

Gloves are useful, especially when driving. Wide-brimmed hats can help to protect the face. Also, using an umbrella (especially a UV reflective umbrella) whilst shadow hopping can help.

**Window films:** Some EPP sufferers have found that using film on car (and home) windows helps. With regards to car windows the rules from DVLA are:

*75% of visible light has to be able to come through the windscreen and 70% through the front side windows. There are no restrictions on rear windows.*

Doctors recommend filter films such as the following: ARC UVTA (ARC Window Films) give almost 100% protection for EPP patients. These films are amber in colour and are suitable for almost any window. However, they cannot be installed on the windscreen, or front side windows of a car, as these must be clear. UVCL (ARC Window Films) or Dermagard (available from Bonwyke, ARC Window Films and SunSibility) give about 50% protection to EPP patients - they are legal on car windscreens as they are clear/colourless.

**Phototherapy:** Phototherapy involves exposure to artificial ultraviolet light, usually for a few times a week for a few weeks in the spring, to allow the skin to thicken slightly and develop a tan. This acts as a natural sunblock and may improve tolerance to sunlight. Please see your porphyria specialist/dermatologist for more information.

**Beta-carotene:** A few people respond to high doses of beta-carotene (available on prescription). The capsules are taken orally and may turn the skin slightly orange. Self treatment is not recommended as foods and some supplements may contain chemicals that are dangerous in high doses.

**SCENESSE® (Afamelanotide):** Clinical trials are being undertaken by Clinuvel on an implant which increases the levels of melanin in the skin to shield against UV radiation (UVR) and visible light. The trial results are due to be finalised soon; we will keep you updated on the outcome.

## What can be done to relieve the pain from sun exposure?

Individuals each have their own methods of dealing with painful reactions. Avoidance of further light is essential to allow the skin time to recover. Some EPP sufferers attempt to relieve the pain with cold water or cold compresses. Some have suggested that fanned air helps to relieve the pain to a certain extent, while others have reported that immersing skin in lukewarm water can help to alleviate the pain.

### Useful contact details

**BPA telephone helpline:** 0300 30 200 30

**BPA email helpline:** [helpline@porphyria.org.uk](mailto:helpline@porphyria.org.uk)

**European Porphyria Network:** [www.porphyria-europe.com](http://www.porphyria-europe.com)

**British Association of Dermatologists:** [www.bad.org.uk](http://www.bad.org.uk)

**Rare Connect:** [www.rareconnect.org](http://www.rareconnect.org)

### Window film suppliers

- Bonwyke Ltd: 01329 289621 or [sales@bonwyke.co.uk](mailto:sales@bonwyke.co.uk)  
[www.bonwyke.co.uk](http://www.bonwyke.co.uk)
- ARC Window Films: 01942 523 078 or [sales@arcwf.com](mailto:sales@arcwf.com)  
[www.arcwindowfilms.com](http://www.arcwindowfilms.com)
- SunSibility: 0208 224 2299 or [info@sunsibility.co.uk](mailto:info@sunsibility.co.uk)  
[www.sunsibility.co.uk](http://www.sunsibility.co.uk)

### Protective clothing

A range of protective clothing and products can be found from:

- SunSibility: 0208 224 2299 or [www.sunsibility.co.uk](http://www.sunsibility.co.uk)
- Rohan: 0800 840 1411 or [www.rohan.co.uk](http://www.rohan.co.uk)
- Suntogs: 01733 765030 or [www.sun-togs.co.uk](http://www.sun-togs.co.uk)
- Equatorsun: 01932 230907 or [www.equatorsun.com](http://www.equatorsun.com)

### Dundee Cream

Dundee Pharmaceuticals, Ninewells Hospital, Dundee. DD1 9SY. Tel: 01382 632052.

### Specialist porphyria laboratories

- For an up-to-date list see the British and Irish Porphyria Network (BIPNET) website: [www.bipnet.org.uk](http://www.bipnet.org.uk).
- Also see [www.kingspath.co.uk](http://www.kingspath.co.uk) for details of tests available and sample requirements.

# British Porphyria Association

Charity No: 1089609



## Erythropoietic Protoporphyrria (EPP)

*Information for patients*



## Introduction: what is porphyria?

The 'porphyrias' are a group of eight rare disorders. Most are inherited and result from a faulty gene leading to difficulty making a chemical called haem, which is a constituent of many important proteins in the body. Haem precursor chemicals accumulate which can cause severe medical problems.

The cutaneous (skin) porphyrias give sensitivity to sunlight on exposed areas of skin. Sunlight should be avoided as much as possible. As with all porphyrias, the severity of the problem varies enormously. People with EPP do NOT need to stick to the SAFE drugs list used by those with acute porphyrias.

