sema4

123456-2-X

Please place green collection kit barcode here.

# BIOCHEMICAL GENETIC TESTING REQUISITION

Phone: **800-298-6470** / Fax: 646-859-6870
Branford CT Lic#: CL-0830
Stamford CT Lic#: CL-1016

**Please fill out all the highlighted fields. Failure to do so may result in delayed testing and delivery of results.**

## PATIENT INFORMATION

*Sema4 will use this information to contact the patient via automatic email, SMS, and/or phone regarding payment, testing status, and online results access. By submitting this requisition, I confirm that I have obtained the patient's authorization to be contacted by Sema4 by these means (email address must be specific to patient listed on form).*

| | |
|---|---|
| PATIENT EMAIL ADDRESS RECOMMENDED | PATIENT MOBILE/PRIMARY NUMBER REQUIRED |
| LAST NAME REQUIRED | FIRST NAME REQUIRED MI |
| DATE OF BIRTH MM / DD / YYYY | SEX ASSIGNED AT BIRTH REQUIRED ☐ MALE ☐ FEMALE ☐ INTERSEX |
| CLIENT MRN | |
| ADDRESS REQUIRED | CITY / STATE / ZIP REQUIRED |

## BILLING INFORMATION

Bill to: ☐ Insurance (Provide ICD10 in Indications for Testing) ☐ Client/Institution ☐ Self Pay/No Insurance

| | | |
|---|---|---|
| POLICYHOLDER LAST NAME REQUIRED | POLICYHOLDER FIRST NAME REQUIRED | POLICYHOLDER DOB MM / DD / YYYY |
| INSURANCE CARRIER REQUIRED | INSURANCE ID REQUIRED | GROUP NO. REQUIRED |

BILLING ADDRESS REQUIRED

SECONDARY INSURANCE ☐ YES ☐ NO

| | |
|---|---|
| SECONDARY INSURANCE NAME | GROUP NO. |

Pre-Authorization #: _____ *Please include a copy of all insurance paperwork.*

**ASSIGNMENT AND RELEASE:** I hereby authorize my insurance benefits be paid directly to the provider and I understand that I am financially responsible for uncovered services. I also authorize the release of any information required to process the claim. Billing inquiries, please call 800-298-6470.

SIGNATURE DATE MM / DD / YYYY

## ORDERING PROVIDER INFORMATION

| | |
|---|---|
| NAME REQUIRED | GENETIC COUNSELOR |
| ADDRESS REQUIRED | CLINIC / INSTITUTION REQUIRED |
| | TELEPHONE |
| | FAX |

**PROVIDER SIGNATURE OF CONSENT (REQUIRED):** I certify that this patient (and/or their legal guardian, as necessary) has been informed of the benefits, risks, and limitations of the laboratory test(s) requested. I have answered this person's questions. I have obtained a signed informed consent from this patient or their legal guardian for this testing in accordance with applicable laws and regulations, including N.Y. Civil Rights Law Section 79-L, and will retain this consent in the patient's medical record.

SIGNATURE DATE MM / DD / YYYY

## CLINICAL INDICATION (PLEASE FILL OUT ADDITIONAL INDICATIONS ON BACK)

SPECIMEN TYPE:

For Aminolevulinic Acid, Porphobilinogen, and Tamoxifen Metabolites, specimen should be protected from light

☐ Blood ☐ Plasma ☐ Serum ☐ DBS ☐ Urine ☐ CSF ☐ Other _____

COLLECTION DATE MM / DD / YYYY COLLECTION TIME: ____: ____ AM/PM

**CLINICAL INFORMATION:** Please provide relevant clinical history information/records

**PLEASE PROVIDE ICD10(s) HERE**

## LABORATORY TESTING INFORMATION

History of bone marrow transplant? ☐ YES ☐ NO History of recent blood transfusion within the last 4 weeks? ☐ YES ☐ NO **Note: If Yes, please contact Sema4 to confirm if an alternate specimen is needed for testing.**

