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|--------------------------|--------------------------------------|
| Patient First Name       | Patient Last Name                    |
| Patient DOB (mm/dd/yyyy) | GeneDx Portal Order ID (if relevant) |

For the purposes of this consent, “I”, “my”, and “your” will refer to me or to my child, including my unborn child, if my child is the person for whom the healthcare provider has ordered testing.

## PURPOSE OF THIS TEST

The purpose of this test is (a) to see if I may have a genetic variant or chromosome rearrangement causing a genetic disorder; or (b) to evaluate the chance that I will develop or pass on a genetic disorder in the future. If I already know the specific gene variant(s) or chromosome rearrangement that causes the genetic disorder in my family, I agree to inform the laboratory of this information.

## WHAT TYPE OF TEST RESULTS CAN I EXPECT FROM GENETIC TESTING?

- Positive:** A change in your DNA was found, which is very likely the cause of your features/symptoms. This is the most straightforward test result, which can be used as the basis to test other family members to determine their chances of having either the disease or a child with the disease.
- Negative:** No variants were found to explain your symptoms. This does not mean that you do not have a genetic condition. It is still possible that there is a genetic variant not found by the test that was ordered. Your healthcare provider or genetic counselor may discuss more testing either now or in the future.
- Variant of Uncertain Significance (VUS):** A change in a gene was found. However, we are not sure whether this variant is the cause of your symptoms/features. More information is needed. We may suggest testing other family members to help figure out the meaning of the test result.
- 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 find you are at risk for another genetic condition I am not aware of or it may indicate differences in the number or rearrangement of sex chromosomes. We may disclose this information to the ordering healthcare provider if it likely affects medical care.

Because medical and scientific knowledge is constantly changing, new information that becomes available may supplement the information GeneDx used to interpret my results. Healthcare providers can contact GeneDx at any time to discuss the classification of an identified variant.

## WHAT IS TRIO/DUO-BASED GENETIC TESTING?

For some genetic tests, including samples from the biological parents and/or other biological relatives along with the patient’s sample can help with the interpretation of the test results. These tests are often referred to as “trio tests” since they typically include samples from the patient and both parents.

Samples from relatives should be submitted with the patient’s sample. Clinical information must be provided for the patient and any relative who submits a sample.

I understand that GeneDx will use the relative sample(s) when needed for the interpretation of my test results and that my test report may include clinical and genetic information about a relative when it is relevant to the interpretation of the test results. I further understand that relatives will not receive an independent analysis of data nor a separate report.

## RISKS AND LIMITATIONS OF GENETIC TESTING

- 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. I understand that if I fail to accurately state the biological relationships in my family, it could lead to incorrect interpretation of the test results, incorrect diagnoses, and/or inconclusive test results. If genetic testing reveals that the true biological relationships in a family are not as I reported them, including non-paternity (the reported father is not the biological father) and consanguinity (the parents are related by blood), I agree to have these findings reported to the healthcare provider who ordered the test.
- Although genetic testing is highly accurate, inaccurate results may occur. These reasons include, but are not limited to mislabeled samples, inaccurate reporting of clinical/medical information, rare technical errors, or other reasons.
- I understand that this test may not detect all of the long-term medical risks that I might experience. The result of this test does not guarantee my health and that additional diagnostic tests may still need to be done.
- I agree to provide an additional sample if the initial sample is not adequate.

## PATIENT CONFIDENTIALITY AND GENETIC COUNSELING

It is recommended that I receive genetic counseling before and after having this genetic test. I can find a genetic counselor in my area at [www.nsgc.org](http://www.nsgc.org). Further testing or additional consultations with a healthcare provider may be necessary.

To maintain confidentiality, test results will only be released to the referring healthcare provider, the ordering laboratory, to me, to other healthcare providers involved in my care, diagnosis and treatment, or to others with my consent or as permitted or required by law. Federal laws prohibit unauthorized disclosure of this information. More information can be found at: [www.genome.gov/10002077](http://www.genome.gov/10002077)

## INTERNATIONAL SAMPLES

If I 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 residence.

