

MAY 13 2013

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UNIVERSITY OF WASHINGTON

CONSENT FORM

Adults 18 years and older

Parents/Guardians of Children (less than 18 years)

Assent Form for Children Age 13-17

MOLECULAR ANALYSIS OF GENETIC NEURODEVELOPMENTAL DISORDERS

Researchers:

Ian A. Glass, M.D.	Professor, Dept. of Pediatrics	206-987-5142
Daniel Doherty, M.D., Ph.D.	Associate Professor, Dept. of Pediatrics	206-987-2590
William Dobyns, M.D., Ph.D.	Professor, Dept. of Pediatrics	206-884-1025
Heather Mefford, M.D., Ph.D.	Assistant Professor, Dept. of Pediatrics	206-543-9572
Jennifer C. Dempsey, M.P.H.	Research Coordinator, Dept. of Pediatrics	206-616-3788

Division of Genetic Medicine, Department of Pediatrics, School of Medicine  
206-987-2590, Toll-Free 1-800-246-6312

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 one of the physicians listed above 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 help you 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, or your rights as a volunteer. You may ask questions about anything else related to the 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 you to be in a research study because you, or someone in your family, have a neurodevelopmental (NDV) disorder. NDV disorders affect the brain, spinal cord, muscles, and other organs. We are doing this research study to answer two questions:

- What are the genes that cause NDV disorders?
- What are the functions of these genes?

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

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We would like to figure out whether differences in your genetic material, called DNA, cause these disorders. We expect that our studies will take many years to complete. We may never find the specific genetic change responsible for the disorder in you or your family. If we do find the specific cause, we may be able to develop better tests and treatments for this disorder in the future.

## STUDY PROCEDURES

If you join this study, there will be two parts: information collection and sample collection.

### Information Collection

Dr. Doherty and his research team will collect information about you from your medical records, including:

- Medical history and exams
- Photographs
- Brain scans, especially MRI scans
- Genetic and other test results

We will use this information to confirm, or sometimes re-classify, your 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 information for many years.

We will also ask questions about your family history. You can decide not to answer any question. Photos may be taken during a visit or you can send them by email or regular mail. This will also help us understand your disorder.

### Sample Collection

We will ask to collect a small sample of your blood (2-4 teaspoons) for our study. A cream, like LMX, may be used to lessen the pain of the needle. The blood will be used to obtain your DNA. In rare cases where a blood sample is not an option, DNA will be collected in other ways such as:

- A saliva (spit) sample (½ to 1 teaspoon)
- Previously collected and stored DNA sample ("banked" DNA)
- Previously collected and stored tissue (collected for another reason)
- Skin sample collected for this research study
- Blood or tissue sample left over after all clinical needs have been met

You can give the samples at the University of Washington, your local doctor's office, or a laboratory close to your home. We will help arrange to have the samples shipped to us at the University of Washington.

We will study these samples in a laboratory and look at your DNA. If the genetic tests find a difference in your gene(s), we will study this gene(s) to help us understand how it caused your NDV disorder.

We may send small samples of your DNA or growing cells to other qualified researchers. However, any samples sent to them will be labeled with a code that cannot be traced back to you.

### Genetic Analysis

In our search for genes that cause or influence NDV disorders, we may perform several gene searches on your DNA. Usually, we study just parts of your genetic code that are linked to a disease or condition. This is called "genotyping and targeted gene sequencing." Other gene searches 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 genes would be analyzed and used to study links to the disease under study. Even if your sample is whole genome sequenced, we cannot promise any direct benefit to you personally, though some people might find satisfaction in contributing to scientific knowledge about genetic problems and medical 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.

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### **1. Photography (Optional)**

If you agree, pictures and/or video clips may be taken during a visit or sent digitally by email. They will help us understand your clinical findings. Some pictures and/or video clips may include features that allow people to recognize you, such as your face. If we use these pictures and/or video clips for scientific publication, we will try to hide these features as much as possible before publication. Once the photo and/or video clip has been published, it may be shared with additional organizations for educational purposes if the journal has policies to allow this.

