PORPHYRIA CUTANEA TARDA (PCT)

What are the aims of this leaflet?

This leaflet has been written to help you understand more about porhyria 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 seven individual "porphyrias" each of which is caused by a different blockage in a biochemical pathway that make a substance called "haem". Haem is made in all cells of the body, although the majority is used to make haemoglobin, the oxygen carrying pigment in the red blood cells.

In PCT there is a blockage in the liver of the fifth step in the pathway, an enzyme called uroporphyrinogen decarboxylase (abbreviated to UROD). This causes a build up of chemicals, termed "porphyrins", which fail to be converted into haem. These porphyrins leak out of the liver into the bloodstream and are carried to the skin where they cause the characteristic skin problems associated with this type of porphyria. PCT DOES NOT CAUSE ACUTE ATTACKS.

Although it is the commonest of the porphyrias, it is still a rare condition which affects about one in 25,000 of the population.

What causes PCT?

Unlike most porphyrias, the majority of patients with PCT do not inherit the condition, and this form of PCT is known as sporadic (or Type I) PCT.

In a small percentage of patients, approximately 20%, the condition may be in part due to an inherited mutation in the UROD gene. This form of PCT is known as familial (or type II) PCT. However not all family members who inherit a UROD mutation will develop the condition, as PCT requires other factors described below to be present as well.

The following are the most common factors associated with PCT:
Increase in liver iron accumulation. Nearly all patients have an increase in iron, which is believed to partly responsible for the enzyme blockage. However some patients also have an inherited condition called haemochromatosis that is responsible for the iron accumulation.

Regular alcohol consumption.

Viral infections of the liver; e.g. hepatitis C.

Oestrogen therapy; e.g. oral contraception or hormone replacement therapy (HRT).

What are the symptoms of PCT?

Sores, blisters and little white spots called milia occur on any area of sun exposed sites, particularly backs of the hands and sometimes on the face. The blisters rupture forming sores which heal slowly and can cause scarring.
The skin in these areas is also very fragile, breaks down easily and heals slowly.
Excessive fine hair growth on cheeks and forehead is sometimes seen, and brown discolouration of skin on the face can happen.
Changes in skin pigmentation, which may be increased or decreased, can occur.
In addition to the skin problems, persistently dark urine may also be noticed

How is PCT diagnosed?
PCT is diagnosed by measuring porphyrins in samples of blood, urine and faeces. Other tests are done to investigate for the conditions described above which are associated with PCT. These will include blood tests for liver function, viral hepatitis, haemochromatosis and total iron levels, and in some patients more detailed liver tests may be advised. Depending on the results of these tests, you may also be referred to see another doctor, for example a liver specialist.

If other family members are affected with similar signs and symptoms they can easily be tested for PCT. However screening of family members who do not have symptoms is not required.

Can PCT be cured?
Although PCT itself can be "cured", sometimes it is not possible to cure the predisposing condition. Relapses are therefore common, and require a further phase of active treatment. Patients with familial PCT cannot be "cured" as such, but treatment can induce permanent remission if the advice that is given is adhered to.

How can PCT be treated?
The treatment of PCT will be tailored to each individual's circumstances. The main focus of treatment is to remove or decrease any triggers for PCT, to unblock the enzyme and to remove the excess porphyrin that has accumulated in the body.
While the porphyrin levels remain high, which is usually for a number of months after treatment has been started, patients remain photosensitive and should protect their skin from exposure to bright sunlight. Occasionally skin symptoms may get worse and the urine may go dark on starting treatment.

Specific treatments aimed at reducing porphyrin levels are as follows:

- If raised iron levels are found then these are reduced by regularly taking a unit of blood, the same process as followed by blood donors. The excess iron is then used by the body to make more blood and this process is repeated until the excess iron has been removed.
- Low-dose chloroquine or hydroxychloroqine may be prescribed (usually one tablet of either drug twice weekly). These drugs help 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 this very small dose is used, as larger doses can cause acute illness.

- In patients who cannot tolerate either of these two treatments, other options are available, although these are more complicated to administer.
- Women on oestrogen treatments 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.

What can I do?

- ***While treatment takes effect:*** Treatment can take several months to become effective. While the treatments take effect the skin will remain fragile and sensitive to bright sunlight. The wavelengths responsible for the photosensitivity in PCT are in the visible part of the sun's spectrum, which means that conventional sunscreens (which protect against ultraviolet) are ineffective. Therefore, you should avoid damaging the skin by wearing protective clothing (e.g. gloves) and protect the skin from sun exposure during this time.

- ***Alcohol.*** Avoiding alcohol is advised.

- ***Reactions to other medications:*** Unlike other types of porphyria PCT does not cause acute attacks or 'porphyria crises' as an effect of certain medications and anaesthetics. Some doctors and pharmacists confuse PCT with these other porphyrias and may tell you to avoid certain medicines, which is not necessary.

Where can I get more information about PCT?
PCT is uncommon, and many general practitioners will have had little experience of dealing with it. However, most dermatologists see people with PCT and are a useful source of information.

Organisations dealing with all forms of porphyria

British Porphyria Association: www.porphyria.org.uk
E-mail address: chair@porphyria.org.uk

UK Supra Regional Porphyria Services www.sas-centre.org/

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

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

American Porphyria Foundation www.porphyriafoundation.com

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