

Congenital erythropoietic porphyria (CEP) is a very rare inherited metabolic disorder resulting from the deficient function of the enzyme uroporphyrinogen III cosynthase (UROS), the fourth enzyme in the heme biosynthetic pathway. Due to the impaired function of this enzyme, excessive amounts of particular porphyrins accumulate, particularly in the bone marrow, plasma, red blood cells, urine, teeth, and bones. The major symptom of this disorder is hypersensitivity of the skin to sunlight and some types of artificial light, such as fluorescent lights (photosensitivity). After exposure to light, the photo-activated porphyrins in the skin cause bullae (blistering) and the fluid-filled sacs rupture, and the lesions often get infected. These infected lesions can lead to scarring, bone loss, and deformities. The hands, arms, and face are the most commonly affected areas. CEP is inherited as an autosomal recessive genetic disorder. Typically, there is no family history of the disease. Neither parent has symptoms of CEP, but each carries a defective gene that they can pass to their children. Affected offspring have two copies of the defective gene, one inherited from each parent. CEP is a very rare genetic disorder that affects males and females in equal numbers. Over 200 cases have been reported worldwide.