## LABORATORY TEST(S) ORDERED

### Analyte Testing

| | TEST CODE | TEST NAME | SAMPLE TYPE |
|---|---|---|---|
| ☐ | AACSF | Amino Acids Full Panel, CSF | CSF |
| ☐ | AAPL | Amino Acids Full Panel, Plasma | Plasma |
| ☐ | AAUR | Amino Acids Full Panel, Urine | Urine |
| ☐ | SAA | Amino Acids Selective Panel (PKU/MSUD) | Plasma |
| ☐ | PKU | Phenylalanine/Tyrosine | DBS |
| ☐ | ACD | Acylcarnitine Profile, DBS | DBS |
| ☐ | ACP | Acylcarnitine Profile, Plasma | Plasma |
| ☐ | CARN | Carnitine Concentration, Plasma/Serum | Plasma/Serum |
| ☐ | CARU | Carnitine Concentration, Urine | Urine |
| ☐ | UOA | Organic Acids | Urine |
| ☐ | MMAPL | Methylmalonic Acid, Plasma | Plasma |
| ☐ | MMAUR | Methylmalonic Acid, Urine | Urine |
| ☐ | ORO | Orotic Acid | Urine |
| ☐ | SUCC | Succinylacetone | Urine |

### Biomarkers for LSD

| | TEST CODE | TEST NAME | SAMPLE TYPE |
|---|---|---|---|
| ☐ | CHITO | Chitotriosidase activity (Gaucher Disease) | Plasma |
| ☐ | LGL1 | Lyso-GL1 (Gaucher Disease) | Plasma |
| ☐ | LGB3 | Lyso-Gb3 (Fabry Disease) | Plasma |
| ☐ | LSPM | Lyso-SPM (Niemann-Pick Disease) | Plasma |
| ☐ | PSYC | Psychosine (Krabbe Disease) | Plasma |
| ☐ | GAG | Quantitative GAG (DS. HS, CS) | Urine |
| ☐ | KSU | Quantitative GAG (KS) | Urine |

### Congenital Disorders of Glycosylation (CDG)

| | TEST CODE | TEST NAME | SAMPLE TYPE |
|---|---|---|---|
| ☐ | CDT | Carbohydrate Deficient Transferrin | Plasma |
| ☐ | NGLY | N-Glycan Profiling | Plasma |
| ☐ | OGLY | O-Glycan Analysis | Plasma |

### Porphyria Analytes

| | TEST CODE | TEST NAME | SAMPLE TYPE |
|---|---|---|---|
| ☐ | ALAU | Aminolevulinic Acid (ALA), Urine | Urine |
| ☐ | ALAP | Aminolevulinic Acid (ALA), Plasma | Plasma |
| ☐ | PBGU | Porphobilinogen (PBG), Urine | Urine |
| ☐ | PBGP | Porphobilinogen (PBG), Plasma | Plasma |
| ☐ | UPP | Aminolevulinic Acid/Porphobilinogen | Urine |
| ☐ | PPP | Aminolevulinic Acid/Porphobilinogen | Plasma |

### LSD Enzyme Testing

| | TEST CODE | TEST NAME | SAMPLE TYPE |
|---|---|---|---|
| ☐ | ASM | Acid Sphingomyelinase (Niemann-Pick A/B) | Blood |
| ☐ | GDSCRN | Acid-β-Glucosidase (Gaucher Disease) | Blood |
| ☐ | LAL | Lysosomal Acid Lipase (Wolman/Cholesteryl Ester Storage Disease) | Blood |
| ☐ | FABRY | α-Galactosidase A (Fabry Disease) | Blood |
| ☐ | GAA | Acid α-Glucosidase (Pompe disease) | Blood |
| ☐ | IDUA | α-L-Iduronidase (MPS-I) | Blood |
| ☐ | GALC | b-Galactocerebrosidase (Krabbe Disease) | Blood |
| ☐ | TAYSAC | Hexosaminidase A (Tay-Sachs Disease) | Blood |
| ☐ | SANDHO | Hexosaminidase B (Sandhoff Disease) | Blood |

