



## WHAT ARE THE AIMS OF THIS LEAFLET?

This leaflet has been written to help you understand more about congenital erythropoietic porphyria. It explains what it is, what causes it and what treatment is available. It also explains how to manage the symptoms, and where you can find more information about it.

## WHAT IS CONGENITAL ERYTHROPOIETIC PORPHYRIA?

Congenital erythropoietic porphyria (CEP) is an extremely rare type of porphyria. It is also called Günther disease, named after the doctor who first described it. CEP affects males and females equally and is thought to affect about 1 in every 2 - 3 million people.

- The word 'congenital' describes a condition that a person is born with. This can mean the condition was present before birth. It can also mean the condition develops after birth.
- 'Erythropoietic' means the condition is related to red blood cells and their production in the body.
- 'Porphyrias' are a group of genetic conditions where the body makes too much of substances called *porphyrins*.
- Porphyrins are very important because they help the body make haemoglobin. Haemoglobin is the iron-containing protein pigment in red blood cells. It carries oxygen around the body.

The porphyrins go through a chain of reactions to make haemoglobin. If there is a 'block' in the chain, a specific porphyrin builds up in the body. Which porphyrin builds up depends on where the block happens. The high concentration of porphyrins in the body can damage the

tissues and cause the symptoms seen in people with porphyria.

## WHAT CAUSES CONGENITAL ERYTHROPOIETIC PORPHYRIA (CEP)?

An enzyme is a protein that helps convert one chemical substance into another. In CEP, an important enzyme called 'uroporphyrinogen III co-synthase' does not work correctly in all cells. This leads to the build-up of porphyrins particularly in the red blood cells. When porphyrins react with sunlight, damage to the skin and surrounding tissues occurs.

## IS CONGENITAL ERYTHROPOIETIC PORPHYRIA HEREDITARY?

Yes. People who carry the gene for CEP but have no symptoms are called *carriers*.

Each carrier has a mutation in one of their two CEP genes. If both parents are carriers, there is a 1 in 4 chance that their children will inherit this gene mutation and develop CEP. This form of genetic inheritance is known as *autosomal recessive*.

## WHAT DOES IT FEEL AND LOOK LIKE?

Around half of the people affected by CEP have a mild form of the condition. CEP can cause skin that:

- blisters easily
- is fragile
- may cause scarring

However, this mild form does not affect the eyes or cause anaemia.

The other half of affected people have the severe form. This causes their skin to be very sensitive to light (known as severe photosensitivity). Their skin blisters and is

painful after daylight exposure. In the long term, severe scarring develops.

Severe CEP also affects the eyes. It causes eye inflammation, scarring and sometimes visual impairment. A major problem caused by severe CEP is severe and long-term anaemia (a lower-than-normal level of haemoglobin in the blood).

Symptoms usually start soon after birth or early childhood. In some people, symptoms do not appear until the teenage years or early adulthood. Common symptoms include:

- Reddish pink urine. This is often the first sign noticed in newborn babies. The colour can change from day to day.
- Teeth that appear reddish-brown in colour.
- Skin that is very sensitive to light, especially direct sunlight. Even a short time in sunlight can cause painful blisters or open sores (ulcers). These usually affect areas of skin exposed to the sun, such as the backs of the hands, the face, ears and nose.
- Slow healing of blisters and ulcers or other injury. Repeated damage can leave permanent scars.
- Damage to exposed areas of skin and to other tissues deeper tissue, such as bones and tendons. This can lead to infection, scarring and disfigurement. This can include the face and hands.
- The eyes may also be sensitive to sunlight. In severe cases, long-term damage to the surface of the eye can cause scarring, and visual impairment.
- Some people affected by the more severe type of CEP can develop anaemia (a lower-than-normal level of haemoglobin in the blood) This happens because porphyrins damage red blood cells. Severe anaemia may require regular blood transfusions in

the long term. Over time, repeated blood transfusions can cause build-up of iron in the body, which may cause other health problems. Blood transfusions are given directly into a vein, to restore the body's levels of healthy red blood cells to treat severe anaemia

- Bones can become thinner and weaker, a condition called osteoporosis, which can increase the risk of fractures after minor injuries.
- Permanent patches of hair loss on the scalp may be experienced.
- Some people may develop darkening of skin that is exposed to the sun.
- Excess hair growth can occur, especially on the face and backs of the hands.

## HOW IS CEP DIAGNOSED?

The diagnosis is confirmed by measuring porphyrin levels in the blood, urine and faeces samples. These samples must be protected from light until testing is complete. A blood sample may also be taken to look for changes in the genes.

During pregnancy, prenatal testing is available for high-risk families with a known mutation if they wish. It is usually performed at 3-4 months into the pregnancy via chorionic villus sampling (CVS) or amniocentesis.

## CAN CEP BE CURED?

Severe CEP can be cured by bone marrow transplantation (BMT). This involves replacing the bone marrow of the patient with bone marrow from a healthy person (the donor).

Bone marrow is the soft, spongy, jelly-like tissue inside bones. It is where essential red blood cells, white blood cells, and platelets are produced.

If BMT is successful, the bone marrow produces healthy blood cells. This can



improve the symptoms of CEP, such as photosensitivity and anaemia. However, it cannot repair scarring caused by earlier skin or eye damage.

For BMT to be successful, the bone marrow of the donor needs to be a close match with the patient. BMT can be effective, but it is a high-risk procedure and can lead to life-threatening complications. Because of this, it is usually considered for people who are severely affected by CEP and who have a suitable donor.

