

Name: \_\_\_\_\_

DOB: \_\_\_\_\_

MRN: \_\_\_\_\_

Name of authorizing provider: \_\_\_\_\_

This form is to help the consent process. It is also to support the pre-test counseling discussion. It is your choice to have this test or not. Another option is to not have this test. Please ask any questions about this test.

### **PURPOSE OF EXOME SEQUENCING (ES)**

- To find genetic cause(s) of the patient's health issues.

### **THE TEST PROCESS**

- This genetic test looks at most of the genes in the body at one time. This test is different from most genetic tests that look at one gene at a time.
- This test needs a small amount of DNA from the patient. Typically, this comes from blood or saliva.
- The laboratory will compare the patient's DNA sequence to other family members' (if applicable) DNA sequences and to the reference DNA sequence to see if there are differences compared to the expected spelling of the DNA.
- The laboratory will use clinical information and family history to decide which of the genetic changes found may be responsible for the patient's signs and symptoms.
- The laboratory will report the genetic changes that may relate to the patient's signs and symptoms to the provider who ordered the test.

### **LIMITATIONS OF ES TECHNOLOGY**

- This test does not sequence every exon.
- The test may not find all changes in each gene.
- ES finds only small genetic misspellings or small extra or missing pieces of DNA. This test does not detect other types of disease-causing variants. This test may not detect mosaic DNA changes, which are changes that are not present in all cells.
- It is important to have reliable clinical information and a correct family history to interpret results from ES.
- About 25% of patients get a confirmed diagnosis or possible diagnosis from this test.
- ES testing is more likely to find a genetic cause of a patient's disease when samples from biological parents are analyzed at the same time.
- ES results do not predict how severe a condition will be. ES results do not predict the age of onset of conditions.

### **POTENTIAL RISKS OF ES**

- No laboratory test, including ES, is 100% accurate.
  - A possible genetic diagnosis may be incorrect.
  - The patient's diagnosis may not be found with this test.
  - The patient may get uncertain results.
  - The results may be reclassified in the future as genetic knowledge evolves, which could change the recommendations for treatment.
- This test may show that the biological relationships in a family, such as fatherhood or blood relation, are not as suspected. ES is not used to establish paternity or biological relationships.
- This test may find genetic changes unrelated to the patient's current signs and symptoms. These changes could be connected with other health problems (see sections about incidental and secondary findings for more information). These results may be upsetting.



## WHAT IS REPORTED

- Genetic changes that may have caused the patient's signs and symptoms.
- Genetic changes found in genes not related to the patient's condition that may have an important impact on health. You can decide if you do or do not want this information.
- Genetic changes found in family members that are related to the patient's signs and symptoms will be included in the patient's report. Family members will not get separate written reports.

## WHAT IS NOT REPORTED

- Variants in genes that are not thought to affect one's health.
- Variants found in research studies that may not be connected to disease.
- Variants that predict an increased risk of a disease, but do not cause a disease by themselves.
- Variants that may show carrier status but that are not associated with the patient's symptoms.

## SECONDARY FINDINGS

ES may find some genetic changes that are not related to the patient's current signs and symptoms. These are called secondary findings. These findings may have important health effects for patients and their family members. For example, the American College of Medical Genetics and Genomics recommends that all labs that perform ES report disease-causing changes in genes that cause certain inherited disorders. These disorders may lead to serious health problems that can be monitored or treated. These disorders include some cancer syndromes, connective tissue disorders associated with sudden cardiac events, certain types of heart disease, high cholesterol and susceptibility to complications from anesthesia. On the other hand, some types of genetic disorders do not have any effective treatment. These may lead to death or lifelong disability. Secondary findings can be included in the patient's report. We will not look for or report these findings if you tell us that you do not want these results. If you choose to get these results, the patient's report will include this information on disease-causing variants. In addition, the report will note if these variants were found in family members who submitted samples.

Please initial one of the following options (adult patient or parent/guardian of minor child must initial):

\_\_\_\_\_ Option 1: I want results about secondary findings.

\_\_\_\_\_ Option 2: I do not want results about secondary findings. I understand that I may not be able to get these results later.

## CONFIDENTIALITY

- The laboratory will report test results to the provider who ordered the test.
- The laboratory will not give test results to anyone else without your written permission.
- The written report will become part of the patient's medical record. The patient's health insurance provider or other parties may have legal access to this information.
- The laboratory can give raw data from the ES testing after the testing is complete to a healthcare provider or researcher. This will only be done with consent from the patient or parents, and upon request from a healthcare provider.

## FUTURE OF THE DATA

- The laboratory will store any remaining sample(s) for two years. They may get rid of those samples after two years.
- The laboratory will keep the test report for 20 years.
- The laboratory will keep the genetic information on a secured computer that can only be accessed by certain people.
- The laboratory may contact your provider if new information is available later about the findings of this test that could affect the patient's medical care.



**INFORMED CONSENT FOR  
CLINICAL EXOME SEQUENCING**

Name: \_\_\_\_\_

DOB: \_\_\_\_\_

MRN: \_\_\_\_\_

**POST-TEST COUNSELING AND INTERPRETATION**

It is recommended that patients get genetic counseling before signing this consent and when results are available. To find a genetic counselor near you, ask your doctor to refer you to a genetic counselor. Or you can go to [www.nsgc.org](http://www.nsgc.org). Signing this document is saying that the test and its limitations and risks have been explained to you.

By signing below, I am saying that I have talked about the benefits, risks, and limitations of this genetic test with my provider. ES is an ever-changing field of medicine. The laboratory will use a current clinically appropriate methodology available to the laboratory at this time to find genetic changes that might be causing my or my child's signs and symptoms. Better technology might be available in the future. I understand the limitations in current laboratory testing that might be surpassed by future testing. Whether or not I am eligible or appropriate for any future testing is an issue to discuss with my healthcare provider when and if that technology becomes available. My questions about the test have been answered. I consent to whole exome sequencing. I will get a copy of this consent form for my records.

	Time: _____	Date: _____
Patient / Parent / Caregiver Signature	Printed Name	

	Time: _____	Date: _____
Patient / Parent / Caregiver Signature	Printed Name	

	Time: _____	Date: _____
Witness Signature/Credentials	Printed Name	

Note: Both parents must sign above if they are submitting specimens for analysis. Only one parent or guardian must sign if the child's sample is submitted without parental samples.

*Clinician to complete this section:*

**WES ANALYSIS:**  Trio  Duo  Proband Only  Additional Family Member(s)

Mother Name: \_\_\_\_\_ Date of Birth: \_\_\_\_\_

Father Name: \_\_\_\_\_ Date of Birth: \_\_\_\_\_

Additional Family Member Name: \_\_\_\_\_ Date of Birth: \_\_\_\_\_

Relationship to Proband: \_\_\_\_\_

**Physician's/Genetic Counselor's statement:** I have explained exome sequencing to this person. I have addressed the limitations of the test. I understand that interpretation of these results within a clinical context is my responsibility.

Physician/Genetic Counselor Signature/Credentials	Printed Name	Date/Time
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