sema4

123456-2-X

Please place green collection kit barcode here.

CARRIER SCREENING WITH PERSONALIZED RESIDUAL RISK REQUISITION

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

Please be sure to fill out all highlighted fields. Failure to fill them in 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
DATE OF BIRTH MM / DD / YYYY	SEX ASSIGNED AT BIRTH REQUIRED ☐ MALE ☐ FEMALE ☐ INTERSEX / PATIENT IS A SPERM/EGG DONOR ☐ YES ☐ NO
PARTNER / SPOUSE LAST NAME	PARTNER / SPOUSE FIRST NAME
PATIENT/CLIENT MRN	PARTNER / SPOUSE DATE OF BIRTH MM / DD / YYYY
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

OTHER HEALTH COVERAGE (IDENTIFY)

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

INDICATIONS FOR TESTING

ICD10 Dx CODE(S) (Required)

Carrier Screening Indications

☐ **Z84.81** Family history of carrier of genetic disease
☐ **Z31.430** Encounter of female for testing for genetic disease carrier status for procreative management
☐ **Z31.440** Encounter of male for testing for genetic disease carrier for procreative management

☐ **Other** _____

FAMILY HISTORY OF: _____

PARTNER CARRIER OF: _____

COLLECTION DATE ___ / ___ / ___ (MM / DD / YYYY)

SPECIMEN TYPE: ☐ Peripheral Blood ☐ Saliva
☐ Other: _____

OF BLOOD TUBES SENT: YELLOW____ PURPLE____

LABORATORY TESTING INFORMATION

Patient Ancestry: _____ Preferred Language: _____

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.

Is the patient or their partner pregnant? ☐ YES ☐ NO
Is the patient currently using birth control medication? ☐ YES ☐ NO
Previous Carrier Screening? ☐ YES ☐ NO
If yes, what gene/variant: _____

LABORATORY TEST(S) ORDERED

Carrier Screening

(see reverse side for genes in each panel, all screening options include personalized residual risk (PRR))

☐ Standard Pan-ethnic Carrier Screen *(S4) (4 genes)* (**) (includes carrier screening for cystic fibrosis, fragile X syndrome, spinal muscular atrophy, and Smith-Lemli-Opitz syndrome)

Select **BOTH** S4 above and one of the options below to order additional carrier screening:

NEW
- ☐ Expanded Carrier Screen 502 *(S4+498 genes)* (**^)
- ☐ Expanded Carrier Screen 283 *(S4+279 genes)* (**^)
- ☐ High Frequency Pan-ethnic Carrier Screen *(11 genes; S4+7 genes)* (**)
- ☐ Expanded Carrier Screen 39 *(S4+35 genes)* (**^)
- ☐ Expanded Carrier Screen 152 *(S4+148 genes)* (**^)
- ☐ Comprehensive Jewish Carrier Screen *(101 genes; S4+97 genes)* (**^)
- ☐ Ashkenazi Jewish Carrier Screen *(64 genes; S4+60 genes)* (**^)
- ☐ Sephardic Jewish Carrier Screen *(54 genes; S4+50 genes)* (**^)

☐ Supplemental X-linked panel *(32 genes)* (**) (for female patients being screened after their male reproductive partner)
☐ AR disorders partner screened positive for (sample will be held if partner screening has not completed) (**)
☐ Custom Carrier Screen (**) - Please write in each gene that should be screened below:
Gene(s): _____
☐ Tay-Sachs Enzyme only (^)
☐ Sandhoff Disease Enzyme only (^)
☐ ECS Re-analysis
Previous ECS lab number (required): _____
Note: If a personalized residual risk was not provided with the initial testing this cannot be provided through re-analysis.
☐ Ultra-High Resolution Microarray (targeted follow-up to ECS result) (*)
Previous ECS lab number: _____
Gene requiring follow-up: _____

☐ Family Studies (for phasing analysis on a first-degree relative as follow-up to a patient's Sema4 carrier screening) (**)
Gene(s): _____
Variant(s): _____
Please provide the previously tested patient's information:
Sema4 ID: _____
Name: _____
Date of Birth: _____
Patient's relationship to relative: _____

☐ Targeted Mutation Analysis (PGT-M setup, or for assessment for a known familial variant) (**)
Gene(s): _____
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: _____

Infertility/Pregnancy Loss:

