GENERAL TEST REQUISITION

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

123456-2-X

Please place green collection kit barcode here.

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 | BIOLOGICAL SEX ☐ M ☐ F REQUIRED | PATIENT IS A SPERM/EGG DONOR ☐ YES ☐ NO

PARTNER / SPOUSE LAST NAME | PARTNER / SPOUSE FIRST NAME

CLIENT MRN | PARTNER / SPOUSE DATE OF BIRTH MM / DD / YYYY

ADDRESS REQUIRED | CITY / STATE / ZIP REQUIRED

BILLING INFORMATION

Bill to: ☐ Client/Institution ☐ Insurance ☐ 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

REFERRING 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 / YYY

CLINICAL INDICATIONS

SPECIMEN TYPE (Please contact laboratory for alternate specimen types)

☐ AMNIOTIC FLUID ☐ BLOOD ☐ CVS
☐ DBS ☐ PLASMA ☐ URINE
☐ OTHER _____
☐ CULTURED CELLS (Origin) _____

DATE / TIME SPECIMEN DRAWN AM PM MM / DD / YYYY

DATE SPECIMEN SENT MM / DD / YYYY

☐ LMP MM / DD / YYYY | GESTATIONAL AGE ON SONO

INDICATIONS FOR TEST:

Is the patient pregnant? ☐ Yes ☐ No Currently using birth control medication? ☐ Yes ☐ No

ICD10 Dx CODE(S)

LABORATORY TEST(S) ORDERED

Test Selection (Required)

Cytogenetics and Cytogenomics

Chromosome Analysis

☐ Chromosome Analysis (includes AFP with amniotic fluid)
Includes reflex to array if no growth for POC specimens
☐ Reflex to array if normal chromosomes
☐ Additional Cell Culture: ☐ Hold ☐ Grow
☐ Mosaicism study

Chromosomal Microarray: Array Comparative Genomic Hybridization (aCGH) 180K + SNP

For prenatal specimens, please submit maternal blood for Maternal Cell Contamination (MCC)

☐ Prenatal chromosomal microarray (lower resolution) ☐ High Resolution Chromosomal Microarray

Include blood (1 EDTA purple top, 1 Sodium heparin green top) from the parents of the proband/pregnancy if available. ☐ included _____ mother _____ father

☐ **POC Microarray PLUS:** Includes high resolution microarray analysis, triploidy detection, UPD analysis, molar pregnancy analysis and MCC studies with submission of maternal blood or saliva sample. Include blood (1 EDTA purple top, 1 Sodium heparin green top) from t he parents of the pregnancy if available.
☐ Included _____ mother _____ father

Fluorescent in situ Hybridization (FISH)

☐ Aneuploidy FISH (chromosomes 13,18,21,X,Y)
☐ Microdeletion FISH Panel (individually or as a panel)
- ☐ Angelmann Syndrome (15q11.2)
- ☐ CHARGE (8q12.1 - q12.2)
- ☐ Cri-du-chat Syndrome (5p15.2)
- ☐ DiGeorge/Velo-Cardio-Facial Syndrome (22q11.2)
- ☐ Langer-Giedion (8q23.3 - 8q24.11)
- ☐ Miller-Dieker Syndrome (17p13.3)
- ☐ Prader-Willi Syndrome (15q11.2)
- ☐ Rubinstein–Taybi (16p13.3)
- ☐ Smith-Magenis Syndrome (17p11.2)
- ☐ Sotos Syndrome (5q35)
- ☐ Williams Syndrome (7q11.23)
- ☐ Wolf-Hirschhorn Syndrome (4p16.3)
- ☐ 1p36 deletion syndrome (1p36.3)

☐ FISH other: _____
☐ FISH for Kallman Syndrome
☐ FISH for *STS* Deficiency
☐ FISH for *SRY* deletion

Pharmacogenetic Tests

☐ Comprehensive PGx Panel
- ☐ Cardiovascular PGx Panel
- ☐ Psychiatry PGx Panel
- ☐ Pain PGx Panel
- ☐ Oncology PGx Panel
- ☐ Pediatric PGx Panel
- ☐ Epilepsy PGx Panel

☐ Custom PGx Testing: gene(s):_____
☐ Tamoxifen Metabolites, Plasma

Molecular

For all testing related to Carrier Screening and Natalis, please refer to our test-specific requisition forms.

