# INFORMED CONSENT FOR CLINICAL EXOME SEQUENCING (ES) and CLINICAL GENOME SEQUENCING (GS)



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 whether to have this test. Another option is to not have this test. Please ask any questions about this test.

**PURPOSE OF ES/GS**

- To find the 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.
- ES looks at a type of DNA called exons. These pieces of DNA tell the body what proteins to make.
- GS looks at exons and another type of DNA called introns. Introns do not directly make proteins. Some introns are important for DNA function.
- This test needs a small amount of DNA from the patient. Typically, this comes from blood or saliva (spit).
- The laboratory will compare the patient's DNA sequence to other family members' (if applicable) DNA sequences and to a reference DNA sequence to see if there are differences compared to the expected sequence 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/GS TECHNOLOGY**

- ES does not sequence every exon. GS does not sequence every exon and every intron.
- The test may not find all genetic changes in each gene.
- ES finds only single letter changes or small additions or deletions of DNA. This test does not detect other types of disease-causing changes in the DNA. GS finds single base pair changes, small additions or deletions of DNA, and larger additions or deletions of DNA. This test may not detect "mosaic" DNA changes, which are changes that are not present in all cells.
- Accurate clinical information and a correct family history will help interpret results from ES/GS. ES/GS testing is more likely to find a genetic cause of a patient's disease when several family members are tested at the same time.
- About 25% of patients get a confirmed diagnosis or possible diagnosis from ES. In the neonatal setting, up to 40% of people who undergo GS as a first-line test get a diagnosis. In the pediatric setting, the diagnostic yield may be lower, especially for patients who have had previous genetic testing, such as single nucleotide polymorphism (SNP) microarray and/or ES.
- ES/GS results do not predict how severe a condition will be. ES/GS results do not predict the age at which symptoms may appear.

**POTENTIAL RISKS OF ES/GS**

- No laboratory test, including ES/GS, is 100% accurate.
  - A possible genetic diagnosis may be incorrect.
  - The patient's true 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 changes. This 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 reported on the test requisition. ES/GS 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 relate to other health problems (see sections about 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 (changes) in genes that are not thought to affect one's health.
- Variants found in research studies that may not be connected to the disease.
- Variants that predict an increased risk of a disease, but do not cause a disease by themselves.
- Variants that may indicate carrier status but that are not associated with the patient's symptoms.

**SECONDARY FINDINGS**

ES/GS may find some genetic changes that are not related to the patient's current signs and symptoms (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/GS 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. Also, the report will note if these variants were found in family members who submitted samples. Secondary findings that are present in a family member but not present in the patient will not be reported.

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

_____ I want results about secondary findings.

_____ 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/GS 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 at the request of a healthcare provider.

**FUTURE OF THE SAMPLE AND RESULT 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 at least 20 years.
- 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.

**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. 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/GS is a rapidly 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, more precise technology might be available in the future. I understand and acknowledge 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 providers at the time that technology becomes available. My questions about the test have been answered. I consent to:

☐ Exome sequencing

☐ Genome 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

Via: ☐ Phone ☐ Video ☐ On-site

Print name of interpreter and ID number *Note: The interpreter cannot sign as the witness above.*

Name: _____
DOB: _____
MRN: _____

**ES/GS 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/genome sequencing to this person. I have addressed the limitations of the test and have answered all questions. I understand that interpretation of these results within a clinical context is my responsibility.

_____

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