

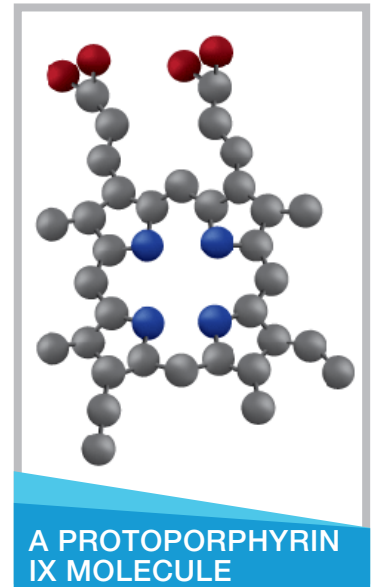
About erythropoietic protoporphyria (EPP)

What is EPP?

Erythropoietic protoporphyria (EPP) is a rare genetic blood disorder which causes an absolute intolerance to light. EPP is caused by a deficiency of an enzyme (ferrochelatase) which leads to build up of chemical called protoporphyrin IX (PPIX - right) in the body, mostly in the skin and liver.

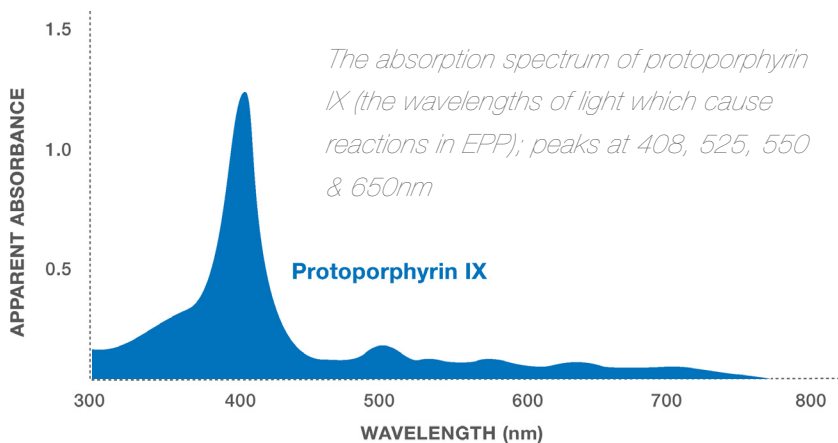
Protoporphyrin IX is phototoxic in skin: when exposed to light (particularly blue light) the protoporphyrin IX in skin reacts with varying intensity which can cause excruciating pain. This reaction can be caused by sunlight exposure, but also certain types of artificial lights and laptop screens, as well as reflective light off water, snow or concrete. Despite the reaction going on underneath the skin, there are often no immediate visible signs on the skin.

There is no effective treatment for EPP. Rather, individuals with EPP must avoid sunlight and even reflected light, often staying indoors.



In a small number of EPP cases (around 5%), liver failure can occur, so liver function test should be conducted annually. EPP can lead to vitamin D deficiency due to a lack of sunlight exposure. It can also cause anemia as those with EPP are unable to properly produce heme (haem) due to the ferrachelatase deficiency.

EPP is diagnosed by testing the levels of protoporphyrin IX in the blood. While this testing can be done by a local hospital with the correct equipment, a number of specialist centres exist globally (see below). If correctly handled, a blood sample taken at a local clinic can be shipped to a specialist centre for testing.



What happens when you have a 'reaction'?

Phototoxic reactions in EPP occur when the skin is exposed to certain wavelengths of light and the protoporphyrin IX molecules react; see the action spectrum of EPP below to understand exactly which wavelengths of light cause reactions.

Reactions are often invisible – there may be no visible sign on the skin that a reaction has occurred. Firstly the skin starts tingling or prickling then you experience an intense burning pain likened to immersing skin in boiling water. This is not like sunburn, but is a toxic reaction under the skin and can occur after only a few minutes of light exposure. For several days following an EPP reaction, skin can be painful and swollen or develop into blisters, a rash, scabs or crusts. Longer term, multiple reactions can cause scarring of the skin, especially on the hands and face.

How do you prevent an EPP reaction?

Avoiding sunlight is the most common form of prevention. For most people this means staying indoors or wearing fully sun protective clothing (hats, balaclavas, gloves, full length clothes) when outdoors. Tinted windows and specially formulated sunscreens ('Dundee cream') can help some individuals with EPP. In some cases, people with EPP change their lifestyle to live and work at night. Most people with EPP also describe 'shade hopping' when they're outdoors: moving from one location to another in shadows to minimise their exposure to light. For most people with EPP, conventional sunscreen is of little or no use as it does not block the wavelengths of light which cause EPP. High doses of beta-carotene can help in some cases of EPP, although many people describe it as ineffective.

How do you relieve an EPP reaction?

There is no set method for relieving an EPP reaction and each individual will have their own ways of dealing with a reaction. For most people, avoiding further light is essential, followed by attempting to cool the skin where a reaction has occurred using water, ice and/or fans. For some people, however, air movement can actually exacerbate an EPP reaction or symptoms. Others with EPP have reported that immersing skin in very warm water can help relieve the pain of a reaction. Finally, the use of pain killers and sleeping pills has been discussed widely, although their efficacy is debated and they should only be used in consultation with a doctor.

What is important for me to know about EPP?

When compiling this information sheet, we asked people with EPP (and their families and friends) what was most important for others to know about EPP. Here are some of the most common responses:

- ▶ People with EPP are normal, we just have a disease which prevents us from exposing our skin to sunlight;
- ▶ EPP is not an 'allergy', nor is it like sunburn; it is a rare disease which causes toxicity to light;
- ▶ Only a small amount of light exposure can cause a seriously painful reaction in EPP, but sometimes people with EPP can tolerate a little bit of sun or light exposure;
- ▶ An EPP reaction might be invisible, but it can be very painful.

Where can I learn more about EPP?

There is a very active EPP community on **Facebook**; just search groups for 'erythropoietic protoporphyria'. There are a number of associations dedicated to helping those with EPP and other forms of porphyria: The **American Porphyria Foundation**'s website has information on porphyrias, diagnosis and testing, regular meetings, clinical research and a list of [specialist centres](#).

<http://www.porphyrifoundation.com/>

The **European Porphyria Initiative** and **European Porphyria Network** were established to help physicians and Europeans living with EPP.

<http://www.porphyrria-europe.com/>

A number of regional support associations exist across Europe, South America and Australasia. For a full list of global associations, [click here](#).

Clinuvel maintains a website at <http://www.clinuvel.com/erythropoietic-protoporphyrria> with the latest news on EPP, videos and blogs from individuals with EPP and an extensive overview of the disease.



Clinuvel would like to thank all the individuals involved in the production of this information sheet for their time, energy and effort to help improve EPP resources.

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