DOS

Lexington, Kentucky

MEDICAL EXOME TESTING CONSENT

Clinical Molecular Genomic Pathology Laboratory, Department of Pathology and Laboratory Medicine

(Patient Label Here)

Test Information

Your health care provider recommends that you or your child have a Medical Exome Test. This tests about 4,800 genes for differences (variants) that cause the specific disease or condition you are being tested for. Finding the variant(s) involved may help diagnose and treat you or your child. It may also let family members who are at risk to be tested. In some cases, it is not certain if a variant found is actually causing the disease.

Getting the Test

If you consent to this test, we will take a sample of your or your child's blood or buccal (cheek swab) specimen. We then send it for testing at the Genomics Laboratory of UK HealthCare, where the DNA will be removed from the blood and a Medical Exome Test performed. Another sample may be needed for testing but this is rare.

Test Results

The results of this test will fall into one or more of the following possibilities:

1. **Positive:** A positive result means that a pathogenic or likely pathogenic (disease-causing) genetic variant was found that explains the cause of a person's symptom(s), health history or that the person is at increased risk for this condition in the future. A patient may test positive for more than one genetic variant.
2. **Negative:** A negative result means that no disease-causing genetic variant known to be related to a person's symptoms or health history was found by the test. A negative result does not mean that the person is completely free from all genetic disorders or risks.
3. **Inconclusive/Variant of Unknown Significance (VUS):** A result of a variant of unknown significance (VUS) means that the test found a genetic variant but that it is not now known if this variant is related to the patient's genetic disorder. In some cases, we may need more information, such as genetic and/or medical information from other family members -- to see if a VUS is disease-causing or not. If this is the case, your health care provider will discuss this with you.
4. **Secondary Finding:** Secondary findings are pathogenic or likely pathogenic variants found by the test that are important for a person's health care but unrelated to the reason for ordering the test. You must check the box next to "I wish to receive Secondary Findings" on page 2 to get these results. Please see the section on secondary findings below.

Test Report

Variants found in the Medical Exome Test will be reported if they are pathogenic, likely pathogenic, or variants of unknown significance in genes known to be related to your or your child's symptoms, health history, or risk for a condition. Only pathogenic and likely pathogenic secondary findings will be reported if your select this option.

Parental and Familial Testing

It may be helpful to test other family members. The Medical Exome test can be performed as a TRIO test, using the DNA of a person and their parents. Your health care provider will discuss this with you if parental or familial testing could help interpret the test results.

Test Limitations

- Genetic testing is highly accurate. Inaccurate results happen but they are rare. They can be caused by technical limitations such as: high guanine/cytosine content genomic sequence, poor or non-probe coverage, sample contamination, or inaccurate clinical information.
- Genetic testing may also lead to inaccurate results in patients who have had bone marrow transplants or who have variants in only small percentage of their cells (a condition called mosaicism).
- Accurate interpretation of Medical Exome test results relies on the correct reporting of biological relationships. To prevent inaccurate interpretation of results, it is important to share how family members are related biologically (or not) to the person being tested.

Test Risks

The Medical Exome test has some risks, including:

1. **Physical:** Taking a blood sample for DNA testing carries the risks normally of a blood draw. These include mild pain and/or bruising at the site, skin redness, and a slight risk of infection. Sometimes the blood draw can cause light-headedness.
2. **Mental:** Testing may cause patients or family members to feel anxiety or stress.
3. **Family Relationships:** Genetic testing may reveal that the biological relationships within a family are different from the history provided or that the parents of someone are related by blood (consanguinity).

HealthCare

Lexington, Kentucky

MEDICAL EXOME TESTING CONSENT

Clinical Molecular Genomic Pathology Laboratory, Department of Pathology and Laboratory Medicine

(Patient Label Here)

4. **Insurance:** In some cases, a positive test may result in different treatment (such as denial of coverage or increased cost) from certain types of insurance plans. Please refer to information on the Genetic Information Nondiscrimination Act (GINA) for more information.

Confidentiality

The confidentiality of your test results is protected by HIPAA. Providers can only share your results for diagnosis and treatment purposes only.

Sample and Data Retention

Your DNA sample will be stored in the laboratory for at least 2 years. Sequencing data will be kept in the laboratory for at least 2 years. Final reports will be stored in laboratory for at least 10 years.

Withdrawal of Consent

You may withdraw your consent at any time before the laboratory processing starts. If you want to withdraw consent for Medical Exome testing, your provider will need to contact the laboratory immediately. You cannot withdraw consent once laboratory processing has started.

Information on Secondary Findings

Since many genes are analyzed in the Medical Exome test, these tests may reveal some findings that are not directly related to the reasons for ordering the test. These findings can provide information that was not expected. Such findings are called "incidental findings" or "secondary findings (SF)."

The American College of Medical Genetics and Genomics (ACMG) publishes a list of medically actionable genes associated with various inherited disorders. It recommends that secondary findings in these genes be reported to the patients and health care providers.

Patients must "opt in" to receive these secondary findings. To opt in, check the box next to "I wish to receive Secondary Findings" below.

The absence of reportable secondary findings for any particular gene does mean that a genetic disorder or risk of related conditions do not exist.

NOTE: Interpretive services must be offered for preferred languages other than English.

Signatures

Patient Consent

q I have read this Medical Exome test consent document and I give permission for Medical Exome testing to be performed as described.

Patient _____ Date _____

Legal Representative and Relationship to Patient _____ Date _____

Provider Signature _____ Date / Time _____

q I wish to receive Secondary Findings

Legal Representative and Relationship to Patient _____ Date _____

Interpreter Name or ID # _____ In person or via Cyracom (circle one)