



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

PATIENT INFORMATION

Form containing patient information fields: PATIENT EMAIL ADDRESS, LAST NAME, DATE OF BIRTH, SEX ASSIGNED AT BIRTH, PARTNER / SPOUSE LAST NAME, CLIENT MRN, ADDRESS, CITY / STATE / ZIP.

ORDERING PHYSICIAN INFORMATION

Form containing ordering physician information fields: NAME, ADDRESS, CLINIC / INSTITUTION, TELEPHONE, FAX.

PHYSICIAN 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.

Form containing signature and date fields: SIGNATURE, DATE MM / DD / YYYY.

BILLING INFORMATION

Form containing billing information fields: Bill to, POLICYHOLDER LAST NAME, POLICYHOLDER FIRST NAME, POLICYHOLDER DOB, INSURANCE CARRIER, INSURANCE ID, GROUP NO., BILLING ADDRESS, SECONDARY INSURANCE, SECONDARY INSURANCE NAME, GROUP NO.

INDICATORS FOR TESTING

ICD10 Dx CODE(S) (Required) Please see each testing section below and write in ICD10s as needed for each test type.

Form containing collection date and blood tubes sent fields: COLLECTION DATE MM/DD/YYYY # OF BLOOD TUBES SENT: YELLOW, PURPLE, BCT, GREEN.

SPECIMEN TYPE: (Please contact laboratory for alternate specimen types)

Form containing specimen type options: Maternal, Paternal, Fetal. Options include Peripheral Blood, Saliva, Amniotic Fluid, Chorionic Villi, etc.

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.

Form containing signature and date fields: SIGNATURE, DATE MM / DD / YYYY.

PREGNANCY HISTORY: Gestational Age: Weeks Days or EDD: (Required for fetal samples.)

Form containing pregnancy history fields: Pregnancy conceived: IVF, Egg donor/gestational carrier, Age of genetic mother.

LABORATORY TEST(S) ORDERED

Parental Carrier Screening

Form containing parental carrier screening questions: Patient ancestry, History of bone marrow transplant, History of recent blood transfusion, Note: If Yes, please contact Sema4 to confirm if an alternate specimen is needed for testing.

Form containing parental carrier screening options: Standard Pan-ethnic Carrier Screen (S4), Expanded Carrier Screen 502, Expanded Carrier Screen 283, High Frequency Pan-ethnic Carrier Screen (11 genes), Expanded Carrier Screen 39, Expanded Carrier Screen 152, Comprehensive Jewish Carrier Screen (101 genes).

Form containing parental carrier screening indications: Z31.430, Z31.440, Other.

Infertility/Pregnancy Loss:

Form containing infertility/pregnancy loss test options: Test for Microdeletions of Y Chromosome (male), Cystic Fibrosis with CFTR Intron 9 PolyT (male), Infertility/Pregnancy Loss Testing Indications.

Please contact the laboratory for all prenatal testing

Prenatal Diagnostic Testing

Form containing prenatal diagnostic testing options: FGFR3 Hotspot Panel, Limb Defects Next Gen Sequencing Panel (7 genes), Noonan Syndrome Next Gen Sequencing Panel (18 genes), Prenatal Diagnostic Selective Gene.

Biochemical testing: Tay-Sachs enzyme analysis, Sandhoff enzyme analysis. Maternal blood is required for all prenatal specimens for maternal cell contamination.

For samples received for prenatal molecular and/or biochemical testing, cytogenetic and cytogenomic testing can be ordered in conjunction as shown below. Please note that prenatal diagnostic testing must be ordered for a sample to be accepted for cytogenetic and cytogenomic testing.

Noninvasive Prenatal Testing (NIPT) (Must be at least 9 weeks gestation)

Form containing noninvasive prenatal testing options: Sema4 Noninvasive Prenatal Select, Sequenom MaternIT GENOME.

NIPT REQUIRED CLINICAL INFORMATION Specimen Required: Two 10 mL Whole Blood BCT Streck Tubes

Form containing NIPT clinical information fields: Gestation, Gestational age, Maternal height, Maternal weight, Pregnancy conceived, If IVF, were multiple embryos transferred?