Diagnostic Testing

(please refer to our website for additional diagnostic testing offerings)

☐ Single gene: _____
- ☐ Targeted Testing: variant _____ (please include previous report if available)
- ☐ Phase analysis

Infertility/Pregnancy Loss

☐ Test for Microdeletions of Y Chromosome (male)
☐ Cystic Fibrosis with *CFTR* Intron 9 PolyT (male)
☐ *MTHFR* - c.665C>T (p.Ala222Val) add-on
☐ Thrombophilia Test (2 variants below)
- ☐ F2 - c.*97G>A
- ☐ F5 - c.1601G>A (p.Arg534Gln)

Please refer to our test-specific requisition forms for more defined or smaller panels

Hearing and Vision Loss Panels

☐ Comprehensive Hearing and Vision Loss (308 genes)
- ☐ Comprehensive Hearing Loss (92 genes)
- ☐ Comprehensive Vision Loss (250 genes)

Neurodevelopmental Panels

☐ Comprehensive Epilepsy and Autism Panel (401 genes)
- ☐ Comprehensive Epilepsy Panel (226 genes)
- ☐ Comprehensive Autism Panel (228 genes)
  - ☐ STAT Autism Panel (30 genes)

☐ Microcephaly (78 genes)

Skeletal Panels

☐ Craniosynostosis (8 genes)
☐ Limb defects (7 genes + ZRS regulatory region)
☐ *FGFR3* Hotspot Panel ☐ Reflex to sequencing if negative
- ☐ F*GFR3* Full Gene Sequencing

Cardiovascular Panels

☐ Comprehensive Cardiovascular Panel (241 genes)
- ☐ Comprehensive Cardiomyopathy Panel (190 genes)
- ☐ Noonan Spectrum Disorders Panel (19 genes)

☐ Comprehensive Immunodeficiency Panel (250 genes)

Immunodeficiency Panels

☐ Comprehensive Immunodeficiency Panel (250 genes)

Genotyping and Targeted Analysis

☐ Chitotriosidase
☐ Chronic Kidney Disease *APOL1* genotyping (African American)

Craniosynostoses

Please inquire regarding which exons are tested & which genes are analyzed on a reflex basis

☐ Antley-Bixler syndrome *(FGFR2)*
☐ Apert syndrome *(FGFR2)*
☐ Beare-Stevenson Syndrome *(FGFR2)*
☐ Carpenter Syndrome *(RAB23)*
☐ Craniofrontonasal Syndrome (CFNS) *(EFNB1)*
☐ Craniosynostosis, Boston Type (CRS2) *(MSX2)*
☐ Craniosynostosis with Radial Defects *(TWIST1, REC QL4)*
☐ Crouzon Syndrome *(FGFR2, FGFR3)*
☐ Crouzon and Acanthosis Syndrome (Crouzoodermoskeletal Syndrome) *(FGFR3)*
☐ Jackson-Weiss Syndrome *(FGFR2, FGFR3)*
☐ Non-Syndromic Coronal Syndrome *(FGFR2, FGFR3)*
☐ Muenke Syndrome *(FGFR3)*
☐ Pfeiffer Syndrome *(FGFR1, FGFR2, FGFR3)*
☐ POR Deficiency *(POR)*
☐ Saethre-Chotzen Syndrome (SC2) *(TWIST1, FGFR2, FGFR3)*

Biochemical

Please circle the specimen type for each biochemical test selected below

Analyte Tests

☐ Amino Acids Full Panel: P, U, C
☐ Phenylalanine/Tyrosine, DBS
☐ Amino Acids Selective Panel (PKU/MSUD): P
☐ Acylcarnitine Profile: P, D
☐ Carnitine: P, U
☐ Organic Acids Profile: U
☐ Orotic Acid: U
☐ Methylmalonic Acid: P, U
☐ Succinylacetone: U
☐ Quantitative Glycosaminoglycans: U (chondroitin,dermatan, and heparan sulfates)
☐ Aminolevulinic Acid and Porphobilinogen: U, P
☐ Quantitative Keratan Sulfate: U
☐ Lyso-GL1, P (Gaucher Disease)
☐ Psychosine: P (Krabbe Disease)
☐ Carbohydrate Deficient Transferrin: P
☐ N-Glycan Profiling: P
☐ O-Glycan Profiling: P

Enzyme Tests

☐ Hexosaminidase A (Tay-Sachs Disease): W, S
☐ Hexosaminidase B (Sandhoff Disease): W, S
☐ Acid-β-Glucosidase (Gaucher Disease): W
☐ Chitotriosidase (Gaucher Biomarker): P
☐ α-Galactosidase A (Fabry Disease): W, P
☐ Lysosomal Acid Lipase: W (Wolman Disease/Cholesteryl Ester Storage Disease)
☐ α-L-Iduronidase (MPS-I): W
☐ α-Glucosidase (Pompe Disease): W
☐ Acid Sphingomyelinase (Niemann-Pick A/B): W
☐ β-Galactocerebrosidase (Krabbe Disease): W

***Legend:* P = Plasma, U = Urine, S = Serum, C = Cerebrospinal Fluid (CSF), D = Dried Blood Spot (DBS), W = White Blood Cells (WBC)**

