# Congenital Erythropoietic Porphyria (CEP)

## OVERVIEW

CEP, also known as Günther disease, is very rare, with only several hundred cases reported in the world literature. The prevalence is not known, but probably is less than 1 in 1,000,000. It is due to the markedly deficient activity of the heme biosynthetic enzyme, uroporphyrinogen III synthase (UROS).

## SYMPTOMS

CEP is one of the most severe porphyrias. Symptoms usually begin soon after birth or in early childhood. Some severe cases have been diagnosed prenatally with hemolytic anemia and non-immune fetal hydrops. Severe early-onset patients typically become transfusion-dependent secondary to hemolytic anemia and ineffective erythropoiesis, and have extreme photosensitivity. Less severe patients, who have more residual UROS enzymatic activity, may not be transfusion-dependent, but will have cutaneous photosensitivity. Adult-onset cases may occur due to myelodysplasia.

The cutaneous photosensitivity results in severe blistering and, following their rupture, can lead scarring and to secondary infections of the skin and bone.

## LONG TERM COMPLICATIONS

Photomutilation can result in the loss of facial features (nose, ear and lids) and digits. Hypertrichosis on sun-exposed skin, reddish-brown colored teeth (erythrodontia), and reddish-colored urine are common features. There may be bone fragility due to expansion of the bone marrow and vitamin D deficiency. In severe causes, erythrocytes have a shortened life-span, and mild or severe hemolytic anemia results, along with increased erythroid synthesis and splenomegaly.

## TREATMENT & PROGNOSIS

Chronic erythrocyte transfusions to maintain a hematocrit of >35% are required in severe transfusion-dependent cases to reduce porphyrin production by the marrow. In transfusion-dependent patients, bone marrow transplantation may be considered as this is a curative treatment for severely affected CEP patients. Recent results indicate that iron reduction therapy with phlebotomies lead to decreases in porphyrin levels and in severity of CEP.

## MANAGEMENT

Newborns with red-colored urine in their diapers should not undergo phototherapy for hyperbilirubinemia.




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## MANAGEMENT (continued)

### PREVENTION

Avoidance of sunlight is most important in the management of CEP. Protective clothing is a must, and special tinted glass on house and car windows is strongly recommended.

### FAMILY TESTING & COUNSEING

CEP is an autosomal recessive disorder. Identifying the causative UROS mutations in a family enables prenatal and pre-implantation genetic diagnoses for at-risk pregnancies.

References

1. Erwin A, Desnick RJ. Congenital Erythropoietic Porphyria: Recent Advances. 2019




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