

In special circumstances, where risk is low and the benefits high, your doctor may consider (after discussion with your porphyria specialist) the use of hormone-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 occur just before periods. One possible treatment is to suppress 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. It is therefore very important for the doctors providing care during pregnancy to know that you have AIP.

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. An immediate increase in carbohydrate intake (sweet or starchy food and drinks) may help to reduce the severity of the symptoms.

If symptoms are severe, or don't settle within 24 hours, you should contact your doctor, as you may need to be admitted to hospital. This will allow for:

- Confirmation of the acute attack by measuring PBG in urine.
- Early start of specific treatment: for example, with haem arginate (Normosang).
- Treatment for the various symptoms accompanying the attack, such as drugs to relieve pain and nausea, or actions to maintain an adequate intake of calories.

Your doctor should contact the National Acute Porphyria Service (NAPS: *see back page*) who will give advice about treatment and provide haem arginate if needed.

Conclusion

Most people with AIP lead a normal healthy life. A few simple measures can decrease the risk of illness. 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. With age, the risk of an acute attack decreases, particularly after forty, but it never disappears completely.

Useful contact details

BPA telephone helpline: 0300 30 200 30

BPA email helpline: helpline@porphyria.org.uk

European Porphyria Network: www.porphyria-europe.com

Rare Connect: www.rareconnect.org

Medic Alert: www.medicalert.org.uk

Welsh Medicines Information Centre (WMIC)

WMIC provides advice and a list of SAFE drugs which can be downloaded at www.wmic.wales.nhs.uk/porphyria_info.php. WMIC can also be contacted by telephone on 029 2074 3877 or 029 2074 2251.

National Acute Porphyria Service (NAPS)

NAPS provides clinical advice and haem arginate where appropriate for patients having either one-off acute attacks or recurrent attacks of porphyria. There are two full NAPS centres and two associate centres:

- King's College Hospital, London
- University Hospital of Wales, Cardiff
 - * Salford Royal Hospital, Salford
 - * St James University Hospital, Leeds

Initially, your doctor would need to contact the emergency number at the University Hospital of Wales: 029 2074 7747. This 24/7 number should be used at all times for new patients, and out of working hours for existing NAPS patients.

Specialist porphyria laboratories

- For an up-to-date list see the British and Irish Porphyria Network (BIPNET) website: www.bipnet.org.uk.
- Also see www.kingspath.co.uk for details of tests available and sample requirements.

British Porphyria Association



Charity No: 1089609

Acute Intermittent Porphyria (AIP)

Information for patients



Introduction: what is porphyria?

The 'porphyrias' are a group of eight rare disorders. Most are inherited and result from a faulty gene leading to difficulty making a chemical called haem, which is a constituent of many important proteins in the body. Haem precursor chemicals accumulate which can cause severe medical problems.

Acute intermittent porphyria (AIP) is one of four porphyrias that may present with 'acute attacks'. The others are variegate porphyria (VP), hereditary coproporphyria (HCP) and ALA-dehydratase deficiency porphyria (ADP). These attacks are uncommon and are often difficult to diagnose.

In AIP, only acute attacks occur - the skin is never affected - whereas in VP and HCP the skin may be affected.

Acute attacks:

- **Severe pain** is almost always present in an acute attack - usually in the abdomen, but it may be felt in the back, arms or legs.
- **Nausea, vomiting** and **constipation** are common.
- **Low sodium (salt) levels** in the blood are often found.
- **Pulse rate** and **blood pressure may increase**, but rarely to dangerous levels.
- **Confusion** may occur during an acute attack and rarely severe mental disturbance.
- **Convulsions** and **muscular weakness**, which may lead to paralysis, can arise, often several weeks later.
- An acute attack usually lasts for no longer than one or two weeks. However, if severe neurological complications like motor paralysis occur, the recovery will be gradual but slow.

Occurrence

Most people with AIP will never have any problems. Attacks occur in about 1 in 5 patients, more often in

females than males. The most common age for an attack is from the late teens to the early forties and 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, nowadays they are rarely fatal and most people make a full recovery. However, everyone with AIP needs to take a few simple precautions to reduce the risk of attacks.

How is acute porphyria diagnosed?

To identify an **acute attack** of porphyria, the first urgent test, urine porphobilinogen (PBG), may be done in your local hospital. To identify the **type of porphyria**, samples of urine, blood and stools (faeces) need to be tested in a laboratory which specialises in 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 AIP, it may be difficult to decide whether an illness is an attack of acute porphyria or something else. In this situation, urinary PBG can sometimes help your doctor to decide whether you are suffering from your porphyria or some other illness. It is important that people with porphyria do not assume that all of their illnesses are porphyria related, as common but potentially serious conditions, such as appendicitis, may be overlooked.

How can I reduce the risk of attacks?

Acute attacks can usually be reduced by avoiding any triggers.

Triggers include:

- Numerous drugs, including prescribed, over-the-counter or herbal remedies
- Alcohol
- Fasting - including dieting and gastric infections
- Hormones
- Infections and stressful situations

Drugs: Many drugs can trigger attacks. The Welsh Medicines Information Centre (WMIC) has compiled a list of SAFE drugs for easy reference. The SAFE list is updated yearly and is available to view online. Alternatively, patients or their doctors can call **029 2074 3877** (*more details overleaf*). Drugs not on the safe list should only be taken after obtaining expert advice from WMIC or a porphyria specialist.

The European Porphyria Initiative (EPI) website www.porphyrria-europe.com contains further detailed information on prescribing in acute porphyria, including common prescribing problems (pain relief, anaesthesia, hormonal contraception). *Please also see our 'Drugs and Porphyria' leaflet for further information.*

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

It is important to ALWAYS check the safety of any medicine with your doctor or pharmacist using the SAFE list

Alcohol: This is a common trigger for attacks, especially binge drinking, so the best advice is don't drink. If you do drink, keep intake low.

Diet: Low calorie diets, 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. Some people 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. Women with an acute porphyria should avoid oral contraceptives 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.