Molecular Genes by Panel

PANELS	GENES
Comprehensive Hearing and Vision Loss Panel:	*ABCA4, ABHD12, ACTG1, ADAM9, ADGRV1, AGK, AHI1, AIFM1, AIPL1, ALMS1, AP3B1, ARL13B, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, BLOC1S6, BMP4, C2ORF71, C5ORF42, C8ORF37, CA4, CABP4, CACNA1D, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CCDC50, CDH23, CDH3, CDHR1, CEACAM16, CEP164, CEP290, CEP41, CERKL, CHM, CIB2, CLDN14, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COCH, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, CRB1, CRX, CRYAA, CRYAB, CRYBB1, CRYBB3, CTSD, CYP1B1, CYP4V2, DHDDS, DIABLO, DIAPH1, DNAJC5, EDN3, EDNRB, EFEMP1, ELOVL4, ERCC6, ESPN, ESRRB, EYA1, EYA4, EYS, FAM161A, FLVCR1, FOXC1, FOXE3, FRAS1, FREM1, FREM2, FRMD7, FSCN2, FYCO1, FZD4, GCNT2, GIPC3, GJA8, GJB2, GJB6, GNAT1, GNAT2, GNPTG, GPR143, GPR179, GPSM2, GRHL2, GRIP1, GRK1, GRM6, GRXCR1, GSDME, GUCA1A, GUCA1B, GUCY2D, HARS, HCCS, HESX1, HGF, HPS1, HPS3, HPS4, HPS5, HPS6, HSF4, IFT140, ILDR1, IMPDH1, IMPG2, INPP5E, IQCB1, JAG1, KARS, KCNJ13, KCNQ1, KCNQ4, KCNV2, KIF11, KIF7, KLHL7, LCA5, LHFPL5, LOXHD1, LRAT, LRIT3, LRP5, LRTOMT, LYST, LZTFL1, MAK, MARVELD2, MERTK, MFRP, MFSD8, MITF, MKKS, MKS1, MSRB3, MT-RNR1, MTTP, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, MYOC, NDP, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1SW, OTOA, OTOF, OTOG, OTOGL, OTX2, P2RX2, PANK2, PAX3, PAX6, PCDH15, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PEX1, PEX10, PEX14, PEX16, PEX19, PEX2, PEX5, PEX6, PEX7, PGK1, PHYH, PITPNM3, PITX2, PITX3, PJVK, PLA2G5, POU3F4, POU4F3, PPT1, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, PRPS1, PTPRQ, PXDN, RAB28, RAX2, RBP3, RBP4, RD3, RDH12, RDH5, RDX, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, SAG, SDCCAG8, SEMA4A, SERPINB6, SIX1, SIX5, SLC24A1, SLC26A4, SLC45A2, SMOC1, SMPX, SNRNP200, SOX10, SOX2, SPATA7, STRA6, STRC+, TBC1D24, TCTN1, TCTN2, TCTN3, TDRD7, TECTA, TGFBI, TIMM8A, TIMP3, TMC1, TMEM126A, TMEM216, TMEM231, TMEM237, TMEM67, TMIE, TMPRSS3, TOPORS, TPP1, TPRN, TREX1, TRIM32, TRIOBP, TRPM1, TSPAN12, TSPEAR, TTC21B, TTC8, TULP1, TYR, TYRP1, UBIAD1, USH1C, USH1G, USH2A, VCAN, VSX2, WDR19, WFS1, WHRN, ZNF423*
Comprehensive Epilepsy and Autism Panel:	*ABAT, ABCD1, ACSL4, ACY1, ADGRG1, ADGRV1, ADNP, ADSL, AFF2, AGO1, AHI1, AIFM1, ALDH5A1, ALDH7A1, ALG13, AMT, ANK3, ANKRD11, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, ARFGEF2, ARHGEF9, ARID1A, ARID1B, ARX, ASPM, ATP13A2, ATP1A2, ATP2A2, ATP6AP2, ATP6V0A2, ATP7A, ATP8A2, ATR, ATRX, AUTS2, BCKDK, BCL11A, BCOR, BRAF, BRWD3, C12ORF57, CA8, CACNA1A, CACNA1C, CACNA1H, CACNA2D2, CACNB4, CASK, CASR, CBL, CC2D1A, CCDC22, CCDC88C, CCM2, CDKL5, CDKN1C, CHD2, CHD7, CHD8, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CNTNAP2, COL18A1, COL4A1, CPA6, CREBBP, CSTB, CTCF, CTNNB1, CTSD, CUL3, CUL4B, CYP27A1, D2HGDH, DCX, DDHD2, DDX3X, DEAF1, DEPDC5, DHCR7, DIS3L2, DKC1, DLG3, DMD, DNAJC5, DNM1, DNMT3A, DOCK7, DPYD, DYNC1H1, DYRK1A, EBP, EEF1A2, EFHC1, EHMT1, EIF2S3, ELP4, EMX2, EPM2A, EZH2, FGD1, FGFR3, FKRP, FLNA, FLVCR2, FMR1, FOLR1, FOXG1, FOXP1, FOXP2, FTSJ1, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GAMT, GATAD2B, GATM, GCSH, GDI1, GK, GLDC, GLI2, GLI3, GNAO1, GNB1, GOSR2, GPC3, GPHN, GRIA3, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIP1, HCCS, HCN1, HCN4, HDAC8, HGSNAT, HIP1, HNRNPU, HPRT1, HRAS, HSD17B10, HTRA1, HUWE1, IDS, IL1RAPL1, IQSEC2, ITPA, KANSL1, KAT6A, KAT6B, KCNA1, KCNA2, KCNB1, KCNJ1, KCNJ10, KCNJ11, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD13, KCTD7, KDM5C, KDM6A, KIF1A, KIF1BP, KIRREL3, KMT2A, KMT2D, KRAS, KRIT1, L1CAM, L2HGDH, LAMC3, LAMP2, LARGE1, LAS1L, LBR, LGI1, LIAS, LINS1, LRP2, MAGEL2, MAGI2, MAN1B1, MAOA, MAP2K1, MAP2K2, MAPK10, MBD5, MBTPS2, MCPH1, MECP2, MED12, MED13L, MED23, MEF2C, MFSD8, MID1, MTHFR, MTOR, MYT1L, NAA10, NAGLU, NDE1, NDP, NDUFA1, NECAP1, NEXMIF, NF1, NFIX, NHLRC1, NHS, NIPBL, NLGN4X, NOTCH3, NPRL2, NPRL3, NRAS, NRXN1, NRXN3, NSD1, NSDHL, NSUN2, OCRL, OFD1, OPHN1, OTC, PACS1, PAFAH1B1, PAK3, PANK2, PAX6, PCDH19, PDCD10, PDHA1, PGK1, PHF6, PHF8, PIGA, PIGN, PIGO, PIGV, PLA2G6, PLCB1, PLP1, PNKP, PNPO, POGZ, POLG, POMGNT1, POMT1, POMT2, PORCN, PPT1, PQBP1, PRICKLE1, PRICKLE2, PRPS1, PRRT2, PTCH1, PTCHD1, PTEN, PTPN11, PURA, RAB39B, RAD21, RAF1, RAI1, RARS2, RBM10, RELN, RIT1, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, RPL10, RPS6KA3, SAMHD1, SATB2, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN8A, SCN9A, SERPINI1, SETBP1, SETD2, SETD5, SGSH, SHANK2+, SHANK3+, SHOC2, SIK1, SLC12A5, SLC13A5, SLC16A2, SLC19A3, SLC1A2, SLC25A1, SLC25A19, SLC25A22, SLC2A1, SLC35A2, SLC4A10, SLC6A1, SLC6A4, SLC6A8, SLC9A6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMS, SNAP25, SNAP29, SOS1, SOX11, SOX5, SPRED1, SPTAN1, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SYP, SZT2, TAF1, TBC1D24, TBL1XR1, TBR1, TBX1, TCF12, TCF20, TCF4, THOC2, TIMM8A, TMEM231, TMLHE, TPP1, TRAPPC9, TREX1, TRIO, TRPM6, TSC1, TSC2, TSPAN7, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, TUSC3, UBE2A, UBE3A, UNC80, UPF3B, USP9X, VANGL1, VPS13A, VPS13B, WAC, WDR45, WDR62, WDR81, WWOX, YWHAE, ZC4H2, ZDHHC9, ZEB2, ZMYND11, ZNF407, ZNF711*
Comprehensive Epilepsy Panel:	*ABAT, ACY1, ADGRG1, ADGRV1, ADSL, ALDH5A1, ALDH7A1, ALG13, AMT, AP1S2, ARFGEF2, ARHGEF9, ARX, ASPM, ATP1A2, ATP2A2, ATP6AP2, ATP6V0A2, ATR, ATRX, BCKDK, CACNA1A, CACNA1H, CACNA2D2, CACNB4, CASK, CASR, CCDC88C, CCM2, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLCN2, CLN3, CLN5, CLN6, CLN8, CNTNAP2, COL18A1, COL4A1, CPA6, CREBBP, CSTB, CTSD, CUL4B, DCX, DEPDC5, DNAJC5, DNM1, DOCK7, DYRK1A, EEF1A2, EFHC1, EHMT1, EMX2, EPM2A, FGD1, FGFR3, FKRP, FLNA, FLVCR2, FOLR1, FOXG1, GABRA1, GABRB2, GABRB3, GABRD, GABRG2, GAMT, GATM, GCSH, GLDC, GLI2, GNAO1, GNB1, GOSR2, GPC3, GPHN, GRIA3, GRIN1, GRIN2A, GRIN2B, HCN1, HCN4, HIP1, HNRNPU, HSD17B10, HTRA1, IQSEC2, ITPA, KANSL1, KCNA1, KCNA2, KCNB1, KCNJ1, KCNJ10, KCNJ11, KCNMA1, KCNQ2, KCNQ3, KCNT1, KCTD7, KDM5C, KIF1BP, KMT2D, KRIT1, LARGE1, LBR, LGI1, LIAS, MAGI2, MAPK10, MBD5, MCPH1, MECP2, MEF2C, MFSD8, MTHFR, MTOR, NDE1, NDUFA1, NECAP1, NHLRC1, NIPBL, NOTCH3, NPRL2, NPRL3, NRXN1, OPHN1, PAFAH1B1, PAK3, PANK2, PAX6, PCDH19, PDCD10, PHF6, PIGA, PIGO, PIGV, PLA2G6, PLCB1, PLP1, PNKP, PNPO, POLG, POMGNT1, POMT1, POMT2, PPT1, PQBP1, PRICKLE1, PRICKLE2, PRRT2, PTEN, RAB39B, RAI1, RARS2, RELN, RNASEH2A, RNASEH2B, RNASEH2C, ROGDI, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN5A, SCN8A, SCN9A, SERPINI1, SETBP1, SHANK3+, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC25A19, SLC25A22, SLC2A1, SLC35A2, SLC4A10, SLC6A1, SLC6A8, SLC9A6, SMC1A, SMC3, SMS, SNAP25, SNAP29, SPTAN1, ST3GAL3, ST3GAL5, STX1B, STXBP1, SYN1, SYNGAP1, SYP, SZT2, TBC1D24, TBX1, TCF4, TPP1, TREX1, TRPM6, TSC1, TSC2, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, UBE3A, VANGL1, VPS13A, VPS13B, WDR62, WWOX, ZEB2*
Comprehensive Autism Spectrum Disorder Panel:	*ABCD1, ACSL4, ADNP, ADSL, AFF2, AGO1, AHI1, AIFM1, ALDH5A1, ANK3, ANKRD11, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, ARID1A, ARID1B, ARX, ATP13A2, ATP7A, ATP8A2, ATRX, AUTS2, BCKDK, BCL11A, BCOR, BRAF, BRWD3, C12ORF57, CA8, CACNA1C, CASK, CBL, CC2D1A, CCDC22, CDKL5, CDKN1C, CHD2, CHD7, CHD8, CLCN4, CNTNAP2, CREBBP, CTCF, CTNNB1, CUL3, CYP27A1, D2HGDH, DDHD2, DDX3X, DEAF1, DHCR7, DIS3L2, DKC1, DLG3, DMD, DNMT3A, DPYD, DYNC1H1, DYRK1A, EBP, EHMT1, EIF2S3, ELP4, EZH2, FGD1, FMR1, FOLR1, FOXG1, FOXP1, FOXP2, FTSJ1, GATAD2B, GDI1, GK, GLI3, GNS, GPC3, GRIA3, GRIK2, GRIN2B, GRIP1, HCCS, HDAC8, HGSNAT, HPRT1, HRAS, HUWE1, IDS, IL1RAPL1, KAT6A, KAT6B, KCTD13, KDM5C, KDM6A, KIF1A, KIRREL3, KMT2A, KRAS, L1CAM, L2HGDH, LAMC3, LAMP2, LAS1L, LINS1, LRP2, MAGEL2, MAN1B1, MAOA, MAP2K1, MAP2K2, MBD5, MBTPS2, MECP2, MED12, MED13L, MED23, MEF2C, MID1, MYT1L, NAA10, NAGLU, NDP, NEXMIF, NF1, NFIX, NHS, NIPBL, NLGN4X, NRAS, NRXN1, NRXN3, NSD1, NSDHL, NSUN2, OCRL, OFD1, OPHN1, OTC, PACS1, PAFAH1B1, PCDH19, PDHA1, PGK1, PHF6, PHF8, PIGN, PNKP, POGZ, PORCN, PQBP1, PRPS1, PTCH1, PTCHD1, PTEN, PTPN11, PURA, RAB39B, RAD21, RAF1, RAI1, RARS2, RBM10, RIT1, RPL10, RPS6KA3, SATB2, SCN1A, SCN2A, SETBP1, SETD2, SETD5, SGSH, SHANK2+, SHANK3+, SHOC2, SLC16A2, SLC25A1, SLC6A1, SLC6A4, SLC6A8, SLC9A6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SOS1, SOX11, SOX5, SPRED1, STXBP1, SYN1, SYNGAP1, TAF1, TBL1XR1, TBR1, TCF12, TCF20, TCF4, THOC2, TIMM8A, TMEM231, TMLHE, TRAPPC9, TRIO, TSC1, TSC2, TSPAN7, TUBA1A, TUSC3, UBE2A, UBE3A, UNC80, UPF3B, USP9X, VPS13B, WAC, WDR45, WDR81, YWHAE, ZC4H2, ZDHHC9, ZEB2, ZMYND11, ZNF407, ZNF711*
