

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

ACCESSION NO.	DATE / /
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PATIENT INFORMATION	
LAST NAME	FIRST NAME
DATE OF BIRTH / /	BIOLOGICAL GENDER <input type="checkbox"/> M <input type="checkbox"/> F
PARTNER / SPOUSE LAST NAME	PARTNER / SPOUSE FIRST NAME
TELEPHONE (DAY)	PARTNER / SPOUSE DATE OF BIRTH / /
TELEPHONE (NIGHT)	EMAIL
ADDRESS	CITY / STATE / ZIP

ORDERING PROVIDER INFORMATION	
NAME	GENETIC COUNSELOR
ADDRESS	CLINIC / INSTITUTION
	TELEPHONE
	FAX
<input type="checkbox"/> Patient is a donor <input type="checkbox"/> Bill Clinic <input type="checkbox"/> Self Pay	
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		
POLICYHOLDER LAST NAME	POLICYHOLDER FIRST NAME	POLICYHOLDER DOB / /
INSURANCE CARRIER	INSURANCE ID	GROUP NO.
BILLING ADDRESS		
OTHER HEALTH COVERAGE (IDENTIFY)		
ASSIGNMENT AND RELEASE: I hereby authorize my insurance benefits be paid directly to the provider and I understand that I'm financially responsible for uncovered services. I also authorize the release of any information required to process the claim.		
SIGNATURE	DATE / /	

INDICATIONS FOR TESTING	
ICD10 Dx CODE(S) (Required if indication is not specified above)	
<input type="checkbox"/> Z31.430 Encounter of female for testing for genetic disease carrier status for procreative management. <input type="checkbox"/> Z31.440 Encounter of male for testing for genetic disease carrier for procreative management. <input type="checkbox"/> Z84.81 Family history of carrier of genetic disease. <input type="checkbox"/> Other (please specify ICD10): _____	
FAMILY HISTORY OF: _____	PARTNER CARRIER OF: _____
OTHER: _____	
COLLECTION DATE / /	
# OF BLOOD TUBES SENT: YELLOW _____ PURPLE _____ GREEN _____	

LABORATORY TESTING INFORMATION			
Are you of 100% Ashkenazi Jewish descent?	<input type="checkbox"/> YES <input type="checkbox"/> NO	If not, ethnic background: _____	Are you or your partner pregnant?
Previous Carrier Screening?	<input type="checkbox"/> YES <input type="checkbox"/> NO	Specify: _____	Currently using birth control medication?
			<input type="checkbox"/> YES <input type="checkbox"/> NO

