sema4

Please place green collection kit barcode here.

123456-2-X

PRENATAL TESTING REQUISITION

SPECIMENS: 1428 Madison Ave., Rm AB2-25, New York, NY 10029
Phone: **800-298-6470** / Fax: 212-241-0139
Tax ID# 47-5349024/ CLIA# 33D2097541

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 GENDER ☐ 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, Option 3.

SIGNATURE DATE MM / DD / YYYY

ORDERING PHYSICIAN INFORMATION

NAME REQUIRED	GENETIC COUNSELOR
ADDRESS REQUIRED	CLINIC / INSTITUTION REQUIRED
	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. 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)

- ☐ **O09.511** Advanced Maternal Age, First Trimester
- ☐ **O09.512** Advanced Maternal Age, Second Trimester
- ☐ **N96** Recurrent pregnancy loss
- ☐ **Z82.7** Family history of congenital malformations, deformations, and chromosomal abnormalities
- ☐ **Z84.81** Family history of carrier of genetic disease
- ☐ **O28.3** Abnormal ultrasonic finding on antenatal screening of mother
- ☐ **O28.5** Abnormal chromosomal and genetic finding on antenatal screening of mother
- ☐ **N46** Male infertility
- ☐ **N97** Female infertility
- ☐ **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
- ☐ **O02.89** Other abnormal products of conception
- ☐ **Other** _____

COLLECTION DATE ___ /___ /___ # OF BLOOD TUBES SENT: YELLOW____ PURPLE____ BCT____ GREEN____

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

Maternal: ☐ Peripheral Blood ☐ Saliva ☐ Other_____
Paternal: ☐ Peripheral Blood ☐ Saliva ☐ Other_____
Fetal: ☐ Amniotic Fluid ☐ Chorionic Villi ☐ POC ☐ Other_____

PREGNANCY HISTORY: **Gestational Age:** _____Weeks _____Days *or* EDD: ____ /____ /____ (Required for fetal samples.)

Pregnancy conceived: ☐ IVF ☐ Egg donor/gestational carrier Age of genetic mother (at time of retrieval):_____

LABORATORY TEST(S) ORDERED

Test Selection (Required)

Parental Carrier Screening

Carrier Screening Clinical Information:

Patient ancestry: _____ Preferred Language: _____

History of BMT or recent blood transfusion in the last 4 weeks? ☐ YES ☐ NO
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: _____

Family History of: _____ Partner Carrier of: _____

- ☐ Expanded Carrier Screen (283 genes)
- ☐ Standard Pan-ethnic Panel (4 genes)
- ☐ High Frequency Pan-ethnic Panel (11 genes)
- ☐ ECS 39 (39 genes)
- ☐ ECS 152 (152 genes)
- ☐ Comprehensive Jewish Carrier Screen (101 genes)
- ☐ Ashkenazi Jewish Disorders (47+17 genes)
- ☐ Sephardi-Mizrahi Jewish Disorders (37+17 genes)
- ☐ X-Linked Supplemental Panel (21 genes)
- ☐ Single Gene_____ Variant(s)_____
  ☐ Phase analysis ☐ PGT set-up
- ☐ Test only for AR disorders partner screened positive for - hold sample pending partner results (mark on reverse side or list here):_____
- ☐ NGS re-analysis of ECS 281 + enhancements included in ECS 283. Previous test order date: ____/____/____
- ☐ Other _____

NONINVASIVE PRENATAL TEST (NIPT) MENU - *select only one*

☐ **MaterniT GENOME**
Genome-wide fetal aneuploidies (singleton only)

OR

MaterniT21 PLUS
Select fetal aneuploidies
☐ Core (chr 21, 18, 13, sex)
☐ Core +ESS*
☐ Core + SCA**
☐ Core + ESS + SCA

*ESS = chr 16, chr 22, and select microdeletions **SCA = sex chromosome aneuploidies

REQUIRED CLINICAL INFORMATION

Specimen Required: Two 10 mL Whole Blood BCT Streck Tubes

Gestational age: _____ weeks _____ days *or* **EDD:** _____ /_____ /_____

Gestation ☐ Singleton ☐ Twins ☐ Triplets ☐ Other:_____

Maternal height: _____ ft. _____ in. Maternal weight: _____ lbs.

