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New York, NY 10029-6574
Phone: 212-241-7518 / Fax: 212-241-0139
Tax ID# 13-6171197
CLIA# 33D0653419

PRENATAL TESTING AND GENETIC SCREENING REQUISITION

Mount Sinai Genetic Testing Laboratory
Mount Sinai Medical Center

ACCESSION NO.

DATE / /

LabForms 888-200-5114

PATIENT INFORMATION

LAST NAME | FIRST NAME

DATE OF BIRTH / / | SEX M ☐ F ☐

PARTNER / SPOUSE LAST NAME | PARTNER / SPOUSE FIRST NAME

TELEPHONE (HOME) | TELEPHONE (CELL) | TELEPHONE (WORK)

ADDRESS

CITY / STATE / ZIP

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 am financially responsible for uncovered services. I also authorize the release of any information required to process the claim.

SIGNATURE DATE / /

REFERRING PHYSICIAN INFORMATION

PHYSICIAN SIGNATURE OF CONSENT REQUIRED BELOW: I certify that the patient specified above and/or their legal guardian 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 informed consent from the patient or their legal guardian for this testing.

SIGNATURE DATE (MM/DD/YY) / /

INDICATIONS FOR TESTING

☐ GENERAL CARRIER SCREENING (NO FAMILY HISTORY)
☐ FAMILY HISTORY OF _____ ☐ PARTNER CARRIER OF:_____
☐ ADVANCED MATERNAL AGE ☐ ULTRASOUND FINDING_____
☐ POSITIVE PRENATAL ANEUPLOIDY SCREEN _____
☐ OTHER _____

ICD9 Dx CODE(S) (Required if indication is not specified above)

COLLECTION DATE: _____ / _____ / _____

OF BLOOD TUBES SENT: YELLOW_____ PURPLE_____ RED_____ GREEN_____

LABORATORY TEST(S) ORDERED

Cytogenetics and Cytogenomics

Chromosome Analysis

☐ Amniotic Fluid + AFP **G.A.**_____
☐ By LMP ☐ By Ultrasound **Date**_____
☐ CVS
☐ Peripheral Blood
☐ Peripheral Blood Mosaicism Study (50 Cells)
☐ Peripheral Blood STAT (newborn blood includes Aneuploidy FISH)
☐ Fetal Blood (PUBS)
☐ Skin Biopsy
☐ Products of Conception (In case of growth failure, reflex to P.O.C. FISH Panel is included)

Additional Cell Culture

☐ Hold ☐ Grow_____

☐ Aneuploidy FISH (prenatal specimens)
☐ Single Microdeletion FISH
Please specify disease: _____
☐ Microdeletion FISH Panel
☐ Subtelomere FISH - single probe only
☐ 180K Array CGH + SNPs (pre-/postnatal)
☐ FISH STAT
Please specify disease:_____

Molecular

☐ FGFR3 Hotspot Panel
☐ reflex to sequencing if negative
☐ Limb Defects Next Gen Sequencing Panel (7 genes)
☐ Noonan Syndrome Next Gen Sequencing Panel (14 genes)
☐ Maternal Cell Contamination
☐ Other: _____

☐ MaterniT21 PLUS*
MaterniT21 PLUS is a trademark of Sequenom

Specimen Required:
Two 10 ML Whole Blood BCT Streck Tubes (Black/Tan Top)
***Please Call 212-241-7518 for Supplies and Specimen Pickup**

SCMM ID #: _____
☐ Opt-out for subchromosomal copy variants (microdeletions), chromosomes 22 and 16

Ordering Physician_____ Fax #: _____
Referring Physician _____ Fax #: _____

Gestational Age:_____ **Method For Determining Gestational Age**:
☐ LMP ☐ Ultrasound

Patient Height: _____ **Weight:** _____

Increased Risk due to (please check one)	If Multifetal Gestation (please check one)	ICD9 Dx CODE(S) (Required)
☐ Advanced Maternal Age	☐ Twins	☐ 659.63 ☐ 796.5
☐ Serum Biochemical Screening	☐ Triplets	☐ 659.53 ☐ Other:
☐ Ultrasound Finding		☐ 655.13 _____
☐ Personal or Family History		