STAT Autism Spectrum Disorder Panel:	*AHI1, AP1S2, ARX, CACNA1C, CDKL5, CNTNAP2, DHCR7, FMR1, GPC3, GRIA3, IL1RAPL1, KDM5C, MECP2, NLGN4X, NRXN1, NSD1, OPHN1, OTC, PCDH19, PTCHD1, PTEN, RAB39B, SHANK2+, SHANK3+, SLC6A8, SLC9A6, TSC1, TSC2, UBE3A, UPF3B*
Microcephaly Panel:	*ACTB, ACTG1, ADGRG1, ARFGEF2, ARX, ASPM, ATR, ATRIP, CASK, CDK5RAP2, CDKL5, CDON, CENPJ, CEP135, CEP152, CEP63, CREBBP, DCX, DHCR7, DISP1, DLL1, DYNC1H1, EP300, ERCC6, ERCC8, FGF8, FKRP, FKTN, FOXG1, FOXH1, GAS1, GLI2, HDAC8, KIF11, KIF2A, KNL1, LARGE1, MCPH1, MECP2, MED17, MRE11, NBN, NDE1, NHEJ1, NIN, NIPBL, NODAL, PAFAH1B1, PCNT, PHGDH, PNKP, POMGNT1, POMT1, POMT2, PTCH1, RAB18, RAB3GAP1, RAB3GAP2, RAD21, RBBP8, RELN, RNU4ATAC, SHH, SIX3, SLC25A19, SLC9A6, SMC1A, SMC3, STAMBP, STIL, TDGF1, TGIF1, TUBA1A, TUBG1, UBE3A, VLDLR, WDR62, ZIC2*
Craniosynostosis Panel:	*ENFB1, FGFR1, FGFR2, FGFR3, MSX2, POR, RAB23, TWIST1*
Limb Defects Panel:	*GLI3, HOXD13, ROR2, SALL1, SALL4, TBX5, WNT7A,* ZRS
Comprehensive Cardiovascular Panel:	*A2ML1, ABCC9, ACADVL, ACTA2, ACTC1, ACTN2, ACVR2B, ACVRL1, ADAMTS2, AGK, AGL, AKAP9, ALG1, ALG12, ALMS1, ALPK3, ANK2, ANKRD1, APOB, ARSB, ATP7A, BAG3, BCOR, BMPR1B, BMPR2, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV1, CAV3, CAVIN4, CBL, CBS, CHD7, CHRM2, CHST14, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COX10, COX15, CPT2, CRELD1, CRYAB, CSRP3, CTNNA3, DES, DMD, DNAH5, DOLK, DSC2, DSG2, DSP, DTNA, EFEMP2, EIF2AK4, ELAC2, ELN, EMD, ENG, EYA4, FBN1, FBN2, FHL1, FKBP14, FKRP, FKTN, FLNA, FLNC, FOXH1, GAA, GATA4, GATA6, GATAD1, GBA, GBE1, GDF1, GDF2, GJA1, GJA5, GLA, GLB1, GPC3, GPD1L, GUSB, HADHA, HAND1, HCN4, HEXB, HRAS, IDUA, ILK, JAG1, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK3, KCNQ1, KRAS, LAMA4, LAMP2, LDB3, LDLR, LDL-RAP1, LEFTY2, LMNA, LZTR1, MAP2K1, MAP2K2, MAT2A, MED12, MED13L, MEIS2, MFAP5, MIB1, MLYCD, MTO1, MUT, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYOM1, MYO22, MYPN, NDUFA10, NDUFA12, NDUFA2, NDUFA9, NDUFAF2, NDUFAF6, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NODAL, NOTCH1, NPPA, NR2F2, NRAS, NSD1, OBSCN, PCCA, PCCB, PCSK9, PDLIM3, PET100, PGM1, PHYH, PKP2, PLEKHM2, PLN, PLOD1, PMM2, PPP1CB, PRDM16, PRKAG2, PRKG1, PTPN11, RAF1, RANGRF, RASA1, RASA2, RBM20, RIT1, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO1, SDHA, SDHAF1, SGCD, SGSH, SHOC2, SKI, SLC22A5, SLC25A20, SLC2A10, SLC39A13, SLMAP, SMAD3, SMAD4, SMAD6, SMAD9, SNTA1, SOS1, SOS2, SPRED1, SURF1, TAZ, TBX1, TBX5, TCAP, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, TXNRD2, VCL, ZFPM2, ZIC3*
