefit in using birth control pills to control your periods.

SEEK MEDICAL CARE IF:

- You develop a recurrence of any of the symptoms noted above.
- You have been advised to start any new prescription medicines. There is a long list of medicines that can possibly trigger an attack or porphyria. Talk to your caregiver to make sure a new medicine is not included in that list.

SEEK IMMEDIATE MEDICAL CARE IF:

You develop a recurrence of any symptoms noted above not helped by the “initial treatment” recommendations described above.

FOR MORE INFORMATION

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