Gunther’s Disease

History
Congenital erythropoietic porphyria (CEP) is a very rare metabolic disorder affecting the synthesis of haem, the iron-containing pigment that binds oxygen onto red blood cells. It was initially described by Hans Gunther so is also known as Gunther disease. The typical complaint is blistering and fragility of light-exposed skin in an individual with discolored urine. The presentation of erythropoietic porphyria at birth in a patient with a history of a difficult perinatal course and concomitant jaundice usually indicates severe disease. Patients may have a history of hemolytic anemia before the complete diagnosis was recognized. Very early prenatal expression with nonimmune hydrops fetalis has been reported.

Symptoms
Blistering, scarring, and increased hair growth. Affected skin may harbor bacteria, and facial features and fingers may be lost through sun damage and infection.

Treatment
CEP persists lifelong. It is essential to protect the skin from all forms of daylight to reduce pain, burning, swelling, and itching. Indoors, incandescent lamps are more suitable than fluorescent lamps and protective films can be placed on the windows to reduce the light that provokes porphyria.

Tests And Diagnosis
A test done on a baby that died from Gunther’s disease showed that determination of the enzymes of haem biosynthesis in erythrocytes and lymphocytes showed that both parents possessed only 50% of the normal activity of cosynthase. A previously described point mutation in codon 73 was observed in one parent. Fatal cases of neonatal Günther’s disease are extremely rare and such an observation, according to our knowledge, is probably one of the first described.

Definition
Congenital erythropoietic porphyria (CEP) or Günther’s disease is the rarest form of the porphyrias. The disease is usually diagnosed at birth or during early infancy, but rarely in utero.

Management
Management of the disease is often difficult because hemolytic anemia, splenic sequestration, and thrombocytopenia often necessitate repeated transfusions, which in turn can lead to iron overload. Splenectomy is often indicated. Intense photoprotection is required to prevent the appearance and aggravation of cutaneous lesions. There is a constant risk of the lesions becoming infected, but generally this is controlled by antibiotic therapy.

Sources
- Pannier, Dr. E, et al. (2002) Congenital erythropoietic porphyria (Günther’s disease): two cases with very early prenatal manifestation and cystic hygroma.