MEDICAL INDICATION FOR TESTING *Select one or more ICD10 codes*

High risk for fetal chromosomal aneuploidies

Maternal age: Primigravida ☐ O09.511 1st tri ☐ O09.512 2nd tri ☐ O09.513 3rd tri
Multigravida ☐ O09.521 1st tri ☐ O09.522 2nd tri ☐ O09.523 3rd tri

Abnormal serum biochemical screening: ☐ O28.1

Ultrasound finding: (specify type[s] in Comments) ☐ O35.1XX0

Personal family history (specify type in Comments)
Prior pregnancy with trisomy ☐ O09.291 1st tri ☐ O09.292 2nd tri ☐ O09.293 3rd tri
Robertsonian translocation ☐ Q95.0 ☐ Q95.1

Other high risk factor: ICD10 code_____

No known high risk for fetal chomosomal aneuploidies (MaterniT21 PLUS or VisibiliT only)
☐ Z34.91 1st tri ☐ Z34.92 2nd tri ☐ Z34.93 3rd tri ☐ Other ICD10 code:_____

Infertility/Pregnancy Loss

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

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 (select option below)
- ☐ 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) For all specimens, please include blood (1 EDTA purple top, 1 Sodium heparin green top) from parents of the proband/pregnancy for array follow up if available

- ☐ **Prenatal Chromosomal Microarray** (lower resolution)
- ☐ **High Resolution Chromosomal Microarray** prenatal/postnatal/POC
- ☐ **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.**
- ☐ **Parental array followup**

Proband SEMA4 Lab ID: _____ Name: _____ DOB: _____

FLUORESCENT *in situ* HYBRIDIZATION (FISH)

- ☐ Aneuploidy FISH (chromosomes 13,18,21,X,Y)
- ☐ 1p36 deletion syndrome (1p36.3)
- ☐ 22q deletion/DiGeorge syndrome (22q11.2)
- ☐ CHARGE syndrome (8q12.1-q12.2)
- ☐ Cri-du-chat syndrome (5p15.2)
- ☐ Kallman syndrome (Xp22.3)
- ☐ Langer-Giedion syndrome (8q23.3-q24.11)
- ☐ Miller-Dieker syndrome (17p13.3)
- ☐ SRY (Yp11.3)
- ☐ Prader-Willi/Angelman (15q11.2)
- ☐ Rubenstein-Taybi syndrome (16p13.3)
- ☐ Smith-Magenis syndrome (17p11.2)
- ☐ Sotos syndrome (5q35)
- ☐ STS deficiency (Xp22.3)
- ☐ Williams syndrome (7q11.23)
- ☐ Wolf-Hirschhorn syndrome (4p16.3)
- ☐ FISH other: _____

Prenatal Diagnostic Testing

- ☐ *FGFR3* Hotspot Panel
  ☐ Reflex to sequencing if negative
- ☐ Limb Defects Next Gen Sequencing Panel (7 genes)
- ☐ Noonan Spectrum Disorders Panel (18 genes)
- ☐ Single gene/Diagnostic testing
  ☐ Gene:_____
  ☐ Targeted testing: Maternal variant:_____ Paternal variant: _____

Biochemical testing: ☐ Tay-Sachs enzyme analysis ☐ Sandhoff enzyme analysis

- ☐ Maternal Cell Contamination
- ☐ Other: _____

- *Maternal blood is required for all prenatal specimens for maternal cell contamination.*
- *If patient/partner was NOT tested at Sema4, parental bloods are required (5-10mL EDTA) to confirm the variant in-house. Please also provide a copy of any previous results.*
- ***Please contact the laboratory for all prenatal molecular/biochemical testing***

COMPLETE AND SUBMIT ALL PAGES TO LABORATORY

Expanded Carrier Screen Panel includes:

