Erythropoietic Protoporphyria

National Cancer Institute

Source

National Cancer Institute. Erythropoietic Protoporphyria. NCI Thesaurus. Code C84698.

A rare congenital metabolic disorder characterized by an inborn error of porphyrin-heme biosynthesis. It is caused by deficiency of the enzyme ferrochelatase. Signs and symptoms include painful cutaneous photosensitivity leading to blistering and scarring of the exposed skin areas, erythrodontia, red discoloration of urine, hemolytic anemia, and splenomegaly.