sema4
a Mount Sinai venture

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GENERAL TEST REQUISITION

SPECIMENS: 1428 Madison Ave., Rm AB2-25, New York, NY 10029
MAIL: One Gustave L. Levy Place, Box 1497, New York, NY 10029-6574
Phone: **800-298-6470** / Fax: 646-859-6870
Tax ID# 47-5349024/ CLIA# 33D2097541

ACCESSION NO.	DATE / /

PATIENT INFORMATION

LAST NAME	FIRST NAME
DATE OF BIRTH / /	BIOLOGICAL GENDER ☐ M ☐ F
PARTNER / SPOUSE LAST NAME	PARTNER / SPOUSE FIRST NAME
CLIENT MRN	PARTNER / SPOUSE DATE OF BIRTH / /
TELEPHONE	EMAIL
ADDRESS	CITY / STATE / ZIP

REFERRING PROVIDER INFORMATION

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 / /**

BILLING INFORMATION ■ Bill Clinic ■ Bill Insurance Below ■ Self Pay

POLICYHOLDER LAST NAME	POLICYHOLDER FIRST NAME	POLICYHOLDER DOB / /
INSURANCE CARRIER	INSURANCE ID	GROUP NO.

BILLING ADDRESS

OTHER HEALTH COVERAGE (IDENTIFY)

SELF-PAY: ☐ Credit Card ☐ Check

Make Checks Payable to: Mount Sinai Genomics Inc., P.O. Box 21312, New York, NY 10087-1312

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 800-298-6570, Option 3.

SIGNATURE DATE / /

CLINICAL INDICATIONS

SPECIMEN TYPE
(Please contact laboratory for alternate specimen types)
☐ AMNIOTIC FLUID ☐ BLOOD ☐ CVS
☐ DBS ☐ PLASMA ☐ URINE
☐ OTHER _____
☐ CULTURED CELLS TYPE _____

DATE / TIME SPECIMEN DRAWN
AM PM / /

DATE SPECIMEN SENT / /

☐ LMP / / 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

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
- ☐ *FGFR3* 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)**

50000	50000	50000	50000
Patient Name:_____	Patient Name:_____	Patient Name:_____	Patient Name:_____

CLIENT SERVICES

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, MY015A, MY03A, MY06, MY07A, MY0C, 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, AG01, 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, GNS, 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, AG01, 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, LDLRAP1, 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, MYO22, 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, SLC2A10, 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, COR01A, 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, INO80, 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, MY05B, 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*

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, _____ , 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 this consent and my doctor.

De-identified research

Sema4 may also give the de-identified information to its research partners and may submit this de-identified information 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. Any information that could directly identify you (such your name or address) will not be provided to a scientific database.

If I prefer not to have any of my/my child's de-identified health information used in research consistent with this consent, I may initial here _____ or request this 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.

Sample storage

_____
Initials

By initialing here, I agree that Sema4 may store, de-identify, and use my/my child's sample and information to support medical and academic research relating to health, disease prevention, drug development, and other scientific purposes, and that I/my child will receive no compensation in connection with such research. If I do not initial here, my/my child's sample will be destroyed at the end of the testing process or not more than 60 days after collection. I understand that I may withdraw this consent by contacting Sema4 (including by emailing privacy@sema4.com).

_____ _____

Signature of person being tested (or guardian) Date