- ☐ Abetalipoproteinemia ◆ ● ▼
- ☐ Achromatopsia ▼
- ☐ Acrodermatitis Enteropathica
- ☐ Acute Infantile Liver Failure ◆ ■ ▼
- ☐ Acyl-CoA Oxidase I Deficiency
- ☐ Adenosine Deaminase Deficiency ▼
- ☐ Adrenoleukodystrophy, X-Linked ◆ ■ ▼
- ☐ Aicardi-Goutières Syndrome (SAMHD1-Related)
- ☐ Alpha-Mannosidosis
- ☐ Alpha-Thalassemia ▲ ✠ ◆ ● ■ ▼
- ☐ Alpha-Thalassemia Mental Retardation Syndrome
- ☐ Alport Syndrome (COL4A3-Related) ◆ ● ▼
- ☐ Alport Syndrome (COL4A4-Related)
- ☐ Alport Syndrome (COL4A5-Related)
- ☐ Alstrom Syndrome
- ☐ Andermann Syndrome ▼
- ☐ Argininosuccinic Aciduria ▼
- ☐ Aromatase Deficiency
- ☐ Arthrogryposis, Mental Retardation, and Seizures ◆ ● ▼
- ☐ Asparagine Synthetase Deficiency ◆ ■ ▼
- ☐ Aspartylglycosaminuria ▼
- ☐ Ataxia With Isolated Vitamin E Deficiency
- ☐ Ataxia-Telangiectasia ◆ ■ ▼
- ☐ Autosomal Recessive Spastic Ataxia of Charlevoix-Saguenay ▼
- ☐ Bardet-Biedl Syndrome (BBS10-Related)
- ☐ Bardet-Biedl Syndrome (BBS12-Related)
- ☐ Bardet-Biedl Syndrome (BBS1-Related) ▼
- ☐ Bardet-Biedl Syndrome (BBS2-Related) ◆ ● ▼
- ☐ Bare Lymphocyte Syndrome, Type II
- ☐ Bartter Syndrome, Type 4A
- ☐ Bernard-Soulier Syndrome, Type A1
- ☐ Bernard-Soulier Syndrome, Type C
- ☐ 3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency
- ☐ Beta-Ketothiolase Deficiency
- ☐ Beta-Globin-Related Hemoglobinopathies ▲ ◆ ■ ● ✠ ▼
- ☐ Bilateral Frontoparietal Polymicrogyria
- ☐ Biotinidase Deficiency ▼
- ☐ Bloom Syndrome ◆ ● ✠ ▼
- ☐ Canavan Disease ◆ ● ✠ ▼
- ☐ Carbamoylphosphate Synthetase I Deficiency
- ☐ Carnitine Palmitoyltransferase IA Deficiency
- ☐ Carnitine Palmitoyltransferase II Deficiency ◆ ● ▼
- ☐ Carpenter Syndrome
- ☐ Cartilage-Hair Hypoplasia ▼
- ☐ Cerebral Creatine Deficiency Syndrome 1
- ☐ Cerebral Creatine Deficiency Syndrome 2
- ☐ Cerebrotendinous Xanthomatosis ◆ ■ ▼
- ☐ Charcot-Marie-Tooth Disease, Type 4D
- ☐ Charcot-Marie-Tooth Disease, Type 5 / Arts syndrome
- ☐ Charcot-Marie-Tooth Disease, X-Linked
- ☐ Choreoacanthocytosis ◆ ● ▼
- ☐ Choroideremia
- ☐ Chronic Granulomatous Disease (CYBA-related) ◆ ■ ▼
- ☐ Chronic Granulomatous Disease (CYBB-related)
- ☐ Citrin Deficiency ▼
- ☐ Citrullinemia, Type 1 ✠
- ☐ Cohen Syndrome
- ☐ Combined Malonic and Methylmalonic Aciduria ▼
- ☐ Combined Oxidative Phosphorylation Deficiency 1
- ☐ Combined Oxidative Phosphorylation Deficiency 3 ▼
- ☐ Combined Pituitary Hormone Deficiency 2 ▼
- ☐ Combined Pituitary Hormone Deficiency 3
- ☐ Combined SAP Deficiency
- ☐ Congenital Adrenal Hyperplasia due to 17-Alpha-Hydroxylase Deficiency
