sema4
a Mount Sinai venture

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CARRIER SCREENING 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	PATIENT IS A DONOR ☐ YES ☐ NO
PARTNER / SPOUSE LAST NAME	PARTNER / SPOUSE FIRST NAME	
CLIENT MRN	PARTNER / SPOUSE DATE OF BIRTH / /	
TELEPHONE	EMAIL	
ADDRESS	CITY / STATE / ZIP	

BILLING INFORMATION ■ Institutional Bill

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., One Gustave L. Levy Place, Box 1497, New York, NY 10029

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 / /

ORDERING PROVIDER INFORMATION

NAME	GENETIC COUNSELOR
ADDRESS	CLINIC / INSTITUTION
	TELEPHONE
	FAX

PROVIDER SIGNATURE OF CONSENT (REQUIRED): I certify that this patient (and/or their legal guardian, as necessary) has been informed of the benefits, risks, and limitations of the laboratory test(s) requested. I have answered this person's questions. I have obtained a signed informed consent from this patient or their legal guardian for this testing in accordance with applicable laws and regulations, including N.Y. Civil Rights Law Section 79-L, and will retain this consent in the patient's medical record.

SIGNATURE DATE / /

INDICATIONS FOR TESTING

ICD10 Dx CODE(S) (Required if indication is not specified above)

☐ 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.
☐ Z84.81 Family history of carrier of genetic disease.
☐ Other (please specify ICD10): _____

FAMILY HISTORY OF: _____

PARTNER CARRIER OF: _____

OTHER: _____

COLLECTION DATE ____/____/____ SPECIMEN TYPE: ☐ Peripheral Blood ☐ Saliva

OF BLOOD TUBES SENT: YELLOW____ PURPLE____ GREEN____

LABORATORY TESTING INFORMATION

Are you of 100% Ashkenazi Jewish descent? ☐ YES ☐ NO If not, ethnic background: _____

Previous Carrier Screening? ☐ YES ☐ NO Specify: _____

Are you or your partner pregnant? ☐ YES ☐ NO

Currently using birth control medication or hormone replacement therapy? ☐ YES ☐ NO

LABORATORY TEST(S) ORDERED

Carrier Screening *(see reverse side for genes in each panel)*

☐ NEW Expanded Carrier Screen (283 genes) - this test replaced ECS 281

☐ 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)

☐ Single Gene _____
Variant(s)_____
☐ Phase analysis

☐ 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:

☐ Test for Microdeletions of Y Chromosome (male)
☐ Cystic Fibrosis with CFTR Intron 9 PolyT (male)
☐ Chromosome analysis (male or female)

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

CLIENT SERVICES

FFP0090G0418

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 38
- ▼ ECS 151

rev 0218

Informed Consent for Genetic Carrier Screening Testing

I, _____, hereby request genetic carrier screening testing which may include molecular and/or biochemical analyses. I have received verbal and written information (please see https://sema4genomics.com/products/test-catalog/ for specific 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 a specimen(s), such as peripheral blood or saliva will be taken from me. I understand that the samples will be used for determining if I am a carrier of a genetic variant that may affect the health of my future potential offspring. I also understand that the results of this test may inform me about my own health and my susceptibility to develop a genetic disease or medical condition.

The nature of the carrier screening options available to me—also available at https://sema4genomics.com/products/for-pregnancy/ —have been explained to me, including details regarding the accuracy of such test and its risks and limitations. Several technologies are used to perform Carrier Screening, which may include:

- High-throughput, next generation sequencing
- Sanger sequencing
- Targeted mutation analysis
- Capillary electrophoresis
- Southern blot analysis
- Multiplex Ligation-Dependent Probe Amplification
- Enzymatic analysis

Il understand that although the likelihood of an incorrect diagnosis or a misinterpretation of the result is extremely small, infrequent errors may occur. 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 carry a mutation or mutations in the genes analyzed. I understand that carrier screening includes assessment of a particular set of genes and that no determination may be made regarding other genes that are not included in this test. I understand that this testing may yield incidental findings that indicate that I am affected with one of the diseases on the panel or with another genetic abnormality.

No test will be performed on my sample other than the one(s) authorized by this consent. I hereby give consent to have my specimen tested for:

☐ The Sema4 Expanded Carrier Screen (ECS) - 283 gene panel, **OR**

☐ the following panel/genes: _____

By signing this consent form, I agree that Sema4 may store, de-identify, and use my sample and information to support medical and academic research. Specimens from residents of New York will not be retained for more than 60 days after collection and will not be included in research studies unless I consent by initialing below. I understand that I may withdraw this consent at any time and that my specimen will be promptly destroyed.

_____ *Initials*

For residents of New York only, I give consent to have my specimen anonymously used by Sema4 for scientific research related to genetic disease and stored for as long as the specimen is useful for such research purposes, not to exceed 10 years. I understand that I may withdraw this consent at any time and that my specimen will be promptly destroyed.

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). I understand that I will receive no compensation in connection with such research. If I prefer not to have any of my de-identified sample and health information used in research consistent with this consent, I may request this by contacting the laboratory.

I understand that, because of the nature of carrier screening tests, my results may be shared with my reproductive partner, and I hereby authorize Sema4 and my physician to disclose and discuss my results with my reproductive partner to the extent necessary to provide the testing and genetic counseling services that I have requested.

I understand that the Laboratory may wish to contact me in the future 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 sample(s). I understand that I may notify the Laboratory to opt out of such future contact and doing such will not affect my clinical care.

The results of my 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.

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 / Date

_____ Signature of witness / Date