### **2. Recontact for Future Research Study (Optional)**

If you agree, we may contact you in the future to learn about any changes in your condition. 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. It may take several years to find this cause. 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, this will require genetic counseling. We will help you arrange for genetic counseling before and after the results are given to you.

### **4. Skin Biopsy for Establishment of Fibroblast Cultures (Optional)**

If you agree, we may ask you to provide a small skin sample. We get the skin by performing a minor procedure known as a "skin biopsy." The purpose of the skin biopsy is to get a small amount of skin to grow skin cells called fibroblasts. A doctor who routinely performs skin biopsies will explain the procedure to you. Your skin will be numbed with a special numbing cream before the skin is removed.

### **5. Participation in the Database of Genotypes and Phenotypes (Optional)**

dbGaP is the database of Genotypes (genes) and Phenotypes (observable qualities such as race, age, gender). In order to allow researchers to share test results, the National Institutes of Health (NIH) and other central repositories have developed special data banks that collect the results of genetic studies. The NIH and other data banks will store your genetic information and give it to other qualified researchers to do more studies. Qualified researchers that can access the national databases can be from the government, academic, or commercial institutions. Your genetic and disease status information will be sent with only a code number attached. Your name and other information that could identify you will never be given to them. There are many safeguards in place to protect your information while it is stored in repositories and used for research. You will not receive any results produced from participation in the national databases unless it is considered medically relevant. Usually researchers study just a few areas of your genetic code that are linked to a disease or condition, like NDV disorders. Research using your whole genome information involves using all or most of your genetic code and could be important for the study of virtually all diseases and conditions. Therefore, the databank will not restrict information for researchers working on any disease. You can withdraw your consent at any time you no longer want your data in the national databases. There will be no consequences for withdrawing consent. However, data that has already been sent to researchers cannot be retrieved from those researchers.

### **6. Participation in the Neurogenetics Laboratory Repository (Optional)**

If you agree, we will store your sample and medical information in the University of Washington Neurogenetics Laboratory Repository. The medical information and samples will be retained indefinitely and used in future studies of NDV disorders. We will ask you to sign a separate consent form. If you do not want your samples added to the repository, the samples will be kept for five years and then discarded.

## **RISKS, STRESS, OR DISCOMFORT**

### **Information Collection**

You may feel badly when discussing your NDV disorder. You may feel uneasy sharing your medical or family history. If you do feel uneasy, you may refuse to answer any question. You may learn about family members you did not know.

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### **Specimen Collection**

Drawing blood may cause a small amount of pain. You might get a bruise. Very rarely, an infection may develop. There may be side effects, like a rash, from using a numbing creaming. In general, people do not have any long-term effects from a blood draw. We do not think there will be any discomfort when giving saliva.

### **Optional Participation**

#### **1. Photography (Optional)**

We will attempt to hide features on a photo and/or a video clip that would allow someone to recognize you before scientific publication. Rarely, we will ask you for your permission to publish, in a peer-reviewed medical journal, a specific recognizable photo and/or video clip to show features of the disorder. If someone recognizes you, the confidentiality of participating in the research study may be broken.

#### **2. Recontact for Future Research Study (Optional)**

You may not want to answer more questions about our study. You can say no and ask not to be contacted again in the future.

#### **3. Reporting Results (Optional)**

You may feel stressed or anxious if you learn the genetic cause of your NDV disorder or that you are a carrier for an NDV disorder. It is possible that information regarding the genetic cause of your NDV disorder could lead to denial of insurance or employment. Although we may report clinical results to you, it is up to you and your doctor to decide whether these results should be included in your medical files. We suggest that you DO NOT report participation in this study to a third party. Third parties include health, life, disability or long-term care insurance. This would also include your mortgage company and employer. Rarely, genetic studies show 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. Skin Biopsy for Establishment of Fibroblast Cultures (Optional)**

You may feel pain from the needle for the medicine used to numb your skin. Medicines used to numb skin can give some people a rash. Skin biopsies can leave a small scar.

