



Hepatoerythropoietic Porphyria (HEP)

OVERVIEW & SYMPTOMS

HEP is a very rare type of porphyria, due to mutations in both copies of the UROD gene resulting in severe deficiency of UROD enzyme activity in all cells, the same enzyme that causes PCT. The main manifestation of HEP is skin blistering and is more severe than that observed in PCT. The blistering begins in infancy and resembles other severe cutaneous porphyrias such as CEP. Sun protection is critical for management. HEP is an autosomal recessive disorder, all children of an affected individual will have familial PCT, but like likelihood of developing symptoms is low. The parents of a child with HEP will also have familial PCT.