Informed Consent and Authorization Form

I understand that my health care provider has ordered the following genetic testing for {me/my child}:_____.

General Information About Genetic Testing

What is genetic testing?

Genetic disorders are caused by changes in a person's DNA. DNA is the material that provides instructions for our body's growth and development. For example, DNA determines such things as eye color and how our lungs work. DNA is compacted into 46 chromosomes, which are found in almost every cell of the body. A gene is a stretch of DNA on a chromosome that has the instructions for making a protein.

Genetic testing is a type of medical test that identifies changes in chromosomes and the DNA of a gene. The purpose of this test is to see if I, or my child, have a genetic variant or chromosome rearrangement causing a genetic disorder or to determine the chance I, or my child, will develop or pass on a genetic disorder in the future. For the purposes of this Consent, 'my child' can also mean my unborn child.

Additional information about the specific test being ordered is available from my health care provider or I can go to the GeneDx website, www.genedx.com. This information includes the specific types of genetic disorders that can be identified by the genetic test, the likelihood of a positive result, and the limitations of genetic testing.

What could I learn from this genetic test?

If {I/my child} have a family history of one of the conditions that is being tested, I should inform the laboratory of the specific gene variant(s) or chromosome rearrangement present in the family if it is known. The genetic test may identify the cause of the genetic disease that {I/my child} have or a normal genetic result may significantly reduce, but cannot eliminate, the likelihood that the condition in {me/my child} is genetic or that {I/my child} will develop the genetic disorder in the future. The following describes the possible results from the test:

1) Positive: A positive result indicates that a gene or chromosome variation has been identified that explains the cause of {my/my child's} genetic disorder or that {I/my child} am at increased risk to develop the disorder in the future. It is possible to test positive for more than one genetic variant.

2) Negative: A negative result indicates that no disease-causing genetic variant was identified for the test performed. It does not guarantee that {I/my child} will be healthy or free from other genetic disorders or medical conditions.

If {I/my child} test negative for a variant known to be present in other members of {my/my child's family}, this result rules out a diagnosis of the same genetic disorder in {me/my child}.

3) Inconclusive/Variant of Uncertain Significance (VUS): A finding of a variant of uncertain significance indicates that a change in a gene was detected, but it is currently unknown whether that change is associated with a genetic disorder. A variant of uncertain significance is not the same as a positive result and does not clarify whether {I/my child} am at increased risk to develop a genetic disorder. The change could be a normal genetic variant or it could be disease-causing. Further analysis may be recommended, including testing both parents and other family members. Detailed medical records or information from other family members also may be needed to help clarify results.

4) Unexpected results: In rare instances, this test may reveal an important genetic change that is not directly related to the reason for ordering this test. For example, this test may tell me about the risk for another genetic condition {I/my child} am not aware of or it may indicate differences in the number or rearrangement of sex chromosomes. This information may be disclosed to the ordering health care provider if it likely impacts medical care.

Result interpretation is based on currently available information in the medical literature, research and scientific databases. Because the literature, medical and scientific knowledge are constantly changing, new information that becomes available in the future may replace or add to the information GeneDx used to interpret {my/my child's} results. GeneDx does not routinely re-analyze test results or issue new test reports, and has no obligation to do so. I, or {my/my child's} health care providers may monitor publicly available resources used by the medical community, such as ClinVar (www.clinvar.com), to find current information about the clinical interpretation of my/my child's variant(s).

What are the risks and limitations of this genetic test?

- Genetic testing is an important part of the diagnostic process. However, genetic tests may not always give a definitive answer.
- In some cases, testing may not identify a genetic variant even though one exists. This may be due to limitations in current medical knowledge or testing technology.
- Accurate interpretation of test results may require knowing the true biological relationships in a family. Failing to accurately state the biological relationships in {my/my child's} family may result in incorrect interpretation of results, incorrect diagnoses, and/or inconclusive test results.
- In some cases, genetic testing can reveal that the true biological relationships in a family are not as they were reported. This includes non-paternity (the stated father of an individual is not the biological father) and consanguinity (the parents of an individual are related by blood). It may be necessary to report these findings to the health care provider who ordered the test.
- Genetic testing is highly accurate. Rarely, inaccurate results may occur for various reasons. These reasons include, but are not limited to: mislabeled samples, inaccurate reporting of clinical/medical information, rare technical errors, or unusual circumstances such as bone marrow transplantation, blood transfusion, or the presence of change(s) in such a small percentage of cells that may not be detectable by the test (mosaicism).
- This test does not have the ability to detect all of the long-term medical risks that {I/my child} might experience. The result of this test does not guarantee my health or the health of my child/fetus.
- Occasionally, an additional sample may be needed if the initial specimen is not adequate.

Specimen Retention, De-identified Scientific and Medical Research

DNA samples are not returned to individuals or to referring health care providers. De-identified samples and de-identified test results may be stored in a repository and used for internal validation, educational, and/or research purposes or presented in scientific presentations or papers. In addition, de-identified information may be submitted in a HIPAA-compliant manner to research databases.

Any such research with such de-identified samples and test data that results in medical advances, including new products, tests or discoveries, may have potential commercial value and may be developed and owned by GeneDx or the researchers who analyze the data. If any individuals or corporations benefit financially from studying {my/my child's} de-identified genetic material, no compensation will be provided to {me/my child} or {my/my child's} heirs.