NIPT MEDICAL INDICATIONS FOR TESTING Select one or more ICD10 codes

Form containing NIPT medical indications for testing fields: No known high risk for fetal chromosomal aneuploidies, High risk for fetal chromosomal aneuploidies, Advanced Maternal age, Abnormal serum biochemical screening, Ultrasound finding, Personal/Family History, Other high risk factor.

Array Comparative Genomic Hybridization (aCGH) 180K + SNP Please submit maternal blood (1 EDTA purple top) for Maternal Cell Contamination (MCC) with any microarray order.

Form containing array comparative genomic hybridization options: Chromosome Analysis, Reflex to array if normal chromosomes, Additional Cell Culture, Mosaicism study.

Form containing array comparative genomic hybridization fields: Proband SEMA4 Lab ID, Name, DOB, Aneuploidy FISH, FISH other.

Prenatal Diagnostic Testing Indications: 009.511, 009.512, N96, Z82.7, Z84.81, 028.3, 028.5, Other.

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.

Available Carrier Screening Genes:

AAAS	CASQ2	DKC1	GFM1	LDLRAP1	OTC	RLBP1 +	ST3GAL5
ABCA12	CASR	DLD ◆●※▼	GHR	LHCGR +	OTOF	RMRP ▼	STAR
ABCA3	CBS ▼	DLL3	GHRHR	LHX3	PAH ◆●■※▼	RNASEH2C	SUCLA2
ABCA4 +	GC2D1A	DMD ▲◆●■※▼	GJB1	LIFR	PC ▲	ROGDI	SUMF1 ◆●▼
ABCB11	CCDC103	DNAH5 ◆●▼	GJB2 ◆●▼	LIPA ◆■▼	PCBD1	RPE65 ◆■▼	SURF1
ABCC8 ◆●※▼	CCDC151	DNAI1 ◆●▼	GLA	LMAN1	PCCA	RPGRIP1L	SYNE4
ABCD1 ◆■▼	CCDC39	DNAI2 ◆●▼	GLB1 ▼	LMBRD1	PCCB	RS1	TAT
ACAD9	CD3D	DOK7	GLDC	LOXHD1 ◆●▼	PCDH15 ◆●※▼	RSPH9	TAZ
ACADM ▲◆●■※▼	CD3E	DOLK	GLE1 ▼	LPAR6	PDHA1	RTEL1 ◆●▼	TBCE
ACADS	CD40LG	DPYD +	GNE ◆■▼	LPL	PDHB	RYR1 +	TBX19
ACADSB	CD59	DUOX2	GNPTAB ▼	LRPPRC ▼	PEPD	SACS ▼	TCIRG1 ◆●▼
ACADVL ▼	CDAN1	DUOX2	GNPTG	LYST	PET100	SAMHD1	TECPR2 ◆■▼
ACAT1	CDH23 ▼	DYSF ◆■▼	GNS	MAN2B1	PEX1 ※	SARS2	TFR2
ACOX1	CEP152	EDA	GORAB	MANBA	PEX10	SBDS	TG
ACSF3 ▼	CEP290 ▼	EIF2AK3	GP1BA	MAT1A	PEX12	SC01	TGM1 ▼
ADA ▼	CERKL ◆■▼	EIF2B5	GP9	MCCC1	PEX2 ◆●▼	SEC23B	TH
ADAMTS2 ◆●▼	CFTR ▶▲◆●■※▼	EMD	GPR56	MCCC2	PEX26	SEPSECS ◆■▼	TK2
AGA ▼	CHAT	EOGT	GRHPR	MCEE	PEX6 ◆■▼	SERPINA1 +	TMCM1
AGL ◆■▼	CHM	EPB42	GSS	MCOLN1 ◆●※▼	PEX7 ※	SGCA	TMEM216 ◆●※▼
AGPS	CHRNE	ERBB3	GUCY2D	MED17 ◆■▼	PEX7 ※	SGCB	TPMRS3
AGXT	CHRNA	ERCC5	GUSB	MEFV ◆●■▼	PHGM ◆●▼	SGCD	TPO
