Life doesn't stop

A guide to living with acute porphyria and staying informed

for patients and their families

Provided as a service to patients
by Recordati Rare Diseases.

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Introduction

If you, or someone close to you, have been diagnosed with a form of acute porphyria, this leaflet will help to explain exactly what the condition is, why it occurs and what can be done to help those who live with it.

It is designed to give you the basic facts, but if, after reading the leaflet, you have unanswered questions, please consult your doctor or a healthcare professional for further information.

What is acute porphyria?

Our bodies in balance

A healthy body needs a constant supply of essential elements, and we produce just enough enzymes to control how these important supplies are used. It's a system that is finely tuned and in perfect balance in healthy individuals.

One of the most familiar of essential elements is iron and our bodies use iron to make hemoglobin – the substance that colors blood red and carries oxygen to all major organs, including the brain and the heart.

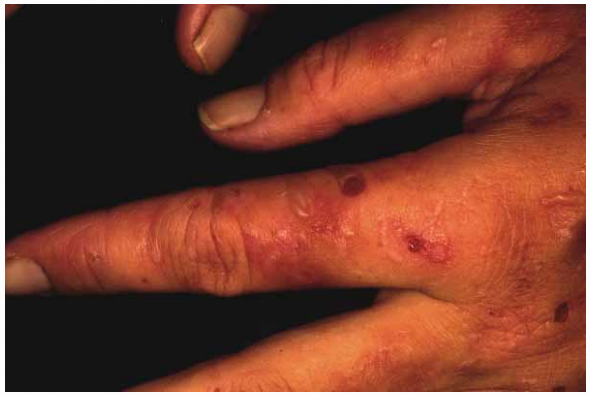
In order to make hemoglobin with the iron that we take in as part of our diet, our bodies make proteins called porphyrins.

In people with porphyria, the system is out of balance and there is an over-production of porphyrins. The excess porphyrins build up inside the body, giving rise to unpleasant symptoms. If they build up in the liver, for example, it can cause severe abdominal pain.

These unpleasant symptoms usually have a rapid onset and last for a relatively short period and that's how doctors define an 'acute' attack – hence the name acute porphyria.

Certain types of acute porphyria (see 'Types') involve a build-up of porphyrins in the skin. When that happens, it can make the skin sensitive to sunlight and result in painful sores and blisters.

Typical skin lesions



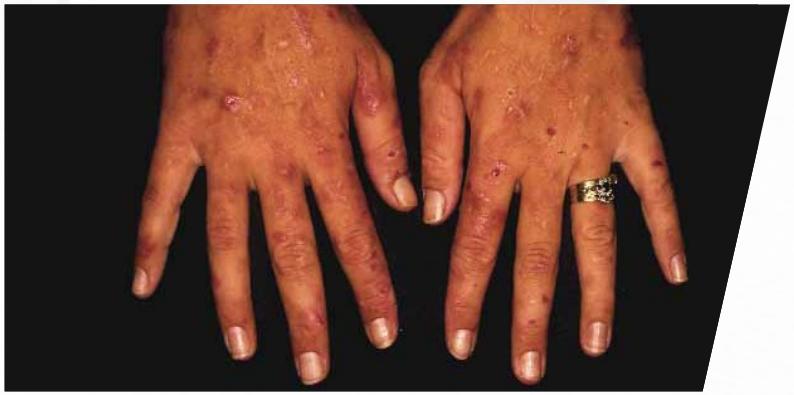


Image source: DermNet NZ

What happens during an attack?

Acute attacks almost always start with severe unexplained pain, usually in the abdomen but sometimes in the back or thighs. This may be preceded by anxiety, restlessness and insomnia. It's also quite common to experience nausea, vomiting and constipation.

Acute porphyria can also affect the nervous system and some people develop muscle weakness. In the chest wall, this can lead to breathing difficulties. Effects on the nervous system can lead to speeding up of heart rate and increased blood pressure too.

These symptoms can become very severe and even life-threatening if not managed early, so it's vital that a doctor is seen as soon as an attack begins.

> "As a teenager, I began to have extremely severe abdominal pain and rapid pulse and even a little bit of confusion. But this pain was so severe that I'd never felt anything like it. I was doubled over in pain."

