RESEARCH REPOSITORY (RR) FOR HERITABLE DISORDERS OF BONE, BLOOD VESSELS AND SKIN

UNIVERSITY OF WASHINGTON, SCHOOL OF MEDICINE, DEPARTMENT OF PATHOLOGY
SEATTLE, WA 98195-7470

RR Adult Consent Form

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Toll Free Number: 1-888-288-7362
Fax: 1-206-616-1899

RESEARCHER'S STATEMENT

We are asking you to be in a research study. The purpose of this consent is to give you the information you will need to help you decide whether to participate in the study. Please read the form carefully. You may ask questions about the purpose of this research, what we would ask you to do, the possible risks and benefits, your rights as a volunteer, and anything else about the research or this form that is not clear. When we have answered all of your questions, you can decide if you want to be in the study. This process is called "informed consent". We will give you a copy of this form for your records.

PURPOSE OF THE STUDY

We are trying to find out what causes inherited connective tissue disorders that involve bones, skin, and blood vessels. These disorders are passed down in families. The diseases include osteogenesis imperfecta (OI), Ehlers -Danlos syndrome (EDS), and others. Our research lab studies proteins called collagens. We also study the genes (the DNA) that tell the cells how to make collagens. We work with cells and tissues from people with these disorders because we want to see how the changes in the DNA and proteins cause the disorders. We also gather medical information about these people to understand the clinical aspects of the disorders.

We are asking you to give consent to take part in our research study because your doctor believes that you or your family may have a genetic collagen disorder. There are two ways to be involved in this study. One is to sign up at the time that a sample from you is already being tested in the Collagen Diagnostic Lab*. Another way is to have a sample sent in for the study only. In either case, we are asking to store and use your cells and/or DNA for research and to review medical information about you from your medical records. We are also asking you to consent to share your sample and your genetic information with other collagen researchers. The sample and information that we share with them will not identify you. (*housed at the same location)

STUDY PROCEDURES

Sample Collection & Storage: The sample collection is the same whether you are having clinical testing or sending a sample for research only. We will ask you for a saliva (spit) sample, a blood draw or a skin biopsy or a combination of these. If we want to look at the genes, we will ask for saliva or a blood sample. To look at proteins, we need a skin biopsy.

□ Saliva sample: A "spit kit" with step by step instructions will be provided to you to collect the saliva sample.

□ Blood draw: About 3 teaspoons of blood will be drawn from a vein in your arm.

□ Skin biopsy: A circle of skin about 1/8th of an inch in diameter and 1/8th of an inch deep will be taken by using a small punch instrument. The skin is first numbed using local anesthetic injected through a small needle.

We will store the sample and medical information for an open-ended period of time in our Laboratory in the Department of Pathology at the University of Washington.

Review of Health Information: We will look at your medical history and family history as part of the study. When the sample is sent to us for testing or research we will also get medical information from your doctor. If you are a patient at the University of Washington we will get medical information from the hospital chart. We may also ask to look at records from another clinic or hospital. If so, we will ask you to sign a separate "request for medical records" .

Optional Contact of Relatives: When we look at your family history, we may find family members that we want to include in the study. We will ask you to contact the relative and give them a consent form that describes the study. This is optional and will not change your participation in the study if you choose not to do so.

Database Data Entry: We may choose to share your genetic information and medical data with a "disorder specific mutation database". In addition, the National Institutes of Health have set up a health research database of genetic (genotype) and health (phenotype) data. Genotype data includes genetic information. Phenotype data is health information like blood pressure, cholesterol levels, or information that is specific to the disorder.) The purpose of this database is to allow many different researchers to use a large pool of data. This data will help them to understand how genes cause human disease. If you agree to allow us to send your data to these databases it will only be identified by a code and never your name. There is a question at the end of this consent form where you can choose whether or not we can share this information with these databases.

BENEFITS OF THE STUDY

Information learned from studying your cells may benefit others with the disorder. The research may or may not benefit you or your family.

