The cutaneous (skin) porphyrias

can all give *sensitivity to sunlight*, on exposed areas of skin. As with all porphyrias, *the severity* of the problem varies enormously.

Sunlight should be avoided as much as possible. When avoiding sunlight is not possible, special sun-blocks which block violet light can help. (In mild cases, high-factor sun cream with high-UVA protection may be enough.) Hats and clothing which cover up the skin are also recommended. The BPA keeps a list of manufacturers of light-dense clothing.

Symptoms

CEP, PCT and EPP never cause acute attacks, so the "safe" drugs list is not needed.

In **EPP** (Erythropoietic Protoporphyria), the symptoms are rather different from the rest, so turn to *section 4* for a description.

The other cutaneous porphyrias (1.**CEP** and 2.**PCT**) and the two acute porphyrias with skin problems (3.**HCP** and **VP**) can all cause *blistering* (leading to *scarring*). *White spots* (milia) can form. There can also be *abnormal hair growth* (usually on the face, mostly the cheeks or temples).

There is likely to be *fragility* of the skin. For example, the skin on the knuckles gets damaged when knocked fairly lightly, and can take longer than normal to heal. So care is needed doing anything which might knock or rub the skin, and knocks need to be kept clean. Gardening or DIY is best done in gloves, if possible.

1. Cutaneous Erythropoietic Porphyria (CEP)

This is very rare, as the gene which causes it is a *recessive* (you have to have 2 faulty copies, one from each parent). It usually causes sun-sensitivity *from birth*, or soon after. Severely affected people can get pain immediately on sun exposure, with crops of blisters later which easily get infected. Severe scarring can affect the cartilage in ears and nose, and damage the bones of the fingers. So sunlight must be avoided.

It can also cause *anaemia*. With regular blood transfusions, however, life expectancy is normal.

Since the faulty part of the haem-building sequence happens in the bone marrow, *a bone marrow transplant* is a possible cure.

2. Porphyria Cutanea Tarda (PCT)

This is the most common of the cutaneous porphyrias. It is the only one which normally develops in *middle age* (hence "Tarda"). It is also the only one which can be *acquired.*

A few people (about 1 in 5) have a faulty gene causing PCT. They usually develop symptoms earlier in life. The gene is *dominant* – of the two copies, only one needs to be faulty to cause PCT. The parent who passed it on may have problems.

Most people with PCT acquire it (develop it) because iron in the liver, or the systems using iron, slow down the relevant enzyme. This is often triggered by alcohol (moderately heavy drinking), though some people are sensitive to iron supplements, artificial oestrogens (found in the pill, HRT and prostate cancer treatment) or other drugs. Hepatitis C and HIV can also trigger it and *haemochromatosis*, another genetic condition, contributes in many cases.

PCT causes *blistering & fragility* of sun exposed skin, and the skin can *thicken* and *darken.* (It sometimes turn lighter where blisters have been).

After diagnosis it is important to find out if there is a trigger for the sun-sensitivity.

Treatment is the same, whatever the origin, acquired or genetic - *removal of any identified cause (trigger) where possible*, and treatment either with a *drug* (a low dose of chloroquine) or by *venesection* (removing blood, as for a blood donation), depending on the exact details of the person's condition.

The body then uses iron to make new red blood cells, reducing the stores in the liver, and the skin problems subside. The PCT may recur a few years after treatment has finished.

3. Hereditary Coproporphyria (HCP) and Variegate Porphyria (VP)

These usually develop in the *teens or twenties*, and both are caused by a *dominant* gene. So one parent carries the gene and can also be affected. They are classed as *acute porphyrias*, as they can cause acute attacks. However, they can also *cause sun-sensitivity* at times.

Skin fragility is common. Blistering may occur in the sun, coinciding with attacks in HCP, but often happening at other times in VP.

Those who get blistering should *keep covered up* as much as possible, and use a *sun-cream* when outside, especially in the summer. Usually a cream with high UVA protection is enough in Britain, but a sun block may be needed in direct sunlight or in hotter countries.

Sometimes skin pigment can *increase (darken)* or *decrease (turn lighter*) in sun-exposed skin, most often hands and face. Whiter areas need extra protection from the sun, or they will burn.

Those with HCP and VP should stick to "safe" drugs, if at all possible (see websites listed overleaf). "Unsafe" drugs can trigger attacks, & make skin problems worse.

4. Erythropoietic Protoporphyria (EPP)

The inheritance of EPP is rather complex. It is now believed that the faulty gene, paired with one particular "normal" variant of the gene, can cause EPP. In combination with other variants, the EPP gene gives no problems.

EPP is rather different from the other cutaneous porphyrias, as it *doesn't* cause blistering. In sunlight, the skin becomes very *painful (itching & burning)* and sometimes *swells up slightly*. Attacks of pain often last for about 2 days. The time of exposure to sunlight, before the pain starts, varies from one person to another, as does the time taken to recover once out of the light. Fanned air and cold water can help relieve the pain to some extent.

Some people get *reddening* of the skin, but many have nothing visibly wrong. Over time, the skin can thicken on the knuckles and there can be scarring on the face.

EPP normally develops in early childhood. An affected baby will often get fractious if taken outside, or put near bright lights, or by a window during daylight. The pain makes some scream. Even when brought indoors, the baby can still be fractious while the skin recovers.

EPP does **not** cause acute attacks, so there is no need to stick to the "safe" drugs list. However, it is not advisable to drink excessively, as the combination of alcohol with EPP can affect the liver. Routine blood tests are needed, as a few people develop gall-stones or liver problems.

Unfortunately, there is no easy treatment. Mostly it is a matter of:
avoiding being out in the sun, under bright artificial light or close to a window (tinted film can be attached to windows, very useful in cars);
covering up with a hat and light-dense clothing when outside; and
using a special sun-block cream (e.g. Dundee cream). Sun block can be mixed with normal cosmetics to get a more realistic colour.

Some people respond to high doses of *pure beta-carotene*, which turns the skin slightly orange. (Self treatment is not recommended. Foods and some supplements may contain chemicals dangerous in the high doses needed.)

Recently, there have been hospital trials of UV light treatment. It may harden the skin & reduce sensitivity. First results are encouraging.

Further information:

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

www.porphyria.org.uk BPA has patient info on porphyrias

www.cardiff-porphyria.org Cardiff University Hospital's web site, (which has a printable *drugs* list for "acutes")

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

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: ☎ 0147 4 36 9231

The Cutaneous (Skin) Porphyrias

British Porphyria Association

Charity no.1089609
www.porphyria.org.uk