MedStar Research Institute

Informed Consent for Skin Biopsy

SITE:

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INTRODUCTION

We invite you to take participate in a procedure called a Skin Biopsy which is part of the research study titled: "Clinical and Molecular Manifestations of Heritable Disorders of Connective Tissue". The Skin Biopsy procedure will be performed by a medical doctor or a nurse practitioner connected to the study. You were selected as a possible participant in this study because you or a close relative have been diagnosed with or suspected to have a heritable disorder of connective tissue, such as Ehlers-Danlos, Stickler or Marfan Syndrome, or an Overlap Connective Tissue Disorder. Please take your time to read this form, ask any questions you may have and make your decision. We encourage you to discuss your decision with your family, friends and your doctor(s).

WHAT IS THE PURPOSE OF THIS STUDY?

As part of this study you are being asked to have a **skin biopsy procedure** in order to investigate a genetic disorder that you or your family may have. This study may lead to better medical care for patients with Hereditary Disorders of Connective Tissue. Your tissue sample will be kept by the NIA and will be used for research about the genetic disorder in you or your family. Samples will be stored in a secured building at the NIA-IRP or sent to a certified clinical laboratory for analysis. This test is being performed for research purposes. In the event that the results of this study are expected to have clinical implications for your management, we will discuss this with you at the time of the procedure.

WHAT ELSE SHOULD I KNOW ABOUT THIS RESEARCH STUDY?

It is important that you read and understand several points that apply to all who take part in our studies:

- Taking part in the skin biopsy procedure is entirely voluntary and refusal to participate will not affect any rights or benefits you normally have;
- You may or may not benefit from taking part in the procedure, but knowledge may be gained from your participation that may help others; and
- You may decide not to undergo the procedure at any time without any penalty or losing any of the benefits you would have normally received.

The nature of the study, the benefits, risks, discomforts and other information about the study are discussed further below. If any new information is learned, at any time during the research, which might affect your participation in the study, we will tell you. We urge you to ask any questions you have about this study with the staff members who explain it to you and with your own advisors before agreeing to participate.

WHO IS IN CHARGE OF THIS STUDY?

The research is being conducted and sponsored by the National Institute on Aging with Nazli B. McDonnell MD PhD as the principal investigator.

WHO CANNOT PARTICIPATE IN THIS STUDY?

You cannot be in this study if any of the following apply to you:

- If you are unable to give informed consent or do not have a legal guardian who is able to provide such consent in your behalf.
- If you have a significant bleeding disorder that will result in undue risk to you by your undergoing the procedure.
- If you have a known allergy to lidocaine.

While participating in this study, you should not take part in any other research project without approval from the people in charge of each study. This is to protect you from possible injury arising from such things as extra blood drawing, extra x-rays, interaction of research drugs, or similar hazards.

WHAT IS INVOLVED IN THE SKIN BIOPSY PROCEDURE?

If you decide to undergo this procedure it will take place at the National Institute on Aging (NIA) Clinical research unit, named ASTRA (Advanced Studies in Translational Research on Aging).

A skin biopsy is a procedure that involves removing a piece of skin measuring 3-4 mm (about the size of a pencil eraser), usually taken from your inner arm using a small instrument. The area will first be anesthetized (made numb) by injecting a special medicine called Xylocaine, or by the application of Emla topical cream. We will also ask you to make a list of medications that you are taking and any that you are allergic to. At the completion of the procedure, a small dry sterile dressing will be applied to the biopsy area and kept in place for 24 hours. You will be given instructions for care of the biopsy site to take home with you along with the phone number of the doctor and nurse to contact if you have questions. You will be asked to contact the study investigator or nurse to report on the condition of the biopsy site the day of your biopsy.

A separate consent form regarding donation of your de-identified tissue sample for future scientific use to a tissue repository will be discussed with you during your enrollment. Your participation in this program will not affect your eligibility or participation in the "Clinical and Molecular Manifestations of Heritable Disorders of Connective Tissue" study.

HOW LONG WILL I BE IN THE STUDY?

Participation in the skin biopsy procedure is a one-time process beginning when you agree to the procedure and ending when it is concluded.

