

## ASSENT FORM TRANSLATIONAL RESEARCH IN THE DYSTROPHINOPATHIES

### Purpose of the Research

Genes determine who we are and what we look like. A mutation in a gene called the dystrophin gene results in the muscle diseases called Duchenne Muscular Dystrophy, Becker Muscular Dystrophy, and isolated cardiomyopathy. Because your physician has determined that you or members of your family have a form of muscle weakness is probably due to muscular dystrophy, we are inviting you to participate in this study.

This study is to help us understanding how different gene mutations (or changes in genes) cause muscle weakness. In some families or patients with muscular dystrophy, a large part of the dystrophin gene missing. Others have smaller changes in the gene. How these different mutations influence disease severity is not fully understood.

We hope to get a better understanding of how different mutations in the dystrophin gene affect the symptoms and severity of muscular dystrophy. A second goal of our study is to establish a registry of patients with muscular dystrophy. We are not asking you to participate in any clinical trials, which might treat muscular dystrophy. However, if you give us permission to do so, we will notify you of clinical trials in which you might take part.

### Study procedure

There are several parts to the study.

1. You will be examined about once a year. The examination will be essentially the same as you go through now when you visit your doctor. For example, your strength will be tested by the examiner pushing against your limbs. You will be timed performing tests such as getting up off of the floor walking about thirty feet down a hall. You will have a breathing test to measure the volume of air you can inhale. The examiner will score your performance on these tests, and record it.
2. You (and your parents) will be asked to fill help the study coordinator fill out a questionnaire each year. The questionnaire will ask about your medical history. You may be asked for a sample of blood for analysis of the dystrophin gene. If you agree to have blood drawn, will draw 3-4 tablespoons of blood from which we will extract DNA for study. The blood draw will only require 5-10 minutes of your time.
3. If you have already had a muscle biopsy as part of your doctor appointments, a leftover piece of muscle is probably stored in a freezer at the hospital. We would like to obtain that muscle biopsy sample in order to perform special studies with it. These studies include looking at the size and amount of the dystrophin protein, as well as how the protein is put together (the DNA and RNA).
4. Your information will be stored in a database maintained by Dr. Flanigan and his colleagues. Other research programs may be interested in your and other people's information. We want to share any information with doctors which will help in more research programs. Your information shared with other researchers will not include your name or other information that can identify you.

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### **Risks**

When blood is drawn, there is a small risk of getting a bruise, or infection, and it might be slightly uncomfortable. These risks are extremely small when a skilled person draws blood.

It might be possible for someone to hold it against you (discriminate against you) in the future if you have a genetic disorder. However, because of the symptoms in you or your family, your doctor already suspects that you might have a muscular dystrophy.

Our additional testing does not make it more likely that you will be discriminated against. In order to protect against discrimination, we will keep all records in locked files. Only Dr. Flanigan and his colleagues working on muscular dystrophy will be able to look at your records.

### **Benefits**

We hope that muscular dystrophy researchers around the country (or around the world) will tell us about clinical trials. Clinical trials may be tests of treatments for muscular dystrophy. They will tell us what type of patients they are looking for (for example, if they are walking, running, or in a wheelchair). We will ask them to show that they have received proper approval for their research. If our records show that you might be the kind of muscular dystrophy patient they are looking for, we can notify you about their trial.

**We will not give your name or any personal information to these researchers.** We will give your or your parents the information you need to contact them, so that if you wish you can contact them directly for more information about their trial.

**Just because we tell you about a study at another university or hospital, it does not mean we think recommending that you take part in it. It also does not mean that Dr. Flanigan or the University of Utah's Institutional Review Board knows that it is safe, or will do you any good. You need to carefully review any other studies. You will need to review and sign a separate consent form for any other research study in which you choose to participate.**

- a.  \_\_\_\_ Please notify me of any research studies for which I may be a candidate. I understand that if you notify me it does not guarantee that I will be enrolled in that study. I also understand that if you notify me, I will need to contact those researchers myself, and determine for myself whether I wish to participate in their study.
- b.  \_\_\_\_ Please do not notify me of any research studies for which I may be a candidate

We cannot guarantee any benefits to you for being in this study. You being in this study may help others, and help society. Anything new we find during this study will be shared with you.

### **Alternative procedures**

You can choose not to participate in this study.

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**Confidentiality**

We will keep all the information that identifies you to ourselves. We will keep your information locked so that no one can see it. If your information is on the computer, it will be protected with a password. We will return information to your physician or physicians only if you ask us. Please feel free to ask.

**Person to contact**

You can ask any questions that you have about the study. If you have a question later that you didn't think of now, you can call me Dr. Kevin Flanigan at 801-587-9540 or ask me next time. You can also call my Research Coordinator at 801-585-1299.

**Voluntary participation**

If you don't want to be in this study, you don't have to participate. Remember, being in this study is up to you and no one will be upset if you don't want to participate or even if you change your mind later and want to stop. Please talk this over with your parents before you decide whether or not to participate. We will also ask your parents to give their permission for you to take part in this study. But even if you parents say "yes" you can still decide not to do this.

**ASSENT**

Signing my name at the bottom means that I agree to be in this study. (if the study is related to treatment insert the following: My doctors will continue to treat me whether or not I participate in this study.) My parents and I will be given a copy of this form after I have signed it.

\_\_\_\_\_  
Printed Name of Child

\_\_\_\_\_  
Signature of Child

\_\_\_\_\_  
Date

\_\_\_\_\_  
Printed Name of Witness

\_\_\_\_\_  
Signature of Witness

\_\_\_\_\_  
Date

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