Limitations of the MaterniT21 PLUS Test
DNA test results do not provide a definitive genetic risk in all individuals. Cell-free fetal DNA does not replace the accuracy and precision of prenatal diagnosis with CVS or amniocentesis. A patient with a positive test result should be referred for genetic counseling and offered invasive prenatal diagnosis for confirmation of test results. A negative test result does not ensure an unaffected pregnancy. While results of this testing are highly accurate, not all chromosomal abnormalities may be detected due to placental, maternal or fetal mosaicism, or other causes. Sex chromosomal aneuploidies will not be reported for multiple gestations.

Molecular - Carrier Screening Panels

☐ Basic Pan-Ethnic (CF, Fragile X, SMA and SLOS)
☐ Expanded Ashkenazi Jewish (38 diseases individually listed below)
☐ NEW Ashkenazi Jewish (18 diseases)[E]
☐ Mt Sinai – Counsyl Expanded Pan-Ethnic (73 additional diseases only, listed on back)
☐ All-inclusive Pan-Ethnic (includes all 111 disorders above)

Molecular - Individual Tests

☐ Abetalipoproteinemia[E]
☐ Alport Syndrome, AR[E]
☐ Arthrogryposis, Mental Retardation & Seizures[E]
☐ Bardet-Biedl (BBS2)[E]
☐ Bloom Syndrome
☐ Canavan Disease
☐ Carnitine Palmitoyltransferase II Deficiency[E]
☐ Congenital Amegakaryocytic Thrombocytopenia[E]
☐ Congenital Disorder of Glycosylation Ia[E]
☐ Cystic Fibrosis (CF)
☐ Dyskeratosis Congenita, AR[E]
☐ Ehlers-Danlos VIIC[E]
☐ Familial Dysautonomia
☐ Familial Hyperinsulinism (*ABCC8*)
☐ Fanconi Anemia C
☐ Fragile X Syndrome (females only)
☐ Galactosemia[E]
☐ Gaucher Disease
☐ Glycogen Storage Disease Ia
☐ Joubert syndrome 2
☐ Lipoamide Dehydrogenase Deficiency (E3)
☐ Maple Syrup Urine Disease Ib
☐ Mucolipidosis IV
☐ Multiple Sulphatase Deficiency[E]
☐ Nemaline Myopathy (*NEB*)
☐ Niemann-Pick Disease A and B
☐ 3-Phosphoglycerate Dehydrogenase Deficiency[E]
☐ Polycystic Kidney Disease, AR[E]
☐ Retinitis Pigmentosa 59[E]
☐ Smith-Lemli-Opitz (SLOS)[E]
☐ Spinal Muscular Atrophy (SMA) (includes Enhanced SMA Testing)*
☐ Tay-Sachs Disease
☐ Tyrosinemia I[E]
☐ Usher IF
☐ Usher III
☐ Walker-Warburg (*FKTN*)
☐ Wilson Disease[E]
☐ Zellweger Syndrome (*PEX2*)[E]
☐ Other: _____

*Enhanced SMA testing includes analysis for presence/absence of g.27134T>G to identify (2+0) silent carriers.

LABORATORY TESTING INFORMATION

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

Are you or your partner pregnant? ☐ YES ☐ NO
Currently using birth control medication? ☐ YES ☐ NO

Previous Carrier Screening? ☐ YES ☐ NO
Specify:_____

LAB USE ONLY

We accept VISA, MasterCard, AMEX and personal checks.
Make checks payable to: Mt. Sinai Genetic Testing Lab

For billing questions, call: 212-241-8717

TOTAL CHARGES:
AMOUNT PAID:
BALANCE DUE:

Informed Consent for Genetic Testing

I, _____, hereby request genetic testing for me/or my child (name of child if applicable) _____, which may include molecular, cytogenetic and/or biochemical analyses. I have received verbal and/or written information from my physician or from a genetic counselor that described, in words that I understood, the nature of the genetic testing that I/or my child am about to undergo.

