# PEDIATRICIAN NATALIS TEST REQUISITION

sema4
a Mount Sinai venture

50000

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 | |
|---|---|---|
| CLIENT MRN | DATE OF BIRTH / / | BIOLOGICAL SEX ☐ M ☐ F |

## LEGAL GUARDIAN INFORMATION

| LAST NAME | FIRST NAME |
|---|---|
| TELEPHONE | |
| ADDRESS | |
| PHONE | EMAIL |

***Select patient pay or institutional billing below. For patient pay orders, Sema4 will send the guardian an email to complete payment online after our lab receives the specimen(s).***

☐ Patient pay ☐ Institutional billing

**Guardian Payment Agreement:** I understand that I am financially responsible for these service and that my insurance will not be billed. I will contact Sema4 with any billing inquiries at 800-298-6470, option 3.

**SIGNATURE** **DATE** / /

## 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** / /

## PATIENT SPECIMEN INFORMATION

SPECIMEN TYPE

☐ BLOOD ☐ SALIVA ☐ CHEEK SWAB **(preferred)** ☐ OTHER _____

DATE OF COLLECTION:

INDICATION FOR TEST:

**ICD10 Dx CODE(S)**

☐ Z13.9: Encounter for screening, unspecified
☐ P09: Abnormal findings on neonatal screening
☐ Z13.228: Encounter for screening for other metabolic disorders
☐ Other (please specify ICD10): _____

## PARENTAL SPECIMEN INFORMATION

Please also provide specimen for one biological parent

☐ Biological Mother ☐ Biological Father ☐ Not available

SPECIMEN TYPE

☐ BLOOD ☐ SALIVA ☐ CHEEK SWAB **(preferred)** ☐ OTHER _____

DATE OF COLLECTION:

## LABORATORY TEST(S) ORDERED

### Molecular

☐ NEW **Natalis**
Includes (individual genes listed on reverse side):
166 Disease genes, 10 Pharmacogenetic genes

**Please Note:** Consent must be collected on all individuals who are submitting a sample to Sema4. Please complete Page 3 for Patient consent and page 4 for Biological Parent consent.

## SEMA4 NATALIS FAMILY MEDICAL HISTORY QUESTIONNAIRE

**1. What is the ethnicity of the child?**
*(Please check all that apply.)*

☐ African American
☐ Ashkenazi Jewish
☐ Caucasian
☐ Cajun/French Canadian
☐ East Asian
☐ Hispanic Caribbean
☐ Mizrahi Jewish
☐ North African
☐ North/Central American
☐ Sephardic Jewish
☐ South American
☐ South Asian
☐ Sub-Saharan African
☐ Other: _____

**2. Were there any abnormal prenatal testing results during the pregnancy?** *(Please check all that apply.)*

☐ Abnormal ultrasound
☐ Fetal AFP analysis
☐ Fetal chromosome analysis
☐ Fetal chromosomal microarray
☐ Maternal serum screening (First Trimester Screening, AFP, Quadruple Screen, etc.)
☐ Noninvasive prenatal testing (NIPT)
☐ None
☐ Not sure

**3. Do the biological parents/relatives have any of the conditions tested in this screening?**

☐ Yes If yes, what is the condition(s) name(s)?
_____
Who is the family member with the condition(s)?
_____
☐ No

**4. What is the primary reason for pursuing testing?**

☐ The family is interested in additional screening for their healthy child
☐ The child is suspected of having an inherited condition and seeking a diagnosis.

**5. Has the child had a bone marrow transplant or a stem cell transplant?**

☐ Yes
☐ No

**6. Has the child had routine state newborn screening?**

☐ Yes
☐ No
☐ Unsure

**7. Is the child generally healthy with no major physical medical problems and not currently under the care of a medical specialist with exception of optometrist, dentist, and allergist, or specialist associated with traumatic injury?**

☐ Yes
☐ No

**8. Is the child currently receiving any medications other than antibiotics?**

☐ Yes
☐ No

| 50000 | 50000 | 50000 | 50000 |
|---|---|---|---|
| Patient Name: _____ | Patient Name: _____ | Patient Name: _____ | Patient Name: _____ |