WHAT ARE THE RISKS AND SIDE EFFECTS OF THIS STUDY?

Although there can be no absolute guarantees, every reasonable effort will be made to keep your personally identifiable information secret so that there will be no misuse. Even when the information is kept secret, if you are asked if you have ever been tested for a genetic disorder, answering "yes" could cause benefits to be denied or could cause other problems including discrimination. Participation in this protocol includes an investigation of genes that can or are suspected to cause hereditary disorders of connective tissue. If a mutation in one of these genes is identified, you will be notified of the result. This information will not be shared with anyone without your consent. If you do not want genetic analysis performed on your blood or tissue samples, or if you do not wish to be notified of the results, please indicate so at the time of the consent.

ARE THERE ANY BENEFITS TO TAKING PART IN THE STUDY?

You may or may not get any direct benefit from being in this study. We cannot promise that you will experience any benefits from participating in this study. We hope the information learned from this study will benefit others in the future.

WHAT OTHER OPTIONS ARE THERE?

Instead of being in this study, you have these options:

- This is a natural history study. You have the option of not participating in the study.

WHAT ABOUT CONFIDENTIALITY?

Your personal health information (PHI) will be kept private to the extent allowed by law. You will not be identified by name in any publications resulting from this study. You will be asked to sign a separate form that will give permission to the investigator, the sponsor, and certain other people, agencies or entities to look at and review the records related to this study including your personal health information and the information discovered during this study.

Organizations that may request, inspect and/or copy your research and medical records for quality assurance and data analysis include groups such as: the National Institute on Aging, Food and Drug Administration, Office of Human Research Protection, MedStar Research Institute, Institutional Review Board (IRB).

A Data Safety and Monitoring Board (DSMB), which is a group of experts not connected to the study, may be reviewing the data from this research throughout the study. We will tell you about the new information from this or other studies that may affect your health, welfare, or willingness to stay in this study. Currently, this study does not have a DSMB.

Records will be kept using Clinical Management Software products called Study Manager and Oracle Clinical Applications. These softwares are HIPAA (Health Insurance Portability and Accountability Act) compliant softwares developed by Advanced Clinical Software and Oracle Corporation respectively. These databases are password protected and maintained on a secure NIA/NIH internet with access limited to authorized NIA staff members. All NIA members who have access to these databases have the proper training on patient confidentiality as well as the required Human Subject Protection Training.

We may also use the Cardiff Teleform Information Capture system for data collection and automated data entry. The Cardiff Teleform system produces machine-readable data collection forms that will be read by a dedicated scanner and entered into a secure, limited access database, maintained by the NIA.

You can stop participating at any time. Any data or blood collected until that point in time would remain part of the study. All data and blood collected is available only to authorized staff working on this protocol.

WILL I BE PAID FOR PARTICIPATING IN THIS STUDY?

You will not be paid for participating in this study. Materials and information obtained from you in this research may be used for commercial or non-commercial purposes. It is the policy of NIA, MedStar Research Institute, MedStar Health, Inc. and its affiliated entities not to provide financial compensation to you should this occur.

WHAT ARE THE COSTS?

You do not have to pay anything to be in this study. However, it is possible that taking part in this study may lead to procedures or care not included in the study. This may lead to added costs for you or your insurance company. **You will not be charged by the National Institute on Aging for tests and procedures that are listed in this consent as a part of this research study.** However, you, or your insurance company, will be charged for any other portion of your care that is considered clinically indicated and standard of care. You may be responsible for any co-payments and deductibles that are standard for your insurance coverage.

WHAT IF I'M INJURED OR BECOME ILL DURING THE STUDY?

We will make every effort to prevent injuries and illness from being in the study. However, you should not expect anyone to pay your any pain, worry, lost of income or non-medical care costs that may result from taking part in this study. No long term medical care or financial compensation for research related injuries will be provided by the National Institutes of Health, the Federal Government, Harbor Hospital, MedStar Research Institute or MedStar Health.

You or your insurance company will be charged for continuing medical care and/or hospitalization that are not apart of the study. By agreeing to this you do not give up your rights to seek compensation in the courts.