LABORATORY TEST(S) ORDERED			
<input type="checkbox"/> Expanded Carrier Screen Pan-ethnic Panel (281 genes, see reverse side for comprehensive list)		<input type="checkbox"/> Test only for AR disorders female partner screened positive for (mark on reverse side or list here):	
<input type="checkbox"/> Standard Pan-ethnic Panel (4)	<input type="checkbox"/> High Frequency Pan-ethnic Panel (10)		
<input type="checkbox"/> Cystic Fibrosis <input type="checkbox"/> Fragile X Syndrome (females only) <input type="checkbox"/> Spinal Muscular Atrophy (includes Enhanced SMA testing) <input type="checkbox"/> Smith-Lemli-Opitz-Syndrome	<input type="checkbox"/> Alpha-Thalassemia <input type="checkbox"/> Beta-Globin-Related Hemoglobinopathies <input type="checkbox"/> Congenital Disorder of Glycosylation Ia <input type="checkbox"/> Cystic Fibrosis <input type="checkbox"/> Duchenne Muscular Dystrophy / Becker Muscular Dystrophy	<input type="checkbox"/> Fragile X Syndrome (females only) <input type="checkbox"/> Medium Chain Acyl-CoA Dehydrogenase Deficiency <input type="checkbox"/> Phenylalanine Hydroxylase Deficiency	<input type="checkbox"/> Smith-Lemli-Opitz-Syndrome <input type="checkbox"/> Spinal Muscular Atrophy (includes Enhanced SMA testing) <input type="checkbox"/> Other _____
<input type="checkbox"/> Comprehensive Jewish Carrier Screen (96 genes)		<input type="checkbox"/> Single Gene _____	
<input type="checkbox"/> Ashkenazi Jewish Disorders (48+10) <input type="checkbox"/> Abetalipoproteinemia <input type="checkbox"/> Alport Syndrome (COL4A3) <input type="checkbox"/> Arthrogryposis, Mental Retardation and Seizures <input type="checkbox"/> Bardet-Biedl (BBS2) <input type="checkbox"/> Bloom Syndrome <input type="checkbox"/> Canavan Disease <input type="checkbox"/> Carnitine Palmitoyltransferase II Deficiency <input type="checkbox"/> Choreoacanthocytosis <input type="checkbox"/> Congenital Amegakaryocytic Thrombocytopenia <input type="checkbox"/> Congenital Disorder of Glycosylation Ia <input type="checkbox"/> Deafness, AR 77 <input type="checkbox"/> Dyskeratosis Congenita (RTEL1)	<input type="checkbox"/> Ehlers-Danlos VIIC <input type="checkbox"/> Enhanced S-Cone Syndrome <input type="checkbox"/> Factor XI Deficiency <input type="checkbox"/> Familial Dysautonomia <input type="checkbox"/> Familial Hypercholesterolemia <input type="checkbox"/> Familial Hyperinsulinism (ABCC8) <input type="checkbox"/> Fanconi Anemia, Group C <input type="checkbox"/> Galactosemia <input type="checkbox"/> Gaucher Disease <input type="checkbox"/> Glycogen Storage Disease Ia <input type="checkbox"/> Glycogen Storage Disease IV <input type="checkbox"/> Glycogen Storage Disease VII	<input type="checkbox"/> Hermansky-Pudlak Syndrome 3 <input type="checkbox"/> Joubert Syndrome 2 <input type="checkbox"/> Lipoamide Dehydrogenase Deficiency <input type="checkbox"/> Maple Syrup Urine Disease Ib <input type="checkbox"/> Mitochondrial Complex I Deficiency (NDUFA5) <input type="checkbox"/> Mucopolidiosis IV <input type="checkbox"/> Multiple Sulphatase Deficiency <input type="checkbox"/> Nemaline Myopathy 2 <input type="checkbox"/> Niemann-Pick Disease A & B <input type="checkbox"/> Non-Syndromic Hearing Loss (GJB2) <input type="checkbox"/> Osteopetrosis 1 <input type="checkbox"/> 3-Phosphoglycerate Dehydrogenase Deficiency	<input type="checkbox"/> Polycystic Kidney Disease, AR <input type="checkbox"/> Pontocerebellar Hypoplasia 1A <input type="checkbox"/> Primary Ciliary Dyskinesia (DNAH5) <input type="checkbox"/> Primary Ciliary Dyskinesia (DNAI1) <input type="checkbox"/> Primary Ciliary Dyskinesia (DNAI2) <input type="checkbox"/> Primary Hyperoxaluria 3 <input type="checkbox"/> Retinitis Pigmentosa 59 <input type="checkbox"/> Tyrosinemia I <input type="checkbox"/> Usher Syndrome IF <input type="checkbox"/> Usher Syndrome III <input type="checkbox"/> Walker Warburg Syndrome and other FKTN-Related Dystrophies <input type="checkbox"/> Zellweger Syndrome Spectrum (PEX2)
<input type="checkbox"/> Sephardi-Mizrahi Jewish Disorders (38+10) <input type="checkbox"/> Acute Infantile Liver Failure <input type="checkbox"/> Adrenoleukodystrophy, XL <input type="checkbox"/> Asparagine Synthetase Deficiency <input type="checkbox"/> Ataxia-Telangiectasia <input type="checkbox"/> Beta-Globin-Related Hemoglobinopathies <input type="checkbox"/> Cerebrotendinous Xanthomatosis <input type="checkbox"/> Chronic Granulomatous Disease (CYBA) <input type="checkbox"/> Congenital Insensitivity to Pain with Anhidrosis <input type="checkbox"/> Congenital Myasthenic Syndrome (RAPSN) <input type="checkbox"/> Corticosterone Methyloxidase Deficiency <input type="checkbox"/> Cystinosis	<input type="checkbox"/> Fanconi anemia, Group A <input type="checkbox"/> Glycogen Storage Disease III <input type="checkbox"/> Glycogen Storage Disease V <input type="checkbox"/> Hereditary Spastic Paraparesis 49 <input type="checkbox"/> Homocystinuria due to MTHFR Deficiency <input type="checkbox"/> Inclusion Body Myopathy 2 <input type="checkbox"/> Infantile Cerebral and Cerebellar Atrophy <input type="checkbox"/> Leber Congenital Amaurosis 2 / Retinitis pigmentosa 20 <input type="checkbox"/> Limb Girdle Muscular Dystrophy 2B	<input type="checkbox"/> Megalencephalic Leukoencephalopathy with Subcortical Cysts <input type="checkbox"/> Metachromatic Leukodystrophy <input type="checkbox"/> 3-Methylglutaconic Aciduria, III / Optic Atrophy 3, with Cataract <input type="checkbox"/> Microphthalmia / Anophthalmia <input type="checkbox"/> Mitochondrial Complex I Deficiency (NDUFS6) <input type="checkbox"/> Mitochondrial Myopathy and Sideroblastic Anemia 1 <input type="checkbox"/> Myoneurogastrointestinal Encephalopathy <input type="checkbox"/> Omenn Syndrome (RAG2) <input type="checkbox"/> Ornithine Aminotransferase Deficiency	<input type="checkbox"/> Polyglandular Autoimmune Syndrome 1 <input type="checkbox"/> Pontocerebellar Hypoplasia 6 <input type="checkbox"/> Progressive Cerebello-Cerebral Atrophy <input type="checkbox"/> Renal Tubular Acidosis and Deafness <input type="checkbox"/> Retinitis Pigmentosa 25 <input type="checkbox"/> Retinitis Pigmentosa 26 <input type="checkbox"/> Usher Syndrome IIA <input type="checkbox"/> Wolman Disease / Cholesteryl Ester Storage Disease <input type="checkbox"/> Zellweger Syndrome Spectrum (PEX6)