Desiree *Acute Intermittent Porphyria patient*

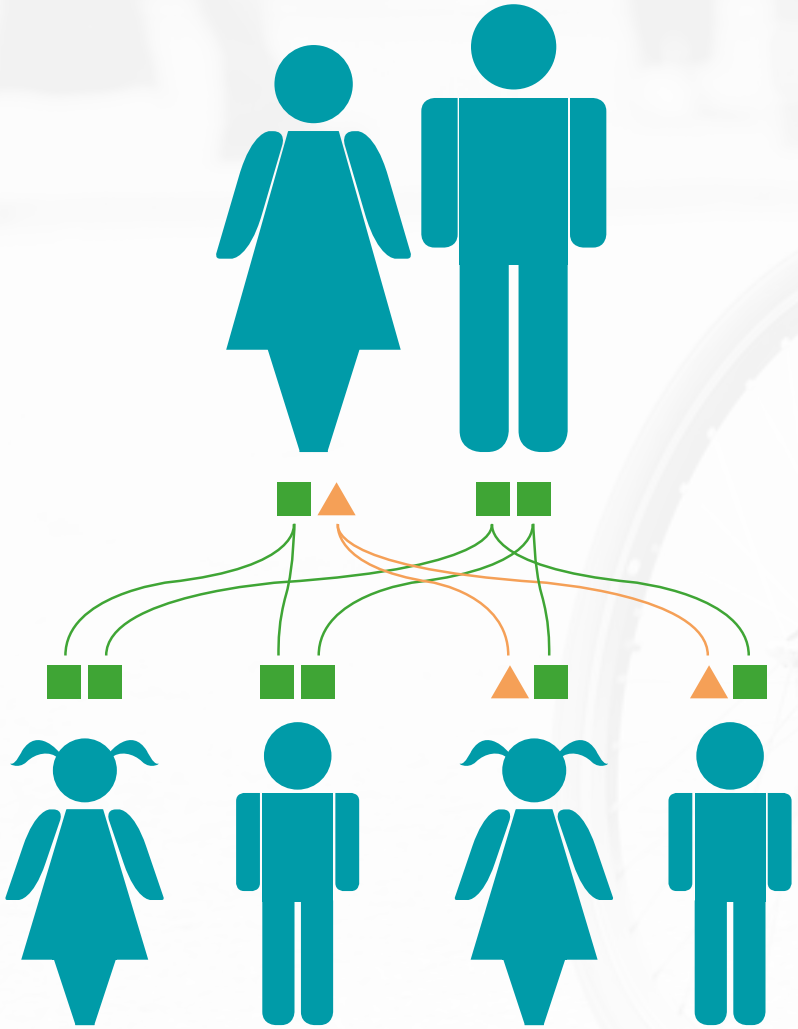
Can anyone get acute porphyria?

Acute porphyria is inherited so is passed down in the genes, usually from one parent and very rarely from both parents. It's rare, with about 1 in 75,000 people in European countries affected by the most common type of porphyria.[1]

If someone has inherited a faulty gene, it does not necessarily mean that they will suffer from acute attacks; in fact most people with the gene DON'T experience them. That means that the gene can be passed on through the generations without an individual realizing that it is present in the family – until someone has an acute attack. There is currently no way of predicting who will suffer from acute attacks.

1. Puy et al. (2010), Porphyrias, *Lancet;* 375: 924-937

The diagram below explains how acute porphyria may be inherited by some children in a family and not by others.



Father and 2 children have not got porphyria

Mother and 2 children have acute porphyria and are at risk of suffering from an acute attack

This is an example of the inheritance of an acute porphyria. In this diagram the mother is suffering from porphyria. She is carrying one normal gene ■ and one faulty (mutated) gene ▲. Of course, in another family it could be the father suffering from porphyria. In each case the probability of each child inheriting porphyria is one in two (50%).

What's the connection between genes and acute porphyria?

We depend on the genes that we inherit from our parents, as they code for all proteins and carry the information needed to build and maintain our cells and pass our genetic traits to our children. These traits include those that are visible, such as eye and hair color, and those that are not, such as blood type and the thousands of basic chemical processes that comprise life.

Sometimes, individual genes can change (mutate) and become faulty, so that the proteins they code for can't perform the tasks that they are supposed to. This is what's happened in acute porphyria: the gene that usually codes for a protein involved in the production of hemoglobin has become faulty, leading to a build-up of porphyrins.

