I understand that my blood sample will be collected for the purpose of Whole Genome Sequencing (WGS). If I agree to have WGS performed on the blood sample, I will sign the last page of this form.

I understand that because of the complicated nature of this testing, and the information that might be learned, it is recommended that I or my family meet with a genetic counselor based on the results of the testing. Genetic counseling is an additional resource to answer questions and help me understand the results of the testing and information that may be learned.

DESCRIPTION OF THE TESTING

The purpose of this testing is to try and identify the reason for my symptoms. This test will look at all of my genes and complete DNA sequence at the same time.

I understand that DNA will be extracted from my blood. DNA is the genetic material in our cells that makes up our genes, which are the recipes for making us who we are. Everyone's genes are different and cause differences between people such as eye color, hair color and blood type. Genetic differences can be the reason why some people get certain diseases and others don't.
My DNA will be sequenced and then compared to the DNA of other family members (if possible) and thousands of unrelated "control" individuals to look for differences that could explain my symptoms. I understand that multiple family members may need to be tested in order to increase the chances of finding a genetic difference that is causing my symptoms. I understand that if additional family members are not available for testing it may lower the chance that the genetic difference that explains my symptoms will be able to be identified by this test.

Once the genetic differences between my family members and me are identified, a group of scientists in the Center for Pediatric Genomic Medicine at Children's Mercy Hospital will determine which, if any, are related to my disorder. A detailed written report discussing the possible disease-causing genetic differences will be given to my doctor. I understand that any other family members who are tested will not receive a separate report.

LIMITATIONS OF THE TESTING

The WGS test attempts to look at the sequence of my entire DNA; however the lab machines are sometimes not able to look at all of the sequence. I understand that usually only a part of the genome is analyzed (the "exome") and about 97% of this will have enough data to be examined by the scientists in the Center for Pediatric Genomic Medicine. Usually the missing parts are small portions of many different areas, rather than an entire gene sequence. I understand that this is part of the reason that WGS cannot find a diagnosis for every person.

I understand that even if this test finds genetic differences that are responsible for my symptoms, the testing will not completely predict the severity of my disease, possible future problems, or response to treatment.

RESULTS

I understand that WGS will find millions of genetic differences, called variants, in all people, including me. These variants fall into several categories and not all of them will be included in the report given to my doctor.

Variants reported -

- Variants in genes that may be related to my symptoms.
- Variants in genes that are not related to my symptoms but may cause additional medical problems as an adult WILL BE provided in the report only if I choose this option–see section called "Incidental Findings" for more information.

Variants NOT reported –

- Variants known to be common harmless differences found in many healthy people.
- Variants that may be linked to a small increase or decrease in the risk to develop certain common diseases such as diabetes, high blood pressure, or arthritis.
- Variants in genes which are not known to cause disease in humans. These variants may be re-evaluated in the future if my child's doctor requests re-evaluation of the data.

I understand that WGS may discover that relationships between family members were unknown or were reported incorrectly to my doctor. While this type of discovery (such as non-paternity) will not be specifically noted in the report, it may be apparent from the report.

I understand that this report will be placed in my medical record and will be available for review by my doctors in the future.

In providing this Consent for Whole Genome Sequencing, I understand that the sequencing data created as a result of this testing may be subject to reanalysis in the future. I understand that if I want to obtain or authorize the use and/or disclosure of the sequencing data, I will be required to complete an Authorization from the Center for Pediatric Genomic Medicine.

INCIDENTAL FINDINGS

I understand that WGS might identify genetic differences that are associated with disease(s) that are not related to my symptoms. These differences are called "incidental findings." I understand that I can choose whether or not to have my doctor receive these findings.

The American College of Medical Genetics and Genomics (ACMG) has recommended that incidental findings in 56 genes associated with certain inherited disorders should be offered to all individuals who have WGS. Some of these genes are associated with increased risks of developing tumors or cancer. Other genes on the list are associated with an increased risk of disease affecting the heart or blood vessels. There are also some genes on the list that are associated with other disorders, such as an increased risk for complications from anesthesia. I will need to decide whether you want to receive this information. My doctor will show me this list upon my request.

I understand that if I choose for my doctor to receive incidental findings, there may be implications for other family members. For example, if there is a genetic difference that indicates I am at an increased risk to develop cancer, it may mean that my brothers and sisters and my parents may be at an increased risk to develop cancer as well. I understand that additional family members will not receive a separate report with their own results, and may need to pursue clinical genetic testing through their physician if they are concerned about their own risk based on my incidental findings.

I understand that if I choose for my doctor to receive incidental findings, only findings in the ACMG list will be reported, and only if there is enough data. If no genetic differences are found in these genes, it does NOT mean that there are no disease-causing differences.

By signing below, I acknowledge that I understand the information described in this Informed Consent, including the risks, benefits and alternatives to Genome Sequencing. I have been given an opportunity to ask my questions about my child's condition and Genome Sequencing and I have sufficient information and understanding to give this informed consent.

PLEASE CHOOSE ONE BOX AND SIGN IF YOU AGREE TO WGS TESTING

☐ **Whole Genome Sequencing Without Incidental Findings**

By signing in this box, I agree to have the Children's Mercy Hospital Center for Pediatric Genomic Medicine perform whole genome sequencing on a sample of my blood. I DO NOT wish to have incidental findings for the ACMG recommended list of genes listed on the report provided to my doctor.

☐ **Whole Genome Sequencing With Incidental Findings**

By signing in this box, I agree to have the Children's Mercy Hospital Center for Pediatric Genomic Medicine perform whole genome sequencing on a sample of my blood. I DO wish to have incidental findings for the ACMG recommended list of genes listed on the report provided to my doctor.

Primary Care Physician: _____

Patient's Name: _____

Date of Birth (month/day/year): _____ Phone Number: _____

Patient's Address: _____

City: _____ State: _____ Zip Code: _____

Consenting Adult: _____ Date: _____ Time: _____

Witness: _____ Date: _____ Time: _____