- ☐ Congenital Adrenal Hyperplasia due to 21-Alpha-Hydroxylase Deficiency ◆ ● ■
- ☐ Congenital Amegakaryocytic Thrombocytopenia ◆ ● ▼
- ☐ Congenital Disorder of Glycosylation, Type Ia ▲ ◆ ● ✠ ■ ▼
- ☐ Congenital Disorder of Glycosylation, Type Ib
- ☐ Congenital Disorder of Glycosylation, Type Ic
- ☐ Congenital Insensitivity to Pain with Anhidrosis ◆ ■ ▼
- ☐ Congenital Myasthenic Syndrome (CHRNE-Related)
- ☐ Congenital Myasthenic Syndrome (RAPSN-Related) ◆ ■ ▼
- ☐ Congenital Neutropenia (HAX1-Related)
- ☐ Congenital Neutropenia (VPS45-Related)
- ☐ Corneal Dystrophy and Perceptive Deafness
- ☐ Corticosterone Methyloxidase Deficiency ◆ ■ ▼
- ☐ Cystic Fibrosis ◗ ▲ ◆ ● ■ ✠ ▼
- ☐ Cystinosis ◆ ■ ▼
- ☐ D-Bifunctional Protein Deficiency
- ☐ Deafness, Autosomal Recessive 77 ◆ ● ▼
- ☐ Duchenne Muscular Dystrophy / Becker Muscular Dystrophy ▲ ✠ ◆ ● ■ ▼
- ☐ Dyskeratosis Congenita (RTEL1-Related) ◆ ● ▼
- ☐ Dystrophic Epidermolysis Bullosa ▼
- ☐ Ehlers-Danlos Syndrome, Type VIIC ◆ ● ▼
- ☐ Ellis-van Creveld Syndrome (EVC-Related) ▼
- ☐ Emery-Dreifuss Myopathy 1
- ☐ Enhanced S-Cone Syndrome ◆ ● ▼
- ☐ Ethylmalonic Encephalopathy
- ☐ Fabry Disease
- ☐ Factor IX Deficiency
- ☐ Factor XI Deficiency ◆ ● ▼
- ☐ Familial Autosomal Recessive Hypercholesterolemia ◆
- ☐ Familial Dysautonomia ◆ ● ✠ ▼
- ☐ Familial Hypercholesterolemia ●▼
- ☐ Familial Hyperinsulinism (ABCC8-Related) ◆ ● ✠ ▼
- ☐ Familial Hyperinsulinism (KCNJ11-Related) ▼
- ☐ Familial Mediterranean Fever ◆ ● ■ ▼
- ☐ Fanconi Anemia, Group A ◆ ■ ▼
- ☐ Fanconi Anemia, Group C ◆ ● ✠ ▼
- ☐ Fanconi Anemia, Group G ▼
- ☐ Fragile X Syndrome ◗ ▲ ◆ ● ■ ✠ ▼
- ☐ Fumarase Deficiency ▼
- ☐ Galactokinase Deficiency ▼
- ☐ Galactosemia ◆ ● ✠ ▼
- ☐ Gaucher Disease ◆ ● ✠ ▼
- ☐ Gitelman Syndrome ▼
- ☐ Glutaric Acidemia, Type I ▼
- ☐ Glutaric Acidemia, Type IIa
- ☐ Glutaric Acidemia, Type IIc ▼
- ☐ Glycine Encephalopathy (AMT-Related)
- ☐ Glycine Encephalopathy (GLDC-Related)
- ☐ Glycogen Storage Disease, Type Ia ◆ ● ✠ ▼
- ☐ Glycogen Storage Disease, Type Ib
- ☐ Glycogen Storage Disease, Type II ◆ ● ■ ▼
- ☐ Glycogen Storage Disease, Type III ◆ ■ ▼
- ☐ Glycogen Storage Disease, Type IV / Adult Polyglucosan Body Disease ◆ ● ▼
- ☐ Glycogen Storage Disease, Type V ◆ ■ ▼
- ☐ Glycogen Storage Disease, Type VII ◆ ● ▼
- ☐ GRACILE Syndrome and Other BCS1L-Related Disorders ▼
- ☐ Hemochromatosis, Type 2A
- ☐ Hemochromatosis, Type 3
- ☐ Hereditary Fructose Intolerance ▼
- ☐ Hereditary Spastic Paraparesis 49 ◆ ■ ▼
