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Sep 05, 2023

RESEARCH SUBJECT INFORMATION AND CONSENT FORM

TITLE: Pilot study to assess clinical and pivotal biomarkers in the urine to predict the progression of nephropathy in Fabry disease.

PROTOCOL NO.: 23-LDRTC-01
WCG IRB Protocol #20233966

SPONSOR: Lysosomal & Rare Disorders Research & Treatment Center, Inc

INVESTIGATOR: Ozlem Goker-Alpan MD
Lysosomal & Rare Disorders Research & Treatment Center, Inc
3702 Pender Drive, Suite 170
Fairfax, VA 22030
United States

STUDY-RELATED PHONE NUMBER(S): 571-732-4655
703-261-6220
240-643-6003 (24 hours)

Taking part in this research is voluntary. You may decide not to participate, or you may leave the study at any time. Your decision will not result in any penalty or loss of benefits to which you are otherwise entitled.

If you have any questions, concerns, or complaints or think this research has hurt you, talk to the research team at the phone number(s) listed in this document.

RESEARCH CONSENT SUMMARY

In this consent form ”you” refers to the patient or study subject (child or adult).

You are being asked for your consent to take part in a research study. This document provides a concise summary of this research. It describes the key information that we believe most people need to decide whether to take part in this research. Later sections of this document will provide all relevant details.

What should I know about this research?

- Someone will explain this research to you.
- Taking part in this research is voluntary. Whether you take part is up to you.
- If you don’t take part, it won’t be held against you.
- You can take part now and later drop out, and it won’t be held against you
- If you don’t understand, ask questions.
- Ask all the questions you want before you decide.

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How long will I be in this research?
We expect that your taking part in this research will last for 24 months.

Why is this research being done?
The purpose of this research is to collect biological samples (urine) to develop assays for immune biomarkers to possibly in the future be able to screen subjects with Fabry disease and be able to understand better progression of nephropathy in Fabry disease and predict nephropathy in Fabry disease.

What happens to me if I agree to take part in this research?
If you decide to take part in this research study, the general procedures include the collection of medical history, a physical exam, and urine sample collection.

Could being in this research hurt me?
The most important risks that you may expect from taking part in this research include the potential for loss of confidentiality of your data and genetic information.

Will being in this research benefit me?
It is not expected that you will personally benefit from this research. Others may benefit in the future.

What other choices do I have besides taking part in this research?
Instead of being in this research, you can choose not to participate.

This Subject Information and Consent Form may contain words you do not understand. Please ask the study doctor or the study staff to explain any words or procedures that you do not clearly understand. You may take home an unsigned copy of this consent form to think about it or discuss it with your family or friends before making your decision.

A person who participates in a research study is called a research or study subject. In this consent form, "you" generally refers to the research subject. If you are a parent/guardian, please remember that "you" means the research (study) subject.

PURPOSE OF THE SUBJECT INFORMATION AND CONSENT FORM

You are being asked to participate in a clinical research study. This form gives you information about the research study and, if signed, will give your permission to take part in the study. The form describes the purpose, procedures, risks, discomforts, and benefits of the research study. Please read this Subject Information and Consent Form and ask as many questions as needed. You should not sign this form if you have any questions that have not been answered to your satisfaction.

Your doctor will be paid by the Sponsor to conduct this research study.

If you take part in this research study, you will be given a copy of this signed and dated consent form.

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PURPOSE OF THE STUDY

This prospective study aims to establish novel biomarkers for non-invasive detection of kidney involvement in patients with Fabry disease.

SUBJECT PARTICIPATION

You are being invited to take part in this research because you:

1. Have been diagnosed with Fabry disease (FD) with or without kidney involvement.
   Or
2. Have been determined to be a healthy control subject for the study.

A total of 50 subjects, male or female, between the ages of 18 to 80 years of age, including 25 healthy individuals and 25 patients with Fabry disease (5 patients with Fabry disease without clinical evidence of nephropathy, 15 patients with clinical evidence of nephropathy, and 5 NAÏVE (non treated) patients).

If you meet the eligibility criteria and agree to participate in the study, you may qualify to take part in the study. You should take part in the study only if you want to do so. Your signature on this consent form means that you agree to participate in the study, as described in this consent form. You may object to any one of the planned procedures and undergo only those you are comfortable with. You may refuse to take part or withdraw from this study at any time without penalty or loss of benefits to which you are otherwise entitled. If you have Fabry disease, you do not need to participate in this study to receive medical care from your doctor for your Fabry disease.

PROCEDURES

If you agree to take part in this study, first, you will sign this Subject Information and Consent Form before any study-related procedures are performed. We will discuss the study with you in detail so that you can ask questions and give us your permission.

Medical Questions: You will be asked about your medical history. You may also be asked to provide consent for the release of your medical records from previous doctors to your current study doctor.

Physical Exam: Physical examination includes your general appearance, skin, neck, eyes, ears, nose, throat, abdomen, hands, feet, lymph nodes, lungs, and heart, which will be obtained at baseline, 6 months, and 12 months.

