BOLER-PARSEGHIAN
Center for Rare & Neglected Diseases

UNIVERSITY OF NOTRE DAME

DEPARTMENT OF BIOLOGICAL SCIENCES
BOLER PARSEGHIAN CENTER FOR RARE AND NEGLECTED DISEASES

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UNIVERSITY OF NOTRE DAME CONSENT TO BE PART OF A RESEARCH STUDY

(ONE-TIME BLOOD, STOOL and TISSUE SAMPLE — MINIMAL RISK)

Co-Investigator: Kasturi Haldar, PhD

Co-Investigator: Suhail Alam, PhD

GENERAL INFORMATION

Purpose of the study

We invite you to participate in a procedure called a Skin Biopsy, which is part of the research study titled, "Isolation of Skin Fibroblasts, Blood and Stool specimens from Rare Disease Patients: Studying Cellular Defects and Developing Novel Therapies". In addition, we ask that you provide a blood sample and/or stool sample.

We are conducting this study to learn more about rare diseases that we research in the Haldar Lab. Currently, we are working on 6 different rare diseases and hope to expand this further. The purpose of this study is to obtain skin fibroblasts, blood and stool specimens from rare disease patients that will be used for cellular and molecular analysis to advance our understanding of a particular rare disease. By studying patient's own cells, we are able to better understand the mechanisms of action that can assist with development of treatments.

What do I need to know about this study?

There is no charge to you or your health insurance for being in this study and your participation in this skin biopsy procedure is entirely voluntary. Even if you agree to join the study now, you can change your mind later and quit. Please note, however, that once we've analyzed information about your cells, we may not be able to take that information out of our research.

1. You are not waiving any of your legal rights by signing this informed consent document.
2. Participation in this study will not affect any rights or benefits you normally have.
3. None of the information that we publish or discuss will enable others to figure out who took part in this study.
4. Like the information in your medical record, the records we create in this study will remain confidential and protected.

What is involved in a skin biopsy?

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 an anesthetic 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. .

How long will I be in this 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 risks and side effects?

If you decide to participate in this procedure, you should know there may be risks. You should discuss these with the investigators and/or your regular doctor and you are encouraged to speak with your family and friends about any potential risks before making a decision. Potential risks and side effects related to the skin biopsy procedure include:

- Local infection that can usually be managed with antibiotic applied to the skin.
- Local bleeding and bruising in the area surrounding the biopsy site.
- An allergic reaction to the local anesthetic is possible.
- Scarring in the area that would measure about 1/8 of an inch long.
- Minor discomfort related to injection of the local anesthetic.

There may also be risks and side effects, other than those listed above that we cannot predict. Many side effects go away shortly after the procedure is stopped, but in some cases side effects can be serious, long lasting and/or life threatening. If you have any unwanted side effects, you should ask the investigator whether there are any medications or other things that may be done to make the side effect less uncomfortable.

What is involved in obtaining a blood and/or stool specimen?

A blood sample is obtained by a licensed phlebotomist or nurse. The procedure involves obtaining 2-5ml of blood through the use of a sterile venipuncture needle apparatus. The area will first be cleaned with an alcohol swab and collection takes place from a superficial vein in the upper limb, generally the median cubital vein. The area will be covered with a sterile Band-Aid.

What are risks and side effects?

If you decide to have a blood sample taken, you should know that there are few risks. You should discuss these with your investigators and/or regular doctor and are encouraged to speak with family and friends about any potential risks.

Potential risks and side effects related to obtaining a blood sample include:

- Local bleeding and bruising in the area surrounding the venipuncture site
- Local infection that can usually be managed with antibiotic applied to the skin
- Soreness at the site of puncture.

What is involved with obtaining a stool specimen?

