

Introduction - What is Porphyria?

The 'Porphyrias' are a group of seven rare genetic disorders. They are called the porphyrias because they cause accumulation of chemicals called porphyrins (purple-red pigments named from the Greek for purple); or the simpler chemicals (ALA and PBG), which are used by the body to make porphyrins. In each porphyria a specific enzyme, which is needed to complete each step on the pathway to produce Haem (a red pigment containing iron and porphyrin), is deficient. As a result these porphyrins accumulate causing severe medical problems. The type of porphyria varies according to the enzyme/step which is affected.

Acute Porphyria

Acute porphyria is the term used for three similar inherited disorders: Acute Intermittent Porphyria (AIP), Variegate Porphyria (VP), Hereditary Coproporphyria (HCP), ALA-dehydratase Deficiency Porphyria (ADP). They are grouped together because acute attacks of porphyria occur in each one. These attacks are uncommon and are often difficult to diagnose. In most European countries, about 1 in 75,000 people suffer from them. AIP is the commonest type. In AIP, only acute attacks occur and the skin is never affected. If you have variegate porphyria or hereditary coproporphyria, your skin may also be affected.

Acute Attacks

- Acute attacks almost always start with severe pain - usually in the abdomen but may also be felt in the back, arms or legs.
- Nausea, vomiting, and constipation are also very common.
- Low levels of sodium (salt) in the blood are often found.
- Pulse rate and blood pressure may increase but rarely to dangerous levels.
- Some people may become very confused during an acute attack and later find it difficult to remember details of their illness.
- Convulsions and muscular weakness, which may lead to paralysis, can occur, often several weeks later.
- An acute attack usually lasts for no longer than one or two weeks, but may be life threatening because of severe neurological complications like motor paralysis. If paralysis occurs, recovery is gradual but slow.

Occurrence

Acute attacks are often provoked by drugs, alcohol, low food intake or hormones. Infections or stressful situations may also trigger an acute attack.

The most common age for an acute attack is from the late teens to the forties. They are extremely rare in children before puberty. Most people have only one or a few acute attacks; only a minority suffer repeated attacks, sometimes over several years. Although acute attacks can be very severe, particularly if precipitated by drugs or alcohol, nowadays they are rarely fatal.

Most people who have one or a few attacks of acute porphyria make a full recovery. They are then able to lead a perfectly normal life, however, they need to take a few simple precautions to reduce the risk of having another attack.

How is acute porphyria diagnosed?

Samples of urine, blood and stools (faeces) need to be carefully tested in a laboratory which specialises in porphyria. This can identify the type of porphyria. It is important that these tests are carried out as soon as possible after the start of the illness, as an accurate diagnosis may be difficult after recovery from an acute attack, especially after several months or years.

In a patient who is known to have inherited an acute porphyria, it may be difficult to decide whether an illness is an attack of acute porphyria or caused by something else. In this situation, urine and other tests can help your doctor to decide whether you are suffering from your porphyria or some other illness.

It is important that people with porphyria should not assume that all their illnesses are porphyria. If they do, common but potentially serious conditions like appendicitis may be overlooked.

How can I reduce the risk of attacks?

Many acute attacks are precipitated by drugs, alcohol, fasting (including dieting and gastric infections), or hormones. Infections and stressful situations may also precipitate an acute attack.

Drugs – Stick to drugs on the safe list except in extreme need. Drugs not on the safe list should only be taken after obtaining expert advice.

It is important to ALWAYS check the safety of any medicine or remedy with your doctor.

Many drugs can trigger attacks, including prescription medicines, as well as over-the-counter treatments, tonics and herbal remedies. While over 100 drugs are considered suspect, there are many other drugs available and good alternatives can almost always be found. (Please see our Drugs and Porphyria leaflet or www.porphyria-europe.com.)

Even though acute attacks are very rare before puberty, it is safest for children if they also avoid all drugs that are not known to be safe in porphyria.

Alcohol – This is a common trigger for attacks, so the best advice is don't drink. This is particularly important for those who have had attacks. Even if you have not had an attack, it is still safest to avoid alcohol. If you do drink, keep intake low and avoid heavy red wines, brandy and other liqueurs.

Diet - Low calorie diets, such as those used to reduce weight, or prolonged periods with little food may provoke an acute attack. It is therefore important to keep to a normal diet with regular meals, eating enough to maintain a desirable body weight. At least three regular meals should be taken each day. Some people, particularly women with pre-menstrual problems, may find it easier to eat small meals every three hours rather than three normal sized meals.

Particular issues for women

Women are at least three times more likely than men to experience an acute attack, mostly due to female hormones, particularly progesterone. This hormone is found in the pill, as well as in hormone replacement therapy (HRT). Women with an acute porphyria should avoid oral contraceptive and HRT preparations containing progesterone or related compounds (progestogens), if at all possible. Injectable and implantable long-term hormone preparations are very dangerous and must always be avoided.

In special circumstances, where the risk is low and the benefits high, your doctor may consider (after discussion with you and a porphyria specialist) the use of progestogen-containing preparations. Replacement doses, given through the skin from patches, are preferred as they can be rapidly removed.

Pre-menstrual symptoms

In some women, attacks are related to the pre-menstrual phase of the menstrual cycle. One possible treatment is to suppress your periods for up to two years.

Pregnancy

Though nearly all pregnancies are uneventful, there is a small increased risk of having an acute attack during or after pregnancy. However, the chances and dangers of such an acute attack are much diminished if porphyria has been previously diagnosed. It is therefore very important for the doctors providing care during pregnancy to know that you have an acute porphyria.

Treatment of acute attacks

Early recognition of an acute attack allows early treatment. The early symptoms are often easily recognised by those who have previously experienced an attack. When these symptoms are recognised you should stop any provoking agents such as any kind of medication. Immediate intake of sugary substances such as soft drinks or glucose tablets may help to reduce the severity of the symptoms.

As soon as an acute attack is suspected, you should contact your physician for rapid hospitalisation if the acute episode is severe. This will allow for:

- Diagnosis of the acute attack by measuring PBG in urine.
- Early start of specific treatment of the acute attack: for example, with intravenous human haemin or Haem arginate.
- Treatments for the various symptoms accompanying the attack. These are likely to include drugs to relieve pain and nausea and to provide sedation. It is also important to maintain an adequate intake of calories and this may require feeding intravenously or through a nasogastric tube.

Haem arginate is now one of the first choices of treatment in the UK. This helps to overcome the relative deficiency of haem in the liver and takes away the body's desire to increase the supply of the chemicals (porphyrins and precursors) needed for its production. If it cannot be obtained soon enough, large quantities of glucose, which have a similar but lesser effect, may be given in the meantime.

Conclusion

It is important to remember that most people with acute intermittent porphyria lead a normal healthy life. A few simple measures can decrease the risk of illness described here. Even the few who do become ill usually make a complete recovery and have no more than one or two acute attacks in early adult life. As one grows older, the risk of an acute attack decreases, particularly after the age of forty, but it never disappears completely.

Important Contact Information

Patient Support Group

The British Porphyria Association (BPA)

☎ **Help-line: 01474 369231**

Email: helpline@porphyria.org.uk

Website: www.porphyria.org.uk

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Website: www.cardiff-porphyria.org

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Useful Website Addresses

www.porphyria.org.uk

www.cardiff-porphyria.org

www.porphyria-europe.com

www.porphyriafoundation.com (USA)

www.cpf-inc.ca (Canada)

Acute Intermittent Porphyria

Information for Patients



British Porphyria Association

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