

# INFORMED CONSENT

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

## 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 have 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 you 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 a guide to help 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 that was not tested. 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 reveal that 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 your 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 the test results. Healthcare providers can contact GeneDx at any time to discuss the classification of an identified variant.

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

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## EXOME/GENOME SEQUENCING SECONDARY FINDINGS

- Applicable only for full exome and genome sequencing tests
- Does not pertain to Xpafid 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 testing. 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 be reported to patients.