I authorize the Mount Sinai Genetic Testing Laboratory of the Department of Genetics & Genomic Sciences, Mount Sinai School of Medicine, New York to analyze a sample of my (my child's/my ward's) blood for the purpose of determining if I (my child/my ward) have (has) a mutation(s) which causes one of the acute porphyrias, acute intermittent porphyria (AIP), hereditary coproporphyria (HCP), and variegated porphyria (VP), or one of the cutaneous porphyrias, congenital erythropoietic porphyria (CEP), familial porphyria cutanea tarda (f-PCT), hepatocerebrohepatic porphyria (HCP), and erythropoietic protoporphyria (EPP). The Mount Sinai Genetic Testing Laboratory of the Mount Sinai School of Medicine is New York State/CLIA approved for these tests.

I understand that:

- The purpose of this testing is to determine if I (my child/my ward) have (has) a mutation(s) which causes one of the porphyrias.
- Mutation analysis will only be performed for the porphyria(s) that have been requested by my/my child's/my ward's referring physician.
- In diagnostic testing, rare errors can occur, for example, due to sample mix-up, and/or laboratory errors. Genetic polymorphisms may also cause rare errors in mutation analysis.
- A “positive” result from the testing indicates that a mutation (or my family’s mutation) in the gene causing porphyria has been identified in my sample.
- A “negative” result from the testing indicates either: 1) the lab did not find my family’s mutation and, therefore, I do not have the porphyria present in my family, or 2) the lab did not detect a mutation in the gene that causes my suspected porphyria. This could be because: a) I do not have the suspected porphyria; b) I have another porphyria caused by a mutation in a different gene or c) there are limitations of the testing.
- Genetic counseling will be available to me about the results of the tests and their effect on my family by a genetic counselor.
- A written report of the results will be sent to my physician(s).
- The results of this diagnostic testing and any medical information that I provide are confidential and will not be used for other purposes unless I give MY signed consent.
- I give the Mount Sinai Genetic Testing Laboratory permission to store my DNA labeled with my name indefinitely.

Please Initial: □ Yes □ No

Signature: ___________________________ Date: ______________________
Printed Name: ___________________________ Witness: ___________________________

If you have any questions, please contact Dana O. Doheny, MS, CGC, Genetic Counselor, by telephone (212-659-6779, direct, or 866-322-7963, toll-free) or email (dana.doheny@mssm.edu) to discuss our porphyria testing and sample requirements. THANK YOU.