UNIVERSITY OF WASHINGTON
CONSENT FORM
Leftover Fetal Samples
MOLECULAR ANALYSIS OF GENETIC NEURODEVELOPMENTAL DISORDERS

Researchers:

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24-hour emergency telephone number: 1-866-987-2000 (within Washington, Wyoming, Alaska, Montana and Idaho) Seattle Children's Hospital Switchboard, Seattle, Washington. Please ask the operator to page Dr. Doherty or the physician on call for Medical Genetics. If outside these states, place a collect call to 206-987-2000.

Researchers' Statement:
We are asking you to be in a research study. The purpose of this consent form is to give you the information you will need to decide whether to be in the study or not. Please read the form carefully. You may ask questions about the purpose of the research, what we would ask you to do, the possible risks and benefits, your rights as a volunteer, or anything else about this research or this form that is not clear. When we have answered all your questions, you can decide if you want to be in the study or not. This process is called "informed consent." We will give you a copy of this form for your records.

PURPOSE OF THE STUDY

We are asking your fetus to be in a research study because your fetus, or someone in your family, has a neurodevelopmental (NDV) disorder. NDV disorders affect the brain, spinal cord, muscles, and other organs. We expect that our studies will take many years to complete. We may never find the specific genetic change responsible for the disorder in your family. During the course of this research, it may be beneficial to share samples and data with other researchers who use additional techniques that will move the research forward more quickly. The goals of our research are to:

- Identify specific brain malformation conditions and other NDV disorders
- Determine how people with specific NDV disorders do over time ("natural history")
- Identify the genetic causes of NDV disorders
- Determine the biological mechanisms of NDV disorders

Your fetus, or someone in your family, may have one of the following disorders (check one):

☐ Joubert syndrome and related disorders (JSRD)

☐ Differences of the mid-hindbrain (the back part of the brain)

☐ Differences of the forebrain (front of the brain) and cortical brain development (outer part of the brain), including developmental delay, intellectual disability, autism, and seizures

We will study your fetus's leftover genetic material, called DNA, to understand the causes of these disorders. We will use DNA from samples left over from the clinical care of your pregnancy. We do not perform clinical prenatal genetic diagnosis in our research laboratory. We will not test any leftover fetal samples until we know that the pregnancy has ended to ensure that the research does not influence decisions about prenatal testing or termination of pregnancy. We expect that our studies will take many years to complete. We may never find the specific genetic change responsible for the disorder in your family.

STUDY PROCEDURES

Participation consists of information collection and sample collection.

Information Collection

Dr. Doherty and his research team will collect information about your pregnancy from phone or email interviews and your medical records, which may include the following:

- Medical history and exams
- Prenatal imaging results
- Photographs
- Genetic and other test results

We will use this information to confirm, or sometimes re-classify, your fetus's NDV disorder. We will compare your medical information with information from other people with NDV disorders. Medical information from many people with NDV disorders is helpful for understanding these disorders. Therefore, we plan to keep your fetus's information until the end of this study.

We will also ask questions about your family history. You can decide not to answer any question.

Sample Collection

We will collect leftover DNA samples from chorionic villus sampling (CVS), amniocentesis, umbilical cord blood sampling and/or autopsy (miscarriage, stillbirth, or pregnancy termination). These samples will be collected after all clinical needs have been met and when the samples are ready to be thrown away. We will not test any leftover fetal samples until we know that the pregnancy has ended. We may test fetal tissues from autopsy to help understand the causes of NDV disorders. If samples were donated to another research study, it may be possible for us to share these samples.

We will study these samples in a laboratory and look at your fetus's DNA. If the genetic tests find a difference in your fetus's gene(s), we will study this gene(s) to help us understand how it caused your fetus's NDV disorder. An immortal cell line may be created so we can continue to study your fetus's DNA indefinitely.

We will keep your fetus's DNA until the end of this study and may send small samples of your fetus's DNA or growing cells to other researchers. However, any samples sent to them will be labeled with a code that cannot be traced back to your fetus. Future research with your fetus's information and DNA may include topics that are unrelated to your fetus's disorder.

Genetic Analysis

In our search for genes that cause or influence NDV disorders, we may perform several tests on your fetus's DNA. Usually, we study just the parts of a genetic code that are linked to a disease or condition. This is called "genotyping and targeted gene sequencing." Other tests look at much larger areas such as parts of the DNA that code for proteins. This is called "whole exome" sequencing. In "whole genome" studies, all or most of your fetus's genes would be analyzed. We cannot promise any direct benefit to you personally, though some people might find satisfaction in contributing to scientific knowledge about genetic conditions.

