Prenatal Symptom-Driven Exome Informed Consent

I understand that my prenatal medical team at Children's Mercy Hospital ("CM") has recommended that I consider undergoing a genetic test known as a Prenatal Symptom-Driven Exome ("Prenatal Exome Testing").

I understand that because of the complicated nature of Prenatal Exome Testing, and the information that might be learned, I will need to meet with a genetic counselor to talk about the results of the Prenatal Exome testing. Genetic counseling is an additional resource to answer questions and help me understand the results of the Prenatal Exome Testing and information that may be learned.

DESCRIPTION OF THE TESTING

Prenatal Exome Testing is a highly complex genetic test that aims to identify changes in a baby's DNA that might explain the physical differences identified on sonogram.

I understand that my baby's DNA will be extracted from either the amniotic fluid (collected during an amniocentesis) or the placenta (collected during a chorionic villus sampling or CVS). DNA is the genetic material in our cells that are the "instructions" (known as genes) that help make 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 do not.

My baby's DNA will be sequenced and then compared to the DNA of other family members and thousands of unrelated "control" individuals to look for differences that could explain the ultrasound findings. I understand that the baby's father and I will also need to provide a blood sample, and that if the baby's father does not provide a sample, it will lower the chance that the test will be able to find an answer for my baby's ultrasound findings.

Once the genetic differences between my baby and other family members are identified, a group of scientists in the CM Center for Pediatric Genomic Medicine will determine which, if any, are related to my baby's ultrasound findings. A detailed written report discussing the possible disease-causing genetic differences will be given to my prenatal medical team. I understand that any other family members who are tested will not receive a separate report.

I understand that any DNA remaining after Prenatal Exome Testing is complete may be stored by CM indefinitely.

LIMITATIONS OF THE TESTING

Prenatal Exome Testing examines many genes at the same time, looking at the parts of genes called "exons." Exons are the parts of genes that are best understood by scientists and doctors, and changes in the exons are thought to be the cause of most genetic disorders. However, some genetic conditions are caused by changes outside the exons and those changes might not be detected by this test. I understand that sometimes the lab machines are not able to look at all of the DNA sequence, although typically more than 97% of the exons have enough data to be examined by the scientists in the CM 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 Prenatal Exome Testing cannot find a diagnosis for every baby.

Prenatal Exome Testing is not currently able to detect every type of change associated with genetic disorders, such as repeat expansions or methylation changes. In addition, scientific knowledge about the function of all genes is incomplete and will change over time.

I understand that even if Prenatal Exome Testing finds genetic differences that are responsible for my baby's symptoms, Prenatal Exome Testing will not completely predict the severity of my baby's disease, possible future problems, or response to treatment.

RESULTS

I understand that this Prenatal Exome Testing will find many genetic differences in all people, including my baby. These differences fall into several categories and not all of them will be included in the report given to my prenatal team.

INFORMATION THAT IS NOT PROVIDED WITH PRENATAL EXOME TESTING –

- Differences that are known to be common harmless differences found in many healthy people.
- Differences 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.
- Differences and/or genes which are too poorly understood at the current time to be evaluated completely. These are called "variants of unknown significance" or "genes of unknown significance." If this Prenatal Exome Testing does not find a clear explanation for my baby's ultrasound findings, these variants may be reported after delivery (see the Reanalysis section for more details).
- Differences in genes that cause medical problems as an adult.
- Differences in my and/or my partner's DNA that may be medically relevant to us but are not relevant to my baby's ultrasound findings.

INFORMATION THAT WILL BE PROVIDED WITH PRENATAL EXOME TESTING -

- Medically relevant differences in genes that may be related to my baby's ultrasound findings.
- Medically relevant differences in genes that may not be related to my baby's ultrasound findings, but could impact their medical care during childhood.

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

I understand that this report will be placed in my prenatal medical record and will also be copied to my baby's medical record after delivery. I understand that should I need to have the raw sequencing data sent to a non-CMH provider, I must specifically request the release from the Center for Pediatric Genomic Medicine, and that the data will not be released without my written permission.

REANALYSIS

I understand that if my baby's Prenatal Exome Testing does not find a genetic diagnosis, I have the option of having the Prenatal Exome Testing reanalyzed under the following conditions:

- If physical exam, imaging, or testing after delivery reveals additional or different medical issues. There will be an additional charge for this type of reanalysis and the report will include variants of unknown significance.
- If at least a year has passed since my baby's Prenatal Exome Testing was completed and there are still concerns about the possibility of a genetic disorder as an explanation for medical issues. There will be an additional charge for this type of reanalysis and the report will include variants of unknown significance.
- If my baby does not survive until delivery and is unable to be examined for additional issues, a list of the variants of unknown significance can be added to the original report. There is no charge for this type of reanalysis.

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

_____		____/____/____	_____ hours
Signature of Patient/Parent/Legal Guardian	**Printed Name/Relationship**	**Date**	**Time**
_____		____/____/____	_____ hours
Witness to Signature	Printed Name/Relationship	Date	Time

Telephone and Interpreter Consent: **STAFF USE ONLY**

I read the above statement to _____, reached at (___)____-_____ at ____ hours; he/she stated understanding and approval.

_____		____/____/____	_____ hours
Signature of 1st Witness	Printed Name	Date	Time
_____		____/____/____	_____ hours
Signature of 2nd Witness	Printed Name	Date	Time
_____		____/____/____	_____ hours
Signature of Interpreter			