Comprehensive Cardiomyopathy Panel:	*ABCC9, ACADVL, ACTA2, ACTC1, ACTN2, AGK, AGL, AKAP9, ALG1, ALG12, ALMS1, ALPK3, ANK2, ANKRD1, ARSB, BAG3, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CAVIN4, CBL, CBS, CHRM2, COL3A1, COL5A1, COL5A2, COX10, COX15, CPT2, CRYAB, CSRP3, CTNNA3, DES, DMD, DOLK, DSC2, DSG2, DSP, DTNA, ELAC2, EMD, EYA4, FBN1, FBN2, FHL1, FKRP, FKTN, FLNA, FLNC, GAA, GATA4, GATA6, GATAD1, GBA, GBE1, GJA5, GLA, GLB1, GPD1L, GUSB, HADHA, HCN4, HEXB, HRAS, IDUA, ILK, JAG1, JPH2, JUP, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNQ1, KRAS, LAMA4, LAMP2, LDB3, LMNA, MAP2K1, MAP2K2, MED12, MIB1, MLYCD, MTO1, MUT, MYBPC3, MYH11, MYH6, MYH7, MYL2, MYL3, MYLK, MYLK2, MYOM1, MYOZ2, MYPN, NDUFA10, NDUFA12, NDUFA2, NDUFA9, NDUFAF2, NDUFAF6, NDUFS3, NDUFS4, NDUFS7, NDUFS8, NEBL, NEXN, NKX2-5, NOTCH1, NPPA, NRAS, OBSCN, PCCA, PCCB, PDLIM3, PET100, PGM1, PHYH, PKP2, PLEKHM2, PLN, PLOD1, PMM2, PRDM16, PRKAG2, PRKG1, PTPN11, RAF1, RANGRF, RASA1, RBM20, RIT1, RYR2, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCO1, SDHA, SDHAF1, SGCD, SGSH, SHOC2, SKI, SLC22A5, SLC25A20, SLC2A1O, SLMAP, SMAD3, SMAD4, SNTA1, SOS1, SURF1, TAZ, TBX1, TBX5, TCAP, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNT2, TPM1, TRDN, TRPM4, TTN, TTR, TXNRD2, VCL*
Noonan Spectrum Disorders Panel:	*A2ML1, BRAF, CBL, HRAS, KRAS, LZTR1, MAP2K1, MAP2K2, NF1, NRAS, PPP1CB, PTPN11, RAF1, RASA2, RIT1, SHOC2, SOS1, SOS2, SPRED1*
Comprehensive Immunodeficiency Panel	*ACD, ACP5, ADA, ADA2, ADAM17, ADAR, AICDA, AIRE, AK2, AP3B1, B2M, BTK, C1QA, C1QB, C1QC, C1S, C2, C3, C4A, C4B, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD9, CASP10, CASP8, CCDC39, CCDC40, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD59, CD79A, CD79B, CD81, CD8A, CEBPE, CFB, CFD, CFH, CFHR1, CFHR3, CFI, CFP, CHD7, CIITA, CLPB, COPA, CORO1A, CR2, CSF2RA, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DCLRE1B, DCLRE1C, DGKE, DKC1, DNMT3B, DOCK2, DOCK8, DUOX2, ELANE, EPCAM, EPG5, FADD, FAS, FASLG, FCN1, FERMT1, FERMT3, FOXN1, FOXP3, G6PC3, G6PD, GATA2, GATA3, GFI1, GUCY2C, HAX1, HPS1, HPS4, HPS6, ICOS, IFIH1, IFNGR1, IFNGR2, IGHM, IGLL1, IKBKB, IKBKG, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17F, IL17RA, IL17RC, IL18, IL18RAP, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RG, IL36RN, IL7R, IN080, IRAK4, IRF7, IRF8, IRGM, ISG15, ITGAM, ITGB2, ITK, JAGN1, JAK1, JAK3, LAMTOR2, LCK, LIG1, LIG4, LPIN2, LRBA, LRRC8A, LYST, MAGT1, MALT1, MAP3K14, MASP2, MEFV, MRE11, MVK, MYD88, MYO5B, NBN, NCF1, NCF2, NCF4, NFAT5, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP12, NLRP3, NOD2, NOP10, NRAS, ORAI1, PGM3, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, POLE, PRF1, PRKCD, PRKDC, PTEN, PTPRC, RAB27A, RAC2, RAG1, RAG2, RBCK1, RET, RFX5, RFXANK, RFXAP, RHOH, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RORC, RTEL1, SAMHD1, SBDS, SERPING1, SH2D1A, SKIV2L, SLC35C1, SLC37A4, SLC9A3, SMARCAL1, SP110, SPINK5, STAT1, STAT3, STAT5B, STIM1, STK4, STX11, STXBP2, TAP1, TAP2, TAPBP, TAZ, TBX1, TCN2, TERC, TERT, TINF2, TLR3, TMEM173, TNFAIP3, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFSF12, TREX1, TRIM22, TRNT1, TTC37, TTC7A, TYK2, UNC13D, UNG, USB1, VPS13B, VPS45, WAS, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24*