# GENES INCLUDED ON NATALIS

| DISEASE GENES | |
|---|---|
| ABCC8 | Familial Hyperinsulinism (ABCC8-Related) |
| ABCD1 | Adrenoleukodystrophy, X-Linked |
| ACADM | Medium Chain Acyl-CoA Dehydrogenase Deficiency |
| ACADVL | Very Long Chain Acyl-CoA Dehydrogenase Deficiency |
| ACAT1 | Beta-Ketothiolase Deficiency |
| ADA | Adenosine Deaminase Deficiency |
| AGL | Glycogen Storage Disease, Type III |
| AGXT | Primary Hyperoxaluria, Type 1 |
| AKR1D1 | Congenital Bile Acid Synthesis Defect (AKR1D1-Related) |
| ALDH7A1 | Pyridoxine-Dependent Epilepsy |
| ALDOB | Hereditary Fructose Intolerance |
| ALPL | Hypophosphatasia |
| ANK1 | Spherocytosis, Type 1 |
| AQP2 | Nephrogenic Diabetes Insipidus, Type II |
| ARG1 | Argininemia |
| ARSA | Metachromatic Leukodystrophy |
| ARSB | Mucopolysaccharidosis Type VI |
| ASL | Argininosuccinic Aciduria |
| ASS1 | Citrullinemia, Type 1 |
| AVPR2 | Nephrogenic Diabetes Insipidus (AVPR2-Related) / Nephrogenic Syndrome of Inappropriate Antidiuresis |
| BCKDHA | Maple Syrup Urine Disease, Type 1a |
| BCKDHB | Maple Syrup Urine Disease, Type 1b |
| BTD | Biotinidase Deficiency |
| CASR | Neonatal Hyperparathyroidism / Autosomal Dominant Hypocalcemia |
| CBS | Homocystinuria (CBS-Related) |
| CD3D | Immunodeficiency 19 |
| CD3E | Immunodeficiency 18 |
| CFTR | Cystic Fibrosis |
| COL4A3 | Alport Syndrome (COL4A3-Related) |
| COL4A4 | Alport Syndrome (COL4A4-Related) |
| COL4A5 | Alport Syndrome (COL4A5-Related) |
| CPS1 | Carbamoylphosphate Synthetase I Deficiency |
| CPT1A | Carnitine Palmitoyltransferase IA Deficiency |
| CPT2 | Carnitine Palmitoyltransferase II Deficiency |
| CTNS | Cystinosis |
| CYBA | Chronic Granulomatous Disease (CYBA-related) |
| CYBB | Chronic Granulomatous Disease (CYBB-related) |
| CYP11B1 | Congenital Adrenal Hyperplasia due to 11-Beta-Hydroxylase Deficiency |
| CYP11B2 | Corticosterone Methyloxidase Deficiency |
| CYP27A1 | Cerebrotendinous Xanthomatosis |
| DBT | Maple Syrup Urine Disease, Type 2 |
| DCLRE1C | Omenn Syndrome / Severe Combined Immunodeficiency, Athabaskan-Type |
| DLD | Lipoamide Dehydrogenase Deficiency |
| DUOX2 | Thyroid Dyshormonogenesis 6 |
| DUOXA2 | Thyroid Dyshormonogenesis 5 |
| EPB42 | Spherocytosis, Type 5 |
| ETFA | Glutaric Acidemia, Type IIa |
| ETFB | Glutaric Acidemia, Type IIb |
| ETFDH | Glutaric Acidemia, Type IIc |
| ETHE1 | Ethylmalonic Encephalopathy |
| F9 | Factor IX Deficiency |
| FAH | Tyrosinemia, Type I |
| FBN1 | Marfan syndrome and other FBN1-related disorders |
| FBP1 | Fructose-1,6-Bisphosphatase Deficiency |
| FOLR1 | Neurodegeneration due to Cerebral Folate Transport Deficiency |
| G6PC | Glycogen Storage Disease, Type Ia |
| G6PD | Hemolytic Anemia (G6PD-Related) |
| GAA | Glycogen Storage Disease, Type II |
| GALE | Galactose Epimerase Deficiency |