## SAMPLE RETENTION

After testing is complete, my sample may be de-identified and be used for test development and improvement, internal validation, quality assurance, and training purposes. GeneDx will not return DNA samples to you or to referring healthcare providers, unless specific prior arrangements have been made.

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 GeneDx will not retain them for more than 60 days after test completion, unless specifically authorized by my selection. The authorization is optional, and testing will be unaffected if I do not check the box for the New York authorization language. GeneDx will not perform any tests on the biological sample other than those specifically authorized.

## DATABASE PARTICIPATION

De-identified health history and genetic information can help healthcare providers and scientists understand how genes affect human health. Sharing this de-identified information helps healthcare providers to provide better care for their patients and researchers to make new discoveries. GeneDx shares this type of information with healthcare providers, scientists, and healthcare databases. GeneDx will not share any personally identifying information and will replace the identifying information with a unique code not derived from any personally identifying information. Even with a unique code, there is a risk that I could be identified based on the genetic and health information that is shared. GeneDx believes that this is unlikely, though the risk is greater if I have already shared my genetic or health information with public resources, such as genealogy websites.

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## EXOME/GENOME SEQUENCING SECONDARY FINDINGS

- Applicable only for full exome and genome sequencing tests
- Does not pertain to Xpanded® or Slice tests

As many different genes and conditions are analyzed in an exome or genome sequencing test, these tests may reveal some findings not directly related to the reason for ordering the test. Such findings are called “incidental” or “secondary” and can provide information that was not anticipated.

Secondary findings are variants, identified by an exome or genome sequencing test, in genes that are unrelated to the individual’s reported clinical features.

The American College of Medical Genetics and Genomics (ACMG) has recommended that secondary findings identified in a specific subset of medically actionable genes associated with various inherited disorders be reported for all probands undergoing exome or genome sequencing. Please refer to the latest version of the ACMG recommendations for reporting of secondary findings in clinical exome and genome sequencing for complete details of the genes and associated genetic disorders. Reportable secondary findings will be confirmed by an alternate test method when needed.

### WHAT WILL BE REPORTED FOR THE PATIENT?

All pathogenic and likely pathogenic variants associated with specific genotypes identified in the genes (for which a minimum of 10X coverage was achieved by exome sequencing or a minimum of 15X coverage was achieved by genome sequencing), as recommended by the ACMG.

### WHAT WILL BE REPORTED FOR RELATIVES?

The presence or absence of any secondary finding(s) reported for the proband will be provided for all relatives analyzed by an exome or genome sequencing test.

### LIMITATIONS

Pathogenic and/or likely pathogenic variants may be present in a portion of the gene not covered by this test and therefore are not reported. The absence of reportable secondary findings for any particular gene does not mean there are no pathogenic and/or likely pathogenic variants in that gene. Pathogenic variants and/or likely pathogenic variants that may be present in a relative, but are not present in the proband, will not be identified, or reported. Only changes at the sequence level will be reported in the secondary findings report. Larger deletions/duplications, abnormal methylation, triplet repeat or other expansion variants, or other variants not routinely identified by clinical exome and genome sequencing will not be reported.

## FINANCIAL AGREEMENT AND GUARANTEE

For insurance billing, I understand and authorize GeneDx to bill my health insurance plan on my behalf, to release any information required for billing, and to be my designated representative for purposes of appealing any denial of benefits. I irrevocably assign to and direct that payment be made directly to GeneDx.

I understand that my out-of-pocket costs may be different than the estimated amount indicated to me by GeneDx as part of a benefit investigation. I agree to be financially responsible for any and all amounts as indicated on the explanation of benefits issued by my health insurance plan. If my insurance provider sends a payment directly to me for services performed by GeneDx on my behalf, I agree to endorse the insurance check and forward it to GeneDx within 30 days of receipt as payment towards GeneDx’s claim for services rendered.

