



Porphyria Cutanea Tarda (PCT)

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1. What is Porphyria Cutanea Tarda (PCT)?

Porphyria cutanea tarda (PCT) is the commonest type of porphyria, affecting about one in 25,000 of the population. In PCT, porphyrins produced in excess by the liver, accumulate in the body and cause the skin to become sensitive to light.

2. What causes PCT?

PCT is caused by partial inactivation of an enzyme called uroporphyrinogen decarboxylase (UROD) (an enzyme is a protein inside our cells that converts one chemical substance into another). This leads to a build up of porphyrins in the liver from which they overflow into the blood stream and thus reach the skin. Skin problems usually start during adult years; 'tarda' means 'late'. The cause of the partial inactivation of UROD is not fully understood but it is believed to result from interaction with various factors (predisposing factors – see list below) which increase individual susceptibility to PCT.

3. Is PCT inherited?

Most patients with PCT have a form of the condition that is not inherited. This type of PCT is called **sporadic or Type I PCT**. However, in Europe, about one quarter of patients have **familial or type II PCT**. This is caused by inheritance of a mutation which inactivates one copy of the UROD gene, leaving the other to function normally (everyone has two genes for UROD in each cell in their body; one inherited from their mother and one from their father). However, most family members who inherit the gene mutation will not develop PCT because this requires other causes (predisposing factors) to be present as well.

4. What are the most important predisposing factors?

The following are the most important predisposing factors that have been identified. Most patients will have at least one.

- **Iron accumulation in the liver.** Almost all PCT patients have an increase in iron in the liver, which is believed to be the most important factor involved in the inactivation of the UROD enzyme. Some of these patients, may also have inherited a condition called haemochromatosis that is responsible for too much iron accumulating in the body.
- **Regular alcohol consumption.**
- **Viral infections of the liver.** Hepatitis C is an important predisposing factor for PCT in many European countries.
- **Oestrogen therapy.** For example the oral contraceptive or hormone replacement therapy (HRT).

5. What are the symptoms of PCT?

Only areas of the skin exposed to sunlight are affected. These are most commonly the backs of the hands, the face and scalp and any other areas of skin regularly exposed. The skin is fragile, and even mild injury or everyday tasks may cause the skin to break or form large fluid filled blisters that burst and heal slowly leaving scars and tiny white raised spots called milia. There may be changes in skin pigmentation, which can be increased or decreased, and an increased growth of fine hair on the cheeks and forehead. In addition to the skin problems, the urine may take on a dark colour.

Acute crises (the abdominal pain and nerve damage, often provoked by drugs or hormones, that occur in a group of porphyrias, called the **acute porphyrias** **DO NOT** occur in PCT.

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6. How is PCT diagnosed?

PCT is diagnosed by measuring porphyrins in samples of blood, urine and faeces. These tests look for the particular pattern of porphyrins associated with PCT. This is very important because other porphyrias can cause similar skin problems.

Your doctor should also test for the conditions described above which are associated with PCT.

These will include blood tests for

- liver function
- viral hepatitis
- Iron status and haemochromatosis



Depending on the results of these tests, you may be advised to see another doctor, for example a liver specialist or haematologist.

Identification of the type of porphyria is not normally required. Members of families in which there is more than one patient with PCT should be offered genetic counselling and may require screening for Type II PCT.

7. Can PCT be cured?

Although the underlying causes of PCT may not be curable, the condition can be well controlled and the porphyrin levels reduced to normal. It is important that known predisposing factors are controlled or treated. With appropriate treatment, the skin problems resolve slowly and eventually disappear though they may leave some scars. Relapse 2-3 years after successful treatment may occur. *This is why patients with PCT should continue seeing their doctor regularly.*

8. How can PCT be treated?

The aim of treatment is to remove or decrease any predisposing factors, to reverse the inactivation of UROD enzyme and to remove the excess porphyrin that has accumulated in the body.

You are likely to be offered one of two specific treatments:

- Regular removal (usually every two weeks) of a unit of blood (the same amount as given by blood donors) to decrease iron levels. The medical term for this treatment is venesection. The body uses iron to make more blood and the process is repeated until enough iron has been removed. This will take several months in most cases. If you also have haemochromatosis, venesection may be needed on a long-term basis.
- Low-dose chloroquine or hydroxychloroquine (usually half a tablet twice weekly). It is important that only this very small dose is used, as larger doses can cause an acute illness.

Both treatments are effective in most patients. The choice will depend on various factors, including which are considered to be the most important predisposing conditions in each patient. Occasionally both treatments may be required. In patients who cannot tolerate either of these two treatments, other options are available, although these are more complicated to administer.

Other measures:

- **Alcohol.** All alcoholic drinks should be avoided.
- **Oestrogen therapy.** Women on oestrogen treatments will be asked to stop taking them while the PCT is being treated. However once the PCT has been treated it may be possible to restart hormone treatment.

Occasionally skin symptoms may get worse and the urine can darken when treatment is started.

9. What can I do?

- **While treatment takes effect:** The skin will remain fragile and sensitive to bright sunlight for several months until treatment becomes fully effective. The skin damage in PCT is caused by the visible part of the sun's spectrum, which means that ordinary sunscreens (which protect against ultraviolet light) do not work. Reflectant sunscreens based on titanium dioxide or zinc oxide cover will be more effective. You should avoid damaging the skin by wearing protective clothing such as gloves (white cotton preferably), a hat (ideally brimmed or Foreign Legion-style), shoes rather than sandals, and minimising your exposure to the sun during this time.
- **Reactions to other medications:** Unlike other types of porphyria, PCT does not cause acute porphyria crises due to other medications. Unfortunately some doctors and pharmacists confuse PCT with the "acute porphyrias" and may tell you to avoid certain medicines. In general, you can take whatever medicines your health requires.

10. Where can I get more information about PCT?

As PCT is uncommon, most general practitioners will have little experience of the condition. However, dermatologists see most people with PCT and may sometimes ask advice from a porphyria **specialist centre**.

If you are concerned about the likelihood of passing the condition onto your children (familial PCT), you may be referred to a geneticist or porphyria specialist center for information.

The content on this website is based on a consensus agreed by EPNET partners. Although there are a number of other sources of information, the majority of which are on the internet, they may not have been validated by porphyria specialists. Most give details about all the forms of porphyria,

Dedicated Internet sources on all forms of porphyrias

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