| GALK1 | Galactokinase Deficiency |
| GALNS | Mucopolysaccharidosis Type IVa |
| GALT | Galactosemia |
| GAMT | Cerebral Creatine Deficiency Syndrome 2 |
| GATM | Cerebral Creatine Deficiency Syndrome 3 |
| GCDH | Glutaric Acidemia, Type I |
| GCH1 | Dopa-Responsive Dystonia / BH4-Deficient Hyperphenylalaninemia B |
| GLA | Fabry Disease |
| GLUD1 | Hyperinsulinism-Hyperammonemia Syndrome |
| GRHPR | Primary Hyperoxaluria, Type 2 |
| GSS | Glutathione Synthetase Deficiency |
| GYS2 | Glycogen storage disease, Type 0 |
| HADH | Familial Hyperinsulinemic Hypoglycemia 4 / 3-Hydroxyacyl-CoA Dehydrogenase Deficiency |
| HADHA | Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase Deficiency |
| HADHB | Mitochondrial Trifunctional Protein Deficiency (HADHB-Related) |
| HAX1 | Congenital Neutropenia (HAX1-Related) |
| HBA1/HBA2 | Alpha-Thalassemia (copy number) |
| HBB | Beta-Globin-Related Hemoglobinopathies |
| HLCS | Holocarboxylase Synthetase Deficiency |
| HMGCL | HMG-CoA Lyase Deficiency |
| HMGCS2 | HMG-CoA Synthase 2 Deficiency |
| HOGA1 | Primary Hyperoxaluria, Type 3 |
| HPD | Tyrosinemia, type III |
| HSD3B2 | 3-Beta-Hydroxysteroid Dehydrogenase Type II Deficiency |
| HSD3B7 | Congenital Bile Acid Synthesis Defect (HSD3B7-Related) |
| IDS | Mucopolysaccharidosis Type II |
| IDUA | Mucopolysaccharidosis Type I |
| IGSF1 | Central Hypothyroidism and Testicular Enlargement |
| IL2RG | X-Linked Severe Combined Immunodeficiency |
| IL7R | Severe Combined Immunodeficiency (IL7R-Related) |
| INS | Permanent Neonatal Diabetes Mellitus (INS-Related) |
| IVD | Isovaleric Acidemia |
| IYD | Thyroid Dyshormonogenesis 4 |
| JAG1 | Alagille syndrome 1 / Tetralogy of Fallot |
| JAK3 | Severe Combined Immunodeficiency (JAK3-Related) |
| KCNJ11 | Familial Hyperinsulinism (KCNJ11-Related) |
| KCNQ2 | Early Infantile Epileptic Encephalopathy 7 / Benign Neonatal Seizures 1 |
| LDLR | Familial Hypercholesterolemia |
| LHX3 | Combined Pituitary Hormone Deficiency 3 |
| LIPA | Wolman Disease / Cholesteryl Ester Storage Disease |
| LMBRD1 | Methylmalonic Aciduria and Homocystinuria, Cobalamin F Type |
| LPL | Lipoprotein Lipase Deficiency |
| MAT1A | Methionine Adenosyltransferase I/III Deficiency |
| MCCC1 | 3-Methylcrotonyl-CoA Carboxylase Deficiency (MCCC1-Related) |
| MCCC2 | 3-Methylcrotonyl-CoA Carboxylase Deficiency (MCCC2-Related) |
| MCEE | Methylmalonyl-CoA Epimerase Deficiency |
| MLYCD | Malonyl-CoA Decarboxylase Deficiency |
| MMAA | Methylmalonic Acidemia (MMAA-Related) |
| MMAB | Methylmalonic Acidemia (MMAB-Related) |
| MMACHC | Methylmalonic Aciduria and Homocystinuria, Cobalamin C Type |
| MMADHC | Methylmalonic Aciduria and Homocystinuria, Cobalamin D Type |
| MPI | Congenital Disorder of Glycosylation, Type Ib |
| MPL | Congenital Amegakaryocytic Thrombocytopenia |
