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<b>MEDICAL RECORD</b>	<b>MINOR PATIENT'S ASSENT TO PARTICIPATE IN A CLINICAL RESEARCH STUDY</b> • Attach to NIH-2514-2, Consent to Participate in a Clinical Research Study
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INSTITUTE: National Human Genome Research Institute

STUDY NUMBER: 14-HG-0071 PRINCIPAL INVESTIGATOR: Carlos R. Ferreira, M.D.

STUDY TITLE: Clinical and Basic Investigations into Known and Suspected Congenital Disorders of Glycosylation

Continuing Review Approved by the IRB on 07/13/17

Amendment Approved by the IRB on 07/06/18 (E)

Date Posted to Web: 07/11/18

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Standard Assent

The National Institutes of Health (NIH) has a hospital in which doctors study many diseases. We would like to ask you to help us with our study. We use the words "research" and "study" a lot. Medical research means asking questions about diseases and what they do to people. We will explain to you how the study works. We want you to ask any questions you have. There is no such thing as a silly or stupid question.

#### **WHY ARE WE DOING THIS STUDY?**

We are currently studying children and adults like you who have congenital disorders of glycosylation (or more simply CDG). All this means is that you cannot put sugars into proteins, which are the building blocks of your body; most proteins need to have sugars attached in order to work properly. People with CDG may have changes in the way their nerves, muscles, liver, and immune system work, among others. We would like you to help us understand more about CDG, and so we want to tell you about our study and ask you to participate.

#### **WHAT WILL HAPPEN IN THIS STUDY?**

1. Your parents and/or your doctor at home will need to tell us about any medical problems you may have. Anything they tell us will be kept private.
2. We would like you to stay in the hospital for 3 to 5 days. We might also ask you to come back again in the future.
3. We will get to know you and how the CDG affects you and your body. We will look at you from head to toe.
4. While you are at the NIH you will be examined by many different doctors and both you and your parents will be asked a lot of questions. These doctors will test various things like hearing or thinking in addition to regular examinations that you get when you visit other doctors at home.
5. Because we can learn so much from the blood in your body, we will take some blood tests at different times on each visit. Having your blood taken can hurt. To help, we first will numb your skin and then place a needle called an IV in your arm vein. Once the needle is in your arm, you will not feel it. We will draw blood from the IV.
6. We also want to do some tests that need a little bit of your urine, so we will ask you to urinate in a container.
7. If you are girl and have begun having your period, we will do a pregnancy test on your blood or urine. Your parents or guardian will be told the result of this test.
8. We will take a picture of you to keep for our files. You may be asked if it is OK to take your clothes off, except for your underwear, for these pictures. These pictures may be put in medical journals. You may ask that your eyes be covered in the pictures so that other people will not recognize your face.
9. You may also have a special picture taken of your body. This X-ray picture does not hurt at all. It will show the bones and other parts of your body.
10. We may take a small piece of skin from your arm. It will be half the size of an M&M. Taking a piece of skin can hurt.

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**PATIENT IDENTIFICATION**

**MINOR PATIENT'S ASSENT TO PARTICIPATE IN A CLINICAL RESEARCH STUDY**

NIH-2514-2 (10-09)

P.A.: 09-25-0099

File in Section 4: Protocol Consent (4)

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We will use numbing medicine on your skin to make it hurt less. The cut will bleed a little bit but we will put a band-aid over it and it will heal in a few days. The biopsy will leave a little scar but we can do it in a place that is covered by your clothes so no one will see it.

11. We will take pictures of your brain using a special machine called an MRI. The MRI test does not hurt but can be noisy. Because you have to hold still for this test and because this test can be scary for some kids, we may give you a medicine so you can sleep through this test.

12. Because CDG causes problems with the brain, we might want to do some tests on a fluid called cerebral spinal fluid (CSF). We collect CSF by using a needle like we do when we collect blood. However, in this case we need to stick the small needle in your lower back. If we give you medicine to make you sleep for the MRI exam, we will do this other test while you are asleep. If we need to do this while you are awake we will use numbing medicine to make this hurt less. Some kids get a headache after CSF is collected. If you get a headache, we will have you lie in bed and give you a medicine to make the headache go away.

13. Because CDG can cause problems with the nerves and muscles, we might need to perform a test in which we stickers in your skin or place needles in your muscles and then administer a small electric shock. This might hurt some, but we would most likely do this test while you are asleep for other procedures.

### **WHAT YOU MIGHT NOT LIKE ABOUT THIS STUDY**

We will want you to do some tests that involve putting a needle through your skin, like blood draws, obtaining CSF, or getting an EMG. For the blood draw, we will numb the skin before putting the needle, and for the CSF study and EMG we will give you medicine that will help you go to sleep. When we do the MRI we may also give you medicine that will make you go to sleep.

Collecting urine for a whole day can be difficult because you will have to carry around a container for a day.

Taking a piece of skin can hurt. We can give you some numbing medicine to reduce the pain.

You will be exposed to some radiation during the X-rays. This doesn't hurt at all. We are careful about the amount of radiation someone gets because it may not be completely safe. Experts at the NIH have decided that the amount of radiation you will get by being in this study is reasonable.

### **RIGHT OF REFUSAL AND RIGHT TO WITHDRAW**

You can ask us any questions that you want to about the study. We want to make sure that you understand what we are doing, and why we are doing it. You do not have to be in this study if you do not want to be. You can decide to be in the study and then change your mind at any time if you want to stop being in the study.

### **COMPENSATION**

You will not be paid to take part in this study.

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I have had this study explained to me in a way that I understand, and I have had the chance to ask questions. I agree to take part in this study.

Signature of Minor Patient: \_\_\_\_\_ Date: \_\_\_\_\_

Print Name: \_\_\_\_\_

Signature of Investigator: \_\_\_\_\_ Date: \_\_\_\_\_

Print Name: \_\_\_\_\_