

UNIVERSITY OF MICHIGAN CONSENT TO BE PART OF A RESEARCH STUDY

INFORMATION ABOUT THIS FORM

You, or your child, may be eligible to take part in a research study. This form gives you important information about the study. It describes the purpose of the study, and the risks and possible benefits of participating in the study. Parents or legal guardians who are giving permission for a child, please note: in the sections that follow the word 'you' refers to 'your child.'

Please take time to review this information carefully. After you have finished, you should talk to the researchers about the study and ask them any questions you have. You may also wish to talk to others (for example, your friends, family, or other doctors) about your participation in this study. If you decide to take part in the study, you will be asked to sign this form. *Before you sign this form, be sure you understand what the study is about, including the risks and possible benefits to you.*

1. GENERAL INFORMATION ABOUT THIS STUDY AND THE RESEARCHERS

1.1 Study title:

Genes In Inherited Neurologic Disorders

1.2 Company or agency sponsoring the study:

University of Michigan funds (CCMB, rare disorders consortium and internal), National Institutes of Health

1.3 Names, degrees, and affiliations of the researchers conducting the study:

Margit Burmeister, Ph.D., Professor, Molecular & Behavioral Neuroscience Institute, Department of Psychiatry and Department of Human Genetics

Haiming Chen, M.D. Research Assistant Professor, Depression Center, Department of Psychiatry

James Dowling, M.D., Ph.D., Lecturer, Department of Pediatrics and Communicable Diseases and Department of Neurology

Johann E Gudjonsson, M.D., Ph.D. Clinical Lecturer, Department of Dermatology

Jun Li, Ph.D., Assistant Professor, Department of Human Genetics

Karen Majczenko, M.D., Research Laboratory Specialist Senior, Molecular & Behavioral Neuroscience Institute

Sandra Villafuerte, Ph.D., Research Investigator, Molecular & Behavioral Neuroscience Institute and Department of Psychiatry

2. PURPOSE OF THIS STUDY

2.1 Study purpose:

This study is designed to find what changes in the genetic material (DNA) cause inherited neurologic disorders, such as Ataxia (incoordination), Myoclonus (muscle jerks) or Epilepsy, or a combination of these neurological symptoms.

3. INFORMATION ABOUT STUDY PARTICIPANTS (SUBJECTS)

Taking part in this study is completely **voluntary**. You do not have to participate if you don't want to. You may also leave the study at any time. If you leave the study before it is finished, there will be no penalty to you, and you will not lose any benefits to which you are otherwise entitled.

3.1 Who can take part in this study?

Anyone who has an unexplained neurologic disorder that is thought to be genetic (inherited) can participate. However, if your health care provider thinks that you may have a known genetic form of ataxia or epilepsy, you should first be tested for these known causes before participating in this study. In order to confirm some of our experiments, we may ask persons with known ataxia mutations to participate.

Please indicate here the name of the disorder you have been told by your referring health care provider (Doctor or genetic counselor) that you have or might have:

(please fill in the specific diagnosis of the neurological disorder as you know it)

3.2 How many people (subjects) are expected to take part in this study?

We anticipate enrolling between 5 and 30 subjects each year in this ongoing study. In addition, in some cases we are interested in participation of unaffected relatives, approximately 10-30 relatives per year.

If there are other family members who have the same disorder, please let the study group know that this is so but don't give names of family members. You may choose to let other family members know about this study so they can participate and give their own consent if they so wish.

4. INFORMATION ABOUT STUDY PARTICIPATION

4.1 What will happen to me in this study?

You will be asked questions about your health by your health care provider or the researcher about your symptoms. You are asked to give permission for your health care provider to discuss with and provide

your health information to the researchers. In some cases, if you do not have a regular health care provider or have never seen a neurologist, the researchers may perform a brief physical examination focusing on the nervous system. You will also be asked about your ethnic background. You will donate up to 40 cc (8 teaspoons) of blood for DNA testing. If you are a child under the age of 16, you will donate 1 cc/kg body weight (about 1 teaspoon/10 lbs of weight). If you are unable or unwilling to donate a blood sample, you may donate approximately 2.5cc (1/2 teaspoon) saliva, or 2 cheek swabs. You will take this consent to your next scheduled visit with your health care provider who will perform the blood draw. You or the health care provider will then ship your blood samples to the researchers using the prepaid envelopes provided. If you are donating saliva or cheek swabs, you can collect the samples at home following the enclosed instructions and ship them using the prepaid envelopes. The researchers will isolate the genetic material, DNA, from some of the blood tubes or from the saliva/cheek swabs. From another blood tube, the researchers will isolate certain cells and change them in a way that they can grow them in a test tube for studies and to make more DNA; this can only be done from a blood sample.

