

If you have been handed this leaflet, it is possible that this is the first time you have ever come across the term **porphyria**. So what is it, and what does the British Porphyria Association do?

What is Porphyria?

The **porphyrias** are a group of seven, relatively rare, genetic disorders. In each porphyria a specific enzyme, which is needed to complete each step on the pathway to produce **haem**, is faulty. (**Haem** is used in **haemoglobin**, the red blood pigment which carries oxygen.) As a result, porphyrins accumulate causing severe medical problems but haem levels are maintained so porphyrias rarely cause anaemia. The type of porphyria varies according to the enzyme/step which is affected. The porphyrias are broadly categorised into acute and cutaneous.

What are the main symptoms?

Acute Porphyrias: (*Acute Intermittent Porphyria, Variegate Porphyria, Hereditary Coproporphyrinuria and ALA-dehydratase Deficiency Porphyria*)

Commonly suffer from attacks of –

- severe stomach pain, or pain in back, legs or arms,
- constipation, nausea, vomiting,
- dark, purple-red or brown coloured urine, and sometimes -
- muscular weakness (from nerve damage) particularly in arms is common – but this can progress to complete paralysis,
- psychiatric symptoms, convulsions.

Cutaneous (Skin) Porphyrias: (*Porphyria Cutanea Tarda, Erythropoietic Protoporphyrinuria and Congenital Erythropoietic Porphyria*)

Suffer from -

- photosensitivity leading to burning and itching or skin lesions and fragile skin after exposure to the sun,

- many people with PCT suffer from liver cell damage.

Two of the acute porphyrias, Variegate Porphyria and Hereditary Coproporphyrinuria, also suffer from skin problems when exposed to sunlight.

What are attacks triggered by?

Acute Porphyria attacks are often triggered by exposure to commonly prescribed drugs, illegal drugs, alcohol, dieting, stress, infections and hormonal fluctuations.

Cutaneous Porphyria attacks are triggered by exposure to the sun and in PCT also from alcohol, oestrogens, iron and chemicals.

How is it treated?

Acute attacks are treated by –

- withdrawing precipitating factors,
- haem arginate (via a drip),
- maintaining nutrition.

Cutaneous symptoms are treated by –

- avoiding the sun and attention to skin care,
- PCT is treated by phlebotomy or low-dose oral chloroquine. PCT sufferers also need to avoid alcohol and oestrogens.

It is IMPORTANT to remember that the severity of symptoms varies dramatically in all types of porphyria but the more knowledgeable a sufferer is about the condition, the more likely they are to stay well. This is where the BPA comes in.

What is the BPA?

The BPA is the British Porphyria Association.

It was established in 1999 by group of patients and relatives who had experienced isolation and difficulties due to their diagnosis, and 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.

What does the BPA aim to do?

The Association's aim is to reach out to as many people as possible;

- patients and relatives,
- doctors and medical staff,
- hospitals and research establishments,

- in order to improve the understanding of this condition. Early diagnosis is vital if we are to improve the quality of life for those affected by it.

Porphyria is a rare condition and as the complaint thought by some to have been suffered by 'mad' King George III, it is often portrayed in a dismal light. Although the disorder can in some cases be very severe, there are many reasons to be optimistic and with the right information, many sufferers can live an absolutely normal life. With this in mind the BPA aims not only to raise awareness of the condition, but also to ensure that the portrayal of the disorder is always accurate.

The BPA also provide an emotional/social support system for sufferers and their families; simply knowing that they are not alone is, in many cases, a great support and comfort to sufferers and their families.

What happens to the funds raised?

Funds help the BPA to do the following:

for patients:

- ? Educate patients and their families about their condition – this enables them to deal with it from a more informed perspective.
- ? Support those who feel isolated and find it helpful to speak to others in similar circumstances.

- Provide helpful contacts with doctors and 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.

for medical staff

- Educate doctors and medical staff in order to improve their understanding of the condition – early diagnosis is vital if we are to improve the quality of life of those affected by it.
- Provide doctors with links to other medical professionals.

for research:

- Provide funding for new research.
- Encourage research establishments/ organisations to consider porphyria for research.

To do this, some of the funds are currently being used to:

- Produce and distribute handouts for patients, their families and for doctors.
- Produce and distribute the twice yearly newsletter.
- Cover costs incurred through holding meetings in various places over the country in order to make it easier for sufferers to attend.
- Cover costs involved with a BPA representative attending other relevant porphyria research meetings.
- Cover costs incurred providing the helpline, the website, and other running costs of the charity including stationery and postage.

National Porphyria Awareness Day

In 2005, the BPA joined the Canadian Porphyria Foundation in celebrating 1st June as National Porphyria Awareness Day. This will continue each year and provides an opportunity to try and raise awareness of the condition through the media and education days.

If you have come across this handout while searching for more information about porphyria, then please do not hesitate to contact us.

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

Specialist Porphyria Laboratories

Department of Medical Biochemistry and Immunology
University Hospital of Wales
Heath Park
Cardiff CF14 4XW
Tel: 02920 743565

Department of Clinical Biochemistry
King's College Hospital
Denmark Hill
London SE5 9RS
Tel: 020 7346 3856

Useful Website Addresses

www.porphyria-europe.com
www.porphyriafoundation.com
www.cpf-inc.ca
www.porphyria.org.uk

What is Porphyria?

and

What is the British Porphyria Association (BPA)?



British Porphyria Association

Charity No: 1089609