ARUP LABORATORIES | NATIONAL REFERENCE LABORATORY

A nonprofit enterprise of the University of Utah and its Department of Pathology

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INFORMED CONSENT FOR EXOME SEQUENCING

Patient Name _____ Date of Birth _____ Sex ☐F ☐ M

Symptoms ☐ No ☐ Unknown ☐ Yes (please describe) _____

If this individual is a parent of a child being tested, provide child's name: _____

Test Description/Purpose

Genes hold the DNA code for making proteins. Exome sequencing involves examining the DNA code of ~19,000 genes. Thousands of DNA variants are detected. Some variants are disease-causing while others are harmless or have an unknown effect. The purpose of the test is to identify the variant(s) causing the patient's suspected genetic disorder.

Ordering Considerations

Participation in exome sequencing is voluntary. Genetic counseling is required prior to, as well as following, this complex test.

Parental samples are critical to the interpretation of the patient's results. A cause for the patient's medical issue(s) is determined in ~45% of cases when both parents undergo exome sequencing, in ~35% of cases when only targeted sequencing of parental samples is ordered, and in ~20% of cases when parental samples are not provided.

Exome sequencing results may provide unexpected information, such as:

- Identifying a genetic risk unrelated to the original reason for testing.
- Predicting another family member has, is at risk for, or is a carrier of a genetic condition.
- Revealing non-paternity (the person stated to be the biological father is not, in fact, the biological father).
- Suggesting the parents of the individual tested are blood relatives.

If a disease-causing genetic variant is identified, insurance rates, the ability to obtain disability and life insurance, and employability could be affected. The Genetic Information Nondiscrimination Act of 2008 extends some protections against genetic discrimination (http://www.genome.gov/10002328). All test results are released to the ordering healthcare provider and those parties entitled to them by federal, state, and local laws.

The American College of Medical Genetics and Genomics (ACMG) recommends reporting disease-causing variants in certain cancer, cardiovascular and other genes, even if they are not related to the patient's condition because monitoring or early treatment may be available. Other medically actionable incidental variants in non-ACMG genes may be reported at ARUP's discretion. If a patient or family member has symptoms of a condition related to an ACMG-recommended gene, separate testing should be ordered, as coverage of the ACMG genes is incomplete.

Initial here _____ if findings or variants detected in ACMG genes, or other actionable genes should NOT be reported.

Limitations and Reporting of Results

- Often exome sequencing is not able to identify the cause of a patient's medical issues. This does not exclude the possibility that the patient has a genetic condition. Some disease-causing variants are in genes with unknown function while others are in areas not tested (between genes or within the non-coding regions of genes). Still others are in genes that are not possible to analyze with this testing method.
- Variants related to the patient's medical issues are reported. De novo variants (not inherited from either parent) or variants inherited from both parents in the same gene may be reported even if the function of the gene is unknown. Because genetic knowledge continues to advance at a rapid pace, interpretation of the result may change in the future. If the report is amended, the patient's provider will be contacted with an updated report.
- Variants unrelated to the patient's medical condition are not reported except disease-causing ACMG variants, or possibly other actionable variants, if elected on this consent form.
- Exome sequencing may fail to detect variants in ACMG-recommended genes. Only disease-causing variants in ACMG genes identified with routine exome analysis are reported. Single disease-causing variants in recessive ACMG genes are not reported. Family members undergoing exome sequencing who wish to receive their own ACMG variant report must complete an exome consent form to receive a separate report.
- Results are typically reported in 4-8 weeks.
- Although genetic test results are usually accurate, several sources of error are possible, including: clinical misdiagnosis of a condition, inaccurate information provided regarding family relationships, sample mislabeling or contamination, transfusion, bone marrow transplantation, and maternal cell contamination of prenatal or cord blood samples.

INFORMED CONSENT FOR EXOME SEQUENCING

Access to Sequence Data / Data Sharing / Sample Storage

- ARUP Laboratories will have access to the patient's sequence data from exome testing. Your health care provider and the hospital that submitted the test to ARUP can also request a copy of the sequence data.
- Because ARUP is not a storage facility, most samples are discarded after testing is completed. Some samples may be stored indefinitely for test validation or education purposes after personal identifiers are removed. All New York samples are discarded 60 days following test completion. You may request disposal of your sample by calling ARUP Laboratories at (800) 242-2787, ext. 3301.
- In cooperation with the National Institutes of Health's effort to improve understanding of specific genetic variants, ARUP submits HIPAA-compliant, de-identified (cannot be traced back to the patient) genetic test results and health information to public databases. The confidentiality of each sample is maintained. If you prefer that your test result not be shared, call ARUP at (800) 242-2787, ext. 3301. Your de-identified information will not be disclosed to public databases after your request is received, but a separate request is required for each genetic test. Additionally, patients have the opportunity to participate in patient registries and research. To learn more, visit www.aruplab.com/genetics/resources.

Patient/Legal Guardian: I authorize ARUP Laboratories to perform exome sequencing on my (or my child's) sample. The risks, benefits, and limitations have been explained to my satisfaction by a qualified health professional.

Patient/Guardian Printed Name	Signature	Date

Ordering Healthcare Provider: I have explained exome sequencing, including its risks, benefits and alternatives to the patient or legal guardian, and answered all their questions.

Healthcare Provider Printed Name	Signature	Date

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