Weill Cornell Medical College
The Ronald O. Perelman and Claudia Cohen Center for Reproductive Medicine

NewYork-Presbyterian Hospital
Weill Cornell Medical Center

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Dear Patient,

The Center for Reproductive Medicine is committed to providing you with the best care possible.

CRM now uses **Mt. Sinai's Expanded Pan-ethnic Panel (281 disorders)** for more comprehensive genetic testing on all patients (280 for male testing). There are many disorders that are prevalent in certain ethnic backgrounds and not everyone can properly identify their ancestry. This makes appropriate ethnic-based carrier screening quite challenging. Mt. Sinai has compiled 281 genetic disorders that occur at increased frequencies in certain populations to make expanded pan-ethnic carrier screening more streamlined.

CRM Donor Egg Program requires genetic testing to be done on all male partners, directed/known sperm donors and anonymous, agency and directed/known egg donors. Since this is different genetic testing than what was offered in the past, all patients are required to do this and it cannot be waived. Please find the attached list of diseases that are tested in this panel.

If the sperm source is anonymous, it will be up to the patient to request the sperm bank to have the sperm donor tested for the same disorder(s). We will be requiring an "Acknowledgement of Risk" consent signed by the recipient in order to proceed with the anonymous sperm donor without testing should she decide to do so.

- *Carrier screening on all male partners/directed sperm donors must be completed to become active on the wait list.*
- *We cannot match you with an egg donor until we have these results because we cannot offer a donor who carries the same disorder.*
- *You will be informed of the disorder/s the donor is a carrier of; all recipients receive a genetic report on the donor.*
- *Counseling will be provided and you may proceed with the cycle if you choose to do so.*

We have a special patient price arranged with Mt. Sinai. In order to qualify for this pricing, a blood sample can be collected at CRM. It may take up to 14 days for results to be available so please have this done as soon as possible. Please call Mari Santocildes (646)-962-3447 for an appointment.

If you have any questions regarding genetic testing or have a family history of a genetic disease not listed, please contact our genetic counselor, Debra Lilienthal, at (646)-962-3434.

If you have any other questions please feel free to contact our program manager, Dee Svedberg, at (646)962-3345.

Sincerely,
Ina Cholst, M.D.
Donor Egg Program Director

Expanded Pan-ethnic Panel includes:

- ☐ Abetalipoproteinemia
- ☐ Achromatopsia
- ☐ Acrodermatitis Enteropathica
- ☐ Acute Infantile Liver Failure
- ☐ Acyl-CoA Oxidase I Deficiency
- ☐ Adenosine Deaminase Deficiency
- ☐ 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
- ☐ 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 (*DNAI1*-Related)
- ☐ Primary Ciliary Dyskinesia (*DNAH5*-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
- ☐ Adrenoleukodystrophy
- ☐ 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)