Icahn School of Medicine at Mount Sinai

MOUNT SINAI GENETIC TESTING LABORATORY

Porphyria DNA Testing
Atran Building, 1428 Madison Avenue, Room 2-25
New York, NY 10029
Tel: 212-241-7518; Fax: 212-659-6780; Email: Porphyria@mssm.edu

MOLECULAR TESTING OF ACUTE AND CUTANEOUS PORPHYRIAS
Blood Collection and Shipping Instructions

The molecular diagnosis of Acute Intermittent Porphyria (AIP), Congenital Erythropoietic Porphyria (CEP), familial Porphyria Cutanea Tarda (f-PCT) Hepatoerythropoietic Porphyria (HEP), Hereditary Coproporphyria (HCP), Variegate Porphyria (VP), autosomal recessive Erythropoietic Protoporphyria (EPP), and X-Linked Protoporphyria (XLP) requires one of the following specimens:

Sample Requirements:

Assay	Sample Type	Adults / Older Children	Infants/ Children
DNA Analysis	EDTA (lavender top) tube AND	20 ml whole blood	5 ml whole blood
	Or DNA (from whole blood)	For 3 genes, at least 100 µl; for 1 gene, at least 30 µl; concentration >200 ng/µl	
	Or Buccal Cells (only available for individuals with known family mutation)	2 buccal brushes per person; buccal brushes must be requested from laboratory	
Cell Line (special request)	ACD (yellow-top) tube	7-10 ml whole blood (wipe top with alcohol first)	
Prenatal Testing	(1) 2-3 T-25 flasks of cultured cells AND 1 control flask, AND (2) 7-10 whole blood in EDTA (lavender-top) tube from mother (for MCC)		

The laboratory is New York State Department of Health and CLIA approved for molecular diagnosis of the porphyrias.

Fees for Mutation Analysis (effective 05/01/2014):
Fees schedule is on the requisition.

Payment must be sent with the blood samples, either in the form of a check, made payable to The Mount Sinai Genetic Testing Laboratory or the patient's credit card information (see attached form). The patient will receive a bill which can be submitted by the patient to his/her insurance company. Arrangements for institutional billing can be made by contacting the genetic counselor by telephone (866-322-7963, toll-free, or 212-659-6779, direct-line) or email (dana.doheny@mssm.edu).

TO OBTAIN CPT CODES FOR THIS TESTING OR FOR ANY BILLING QUESTIONS, PLEASE CALL THE BILLING OFFICE AT 212-241-8717.

Shipping Instructions:
Please include with the blood sample: 1) the signed requisition form, 2) the patient's pedigree, and the 3) consent form signed by the patient or patient's parent or guardian, 4) payment, and 5) copies of any biochemical tests that suggest the patient's diagnosis of a specific porphyria.

Ship the specimen at room temperature by an overnight carrier (Federal Express or DHL) to the address below. The specimens must arrive within 24 hours of collection. Specimens should be shipped Monday through Wednesday only. Please notify us by telephone (212-659-6779), Fax (212-659-6780), or email (dana.doheny@mssm.edu) prior to shipment. Arrangements for international shipments should be made in advance by fax or email before obtaining the samples.

Shipping Address: **Mount Sinai Genetic Testing Laboratory**
Porphyria DNA Testing
Atran Building, 1428 Madison Avenue, Room 2-25
New York, NY 10029
Tel: 212-241-7518

If you have additional 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 or porphyria@mssm.edu) to discuss our porphyria testing and sample requirements.

_____ _____
Referring Physician Signature *Date*

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FOR LAB USE ONLY
Date rec'd: _____
Payment: _____
Family: _____
Mutation: _____
☐ EDTA blood
☐ Buccal Swab
☐ DNA
☐ Sputum
☐ Cord blood
☐ Amnio / CVS
Note:

REQUISITION FOR PORPHYRIA DNA TESTING

(CLIA/NYS Approved PFI #3516)

Patient's Name: DATE blood drawn:
Last *First* *MI*

Patient's Address:
Street *City* *State* *Zip*

Date of Birth: / / ☐ Male ☐ Female

Telephone: Email:
(Day) *(Evening)*

Ancestry (Countries of Origin): Mother:_____ Father: _____

Race: ☐ Caucasian ☐ Black ☐ Asian ☐ Native American ☐ Hispanic ☐ Other: _____

Referring Physician: *(By New York State Law, A Referring Physician Must be Listed; The Results of our Testing will be sent to this Physician):*

Name of Referring Physician: NPI#:

Address:

Tel: Fax: Email:

If you wish the results sent to another Physician, please provide the Physician's name, address, phone, & fax:

Name of Physician:

Address:

Tel: Fax: Email:

MOLECULAR LABORATORY TEST(S) ORDERED (PLEASE CHECK DESIRED TEST(S)):

Please check	Porphyria Type	Gene Analyzed	Cost Per Assay
☐	Acute Intermittent Porphyria (AIP)	HMBS	$898
☐	Hereditary Coproporphyria (HCP)	CPOX	$874
☐	Variegate Porphyria (VP)	PPOX	$802
☐	Acute Porphyrias Panel (AIP, HCP, & VP)	HMBS, CPOX, & PPOX	$1,882
☐	Congenital Erythropoietic Porphyria (CEP)	UROS + ALAS2 (Exon 11)	$1028
☐	Porphyria Cutanea Tarda (PCT)	UROD	$826
☐	Erythropoietic Protoporphyria Panel	FECH + ALAS2 (Exon 11)	$1148
☐	Targeted Mutation Analysis *(Family mutation has previously been identified)*		
☐	for Autosomal Dominant and X-Linked Porphyrias (AIP, HCP, VP, PCT, XLP)		$226
☐	for Autosomal Recessive Porphyrias (EPP, CEP, HEP)		$552
☐	for FECH low-expression SNP only		$226
	Name of family member previously tested:		

ICD9 Code: 277.1 Tax ID# 13-6171197 CPT codes available upon request.