☐ Test for Microdeletions of Y Chromosome *(male)* (*)
☐ Cystic Fibrosis with *CFTR* Intron 9 PolyT *(male)* (*)

Infertility/Pregnancy Loss Testing Indications
☐ *N96* ☐ *Z82.7* ☐ *N46* ☐ *N97* ☐ Other: _____

☐ Thrombophilia Test *(3 variants below)* (*)
☐ *F2 - c.*97G>A*
☐ *F5 - c.1601G>A (p.Arg534Gln)*
☐ *MTHFR - c.665C>T (p.Ala222Val)*

***LEGEND:** *1 EDTA tube (lavender top) ^1 EDTA tube or 1 ACD tube (yellow top)*
***Note:** One OGD-500 saliva tube can be substituted for two EDTA tubes. Please note Tay-Sachs Enzyme and Sandhoff Enzyme cannot be performed on a saliva sample.*

COMPLETE AND SUBMIT ALL PAGES TO LABORATORY

Available Carrier Screening Genes:

AAAS
ABCA12
ABCA3
ABCA4 ✚
ABCB11
ABCC8 ◆ ● ✠ ▼
ABCD1 ◆ ■ ▼
ACAD9
ACADM ▲ ◆ ● ■ ✠ ▼
ACADS
ACADSB
ACADVL ▼
ACAT1
ACOX1
ACSF3 ▼
ADA ▼
ADAMTS2 ◆ ● ▼
AGA ▼
AGL ◆ ■ ▼
AGPS
AGXT
AIMP1
AIPL1
AIRE ◆ ■ ▼
AKR1D1
ALDH3A2
ALDH7A1
ALDOB ▼
ALG6
ALMS1
ALPL ▼
AMH ✚
AMHR2 ✚
AMN
AMT
ANO5
AP1S1
APOPT1
AQP2
ARG1
ARL6
ARSA ◆ ■ ▼
ARSB
ARSE
ASL ▼
ASNS ◆ ■ ▼
ASPA ◆ ● ✠ ▼
ASS1 ✠
ATM ◆ ■ ▼
ATP6V1B1 ◆ ■ ▼
ATP7A
ATP7B ◆ ● ■ ▼
ATP8B1
ATRX
AVPR2
BBS1 ▼
BBS10
BBS12
BBS2 ◆ ● ▼
BBS4
BCHE ✚
BCKDHA ✠ ▼
BCKDHB ◆ ● ✠ ▼
BCS1L ▼
BLM ◆ ● ✠ ▼
BMPER
BSND
BTD ▼
BTK
C8ORF37
CANT1
CAPN3
CASQ2
CASR
CBS ▼
CC2D1A
CCDC103
CCDC151
CCDC39
CD3D
CD3E
CD40LG
CD59
CDAN1
CDH23 ▼
CEP152
CEP290 ▼
CERKL ◆ ■ ▼
CFTR ◗ ▲ ◆ ● ■ ✠ ▼
CHAT
CHM
CHRNE
CHRNG
CIITA
CLCNKB
CLN3 ✠
CLN5 ▼
CLN6
CLN8
CLRN1 ◆ ● ✠ ▼
CNGA3
CNGB3 ▼
COA7
COL11A2
COL17A1
COL27A1 **NOW INCLUDING Full Gene Sequencing**
COL4A3 ◆ ● ▼
COL4A4
COL4A5
COL7A1 ▼
COQ4
COX10
COX15
COX20
COX6B1
CPS1
CPT1A
CPT2 ◆ ● ▼
CRB1
CTNS ◆ ■ ▼
CTSA
CTSC
CTSK
CYBA ◆ ■ ▼
CYBB
CYP11A1
CYP11B1
CYP11B2 ◆ ■ ▼
CYP17A1
CYP19A1
CYP1B1
CYP21A2 ◆ ● ■
CYP27A1 ◆ ■ ▼
CYP27B1
DBT
DCAF17
DCLRE1C
DDR2
DGUOK
DHCR24
DHCR7 ◗ ▲ ◆ ● ■ ✠ ▼
