Title: Development of a Newborn Screening Assay for Angelman Syndrome and Prader-Willi Syndrome

NCT: 05783791

Date: 01-23-2023

ICF for Angelman Syndrome and Prader-Willi Syndrome Participants

University of Wisconsin-Madison
Consent to Participate in Research
and
Authorization to Use Protected Health Information for Research

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Study Title for Participants: Early Detection for Better Outcomes

Formal Study Title: Development of a Newborn Screening Assay for Angelman Syndrome and Prader-Willi Syndrome

Lead Researcher: ***Mei Baker, MD, (608) 890-1796, 465 Henry Mall, Madison, WI 53706***

Institution: ***University of Wisconsin-Madison***

Key Information

The information in this section is to help you decide whether or not to be a part of this study. You can find more detailed information later on in this form.

If you are the parent or legal guardian of a minor who is invited to take part in this study, or if you are a legally authorized representative for an adult patient, for the rest of this consent form, "you" refers to "your child."

Why are researchers doing this study?

When babies are born in the U.S, a blood test called a "newborn screening" is commonly done to check for various congenital conditions, that are often genetic disorders. Angelman syndrome (AS) and Prader-Willi syndrome (PWS) are genetic disorders that affect an estimated 10,000 – 20,000 people in the U.S.; however, they are not included in the current newborn screening process. As a result, diagnosis and treatment of these syndromes is often delayed until early childhood.

The purpose of this project is to develop a newborn screening test for AS and PWS that can be included with the newborn screening programs in the United States. The hope is that if we can develop a newborn screening for AS and PWS, we can detect these syndromes at birth and begin helping children and families sooner. Currently, earlier detection of AS may help reduce families' medical costs, stress, and anxiety as they await a diagnosis for their baby. Earlier detection of PWS may allow for earlier treatment interventions and improve babies' and families' quality of life sooner.

We invite you to take part in this research study because you have AS or PWS.

What will I need to do in this study?

The research team will ask you to collect a drop of blood using the blood spot card and instructions provided to you. You will then mail the blood spot card back to the researchers using the provided Blood Spot Envelope. You will not need to include your name or any other information with the blood spot card.

We expect that it will take about 5 minutes to complete this collection.

You can find detailed information about the study procedures in the section called **If I take part in the study, what will I do?**

What are some reasons I might – or might not – want to be in this study?

You may want to be in this study if you are:	You may NOT want to be in this study if you:
• Willing to give a blood sample for research tests. • Interested in contributing to scientific knowledge even though you won't benefit directly from the study.	• Want to be in a study that might help improve your own health. • Are nervous about giving blood samples.

Do I have to be in the study?

No, you do not have to be in this study. Taking part in research is voluntary. If you decide not to be in this study, your choice will not affect your healthcare or any services you receive. There will be no penalty to you. You will not lose medical care or any legal rights. You can ask all the questions you would like before you decide.

Detailed Information

The following is more detailed information about this study in addition to the information listed above.

How is research different from health care?

When you take part in a research study, you are helping to answer a research question. Study tests and procedures are not for your health care.

Who can I talk to about this study?

If you have questions, concerns, or complaints, or think that participating in the research has hurt you, talk to the research team by phone at 608-228-9490 or by email at researchnurses@pediatrics.wisc.edu.

If you have any questions about your rights as a research participant or have complaints about the research study or study team, call the confidential research compliance line at 1-833-652-2506. Staff will work with you to address concerns about research participation and assist in resolving problems.

If I take part in the study, what will I do?

A study team member will talk to you about the study and ask if you would like to participate. (As applicable) If your child is able to understand, we will also describe the study to them and ask if they would like to participate. If you both agree, then the study team member will enroll you in the study.

Next, the study team member will ask you to provide a blood sample. Using the instructions provided, you will prick the participant's finger and place a drop of blood onto a card. The study team member will be available to guide you through the sample collection process over the phone, if you would like. You will then mail the blood spot card back to the research team using the provided Blood Spot Envelope.

In addition to collecting the blood sample, the study team will also collect some basic information from your medical records. This information may include age, sex assigned at birth, and diagnosis.

Some of the tests we will perform on your blood will be genetic testing, which is done on your DNA. DNA, or deoxyribonucleic acid, carries the genetic instructions for the cells that make up your body. Genes tell your body how to do things like form your spine, or what color your eyes should be.

Protected health information (PHI) used in this study

Protected health information, also called PHI, is information about your physical or mental health that includes your name or other information that can identify you, like your date of birth or medical record number. To do this study, we will use the following kinds of PHI:

- Information currently in your medical records. This information could include your medical history; your diagnosis; basic demographic information such as age and sex assigned at birth. We will get this information from your health care providers.

What happens if I say yes, but I change my mind later?

You can leave the research at any time. If you choose to leave the study, your choice will not affect your healthcare or any services you receive. No matter what decision you make, and even if your decision changes, there will be no penalty to you. You will not lose medical care or any legal rights.