Will it always be passed on to the next generation?

Medical science isn't advanced enough to be able to "fix" a faulty gene, although this is an active area of research.

This means that anyone with acute porphyria will be at risk of passing it on to any children they may have, just like their parents passed it on to them.

If anyone knows – or realizes after experiencing an acute attack – that a gene for acute porphyria runs in their family, it's important that a geneticist tests the whole family so that all those at risk of an acute attack can be identified.

> "Because my sister had the condition and because we had family awareness, I had genetic testing at the age of 16, so I knew I had the gene."

Sue *Acute Intermittent Porphyria patient*

Are there different types of acute porphyria?

Three main types of acute porphyria and one extremely rare type have been identified, all of which are inherited. They differ in the genes that are faulty, which code for different proteins in the same pathway but all of which lead to a build-up of porphyrins.

Most people with an acute porphyria never experience any health problems. However, about one in five people with an acute porphyria will experience an acute attack.[1] Females are more likely to suffer than males and the most common age for attacks to occur is between the late teens and early 40s.

1. Ventura et al. (2014), A challenging diagnosis for potential fatal diseases: recommendations for diagnosing acute porphyrias, *Eur J Intern Med;* 25: 497-505

Types of acute porphyria

1. Acute Intermittent Porphyria (AIP)

This is the most common type. The symptoms of an AIP attack and their severity vary greatly, but the skin is never affected. Most people make a full recovery from an attack, although about 1 in every 10 people with an attack (mostly women) will suffer repeat attacks.[1]

2. Variegate Porphyria (VP)

People with VP are at risk of both acute attacks and experiencing skin problems, not necessarily at the same time. VP tends to be more common in certain societies and countries around the world and much less so in others. In South Africa for instance, it is estimated that around 1 in 300 of the European immigrant population have the condition.[2] It can affect both men and women, although symptoms are more common in women.

3. Hereditary Coproporphyria (HCP)

HCP is about seven times less common than AIP[3] and the major difference is that people with HCP can have acute attacks at the same time as skin problems.

4. Aminolevulinate dehydratase (ALAD) deficiency porphyria (ADP)

This type is sometimes called plumboporphyria and is extremely rare, but is similar in outlook to AIP.

1. Puy et al. (2010), Porphyrias, *Lancet*; 375: 924-937
2. Hift RJ, Meissner PN (2005) An analysis of 112 acute porphyric attacks in Cape Town, South Africa: Evidence that acute intermittent porphyria and variegate porphyria differ in susceptibility and severity, *Medicine (Baltimore)*; 84(1):48-60
3. Elder et al. (2013), The incidence of inherited porphyrias in Europe, *J Inherit Metab Dis;* 36: 849-857

How is acute porphyria diagnosed?

Unexplained, severe abdominal pain will always initiate urgent tests from the patient's doctor, but because acute porphyria is relatively rare, it may take some time for the diagnosis to be reached.

However, there is one simple diagnostic 'test' that can aid diagnosis – the color of the patient's urine sample on exposure to light. If the urine turns dark on exposure to sunlight (after around 30 minutes), this may indicate that the patient is experiencing the onset of an acute attack. However, the urine may not turn dark even if the patient is experiencing an acute attack, so a doctor should be consulted, who will carry out further tests on urine, blood and stool samples. They can then confirm the diagnosis and determine which type of acute porphyria is present.

These tests measure the concentrations of porphyrins and related chemicals that will be high if the patient is suffering an acute porphyria attack. It is important that these tests, and the urine color 'test,' are performed as soon as possible after the onset of an acute attack. The concentrations of porphyrins may quickly decrease after an acute attack, so if samples are not taken at the right time, acute porphyria may be missed.

To help with diagnosis, it's important for the doctor to be given as much information as possible about the health of relatives, especially parents and siblings.

Can acute porphyria be cured?

At present, there is no cure for acute porphyria, but there are now medicines available that can help to lessen the severity of attacks and possibly prevent future attacks.

Anyone living with an acute porphyria, or anyone who thinks that they may have inherited it from their parents, should talk to their doctor about a confirming diagnosis and potential treatment.

> "The fact that I knew I had the gene meant that I knew what kind of treatment I needed."