- ☐ Hermansky-Pudlak Syndrome, Type 1 ▼
- ☐ Hermansky-Pudlak Syndrome, Type 3 ◆ ● ▼
- ☐ HMG-CoA Lyase Deficiency
- ☐ Holocarboxylase Synthetase Deficiency ▼
- ☐ Homocystinuria (CBS-Related) ▼
- ☐ Homocystinuria due to MTHFR Deficiency ◆ ■ ▼
- ☐ Homocystinuria, cblE Type
- ☐ Hydrolethalus Syndrome ▼
- ☐ Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome
- ☐ Hypohidrotic Ectodermal Dysplasia 1
- ☐ Hypophosphatasia ▼
- ☐ Inclusion Body Myopathy 2 ◆ ■ ▼
- ☐ Infantile Cerebral and Cerebellar Atrophy ◆ ■ ▼
- ☐ Isovaleric Acidemia ✠
- ☐ Joubert Syndrome 2 ◆ ● ✠ ▼
- ☐ Joubert Syndrome 7 / Meckel Syndrome 5 / COACH Syndrome
- ☐ Junctional Epidermolysis Bullosa (LAMA3-Related)
- ☐ Junctional Epidermolysis Bullosa (LAMB3-Related)
- ☐ Junctional Epidermolysis Bullosa (LAMC2-Related)
- ☐ Krabbe Disease ▼
- ☐ Lamellar Ichthyosis, Type 1 ▼
- ☐ Leber Congenital Amaurosis 10 and Other CEP290-Related Ciliopathies ▼
- ☐ Leber Congenital Amaurosis 13
- ☐ Leber Congenital Amaurosis 2 / Retinitis Pigmentosa 20 ◆ ■ ▼
- ☐ Leber Congenital Amaurosis 5
- ☐ Leber Congenital Amaurosis 8 / Retinitis Pigmentosa 12 / Pigmented Paravenous Chorioretinal Atrophy
- ☐ Leigh Syndrome, French-Canadian Type ▼
- ☐ Lethal Congenital Contracture Syndrome 1 / Lethal Arthrogryposis with Anterior Horn Cell Disease ▼
- ☐ Leukoencephalopathy with Vanishing White Matter
- ☐ Limb-Girdle Muscular Dystrophy, Type 2A
- ☐ Limb-Girdle Muscular Dystrophy, Type 2B ◆ ■ ▼
- ☐ Limb-Girdle Muscular Dystrophy, Type 2C
- ☐ Limb-Girdle Muscular Dystrophy, Type 2D
- ☐ Limb-Girdle Muscular Dystrophy, Type 2E
- ☐ Limb-Girdle Muscular Dystrophy, Type 2I
- ☐ Lipoamide Dehydrogenase Deficiency ◆ ● ✠ ▼
- ☐ Lipoid Adrenal Hyperplasia
- ☐ Lipoprotein Lipase Deficiency
- ☐ Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency
- ☐ Lysinuric Protein Intolerance
- ☐ Maple Syrup Urine Disease, Type 1a ✠ ▼
- ☐ Maple Syrup Urine Disease, Type 1b ◆ ● ✠ ▼
- ☐ Meckel-Gruber syndrome 1 / Bardet-Biedl Syndrome 13 ▼
- ☐ Medium Chain Acyl-CoA Dehydrogenase Deficiency ▲ ✠ ▼ ● ■ ▼
- ☐ Megalencephalic Leukoencephalopathy with Subcortical Cysts ◆ ■ ▼
- ☐ Menkes Disease
- ☐ Metachromatic Leukodystrophy ◆ ■ ▼
- ☐ 3-Methylcrotonyl-CoA Carboxylase Deficiency: (MCCC1-Related)
- ☐ 3-Methylcrotonyl-CoA Carboxylase Deficiency: (MCCC2-Related)
- ☐ 3-Methylglutaconic Aciduria, Type III / Optic Atrophy 3, with Cataract ◆ ■ ▼
- ☐ Methylmalonic Acidemia (MMAA-Related)
- ☐ Methylmalonic Acidemia (MMAB-Related)
- ☐ Methylmalonic Acidemia (MUT-Related)