#### **5. Participation in the database of Genotypes and Phenotypes (dbGaP)**

There is a possible risk of loss of confidentiality. Your privacy will be strictly protected but we cannot absolutely guarantee there will be no breach of confidentiality. There is the risk that coded data could be released to the public, insurers, employers, or law enforcement agencies. There is also the risk that breach of computer security could release your information. In the unlikely event that your genetic information is mistakenly shared and linked with a medical condition, this could affect your ability to get or keep some kinds of insurance. If family members were to see this information, it could also affect them. This could hurt family relationships. It is possible that you could be identified from the sample if someone has another sample from you. The two samples could be matched to identify you from the sample given for this study.

#### **6. Participation in the Neurogenetics Laboratory Repository (Optional)**

There are no additional risks.

### **ALTERNATIVES TO TAKING PART IN THIS STUDY**

You do not have to join this study. If you do not join, your medical care 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 NDV disorder will be found. These results may help with your medical care. We will provide you with information about passing on this disorder to your children. If you are not a carrier of the NDV disorder, you may feel a sense of relief.

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### SOURCE OF FUNDING

The study team and/or the University of Washington is receiving financial support from the National Institutes of Health- National Institute of Neurological Disorders and Stroke, University of Washington Department of Pediatrics, and private non-corporate donors.

### CONFIDENTIALITY OF RESEARCH INFORMATION

All of the information you provide will be confidential. Government or university staff sometimes review studies such as this one to make sure they are being done safely and legally. If a review of this study takes place, your records may be examined. The reviewers will protect your privacy. The study records will not be used to put you at legal risk of harm.

### 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 provide consent for a child under 18, he/she can decide to withdraw from the study after he/she turns 18. If you take part, we will make every effort to keep your information confidential. We will store all of your research records in locked cabinets and secure computer files. We will label your information with a study number. The master list that links a person's name to their study number is 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 you unless you agree to publishing photographs.

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 records may be examined. The reviewers will protect your privacy. The study records will not be used to put you at legal risk of harm. The following groups may need to review study records about you: institutional oversight review offices.

We plan to keep your data and samples until the project is completed. You may refuse to participate or may withdraw from the study at any time without penalty or loss of benefits to which you are otherwise entitled.

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 sample may lead to the development of commercial products (although this is not a specific goal of the research). You will not share in any profits that this research may produce.

For pregnant women: Maternal samples collected during pregnancy will not be tested or studied until we have confirmation that the pregnancy has ended.

### COMPENSATION FOR INJURY

If you think you suffered an injury or illness related to this study, contact the study staff (Dan Doherty, M.D., Ph.D., 206-987-2590) right away. Dr. Doherty will recommend treatment or refer you for treatment. You or your insurer will be billed for treatment of your NDV disorder and any very unlikely medical problems that result from this study. No money has been set aside to pay for things like lost wages, lost time, or pain. However, you do not waive any rights by signing this consent form.

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Printed name of study staff obtaining consent

Signature

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### Subject's statement

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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 records as described in this consent form. I will receive a copy of this consent form.

**Molecular Analysis of Genetic Neurodevelopmental Disorders (Adults and Children)**

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

- |   |            |           |
|---|------------|-----------|
| <b>1. You may obtain photographs and/or video clips and use them for educational purposes and peer-reviewed scientific publication.</b> | <b>YES</b> | <b>NO</b> |
| <b>2. You may contact me about future studies.</b>  | <b>YES</b> | <b>NO</b> |
| <b>3. I would like to be contacted with the results of my research testing.</b>   | <b>YES</b> | <b>NO</b> |
| <b>4. You may obtain a skin biopsy and establish a fibroblast culture if requested.</b>   | <b>YES</b> | <b>NO</b> |
| <b>5. I would like to participate in the database of Genotypes and Phenotypes (dbGaP).</b>  | <b>YES</b> | <b>NO</b> |
| <b>6. I would like to participate in the Neurogenetics Laboratory Repository (requires separate consent form).</b>                      | <b>YES</b> | <b>NO</b> |

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Printed name of subject	Signature of subject	Date
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When subject is a minor:

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Printed name of parent	Signature of parent	Date
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When subject is not able to provide informed consent:

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Printed name of representative	Signature of representative	Date
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Relationship of representative to subject

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Printed name of witness	Signature of witness	Date
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Witness signature is required for: ☐ Subjects who are illiterate/educationally disadvantaged

Copies to: Researcher, subject

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