The procedure for obtaining stool specimens varies by age of the patient. In infants and toddlers still in diapers, a stool sample can be taken directly from the diaper and placed in a sterile container with a lid. All patients that are toilet trained can obtain a stool sample by lining the opening of the toilet with loosely applied saran wrap prior to defecation (releasing stool from the body). The sample is placed in a sterile container with a lid. This procedure is generally done in the home setting and the specimen should be kept in the container and delivered to lab or physician office within 24 hours. Until delivery, the specimen container should be kept in a refrigerator. Your physician will discuss this procedure with you and provide a specimen container.

There are no risks or side effects related to obtaining a stool sample but requires patient to have container ahead of time.

Storage of Specimens for Future Use

As part of this study, we would like to store your skin, blood and stool samples for future research. The future research may be conducted by Suhail Alam, PhD and Kasturi Haldar, PhD or by other researchers that obtain IRB approval for their research. The specimens will be labeled with a code that only Drs. Alam and Haldar can link back to you. Results of any future research will not be given to you or your doctor. The specimens obtained from you in this research may help in the development of a future commercial product. There are no plans to provide financial compensation to you should this occur.

You may request at any time that your research samples be removed from storage and not be used for future research. If you decide you want your samples removed, you may contact Kasturi Haldar, PhD at the University of Notre Dame, 574-631-3372. Once the request is received, and if your samples have not already been used for other research, they will be destroyed.

If you do not make such a request, your specimens will be stored indefinitely or until used. As a rule, the researchers will continue to use information about you until the study is over and will keep it secure until it is destroyed. Limited information about you may continue to be used after the study is over, for other research, education, or other activities. However, use of this information would not reveal your identity.

As long as your information is kept within the University of Notre Dame, it is protected by HIPAA and privacy policies

Your signature in the next section means that you have received copies of all of the following documents:

- This "Consent to Be Part of a Research Study" document. (*Note: In addition to the copy you receive, copies of this document will be stored in a separate confidential research.*
- **Other (specify):_____**

CONTACT INFORMATION

To find out more about the study, to ask a question or express a concern about the study, or to talk about any problems you may have as a study subject, you may contact one of the following:

Co-Investigators: Kasturi Haldar, PhD Suhail Alam, PhD Mailing Address: University of Notre Dame 103 Galvin Life Sciences Notre Dame IN 46556 Telephone: 574-631-3372 Email: khaldar@nd.edu salam@nd.edu	Study Coordinator: Corianne Kellems Mailing Address: University of Notre Dame 107 Galvin Life Sciences Notre Dame IN 46556 Telephone: 574-631-3372 Email : ckellems@nd.edu

You may also express a concern about a study by contacting the Institutional Review Board:

University of Notre Dame
Institutional Review Board
University of Notre Dame Office of Research
940 Grace Hall, Notre Dame, IN 46556
Phone (574) 631-1461 Fax (574) 631-6630
compliance@nd.edu

SIGNATURES

Research Subject:

I understand the information printed on this form. My questions so far have been answered.

Signature of Subject:_____ Date:_____

Name (Print legal name): _____

_____ Date of Birth: _____

Waiver of Assent

The assent of _____ (name of child/minor) was waived because of:

Age Maturity Psychological state of the child

Legal Representative (if applicable):

Signature of Person Legally
Authorized to Give Consent_____ Date:_____

Name (Print legal name):_____ Phone: _____

Address:_____

Check Relationship to Subject:

☐Parent ☐Spouse ☐Child ☐Sibling ☐Legal Guardian ☐Other: _____

_____

Co-investigators

I have provided this participant and/or his/her legally authorized representative(s) with information about this study that I believe to be accurate and complete. The participant and/or his/her legally authorized representative(s) indicated that he or she understands the nature of the study, including risks and benefits of participating.

Co-investigator 1 Legal Name: **Kasturi Haldar, PhD**

Title: **Director, Boler Parseghian Center for Rare And Neglected Diseases**

Signature: Kasturi Haldar
Date of Signature (mm/dd/yy): 11/28/2017

Co-investigator 2 Legal Name**: Suhail Alam, PhD**

Title: **Research Scientist, University of Notre Dame**

Signature: 
Date of Signature (mm/dd/yy): 11/28/2017