Porphyria

Summary

- Porphyria is the name for a group of rare conditions that involve a molecule called ‘heme’ or ‘haem’.
- Heme contains iron and is used in metabolic processes throughout the body.
- Porphyria occurs when the body cannot convert compounds called ‘porphyrins’ into heme.
- Porphyria can affect the skin, nervous system, gastrointestinal system or all of these, depending on the specific type.
- Currently, there is no cure, but medical treatment and lifestyle changes can usually manage the symptoms.

Porphyria is the umbrella term for a group of rare conditions that involve a molecule called ‘heme’ or ‘haem’. Heme contains iron and is used in metabolic processes throughout the body. Porphyria occurs when the body cannot convert 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.

More women than men are affected by porphyria.

In most cases, a combination of genetic and environmental factors trigger porphyria symptoms. There is no cure for porphyria, but by avoiding triggers, development of symptoms of porphyria can be prevented. Treatments are available to manage the symptoms.

Symptoms of porphyria

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

- **Acute porphyrias** – this condition mostly affects the nervous system. The skin is occasionally affected. 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’. Symptoms of acute porphyrias may include:
  - muscle pain or paralysis
  - seizures
  - disorientation
  - hallucination
  - bloody (red) urine
  - hypertension
  - gastrointestinal problems such as vomiting, abdominal pain and constipation

- **Cutaneous porphyrias** – this condition affects the skin but not the nervous system. The skin is highly sensitive to sunlight and exposure tends to trigger symptoms within minutes. Cutaneous porphyrias may develop during childhood. Different types of cutaneous porphyria include ‘porphyria cutanea tarda’ and ‘hepatoerythropoietic porphyria’. Symptoms may include:
  - red, itchy, blistered, painful and swollen skin
  - bloody (red) urine

- **Neurocutaneous porphyrias** – this 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’.

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What causes porphyria

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. An altered 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.

Porphyria is usually inherited

Most forms of porphyria are inherited. This means the genetic predisposition for the condition is passed from one generation to the next. The altered gene interferes with the body’s ability to create one or more enzymes needed to turn porphyrins into heme. The pattern of inheritance may include:

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

Porphyria needs an environmental trigger

Not everyone who has the altered gene will develop porphyria. It appears that an environmental trigger is needed for porphyria to develop. Triggers and responses vary between individuals.

All triggers for porphyria involve increased heme production.

Common triggers include:

- prescription drugs such as barbiturates, tranquillisers, sedatives, oral contraceptives and some types of antibiotics
- female sex hormones that arise at puberty
- artificial hormones such as those used in common female contraceptives
- sunlight
- alcohol
- cigarette smoking
- infection
- surgery
- fasting.

Prevention of porphyria symptoms

Be guided by your doctor, but general suggestions for preventing symptoms of porphyria include:

- In all cases avoid known triggers – for example, don’t smoke or drink alcohol.
- 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.
- Consider wearing a medical alert bracelet or pendant, as surgery and some medications can provoke symptoms.

Diagnosis of porphyria

As porphyria is rare, most doctors are unfamiliar with it and may not recognise the symptoms. Porphyria mimics the symptoms and signs of other medical conditions such as Guillain-Barre syndrome, eczema, multiple sclerosis and irritable bowel syndrome. This can delay diagnosis.

Diagnostic tests for porphyria 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 testing.

**Common complications of porphyria**
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.

**Treatment – acute porphyria**
Treatment for acute porphyria 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 for cutaneous porphyria 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.
• therapeutic phlebotomy – a healthcare practitioner can remove blood to reduce iron concentration in the liver.

**Genetic counselling and porphyria**
If you or your child have been diagnosed with porphyria, or if it runs in your family, it may be helpful to speak to a genetic counsellor.

Genetic counsellors are health professionals qualified in both counselling and genetics. As well as providing emotional support, they can help you to understand porphyria and what causes it, how it is inherited, and what a diagnosis means for your child's health and development, and for your family. Genetic counsellors are trained to provide information and support that is sensitive to your family circumstances, culture and beliefs.

If porphyria runs in your family, a genetic counsellor can explain what genetic testing options are available to you and other family members. You may choose to visit a genetic counsellor if you are planning a family – to find out your risk of passing the condition on to your child, or to arrange for prenatal tests.

The **Genetic Support Network of Victoria** (GSNV) is connected with a wide range of support groups throughout Victoria and Australia and can connect you with other individuals and families affected by porphyria.

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Where to get help

- In an emergency, call triple zero (000) for an ambulance
- Your **GP (doctor)**
- **NURSE-ON-CALL** Tel. **1300 60 60 24** – for expert health information and advice (24 hours, 7 days)
- **Porphyria Association Inc.** Tel. **0413 757 892**
- **Genetic Support Network of Victoria** Tel. **(03) 8341 6315**
- **Genetic Alliance Australia** Tel. **(02) 9295 8359**
- **Victorian Clinical Genetics Services** Tel. **1300 118 247**

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