

# CHILDREN'S NATIONAL MEDICAL CENTER

Center for Genetic Medicine, Children's Research Institute  
111 Michigan Avenue, NW  
Washington, DC 20010  
(202) 476 6011

## ASSENT (AGES 12 to 18) TO PARTICIPATE IN A CLINICAL RESEARCH STUDY

### Patient with a Leukodystrophy

---

**TITLE OF STUDY:** New Diagnostic Approaches In Leukodystrophy

**PRINCIPAL INVESTIGATOR:** Adeline Vanderver, MD; Department of Neurology

---

**INTRODUCTION:** We would like to invite you to be part of a research study at Children's National Medical Center. Before you decide if you would like to participate, we want you to know why we are doing the study. We also want you to know about any risks (anything unexpected that might happen) and what you will be expected to do in the study. You can only be in the study if your parent(s) agree(s).

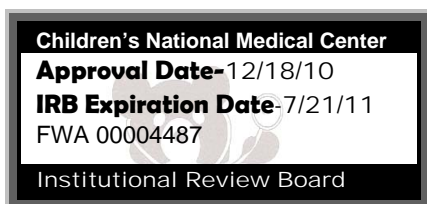
This form gives you information about the study. Your doctor or a research staff member will talk to you about the study and answer any questions you have. We encourage you to discuss this study with your family before making your decision. We will ask you to sign this form to show that you understand the study. We will give you a copy of this form to keep. It is important that you know:

- You do not have to join the study;
- You may change your mind and stop being in the study any time you want and no one will mind.
- If we make any important change to the study we will tell you about it and make sure you still want to be in the study.

#### A. WHAT IS THE REASON FOR THE STUDY?

Your doctor thinks that you have a leukodystrophy and would like to let us look at your medical records, MRI, and clinical examination and see if you can participate in a study to find out more about leukodystrophies. Leukodystrophy is a disease that affects the white matter. The white matter is the area of the brain through which messages are sent. A substance called myelin allows messages to move quickly through this area and causes the area to look white in color, which is why it is called white matter. Leukodystrophies can have many causes, some known and some yet to be recognized.

IRB Protocol No.: { 3415 }  
Date: { 11.10.2010 }  
Page 1 of 5



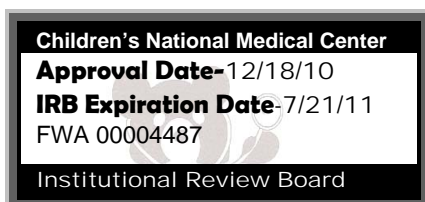
If we think we can learn about leukodystrophies with your help and the help of others like you, we may ask your doctor if you can come to be seen by our doctors at Children's National Medical Center. We may ask your doctor to send us an extra blood sample, extra spinal fluid or cultured cells from you so that we can do research related to your condition. We may ask your doctor to send us a blood sample to isolate DNA, which is the recipe for your genes, to see if we can find a cause for your leukodystrophy. We are asking your permission to look at your medical records, clinical exam and MRI and to use any sample that your doctor sends for research. This research may help to find what causes leukodystrophies as well as find new tests to diagnose them.

## **B. WHAT WILL HAPPEN IN THE STUDY?**

This study uses experiments to look at genes and proteins. Genes carry the instructions for how everything in our body works. Proteins are created from the instructions in the genes and carry that message to the body. Before your doctor sends us any samples, we would like him/her to send us copies of your medical records and MRI testing. In addition, we may ask to meet you and perform an examination like your doctor would normally perform. That means looking at how your nerves work by looking at the strength of your muscles, your reflexes and other things that don't hurt.

1. If, based on that information, we feel that you do have a leukodystrophy then we may ask for a small amount of blood (about two teaspoons). We will use that blood sample to try to identify changes in your genes that might be causing kids like you to have a leukodystrophy. We may also use that sample to save DNA, fluid or cells from your blood to try to identify changes in the proteins or molecules in your blood that may cause a leukodystrophy. When we look at your DNA, it is possible that we will find other genetic changes that could affect you or your family members, that are unrelated to the leukodystrophy. If we find genetic changes that are relevant to your health or the health of your family members, and that have a treatment, we will tell let you or your family members know.
2. Also, if your doctor performs a spinal tap, we would like to save some extra fluid. We will use that sample to look at proteins in the fluid to try to find changes that might be making kids like you sick. Your doctor will get this extra spinal fluid if you are getting testing done for other reasons. You will not have to get an additional spinal tap to participate in the study
3. If your doctor collects a urine specimen, they may send us an additional sample. We will test that urine for proteins and molecules that can be seen in patients with leukodystrophies to try to identify causes for leukodystrophies in kids like you.
4. Finally, if your doctor performs a skin biopsy, your doctor will send that sample to a lab. At the lab, the skin cells that were taken in the skin biopsy will be allowed to grow (so

IRB Protocol No.: { 3415 }  
 Date: { 11.10.2010 }  
 Page 2 of 5



that the sample is larger and there is more material to work with). This process is called "culturing cells." We will then ask your doctor to send us some cultured cells. Asking for these cultured cells does not change in any way the skin biopsy that will be done by your doctor for clinically indicated reasons. We will not ask your doctor to do a skin biopsy for this research study alone. We will use those cells to try to identify changes in your genes that might be causing kids like you to be sick.