### Pharmacogenetic Testing

| | TEST CODE | TEST NAME | SAMPLE TYPE |
|---|---|---|---|
| ☐ | TAM | Tamoxifen Metabolites Profiling **Patient must be on standard dose (20 mg/day) of Tam for at least 3 months prior to testing or until the steady state is ready.* | Plasma |

### Porphyria Molecular Testing

| | TEST CODE | PANEL NAME | GENE |
|---|---|---|---|
| ☐ | APP | Porphyria Acute Panel | *HBMS, CPOX, PPOX* |
| ☐ | CPP | Porphyria Cutaneous Panel | *CEP, EPP, PCT* |
| ☐ | POR | Acute Intermittent Porphyria | *HBMS* |
| ☐ | POR | Hereditary Coproporphyria | *CPOX* |

### Porphyria Molecular Testing (cont)

| | TEST CODE | TEST/PANEL NAME | GENE |
|---|---|---|---|
| ☐ | POR | Variegate Porphyria | *PPOX* |
| ☐ | POR | Congenital Erythropoietic Porphyria | *UROS, ALAS** |
| ☐ | POR | Familial Porphyria Cutanea Tarda | *UROD* |
| ☐ | POR | Erythropoietic Protoporphyria Panel | *FECH, ALAS** |
| ☐ | POR | Porphyria Associated Kidney Disease PEPT2 Genotyping | *SLC15A2^* |

### LSD Molecular Testing

| | TEST CODE | TEST NAME | GENE |
|---|---|---|---|
| ☐ | DSGSV2 | Niemann Pick A/B | *SMPD1* |
| ☐ | DSGSV2 | Gaucher Disease | *GBA* |
| ☐ | DSGSV2 | Wolman/Cholesteryl Ester Storage Disease | *LIPA* |
| ☐ | DSGSV2 | Fabry Disease | *GLA* |
| ☐ | DSGSV2 | Pompe disease | *GAA* |
| ☐ | DSGSV2 | Mucopolysacharridosis type 1 | *IDUA* |
| ☐ | DSGSV2 | Krabbe Disease | *GALC* |
| ☐ | DSGSV2 | Tay-Sachs Disease | *HEXA* |
| ☐ | DSGSV2 | Sandhoff Disease | *HEXB* |
| ☐ | CHITO | Chitotriosidase | *CHIT1* |
| ☐ | CUSTPGX | CYP2D6 Genotyping | *CYP2D6* |

### Molecular Diagnostic Testing

Gene(s): _____

| | TEST CODE | TEST NAME |
|---|---|---|
| ☐ | DSGSV2 | Custom Diagnostic Gene Sequencing |
| ☐ | TARG | Targeted Mutation Analysis (assessment for a known familial variant) |

Variant(s): _____

Was this relative's variant(s) identified at Sema4?

☐ No - Please attach a copy of the relative's test report (required)

☐ Yes - Sema4 ID: _____
Name: _____
Date of Birth: _____
Patient's relationship to relative: _____

**COMPLETE AND SUBMIT ALL PAGES TO LABORATORY**

| PATIENT NAME | DATE OF BIRTH | PHYSICIAN NAME | PRACTICE NAME |
|---|---|---|---|
| | | | |

Please submit separate signed general consent form for each sample submitted (including parents)

## Informed Consent for Genetic Testing

**If you do not have legal authority and capacity to sign this consent under law, a legal representative who is at least eighteen (18) years of age and has the legal authority and capacity to do so must sign this consent and authorization on your behalf.**

I hereby request the genetic testing ordered by my health care provider, which may include molecular, cytogenetic, and/or biochemical analyses of my sample(s). I have received information (please see www.sema4.com/testcatalog for test-specific information sheet) from my physician or from a genetic counselor that described, in words that I understand, the nature of the genetic testing that I am about to undergo.