Optional Participation
There are other OPTIONAL choices to taking part in this research. You will have the option to say YES or NO to each of these at the end of this form.

1. Photography (Optional)
If you agree, pictures of your fetus may be taken during an autopsy. These pictures will be used to help us understand your fetus's clinical findings.

2. Re-contact (Optional)
If you agree, we may contact you in the future to learn about any changes in your family history. We may also want to tell you about new research studies.

3. Results Reporting (Optional)
We may find the genetic cause of the NDV disorder in your family. If you want to know these results, we can contact you to tell you this information. Receiving results is your personal choice. You do not have to receive results. If you choose to know these results, genetic counseling will be recommended, and we can help you arrange this. We will only report clinically significant results. Research results should be confirmed in a clinical laboratory.

4. Genomic Data Sharing (Optional)
The National Institutes of Health (NIH) has developed data (information) banks that collect study data. If you agree, the NIH will store de-identified information from this study in these banks for other researchers to use in future studies on any topic. The researchers could be from government, academic, or commercial institutions. The information from this study may be stored in a public unrestricted data bank that anyone can use. This information will not include your name or other information that could identify you. You will not receive any results from allowing your data to be placed in the NIH data banks. You can withdraw your consent any time you decide you do not want your data in the NIH data banks. There will be no consequences for withdrawing consent. There will be no direct benefit to you if you agree to participate in Genomic Data Sharing.

RISKS, STRESS, OR DISCOMFORT

Information Collection
You may feel badly when discussing your fetus's NDV disorder. You may feel uneasy sharing your medical, pregnancy, or family history. If you do feel uneasy, you may refuse to answer any question.

Sample Collection
Giving a leftover sample to research does not cause any more pain than that associated with clinical care. Your decision to give fetal tissue may cause you emotional stress.

Optional Participation

1. Photography (Optional)
We will try to hide features on a picture that would allow someone to recognize your fetus before scientific publication. Rarely, we will ask for your permission to publish a specific recognizable picture to show features of the disorder in a publication. If someone recognizes your fetus, the confidentiality of participating in the research study may be broken. Once the photo has been published. It may be shared with additional organizations for educational purposes if the journal has policies to allow this.

2. Re-contact (Optional)
You may not want to answer more questions about our research study. You can say no and ask not to be re-contacted in the future.

3. Results Reporting (Optional)

Since we do not perform prenatal diagnosis, results will not be available to you during pregnancy. After the pregnancy, if we find the genetic cause of your fetus's NDV disorder, or that you or your fetus is an unaffected carrier for an NDV disorder, you may feel anxious or stressed. Although we may report research results to you, it is up to you and your doctor to decide whether these results should be included in your or your fetus's medical files. If you results were shared with a third party, this could affect your employability or your ability to get certain types of insurance. Rarely, our testing reveals that the man thought to be the father of a child is not the biological father. Should we find this type of information, we will NOT disclose it to you or others.

4. Genomic Data Sharing (Optional)

It is possible that your genomic information could be used to identify you or your close biological relatives when combined with information from other public sources, but we believe this is unlikely to happen. The current risk of this happening is very small but may grow in the future as new technologies are developed. Theoretically, someone could use this information to learn something about your health or genetic heritage. If linked to a medical condition and inappropriately shared with someone, it could affect your ability to get or keep some kinds of insurance. There is a possibility that this information could affect family members because certain conditions and traits run in families. This could hurt family and other relationships. There may also be other risks of re-identification that are unknown at this time.

ALTERNATIVES TO TAKING PART IN THIS STUDY

You do not have to join this study. If you do not join, your medical care during your pregnancy or in the future will not be affected.

BENEFITS OF THE STUDY

If you agree to be in this study, you may or may not benefit. The study may produce information that is helpful to doctors and to patients with NDV disorders. It is possible that the genetic cause of your or your family's disorder will be found. These results may help with your medical care or reduce the stress associated with uncertainty. If we find that your fetus is not a carrier for an NDV disorder, you may feel a sense of relief. We may provide you with specific information about your child's risk to pass on this disorder to his/her offspring.

SOURCE OF FUNDING

The study team and/or the University of Washington receives financial support from the National Institutes of Health, University of Washington Department of Pediatrics, and private non-corporate donors.