I understand that a specimen(s), such as peripheral blood, dried blood spot, skin biopsy, amniotic fluid, chorionic villi and/or urine sample will be taken from me/or my child. I understand that the samples will be used for determining if I/or my child have a genetic disease, are carriers of a genetic disease or are more susceptible to develop a genetic disease.

The nature of the genetic testing for (disease name) _____ _____ _____ has been explained to me and the accuracy of the test and its limitations have been detailed. 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%.

No test will be performed on my sample other than the one(s) authorized by my doctor.

I give consent to have my specimen be used anonymously by the laboratory for the purposes of quality control or for research related to genetic disease. Please check the box below to consent. If you do not consent your sample will be discarded within 60 days of completion of the testing.

☐ I agree to have my sample used anonymously for research by the laboratory._____

Initials

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 in this study are not as I have stated and this test may detect non-paternity.

The results of my/or 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 clinical 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 also 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 (or guardian) Date

_____ _____

Witness Date

Mt Sinai-Counsyl Expanded Pan-Ethnic Panel includes:

Achromatopsia
Alkaptonuria
Alpha-Mannosidosis
Andermann Syndrome
ARSACS
Aspartylglycosaminuria
Ataxia With Vitamin E Deficiency
Ataxia-Telangiectasia
Bardet-Biedl Syndrome, BSS10-related
Bardet-Biedl Syndrome, BSS1-related
Beta Hemoglobinopathies, Hemoglobins C, S, etc
Beta Thalassemia
Biotinidase Deficiency
Carnitine Palmitoyltransferase IA Deficiency
Cartilage-Hair Hypoplasia
Choroideremia
Citrullinemia Type 1
CLN3-related Neuronal Ceroid Lipofuscinosis
CLN5-related Neuronal Ceroid Lipofuscinosis
Cohen Syndrome
Congenital Disorder of Glycosylation Type Ib
Congenital Finnish Nephrosis
Costeff Optic Atrophy Syndrome
Cystinosis
D-Bifunctional Protein Deficiency
Familial Mediterranean Fever
GJB2-related DFNB1 Nonsyndromic Hearing Loss and Deafness
Glutaric Acidemia Type I
Glycogen Storage Disease Type Ib
Glycogen Storage Disease Type III
Glycogen Storage Disease Type V
GRACILE Syndrome
Hereditary Fructose Intolerance
Hereditary Thymine-Uraciluria
Herlitz Junctional Epidermolysis Bullosa, LAMA3-related
Herlitz Junctional Epidermolysis Bullosa, LAMB3-related
Herlitz Junctional Epidermolysis Bullosa, LAMC2-related
Homocystinuria, CBS-related
Hurler Syndrome
Hypophosphatasia, Autosomal Recessive
Inclusion Body Myopathy 2
Isovaleric Acidemia
Krabbe Disease
Limb-Girdle Muscular Dystrophy Type 2D
Limb-Girdle Muscular Dystrophy Type 2E
Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency (LCHAD)
Medium Chain Acyl-CoA Dehydrogenase Deficiency
Megalencephalic Leukoencephalopathy With Subcortical Cysts
Metachromatic Leukodystrophy
Muscle-Eye-Brain Disease
Neuronal Ceroid-Lipofuscinosis, PPT1-related
Neuronal Ceroid-Lipofuscinosis, TPP1-related
Niemann-Pick Disease Type C, NPC1-related
Nijmegen Breakage Syndrome
Northern Epilepsy
Pendred Syndrome
Phenylalanine Hydroxylase Deficiency (including PKU)
Polyglandular Autoimmune Syndrome Type 1
Pompe Disease
Primary Carnitine Deficiency
Primary Hyperoxaluria Type 1
Primary Hyperoxaluria Type 2
PROP1-related Combined Pituitary Hormone Deficiency
Pycnodysostosis
Rhizomelic Chondrodysplasia Punctata Type 1
Salla Disease
Segawa Syndrome
Sjogren-Larsson Syndrome
Steroid-Resistant Nephrotic Syndrome
Sulfate Transporter-Related Osteochondrodysplasia
Very Long Chain Acyl-CoA Dehydrogenase Deficiency
X-Linked Juvenile Retinoschisis
Zellweger Syndrome Spectrum, PEX1-related