I understand that specimen(s), such as a peripheral blood, saliva, cheek swab, dried blood spot, skin biopsy, amniotic fluid, chorionic villus, and/or urine sample, will be taken from me. I understand that these samples will be used for determining if I have a genetic disease, am a carrier of a genetic disease, or am more likely to develop a genetic disease or condition.

The nature of the genetic test(s) ordered in connection with this consent has been explained to me, and the accuracy of the test and its risks and limitations have been detailed. I understand that infrequent errors may occur, even though the likelihood of an incorrect diagnosis or a misinterpretation of the result is extremely small. I understand that a negative result reduces, but does not eliminate, the possibility that I carry a variant(s) in the gene(s) analyzed or in other genes that are not included in the test. I understand that a positive result is an indication that I may be predisposed to or have a specific disease or condition and I may consider further independent testing, consult my physician or pursue genetic counseling. Knowledge of genetic information will improve over time and new information may become available in the future that could impact the interpretation of my results.

I understand that test results may reveal incidental, unsought information, such as discovering an undiagnosed disorder. I understand that this testing may yield results that are of unknown clinical significance and that parental or other relative's specimens may also be tested to determine whether a specific finding was inherited. This testing may reveal cases of adoption or demonstrate that a person is not the biological father or mother of the patient. An error in the diagnosis may occur if the true biological relationships of the family members involved are not as I have described.

I understand that the results of this testing will become part of my medical record and may only be disclosed to individuals who have legal access to this record or to individuals I designate to receive this information. My test results will be explained to me by a genetic counselor or by my healthcare provider, who will have the opportunity to discuss my results with a geneticist.

There are federal and state laws that address genetic discrimination. The US Genetic Information Nondiscrimination Act may prohibit discrimination based on genetic information by employers and health insurers. This law, however, does not protect people in the military nor protect against discrimination by other types of insurance, such as life, disability, or long-term care insurance.

### Sample management

Sema4 may deidentify and retain your left-over sample to use for operational, quality control, validation and improvement purposes. Other than retention for these uses, your sample will be destroyed at the end of the testing process or within 60 days of sample collection, whichever is longer.

### De-identified research

Sema4 may de-identify and use all data and information generated and received in connection with this test to support medical and academic research relating to health, disease prevention, drug development, and other scientific purposes. I will receive no compensation in connection with such research. Data and information are "de-identified" by removing any information that could be used to identify a specific person, such as a name, email address, or date of birth. Sema4 may also give the de-identified data and information to its research partners and may submit it to research databases for use in scientific and medical research. Examples of such research include projects to understand the risk factors and outcomes for various conditions and can be found at www.sema4.com/research.

If I do not want to have my de-identified data and information used in research as set forth above, I may withdraw this consent by emailing privacy@sema4.com, and I understand that the change will apply to all data generated from tests that I have undergone with Sema4. I further understand that this withdrawal will not apply to any information that has already been de-identified and cannot be identified by Sema4.

### Permission to contact

I understand that Sema4 may wish to contact me in the future, including for the following reasons: research purposes, the provision of general information about research findings, and/or the provision of information about the results of tests on my sample(s). If I wish to opt-out out of future contact for research purposes, I will notify Sema4 by emailing privacy@sema4.com.

My healthcare provider has discussed my test order(s) with me, and I hereby consent to have my specimen tested. I have been encouraged to ask questions and agree that any questions I have asked have been answered to my satisfaction. If my legal representative is signing this consent and authorization, my legal representative is satisfied that they have received enough information to sign on my behalf.

Please complete all required (*) fields and optional applicable fields below:

| Patient Name* | Patient's DOB* | Date* |
|---|---|---|
| Signature of Patient or Legal Representative* | Email Address* | Phone Number* |
| Legal Representative Name (if applicable) | | |