Knowing your ethnic origin can help find DNA changes that occur only in some ethnic groups. Please indicate the countries from which your grandparents came from (if all were US-born, fill in what you know about their heritage, e.g. Eastern Europe, African American, South America):

mother's mother mother's father father's mother father's father

4.2 How much of my time will be needed to take part in this study?

In addition to the time it takes you to read over this information, fill in the information and ask any questions, about 30 minutes, the blood draw is expected to only take a few minutes.

4.3 When will my participation in the study be over?

Your direct participation in the study will be over when the researchers receive the consent document and blood/saliva samples. However, this is an ongoing study and the researchers will continue to study your samples indefinitely, unless you withdraw your permission (see section 7 'Ending the Study' below.)

5. INFORMATION ABOUT RISKS AND BENEFITS

5.1 What risks will I face by taking part in the study? What will the researchers do to protect me against these risks?

The known or expected risks are:

- There may be minimal discomfort from donating the blood sample, bleeding or bruising at the site, lightheadedness or fainting, and rarely, the possibility of infection at the site where the blood is obtained. There are no known risks associated with donating a saliva sample.

- You are providing your confidential private health information for the study. The researchers will keep this information confidential and will minimize the risk of accidental disclosure as outlined in more detail in paragraph 9.

As with any research study, there may be additional risks that are unknown or unexpected.

5.2 What happens if I get hurt, become sick, or have other problems as a result of this research?

The researchers have taken steps to minimize the risks of this study. Even so, you may still have problems or side effects, even when the researchers are careful to avoid them. Please tell the researchers listed in Section 10 about any injuries, side effects, or other problems that you have during this study. You should also tell your regular doctors

5.3 If I take part in this study, can I also participate in other studies?

Being in more than one research study at the same time, or even at different times, may increase the risks to you. It may also affect the results of the studies. You should not take part in more than one study without approval from the researchers involved in each study.

5.4 How could I benefit if I take part in this study? How could others benefit?

You will not receive any personal benefits from being in this study. However, the hope is that future generations of patients with inherited neurologic disorders, such as ataxia or myoclonic epilepsy, may benefit from new knowledge.

5.5 Will the researchers tell me if they learn of new information?

The researchers may learn something about the genetic causes of neurological disorders. The results from this research are not definite and cannot be used for your clinical care. If clinically valid diagnostic tests relevant for your disorder become available, the researchers will try to inform your physician and/or yourself about such tests. With this reservation in mind, you have the following choices regarding research results:

In order to help along the research, Dr. Burmeister often exchanges information with the neurologists who are treating patients with these disorders, such as your physician. But you can ask that your genetic results not be shared. However, Dr. Burmeister does not offer genetic counseling, and if you want to find out anything about your results – even though they are only valid for research - a physician or genetic counselor has to be involved to explain the meaning of the research results to you.

Yes No I give Dr. Burmeister permission to discuss my genetic results with my health care provider named below (9.2). I can choose to contact my health care provider if I want to find out about my results, and have him/her explain them to me.

If you answered **no** to the above question, you have the following options

- I prefer that my personal genetic results will not be discussed with any physician but I understand that research results on my condition without my name may be published in scientific literature.
- I would like to find out about my personal genetic results through a different physician or genetic counselor (contact information):

6. OTHER OPTIONS

6.1 If I decide not to take part in this study, what other options do I have?

Your participation is voluntary; the alternative is not to participate.

7. ENDING THE STUDY

7.1 If I want to stop participating in the study, what should I do?

You are free to leave the study at any time. If you leave the study before it is finished, there will be no penalty to you. You will not lose any benefits to which you may otherwise be entitled. If you choose to tell the researchers why you are leaving the study, your reasons for leaving may be kept as part of the study record. If you decide to leave the study before it is finished, please tell one of the persons listed in Section 10 ‘Contact Information’ (below).

7.2 Could there be any harm to me if I decide to leave the study before it is finished?

No, there are no known risks if you decide to leave the study.