Pharmacogenetic Genes by Panel

PANELS	GENES
Comprehensive PGx Panel:	*ABCB1, ABCG2, ADRA2A, COMT, CYP1A2, CYP2B6, CYP2C, CYP2C19, CYP2C8, CYP2C9, CYP2D6, CYP3A4, CYP3A5, CYP4F2, DPYD, DRD2, F2, F5, G6PD, GRIK4, HTR2A, HTR2C, NUDT15, OPRM1, SLCO1B1, TPMT, UGT1A1, UGT2B15, VKORC1*
Cardiovascular PGx Panel:	*ABCG2, CYP2C, CYP2C19, CYP2C9, CYP2D6, CYP3A4, CYP4F2, F2, F5, SLCO1B1, VKORC1*
Epilepsy PGx Panel:	*ABCG2, COMT, CYP2B6, CYP2C19, CYP2C9, CYP2D6, GRIK4, OPRM1, UGT2B15*
Oncology PGx Panel:	*CYP2C8, CYP2D6, DPYD, G6PD, NUDT15, TPMT, UGT1A1*
Pain PGx Panel:	*ABCG2, COMT, CYP2B6, CYP2C19, CYP2C9, CYP2D6, G6PD, OPRM1*
Pediatric PGx Panel:	*CYP2C19, CYP2C9, CYP2D6, CYP3A5, DPYD, SLCO1B1, TPMT, UGT1A1, VKORC1*
Psychiatry PGx Panel:	*COMT, CYP1A2, CYP2B6, CYP2C19, CYP2C9, CYP2D6, DRD2, GRIK4, HTR2A, HTR2C, UGT2B15*

COMPLETE AND SUBMIT ALL PAGES TO LABORATORY

Informed Consent for Genetic Testing

I, _____, hereby request genetic testing, which may include molecular, cytogenetic and/or biochemical analyses, for

☐ Myself

☐ My child _____

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 understood, the nature of the genetic testing that I/my child 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 villi and/or urine sample, will be taken from me/my child. I understand that the samples will be used for determining if I/my child have a genetic disease, are carriers of a genetic disease, or are more likely to develop a genetic disease or condition.

The nature of the genetic test(s) that have been 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. The likelihood of this occurring has been estimated to be less than 1%. I understand that a negative result reduces, but does not eliminate, the possibility that I/my child carry a mutation(s) in the gene(s) analyzed or in other gene(s) that are not included in the test.

I understand that no test will be performed on my sample other than the one(s) authorized by me and my healthcare provider. I have reviewed the test order made in connection with this consent, and I hereby give consent to have my specimen tested as set forth in the order.

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, and 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, including scientific databases that are maintained by the federal government, such as a database kept by the National Institutes of Health ("NIH") (an agency of the federal government that funds research). Researchers have to apply to the NIH to see the information in the database. If I do not want to have any of my de-identified data and information used in research consistent with this consent, I may initial here_____, or I may withdraw this consent by contacting Sema4, including by emailing privacy@sema4.com.

Permission to contact

I understand that Sema4 may wish to contact me/my child 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/my child's sample(s). I understand that I may notify Sema4 to opt out of such future contact, including by emailing privacy@sema4.com.

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. In addition, incidental findings that are not related to the primary diagnosis may be identified in me/my child. An error in the diagnosis may occur if the true biological relationships of the family members involved are not as I have stated and this test may detect non-paternity.

The results of my/my child's test will be explained to me by a genetic counselor or by my physician who will have the opportunity to discuss my results with a geneticist. I have had the opportunity to have all of my questions answered. If I am signing this form on behalf of a minor for whom I am the legal guardian, I am satisfied that I have received enough information to sign on his or her behalf.

I understand that this consent is being obtained in order to protect my right to have all of my questions answered before testing. 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 who I designate to receive this information.

_____ _____

Signature of person being tested (or guardian) Date

FFP0122GE0121

Revised 01/20/2021

COMPLETE AND SUBMIT ALL PAGES TO LABORATORY