Diseases common to all Jewish Individuals (10): these disorders are included in any Jewish panel selected.

<input type="checkbox"/> Cystic Fibrosis	<input type="checkbox"/> Fragile X Syndrome (females only)	<input type="checkbox"/> Phenylalanine Hydroxylase Deficiency	<input type="checkbox"/> Smith-Lemli-Opitz Syndrome	<input type="checkbox"/> Tay-Sachs Disease
<input type="checkbox"/> Familial Mediterranean Fever	<input type="checkbox"/> Glycogen Storage Disease II	<input type="checkbox"/> Retinitis Pigmentosa 28	<input type="checkbox"/> Spinal Muscular Atrophy (includes Enhanced SMA testing)	<input type="checkbox"/> Wilson Disease

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

Expanded Pan-ethnic 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 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, Autosomal Recessive
- 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
- 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
- Primary Ciliary Dyskinesia (DNAH5-Related)
- Primary Ciliary Dyskinesia (DNAH2-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)

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/or written information from my physician, genetic counselor, and/or other healthcare provider that described, in words that I understood, the nature of the testing that I 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 <http://nextstest.com>—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

I 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.

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

the Expanded Pan-ethnic Carrier Screening panel (281 genes), **OR**

the following panel/genes: _____

_____ I give consent to have my specimen anonymously used by the Laboratory for scientific research related
Initial Initial to genetic disease and stored for as long as the specimen is useful for such research purposes, not to exceed 10 years, and that I will receive no compensation in connection with such research. I understand that I may withdraw this consent at any time and that my specimen will be promptly destroyed.

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. 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.

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