5. We are not testing medicines or other treatments in this study that might make you better. This is only research that is done on your blood sample to try to find out more about the disorder making you sick.
6. We won't ask your doctor to do any extra tests for the purposes of our research except for a blood draw. We will just ask for samples when lumbar punctures, or skin biopsies are being done for other reasons.
7. We will keep your sample until this study is over. If you decide at anytime that you do not want to be in this study anymore, you can call the principal investigator, Dr. Vanderver, and she will take you out of the study. That means that she will destroy any samples that you have donated and she will take your name out of the database.

### **C. WHAT POSSIBLE UNEXPECTED THINGS COULD HAPPEN?**

All samples requested for this study will be taken at the time of a clinically indicated procedure. That means that we will not take any samples (blood, skin biopsy, spinal fluid) for this research study alone. Your doctor will explain any procedure related risks at the time of the procedure. The risks associated with this study are limited to those related to confidentiality. The risks associated with obtaining the samples to find out your leukodystrophy should be explained to you by the doctor obtaining the samples. All the potential risks explained below should be explained to you in detail by your doctor.

1. For tests on your DNA we need one sample of blood (about 2 teaspoons). The needle puncture may cause some bleeding, mild pain or infection. However, the procedure is to be performed by individuals trained and experienced in obtaining blood samples so as to minimize these risks.
2. For the spinal fluid, we need about 1 cc or ml ( less than half a teaspoon ). The lumbar puncture may cause some bleeding, mild pain, headache or infection. However, the procedure is to be performed by individuals trained and experienced in performing spinal taps so as to minimize these risks.
3. For the skin culture from the skin biopsy, we will need cells grown in a lab from your skin biopsy. These cells can be saved and we won t have to repeat the skin biopsy.
4. There is a chance that you may feel stress from participating in our research. Knowing that you a leukodystrophy can lead to feelings of anxiety or sadness. Stress may also occur in knowing that you have a genetic condition, but that the doctors don't have a cure or treatment for your problems. We have a genetic counselor who can answer

IRB Protocol No.: { 3415 }  
Date: { 11.10.2010 }  
Page 3 of 5



questions you have about the research. If you are upset, you should talk to your doctor about this. If you are unsure if you want to be in this research study, the genetic counselor can help you decide if you want to participate.

#### **D. WHAT POSSIBLE GOOD THINGS COULD HAPPEN?**

By doing research on blood, spinal fluid and skin samples from patients like you, Dr. Vanderver's laboratory can identify new causes for health problems and a better understanding of why the disease happens. By doing this research, we expect to find gene and protein changes that cause disease and we hope to learn more about their effect on the body. This research helps us to create accurate tests and may help our group and others to find medicines or other therapies to treat people with genetic conditions. If we find any results through our research that might help you directly, we will let your doctor know so that he (she) can tell you.

#### **E. WHAT OTHER CHOICES DO YOU HAVE IF YOU DO NOT WANT TO BE IN THE STUDY?**

No treatment is offered or recommended for participants in this study. If you decide not to participate it will not affect your care here at CNMC in any way.

#### **F. HOW WILL WE KEEP YOUR RECORDS PRIVATE?**

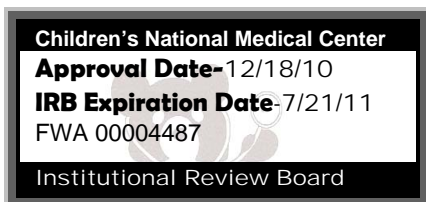
We will keep the records of this study confidential. Only the people working on the study will know your name. They will keep this information in case we have to find you later to let you know of any new information that may affect your health.

#### **ASSENT**

By signing this form, you agree that you have talked to your doctor about the study and understand it, and want to be in the study. You also agree that you have been told about the risks (unexpected things) and benefits (good things) of the study, and about other choices. You may stop being in the study at any time and no one will mind and nothing will change about your medical care other than not being in the study. By signing this form, you agree to participate in the following aspects of this research:

1. Collection of clinical information (medical records, MRI, clinical exam)
2. Collection of blood for gene and other studies
3. Use of extra spinal fluid for protein studies

IRB Protocol No.: { 3415 }  
Date: { 11.10.2010 }  
Page 4 of 5



- 4. Use of culture of skin cells from skin biopsy
- 5. Use of extra urine sample
- 6. After use for this study, my blood, urine, skin and/or CSF samples will be stored to be used in future studies related to leukodystrophies only.

Please call the Principal Investigator, Dr. Adeline Vanderver, at 202-476-6230, if you have any questions.

Printed Name of Participant: \_\_\_\_\_

Medical Record Number: \_\_\_\_\_

Signature of Participant: \_\_\_\_\_

**Witness (to signature):** \_\_\_\_\_ **Date:** \_\_\_\_\_  
(may be investigator)

Translator's Signature (if, applicable): \_\_\_\_\_ **Date:** \_\_\_\_\_  
Language: \_\_\_\_\_

**AFFIDAVIT OF PERSON OBTAINING ASSENT:** I certify that I have explained to the above individual(s) the nature and purpose of the study, potential benefits, and possible risks associated with participation in this study. I have answered any questions that have been raised.

Printed Name of Individual Obtaining Consent: \_\_\_\_\_

Title: \_\_\_\_\_ Signature: \_\_\_\_\_ Date: \_\_\_\_\_

IRB Protocol No.: { 3415 }  
Date: { 11.10.2010 }  
Page 5 of 5