GeneDx has no obligation to retain {my/my child's} sample indefinitely and may destroy it once it no longer has a legal duty to retain it. By consenting to this agreement, I provide authorization for GeneDx and its partners to use {my/my child's} de-identified sample and test results for such purposes as mentioned above *(New York residents: please see specific language on the next page)*.

GeneDx may also contact me in the future regarding the opportunity to participate in research opportunities, including treatment for the condition in my family.

I understand that I may contact the laboratory via email at genedx@genedx.com or by phone at +1-301-519-2100, or if I am located in the United States, toll free at +1-888-729-1206 if I wish to opt out of future contact or have any questions.

I understand that samples from residents of New York State will not be included in the de-identified research studies described in this authorization and will not be retained for more than 60 days after test completion, unless specifically authorized by my selection below. The authorization is optional, and testing will be unaffected if I do not check the box for the New York authorization language.

International Specimens

If {I/my child} reside outside the United States, I attest that by providing a sample for testing, I am not knowingly violating any export ban or other legal restriction in the country of {my/my child's} residence.

Patient Confidentiality and Genetic Counseling

It is recommended that I receive genetic counseling before and after having this genetic test. Further testing or additional consultations with a health care provider may be necessary.

To maintain confidentiality, the test results will only be released to the referring health care provider, to the ordering laboratory, to me, to other health care providers involved in {my/my child's} diagnosis and treatment, or to others as entitled by law.

The United States Federal Government has enacted several laws that prohibit discrimination based on genetic test results by health insurance companies and employers. In addition, these laws prohibit unauthorized disclosure of this information. For more information, I understand that I can visit www.genome.gov/10002077.

Patient Acknowledgment

By agreeing to this authorization, I acknowledge the following:

- I am either (1) the patient providing the sample and am at least 18 years of age or (2) I have legal authorization to provide this informed consent on behalf of another person.
- I have read and agree to the contents of this form.
- I understand the benefits, risks and limitations of genetic testing.
- I have been informed of the availability of genetic counseling services. I can find a genetic counselor in my area at: www.nsgc.org.
- I will be given the opportunity to discuss the results of the test with my health care provider, once I receive them.
- I am responsible for informing my ordering health care provider of changes in {my/my child's} family history.
- I understand that GeneDx may contact me in the future for research opportunities, including treatments for the condition in {my/my child's} family. (Please check the box at the end of this Authorization if you do not wish to be contacted for future research opportunities.)
- I understand that GeneDx may use {my/my child's} de-identified information and test results for validation, educational, and/or research purposes, and this de-identified data may be submitted in a HIPAA-compliant manner to research databases.
- For tests or studies that generate data from multiple family members or my spouse or partner, I consent to all the data being included in a single comprehensive report that will be shared with participating family members, my spouse or partner.
- If GeneDx is billing my medical insurance carrier directly, I represent that I am covered by insurance and authorize GeneDx to give my designated insurance carrier, health plan, or third party administrator (collectively "Plan") the information on this form and other information provided by my health care provider necessary for reimbursement and I authorize Plan benefits to be payable directly to GeneDx.
- I authorize GeneDx to inform my Plan of my test result(s) only if the test result(s) are required for preauthorization of, or payment for, additional testing.
- I will cooperate fully with GeneDx by providing all necessary documents needed for insurance billing and appeals; and understand that I am responsible for sending GeneDx any, and all, of the money that I receive directly from my insurance company in payment for this test. Reasonable collection and/or attorney's fees, including filing and service fees, shall be assessed if the account is sent to collection, as permitted by state law. I permit a copy of this authorization to be used in place of the original.

By agreeing to this informed consent below I am confirming that I understand the benefits, risks and limitations associated with genetic testing. Furthermore, I am affirming that I recognize the seriousness of conditions for which {I/my child} am being tested, and that disease descriptions, prognoses, and treatment options have been made available to me by {my/my child's} health care provider. Finally, if I have the legal authorization to provide this informed consent on behalf of another person, I am attesting that the sample provided belongs to that person.

Patient/Guardian Authorization

By my signature below I attest to the following:

I have read and I understand the information provided on this form.

Opt Out for Research and Contact

❒ I do not wish to participate in any research studies.

❒ I do not wish to be contacted by GeneDx for future research opportunities. I understand that my election to opt out of such follow up contacts will not affect my ability to obtain testing.

Authorization for New York Residents

❒ I am a New York state resident and I give permission for GeneDx to retain any remaining sample longer than 60 days after completion of testing and use my de-identified data for scientific and medical research purposes. Such authorization is optional and is not required for testing.

Patient/Guardian Name: _____ _____
(Please print) First Name Middle Name Last Name Date of Birth: mm/dd/yyyy

Patient/Guardian Signature: _____ Date: _____ mm/dd/yyyy

Health Care Provider's Statement

This test is medically necessary for the risk assessment, diagnosis or detection of a disease, illness, impairment, symptom, syndrome or disorder. The results will determine my patient's medical management and treatment decisions. By my signature below, I indicate that I am the referring physician or authorized health care provider. I have explained the purpose of the test described above. The patient has been given the opportunity to ask questions and/or seek genetic counseling. The patient has voluntarily decided to have the test performed by GeneDx.

Health Care Provider's Signature: _____ Date: _____ mm/dd/yyyy