Current research is focusing on gene therapy to potentially cure CEP. This approach aims to correct the faulty gene that causes CEP. Gene therapy for CEP is still being researched, and it is not yet widely available.

## HOW CAN CEP BE TREATED?

The treatment of CEP aims to reduce damage to the skin and eyes and manage complications. It is very important to stay out of the sun as much as possible.

Some or all of the following measures may be needed:

- **Protective clothing:** Clothing that covers the skin helps prevent blistering and scarring. This should include the routine use of gloves, a broad brimmed hat, scarf, long sleeves, high collars and long trousers.
- **Eye protection:** The eyes should be protected by tinted, wrap-around sunglasses or 'gaming' glasses to reduce exposure to light.
- **Sunscreens:** Most standard sunscreens protect against ultraviolet (UV) light, but the skin problems in CEP are mainly caused by visible light. Because of this, many sunscreens offer little protection. Some sunscreens (such as mineral sunscreens) may provide limited protection against visible light, but they are usually not enough on their own so should be used together with protective clothing.
- For patients who do not suffer from the anaemia, venesection, the regular removal of blood, can decrease porphyrin levels and reduce the severity of the porphyria.
- **Reducing light exposure indoors:** Patients should apply protective window films to the windows of their car and home. This helps protect against the effects of sunlight. These protective films work for around 5 years and may need replacing after this time. Some car manufacturers offer UV protective glass as standard or as an optional extra. However, most car windows do not block UV light. The non-coloured films are required for cars and are only partly protective. The more protective films are tinged orange or yellow and can be used at home. Your healthcare provider or a patient support group may be able to advise about where to purchase UV protective films. The British Photodermatology Group has released a formal, evidence-based document produced by a panel of experts to provide guidance on UV protective films.
- Curtains or blinds in the house and workplace may be needed to reduce the intensity of visible light.
- **Surgery:** Strong operating lights can damage the skin and internal organs of people affected by CEP. If you need surgery, tell your medical team and theatre staff in advance so they can take steps to reduce your exposure to these lights during surgery. Operating lights can be covered with special yellow filters that block the harmful wavelengths of light (below about 530 nm). Examples of yellow filters include TA81 XSR (Madico) or Bexfilm U (Summerside Blinds). Similar filters should also be fitted to nearby windows, including of hospital rooms and wards.



- **Skin care:** Skin affected by CEP is fragile and slow to heal. It is helpful to avoid cuts and grazes where possible, to prevent long-term scarring. To help prevent scarring, keep the skin well-moisturised and wear gloves. Blisters and ulcers should be kept clean, dressed (with bandages) appropriately and treated as soon as possible if infection develops.
- **Managing scarring and movement problems:** Repeated scarring of the skin, especially of the fingers, can limit joint movement. Regular gentle hand exercises may help maintain movement. Occupational therapy may be needed for people who have developed restricted hand movements.
- **Treating anaemia:** Some people affected by CEP develop anaemia and may need blood transfusions. Occasionally, removal of the spleen (a small organ in the chest that stores and filters blood) is necessary. If long term anaemia is a problem, then a haematologist (a doctor who specialises in blood disorders) will be involved in your care. They will monitor the course of treatment and any complications that can result from long-term blood transfusions.
- **Bone health:** CEP can lead to thinning of the bones (a condition known as osteoporosis). Treatment may include medication and taking vitamin D3 supplements long-term. Vitamin D levels should be regularly checked with blood tests.
- **Dental care:** Good oral hygiene is important to reduce the risk of tooth decay. If scarring around the mouth makes it difficult to open your mouth fully, a soft children's toothbrush or an electric toothbrush may be easier to use and cause less damage to the gums.
- **Skin camouflage:** Some people use [skin camouflage](#) products to cover visible scarring or changes in skin colour. If you would like to try this, your healthcare professional may be able to advise you about skin camouflage services and suitable products.

### CAN MEDICATIONS MAKE CEP WORSE?

No. CEP is not known to be made worse by medicines.

This means that people affected by CEP do not usually need to avoid any specific medicines because of their condition. Medicines can be taken as normal, unless the person is allergic or intolerant to a particular medication.

If you are unsure about any medications, you should discuss this with your healthcare professional.

### WHERE CAN I GET MORE INFORMATION ABOUT CEP?

*Patient support groups providing information:*

British Porphyria Association (BPA)

Web: [www.porphyria.org.uk](http://www.porphyria.org.uk)

International Porphyria Network (IPNET)

Web: [ipnet.org/](http://ipnet.org/)

Metabolic Support UK

Web: [metabolicsupport.org/](http://metabolicsupport.org/)

Web links to other relevant sources:

DermNetNZ:

[dermnetnz.org/topics/congenital-erythropoietic-porphyria](http://dermnetnz.org/topics/congenital-erythropoietic-porphyria)

Jargon

Buster: [www.skinhealthinfo.org.uk/support-resources/jargon-buster/](http://www.skinhealthinfo.org.uk/support-resources/jargon-buster/)



Please note that the BAD provides web links to additional resources to help people access a range of information about their treatment or skin condition. The views expressed in these external resources may not be shared by the BAD or its members. The BAD has no control of and does not endorse the content of external links.



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 and the British Photodermatology Group: 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*

## **BRITISH ASSOCIATION OF DERMATOLOGISTS**

### **PATIENT INFORMATION LEAFLET**

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