CONFIDENTIALITY OF RESEARCH INFORMATION

All of the information you provide is confidential. The study records will not be used to put you at legal risk. We have a Certificate of Confidentiality from the federal National Institutes of Health. This helps us protect your privacy. The Certificate means that we do not have to give out identifying information about you even if we are asked to by a court of law. We will use the Certificate to resist any demands for identifying information.

We cannot use the Certificate to withhold your research information if you give your written consent to give it to an insurer, employer, or other person. Also, you or a member of your family can share information about yourself or your part in this research if you wish.

There are some limits to this protection. We will voluntarily provide the information to:

- a member of the federal government who needs it in order to audit or evaluate the research;
- individuals at the University of Washington, the funding agency, and other groups involved in the research, if they need the information to make sure the research is being done correctly;
- the federal Food and Drug Administration (FDA), if required by the FDA;
- Washington state authorities, if we learn of child abuse, elder abuse, or the intent to harm yourself or others.

REQUIREMENTS ABOUT YOUR RESEARCH DECISION

We will obtain your consent to donate fetal tissue only after you have already provided written consent to have an abortion. Your research consent will not be obtained by the same person who obtained your abortion consent. Being in this research will not affect the method used for your abortion.

No payment, other financial benefits, gifts, or incentives can be provided to you for having an abortion or for donating the tissue for research.

OTHER INFORMATION

Taking part in research is always a choice. If you decide to be in the study, you can change your mind at any time. If you take part, we will try to keep your fetus's information private. We will store all of your fetus's records in locked cabinets and secure computer files. We will label your fetus's information with a study number. The master list that links a person's name to their study number will be stored in a locked cabinet or in a secure computer file. If results of this research are published, we will not use information that identifies your fetus unless you agree.

Government or university staff sometimes reviews studies such as this one to make sure they are being done safely and legally. If a review of this study takes place, your fetus's records may be looked at. The reviewers will protect your fetus's privacy. The study records will not be used to put you at legal risk

We plan to keep your fetus's medical information and samples until the project is completed, which is likely to be years or decades. You may refuse to participate or withdraw from the study at any time without penalty or loss of benefits that you deserve.

Neither you nor your insurance company will be billed for any study procedures. You will not receive any money if you take part in this study. Future research using your fetus's sample may lead to the development of commercial products (although this is not a goal of the research). You will not share in any profits that this research may produce.

RESEARCH-RELATED INJURY

If you think you suffered an injury or illness related to this study, contact the study staff right away. We will recommend or refer you for necessary medical treatment at a UW Medicine facility or an institution where you live. The costs of the treatment may be billed to you or your health insurance just like other medical costs, or it may be covered by the UW's discretionary Human Subjects Assistance Program (HSAP), depending on a number of factors. The researcher may request HSAP coverage by following established procedures. If you wish to request HSAP coverage for yourself, contact the researcher or the UW Human Subjects Division at hsdinfo@uw.edu or (206) 543-0098. You may also call collect to the UW Human Subjects Division at (206) 221- 5940 if you do not otherwise have access to a telephone. Ask the researcher if you would like information about the limits and conditions of the HSAP. The UW does not normally provide any other form of compensation for injury. However, the law may allow you to seek payment for injury-related expenses if they are caused by malpractice or the fault of the researchers. You do not waive any right to seek payment by signing this consent form. We will bill your health insurance for treating problems that result from your NDV

disorder or from standard clinical care. If you have no health insurance or your insurance refuses to pay, we will bill you.

Printed name of study staff obtaining consent	Signature	Date

Subject's statement

This study has been explained to me. I volunteer to take part in this research. I have had a chance to ask questions. If I have questions later about the research, I can ask one of the researchers listed above. If I have questions about my rights as a research subject, I can call the Human Subjects Division at (206) 543-0098. I give permission to the researchers to use my medical and pregnancy records as described in this consent form. I will receive a copy of this consent form.

Optional participation as described above: (Please circle YES or NO)

1. You may take pictures and use them for teaching purposes and scientific publication.	**YES**	**NO**
2. You may contact me for health updates and about future studies.	**YES**	**NO**
3. You may contact me with the results of research testing.	**YES**	**NO**
4. You may share my fetus's de-identified genetic data with NIH data banks.	**YES**	**NO**

Printed name of pregnant woman	Signature of pregnant woman	Date
Printed name of father of baby	Signature of father of baby	Date
Printed name of witness	Signature of witness	Date

Witness signature is required for: ☐ Subjects who are illiterate/educationally disadvantaged