Urine Collection: Urine samples will be collected at baseline, 6 months, and 12 months. During the screening period, you will have a urine pregnancy test if you are female and about to have children.

Your samples are collected only for research purposes and will not be sold or distributed for other purposes.

Leftover samples collected throughout the study will be stored and may be used for future exploratory analysis to help learn more about Fabry disease. Samples may be shared with the study sub-investigators. If your samples are used for such purposes, all personal identifiers will be removed from the samples.

Only the study doctor and the research team of LDRTC, Inc. will have access to your file. Collected samples will be analyzed and stored at LDRTC, Inc. Depending on the laboratory test, samples might be shipped to, analyzed, and stored at other institutions.

RISKS AND DISCOMFORTS

This research study may involve specific information on a rare condition called Fabry disease and genetic research. The genetic test is done only for research purposes, and no official reports will be given to you. Sometimes, information obtained through genetic analysis may lead to mental stress to the subject and his/her family in the form of anxiety or guilt if others were to see this information. Family relationships could also be hurt. Stigmatization and discrimination may also result leading to situations in which the research subject may not be able to acquire health or life insurance or benefits otherwise entitled to. To minimize this risk we will protect information about you and your participation in this study to the best of our ability but we do not guarantee this. No personal identity information, but a general identification number will be used to label your samples.

A Federal law called the Genetic Information Nondiscrimination Act (GINA) provides some protection for your genetic information. This law generally will protect you in the following ways:

1. Health insurance companies and group health plans may not request your genetic information collected this research.
2. Health insurance companies and group health plans may not use your genetic information when making decision regarding your eligibility or premiums.
3. Employers with 15 or more employees may not use your genetic information collected in this research when making a decision about your employment.

However, this Federal law does not protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance.

There is also potential loss of confidentiality in the case of giving your information to other people by mistake. To minimize this risk we will protect information about you and your participation in this study to the best of our ability but we do not guarantee this. No personal identifying information will be used to label your blood or urine samples. All blood and urine samples will be given a generic identification number.

The results of this research study may be presented at scientific meetings or in publications. Your identity will not be disclosed in those presentations.

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OTHER RISKS

Your condition may not get better or worse during this study, as this is not a treatment study.

CONFIDENTIALITY AND RELEASE OF MEDICAL RECORDS

We will protect information about you and you are taking part in this research study to the best of our ability. If information about this study is published, your identity will remain confidential. However, the U.S. Food and Drug Administration (FDA) and the WCG Institutional Review Board (IRB) may be granted direct access to your original medical records for verification of clinical trial procedures or data, without violating your confidentiality, to the extent permitted by the applicable laws and regulations. By signing this consent form, you are authorizing such access. A court of law could order medical records shown to other people, but that is unlikely. Therefore, absolute confidentiality cannot be guaranteed. All data obtained during this study will be coded and linked, and be kept locked in a file cabinet in Ozlem Goker-Alpan, MD's office. Data will also be stored on password-protected computers. We might want to publish your pedigree. If so, we will make sure that your family is unrecognized.

A description of this clinical trial will be available on http://www.ClinicalTrials.gov, as required by U.S. Law. 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.

AUTHORIZATION TO USE AND DISCLOSE INFORMATION FOR RESEARCH PURPOSES

What may information be used and given to others?
The study doctor will get your personal and medical information. For example:

- Past and present medical records
- Research records
- Records about phone calls made as part of this research
- Records about your study visits.

Who may use and give out information about you?
The study doctor and the study staff.

Who might get this information?
The Sponsor of this research. "Sponsor" means any persons or companies that are:

- working for or with the Sponsor, or
- owned by the Sponsor.

Your information <u>may</u> be given to:

- The U.S. Food and Drug Administration (FDA),
- Department of Health and Human Services (DHHS) agencies,
- The institution where the research is being done, and
- WCG IRB

Why will this information be used and/or given to others?

- to do the research,
- to study the results, and
- to make sure that the research was done right.

What if I decide not to give permission to use and give out my health information?
Then you will not be able to be in this research study.

May I review or copy my information?
Yes, we will only provide if the information is clinically relevant and after the research is over.

May I withdraw or revoke (cancel) my permission?
Yes, but this permission will not stop automatically. For sites in Maryland, this authorization will expire one year after being signed and dated

You may withdraw or take away your permission to use and disclose your health information at any time. You do this by sending written notice to the study doctor. If you withdraw your permission, you will not be able to stay in this study.

When you withdraw your permission, no new health information identifying you will be gathered after that date. Information that has already been gathered may still be used and given to others.

Is my health information protected after it has been given to others?
There is a risk that your information will be given to others without your permission.

NEW INFORMATION

You will be told about any new information that might change your decision to be in this study. You may be asked to sign a new consent form if this occurs. You may contact the study doctor at any time after your participation ends to find out if any new information about this study has become available.

