



# 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 Bill Clinic 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
  - Z02.3 Abnormal ultrasonic finding on antenatal screening of mother
  - Other \_\_\_\_\_
  - Z02.5 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 precreative management
  - Z31.440 Encounter of male for testing for genetic disease carrier for precreative management
  - Z02.89 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

Is the patient of 100% Ashkenazi Jewish descent?  YES  NO  
 If not, ethnic background: \_\_\_\_\_

Is the patient or their partner pregnant?  YES  NO  
 Is the patient currently using birth control medication?  YES  NO  
 Had the patient had previous carrier screening?  YES  NO  
 Specify: \_\_\_\_\_

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

### 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)  Additional Cell Culture:  Hold  Grow
- Includes reflex to array if no growth*  Mosaicism study
- Reflex to array if normal chromosomes

#### 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

### Verifi™ Prenatal Test

Specimen Required: One 10mL Whole Blood BCT Streck Tube (Black/Tan Top), must be filled to at least 7mL

**Gestational Age:** Weeks \_\_\_\_\_ Days \_\_\_\_\_ **Method For Determining Gestational Age:**  
 As estimated on: \_\_\_\_\_ / \_\_\_\_\_ / \_\_\_\_\_  LMP  Ultrasound  CRL  Date of Implantation  Other \_\_\_\_\_

**Test Indications (choose at least one):**

- Advanced maternal age (≥ 35 years)
- Positive serum screen
- Abnormal ultrasound
- History suggestive of increased risk for the specified chromosome aneuploidies
- Low risk/maternal anxiety
- Other \_\_\_\_\_

**ICD10 Dx Codes(s) (Required)**

- 009.291  009.522
- 009.511  028.3
- 009.512  035.1XX1
- 009.513  Q95
- 009.521

### Prenatal Diagnostic Testing

- FGFR3 Hotspot Panel  Maternal Cell Contamination
- reflex to sequencing if negative  Other: \_\_\_\_\_
- 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 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

<p>Verifi™ Prenatal Test (chromosomes 21, 18, 13)</p> <p><input type="checkbox"/> Singleton <input type="checkbox"/> Twin</p> <p>Additional option: <input type="checkbox"/> Sex chromosome aneuploidies (MX, XXX, XXY, and XYY)</p>	OR	<p>Verifi™ Plus Prenatal Test (chromosomes 21, 18, 13)</p> <p>Singleton</p> <p>Additional options:  <input type="checkbox"/> Microdeletions 1p36 deletion, 4p- (Wolf-Hirschhorn), 5p- (Cri-du-chat), 15q11.2 deletion (Prader-Willi/Angelman), 22q11.2 deletion (DiGeorge)  <input type="checkbox"/> Sex chromosome aneuploidies (MX, XXX, XXY, and XYY)  <input type="checkbox"/> All Chromosomes (including sex chromosome aneuploidies [MX, XXX, XXY, and XYY])</p>
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The Verifi™ Prenatal Test was developed by, and its performance characteristics were determined by Verinata Health, Inc. a wholly owned subsidiary of Illumina, Inc. The VHL laboratory is CAP-accredited and certified under the Clinical Laboratory Improvement Amendments (CLIA) as qualified to perform high complexity clinical laboratory testing. It has not been cleared or approved by the U.S. Food and Drug Administration. The Verifi™ Prenatal Test is validated for singleton and twin pregnancies and Verifi™ Plus Prenatal Test is validated for singleton pregnancies with gestational age of at least 10 weeks 0 days. © 2017 Illumina, Inc. All rights reserved. Illumina, and the pumpkin orange color, are trademarks of Illumina, Inc. in the U.S. and/or other countries.

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
- Arthrogyposis, 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
- Hydrolethals Syndrome ▼
- Hyperomithinemia-Hyperammonemia-Homocitrullinuria Syndrome
- Hypohidrotic Ectodermal Dysplasia 1
- Hypophosphatase ▼
- 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 Arthrogyposis 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 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)
- Niemegeen 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 I ◆●●▼
- 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) ▶▲◆●●■✱▼
- 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 ◆●●✱▼
- 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 ✱ ECS 39
- ▲ High Frequency ▼ ECS 152
- ◆ Comprehensive Jewish
- Ashkenazi Jewish Disorders
- Sephardi-Mizrahi Jewish Disorders

## 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](http://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](mailto: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](mailto: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](mailto:privacy@sema4.com)).

\_\_\_\_\_  
Signature of person being tested (or guardian)

\_\_\_\_\_  
Date