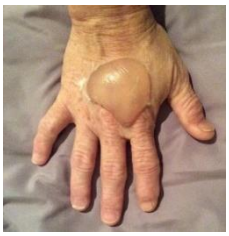
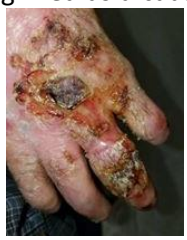
AMERICAN PORPHYRIA FOUNDATION

Congenital Erythropoietic Porphyria (CEP)

What is CEP?

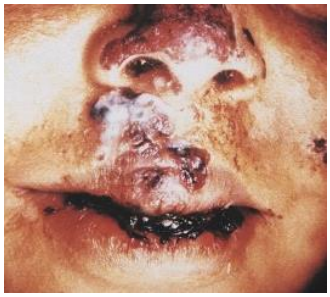
Congenital Erythropoietic Porphyria, also known as Gunther's disease, is an extremely rare autosomal recessive disease. The deficient enzyme is uroporphyrinogen III synthase (UROS). Various mutations in this gene have been identified. Like many of the erythropoietic porphyrias, symptoms appear in infancy. CEP is sometime recognized as a cause of anemia in a fetus prior to birth however, in less severe cases, symptoms will appear during adult life. Adult-onset causes are sometimes associated with expansion of a clone of cells in the bone marrow with mutated UROS. Porphyrins are markedly increased in bone marrow, red blood cells, plasma, urine, and feces in all cases of CEP. Porphyrins are deposited in the teeth and bones before birth.





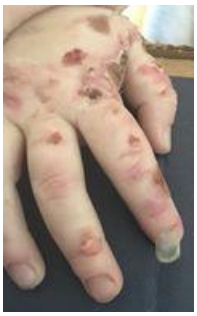
What are the symptoms?

- Skin photosensitivity may be extreme and lead to blistering, severe scarring, and increased hair growth.
- Bacteria may infect the damaged skin.
- Facial features and fingers may be severely affected, or even destroyed, by phototoxic damage and infection.
- Due to the shortened life span of red blood cells, anemia is often a result.
- Deficiency in UROS activity.
- Heme and hemoglobin are increased to compensate for the shortened life of red blood cells.



What is the treatment and prognosis?

- Blood transfusions and possible removal of the spleen may reduce porphyrin production by the bone marrow.
- Marrow stem cell transplantation has been effective and is the preferred treatment in severe childhood causes.
- Gene therapy is being investigated and may be used in the future.



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