You can let the research team know that you want to leave the study by phone call (608-228-4940) or e-mail (researchnurses@pediatrics.edu). You will not be asked why you want to leave the study. Your collected sample will be destroyed. If you stop being in the research, already collected data may not be removed from the study database.

Your authorization for researchers to use your protected health information (PHI) will last until the research study is done. However:

- You can choose to take back your authorization for researchers to use your health information. You can do this at any time before or during your participation in the research.
- If you take back your authorization, information that was already collected may still be used and shared with others, but the researchers will no longer be able to collect NEW information about you.
- If you take back your authorization, you will not be able to take part in the research study.
- To take back your authorization, you will need to tell the researchers by writing to the Lead Researcher, Mei Baker, at 465 Henry Mall, Madison, WI 53706.

Will being in this study help me in any way?

Being in this study will not help you directly. Your participation in the study may benefit other people in the future by helping us improve the lives of children with Angelman syndrome or Prader-Willi syndrome.

What are the study risks?

The most common risks associated with the finger prick are pain and bruising. There is also a risk of loss of confidentiality. Although we will do our best to keep your information safe and secure, there is a risk that your information could become known to someone not involved in this study.

What happens to the information collected for the research?

We have strict rules to protect your personal information and protected health information (PHI). We will limit the use and disclosure of your personal information, including research study and medical records, to people who have a need to review this information.

However, we cannot promise complete confidentiality. Federal or state laws may permit or require us to show information to university or government officials and to study sponsors responsible for monitoring this study. This includes University of Wisconsin

and its representatives and affiliates, including those responsible for monitoring or ensuring compliance, such as the Human Research Protection Program, the Food and Drug Administration, and Ultragenyx.

We may also have to tell appropriate authorities, such as child protective services or health care providers, if we learn during the study that you or others are at risk of harm (for example, due to child or elder abuse, or suicidal thoughts).

Authorizing the research team to use your PHI means that we can release it to the people or groups listed in this form for the purposes described in this form. Once your health information is released outside UW-Madison or UW Health, it may not be protected by privacy laws and might be shared with others.

Also, with appropriate confidentiality protections, we might use information that we collect during this study for other research or share it with other researchers without additional consent from you.

A description of this clinical trial will be available on http://www.ClinicalTrials.gov. This Web site will not include information that can identify you. At most, the Web site will include a summary of the results. You can search this Web site at any time.

Will information from this study go in my medical record?

If you join the study during a clinic visit at UW Health, then a medical record may be created for you if you do not already have one. Your medical record might say that you participated in this study, and a copy of this consent and authorization form might go in your medical record.

If you join the study by returning a blood sample in the mail, then your medical record will not say that you participated in the study.

None of the information we collect for this study will go in your medical record.

Will I receive the results of research tests?

Most tests done as part of a research study are only for research and have no clear meaning for health care. In this study, you will not be informed of any test results or unexpected findings.

Can I be removed from the research without my agreement?

The person in charge of the research study or the sponsor can remove you from the research study without your approval. Possible reasons for removal include:

- you no longer meet the requirements to be in the study
- the study is stopped by the sponsor or researchers

What else do I need to know?

Will I receive anything for participating?
We have included a $10 Amazon gift card in this study packet for considering if you would like to participate in this study. If you complete the blood sample and mail it back to the researchers, you will receive an additional $15 Amazon gift card, for a total of $25.

Your information and blood sample (both identifiable and de-identified) may be used to create products or to deliver services, including some that may be sold and/or make money for others. If this happens, there are no plans to tell you, or to pay you, or to give any compensation to you or your family.

Permission to communicate about the study by email
We are requesting your (or your parent's, if you are under 18) email address so we can remind you about upcoming study visits. Email is generally not a secure way to communicate about your health as there are many ways for unauthorized users to access email. You should avoid sending sensitive, detailed personal information by email. Email should also not be used to convey information of an urgent nature. If you need to talk to someone immediately, please contact the study team at (608) 228-4940. You do not have to provide your email address to participate in this study.

How many people will be in this study?
We expect about 50 people will be in this research study.

Who is funding this study?
This research is being funded by Ultragenyx.

Information about genetic research
This research study involves genetic testing. The Genetic Information Nondiscrimination Act (GINA) prohibits health insurance companies or health plan administrators from requesting or requiring genetic information of you or your family members, or using such information for decisions regarding your eligibility for insurance or your premiums. However, this law does not provide the same protection for disability, life insurance, or long-term care insurance. GINA also prohibits most employers (with 15 employees or more) from using genetic information when making decisions on your employment, including decisions related to hiring, firing, promotion, pay, and job assignments.

Agreement to participate in the research study

You do not have to participate in this research study.

If you participate in this study, it means that:

- You have read this information sheet.
- You and (as applicable) your child have had a chance to ask questions about the research study, and the researchers have answered your questions.
- You are willing to participate in this study.
- You give authorization for your protected health information to be used and shared for research purposes as described in this form.