**Erythropoietic Protoporphyria**

**What is erythropoietic protoporphyria?**

The word ‘erythropoietic’ means associated with red blood cells (‘erythro-’) and their formation (‘-poietic’). The porphyrias are a group of uncommon diseases caused by something going wrong with the production of chemicals known as porphyrins. These chemicals are the building blocks of haem, which, when combined with a protein (globin), forms haemoglobin, the material in red blood cells that carries oxygen round the body. In the case of EPP, there is a build up of one of these porphyrins (protoporphyrin) in the blood, especially in the red blood cells. This leads to a sensitivity to sunlight.

**What causes EPP?**

An enzyme is a protein that helps to convert one chemical substance into another. In EPP, there is a shortage of one particular enzyme (ferrochelatase), which normally helps to convert protoporphyrin into haem by adding iron to it. As a result of this enzyme deficiency, protoporphyrin levels build up in the blood. As blood passes through the skin, the protoporphyrin absorbs the energy from sunlight and this sets off a chemical reaction that can slightly damage surrounding tissues. The nerve endings in the skin interpret this as itching or burning pain, and if the blood vessels are affected, they can leak fluid, causing swelling.

The light that protoporphyrin absorbs is different from that which causes ordinary sunburn. Usually sunburn is caused by the shorter wavelengths of ultraviolet light (UVB), but in EPP the skin is more sensitive to longer ultraviolet wavelengths (UVA) and to visible light.

**Is EPP hereditary?**

Yes, but there is not always a family history of the condition. Everyone has two genes for ferrochelatase in each cell in their body (one coming from their mother and one from their father). In most families, EPP occurs when an affected individual inherits a gene for a severely underactive ferrochelatase enzyme from one parent, and a less severely affected gene from the other parent. The less severely affected gene is quite common, being present in about 10% of the general population, but it never causes EPP by itself. The genetics is quite complex and advice from your local genetics service may be useful.

**What are the symptoms of EPP?**

Typically EPP starts with abnormal sensitivity to sunlight. Exposure to sunlight causes tingling, itching or burning, which may be associated with redness and swelling. These symptoms usually occur within a few minutes of skin exposure to sunlight, and often they take hours or days to resolve. During this time the skin may feel more sensitive than usual to extremes of temperature. The light producing these changes need not be direct – light reflected off water and sand, or passing through window glass, including car windscreens, can also cause the symptoms.

EPP usually starts in childhood, and affects males and females equally. Infants may cry or scream after being taken out into the sunlight; and older children may complain of burning and try to wave their hands in the air, or put them into cold water to try to relieve the pain. A very small number of people who have had with EPP for many years may develop liver damage. Fortunately this is rare.

**What does EPP look like?**

Despite severe discomfort, there may be nothing abnormal to see on the skin. Sometimes there can be swelling of the skin, initially like a nettle rash. With time, some people develop thickening of the skin over their knuckles, and small scars on sun-exposed skin such as that on the cheeks, nose, and backs of the hands. However these skin changes show wide variation between different individuals.

**How is EPP diagnosed?**

The diagnosis is usually suspected from the story, 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 to measure the level of protoporphyrin in the faeces. No urine tests are relevant to this condition except to exclude other types of porphyria.

Although it is unlikely that you will develop liver problems as a complication of EPP, your doctor may monitor the way your liver is working by yearly blood tests. If there is any evidence of a deterioration in liver function, there are certain interventions that may help to halt or reverse this.

As EPP affects the production of haemoglobin, it is not uncommon for people with EPP to be slightly anaemic. Your doctor will probably also measure your blood count to make sure that you are not becoming too anaemic.

**Can EPP be cured?**

At present there is no cure for EPP.

For information on available treatments please go to [this page](http://www.bad.org.uk/site/817/default.aspx)on the website of the British Association of Dermatologists

