

INSTITUTE: National Institute of Child Health and Human Development

STUDY NUMBER: 00-CH-0219 PRINCIPAL INVESTIGATOR: Carolyn A. Bondy, M.D.

STUDY TITLE: Turner Syndrome: Genotype and Phenotype

Continuing Review Approved by the IRB on 5/27/09

Amendment Approved by the IRB on 5/27/09 (V)

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Assent

We are doctors and nurses at the National Institutes of Health (NIH). We would like to learn more about Turner syndrome. We hope that what we learn will help us take care of children with this condition.

What We Are Asking You To Do

We would like you to come to our hospital. Your mother or father will be able to stay with you while you are here. You will be asked to stay overnight in the hospital for 3 to 5 days.

You may be asked to stop taking certain medicines, such as growth hormone or estrogen, two weeks before you come to our hospital. The doctors and nurses here will talk with your parent(s) and your doctor to decide if this is okay. You will be able to start taking your medicines again once you leave our hospital. If you do not feel well when you stop taking your medicines, let your mother or father know. They may need to call your doctor to see if you should start taking your medicines again. If you are not able to stop taking these medicines as we have asked, you will still be able to come to our hospital.

A doctor will ask you and your parents about your health. You will be asked to remove your clothes so that a doctor can examine you. We may take pictures of any parts of your body that look a little different than normal. You will be allowed to wear your underwear for these pictures. We will keep these to show other doctors so they can learn more about Turner syndrome. A doctor will examine your ears and test your hearing. This will not hurt.

You will be weighed and measured by a nurse. We will take some blood from your arm. To do this, we use a needle. It feels like getting a shot. We will also use a needle to put a small plastic tube called an I.V. into your hand or arm. We may put some special cream on your hand or arm to numb the area where the needle goes in. This may help lessen any pain you may feel with the needle. We take the needle out but leave the plastic tube in your hand or arm for a while. We will take blood from that tube. It will not hurt, while we are taking blood. We will also ask you to save your urine. The nurse will give you special containers to help you to do this. You will be on a special diet while you are in the hospital. You will be given a menu to pick out what foods you like.

While you are in the hospital, nurses will measure your blood pressure every 8 hours. While you are in the hospital, a nurse will give you a blood pressure machine to wear on your arm for 12 hours. A nurse will teach you and your parents how to use it.

We will take pictures of the inside of your body with different machines. The tests you will have are called an EKG of your heart, X-rays of your wrists, DEXA scan, MRI, and ultrasounds of your heart and stomach area. These tests do not hurt. Your mother or father can stay with you during these tests. For most of these tests, you will lie down on a table

STUDY NUMBER: 00-CH-0219

CONTINUATION: page 2 of 2 pages

and stay very still for a while. The MRI scan may be a little scary for you. The MRI is a large tube that you lie down inside of. It can make a lot of loud noises while it is taking pictures. If you want to come out of the MRI at any time, you will be allowed to do so.

During the MRI, you may have some special fluid put into one of the your veins in your arm or hand. This fluid will help us to see the picture of your heart better. In order to give you this fluid, we may need to use a needle to put a small plastic tube called an I.V. into your hand or arm. We may put some special cream on your hand or arm to numb the area where the needle goes in. This may help lessen any pain you may feel with the needle. We take the needle out but leave the plastic tube in your hand or arm for a while.

If you are at least 14 years old, we will ask you to answer questions on a written form and in a computer. The answers to these questions will help us to evaluate your memory and learning abilities. This evaluation will take about 2 hours. You will also be asked questions about how you feel about yourself and others and how you act in certain situations. You will be given written questionnaires about these things. These questions will take about 1 hour.

You may request that private information is not shared with your parents or guardian. We will try to keep all information about you private.

You may be asked to give a very small piece of skin from the inside of your arm to help with the study of Turner syndrome. This test will not be required if you do not want it. Before we do this, we will numb the skin on your arm with medication given through a needle. The needle will feel like a shot. After we numb the area, you should not feel any pain. A small piece of skin about the size of a pea will then be removed using a special tool called a skin punch. The wound will be covered with antibiotic and a small bandage. It will form a scab and heal after several days. You may develop a small scar on your arm from this procedure.

We may ask you to return to NIH every 2-5 years for the next 15 years to re-evaluate the way your body reacts to drinking a sugary drink, your heart function, and your liver and thyroid gland function. This will require additional blood draws, having an I.V. for a few hours, and taking pictures of your heart with ultrasound and MRI machines. These tests may be done in 1 or 2 days in the clinic or day hospital, so you will not need to stay overnight in the hospital.

What You Need to Decide

We want you to understand what we will do and why we do it. Please ask us to explain whatever you do not understand. If you agree to what we have explained here, please write your name on this paper to show that you are willing to have these tests done. If you decide that you do not want to do this, you do not have to. Also, you may change your mind at any time later on.

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: _____

Signature of Investigator: _____ Date: _____