AIMP1	CHIT1	ERCC6	GYS2	MES2	PHKB	SGCG	TPP1 ▼
AIPL1	CLCNKB	ERCC8	HADH	MES2	PIGN	SGSH	TREX1
AIRE ◆■▼	CLN3 ※	ESCO2	HADHA	MFSDB	PIP5K1C	SLC12A3 ▼	TRHR
AKR1D1	CLN5 ▼	ETFA	HADHB	MKS1 ▼	PJKV	SLC12A6 ▼	TRIM32
ALDH3A2	CLN6	ETFB	HAX1	MLC1 ◆■▼	PKHD1 ◆●※▼	SLC12A6 ▼	TRIM37
ALDH7A1	CLN8	ETFDH ▼	HBA1/HBA2 ▲◆●■※▼	MLYCD	PLA2G6	SLC17A5 ▼	TRMU ◆■▼
ALDOB ▼	CLRN1 ◆●▼	ETHE1	HBB ▲◆●■※▼	MMAA	PLAA	SLC19A2	TRPM6
ALG6	CNGA3	EVC ▼	HEXA ◆●■▼	MMAB	PLOD1	SLC1A4	TRPM6
ALMS1	CNGB3 ▼	EVC2	HEXB ▼	MMACHC ※	PMM2 ▲◆●■※▼	SLC22A5 ▼	TSEN54
ALPL ▼	COA7	EXOSC3	HFE +	MMADHC	PNPO	SLC25A13 ▼	TSFM ▼
AMH +	COL11A2	EYS ◆■▼	HFE2	MOC51	POC1A	SLC25A15	TSHB
AMHR2 +	COL17A1	F2 +	HGD	MPI	POLG	SLC25A20	TSHR
AMN	COL27A1 NOW	F5 +	HGSNAT	MPL ◆●▼	POLH	SLC26A2 ▼	TTC37
AMT	INCLUDING Full Gene Sequencing	F11 ◆●▼	HLCS ▼	MPV17	POMGNT1 ▼	SLC26A3	TPPA
ANOS	COL4A3 ◆●▼	F7	HMGCL	MRE11	POR	SLC26A4 ▼	TULP1
AP1S1	COL4A3 ◆●▼	F9	HMGCS2	MTHFR NOW INCLUDING Sequencing except variant below ◆■▼ c.665C>T (p.Ala222Val) variant only +	POU1F1	SLC2A2	TYMP ◆■▼
APOPT1	COL4A4	FAH ◆●※▼	HOGA1 ◆●▼	MTM1	PPT1 ▼	SLC34A3	TYR
AQP2	COL4A5	FAM161A ◆●■▼	HPD	MTR	PRCD	SLC35A3 ◆●▼	TYR1
ARG1	COL7A1 ▼	FANCA ◆■▼	HPS1 ▼	MTRR	PRICKLE1	SLC37A4	UGT1A1 +
ARL6	COQ4	FANCC ◆●※▼	HPS3 ◆●▼	MUT	PROP1 ▼	SLC39A4	USH1C ▼
ARSA ◆■▼	COX10	FANCG ▼	HPS4	MYO15A	PRPS1	SLC3A1	USH2A ◆■▼
ARSB	COX15	FBP1	HPS6	MYO7A ▼	PSAP	SLC45A2	VDR
ARSE	COX20	FH ▼	HSD17B3 +	NAGLU	PTPRC	SLC4A1	VPS11
ASL ▼	COX6B1	FKBP10	HSD17B4	NAGS	PTS	SLC4A11	VPS13A ◆●▼
ASNS ◆■※▼	CPS1	FKRP	HSD17B4	NBEAL2	PUS1 ◆■▼	SLC5A5	VPS13B
ASPA ◆●※▼	CPT1A	FKTN ◆●※▼	HSD3B2	NBN	PYGL	SLC6A8	VPS45
ASS1 ※	CPT2 ◆●▼	FMR1 ▶▲◆●■※▼	HSD3B7	NDR1	PYGM ◆■▼	SLC7A7	VPS53
ATM ◆■▼	CRB1	CGG Repeat Analysis and Full Gene Sequencing, Reflex AGG Repeat Analysis for Patients with 55-90 CGG Repeats	HYAL1	NDUFA11	QDPR	SMARCA1	VRK1 ◆●▼
ATP6V1B1 ◆■▼	CTNS ◆■▼		HYLS1 ▼	NDUF2	RAB23	SMN1/SMN2 NOW INCLUDING sequencing of Exons 2a-7 in SMN1 ▶▲◆●■※▼	VX2 ◆■▼
ATP7A	CTSA		IDS	NDUF5	RAG1		VWF +
ATP7B ◆●■▼	CTSC		IDUA ※	NDUFS4	RAG2 ◆■▼		WAS
ATP8B1	CTSK		IGHM1P2	NDUFS6 ◆■▼	RAPSN ◆■▼		WISP3
ATRX	CYBA ◆■▼	FOLR1	IGSF1	NDUFS7	RARS2 ◆■▼	SMPD1 ◆●※▼	WNT10A
AVPR2	CYBB	FOXRED1	IKBKAP ◆●※▼	NDUFV1	RDH12	SNAP29	WRN
BBS1 ▼	CYP11A1	FRMD4A	IL2RG	NEB ◆●※▼	RDH5	SNX19	XPA
BBS10	CYP11B1	FUCA1	IL7R	NEU1	RHAG	SPR	XPC