If I do not have health insurance, I agree to pay for the full cost of the genetic testing that was ordered by my healthcare provider and billed to me by GeneDx. I further understand and agree that, if I fail to make payment for genetic testing, in accordance with the payment policies of GeneDx, my account may be turned over to an external collection agency for non-payment. I agree to pay any associated collection costs, including attorney fees. By my signature on the GeneDx Test Requisition Form or at the bottom of this form, I accept full and complete financial responsibility for all genetic testing ordered by my healthcare provider.

## MEDICARE

A completed Advance Beneficiary Notice (ABN) is required for Medicare patients. Please visit our website, [www.genedx.com/billing](http://www.genedx.com/billing) for more information.

## DIGITAL PATIENT LETTER CONSENT

- Applicable Only for Commercial Insurance
- Estimate is provided by your health insurance company and therefore NO estimate will be sent for any orders placed with federal or state-funded insurance plans (e.g. Medicare, Medicaid, Tricare, etc.), institutional bill, or patient bill (self-pay).

To provide you with the estimated out-of-pocket expenses related to your test, GeneDx will send you an email and/or text with the link to access your personalized Digital Patient Letter. In order to send this information, we need your consent and agreement to the following items:

1. GeneDx can use your email address or mobile phone number solely for the purpose of GeneDx sending your estimated financial obligation. Text message data rates may apply. GeneDx is not responsible for undelivered messages due to incorrect or illegible contact information.
2. GeneDx will send you an email and/or text message containing a link to view your personalized Patient Letter that includes the test out-of-pocket estimate. The link is time-sensitive and will only be available for 72 hours from the time the message is sent. In order to view the estimate, you must click the link in the message.
3. If you take no action, GeneDx will assume that you agree to move ahead with testing and will bill your health insurance. You can approve testing with insurance, switch to self-pay, or cancel the test via the link within the given 72-hour window. In turn, if GeneDx receives your sample(s) and the billing method hasn’t been changed, or the test hasn’t been cancelled, we will move ahead with testing as ordered, and you will be responsible for any out-of-pocket costs for the completion of the test(s).

I have read, or have had read to me, all of the above statements and understand the information regarding molecular genetics testing. I have had the opportunity to ask questions about the testing, the procedure, the risks, and the alternatives. By signing this form, I authorize GeneDx to perform genetic testing as ordered. I understand that, for tests that evaluate data from multiple family members concurrently, test results from these family members may be included in a single comprehensive report that will be made available to all tested individuals and their healthcare providers. More information, including the GeneDx Notice of Privacy Policies, is available on GeneDx’s website: [www.genedx.com](http://www.genedx.com)

- By checking this box, I confirm that I am a New York state resident, and I give permission for GeneDx to retain any remaining sample longer than 60 days after the completion of testing, and to be used as a de-identified sample for test development and improvement, internal validation, quality assurance, and training purposes. Otherwise, New York law requires GeneDx to destroy my sample after 60 days, and it cannot be used for test development studies.
- Check this box if you wish to opt out of being contacted for research studies.
- Check this box if you do not wish to receive ACMG secondary findings (Full Exome Sequencing and Genome Sequencing Tests ONLY; not for Xpanded® or Slice tests).

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|--|--|
| Signature of Patient/Legal Guardian (required) | Date                                       |
| Signature of Relative A/Legal Guardian         | Relative A Relationship to Patient<br>Date |
| Signature of Relative B/Legal Guardian         | Relative B Relationship to Patient<br>Date |

**OPTIONAL AND FOR COMMERCIAL INSURANCE ONLY:** By entering my preferred contact information below, I give my permission to GeneDx to send me an email and/or text with a link to access my personalized Digital Patient Letter. Data rates may apply.

|                |        |
|----------------|--------|
| Mobile Number* | Email* |
|----------------|--------|

\*Contact information provided must be for the individual authorizing the genetic testing.