The Mount Sinai Genetic Testing Laboratory is New York State/CLIA approved for the above tests

Please include all completed and signed paperwork with the sample. For questions, please contact the genetic counselor by telephone (866-322-7963, toll-free or 212-659-6779, direct-line) or email (dana.doheny@mssm.edu).

PAYMENT MUST BE SENT WITH THE BLOOD SAMPLES, either in the form of a check, made payable to The Mount Sinai Genetic Testing Laboratory, credit card information, insurance information, or institutional billing information. Please see payment form for additional information.

_____ _____
Referring Physician Signature *Date*

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PAYMENT INFORMATION

Date: _____

Name: _____ Phone: _____
DOB: _____ Fax: _____
Address: _____ Email: _____
_____

TO OBTAIN CPT CODES FOR THIS TESTING OR FOR ANY BILLING QUESTIONS, PLEASE CALL THE BILLING OFFICE AT 212-241-8717.

☐ I have enclosed a **CHECK** (payable to *Mount Sinai Genetic Testing Lab*); amount of check is $_____.

☐ I have provided **CREDIT CARD information** (below).

THE MOUNT SINAI GENETIC TESTING LABORATORY PARTICIPATES WITH THE FOLLOWING INSURANCE PLANS for which we will bill directly for testing services. If you have one of these insurance carriers, please check the appropriate box, and complete the Medical Insurance information below. I understand that I may be billed for some of the fees not covered by insurance.

☐ Aetna ☐ GHI PPO/ HIP/Emblem Health ☐ United HealthCare

☐ Cigna ☐ Oxford ☐ New York State Medicaid (straight Medicaid, NOT managed care Medicaid)

☐ I have obtained pre-authorization for **INSTITUTIONAL BILLING**, and have discussed arrangements with the Porphyria Laboratory contact person; I have provided mailing information of institution's contact person responsible for payment of services (below).

CREDIT CARD INFORMATION

I hereby authorize Mount Sinai Genetic Testing Lab to charge my ( ) Visa ( ) MasterCard ( ) American Express

Name on Card: _____ Credit Card Number: _____
Address of Card Holder (if different from patient): Expiration Date: _____
_____ Dollar Amount charged: $_____
_____
Card Holder Signature: _____

MEDICAL INSURANCE INFORMATION (including New York State Medicaid))
(please attach copy of front and back of card)

Name of Insured: _____ Insurance Phone: _____
Relationship to patient: _____ Policy #: _____
Insured's DOB: _____ Group #: _____
Insured's Address: _____ Group Name: _____
_____ Group Address: _____
Insured's Phone: _____ _____
Insurance Name: _____ Group Phone: _____

INSTITUTIONAL BILLING INFORMATION

Name of Institution: _____
Name of Contact Person: _____ Billing Phone: _____
Billing Address: _____ Billing Fax#: _____
_____ Billing Email: _____
_____

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Patient Medical and Family History Form (Page 1 of 2)

Patient's Name: _____

Date: _____

Diagnostic Information:

☐ Clinical History: please describe symptoms:

☐ Biochemical testing: please list tests performed to date, including results or attach reports, if available

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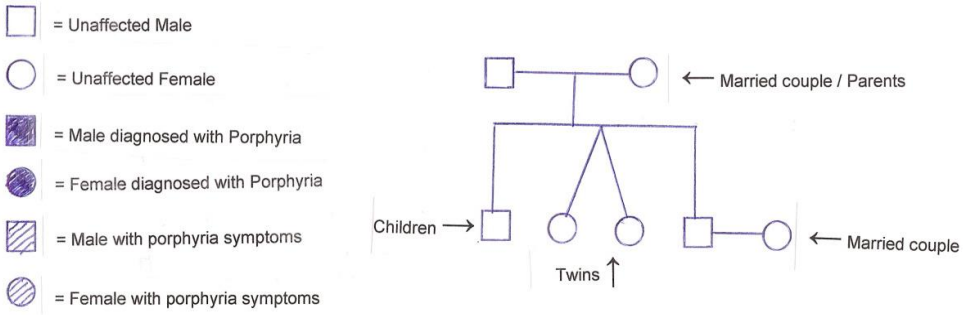
Patient Medical and Family History Form (Page 2 of 2)

☐ Family History:

Please draw a family tree, including family members by first and last names, ages and indicate individuals with porphyria. Try to follow the sample family tree below.

Alternatively, you may list affected individuals and/or individuals who have Porphyria-like symptoms, including their relationship to you.



PEDIGREE / FAMILY HISTORY

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CONSENT FOR COMMUNICATION VIA E-MAIL
(Provider-Patient)

I, _____, hereby consent to have my physician,

Print patient's name

_____, communicate with me or members of his staff, where

Print name of physician

appropriate or other physicians, nurse practitioners and pharmacists via e-mail regarding the following aspects of my medical care and treatment: [test results, prescriptions, appointments, billing, etc.]. I understand that e-mail is not a confidential method of communication. I further understand that there is a risk that e-mail communications between my physician and me or members of my physician's office staff, or between my physician and other physicians, nurse practitioners and pharmacists regarding my medical care and treatment may be intercepted by third parties or transmitted to unintended parties. I also understand that any e-mail communications between my physician and me or members of his office staff, or between my physician and other physicians, nurse practitioners or pharmacists regarding my medical care and treatment will be printed out and made a part of my medical record. I understand that in an urgent or emergent situation I should call my provider or go to the Emergency Room and not rely on e-mail.

Signature:_____ Date: _____

MR-240 (9/03)