

## UCSF Exome Sequencing Test Informed Consent

The purpose of the *UCSF Exome Sequencing* test is to try to identify a genetic cause for a medical condition in you or your child. This document provides information about the exome test. Read this consent form and discuss it with your health care provider, or with your child's provider. If you wish to proceed with the test, please sign and date this form.

### **Whole Exome Sequencing:**

While traditional genetic testing examines only one gene at a time, whole exome sequencing (WES) examines thousands of genes to try to find a genetic cause for a medical condition. This can help diagnose a patient's condition.

The "exome" is the region of the genome that contains information the body needs to make the proteins that do the body's work. The exome includes a small amount (1-2%) of all human DNA (the genome). Most genetic changes that cause medical conditions are found in the exome.

Every person's exome has several thousand DNA changes, known as sequence variants. These variants may be unique to an individual, found only in one family, or present in the general population. Most of these variants do not cause disease.

The purpose of WES is to find variants that contribute to your, or your child's, medical condition. To assist with interpreting your exome:

- The laboratory will use clinical information provided by your health care provider.
- Your health care provider will determine whether samples from other family members will be included in the testing process.

### **Results:**

A test report with your, or your child's, results, will be provided to your health care provider, who will discuss the results with you. WES may find:

- Variants related to, or likely related to, your, or your child's, medical condition
- Variants of uncertain significance for the medical condition
- No variants related to the medical condition

The test report will not include all variants in the exome.

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### Testing Family Members:

Sometimes family members are also tested in order to help understand the patient's genetic variants. A test report will be issued only for the patient with the medical condition. If a relevant variant is identified in a family member who was included in the test, this information will be in the report.

### Limitations:

Diagnosis using WES has limitations. The WES test is not designed to identify all types of DNA changes. Variants may not be detected due to technical problems. In addition, misinterpreting the meaning of a variant may cause mistakes. Limitations include:

- Disease causing genetic variants that are not found in the exome will be missed.
- The *UCSF Exome Sequencing* test does not study every part of the exome.
- Technical accuracy and reliability of results depends on the quality of the sample. Laboratory errors can occur.
- Determining whether or not a variant causes a medical condition is based on current medical and scientific knowledge, and on the clinical information provided by you and your health care provider. Testing may not find a genetic cause even when one exists. Information found in the exome may be reanalyzed in the future at the request of your health care provider.
- If a causative variant is identified, this information may or may not be useful in predicting disease outcome or in guiding treatment.

### Risks:

WES has the following potential risks. Please discuss these risks with your health care provider or genetic counselor:

- Sometimes people feel anxiety about learning uncertain genetic information about themselves or their family.
- Information about family relationships can be learned during testing. Testing may reveal unexpected information about blood-relatedness. It may reveal situations where a father has no biological relationship with a child.
- The Genetic Information Nondiscrimination Act (GINA) is a federal law that protects patients against employment and health insurance discrimination. That means that employers and health insurance companies cannot use genetic information when making hiring or coverage decisions. The law has limits, however. Life insurance, long-term care insurance, and disability insurance, are not included in the law's protections. GINA does not cover those serving in the military. Some state laws also include protection against genetic discrimination but safeguards are not complete.
- The laboratory saves leftover DNA samples after completing the test. These samples are used for improving the test and for educational purposes. To protect your privacy, these samples are "de-identified," meaning they are not stored with your name or other personal information.

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- Only de-identified information from testing is included in scientific presentations and publications in the medical literature.
- UCSF will share your de-identified results with secure scientific databases. The purpose of sharing is to improve the medical community's understanding of the relationship between genetic variants and medical conditions.
- UCSF takes many steps to ensure that your personal health information is kept private. However, unforeseen events could result in a loss of privacy.

### **Additional findings:**

In WES testing, variants in genes that are not related to your medical condition may be found. These findings may have medical significance. They are called "additional" findings. For example, a gene that increases risk of cancer or heart disease may be identified. Such findings are uncommon but may be important to your or your family's health care. UCSF is following guidelines issued by a professional association, the American College of Medical Genetics and Genomics, recommending that additional findings should be offered to patients. However, as knowledge in genetics is constantly changing, variants in additional medically significant genes may also be identified during testing. You will be offered results the UCSF team believes to be medically significant. You have the option of choosing not to learn any additional findings, if you only want information about the primary medical condition. Please note your choice below.

Additional findings will be evaluated in the patient for whom the *UCSF Exome Sequencing* test was ordered. If family members were included in the analysis, these family members will only be evaluated for the specific additional variant(s) found in the patient. The family members will not have all variants in their genes independently assessed.

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### Agreement to Testing:

I have reviewed, or someone has reviewed with me, the information about the *UCSF Exome Sequencing* test provided in this consent form. I have been given the opportunity to ask questions. By signing this form, I agree to testing, or to testing of my minor child. (*Signatures are required from each person to be included in the testing.*)

### Additional Findings:

YES. I do want information about medically significant additional findings to be included in the report.

NO. I do not want information about medically significant additional findings to be included in the report.

Patient Printed Name: \_\_\_\_\_

**Signature:** \_\_\_\_\_ **Date:** \_\_\_\_\_

If signed by someone other than the patient (such as a parent or guardian), please indicate your name and relationship to the patient:

Printed Name: \_\_\_\_\_ Relationship to Patient: \_\_\_\_\_

**Signature:** \_\_\_\_\_ **Date:** \_\_\_\_\_

### Family Members Included in the Exome Sequencing Test (add additional pages if necessary):

Printed Name: \_\_\_\_\_ Relationship to Patient: \_\_\_\_\_

**Signature:** \_\_\_\_\_ **Date:** \_\_\_\_\_

Printed Name: \_\_\_\_\_ Relationship to Patient: \_\_\_\_\_

**Signature:** \_\_\_\_\_ **Date:** \_\_\_\_\_

**Physician/Genetic Counselor statement:** I have discussed the UCSF Exome Sequencing test to this patient/family. I have explained the test limitations as outlined in the consent form and have answered questions about the test.

Physician's or Genetic Counselor's Name: \_\_\_\_\_

**Signature:** \_\_\_\_\_ **Date:** \_\_\_\_\_