PORPHYRIA CUTANEA TARDA (PCT)

What are the aims of this leaflet?

This leaflet has been written to help you understand more about porphyria cutanea tarda (PCT). It tells you what it is, what causes it, what can be done about it and where you can find out more about it.

What is porphyria cutanea tarda?

PCT is one of a group of disorders (known as the porphyrias) caused by a range of enzyme defects in one of the biochemical pathways in the body. Any one of these can result in a build-up of chemicals called porphyrins. In PCT the porphyrins are produced from the liver and cause the skin to become very sensitive to light (photosensitive). Some of the other types of porphyria affect internal organs and can cause symptoms unrelated to the skin but this does **not** happen in PCT.

PCT is an uncommon condition affecting about 1 in 25,000 people.

What causes PCT?

There are two types of PCT:

- Type 1 (sporadic PCT) is the commonest (80% of PCT patients) and is **not** inherited.
- Type 2 (familial PCT) (20% of PCT patients) is due to an inherited gene defect or mutation. Not all family members who inherit the gene mutation will develop the condition, as other factors need to be present in those who have the abnormal gene (described below).

The following are the most common factors associated with exacerbating PCT:

- Excessive alcohol consumption.

- Oestrogen therapy, e.g. oral contraception or hormone replacement therapy (HRT).
- Viral infections of the liver, e.g. hepatitis C.
- Iron accumulation in the liver. Nearly all patients have an increase in iron in the liver, which is believed to be partly responsible for the enzyme blockage. Some patients also have an additional inherited condition called haemochromatosis that is responsible for the iron accumulation.

What are the symptoms of PCT?

Sun-exposed areas of skin are affected, particularly on the very thin skin on the backs of the hands and sometimes the face and scalp. Blisters may occur that burst and heal slowly leaving scars and tiny white raised spots called 'milia'. The skin is fragile, and even mild injury may cause grazes and scars. Darkening of the usual skin colour and an increased growth of hair on the hands and face may occur. In addition to the skin problems, the urine may become a dark colour.

How is PCT diagnosed?

PCT is diagnosed by measuring porphyrins in samples of blood, urine and faeces. Each of these samples must be collected into a special darkened container which excludes light, e.g. wrapping the specimen container in tinfoil before collecting the sample. Other tests, such as blood tests for liver function, glucose and iron levels may be undertaken to investigate for the conditions described above which are associated with PCT. Depending on the results of these tests, you may also be referred to see another doctor, for example a liver specialist (hepatologist).

It is rare for more than one member of a family to have PCT, but if other family members are affected with similar signs and symptoms they can be tested. Screening of family members who do not have skin problems is not generally required.

Can PCT be cured?

Although the underlying cause of PCT cannot be cured, the symptoms can be controlled. Attention to known exacerbating factors, such as sunlight exposure, stopping the combined contraceptive pill and avoiding excess alcohol, is important.

How can PCT be treated?

Specific treatments aimed at reducing porphyrin levels are as follows:

- If raised iron levels are found then these can be reduced by regularly removing a unit of blood (the same amount as given by blood donors). The body uses iron to make more blood and the process is repeated every few weeks or months until the excess iron has been removed. In the genetic disorder haemochromatosis, which may occur in association with PCT, removal of excess iron may need to be on a long-term basis.
- Low-dose hydroxychloroquine may be prescribed (usually one tablet twice weekly for several months or years). This drug helps to mobilise the excess porphyrin from the liver so that it can pass out of the body in the urine. It is important that only small doses are used, as larger doses can cause acute inflammation of the liver in people with PCT.
- In patients who cannot tolerate either of these two treatments, other options are available, although these are more complicated to administer and would be discussed with you individually.
- Women on oestrogen treatments including the combined contraceptive pill will be asked to stop taking them while the PCT is being treated. However it may be possible to restart hormone treatment once the PCT has been treated.

Self care (What can I do?)

- Treatment can take several months to become effective and symptoms may initially get worse. While the treatments take effect the skin will remain fragile and sensitive to bright sunlight. Conventional sunscreens are formulated to protect against ultraviolet A and B light (particularly UVB) and may therefore **not** be effective in those with PCT, as the symptoms are triggered by UVA/visible light. Reflectant sunscreens that are based on titanium dioxide or zinc oxide will be more effective as they cover UVA, UVB, and visible light. A tinted reflectant sunscreen 'Dundee Reflectant Sunscreen', which is available in 3 colours, is available on prescription from Tayside Pharmaceuticals (see below for details) and is effective at blocking visible light. These can be mixed to obtain a good colour match with your skin.
- Avoiding excess alcohol is advised. Some patients may have to abstain from alcohol altogether.
- Some types of porphyria cause acute attacks, or 'porphyria crises', when a sufferer is given certain medications and anaesthetics; this is **not** the case with PCT.

Vitamin D advice

The evidence relating to the health effects of serum Vitamin D levels, sunlight exposure and Vitamin D intake remains inconclusive. Avoiding all sunlight exposure if you suffer from light sensitivity, or to reduce the risk of melanoma and other skin cancers, may be associated with Vitamin D deficiency.

Individuals avoiding all sun exposure should consider having their serum Vitamin D measured. If levels are reduced or deficient they may wish to consider taking supplementary vitamin D3, 10-25 micrograms per day, and increasing their intake of foods high in Vitamin D such as oily fish, eggs, meat, fortified margarines and cereals. Vitamin D3 supplements are widely available from health food shops.

Where can I get more information about PCT?

Links to patient support groups:

British Porphyria Association
Tel: 0300 30 200 30
Email: helpline@porphyria.org.uk
Web: www.porphyria.org.uk

Web links to detailed information:

American Porphyria Foundation
Web: www.porphyriafoundation.com

European Porphyria Initiative
Web: www.porphyria-europe.org (Includes a list of centres specialising in this group of disorders)

UK Supra Regional Porphyria Services
Web: www.sas-centre.org

University of Cape Town Porphyria Service
Web: www.porphyria.uct.ac.za

Other useful information:

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

For details of source materials use please contact the Clinical Standards Unit (clinicalstandards@bad.org.uk).

This leaflet aims to provide accurate information about the subject and is a consensus of the views held by representatives of the British Association of Dermatologists: individual patient circumstances may differ, which might alter both the advice and course of therapy given to you by your doctor.

This leaflet has been assessed for readability by the British Association of Dermatologists' Patient Information Lay Review Panel

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