| MTR | Homocystinuria-Megaloblastic Anemia, Cobalamin G Type |
| MTRR | Homocystinuria, Cobalamin E Type |
| MTTP | Abetalipoproteinemia |
| MUT | Methylmalonic Acidemia (MUT-Related) |
| NAGS | N-Acetylglutamate Synthase Deficiency |
| OAT | Ornithine Aminotransferase Deficiency |
| OTC | Ornithine Transcarbomylase Deficiency |
| PAH | Phenylalanine Hydroxylase Deficiency |
| PAX8 | Congenital Hypothyroidism due to Thyroid Dysgenesis or Hypoplasia |
| PCBD1 | BH4-deficient Hyperphenylalaninemia D |
| PCCA | Propionic Acidemia (PCCA-Related) |
| PCCB | Propionic Acidemia (PCCB-Related) |
| PHGDH | 3-Phosphoglycerate Dehydrogenase Deficiency |
| PHKB | Glycogen Storage Disease, Type IXb |
| PNPO | Pyridoxamine 5'-Phosphate Oxidase Deficiency |
| POU1F1 | Combined Pituitary Hormone Deficiency 1 |
| PROP1 | Combined Pituitary Hormone Deficiency 2 |
| PRRT2 | Familial Infantile Convulsions with Paroxysmal Choreoathetosis |
| PTPRC | Severe Combined Immunodeficiency (PTPRC-Related, CD45) |
| PTS | 6-Pyruvoyl-Tetrahydropterin Synthase Deficiency |
| PYGL | Glycogen storage disease, Type VI |
| QDPR | BH4-deficient Hyperphenylalaninemia C |
| RAG1 | Omenn syndrome and other RAG1-related disorders |
| RAG2 | Omenn Syndrome (RAG2-Related) |
| RB1 | Retinoblastoma |
| SCN2A | Early Infantile Epileptic Encephalopathy 11 / Benign Familial Infantile Seizures 3 |
| SCN8A | Early Infantile Epileptic Encephalopathy 13 / Benign Familial Infantile Seizures 5 |
| SLC22A5 | Primary Carnitine Deficiency |
| SLC25A13 | Citrin Deficiency |
| SLC25A15 | Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome |
| SLC25A20 | Carnitine Acylcarnitine Translocase Deficiency |
| SLC2A1 | Glucose transporter 1 deficiency syndrome and other SLC2A1-related disorders |
| SLC39A4 | Acrodermatitis Enteropathica |
| SLC4A1 | Distal Renal Tubular Acidosis / Spherocytosis, Type 4 |
| SLC5A5 | Thyroid Dyshormonogenesis 1 |
| SLC7A7 | Lysinuric Protein Intolerance |
| SMN1 | Spinal Muscular Atrophy (copy number) |
| SMPD1 | Niemann-Pick Disease (SMPD1-Related) |
| SPR | Sepiapterin Reductase Deficiency |
| STAR | Lipoid Adrenal Hyperplasia |
| TAT | Tyrosinemia, Type II |
| TAZ | Barth Syndrome |
| TCIRG1 | Osteopetrosis 1 |
| TG | Thyroid Dyshormonogenesis 3 |
| TH | Segawa Syndrome |
| THRA | Congenital Nongoitrous Hypothryoidism 6 |
| TPO | Thyroid Dyshormonogenesis 2A |
| TRHR | Generalized Thyrotropin-Releasing Hormone Resistance |
| TRMU | Acute Infantile Liver Failure |
| TSHB | Congenital Nongoitrous Hypothryoidism 4 |
| TSHR | Congenital Nongoitrous Hypothryoidism 1 / Nonautoimmune Hyperthyroidism |
| TTPA | Ataxia With Isolated Vitamin E Deficiency |
| UGT1A1 | Crigler-Najjar Syndrome, Types 1 & 2 / Gilbert Syndrome |
| WT1 | Wilms tumor, type 1 and other WT1-related disorders |

