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## Porphyria

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### Summary

- Porphyria is the umbrella term for a group of rare disorders that involve a particular molecule called 'heme' or 'haem'.
  - Porphyria can affect the skin, nervous system, gastrointestinal system or all of these, depending on the specific type.
  - There is no cure, but medical treatment and lifestyle changes can usually manage the symptoms.
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Porphyria is the umbrella term for a group of rare disorders that involve a particular molecule called 'heme' or 'haem'. Heme contains iron and is used in metabolic processes throughout the body. Porphyria occurs when the body cannot convert naturally occurring compounds (called 'porphyrins') into heme.

While all tissues have heme, those that use it the most are the red blood cells, liver and bone marrow. Porphyria can affect the skin, nervous system and gastrointestinal system, depending on the specific type.

In most cases, the cause is a combination of genetic and environmental factors. More women than men are affected for reasons unknown. There is no cure but treatments are available to manage the symptoms.

### Symptoms

Symptoms vary from one type of porphyria to the next. Cases are generally classified into one of three groups, which include:

- **Acute porphyrias** – the condition mostly affects the nervous system. The skin is occasionally affected. Symptoms may include muscle pain or paralysis, seizures, disorientation, hallucination, bloody (red) urine, hypertension and gastrointestinal problems such as vomiting, abdominal pain and constipation. Acute porphyrias generally occur during adulthood and are rare before puberty or after menopause. Different types of acute porphyria include 'acute intermittent porphyria' and 'erythropoietic protoporphyria'.
- **Cutaneous porphyrias** – the condition affects the skin but not the nervous system. The skin is highly sensitive to sunlight and exposure tends to trigger symptoms within minutes. Symptoms may include red, itchy, blistered, painful and swollen skin and bloody (red) urine. The condition may develop during childhood. Different types of cutaneous porphyria include 'porphyria cutanea tarda' and 'hepatoerythropoietic porphyria'.
- **Neurocutaneous porphyrias** – the condition affects both the skin and the nervous system. Sunlight exposure tends to rapidly trigger symptoms. Different types of neurocutaneous porphyria include 'variegate porphyria' and 'hereditary coproporphyria'.

### Porphyrins build up in the body

The substance heme (or haem) is used in various metabolic processes. The body makes heme from porphyrins, which are metallic compounds found naturally in the tissues of animals and plants. The conversion of porphyrins into heme requires the action of special proteins called enzymes. Genes control the action of enzymes. A flawed gene (or genes) can stop the body from making one or more of these enzymes. This creates a lack of heme and a build-up of porphyrins, which causes the signs and symptoms of porphyria.

### Inherited genes

Most forms of porphyria are inherited, which means the genetic predisposition is passed from one generation to the next. The faulty gene interferes with the body's ability to create one or more enzymes necessary in the conversion of porphyrins into heme. The pattern of inheritance may include:

- **Autosomal dominant inheritance** – the faulty gene is inherited from one parent. This faulty gene overrides the healthy gene inherited from the other parent.
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- **Autosomal recessive inheritance** – the faulty gene is inherited from both parents.

However, about nine in every 10 people with the faulty gene or genes don't have porphyria. It appears that an environmental trigger is needed to allow porphyria to develop.

### **A range of triggers**

Various triggers can prompt the development of porphyria. While the factors in the following list may seem to have nothing in common, each one demands increased heme production, which overwhelms the body's ability to deal with the increased levels of porphyrins.

Common triggers include:

- Prescription drugs such as barbiturates, tranquillisers, sedatives, oral contraceptives and some types of antibiotics
- Female sex hormones
- Sunlight
- Alcohol
- Cigarette smoking
- Infection
- Surgery
- Fasting.

### **Common complications**

Without medical treatment, complications of porphyria may include:

- Permanent hair loss
- Skin scarring
- Permanent skin pigmentation changes
- Dehydration
- Breathing problems
- High blood pressure (hypertension)
- Low salt levels in the blood (hyponatremia)
- Kidney failure
- Liver problems, which may require a liver transplant in severe cases.

### **Diagnosis**

Since porphyria is rare, most doctors are unfamiliar with it and may not recognise the symptoms. Diagnosis can be delayed because porphyria mimics the symptoms and signs of various other medical conditions such as Guillain-Barre syndrome, eczema, multiple sclerosis and irritable bowel syndrome. Diagnostic tests may include:

- Physical examination
- Medical history
- Urine tests to check for elevated substances including porphyrins
- Blood tests to check for high levels of porphyrins in the plasma
- Stool sample to check for excreted porphyrins
- Genetic test.

### **Treatment – acute porphyria**

Treatment may include:

- Pain medication
- Addressing the underlying cause – for example, prescribing antibiotics to treat an infection or ceasing a particular medication
- Medication called 'hematin', which is a type of heme the body can use

- Intravenous fluids and glucose
- Admission to hospital in severe cases.

### **Treatment – cutaneous porphyria**

Treatment may include:

- Oral administration of activated charcoal, which helps to absorb excess porphyrins
- Daily supplementation with beta-carotene (vitamin A) as part of long-term treatment.

### **Self-care options**

Be guided by your doctor, but general suggestions include:

- In all cases avoid known triggers – for example, don't smoke.
- When out in the sun, wear sunglasses, a brimmed hat, a long-sleeved top and long pants. Apply SPF 30+ sunscreen to exposed skin areas.
- Protect your skin every day. For example, wear rubber gloves when handling chemicals or very hot water. Avoid perfumed soaps. Regularly apply barrier cream to the hands.
- Eat regular meals and avoid alcohol.
- You may like to consider wearing a medical alert bracelet or pendant, since surgery and some drugs can provoke symptoms.

### **Where to get help**

- Your doctor
- In an emergency, call triple zero (000)
- NURSE-ON-CALL Tel. 1300 60 60 24 – for expert health information and advice (24 hours, 7 days)
- Porphyria Association Inc. Tel. (03) 9845 2737
- Genetic Support Network of Victoria Tel. (03) 8341 6315
- Australasian Genetic Alliance Tel. (02) 9211 1462

### **Things to remember**

- Porphyria is the umbrella term for a group of rare disorders that involve a particular molecule called 'heme' or 'haem'.
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