7.3 Could the researchers take me out of the study even if I want to continue to participate?

Yes. There are many reasons why the researchers may need to end your participation in the study. Some examples are:

- ✓ You become ineligible to participate.
- ✓ The study is suspended or canceled.

8. FINANCIAL INFORMATION

8.1 Who will pay for the costs of the study? Will I or my health plan be billed for any costs of the study?

The study will pay for the blood draw. If your physician bills you for this blood draw, please ask him or her to bill the researchers instead. If you choose to pay, send us the receipt (at the address listed in section 10, 'Contact Information' below) and we will reimburse you.

You or your health plan will pay for all the things you would have paid for even if you were not in the study, like:

- Health care given during the study as part of your regular care
- Treatment of complications
- Deductibles or co-pays for these items or services.

If you do not have a health plan, or if you think your health plan may not cover these costs during the study, please talk to the researchers listed in Section 10 below or call your health plan's **medical reviewer**.

By signing this form, you do not give up your right to seek payment if you are harmed as a result of being in this study.

8.2 Will I be paid or given anything for taking part in this study?

No, you will not be paid for participating in this study.

8.3 Who could profit or financially benefit from the study results?

The researchers conducting the study:

The University of Michigan:

The University of Michigan may apply for patent protection if a new gene involved in an inherited neurologic disorder is found through this research. In that case, the University and the researchers may gain financially through licensing fees from companies studying treatment or testing for these disorders. You will not receive any proceeds, profits, or other benefits from any commercial product that may result from this study.

9. CONFIDENTIALITY OF SUBJECT RECORDS AND AUTHORIZATION TO RELEASE YOUR PROTECTED HEALTH INFORMATION

The information below describes how your privacy and the confidentiality of your research records will be protected in this study.

9.1 How will the researchers protect my privacy?

The medical information produced as a result of this study will not become part of your medical record. Information will be stored in a research file identified only by a code number. The key connecting your name to the code number will be stored in a separate, secure location. Information used in scientific publications will not contain any identifying information. The University of Michigan collaborates with

other organizations, and data and samples are generally shared. However, no data shared with other research institutions will include your name or other public identifiers. The only exception is the health care provider whom you designated in section 5.5. to receive possible results.

9.2 What information about me could be seen by the researchers or by other people? Why? Who might see it?

Signing this form gives the researchers your permission to obtain, use, and share information about you for this study, and is required in order for you to take part in the study. Specifically, you give permission to the referring health care provider to discuss your diagnosis and condition with the researchers

(please add name, address, phone number of referring physician or genetic counselor you give permission to)

Information about you may be obtained from any hospital, doctor, and other health care provider involved in your care, including:

- Hospital/doctor's office records, including test results (X-rays, blood tests, urine tests, etc.)
- Mental health care records (except psychotherapy notes not kept with your medical records)
- Alcohol/substance abuse treatment records
- Your AIDS/HIV status
- All records relating to your neurologic condition, the treatment you have received, and your response to the treatment
- Billing information
- Results from previous genetic testing

There are many reasons why information about you may be used or seen by the researchers or others during or after this study. Examples include:

- The researchers may need the information to make sure you can take part in the study.
- University, Food and Drug Administration (FDA), and/or other government officials may need the information to make sure that the study is done in a safe and proper manner.
- Study sponsors or funders, or safety monitors or committees, may need the information to:
 - Make sure the study is done safely and properly
 - Analyze the results of the study
- Insurance companies or other organizations may need the information in order to pay your medical bills or other costs of your participation in the study.
- The researchers may need to use the information to create a databank of information about your condition or its treatment.

- Information about your study participation may be included in your regular UMHS medical record.
- Federal or State law may require the study team to give information to government agencies. For example, to prevent harm to you or others, or for public health reasons.

The results of this study could be published in an article or presented at a scientific meeting, but would not include any information that would let others know who you are.

If Dr. Burmeister leaves the University of Michigan, research on your samples may continue at another institution.

