There is no easy treatment for EPP. *(Please see our EPP leaflet for further details on practical tips for living with a light intolerance condition.)*

CEP is a very rare disorder. It is a multi-organ disease predominantly affecting skin, eyes and bone marrow and can cause severe scarring due to sensitivity to visible light. Severe cases may need bone marrow transplants.

Introducing the BPA

The BPA was established in 1999 by a group of patients and relatives who had experienced isolation and difficulties due to their diagnosis. They had found there to be a general lack of understanding and assistance available. The BPA became a registered charity in 2001 and is run by a committee of volunteers.

The BPA aims to:

- Educate via newsletters, leaflets, open days and web information.
- Support those who feel isolated via helplines.
- Provide helpful contacts with doctors/specialists.
- Provide a grant/aid fund through which members can apply for help with certain costs incurred because of the condition.
- Keep patients up to date with new research.
- Educate doctors and medical staff in order to improve their understanding of the condition.
- Provide doctors with links to other medical professionals.
- Provide funding for new research.
- Encourage research establishments to consider porphyria for research.

Join us in raising awareness during **International Porphyria Awareness Week (IPAW).** IPAW starts on the 3rd Saturday in April each year.

Useful contact details

BPA telephone helpline: 0300 30 200 30

BPA email helpline: helpline@porphyria.org.uk

European Porphyria Network: https://porphyria.eu

British Association of Dermatologists: www.bad.org.uk

Medic Alert: www.medicalert.org.uk

Rare Connect: www.rareconnect.org

Acute porphyria patient experience videos: www.youtube.com/user/acuteporphyrias/videos

UK Porphyria Medicines Information Service (UKPMIS)

UKPMIS based with the Welsh Medicines Information Centre (WMIC) provides advice and a list of SAFE drugs for those with acute porphyrias: www.wmic.wales.nhs.uk/porphyria_info.php. UKPMIS can be contacted by telephone on 029 2074 3877 or 029 2074 2251.

Dundee Cream

Dundee Pharmaceuticals, Ninewells Hospital, Dundee. DD1 9SY. Tel: 01382 632052.

National Acute Porphyria Service (NAPS)

NAPS provides clinical advice and haem arginate where appropriate for patients having one-off acute attacks or recurrent attacks of porphyria. There are currently 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.

British Porphyria Association

Charity No: 1089609

Introducing porphyria and the BPA

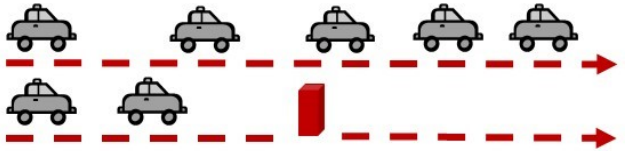
Information leaflet

Introduction: what is porphyria?

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

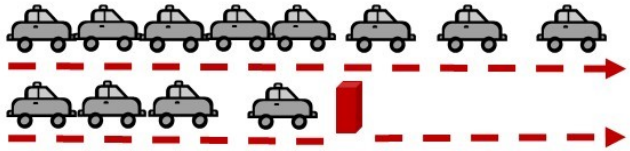
The porphyrias are divided into **acute**, **cutaneous** (skin) or **mixed**, depending on the types of symptoms suffered (*see table*).

Simplified representation of the process



The blocked lane represents an enzyme deficiency caused by a faulty gene. The type of porphyria depends on the enzyme affected.

Normally (i.e. light traffic conditions) haem is still produced fast enough and symptoms do not occur.



However, add the rush hour (e.g. an infection) and porphyrin precursors (ALA, PBG) build up - leading to an acute attack.

For the skin porphyrias, light-sensitive porphyrins build up in the blood and cause sensitivity to sunlight.

Acute attacks are caused by a build-up of ALA and PBG (aminolevulinic acid and porphobilinogen) which result in damage to nerves. ALA and PBG are the raw materials for making haem.

In the **skin porphyrias**, problems are caused by a build-up of light-sensitive porphyrins in the blood and skin. In sunlight, they start to damage the skin.

Acute porphyrias

AIP, **ADP**, **VP** and **HCP** may cause acute attacks. Attacks are uncommon and are often difficult to diagnose. In **AIP** and **ADP** only acute attacks occur and the skin is never affected, whereas in **VP** and **HCP** the skin may also be affected.

Typical features of acute attacks:

- Severe pain in the abdomen, back, arms or legs
- Nausea, vomiting and constipation
- Low sodium (salt) levels in the blood
- Pulse rate and blood pressure may increase
- Confusion
- Convulsions and muscular weakness

Attacks occur in about one in five people with the faulty gene and are more common in women than men. Attacks are extremely rare before puberty and most people have only one or a few acute attacks.

Acute porphyria attacks are often triggered by exposure to commonly prescribed drugs, illegal drugs, alcohol, dieting, stress, infections, viruses and hormonal fluctuations. (*Please see our AIP and Drugs in Porphyria leaflets for more on acute porphyria triggers and treatments*.)

Acute porphyrias with skin symptoms

In addition to acute symptoms, **VP** and **HCP** can cause blistering (skin) in sunlight - the skin can become fragile and should be protected. Skin problems can occur during acute attacks or at different times.

Cutaneous (skin) porphyrias

The skin porphyrias (**PCT**, **EPP**, **XLEPP** and **CEP**) cause sensitivity to sunlight on exposed areas of skin. As with all porphyrias, the severity of the problem varies. People with skin porphyrias do NOT need to stick to the SAFE drugs list used by those with acute porphyrias.

PCT is the most common of the skin porphyrias, and is the only porphyria which is not always inherited. It can be triggered later in life by heavy drinking, iron tablets, oestrogens, certain drugs or by liver infections. In 20% of UK patients, it is caused by hereditary haemochromatosis. **PCT** is treated by phlebotomy (removing blood) or low-dose oral chloroquine, and sufferers need to avoid whatever triggered the problem.

EPP is a less common form of porphyria. Like the acute porphyrias, it is an inherited disorder. There are a variety of genetically different forms of EPP - autosomal recessive, which makes up 97% of cases, and the X-linked dominant form - **XLEPP**.

Sunlight does not always need to be direct – light reflected off water, sand or snow, or passing through window glass may also cause symptoms.

EPP is different from the other cutaneous porphyrias as it doesn't usually cause blistering. In bright light, the skin becomes excruciatingly painful, but there is sometimes little to see.

Acute porphyrias	Mixed: acute porphyrias with skin symptoms	Cutaneous (skin) porphyrias
Acute intermittent porphyria (**AIP**)	Variegate porphyria (**VP**)	Porphyria cutanea tarda (**PCT**)
ALA-dehydratase deficiency porphyria (**ADP** - *also called plumboporphyria*)	Hereditary coproporphyria (**HCP**)	Erythropoietic protoporphyria (**EPP**)
		X-linked erythropoietic protoporphyria (**XLEPP**)
		Congenital erythropoietic porphyria (**CEP**)