WHAT ARE MY RIGHTS AS A PARTICIPANT?

- You have the right to be told about the nature and purpose of the study;
- You have the right to be given an explanation of exactly what will be done in the study and given a description of potential risks, discomforts, or benefits that can reasonably be expected;
- You have the right to be informed of any appropriate alternatives to the study, including, if appropriate, any drugs or devices that might help you, along with their potential risks, discomforts and benefits;
- You have the right to ask any questions you may have about the study;
- You have the right to decide whether or not to be in the study without anyone misleading or deceiving you; and
- You have the right to receive a copy of this consent form.

By signing this form, you will not give up any legal rights you may have as a research participant. You may choose not to take part in or leave the study at any time. If you choose to not take part in or to leave the study, your regular care will not be affected and you will not lose any of the benefits you would have received normally. We will tell you about new information that may affect your health, welfare, or willingness to be in this study.

WHO DO I CALL IF I HAVE QUESTIONS OR PROBLEMS?

For questions about the study or a research-related injury, contact the investigator, Dr. Nazli B. McDonnell at 410 350 7370 or by email mcdonnellna@mail.nih.gov. For medical assistance during the evening or on weekends, call the NIA Security Office at (410) 558-8119 and request that they contact the NIA Physician-on-Call.

If you are having a medical emergency, you should call 911 or go directly to the nearest emergency room.

If you are injured as a result of being in a study, or think you have not been treated fairly, please contact the NIA Clinical Director or Deputy Clinical Director at (410) 350-3922.

For questions about your rights as a research participant, you can call or write the following:

NIA Clinical Director
3001 S. Hanover Street, 5th Floor
Baltimore, MD 21225
Phone (410) 350-3922

NIA Clinical Research Protocol Office
3001 S. Hanover Street, Room 539
Baltimore, MD 21225
Phone: (410) 350-3947
Fax: (410) 350-3979.

MedStar Research Institute
Office of Research Integrity
6495 New Hampshire Avenue, Suite 201
Hyattsville, MD 20783
Phone: (301) 560-7339
Toll Free: (800) 793-7175
Fax: (301) 560-7336

ORAL DOCUMENTATION:

Is there anything you would like me to repeat? (responded) _____*Yes* ___*No*

Have you understood everything I have told you? (responded) ____Yes ___No
Do you have any questions? (responded)____Yes ___No
Do you agree to participate? ___ Yes ___No

I have read the above informed consent to the respondent and he or she agreed to participate in the study titled "Clinical and Molecular Manifestations of Heritable Disorders of Connective Tissue,"
*Signature is recorded on this page.

Print Name of person reading this consent_____
Print Name of witness who observed_____
Date_____ Time_____

SIGNATURES

As a representative of this study, I have explained the purpose, the procedures, the possible benefits and risks that are involved in this research study. Any questions that have been raised have been answered to the individual's satisfaction.

Signature of Person Obtaining Consent _____ Date of Signature _____

I, the undersigned have been informed about this study's purpose, procedures, possible benefits and risks, and I have received a copy of this consent. I have been given the opportunity to ask questions before I sign, and I have been told that I can ask other questions at any time. I voluntarily agree to be in this study. I am free to stop being in the study at any time without need to justify my decision and if I stop being in the study I understand it will not in any way affect my future treatment or medical management. I agree to cooperate with Dr. Nazli McDonnell and the research staff and to tell them immediately if I experience any unexpected or unusual symptoms.

Participant's Signature _____ Date of Signature _____

Signature of Witness _____ Date of Signature _____

Signature of Legally Authorized Representative (When Appropriate) _____ Date of Signature _____

Relationship to Participant (When Appropriate) _____ Date of Signature _____

Principal Investigator's Signature _____ Date of Signature _____

To address Assent or Waiver regarding DHHS protections for children involved in research – 45 CRF 46, Subpart D – add the following section:

Parent's Permission for Minor Patient
I have read the explanation about this study and have been given the opportunity to discuss it and to ask questions. I hereby give permission for my child to take part in this study. (Attach Minor's Assent, if applicable)

Signature of Parent(s)/Guardian _____ Date of Signature _____