Informed Consent for Whole Exome & Whole Genome Sequencing

Notice to Health Care Practitioner:

This document is a consent form for clinical whole exome or whole genome sequencing. Currently, the laboratory will only accept whole exome and whole genome test requests after the patient/parent or legal guardian/next of kin has received genetic counseling from a Healthcare Provider with experience in counseling patients for such a test. Please be aware of any applicable state laws in regards to counseling needs related to the current condition, the possibilities of detecting unsuspected conditions as well as other issues related to health insurance, and possible effects on life insurance. Please explain this consent to the patient, or authorized representative/guardian, and obtain an informed consent. Please explain the list of potential secondary findings that may be reported to the patient.

Consent for WES and WGS Testing

All of the following has been explained to me, to my satisfaction, and my signature below attests to the same. I understand that this is a voluntary test, and I have had the opportunity to ask questions about alternative testing.

Whole Exome Sequencing: ☐
Whole Genome Sequencing: ☐

Whole Exome or Whole Genome Sequencing Participant:

Patient Name:_____

Patient Date of Birth: _____

Signature: _____ Date_____

(Parent/Guardian if person being tested is a minor)

Health Care Provider Obtaining Consent:

Print Name:_____

Signature: _____ Date_____

NPI#: _____

I have provided genetic counseling and have explained the risks, benefits, and limitations of WES/WGS testing to the patient/parent/guardian.

Consent of family members submitting a sample for evaluation of patient's results

I understand that I am submitting my sample to help evaluate the results obtained on the person being tested, and that results obtained from my sample will be used solely for this purpose. I will NOT be informed of any test results on my sample. If I request any test results, I will have to be tested separately.

Name of Family Member: _____

Relationship to Proband: _____

Signature: _____ Date_____

Name of Family Member: _____

Relationship to Proband: _____

Signature: _____ Date_____

Name of Family Member: _____

Relationship to Proband: _____

Signature: _____ Date_____

Additional Consent (If no selection is made, secondary findings will be reported)

1. Secondary Findings Consent (please choose one option)

(initials) _____ I would like to learn of secondary findings to the conditions listed (Proband Only).

(initials) _____ I would like to learn of secondary findings to the conditions listed (Proband + Parents/Comparators).

(initials) _____ I would NOT like to learn of secondary findings to the conditions listed.

2. What are secondary findings?

During testing, disease-causing variants can be identified that are not related to the patient's condition for which the testing was done. These are referred to as "Secondary Findings" and indicate the presence of previously undiagnosed, potentially serious conditions that can be prevented or treated if diagnosed. A list of such conditions based on the recommendation of the American College of Medical Genetics (ACMG) can be found on the ACMG website.

Reset Form

(continued)

Informed Consent for Whole Exome & Whole Genome Sequencing

What is whole exome sequencing (WES) and whole genome sequencing (WGS)?

Whole exome sequencing (WES) and whole genome sequencing (WGS) are genetic tests. They are performed on DNA extracted from blood or other acceptable tissue type. Their purpose is to identify a heritable cause of a disorder. WES examines the "exome" or coding regions of DNA, the most important information-containing segment of the genome, and will often identify the cause of disease. WGS examines all of the DNA in the human genetic code including coding and non-coding regions. The sensitivity of both tests is improved if blood is submitted from biological parents or siblings of the patient. The goal of both tests is to identify the genetic cause of the disease for which the patient is presenting. For this reason, it is crucial that a detailed description of the clinical symptoms of the patient and other affected family members is provided. Results will only be reported on the patient. Because WES and WGS results have potential consequences for the patient's family, we recommend that the consenting process be performed with the assistance of a genetic counselor. Only a physician or other health care provider is authorized to order testing.

What kind of results are reported?

1. Positive: Variant(s) have been identified that are known to cause the disease symptoms based on the available scientific evidence at the time of testing.
2. Indeterminate: Variant(s) have been identified that are likely to cause the disease symptoms based on the available scientific evidence at the time of testing, but there is a lack of definitive scientific evidence available to prove it.
3. Negative: No variant has been identified that is known or likely to cause the disease symptoms based on the available scientific evidence at the time of testing.

