



CONGENITAL ERYTHROPOIETIC PORPHYRIA

What are the aims of this leaflet?

This leaflet has been written to help you understand more about congenital erythropoietic porphyria. It tells you what it is, what causes it, what can be done about it, and where you can find out more about the condition.

What is porphyria?

The porphyrias are a group of diseases caused by abnormalities in the production by the body of chemicals called *porphyrins*. Porphyrins are very important as they form haemoglobin that carries oxygen around the body in the red blood cells. The production of haemoglobin involves a chain of reactions in which one porphyrin is converted to another, and the porphyrias are diseases that result from genetic abnormalities in this process. If there is a block in the chain of reactions, there will be a build-up in the body of a particular porphyrin (which depends on where the block occurs), and porphyrins in high concentration are damaging to tissues. The problems caused by the different porphyrias relate to the particular porphyrin that accumulates.

What is congenital erythropoietic porphyria?

Congenital erythropoietic porphyria (CEP), also called *Günther's disease* after the doctor who first described it, is the rarest of the porphyrias. It is estimated that about 1 in every 2 – 3 million people are affected by CEP, which affects males and females equally, and occurs in all ethnic groups.

What causes congenital erythropoietic porphyria?

In CEP, there are high levels of a porphyrin called *porphyrinogen* in the bone marrow, blood and urine, which cause the symptoms and signs.

Is congenital erythropoietic porphyria hereditary?

Yes. The parents of someone with CEP have no symptoms of the condition themselves (and are called *carriers* of the condition), but each of them has a mutation in one of their genes. There is a 1 in 4 risk that each child born to 2 carriers will inherit the abnormal gene from both parents and thus develop the condition. This form of inheritance is called *autosomal recessive*.

What are the symptoms of congenital erythropoietic porphyria?

Individuals with CEP may not have all of the problems described in this leaflet as the severity of the condition varies. Usually, the disease shows itself soon after birth or in early childhood, but sometimes onset of disease is delayed until the teenage years or early adulthood.

- Red urine is usually the first sign noticed in newborn babies with CEP. The intensity of the redness can vary from day to day.
- The skin is very sensitive to light, especially direct sunlight, which may cause blisters or ulcers, which heal to leave scars. This most commonly happens at sun-exposed sites, for example the backs of the hands, the face, ears and scalp.
- The eyes may also be sensitive to bright sunlight or artificial light, which can cause ulcers and scarring. With time, some patients lose their eyelashes, making their eyes easily irritated by small particles of dust and fibre.
- The skin may take longer to heal after injury or blistering, and become infected.
- Anaemia, which varies in severity, is common in CEP. Anaemia develops because porphyrin damages red blood cells, and causes tiredness, shortness of breath following minimal exertion, and paleness.
- The spleen, which removes the damaged red blood cells, can gradually become bigger and cause worsening of the anaemia and a reduction in the number of platelets (the blood cells that help to form blood clots to stop bleeding) and white cells (the blood cells that fight infections) in the blood. This can lead to an increased risk of bleeding (such as repeated nose bleeds) and infections.
- CEP can occasionally cause thinning of the bones (osteoporosis), which can lead to bone fractures following minor injury.

What does congenital erythropoietic porphyria look like?

- Repeated blisters and ulcers can cause extensive scarring in sun-exposed skin and permanent bald patches on the scalp.
- Some individuals may develop darkening of sun-exposed skin.
- Excess body hair may develop, especially on the face and backs of the hands.

How is CEP diagnosed?

CEP may be suspected in children (or rarely adults) who present with the problems described in this leaflet. The diagnosis is confirmed by measuring porphyrin levels in the blood, urine and faeces. These samples need to be protected from light until tested. A blood sample may also be taken to look for changes in the genes.

Testing for CEP in pregnancy is not offered routinely. However, CEP can be diagnosed in pregnancy in families where there is already a child with CEP. In this situation, cells taken from the fluid surrounding the baby in the womb (amniocentesis) or from the placenta, at 3 to 4 months into the pregnancy, are checked for the gene mutations causing CEP.

Can CEP be cured?

Currently, the only available cure for CEP is a bone marrow transplant (BMT). This involves transplanting healthy bone marrow from another person (the donor) to the person with CEP (the recipient). Following successful BMT, the symptoms of CEP such as photosensitivity and anaemia will improve. However, the scarring from previous damage to the skin is permanent.

For BMT to succeed, the bone marrow of the donor needs to be a good match with the recipient. BMT is a high-risk procedure, and is currently reserved for those severely-affected individuals who also have a bone marrow donor who is a close match.

Research is underway to cure CEP with gene therapy. This would involve correcting the abnormal gene in the affected person. It is hoped that this research will make good progress over the next decade.

How can CEP be treated?

The treatment of CEP is aimed at preventing scarring of the skin and eyes, and reducing the complications. Some or all of the following measures may be needed:

- Protection of exposed skin from direct sunlight is required to prevent blistering and scarring. This should include the routine use of gloves, a broad brimmed hat, scarf, long sleeves, high collars and long trousers.
- The eyes should be protected by tinted, wrap-around sunglasses.
- Conventional sunscreens (that block ultraviolet light) are not effective in CEP where the photosensitivity is to *visible* light. Sunscreens that reflect visible light from the skin surface are required. Tinted reflectant sunscreens, which can be mixed to match the individual skin colour are available on prescription from Tayside Pharmaceuticals, Ninewells Hospital, Dundee, DD1 9SY, UK (Tel: +44 (0)1382 632264).

- Curtains or blinds in the house and workplace may be needed to reduce the intensity of visible light. Additionally, opaque window films may be applied to the windows of buildings and/or vehicles. It is important to check that the window film you select for your vehicle does not break the driving laws in your country.
- Cosmetic camouflage may be used to conceal scarring of the skin.
- Skin on light-exposed areas should be protected from cuts and grazes to prevent long-term scarring. This can be helped by keeping the skin well-moisturised and by wearing gloves.
- Skin ulcers need to be kept clean, dressed appropriately and infection treated in order to help healing.
- Repeated scarring of the skin, especially of the fingers, can restrict movement of joints. Regular, gentle hand exercises may help to delay or prevent this. Occupational therapy may be needed for those who have developed restricted hand movement.
- Blood transfusions may be needed to treat anaemia. Occasionally, removal of the spleen is necessary.
- Thin bones may require treatment with tablets.
- Good oral hygiene is important to prevent tooth decay. If opening the mouth is restricted due to scarring around the mouth, a soft children's toothbrush or an electric toothbrush may be easier to use and cause less damage to the gums.

Can medications make CEP worse?

CEP is not made worse by any medications. Therefore, unless the person is allergic or intolerant to a medication, individuals with CEP have no restrictions in taking any form of medication.

What other precautions are required for CEP?

If an individual with CEP needs an operation, exposure to the very bright lights of an operating theatre may result in damage to the internal organs. Theatre staff should ensure that the amount of light is minimised.

Where can I get more information about CEP?

Links to patient support groups:

British Porphyria Association (BPA): www.porphyrria.org.uk
136 Devonshire Road, Durham City, DH1 2BL, UK. (Tel: 01474 369231)

European Porphyria Initiative (EPI): www.porphyrria-europe.org

Children living with inherited metabolic diseases (CLIMB):
www.climb.org.uk

Carers UK: www.carersuk.org.uk

British Association of Skin Camouflage: www.skin-camouflage.net

British Red Cross Skin Camouflage service:
www.redcross.org.uk/skincamouflage

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.

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