SUBJECT'S RESPONSIBILITIES

While participating in this research study, you will need to share your medical information correctly to your best ability. We would like to know about medical procedures you are planning to undergo (including surgery).

BENEFITS

If you agree to take part in this study, there will be no direct medical benefit to you. However, this study may increase our understanding of Fabry disease and how disease progresses. We are hoping to find biomarkers to help us in deciding on which treatment options should be used to possibly avoid or reduce complications due to bone problems. We hope that information learned from this study will benefit other patients and families in the future.

COSTS

There will be no costs to you for your participation in this study. You will not be charged for blood draws, urine analysis or laboratory tests related to this research study. Tests and procedures that are done only for the study will not be billed to you or your insurance company.

PAYMENT FOR PARTICIPATION

You will not be paid for your participation in this study.

COMPENSATION FOR STUDY RELATED INJURY

If you are injured as a result of study procedures performed during your participation in this study, you should seek medical attention at the medical provider of your choice. We will cover the medical expenses necessary to treat the injury only to the extent that such cost are not covered by your health insurance policy.

You must follow the directions of the study doctor to be eligible for this coverage.

Neither the Sponsor nor the study doctor has a program in place to provide other compensation in the event of an injury.

LEGAL RIGHTS

The above section does not restrict your right to seek legal assistance. You do not waive any legal rights by signing this Subject Information and Consent Form.

ALTERNATIVE TREATMENT

This study is not intended to provide subjects any direct medical benefit. This is not a treatment study. You have the alternative to not participate.

VOLUNTARY PARTICIPATION

Your participation in this study is voluntary. You may decide not to participate or you may leave the study at any time. When you withdraw your permission, no new health information about you will be gathered after that date. Information that has already been gathered may still be used and given to others as some of your information may have already been analyzed and reported. Your decision will not result in any penalty or loss of benefits to which you are entitled. If you choose, you may request to have your records destroyed.

Your participation in this study may be stopped at any time by the study doctor without your consent for any of the following reasons:

- if it is in your best interest;
- you do not consent to continue in the study after being told of changes in the research that may affect you;
- for any other reason.

QUESTIONS

Contact Ozlem Goker-Alpan MD at 571-732-4655, 703-261-6220 (daytime contact number), or at 240-643-6003 (24-hour contact number) for any of the following reasons:

- if you have any questions about your participation in this study,
- if you feel you have had a research-related injury or a reaction, or
- if you have questions, concerns or complaints about the research.

If you have questions about your rights as a research subject or if you have questions, concerns or complaints about the research, you may contact:

WCG IRB
Telephone: 855-818-2289
E-mail: researchquestions@wcgirb.com.

WCG IRB is a group of people who independently review research.

WCG IRB will not be able to answer some study-specific questions, such as questions about appointment times. However, you may contact WCG IRB if the research staff cannot be reached or if you wish to talk to someone other than the research staff.

Do not sign this consent form unless you have had a chance to ask questions and have gotten satisfactory answers.

If you agree to be in this study, you will receive a signed and dated copy of this consent form for your records.

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CONSENT

I have read this consent form (or it has been read to me). All my questions about the study and my part in it have been answered. I freely consent to be in this research study.

I authorize the use and disclosure of my health information to the parties listed in the authorization section of this consent for the purposes described above.

By signing this consent form, I have not given up any of my legal rights.

CONSENT INSTRUCTIONS

Consent: Subjects 18 years and older must sign on the subject line below.

_____
Printed Name of Subject

_____ _____ _____
Signature of Subject (18 years and older) Date Time

_____

PERSON CONDUCTING INFORMED CONSENT DISCUSSION

I confirm that participant was given an opportunity to ask questions about the study, and all questions asked by the participant have been answered correctly and to the best of my ability. I confirm that the individual has not been coerced into giving consent, and the consent has been given freely and voluntarily.

_____
Printed Name of Person Conducting Informed Consent Discussion

_____ _____ _____
Signature of Person Conducting Informed Consent Discussion Date Time

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WITNESS

I observed the process of consent. The prospective subject read this consent form, was given the chance to ask questions, appeared to accept the answers, and signed to enroll in the study.

_____
Printed Name of Witness

_____ _____ _____
Signature of Witness Date Time

ATTESTATION STATEMENT

I confirm that the research study was thoroughly explained to the subject. I reviewed the consent form with the subject and answered the subject's questions. The subject appeared to have understood the information and was able to answer the following questions correctly:

1. What is the purpose of this study?
2. If you decide to be in the study, what will you be asked to do?
3. What is the possible benefit of participating in this study?
4. What are the possible risks of participating in this study?
5. If you decide not to participate in this study, what options do you have?
6. Will participating in this study cost you anything? If so, what will you have to pay for?
7. Do you have to be in this study?
8. If you decide to be in the study, can you leave the study when you want to?

_____ _____
Printed Name of Person Conducting the Informed Consent Discussion Position

_____ _____
Signature of Person Conducting the Informed Consent Discussion Date