BBS12	CYP11B2 ◆■▼	G6PC ◆●※▼	INVS	NPC1		SRD5A2 +	ZFYVE26
BBS2 ◆●▼	CYP17A1	G6PC3	ITGA2B	NPC2			
BBS4	CYP19A1	G6PD +	ITGA6	NPHP1			
BCHE +	CYP1B1	GAA ◆●■▼	ITGB3	NPHS1 ▼			
BCKDHA ※▼	CYP1B1	GALC ▼	ITGB4	NPHS2			
BCKDHB ◆●※▼	CYP21A2 ◆●■	GALE	IVD ※	NROB1			
BCS1L ▼	CYP27A1 ◆■▼	GALK1 ▼	IYD	NR2E3 ◆●▼			
BLM ◆●※▼	CYP27B1	GALN3	JAK3	NTRK1 ◆■▼			
BMPER	DBT	GALNT3	KCNJ11 ▼	OAT ◆■▼			
BSND	DCAF17	GALT ◆●※▼	L1CAM	OCRL			
BTD ▼	DCLRE1C	GAMT	LAMA2	OPA3 ◆■▼			
BTX	DDR2	GATM	LAMA3				
C8ORF37	DGUOK	GBA ◆●※▼	LAMB3				
CANT1	DHCR24	GBE1 ◆●▼	LAMC2				
CAPN3	DHCR7 ▶▲◆●■※▼	GCDH ▼	LCA5				
	DHDDS ◆●▼	GDF5 +	LDLR ◆●▼				

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

Informed Consent for Genetic Testing

If you are under eighteen (18) years of age, 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 genetic testing, which may include molecular, cytogenetic, and/or biochemical analyses, for myself. I have received verbal and written information (please see 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 the 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. 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 (GINA) may prohibit discrimination based on genetic information by employers and health insurances. 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 storage for future clinical purposes

I authorize and consent to Sema4 storing a portion of my sample indefinitely for the sole purpose of performing future clinical testing, provided it meets Sema4's storage criteria. My sample will not be used for any other purpose without my consent, and no test will be performed on my sample other than the one(s) authorized by me and my healthcare provider. The consent preferences that I set now apply to all tests that I have undergone with Sema4 to date. I understand that I can withdraw this consent by emailing privacy@sema4.com, and my sample will be destroyed at the end of the testing process or not more than 60 days after collection, whichever occurs later.

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). I understand that I may notify Sema4 to opt out of such future contact 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)		