Informed Consent for Natalis Testing for Children

If you are under eighteen (18) years of age, a legal representative who is at least eighteen (18) years of age and has the legal authority and capacity to do so must sign this consent and authorization on your behalf.

This informed consent describes the benefits, risks, and limitations of undergoing DNA testing for certain genetic conditions with Sema4 Natalis. I understand that this is a voluntary test and I may wish to seek additional, independent genetic counseling prior to agreeing to this form. If I have any questions about medical care, I should seek the advice of my physician or other qualified healthcare provider.

What is this test?

Natalis has two components. The first screening tests whether a child is affected or at risk to be affected with one of the genetic conditions included on Natalis. If ordered, the second component will test for genes that predict drug response variability to certain medications.

The screening component of this test looks at genetic material for evidence of disease-related changes in specific genes. These genes are associated with diseases that occur in infancy or early childhood. There is treatment or medical management for these diseases 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 (variants) that indicate a child is affected with the disease. This test will not report back whether the child is a "carrier" of genetic changes which are not expected to cause symptoms of a disease. The test will only report variants that have been classified as "pathogenic" or "likely pathogenic", which means that they are known or likely to cause a disease, according to the laboratory standards and guidelines published by The American College of Genetics and Genomics.

If ordered, 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 medications that may be prescribed during childhood. For these genes, only genetic changes that are clinically relevant and that have therapy recommendations affiliated with them will be reported. 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 www.sema4.com/elements/natalis/conditions/.

Is genetic counseling included?

Board eligible/certified genetic counselors are available to support physicians in the event that there are any positive results. Genetic counselors are available to explain any positive pharmacogenetic results. Since the 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?

The screening results may help identify a previously undiagnosed genetic disease that has a specific treatment or medical management plan that could improve clinical outcome and aid in reproductive planning for future pregnancies. Furthermore, the pharmacogenetic component of this test may help guide physicians 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 and PGx genes. It cannot detect every variant associated with each disease, nor does it look for all known genetic diseases.

Negative results do not guarantee a healthy child. No single genetic test can detect all of the possible gene variants that could cause a disease. This test only reports variants that have been classified as pathogenic or likely pathogenic and will not report a variant that is classified as of uncertain significance, benign, or likely benign. Even if a child tests negative, there is a chance that they may still develop one of the genetic conditions on this test. In addition, it is possible that a child may receive a positive result for a disease and be affected differently than expected or not affected at all.

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 that the child does not actually have it. Similarly, a false negative result means the test did not identify a gene variant that a child actually has.

Additionally, I understand that genetic testing may reveal sensitive information about my own health or that of 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. 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 described.

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 I designate to receive this information. My test results will be explained to me by a genetic counselor or by my healthcare provider, who will have the opportunity to discuss my results with a geneticist.

There are federal and state laws that address genetic discrimination. The US Genetic Information Nondiscrimination Act (GINA) may prohibit discrimination based on genetic information by employers and health insurers. This law, however, does not protect people in the military nor protect against discrimination by other types of insurance, such as life, disability, or long-term care insurance.

Sample management

Sema4 may deidentify and retain your left-over sample to use for operational, quality control, validation and improvement purposes. Other than retention for these uses, your sample will be destroyed at the end of the testing process or within 60 days of sample collection, whichever is longer.

De-identified research

Sema4 may de-identify and use all data and information generated and received in connection with this test to support medical and academic research relating to health, disease prevention, drug development, and other scientific purposes. I will receive no compensation in connection with such research. Data and information are "de-identified" by removing any information that could be used to identify a specific person, such as a name, email address, or date of birth. Sema4 may also give the de-identified data and information to its research partners and may submit it to research databases for use in scientific and medical research. Examples of such research include projects to understand the risk factors and outcomes for various conditions and can be found at www.sema4.com/research.

If I do not want to have my de-identified data and information used in research as set forth above, I may withdraw this consent by emailing privacy@sema4.com, and I understand that the change will apply to all data generated from tests that I have undergone with Sema4. I further understand that this withdrawal will not apply to any information that has already been de-identified and cannot be identified by Sema4.

Permission to contact

I understand that Sema4 may wish to contact me 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 sample(s). If I wish to opt-out out of future contact for research purposes, I will notify Sema4 by emailing privacy@sema4.com.

My questions regarding this testing have been answered to my satisfaction, and I hereby consent to have my specimen tested. I understand that I have the option to speak with a healthcare provider should I have additional questions or want counseling regarding this testing. If my legal representative is signing this consent and authorization, my legal representative is satisfied that they have received enough information to sign on my behalf.

Please complete all required (*) fields and optional applicable fields below:

Patient Name*	Patient's DOB*	Date*
Signature of Legal Representative*	Email Address*	Phone Number*
Legal Representative Name*		

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