caused the protoporphyrin concentration to be reduced to a less toxic level while repressing ALA synthetase production and reducing porphyrin synthesis. (ref 32)

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Congenital Erythropoietic Porphyria

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Congenital erythropoietic porphyria, also called G•nther's disease, is extremely rare; fewer than 100 cases have been described. It is inherited as an autosomal recessive trait. The enzymatic basis for this disorder appears to be a deficit of uroporphyrinogen cosynthetase; the enzyme cocatalyzes the synthesis of the asymmetric uroporphyrinogen III molecule. This abnormality leads to erythrocyte overproduction of uroporphyrinogen I and coproporphyrinogen I, which have no biologic function in the synthesis of heme. Oxidation of these products to their respective porphyrins, with accumulation and excretion of uroporphyrin I and coproporphyrin I, begins in the fetus.

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Excess porphyrin causes staining of bones and teeth (erythrodontia), hemolysis, dark urine, and photosensitivity, all of which are usually identified early in infancy. The most dramatic findings are related to repeated vesiculation, scarring, and mutilation, which follow exposure to sunlight. These late changes may be mistaken for scleroderma. Alopecia, conjunctivitis, and corneal inflammation are additional findings. Bone deformities, attributed to the toxic effects of porphyrin deposition in bone and perhaps to rickets secondary to avoidance of sunlight, are sometimes present. Hemolytic anemia is common and is often accompanied by splenomegaly. Hypersplenism with deficiency of platelets may occur.

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Fluorescence of teeth as well as of red blood cells and their precursors in the blood and bone marrow can be demonstrated by irradiation with the Soret band. As with other photosensitive porphyrias, window glass and conventional ultraviolet sunscreens are ineffective in blocking the offending wavelengths. Suppression of erythropoiesis and therefore heme synthesis can be achieved by transfusion and, when hemolysis is present, by splenectomy. Consequently, urinary excretion of uroporphyrin I and coproporphyrin I is reduced. Treatment also includes avoidance of sunlight and topical care for skin lesions.

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Oral sorbents such as cholestyramine and, more recently, charcoal have been advocated to decrease reabsorption of porphyrins from the intestine. Prompt and effective decreases of porphyrin concentrations in the urine, plasma, and skin and resolution of photosensitivity have been documented during therapy. (ref 33) Even with such treatment, however, progressive mutilation and premature