- ☐ Methylmalonic Aciduria and Homocystinuria, Cobalamin C Type ❖
- ☐ Methylmalonic Aciduria and Homocystinuria, Cobalamin D Type
- ☐ Microphthalmia / Anophthalmia ◆ ■ ▼
- ☐ Mitochondrial Complex I Deficiency (ACAD9-Related)
- ☐ Mitochondrial Complex I Deficiency (NDUFAF5-Related) ◆ ● ▼
- ☐ Mitochondrial Complex I Deficiency (NDUFS6-Related) ◆ ■ ▼
- ☐ Mitochondrial DNA Depletion Syndrome 6 / Navajo Neurohepatopathy
- ☐ Mitochondrial Myopathy and Sideroblastic Anemia 1 ◆ ■ ▼
- ☐ Mucolipidosis II / IIIA ▼
- ☐ Mucolipidosis III Gamma
- ☐ Mucolipidosis IV ◆ ● ✠ ▼
- ☐ Mucopolysaccharidosis Type I ✠
- ☐ Mucopolysaccharidosis Type II
- ☐ Mucopolysaccharidosis Type IIIA
- ☐ Mucopolysaccharidosis Type IIIB
- ☐ Mucopolysaccharidosis Type IIIC
- ☐ Mucopolysaccharidosis Type IIID
- ☐ Mucopolysaccharidosis Type IVb / GM1 Gangliosidosis ▼
- ☐ Mucopolysaccharidosis type VI
- ☐ Mucopolysaccharidosis type IX
- ☐ Multiple Sulfatase Deficiency ◆ ● ▼
- ☐ Muscle-Eye-Brain Disease and Other POMGNT1-Related Congenital Muscular Dystrophy-Dystroglycanopathies ▼
- ☐ Myoneurogastrointestinal Encephalopathy ◆ ■ ▼
- ☐ Myotubular Myopathy 1
- ☐ N-Acetylglutamate Synthase Deficiency
- ☐ Nemaline Myopathy 2 ◆ ● ✠ ▼
- ☐ Nephrogenic Diabetes Insipidus, Type II
- ☐ Nephrotic Syndrome (NPHS1-Related) / Congenital Finnish Nephrosis ▼
- ☐ Nephrotic Syndrome (NPHS2-Related) / Steroid-Resistant Nephrotic Syndrome
- ☐ Neuronal Ceroid-Lipofuscinosis (CLN3-Related) ✠
- ☐ Neuronal Ceroid-Lipofuscinosis (CLN5-Related) ▼
- ☐ Neuronal Ceroid-Lipofuscinosis (CLN6-Related)
- ☐ Neuronal Ceroid-Lipofuscinosis (CLN8-Related)
- ☐ Neuronal Ceroid-Lipofuscinosis (MFSD8-Related)
- ☐ Neuronal Ceroid-Lipofuscinosis (PPT1-Related) ▼
- ☐ Neuronal Ceroid-Lipofuscinosis (TPP1-Related) ▼
- ☐ Niemann-Pick Disease A/B (SMPD1-Related) ◆ ● ✠ ▼
- ☐ Niemann-Pick Disease, Type C (NPC1-Related)
- ☐ Niemann-Pick Disease, Type C (NPC2-Related)
- ☐ Nijmegen Breakage Syndrome
- ☐ Non-Syndromic Hearing Loss (GJB2-Related) ◆ ● ▼
- ☐ Odonto-Onycho-Dermal Dysplasia / Schopf-Schulz-Passarge Syndrome
- ☐ Omenn Syndrome (RAG2-Related) ◆ ■ ▼
- ☐ Omenn Syndrome / Severe Combined Immunodeficiency, Athabaskan-Type
- ☐ Ornithine Aminotransferase Deficiency ◆ ■ ▼
- ☐ Ornithine Transcarbomylase Deficiency
- ☐ Osteopetrosis 1 ◆ ● ▼
- ☐ Pendred Syndrome ▼
- ☐ Phenylalanine Hydroxylase Deficiency ▲ ◆ ● ■ ✠ ▼
- ☐ 3-Phosphoglycerate Dehydrogenase Deficiency ◆ ● ▼
- ☐ Polycystic Kidney Disease, Autosomal Recessive ◆ ● ✠ ▼
