



GENERAL TEST 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 / /
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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
CLIENT MRN	PARTNER / SPOUSE DATE OF BIRTH / /
TELEPHONE	EMAIL
ADDRESS	CITY / STATE / ZIP

REFERRING 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

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		

CLINICAL INDICATIONS

SPECIMEN TYPE <input type="checkbox"/> AMNIOTIC FLUID <input type="checkbox"/> BLOOD <input type="checkbox"/> CVS <input type="checkbox"/> DBS <input type="checkbox"/> PLASMA <input type="checkbox"/> URINE <input type="checkbox"/> OTHER _____ <input type="checkbox"/> CULTURED CELLS TYPE _____	DATE / TIME SPECIMEN DRAWN AM PM / / DATE SPECIMEN SENT / / <input type="checkbox"/> LMP / / GESTATIONAL AGE ON SONO
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INDICATIONS FOR TEST _____
Is the patient pregnant? Yes No Currently using birth control medication? Yes No

ICD10 Dx CODE(S)

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 800-298-6570, Option 3.
SIGNATURE _____ DATE / /

LABORATORY TEST(S) ORDERED

Cytogenetics and Cytogenomics

- Chromosome Analysis**
- Amniotic Fluid + AFP
 - Skin Biopsy
 - CVS
 - Peripheral Blood
 - Mosaicism Study
 - Peripheral Blood STAT (newborn blood, includes Aneuploidy FISH)
 - Additional Cell Culture: Hold Grow _____
- Fetal blood (PUBs)
- Products of Conception
- Reflex to aCGH if chromosomes are normal
- Other Tissue
- Fluorescent in situ Hybridization (FISH)**
- Aneuploidy FISH (prenatal specimens)
 - Microdeletion FISH Panel (individually or as a panel)
 - 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)
- Subtelomere FISH – single probe only: _____
- FISH for Kallman Syndrome
- FISH for STS Deficiency
- FISH for SRY deletion

Array Comparative Genomic Hybridization (aCGH) 180K + SNP

- For prenatal specimens, please submit maternal blood for MCC*
- Prenatal Chromosomal Microarray
 - High Resolution Chromosomal Microarray (CV, AF, PB or POC)
- Please include blood specimen from parents of the proband if available
- included _____ mother _____ father
- Other: _____

NEW 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.

Pharmacogenetic Tests

- Comprehensive PGx Panel
 - Cardiovascular PGx Panel
 - Psychiatry PGx Panel
 - Pain PGx Panel
- Oncology PGx Panel
- Pediatric PGx Panel
- Targeted PGx Testing: gene(s): _____

Molecular

- For all testing related to Carrier Screening and Natalis, please refer to our test-specific requisition forms.*
- Genotyping and Targeted Analysis**
- Chitotriosidase
 - Chronic Kidney Disease *APOL 1* Genotyping (African American)
 - FGFR3* Hotspot Panel
 - Reflex to sequencing if negative
 - Fragile X syndrome
 - Maternal Cell Contamination Studies
 - Spinal Muscular Atrophy
 - Y Microdeletion
- Sanger Sequencing**
- Hypophosphatasia (*ALPL*)
 - Porphyria DNA Testing
 - Gene Sequencing
 - Known Mutation Analysis
 - Acute Porphyria Panel (AIP, HCP & VP)
 - Acute Intermittent Porphyria (AIP)
 - Hereditary Coproporphyrin (HCP)
 - Variegated Porphyria (VP)
 - Congenital Erythropoietic Porphyria (CEP)
 - Lysosomal Acid Lipase (*LIPA*) (Wolman Disease/Cholesteryl Ester Storage Disease)
 - Familial Porphyria Cutanea Tarda (PCT)
 - Erythropoietic Protoporphyrin (EPP)
 - PEPT2 (*SLC15A2*) Genotyping for Acute Porphyria Patients
- Targeted Testing: gene _____ variant _____

Next Generation Sequencing and Del/Dup Analysis

- Single Gene Diagnostic Testing
 - Cystic Fibrosis (*CFTR*)
 - Sandhoff Disease (*HEXB*)
 - Tay-Sachs Disease (*HEXA*)
 - Other: _____
- NGS Del/Dup
- NGS Del/Dup
- NGS Del/Dup
- NGS Del/Dup
- NGS Del/Dup
- NGS Del/Dup
- Autism Panel (30 genes)
- Limb Defects Panel (7 genes)
- Microcephaly Panel (78 genes)
- Noonan Syndrome Panel (14 genes)
- Whole Exome Sequencing
- Add Ultra-High Res. Med. Exome Del/Dup Analysis

Hearing and Vision Loss Panels

- (please refer to our test-specific requisition forms and informational brochure)*
- Comprehensive Hearing and Vision Loss (308 genes)
 - Comprehensive Hearing Loss (92 genes)
 - Comprehensive Vision Loss (250 genes)

Craniosynostoses

- Please inquire regarding which exons are tested & which genes are analyzed on a reflex basis*
- Antley-Bixler syndrome (*FGFR2*)
 - Apert syndrome (*FGFR2*)
 - Beare-Stevenson Syndrome (*FGFR2*)
 - Carpenter Syndrome (*RAB23*)
 - Craniofrontonasal Syndrome (CFNS) (*EFNB1*)
 - Craniosynostosis, Boston Type (CRS2) (*MSX2*)
 - Craniosynostosis with Radial Defects (*TWIST1, REC QL4*)
 - Crouzon Syndrome (*FGFR2, FGFR3*)
 - Crouzon and Acanthosis Syndrome, (Crouzonodermoskeletal Syndrome) (*FGFR3*)
 - Jackson-Weiss Syndrome (*FGFR2, FGFR3*)
 - Non-Syndromic Coronal Syndrome (*FGFR2, FGFR3*)
 - Muenke Syndrome (*FGFR3*)
 - Pfeiffer Syndrome (*FGFR1, FGFR2, FGFR3*)
 - POR Deficiency (*POR*)
 - Saethre-Chotzen Syndrome (*SC2*) (*TWIST1, FGFR2, FGFR3*)

Biochemical - Analyte Tests

- Amino Acids Full Panel, Plasma/Urine/CSF
- Phenylalanine/Tyrosine, DBS
- Amino Acids Selective Panel (PKU/MSUD), Plasma
- Acylcarnitine Profile, Plasma/DBS
- Carnitine, Plasma/Urine
- Organic Acids Profile, Urine
- Orotic Acid, Urine
- Methylmalonic Acid, Plasma/Urine
- Succinylacetone, Urine
- Quantitative Glycosaminoglycans, Urine (chondroitin, dermatan, and heparan sulfates)
- Aminolevulinic Acid and Porphobilinogen, Urine/Plasma
- Quantitative Keratan Sulfate, Urine
- Hexosaminidase A (Tay-Sachs Disease), WBC/Serum
- Hexosaminidase B (Sandhoff Disease), WBC/Serum
- Acid-β-Glucosidase (Gaucher Disease), WBC
- Chitotriosidase (Gaucher Biomarker), Plasma
- α-Galactosidase A (Fabry Disease), WBC/Plasma
- Lysosomal Acid Lipase, WBC (Wolman Disease/Cholesteryl Ester Storage Disease)
- α-L-Iduronidase (MPS-I), WBC
- α-Glucosidase (Pompe Disease), WBC
- Acid Sphingomyelinase (Niemann-Pick A/B), WBC
- β-Galactocerebrosidase (Krabbe Disease), WBC

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Patient Name: _____

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Patient Name: _____

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Patient Name: _____

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Patient Name: _____

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 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.

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.

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).

Signature of person being tested (or guardian)

Date