## Erythropoietic protoporphyria (EPP)

In EPP the inheritance pattern is complex. A severe mutation of the ferrochelatase gene (*FECH*), paired with a 'less severely affected' variant of the gene can cause EPP. In combination with other variants, the EPP gene gives no problems. There are other ways to inherit EPP, however, these are not always clear and research is still ongoing. *Please also see our 'Testing and Inheritance' leaflet for more detail.*

## What causes EPP?

In EPP there is a shortage of an enzyme (ferrochelatase), which leads to an increase in protoporphyrin levels in the blood. As blood passes through the skin, the protoporphyrin absorbs energy from sunlight. This sets off a chemical reaction that can damage the surrounding tissues and causes tingling, itching and burning pain, and possibly swelling.

Sunburn is usually caused by short wavelengths of ultraviolet light (UVB), whereas in EPP the skin is more sensitive to the longer ultraviolet wavelengths (UVA) and reacts to visible light. Fluorescent lighting can also affect severe cases of EPP.

## What are the symptoms of EPP?

EPP is different from the other skin porphyrias, in that it doesn't usually cause blistering. In sunlight, the skin becomes very painful (burning and itching) and sometimes swells or reddens (similar to that found with

a nettle rash), but many have nothing visibly wrong, despite severe discomfort.

Exposure to sunlight, for as little as a few minutes in the worst affected patients, causes burning pain which may be so severe and persistent that it prevents sleep for several nights. The time of exposure to sunlight before the pain starts varies from one person to another, as does the time taken to recover once out of the light.

Over time, the skin can thicken and scarring can develop on areas of the body easily exposed to the sun, such as the knuckles and backs of hands, the cheeks, nose and ears.

EPP normally develops in early childhood. An affected baby may cry if taken outside, or put near bright lights, or by a window during daylight. The pain makes some scream. Even when brought indoors, the baby can still be fractious while the skin recovers.

Light does not always need to be direct – light reflected off water, sand or snow, or passing through window glass may also cause symptoms.

**Anaemia:** EPP can cause slight anaemia, as the production of haemoglobin is affected, however, for many this does not affect their ability to function. Iron tablets can increase photosensitivity, so it is vital that EPP-related anaemia is not treated unless there is a clear important need.

**Vitamin D deficiency:** Due to the lack of sunlight, EPP patients can be vitamin D deficient, so levels should be checked by your doctor. If levels are reduced, your doctor may prescribe a supplement. You can also increase your intake of foods high in vitamin D, such as oily fish, eggs, meat and fortified foods.

**Liver problems:** EPP can cause liver damage in a small number of people. Although this is very rare (estimated at less than 5% of EPP sufferers), your doctor should check the way your liver is working by taking yearly blood tests. If there is a problem with liver function, this can be treated.

## How is EPP diagnosed?

The diagnosis of EPP is often suspected from the types of stories mentioned here, and can be confirmed by a blood test. This measures the amount of protoporphyrin in the

blood (serum protoporphyrin) and in the red blood cells (erythrocyte free protoporphyrin). Some doctors will also ask for a stool sample. No urine tests are needed except to exclude the other types of porphyria.

## Is there any treatment?

Unfortunately, there is no easy treatment. It is mostly a matter of avoiding the sun, bright artificial light or being close to windows (although tinted film can be attached to windows in the home and cars). It is vital to cover up with a hat and light-dense clothing when outside, and use sunblock creams.

**Sunscreens:** Conventional sunscreens formulated to protect against ultraviolet (particularly UVB) are not effective. Reflectant sunscreens based on titanium dioxide or zinc oxide will be better, as they cover both UVA, UVB, and visible light to a degree. In the UK, the SPF (sun protection factor) tells you how effective the sunscreen is for UVB, and the star rating (usually found on the back of the bottle, with a maximum four stars) provides a measure of the UVA protection.

If you haven't already used Dundee Cream it is worth a try. Ask your GP to prescribe it for you. Your GP/chemist will need to contact Dundee Pharmaceuticals (*see back page for contact details*). Dundee Cream comes in three colours: beige, white and coffee. It can be mixed with normal cosmetics to get a more realistic colour.

## Reflectant sunscreen products available on prescription and from chemists include:

- Dundee Cream
- Ambre Solaire® lotion SPF60
- Delph® lotion SPF25 or SPF30
- Sensense® Ultra SPF60
- E45 Sun® lotion SPF25 or SPF50
- Ultrablock® cream SPF30
- Vichy factor 60A

**Clothing:** Long-sleeved, close-knit fabric tops and long trousers are a must to protect as much skin as possible. Many companies now provide sun protective clothing, *please see the back page for examples.*