| PHARMACOGENETIC GENES | |
|---|---|
| CYP2C19 | Cytochrome P450-2C19 |
| CYP2C9 | Cytochrome P450-2C9 |
| CYP2D6 | Cytochrome P450-2D6 |
| CYP3A5 | Cytochrome P450-3A5 |
| DPYD | Dihydropyrimidine dehydrogenase |
| MT-RNR1 | Mitochondrially encoded 12S RNA |
| SLCO1B1 | Solute Carrier Organic anion transporter family member 1B1 |
| TPMT | Thiopurine S-methyltransferase |
| UGT1A1 | UDP glucoronosyltransferase family 1 member A1 |
| VKORC1 | Vitamin K epoxide reductase complex subunit 1 |

# Informed Consent for Children

This informed consent describes the benefits, risks, and limitations of undergoing DNA testing on your child for certain genetic conditions with Sema4 Natalis. **Your child's DNA will not be analyzed by Sema4 unless you confirm that you have read and understood the contents of this form.**

This is a voluntary test that you are choosing for your child. You may wish to seek additional, independent genetic counseling on your child's behalf prior to agreeing to this form. If you have any questions about your medical care, you should seek the advice of your physician or other qualified healthcare provider. Never disregard professional medical advice or delay seeking it.

**What is this test?**

This has two components, and tests whether your child is affected or at risk to be affected with one of the genetic conditions included on Natalis and also tests for genes that predict your child's drug response variability to certain medications.

The screening component of this test looks at your child's genetic material for evidence of disease-related changes in 193 different genes. These genes are for diseases that occur in infancy or early childhood and for which there is treatment or medical management that, when administered early in an infant or child's life, may significantly improve their clinical outcome. This test will only report back genetic changes that indicate a child is affected with the disease, but it will not report back whether your child is merely a "carrier" of the variation associated with a disease. The test will also not report changes that have not been classified as "pathogenic" or "likely pathogenic", which means that they are known to cause a disease, according to the laboratory standards and guidelines published by The American College of Genetics and Genomics.

This test also looks at an additional ten genes that are implicated in drug response variability—referred to as "pharmacogenetic" or PGx genes—for a subset of 30 medications that may be prescribed during childhood. For these genes, we will only report those genetic changes that are clinically relevant and that have therapy recommendations affiliated with them. Please note that these genetic changes are much more common than the disease-causing changes included in the screening component of this test, and most people carry at least one genetic change in these ten pharmacogenetic genes.

A complete list of the genes and corresponding conditions screened for by the screening and the PGx components of the test may be found at sema4.com/Natalis/conditions. This test does not screen for any other genetic conditions, and Sema4 will not perform any other analysis on your child's sample without your consent.

**Is genetic counseling included?**

Board Eligible/Certified Genetic Counselors are available to support your physician in the event that there are any positive results. Genetic Counselors are available to explain any positive pharmacogenetic results to you directly. Because our Genetic Counselors conduct focused sessions via telephone and/or video, it is strongly recommended that all positive disease-related results.be discussed in the setting of a formal evaluation by a clinical geneticist (or similar provider).

**What are the possible benefits of this test?**

Your child's screening results may help you identify a previously undiagnosed genetic disease that has a specific treatment or medical management plan that could improve clinical outcome in your child and aid in reproductive planning for future pregnancies. Further, the pharmacogenetic component of this test may help guide your child's physician when selecting appropriate medications.

**What are the limitations and risks of this test?**

This test is designed to detect gene variants associated with only certain genetic diseases. It cannot detect every variant associated with each disease, nor does it look for all known genetic diseases that could affect your child. This test only provides information about the specific conditions and PGx genes tested. Negative results do not guarantee that you or your children will be healthy. No single genetic test can detect all of the possible gene variants that could cause a disease. This test only reports changes (variants) that are pathogenic or likely pathogenic and will not report a variant that is of uncertain significance. This means that, even if your child tests negative, there is a chance that he or she may still develop one of the genetic conditions on this test. In addition, it is possible that your child may receive a positive result for a disease that he/she might not be significantly affected with due to incomplete penetrance. This is because in some disorders, not all individuals who carry genetic variants that have been shown to cause a particular disease will exhibit symptoms of that disease. Please contact your physician or inquire through the genetic counseling services, as this test is not meant to replace medical care from your own physician. It is recommended that you seek the advice of your physician or other appropriate healthcare professional with any questions you may have regarding your results.

