

Informed Consent for Genetic Testing

This consent form is required for all patients in the state of New York

Genetic Testing

Genetic testing looks for changes in DNA, which may be associated with a specific genetic condition. A positive test result is an indication that an individual may be predisposed to or have the specific disease or condition tested. Knowledge of a specific gene change may result in a change in the treatment plan that is currently underway or in medical surveillance for potential future health problems. Discussion of these possible health care changes can take place once a diagnosis is confirmed or a known gene change is found.

Possible Results

- **Positive:** A genetic variant was identified that explains the cause of my/my child's genetic disorder or indicates that I am/my child is at increased risk to develop the disorder in the future. It is possible to test positive for more than one genetic variant.
- **Negative:** No disease-causing genetic variant was identified by the test performed. A negative result does not guarantee that I/my child will not develop the disease tested for. There may be other causes that were not tested for. Other testing may be available to me in the future.
- **Inconclusive/Variant of Uncertain Significance (VUS):** A finding of a variant of uncertain significance indicates that a genetic change was detected, but it is currently unknown whether that change is associated with the tested condition. A variant of uncertain significance is considered an inconclusive result, as it is not clear if the variant is disease causing or not. The change could be a normal genetic variant or it could be disease causing. Further analysis may be recommended, including testing parents and other family members. Detailed medical records or information from other family members may also be needed to help clarify results. These results may take a significant amount of time to understand if they are or are not disease causing.

Test Limitations

- This genetic test may not identify a mutation that is present in an individual due to current limitations in genetic testing technology.
- Results may not be accurate if true biological relationships are not accurately stated. In some cases, genetic testing may reveal biological relationships that are different from those reported (non-paternity, consanguinity). If different biological relationships are identified, it may be required for the testing laboratory to report those findings to the ordering provider.
- Though genetic testing is highly accurate, results may be inaccurate due to mislabeled samples, laboratory error, inaccurate reporting of biological relationships, inconsistent scientific classification systems, or other rare technical errors.
- This test cannot predict all conditions that I/my child may be at risk of developing; likewise, a negative result does not guarantee that I/my child do not have the condition or will not develop the condition. A positive genetic test cannot predict the severity of the disease. Additional testing may be required.

Patient name _____ Date of birth _____

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Confidentiality

- No tests other than those authorized shall be performed on the biological sample.
- Results of this genetic testing will remain confidential and only be shared with the ordering health care provider. Other healthcare providers may receive results if requested as part of my/my child's care, or if I provide consent for the results to be released to them. Results may be shared with individuals as entitled by law. The United States Federal Government has enacted several laws to protect genetic information. For more information, please visit www.genome.gov/10002077.
- My/my child's test results may affect insurance rates and the ability to obtain disability or life insurance and employment. The Genetic Information Non-discrimination Act (GINA), a federal law, provides some protections against genetic discrimination. For information on GINA, visit www.genome.gov/10002328
- My/my child's data and personal information will be stored and protected in compliance with regulatory requirements (e.g., HIPAA and equivalent protections).

Genetic Counseling

Genetic Counseling is recommended prior to undergoing genetic testing, and after results have been received. To find a Genetic Counselor in your area, please visit: www.NSGC.org. Further testing or additional physician consults may be warranted.

The genetic test _____ has been explained to me. I understand the purpose as well as the limitations of the testing and I consent to the testing for me/my child.

De-identified samples may be retained for quality control, test development, validation or training purposes. If you do **NOT** give permission to DDC Clinic to retain any remaining sample longer than 60 days please initial. _____

New York State Residents Only: Samples and any material derived from them (i.e.DNA) will be destroyed after 60 days and will not be used for research or quality control purposes **without the express written consent** of the patient/parent. If you **Do Give Permission** to DDC to retain your/your child's sample for research or quality control purposes, please initial. _____

Patient name (printed) _____ Date of birth _____

Patient or Parent signature _____ Date _____

Parent name (printed) _____