ARUP LABORATORIES
*A nonprofit enterprise of the University of Utah and its Department of Pathology*

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# INFORMED CONSENT FOR RAPID MENDELIAN GENES SEQUENCING PANEL, TRIO

**Patient Name:** _____ **Date of Birth:** _____ **Sex**: ☐ Female ☐ Male

**Symptoms**: ☐ **No** ☐ **Unknown** ☐ **Yes** (describe): _____

**Test Description and Purpose**

The Rapid Mendelian Genes Sequencing Panel, Trio test involves decoding the DNA sequence of approximately 4,900 genes known to cause disease in humans. The purpose is to determine the cause of the patient's medical condition.

**Ordering Considerations**

- Participation in genetic testing is completely voluntary. Genetic counseling is required prior to and following this complex test.
- Because this test targets greater than 4,900 Mendelian genes, thousands of DNA changes (variants) are detected. These variants may be harmless, disease-causing, or have an unknown effect. It may be unclear whether a specific variant identified is contributing to or causing the patient's symptoms.
- This test may identify the cause of an infant's medical condition in approximately 50 percent of cases. The chance of detecting the cause of a condition in older individuals is less.
- Parental samples are required to identify new (de novo) variants present in their child that are not present in either parent. If a healthy parent carries the same variant identified in their affected child, this often decreases the chance the variant is disease-causing. If the variant is de novo, the likelihood that the variant is disease-causing is increased.
- Genetic testing results may provide information that was not anticipated, 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.
- The American College of Medical Genetics and Genomics (ACMG) recommends disease-causing variants in certain cancer, cardiovascular and other genes be reported, 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 has symptoms of a condition related to an ACMG-recommended gene, separate testing should be ordered as coverage of the ACMG genes is incomplete. Parental ACMG variants will not be reported.

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

- 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, and sample mislabeling or contamination.
- If a 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 (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.

**Limitations of the Rapid Mendelian Genes Sequencing Panel, Trio Test**

- If the test is unable to identify the cause of a patient's medical issues, this does not exclude the possibility that the patient has a genetic condition.
- The test does not detect all variants causative for genetic disease as there are about 19,000 genes, but only the targeted Mendelian genes known to be associated with human disease are analyzed.
- Some disease-causing variants in the Mendelian genes analyzed may not be detected as they may lie in the noncoding DNA, occur within repetitive sequences, or reside in other complex regions that are difficult to analyze.

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**Reporting of Results**

- Results are typically reported within 14–28 days. All detected disease-causing variants that may be related to the patient's medical issues will be reported.
- Disease-causing variants, unrelated to the patient's symptoms, will NOT be reported unless they are in a gene included on the ACMG's list of recommended genes or are considered medically actionable variants and are desired as indicated above.
- Because genetic knowledge continues to advance at a rapid pace, the interpretation of results may differ in the future. If your report is amended, your healthcare provider will be contacted and provided a copy of the updated report.

**Test Improvement**

- In cooperation with the National Institutes of Health's effort to improve understanding of specific genetic variants, ARUP submits HIPAA-compliant, deidentified (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 deidentified 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 aruplab.com/genetics/resources.
- 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 at 800-242-2787 ext. 3301.

**Patient/Legal Guardian**: I authorize ARUP Laboratories to perform the Rapid Mendelian Genes Sequencing Panel, Trio test on my, or my child's sample. The benefits, risks, and limitations of this testing have been explained to my satisfaction by a qualified health professional.

| Patient/Guardian | Signature | Date |
|---|---|---|

**Ordering Healthcare Provider, Genetic Counselor**: I have explained this genetic test and its risks, benefits, and alternatives to the patient or legal guardian and addressed all their questions.

| Provider/Genetic Counselor Printed Name | Signature | Date |
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