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Climb™

Supporting those affected by Inherited Metabolic Disorders

# Congenital Erythropoietic Porphyria

The Porphyrias are a group of disorders that are caused by a problem in the production of Haem resulting in an accumulation of either porphyrins, or the chemicals used to make them. Porphyrins are needed to enable Haemoglobin to work properly. This is a vital protein found in red blood cells that carries oxygen from the lungs to the tissues in the body and returns with carbon dioxide back to the lungs. It also gives blood its red colour when it contains oxygen. Porphyrins combine with iron to produce Haem (a component of Haemoglobin) which also helps to form many other proteins. It is found in all body tissues, mainly in red blood cells, bone marrow and in the liver. CEP is the rarest of the Porphyria's and is characterised by hypersensitivity to light, particularly direct sunlight or artificial light, causing blistering, ulcerations and scarring of the skin and eyes which can take a long time to heal and/or become infected. One of the first signs of CEP is usually a red discolouration of the urine noticed in newborns. Varying degrees of anaemia is common.

## Synonyms

Alternative names for this condition are:

- CEP
- Günther's Disease

Further information about this condition is available from Climb.

## Disclaimer



Please read our disclaimer and information on data protection.

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