What implications do positive and negative results have?

When WES/WGS detects known disease-causing variants, the test result is highly accurate. A positive result will help your clinician to better predict the course of the condition and provide you with treatment options, if they exist. The results will also help determine the risk of recurrence of the condition in other children. An indeterminate result will point to a possible cause of a condition, but you may wish to consult a genetic counselor or your physician and undergo further independent testing to confirm or rule out the proposed role. A negative result does not indicate the absence of a genetic cause and will not change the clinical diagnosis.

Are there limitations to WES and WGS testing?

WES and WGS are screening tests. There is a possibility a genetic variant caused a condition that is not identified by the WES/WGS tests either because of the technical limitations of the assays, or because of incomplete understanding of the significance of variants detected. Although WES/ WGS testing is highly accurate, the interpretation of the report is based on current medical knowledge, which is not complete.

1. WES may not be able to detect genetic disorders that are caused by expansion of repetitive regions of the genome. One example is Fragile X Syndrome. If one of these types of conditions is suspected, your physician should order the appropriate test.
2. Not all regions in the human genome can be sequenced due to limitations in technology, so some variants in such regions might go undetected with WGS methodologies.

Are there results that will not be reported?

1. Samples from the patient's relatives may be used to help diagnose the patient's condition, but results for these relatives will not be reported independently. They will only be referred to in the report for the patient if they are directly relevant to the patient's condition. However, the patient's genetic results may have implications for their relatives, and it is important that these implications are discussed with a genetic counselor.
2. Variations in genes that affect susceptibility to a condition, but do not cause the person to develop the condition, will not be reported.
3. Carrier status for recessive disorders: Most people carry variants that are not disease causing but could become disease causing if that person had children with someone who was healthy but had the same variant. This is referred to as being a "carrier" for a disease. This test is not intended for determining carrier status. If you are concerned about carrier status for conditions that might run in your family, you should get tested separately for carrier status. You should discuss these implications with your genetic counselor.
    - Single heterozygous pathogenic/likely pathogenic variants in genes associated with recessive disorders that have potential overlap with a patient's clinical presentation (as provided to our laboratory) will be reported since we cannot definitively exclude that an undetected second variant in trans may be present.
    - Single heterozygous variants of uncertain significance in genes associated with recessive disorders that have potential overlap with a patient's clinical presentation (as provided to our laboratory) will be reported at the discretion of the laboratory director.
    - Single heterozygous variants in genes associated with recessive disorders that do not have overlap with the patient's clinical presentation (as provided to our laboratory) will not be reported.

Who will have access to the results?

Test results are maintained electronically by the laboratory. The results are provided to the ordering physician and/or healthcare facility that ordered the test. Results may also be made available to individuals/organizations with a legal right of access under applicable Federal and/or State law, or as authorized by the patient or the patient's representative.

What are the risks of testing?

1. Non-paternity (when the reported father of the child is not the biological father) or half sibling-ships (when siblings do not share the same father AND mother) would be detected. We do not report these findings unless they have direct clinical significance.
2. Genetic non-discrimination law prevents insurance companies from using your genetic information to deny health insurance coverage, but the law does not cover life insurance, disability insurance or long-term care insurance. The detection of an incidental condition may affect your future ability to buy these forms of insurance or get the best insurance rates. Please be aware of any applicable State laws and applicable terms of any active insurance policies in regards to consent and the release of these results to insurance companies.
3. WES/WGS may identify serious and/or untreatable genetic conditions. It can result in unexpected psychological trauma, both for you and your family. The detection of such a condition or conditions could also affect the health or healthcare needs of your siblings, children, or other close relatives.
4. Although WES/WGS is highly accurate, the interpretation of the report is based on current medical knowledge, which is not complete. We do not report out changes in interpretation of variants automatically, but we do have mechanisms to issue an updated report if requested by the patient's physician.

What will happen to my DNA sample?

Following testing, any remaining extracted DNA will be held for two years, at which time it will be discarded.

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