



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 (the porphyrias) caused by enzyme defects in one of the biochemical pathways in the body. These result in a build-up of chemicals called porphyrins. In PCT the porphyrins are produced from the liver and cause the skin to be very sensitive to light (photosensitive). Some of the other types of porphyria affect internal organs and can cause symptoms unrelated to the skin (acute attacks), but this does not happen in PCT.

PCT is an uncommon condition affecting about one in 25,000 of the population.

What causes PCT?

The commonest type of PCT is called sporadic or Type 1 and is not inherited. In approximately 20% of patients the condition may be due to an inherited gene mutation. This form of PCT is known as familial or type II PCT. However not all family members who inherit the gene mutation will develop the condition, as PCT requires other factors (described below) to be present as well.

The following are the most common pre-disposing factors associated with PCT:

- ***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 inherited condition called haemochromatosis that is responsible for the iron accumulation.
- ***Excessive 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?

Sun-exposed areas of skin, particularly the backs of the hands and sometimes the face and scalp are affected. The skin is fragile, and even mild injury may cause grazes and blisters that burst and heal slowly leaving scars and tiny white raised spots called *milium*. There can be alterations in skin pigmentation and an increased growth of fine hair on the cheeks and forehead. 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. Other tests, such as blood tests for liver function, glucose and iron levels may be done 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. 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. Avoiding known trigger factors, such as 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 are 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 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 chloroquine or hydroxychloroquine 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 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.

- 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 and symptoms may initially get worse. While the treatments take effect the skin will remain fragile and sensitive to bright sunlight. Although, the conventional sunscreens may not entirely be effective in blocking the spectrum of sunlight involved in PCT, using an opaque sun block along with wearing protective clothing (e.g. gloves) and modifying your exposure to the sun 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 unwanted 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 little or no experience in 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.porphyrria.org.uk

E-mail address: chair@porphyrria.org.uk

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

Internet sources:

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

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

American Porphyria Foundation: www.porphyrriafoundation.com

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: its contents, however, may occasionally differ from the advice given to you by your doctor.

**BRITISH ASSOCIATION OF DERMATOLOGISTS
PATIENT INFORMATION LEAFLET
PRODUCED JUNE 2007
UPDATED MAY 2010**

4 Fitzroy Square, London W1T 5HQ
Tel: 020 7383 0266 Fax: 020 7388 5263 e-mail: admin@bad.org.uk
Registered Charity No. 258474