What Is Porphyria?

Porphyria is a group of 7 *inherited metabolic disorders* (diseases), caused by 7 different faulty genes. They cannot be “caught” like a cold.

Metabolic means they affect the chemistry of the body, not its structure. The 7 genes all slow the production of *haem* (used in haemoglobin and other body chemicals which transfer oxygen).

Because the cause is in a gene, there may be several people in a family who have inherited it. *Many of these will have no problems.*

All seven are rare. Two are so rare that even some experts have not seen them. This can make diagnosis difficult.

What effects do the porphyrias have?

They have two possible effects: *acute attacks* (causing very nasty illness) or *skin sensitivity* in sunlight (far worse than normal sunburn).

Two porphyrias* cause *both*.

Acute porphyrias are:

ADP (ALA-dehydratase Deficiency Porphyria), very rare.

AIP (Acute Intermittent Porphyria), the commonest acute porphyria.

***HCP** (Hereditary Coproporphyria)

***VP** (Variegate Porphyria) 2nd commonest.

Acute attacks can cause all or some of these:

- Severe pain (in stomach, back or thighs)
- Nausea; Vomiting; Constipation
- Red or brown urine (can look normal, but change colour when stored before tests)
- Low salt /sodium concentrations in the blood
- Rapid pulse and high blood pressure
- *Loss of movement in the arms or legs*

(The last is unusual and can occur several weeks after an attack.)

Acute porphyrias only cause attacks in about one in five of those with the gene. Often there are only one or two attacks. Many people never have attacks at all, even if a close relative does. Women are more likely to have attacks than men, due to fluctuations in female hormones. Attacks are very rare before puberty.

Skin (cutaneous) porphyrias are:

CEP (Congenital Erythropoietic Porphyria),
PCT (Porphyria Cutanea Tarda),
EPP (Erythropoietic Protoporphyria)

CEP, PCT, *VP and ***HCP** all cause blistering (skin lesions) in sunlight and the skin can become fragile.

CEP is very rare. It can cause severe scarring.
PCT is the most common, as it is the only porphyria which can be caused by something other than a “porphyria” gene**.** It can be triggered later in life, by heavy drinking, iron supplements or certain drugs, and by liver infections.

EPP causes pain in the skin on sun exposure, and there is no blistering and often no redness with it, so it can be puzzling for doctors.

Why does this happen?

Acute attacks are caused by the build-up of ALA and PBG (*aminolevulinic acid* and *porpho-bilinogen*), the raw materials for making haem. ALA and PBG damage the nerves.

Skin problems are caused by the build-up of light sensitive *porphyrins* (purplish chemicals which are on the way to being haem). When they get in the skin, light hits them and they start damaging the skin.

To understand this better, look at the cartoon showing how haem production works.
Haem is made from the common body chemical ALA, represented by the first box on the bench at the left-hand end of the production line.

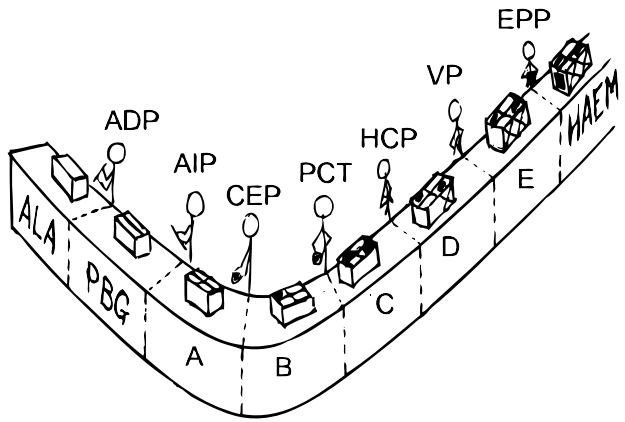
Small changes are made to the ALA “box” as it goes along, changing it into haem (on the right). The stick people in the cartoon represent *enzymes* (special proteins). One *enzyme* makes one change possible, and seven different ones are needed to change ALA to haem.

When someone has a porphyria, the gene producing *one* of these *enzymes* is faulty and the enzyme doesn’t work too well (imagine the stick man has his arm in a plaster!).

For instance, **AIP** is caused by a problem producing the second enzyme (stick man AIP). This means that the raw material PBG (and the ALA from which it is made) isn’t used up as fast as it should be. As long as nothing else slows things down further, haem is still produced fast enough and the person has no attack.

When something slows the enzyme further (the stick man breaks the other arm!) ALA and PBG build up more. This can cause an acute attack. The same happens with **ADP** (though only ALA will build up).

CEP, PCT and **EPP** cause light-sensitive porphyrins to build up (A, B and E respectively), which cause the skin problems, but **they do not cause acute attacks.** ⇨



Unfortunately, in **HCP** and **VP** there is a build-up of light-sensitive porphyrins (C & D) **plus** a feedback system which raises ALA and PBG. So people with HCP or VP can get both skin problems **and** acute attacks.

Triggers for acute attacks

In the **acute porphyrias**, alcohol and some drugs can cause attacks. "Acute" porphyrics should *use only safe drugs.* They should *carry a list* and get a *MedicAlert pendant/bracelet*. They should also eat regular meals, since starvation can bring on attacks. Often stress is a factor and infections (particularly gastric flu) can cause real problems.

What treatments are there?

ACUTE porphyrias: *Mild* attacks can sometimes be stopped by taking high glucose foods/drinks. *Severe* attacks can be treated with *haem arginate* given into a vein (in hospital). A few people need haem arginate regularly to prevent attacks.

SKIN Porphyrias:

PCT is the most easily treated by removing blood (like blood donation). This uses up iron stored in the liver and the skin problems subside. Oral low-dose chloroquine is also effective.

CEP is the opposite – some need blood transfusions for anaemia. Severe cases may need bone marrow transplants.

EPP can sometimes be helped by taking large quantities of beta-carotene. Recently, hospital UV therapy has been surprisingly effective. Mostly, it is a matter of avoiding going out in the sun as much as possible, wearing protective clothing and using thick sun-block creams which block violet light.

Other SKIN porphyrias can also be helped by using sun-blocks and covering up.

Please remember:

If you have a rare disorder, NHS rules allow you to ask to see a medical specialist.
The BPA can advise you about the nearest person to you, specialising in the porphyrias.

Further information:

BPA leaflets (requests to Helpline, below, *or* e-mail secretary@porphyria.org.uk)

www.porphyria.org.uk BPA (info + link to a printable *drugs* list for those with an *acute* porphyria)

www.porphyria-europe.com European Porphyria Initiative **(**info, plus *drug* safety database - useful for doctors)

MedicAlert - ☎ 020 7833 3034 *or* **www.medicalert.org.uk**

What does the British Porphyria Association (BPA) do?

Our aims are to

- Support members, particularly with information (newsletters, leaflets, etc.)

- Increase awareness among the public and medical staff (leaflets, talks by members, etc).

British Porphyria Association,
Helpline: ☎ 01474 369 231
(not manned continuously – answering machine at other times.)

An Introduction to the Porphyrias

British Porphyria Association

Charity No: 1089609