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unit of blood twice monthly for a total of 5 to 10 L, with treatment guided by the patient's hematocrit level, decreases both uroporphyrin excretion and photosensitivity. Improvement occurs within several months to a year. Preexisting significant anemia or hemodynamic instability obviously precludes such treatment. For patients who cannot tolerate phlebotomy, slow subcutaneous infusion of deferoxamine by portable syringe pump (1.0 to 1.5 g in 8 to 10 ml of sterile water for eight to 10 hours nightly, administered five nights a week for two to five months) has produced the same results. (ref 30) Avoidance of sunlight, alcohol, iron, and estrogens is equally important.

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Treatment of photosensitivity with á-carotene or chloroquine may be tried ; however, results have been erratic.

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Erythrohepatic and Erythropoietic Porphyrias

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Two porphyrias are caused by a major enzymatic abnormality in the erythropoietic system: protoporphyria (which also involves a defect in hepatic cells) and congenital erythropoietic porphyria. In contrast to porphyrin synthesis in the liver, porphyrin synthesis in the erythropoietic system is responsive to changes in red blood cell mass and levels of tissue oxygenation. Therefore, anemia, hypoxia, and erythropoietin are important regulators of heme (and hemoglobin) production. Only the reticulocytes and their precursors in the bone marrow are responsive to these stimuli. The mature red blood cells, which lack mitochondria and a nucleus, no longer possess the complete machinery for porphyrin synthesis.

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Protoporphyria

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Because protoporphyria involves overproduction of protoporphyrin by red blood cells as well as liver cells, it has also been referred to by two other terms: EPP and erythrohepatic porphyria. Inherited as an autosomal dominant trait, it is attributed to partial absence of the mitochondrial enzyme ferrochelatase, also called heme synthetase, which binds Fe[2+] to protoporphyrin IX. Normoblasts and hepatocytes are the cells primarily involved in determining the clinical picture, although the enzymatic defect has also been observed in fibroblasts. Protoporphyria is not associated with increased urinary excretion of porphyrins, ALA, or PBG. Levels of fecal protoporphyrin are often, but not regularly, increased.

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The dominant feature of this disorder is photosensitivity, probably caused by increased levels of plasma protoporphyrin. Excess plasma protoporphyrin results from overproduction by hepatocytes as well as by erythrocyte