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PCT exhibits various prominent clinical and diagnostic features [see Table 4]. Because ALA synthetase is not induced, excretion of ALA and PBG is normal and results of the Watson-Schwartz test are negative. As might be expected, PCT is not associated with acute neuropathic symptoms.

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In the familial form of PCT, inherited as an autosomal dominant trait, there is an approximately 50 percent reduction in the activity of uroporphyrinogen decarboxylase in red blood cells as well as in the liver. This finding is compatible with the inheritance of a single defective gene coding for this enzyme. Clinical expression is variable, and latent cases have been documented. The offspring of two heterozygotes may be a phenotypically homozygous individual (i.e., by pedigree and by virtue of more severe symptoms) with severe uroporphyrinogen decarboxylase deficiency (designated HEP) . The childhood onset and greater severity of symptoms in this form of the disease are sometimes accompanied by erythrodontia, hepatosplenomegaly, and hemolytic anemia, thus mimicking CEP (also called G•nther's disease) . (ref 27) The defective gene for uroporphyrinogen decarboxylase has been isolated from two children with hepatoerythropoietic porphyria and has been cloned. The significant sequence change was a point mutation (G • A) at position 860 of the gene. This missense mutation substituted glutamate (coded for by GAG) for glycine (encoded by GGG) at position 281 in the amino acid sequence of the protein. The mutant enzyme underwent very rapid degradation in a cell lysate when compared with the normal protein. (ref 28) Other variants of the enzyme probably exist. (ref 27)

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The presence of abnormal blood and tissue levels of uroporphyrin promotes photosensitivity. The skin lesions of PCT are the same as those that are observed in both VP and HCP. Irradiation (400 to 410 nm) of the skin of patients who have PCT leads to a decrease i

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n the hemolytic titers of C3 and C5 in the serum. This finding was confirmed by in vitro studies that demonstrated activation of the complement system by irradiation of serum from patients with PCT or by irradiation of normal serum to which uroporphyrin or protoporphyrin was added. (ref 29)

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The onset of skin lesions is usually gradual rather than acute. PCT can be chemically distinguished from VP and HCP by the preponderance of uroporphyrin instead of coproporphyrin in the urine and the absence of marked elevation of protoporphyrin levels in the stool.

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Iron overload is not always demonstrable by measurement of serum iron levels. Nevertheless, repeated phlebotomy of one