Some biological factors, such as a history of bone marrow transplantation or recent blood transfusions, may limit the accuracy of results. As with all medical tests, there is a chance of a "false positive" or a "false negative" result. A false positive result means that a gene variant was detected, but it is not actually there. Similarly, a false negative result means the test did not identify a gene variant that you actually have.

Additionally, you understand that genetic testing may reveal sensitive information about your child's health, your own health, or that of your relatives. Test results may reveal incidental, unsought information, such as discovering an undiagnosed disorder, revealing cases of adoption, or demonstrating that a person is not the father or mother of this child.

**Sample storage**

By signing this consent form, I agree that Sema4 may store, de-identify, and use 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 my child will receive no compensation in connection with such research. If my child is a resident of New York, my child's sample will not be retained for more than 60 days after collection unless I give my consent at the end of this document.

I understand that I may decline to have my child's sample retained for de-identified research purposes by Sema4 by initialing here:_____. I understand that I may withdraw this consent at any time by contacting Sema4 (including by emailing privacy@sema4.com) and my child's specimen will be promptly destroyed.

**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 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 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 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.

**For residents of New York state only**

_____
*Initials*

**By initialing here, I give consent to have my child's 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.** Samples from residents of New York will not be retained for more than 60 days after collection without consent. I understand that I may withdraw this consent at any time by contacting Sema4 (including by emailing privacy@sema4.com) and that my child's specimen will be promptly destroyed.

Child's name

Date of Birth

Name and signature of legal guardian

Date

# Informed Consent for Parents

Sema4's Natalis has been ordered for your child. This informed consent describes the benefits, risks, and limitations of your providing a specimen to be used for targeted molecular genetic testing solely so that Sema4 may better interpret test results for your child's test. **Your DNA will not be analyzed by Sema4 unless you confirm that you have read and understood the contents of this form.**

**What is this test?**

Your sample will undergo a DNA extraction process allowing for the storage of your DNA, should it be needed for testing. Should your child's preliminary Natalis results identify a genetic finding that warrants additional interpretation, you understand that your extracted DNA sample will be used for targeted molecular genetic testing so that Sema4 may better interpret test results for your child. The presence/absence of genetic findings in your DNA may impact the interpretation of your child's test results.

Sema4's Natalis has two components, and tests whether your child is affected with one of the genetic conditions included on our panel and also tests for genes that predict your child's drug response variability to certain medications. A complete list of the genes and corresponding conditions screened for by the screening component of the test may be found in the test information sheet and a complete list of the genes and medications included in the PGx component of this test may also be found on the test website: sema4.com/Natalis/conditions. This test does not screen for any other genetic conditions, and Sema4 will not perform any other analysis on your sample without your consent.

**What are the risks of this test?**

You understand that genetic testing may reveal sensitive information about your child's health, your own health, or that of your relatives. Test results may reveal incidental, unsought information, such as discovering an undiagnosed disorder, revealing cases of adoption, or demonstrating that a person is not the father or mother of this child.

**Sample storage**

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 relating to health, disease prevention, drug development, and other scientific purposes, and that I will receive no compensation in connection with such research. If I am a resident of New York, my sample will not be retained for more than 60 days after collection unless I give my consent at the end of this document. I understand that I may decline to have my sample retained for de-identified research purposes by Sema4 by initialing here:_____. I understand that I may withdraw this consent at any time by contacting Sema4 (including by emailing privacy@sema4.com) and my specimen will be promptly destroyed.

**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 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.

**For residents of New York state only**

_____
*Initials*

**By initialing here, 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.** Samples from residents of New York will not be retained for more than 60 days after collection without consent. I understand that I may withdraw this consent at any time by contacting Sema4 (including by emailing privacy@sema4.com) and that my specimen will be promptly destroyed.

_____ _____

Signature of person being tested (or guardian) Date