Table I

Human Porphyrias. Major clinical and laboratory features

	Porphyria	Deficient enzyme	Inheritance	Main symptoms	Increased porphyrin precursors and/or porphyrins: Erythrocytes	Urine	Stool
Hepatic	**5-ALA dehydratase-deficient porphyria (ADP)**	ALA-dehydratase	AR	NV	Zn-protoporphyrin	ALA, coproporhyrin III	-
Hepatic	**Acute intermittent porphyria (AIP)**	HMB-synthase	AD	NV	-	ALA, PBG, Uroporphyrin	-
Hepatic	**Porphyria cutanea tarda (PCT)**	URO-decarboxylase	AD	CP	-	Uroporphyrin 7-carboxylate porphyrin	Isocopro-porphyrin
Hepatic	**Hereditary coproporphyria (HCP)**	COPRO-oxidase	AD	NV & CP	-	ALA, PBG, coproporhyrin III	Coproporhyrin III
Hepatic	**Variegate porphyria (VP)**	PROTO-oxidase	AD	NV & CP	-	ALA, PBG, coproporhyrin III	Coproporhyrin III, protoporphyrin
Erythropoietic	**Congenital erythropoietic porphyria (CEP)**	URO-synthase	AR	CP	Uroporphyrin I, coproporhyrin I	Uroporphyrin I, coproporhyrin I	Coproporhyrin I
Erythropoietic	**Erythropoietic protoporphyria (EPP)**	Ferrochelatase	AD	CP	Protoporphyrin	-	Protoporphyrin

AR, autosomal recessive; AD, autosomal dominant; NV, neurovisceral; CP, cutaneous photosensitivity.