

**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

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RR Proxy Consent Form

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Melanie G. Pepin, LGC, Licensed Genetic Counselor
Dru Leistriz, LGC, Licensed Genetic Counselor
Sam Bailey, LCG, Licensed Genetic Counselor
Department of Pathology

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 or not to allow this. 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 or not. 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 for your family member to take part in our research study because your family member is deceased or not able to provide their own informed consent. We are interested in your relative because they may have a genetic collagen disorder. There are two ways that your relative could be involved in this study. One is to enroll when a sample from your relative is sent for testing to the Collagen Diagnostic Lab*. Another way is to have a sample from your relative sent in for the study only. In either case, we would like to store and use your relative's cells and/or DNA for research. We would also like to review medical information about him/her from his/her medical records. As part of our research, we would like to share your relative's sample and genetic information

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with other collagen researchers. The sample and information that we share with them will not identify your relative. (*housed in the same location)

STUDY PROCEDURES

Sample Collection & Storage: The sample collection is the same whether your relative is having clinical testing or sending a sample for research only. We will use 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.

If your relative is living but unable to provide their own consent:

- 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 the 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.

If your relative is deceased:

- Stored sample of blood, DNA or cultured cells will be submitted.

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 relative's medical history and family history as part of the study. When the sample is sent to us for testing or for research we will also get medical information from your relative's doctor. If your relative was 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 we want to do that we will ask you to sign a separate "request for medical records".

Optional Contact of Relatives: When we look at the 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 relative's participation in the study if you choose not to do so.

Database Data Entry: We may choose to share your relative's 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 includes health information like blood pressure, cholesterol levels or information that is specific to a 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 the data of your relative to these databases it will only be identified by a code and never by your relative's 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

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

RISKS, STRESS, AND DISCOMFORT

If your relative is having a blood draw or a skin biopsy, it 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. The skin biopsy may leave a small white scar after it heals. There is a small chance of a reaction to the skin anesthetic. Please tell the person taking the biopsy if your relative has have had a reaction to anesthetic in the past.

SOURCE OF FUNDING

The study team receives financial support from the Freudmann Foundation.

CONFIDENTIALITY OF RESEARCH INFORMATION

Your relative's 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 without your permission. 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 relative's records may be examined. The reviewers will protect your relative's privacy. The study records will not be used to put your relative 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 your relative 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.

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and to aid further research. If published, the report will not identify your relative. No money or other payment will be given to you or your family 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 your relative has had 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 _____

PROXY'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.

Name of Subject	Printed Name and Signature of Proxy	Date
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Please read below and check the box with your decision for each question:

Sharing Cells: I agree that my relative's 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 relative's 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 relative's cells or DNA.

YES NO

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

YES NO

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Contact information:

Referring Physician: _____
Address: _____
City: _____ State: _____
Telephone: _____ FAX: _____

My contact information:

Address: _____
City: _____ State: _____
Telephone: _____ FAX: _____
E-mail: _____

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