DHDDS ◆ ● ▼
DKC1
DLD ◆ ● ✠ ▼
DLL3
DMD ▲ ◆ ● ■ ✠ ▼
DNAH5 ◆ ● ▼
DNAI1 ◆ ● ▼
DNAI2 ◆ ● ▼
DOK7
DOLK
DPYD ✚
DUOX2
DUOXA2
DYSF ◆ ■ ▼
EDA
EIF2AK3
EIF2B5
EMD
EOGT
EPB42
ERBB3
ERCC5
ERCC6
ERCC8
ESCO2
ETFA
ETFB
ETFDH ▼
ETHE1
EVC ▼
EVC2
EXOSC3
EYS ◆ ■ ▼
F2 ✚
F5 ✚
F11 ◆ ● ▼
F7
F9
FAH ◆ ● ✠ ▼
FAM161A ◆ ● ■ ▼
FANCA ◆ ■ ▼
FANCC ◆ ● ✠ ▼
FANCG ▼
FBP1
FH ▼
FKBP10
FKRP
FKTN ◆ ● ✠ ▼
FMR1 ◗ ▲ ◆ ● ■ ✠ ▼
CGG Repeat Analysis and Full Gene Sequencing, Reflex AGG Repeat Analysis for Patients with 55-90 CGG Repeats
FOLR1
FOXRED1
FRMD4A
FUCA1
G6PC ◆ ● ✠ ▼
G6PC3
G6PD ✚
GAA ◆ ● ■ ▼
GALC ▼
GALE
GALK1 ▼
GALNS
GALNT3
GALT ◆ ● ✠ ▼
GAMT
GATM
GBA ◆ ● ✠ ▼
GBE1 ◆ ● ▼
GCDH ▼
GDF5 ✚
GFM1
GHR
GHRHR
GJB1
GJB2 ◆ ● ▼
GLA
GLB1 ▼
GLDC
GLE1 ▼
GNE ◆ ■ ▼
GNPTAB ▼
GNPTG
GNS
GORAB
GP1BA
GP9
GPR56
GRHPR
GSS
GUCY2D
GUSB
GYS2
HADH
HADHA
HADHB
HAX1
HBA1/HBA2 ▲ ◆ ● ■ ✠ ▼
HBB ▲ ◆ ● ■ ✠ ▼
HEXA ◆ ● ■ ▼
HEXB ▼
HFE ✚
HFE2
HGD
HGSNAT
HLCS ▼
HMGCL
HMGCS2
HOGA1 ◆ ● ▼
HPD
HPS1 ▼
HPS3 ◆ ● ▼
HPS4
HPS6
HSD17B3 ✚
HSD17B4
HSD3B2
HSD3B7
HYAL1
HYLS1 ▼
IDS
IDUA ✠
IGHMBP2
IGSF1
IKBKAP ◆ ● ✠ ▼
IL2RG
IL7R
INVS
ITGA2B
ITGA6
ITGB3
ITGB4
IVD ✠
IYD
JAK3
KCNJ11 ▼
L1CAM
LAMA2
LAMA3
LAMB3
LAMC2
LCA5
LDLR ◆ ● ▼
LDLRAP1
LHCGR ✚
LHX3
LIFR
LIPA ◆ ■ ▼
LMAN1
LMBRD1
LOXHD1 ◆ ● ▼
LPAR6
LPL
LRPPRC ▼
LYST
MAN2B1
MANBA
MAT1A
MCCC1
MCCC2
MCEE
MCOLN1 ◆ ● ✠ ▼
MED17 ◆ ■ ▼
MEFV ◆ ● ■ ▼
MESP2
MFSD8
MKS1 ▼
MLC1 ◆ ■ ▼
MLYCD
MMAA
MMAB
MMACHC ✠
MMADHC
MOCS1
MPI
MPL ◆ ● ▼
MPV17
MRE11
MTHFR **NOW INCLUDING Sequencing except variant below** ◆ ■ ▼
c.665C>T (p.Ala222Val) variant only ✚
MTM1
MTR
MTRR
MTTP ◆ ● ▼
MUT
MYO15A
MYO7A ▼
NAGLU
NAGS
NBEAL2
NBN
NDRG1
NDUFA11
NDUFAF2
NDUFAF5 ◆ ▼
NDUFS4
vNDUFS6 ◆ ■ ▼
NDUFS7
NDUFV1
NEB ◆ ● ✠ ▼
NEU1
NGLY1
NPC1
NPC2
NPHP1
NPHS1 ▼
NPHS2
NR0B1
NR2E3 ◆ ● ▼
NTRK1 ◆ ■ ▼
OAT ◆ ■ ▼
OCRL
OPA3 ◆ ■ ▼
OTC
OTOF
PAH ◆ ● ■ ✠ ▼
PC ▲