Sue *Acute Intermittent Porphyria patient*

Can an acute attack be predicted or avoided?

Many acute attacks are triggered by avoidable causes such as alcohol, medications and low calorie diets. Variations in hormone levels are also a common factor, which is why females tend to experience attacks more than males.

Alcohol – if someone knows they carry an acute porphyria gene, then absolute avoidance of alcohol, in particular heavy red wines, brandy and other liqueurs, is recommended, although a compromise is to keep intake as low as possible.

Drugs – many medications contain ingredients that are capable of inducing an acute attack, so the safety of any medicine or remedy must ALWAYS be checked before it is taken, including any medicines prescribed by a doctor for other conditions, over-the-counter treatments and herbal remedies. Basic lists of safe drugs for acute porphyria patients can be found online, for example that compiled by the European Porphyria Network. However, the lists are subject to change, so a doctor should be the first source of information for which medicines are safe for use by acute porphyria patients.

If an acute porphyria patient requires surgery, it is important to inform the surgeon and anesthetist in advance so that a special anesthetic that has been proven to be safe for acute porphyria patients can be used. Dentists, who often use local anesthetics, should also be informed.

All vaccines are safe for use in acute porphyria patients.

Diet – using low calorie diets to reduce weight and prolonged periods with little food may provoke an acute attack in people with an acute porphyria gene, so it is important to maintain a healthy body weight and stick to a normal or high carbohydrate diet with at least three regular meals a day. Patients who have experienced an acute attack should consult a dietician for advice about how to achieve this balance.

The contraceptive pill and hormone replacement therapy (HRT) – Women are more prone to acute attacks than men due mostly to female hormones, particularly progesterone. This is found in the combined oral contraceptive pill, as well as in HRT. Women with acute porphyria should avoid contraceptives[1] and HRT containing progesterone.

Pregnancy – although most pregnancies are uneventful, there is a slightly increased risk of an acute attack during or after pregnancy, so it is important to inform the doctors providing prenatal care. Women who have experienced a severe acute attack that required hospital care should delay pregnancy until at least 12 months after recovery from the attack.

1. Calvo de Mora Almazán et al. (2012), Acute porphyria in an intensive care unit, *Emergencias;* 24: 454-458

What is the advice for those who suffer repeat attacks?

If an acute porphyria patient experiences more than one acute attack every year, there are several steps that should be taken to avoid attacks happening, and to deal with them when they do happen.

- Avoid alcohol and recreational drugs
- Pay attention to diet
- Consult a doctor about pain management
- Increase carbohydrate intake (sweet or starchy food and drinks) as soon as the symptoms of an acute attack are recognized
- Keep in contact with a doctor, and consult them if symptoms are severe or don't settle within 24 hours
- Consider having the whole family tested for acute porphyria genes so that others at risk of experiencing acute attacks can be identified

Is it possible to speak to other people with acute porphyria?

There are lots of patient support and community groups for people with acute porphyria and they can be relied upon for help, support and information. People with acute porphyria often comment on how welcoming and supportive these groups are.

Some of them are listed here, although there may be others that you find are of particular help to you personally. But remember, the most trusted and first option should be to talk to a doctor or other healthcare professional.

A message from a doctor who truly understands porphyria

Dr Lisa Kehrberg is a practicing clinician in the USA who understands what patients experience because she herself has Acute Intermittent Porphyria.

WATCH VIDEO >

"Porphyria pain is neuropathic pain and this type is the most severe that anyone can experience. It's not comparable to any kind of pain I've ever experienced in my life. Unless you've experienced the extreme, intense neuropathic pain of an acute porphyria attack, then it would be very difficult to truly understand the suffering.*

Fortunately there are treatment options for porphyria, including early, appropriate pain management and then also treatment for the disease."

*Neuropathic pain is different to the common type of pain caused by injury; instead, it comes from problems with signals from affected nerves, which send pain messages to the brain.

www.thinkporphyria.org

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*Laboratories listed on the European Porphyria Network (EPNET) website www.porphyria-europe.org. EPNET has set up a network of specialist porphyria laboratories in which there is collaboration and a quality assurance scheme. These laboratories are able to distinguish, using biochemical testing, between all types of porphyria and are able to offer specialist detailed interpretation of results with clinical advice on management.

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