9.3 What happens to information about me after the study is over or if I cancel my permission?

As a rule, the researchers will not continue to use or disclose information about you, but will keep it secure until it is destroyed. Sometimes, it may be necessary for information about you to continue to be used or disclosed, even after you have canceled your permission or the study is over. Examples of reasons for this include:

- To avoid losing study results that have already included your information
- To provide limited information for research, education, or other activities (This information would not include your name, social security number, or anything else that could let others know who you are.)
- To help University and government officials make sure that the study was conducted properly

As long as your information is kept within the University of Michigan Health System, it is protected by the Health System's privacy policies. For more information about these policies, ask for a copy of the University of Michigan Notice of Privacy Practices. This information is also available on the web at <http://www.med.umich.edu/hipaa/npp.htm>. Note that once your information has been shared with others as described under Question 9.2, it may no longer be protected by the privacy regulations of the federal Health Insurance Portability and Accountability Act of 1996 (HIPAA).

9.4 When does my permission expire?

Your permission will not expire unless you cancel it. You may cancel your permission at any time by writing to the researchers listed in Section 10 'Contact Information' (below). If you are a minor under the age of 18, your permission expires when you turn 18 unless you sign a new consent.

10. CONTACT INFORMATION

10.1 Who can I contact about this study?

Please contact the researchers listed below to:

- Obtain more information about the study
- Ask a question about the study procedures or treatments
- Talk about study-related costs to you or your health plan
- Report an illness, injury, or other problem (you may also need to tell your regular doctors)
- Leave the study before it is finished
- Express a concern about the study

Principal Investigator: Margit Burmeister, PhD
Mailing Address: University of Michigan,
Molecular & Behavioral Neuroscience Institute; 5061 BSRB, 109 Zina Pitcher Place;
Ann Arbor MI 48109-2200
Telephone: (734) 647-2186; email: margit@umich.edu

Co-Investigator/Research Coordinator: Sandra Villafuerte, PhD
Mailing Address: University of Michigan,
Molecular & Behavioral Neuroscience Institute; 5063 BSRB, 109 Zina Pitcher Place;
Ann Arbor MI 48109-2200
Telephone: (734) 647-4241; email: svillafu@umich.edu

You may also express a concern about a study by contacting the Institutional Review Board listed below, or by calling the University of Michigan Compliance Help Line at 1-888-296-2481.

University of Michigan Medical School Institutional Review Board (IRBMED)
Argus I
517 W. William
Ann Arbor, MI 48103-4943

Telephone: 734-763-4768
Fax: 734-615-1622
e-mail: irbmed@umich.edu

If you are concerned about a possible violation of your privacy, contact the University of Michigan Health System Privacy Officer at 1-888-296-2481.

When you call or write about a concern, please provide as much information as possible, including the name of the researcher, the IRBMED number (at the top of this form), and details about the problem. This will help University officials to look into your concern. When reporting a concern, you do not have to give your name unless you want to.

11. RECORD OF INFORMATION PROVIDED

11.1 What documents will be given to me?

Your signature in the next section means that you have received copies of all of the following documents:

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- This "Consent to be Part of a Research Study" document. (*Note: In addition to the copy you receive, copies of this document will be stored in a separate confidential research file and may be entered into your regular University of Michigan medical record. One copy is left with your health care provider who is coordinating this study.*)

12. SIGNATURES

Research Subject:

I understand the information printed on this form. I have discussed this study, its risks and potential benefits, and my other choices with _____. My questions so far have been answered. I understand that if I have more questions or concerns about the study or my participation as a research subject, I may contact one of the people listed in Section 10 (above). I understand that I will receive a copy of this form at the time I sign it and later upon request. I understand that if my ability to consent for myself changes, either I or my legal representative may be asked to re-consent prior to my continued participation in this study.

Signature of Subject: _____ Date: _____

Name (Print legal name): _____

Patient ID: _____ Date of Birth: _____

Legal Representative (if applicable):

Signature of Person Legally Authorized to Give Consent _____ Date: _____

Name (Print legal name): _____ Phone: _____

Address: _____

Check Relationship to Subject:

Parent Spouse Child Sibling Legal Guardian Other: _____

If this consent is for a child who is a ward of the state (for example a foster child), please tell the study team immediately. The researchers may need to contact the IRBMED.

Reason subject is unable to sign for self: _____

Principal Investigator (or Designee):

I have given this research subject (or his/her legally authorized representative, if applicable) information about this study that I believe is accurate and complete. The subject has indicated that he or she understands the nature of the study and the risks and benefits of participating.

Name: _____ Title: _____

Signature: _____ Date of Signature: _____