Diagnosis and Treatment of the Porphyrias- The porphyrias are very rare conditions and there are many other diseases with similar symptoms. If you think you have symptoms of a porphyria please see your primary care physician and refer them to the Porphyrias Consortium website for more information. http://www.rarediseasesnetwork.org/porphyrias/index.htm

Type of Porphyria		Most Common Symptoms	Biochemical Lab Tests to Diagnose Condition	Labs to Use	Treatments
Acute Porphyrias	**Acute Intermittent Porphyria (AIP)**	Acute attacks of severe abdominal pain, nausea, vomiting, rapid heartbeat and other symptoms. These attacks generally last for several days, and can be sporadic or happen frequently (~once/month). Sometimes they can be triggered by certain medications. Generally symptoms start sporadic.	• Urine Porphobilinogen (PBG) done during an acute attack	UTMB, ARUP, Mount Sinai**, Mayo, Quest, LabCorp	Panhematin infusions are given for acute attacks. Avoiding any known triggers of acute attacks; possible triggers can be found on the APF website.
	Variegate Porphyria (VP)	Can have the same acute attacks as in AIP, also can have blistering on sun exposed areas of the skin. Generally symptoms start in adulthood.	• Urine PBG done during an acute attack • Urine total porphyrins • Plasma porphyrins	UTMB, ARUP, Mayo, Quest, LabCorp	Same as AIP for acute attacks, and avoiding sun exposure for blistering.
	Hereditary Coproporphyria (HCP)	Similar symptoms to VP.	• Urine PBG done during an acute attack • Urine total porphyrins • Plasma porphyrins • porphyrins	UTMB, ARUP, Mayo, Quest, LabCorp	
Porphyria Cutanea Tarda (PCT)		Blistering and skin fragility (skin that tears easily) on the sun exposed areas of the skin. Generally symptoms start in adulthood.	• Urine total porphyrins • Plasma porphyrins	UTMB, ARUP, Mayo, Quest, LabCorp	Avoiding sun exposure, and regular phlebotomies (removal of certain amounts of blood), or low doses of hydroxychloroquine (a medication).
Erythropoietic Protoporphyria (EPP) and X-linked Protoporphyria (XLP)		Severe pain on sun exposed areas of the skin, with swelling, lasting several days. Generally there is no blistering with the pain. Symptoms usually start in childhood.	• Erythrocyte protoporphyrin • Plasma porphyrins	UTMB, ARUP or Mayo	Avoiding sun exposure.
Congenital Erythropoietic Porphyria (CEP)		Severe blistering on sun exposed areas of the skin which can result in infections and scarring. Generally symptoms start in early childhood; in very severe cases symptoms can begin right at birth.	• Urine total porphyrins • Plasma porphyrins	UTMB, ARUP, Mayo, Quest, LabCorp	Avoiding sun exposure and treatment of infections if they occur.

NOTE—To diagnose all of the porphyrias GENETIC TESTING is also recommended. Information on genetic testing can be found at: http://icahn.mssm.edu/departments-and-institutes/genomics/genetic-testing/test-catalog

**The Mount Sinai Lab only tests for urine PBG