



PRENATAL TESTING 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: 212-241-0139
 Tax ID# 47-5349024/ CLIA# 33D2097541

| | | | | |
|----|---------------|------|---|---|
| SN | ACCESSION NO. | DATE | / | / |
|----|---------------|------|---|---|

| PATIENT INFORMATION | | |
|----------------------------|--|--|
| LAST NAME | FIRST NAME | |
| DATE OF BIRTH | BIOLOGICAL GENDER <input type="checkbox"/> M <input type="checkbox"/> F | PATIENT IS A DONOR <input type="checkbox"/> YES <input type="checkbox"/> NO |
| PARTNER / SPOUSE LAST NAME | PARTNER / SPOUSE FIRST NAME | |
| CLIENT MRN | PARTNER / SPOUSE DATE OF BIRTH | |
| TELEPHONE | EMAIL | |
| ADDRESS | CITY / STATE / ZIP | |

ORDERING 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 <input type="checkbox"/> Bill Clinic <input type="checkbox"/> Self Pay | | |
|--|---|------------------|
| POLICYHOLDER LAST NAME | POLICYHOLDER FIRST NAME | POLICYHOLDER DOB |
| INSURANCE CARRIER | INSURANCE ID | GROUP NO. |
| BILLING ADDRESS | | |
| OTHER HEALTH COVERAGE (IDENTIFY) | | |
| SELF-PAY: <input type="checkbox"/> Credit Card <input type="checkbox"/> 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 800-298-6470, Option 3. | | |
| SIGNATURE | DATE | / / |

INDICATIONS FOR TESTING

- ICD10 Dx CODE(S) (Required)**
- 009.511** Advanced Maternal Age, First Trimester
 - 009.512** 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
 - Other** _____
 - 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

COLLECTION DATE ___/___/___ # OF BLOOD TUBES SENT: YELLOW ___ PURPLE ___ BCT ___ GREEN ___

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

- Parental: 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

Parental Carrier Screening

- 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 PGD 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 _____

Infertility/Pregnancy Loss

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

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

Sema4 Noninvasive Prenatal Select

- Standard (chromosomes 13, 18, 21 and Fetal Sex)
- Standard Plus ("Standard" plus chromosomes 15, 16, 22 and Sex Chromosome Aneuploidies X, XXX, XXY, XYY)
- Expanded ("Standard Plus" with Microdeletions 22q11.2, 1p36, 4p16, 5p15, 15q11.2-q13, 8q24 and 11q23)

Omit fetal sex

OR

Sequenom MaterniT GENOME
Genome-wide fetal aneuploidies (singleton only).

NIPT REQUIRED CLINICAL INFORMATION

Specimen Required: Two 10 mL Whole Blood BCT Streck Tubes

Gestation Singleton Twins Triplets **G.A.:** _____ Weeks _____ Days or EDD: ___/___/___

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

Is this a repeat test for low fetal fraction? Yes No

Samples from multiple gestations will be referenced out to Sequenom for the equivalent test option (please note: sex chromosome aneuploidy testing is not available for multiple gestations)

NIPT MEDICAL INDICATIONS 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** **DS** **Tri 18/13** **AFP**

Ultrasound finding: (please specify) **O35.1XX0**

Personal/Family History: (please specify)
 Prior pregnancy with trisomy **O09.291** 1st tri **O09.292** 2nd tri **O09.293** 3rd tri
 Translocation/Inversion **Q95.0** **Q95.1**

Other high risk factor: (please specify) ICD10 code _____
No known high risk for fetal chromosomal aneuploidies (Sema4 Noninvasive Prenatal Select only)
 Z34.91 1st tri **Z34.92** 2nd tri **Z34.93** 3rd tri

Cytogenetics and Cytogenomics

- CHROMOSOME ANALYSIS**
- Chromosome Analysis (includes AFP with amniotic fluid)
 - Reflex to chromosomal microarray if chromosomes are normal
 - 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 the 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 (13 disorders)
 - 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

Prenatal Diagnostic Testing

- FGFR3 Hotspot Panel
 - reflex to sequencing if negative
 - Limb Defects Next Gen Sequencing Panel (7 genes)
 - Noonan Syndrome Next Gen Sequencing Panel (14 genes)
 - Single gene/Diagnostic testing
 - Gene: _____
 - Targeted testing: Maternal variant: _____ Paternal variant: _____
 - Biochemical testing: Tay Sachs Enzyme Sandhoff enzyme
 - 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

Expanded Carrier Screen Panel includes:

- Abetalipoproteinemia ◆ ● ▼
- Achromatopsia ▼
- Acrodermatitis Enteropathica
- Acute Infantile Liver Failure ◆ ■ ▼
- Acyl-CoA Oxidase 1 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 ▼
- Hyperomithinemia-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 (NDUFA5-Related) ◆ ● ▼
- Mitochondrial Complex I Deficiency (NDUFS6-Related) ◆ ■ ▼
- Mitochondrial DNA Depletion Syndrome 6 / Navajo Neurohepatopathy
- Mitochondrial Myopathy and Sideroblastic Anemia 1 ◆ ■ ▼
- Mucopolipidosis II / IIIA ▼
- Mucopolipidosis III Gamma
- Mucopolipidosis 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 Transcarbamoylase 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 Immunosseous Dysplasia
- Segawa Syndrome
- Sjogren-Larsson Syndrome
- Smith-Lemli-Opitz Syndrome ◆ ▲ ◆ ● ■ ◆ ✖ ▼
- Spinal Muscular Atrophy (includes Enhanced SMA Testing) ◆ ▲ ◆ ● ■ ◆ ✖ ▼
- Spondyl thoracic 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 ◆ ● ◆ ✖ ▼
- Usher Syndrome, Type IIIA ◆ ● ◆ ✖ ▼
- 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 38
- ▼ ECS 151

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