RISKS, STRESS, AND DISCOMFORT

You may feel embarrassed or as though your privacy has been violated when you answer personal medical questions. Having a blood draw or a skin biopsy may cause some discomfort. The blood draw may cause some pain and bleeding where the needle is inserted. There is a small risk of infection. Some people feel faint when having their blood drawn. Be sure to tell the person drawing your blood if this has happened to you in the past. The skin biopsy may leave a small scar after it heals. There is a small chance of a reaction to the skin anesthetic. Please tell the person taking the biopsy if you have had any reaction to anesthetic in the past.

SOURCE OF FUNDING

The study team receives financial support from the Freudmann Foundation.

CONFIDENTIALITY OF RESEARCH INFORMATION

Your participation in the study is voluntary and confidential. The study records will be stored in a computer that only the researchers can access. We have no plans to share your data with anyone outside the study staff. The data in the records will be identifiable and will remain so in order to connect family members. The data will be stored for as long as the study is open, which may be indefinite. Government or university staff sometimes review studies such as this one to make sure they are being done safely and legally. If a review of this study takes place, your records may be examined. The reviewers will protect your privacy. The study records will not be used to put you at legal risk of harm.

We have a Certificate of Confidentiality from the federal **National Institute of Health; National Institute of Arthritis and Musculoskeletal and Skin Diseases.** This helps us protect your privacy. The Certificate means that we do not have to give out identifying information about you even if we are asked to by a court of law. We will use the Certificate to resist any demands for identifying information.

We can't use the Certificate to withhold your research information if you give your written consent to give it to an insurer, employer, or other person. Also, you or a member of your family can share information about yourself or your part in this research if you wish.

There are some limits to this protection. We will voluntarily provide the information to:

- a member of the federal government who needs it in order to audit or evaluate the research;
- individuals at the University of Washington, the funding agency, and other groups involved in the research, if they need the information to make sure the research is being done correctly;
- the federal Food and Drug Administration (FDA), if required by the FDA;
- someone who is accused of a crime, if he or she believes that our research records could be used for defense;
- Law enforcement authorities, if we learn of child abuse, elder abuse, or the intent to harm yourself or others.

OTHER INFORMATION

There will be no charge to you for laboratory studies undertaken as part of this research. We may publish the results from the research study in a medical journal to educate doctors and

scientists and to aid further research. If published, the report will not identify you. No money or other payment will be given to you for participating in this project. If you want to have your sample destroyed, please contact Dr. Byers directly or make the request through your physician.

Please contact any of research investigators listed on the first page of the consent form to discuss these issues further. There is a toll free number – available specifically for questions about consent. (1-888-288-7362).

COMPENSATION FOR INJURY

If you think you have an injury or illness related to this study, contact the study investigator, Dr. Peter Byers at 206-543-4206, right away. He will treat you or refer you for treatment. The UW will pay up to $10,000 to reimburse for treatment of injury or illness resulting from the study. No money has been set aside to pay for things like lost wages, lost time, or pain. However, you do not waive any rights by signing this consent form.

Investigators Signature: Investigators Printed Name date

SUBJECT'S STATEMENT

This study has been explained to me. I volunteer to take part in this research. I have had a chance to ask questions. If I have questions later about the research, I can ask one of the researchers listed above. If I have questions about my rights as a research subject, I can call the University of Washington Human Subjects Division at (206) 543-0098. I give permission to the researchers to use my medical records as described in this consent form. I will receive a copy of this consent form.

Signature of Subject Printed Name Date

Please read below and check the box with your decision for each question:

Sharing Cells: I agree that my cells or molecules derived from them, like proteins or DNA, may be sent to other investigators for research purposes.

☐ YES ☐ NO

Sharing Data: I agree that my genetic data and a brief medical history may be sent to collective databanks.

☐ YES ☐ NO

Receiving results: I wish to be informed if new information is derived from the study of my cells or DNA.

☐ YES ☐ NO

APPROVED

SEP 23 2013

UW Human Subjects Review Committee

Recontact: You have my permission to contact me in the future to request permission for my participation in additional clinical research studies.

☐ YES ☐ NO

Contact information:

Referring Physician: _____
Address: _____
City: _____ State: _____
Telephone: _____ Phone: _____
E-mail: _____

My contact information:
Address: _____
City: _____ State: _____
Zip: _____ Phone: _____
E-mail: _____

APPROVED

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