PCBD1
PCCA
PCCB
PCDH15 ◆ ● ✠ ▼
PDHA1
PDHB
PEPD
PET100
PEX1 ✠
PEX10
PEX12
PEX2 ◆ ● ▼
PEX26
PEX6 ◆ ■ ▼
PEX7 ✠
PFKM ◆ ● ▼
PHGDH ◆ ● ▼
PHKB
PIGN
PIP5K1C
PJVK
PKHD1 ◆ ● ✠ ▼
PLA2G6
PLAA
PLOD1
PMM2 ▲ ◆ ● ■ ✠ ▼
PNPO
POC1A
POLG
POLH
POMGNT1 ▼
POR
POU1F1
PPT1 ▼
PRCD
PRICKLE1
PROP1 ▼
PRPS1
PSAP
PTPRC
PTS
PUS1 ◆ ■ ▼
PYGL
PYGM ◆ ■ ▼
QDPR
RAB23
RAG1
RAG2 ◆ ■ ▼
RAPSN ◆ ■ ▼
RARS2 ◆ ■ ▼
RDH12
RDH5
RHAG
RLBP1 ✚
RMRP ▼
RNASEH2C
ROGDI
RPE65 ◆ ■ ▼
RPGRIP1L
RS1
RSPH9
RTEL1 ◆ ● ▼
RYR1 ✚
SACS ▼
SAMHD1
SARS2
SBDS
SCO1
SEC23B
SEPSECS ◆ ■ ▼
SERPINA1 ✚
SGCA
SGCB
SGCD
SGCG
SGSH
SLC12A3 ▼
SLC12A6 ▼
SLC17A5 ▼
SLC19A2
SLC1A4
SLC22A5 ▼
SLC25A13 ▼
SLC25A15
SLC25A20
SLC26A2 ▼
SLC26A3
SLC26A4 ▼
SLC2A2
SLC34A3
SLC35A3 ◆ ● ▼
SLC37A4
SLC39A4
SLC3A1
SLC45A2
SLC4A1
SLC4A11
SLC5A5
SLC6A8
SLC7A7
SMARCAL1
SMN1/SMN2 **NOW INCLUDING sequencing of Exons 2a-7 in SMN1** ◗ ▲ ◆ ● ■ ✠ ▼
SMPD1 ◆ ● ✠ ▼
SNAP29
SNX10
SPR
SRD5A2 ✚
ST3GAL5
STAR
SUCLA2
SUMF1 ◆ ● ▼
SURF1
SYNE4
TAT
TAZ
TBCE
TBX19
TCIRG1 ◆ ● ▼
TECPR2 ◆ ■ ▼
TFR2
TG
TGM1 ▼
TH
TK2
TMC1
TMEM216 ◆ ● ✠ ▼
TMPRSS3
TPO
TPP1 ▼
TREX1
TRHR
TRIM32
TRIM37
TRMU ◆ ■ ▼
TRPM6
TSEN54
TSFM ▼
TSHB
TSHR
TTC37
TTPA
TULP1
TYMP ◆ ■ ▼
TYR
TYRP1
UGT1A1 ✚
USH1C ▼
USH2A ◆ ■ ▼
VDR
VPS11
VPS13A ◆ ● ▼
VPS13B
VPS45
VPS53
VRK1 ◆ ● ▼
VSX2 ◆ ■ ▼
VWF ✚
WAS
WISP3
WNT10A
WRN
XPA
XPC
ZFYVE26

KEY FOR SMALLER PANELS

◗ S4 – Standard Pan-ethnic Panel
▲ HF – High Frequency Pan-ethnic Panel
◆ CJ – Comprehensive Jewish Carrier Screen
● AJ – Ashkenazi Jewish Disorders
■ SJ – Sephardi-Mizrahi Jewish Disorders
✠ 39 – ECS 39
▼ 152 – ECS 152
✚ Case by case/opt-in only
Bolded Genes – New to the ECS 502 panel
Underlined Genes – Supplemental X-linked panel

v12/09/21

COMPLETE AND SUBMIT ALL PAGES TO LABORATORY

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)		

P0122GE0122
Revised 01/31/22