- ☐ Polyglandular Autoimmune Syndrome, Type 1 ◆ ■ ▼
- ☐ Pontocerebellar Hypoplasia, Type 1A ◆ ● ▼
- ☐ Pontocerebellar Hypoplasia, Type 6 ◆ ■ ▼
- ☐ Primary Carnitine Deficiency ▼
- ☐ Primary Ciliary Dyskinesia (DNAH5-Related) ◆ ● ▼
- ☐ Primary Ciliary Dyskinesia (DNAI1-Related) ◆ ● ▼
- ☐ Primary Ciliary Dyskinesia (DNAI2-related) ◆ ● ▼
- ☐ Primary Hyperoxaluria, Type 1
- ☐ Primary Hyperoxaluria, Type 2
- ☐ Primary Hyperoxaluria, Type 3 ◆ ● ▼
- ☐ Progressive Cerebello-Cerebral Atrophy ◆ ■ ▼
- ☐ Progressive Familial Intrahepatic Cholestasis, Type 2
- ☐ Propionic Acidemia (PCCA-Related)
- ☐ Propionic Acidemia (PCCB-Related)
- ☐ Pycnodysostosis
- ☐ Pyruvate Dehydrogenase E1-Alpha Deficiency
- ☐ Pyruvate Dehydrogenase E1-Beta Deficiency
- ☐ 6-Pyruvoyl-Tetrahydropterin Synthase Deficiency
- ☐ Renal Tubular Acidosis and Deafness ◆ ■ ▼
- ☐ Retinitis Pigmentosa 25 ◆ ■ ▼
- ☐ Retinitis Pigmentosa 26 ◆ ■ ▼
- ☐ Retinitis Pigmentosa 28 ◆ ● ■ ▼
- ☐ Retinitis Pigmentosa 59 ◆ ● ▼
- ☐ Rhizomelic Chondrodysplasia Punctata, Type 1 ❖
- ☐ Rhizomelic Chondrodysplasia Punctata, Type 3
- ☐ Roberts Syndrome
- ☐ Salla Disease ▼
- ☐ Sandhoff Disease ▼
- ☐ Schimke Immunoosseous Dysplasia
- ☐ Segawa Syndrome
- ☐ Sjogren-Larsson Syndrome
- ☐ Smith-Lemli-Opitz Syndrome ◗ ▲ ◆ ● ■ ✠ ▼
- ☐ Spinal Muscular Atrophy (includes Enhanced SMA Testing) ◗ ▲ ◆ ● ■ ✠ ▼
- ☐ Spondylothoracic Dysostosis
- ☐ Steel Syndrome
- ☐ Stuve-Wiedemann Syndrome
- ☐ Sulfate Transporter-Related Osteochondrodysplasia ▼
- ☐ Tay-Sachs Disease ◆ ● ■ ✠ ▼
- ☐ Tyrosinemia, Type I ◆ ● ✠ ▼
- ☐ Usher Syndrome, Type IB ▼
- ☐ Usher Syndrome, Type IC ▼
- ☐ Usher Syndrome, Type ID ▼
- ☐ Usher Syndrome, Type IF ◆ ● ✠ ▼
- ☐ Usher Syndrome, Type IIA ◆ ■ ▼
- ☐ Usher Syndrome, Type III ◆ ● ✠ ▼
- ☐ Very Long Chain Acyl-CoA Dehydrogenase Deficiency ▼
- ☐ Walker-Warburg Syndrome and Other FKTN-Related Dystrophies ◆ ● ✠ ▼
- ☐ Wilson Disease ◆ ● ■ ▼
- ☐ Wolman Disease / Cholesteryl Ester Storage Disease ◆ ■ ▼
- ☐ X-Linked Juvenile Retinoschisis
- ☐ X-Linked Severe Combined Immunodeficiency
- ☐ Zellweger Syndrome Spectrum (PEX10-Related)
- ☐ Zellweger Syndrome Spectrum (PEX1-Related) ✠
- ☐ Zellweger Syndrome Spectrum (PEX2-Related) ◆ ● ▼
- ☐ Zellweger Syndrome Spectrum (PEX6-Related) ◆ ■ ▼

KEY FOR SMALLER PANELS

◗ Standard
▲ High Frequency
◆ Comprehensive Jewish
● Ashkenazi Jewish Disorders
■ Sephardi-Mizrahi Jewish Disorders
✠ ECS 39
▼ ECS 152

rev 1218

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

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