June 7, 2011

Identification and Characterization of Diagnostic Rare Disease Biomarkers

INFORMED CONSENT

Purpose of Research: This study is being performed under the direction of the principal investigators Drs. Sterling and Osborne who are attempting to identify and characterize better diagnostic biomarkers of rare diseases. The research is funded in part by a grant from the National Science Foundation. The National Organization of Rare Disorders is a partner and plays an advisory role in this project. Many rare diseases are difficult to diagnose and patients with the same rare diseases often have different clinical responses to specific therapies. This research project is designed to give us a better understanding of the similarities and differences in these patient populations. These studies are also designed to help select specific therapy for individual patients (personalized medicine). Whether more knowledge about rare disease biomarkers will actually lead to more effective treatments is not yet known.

Subject Selection: You are invited to participate in an experimental study to identify and characterize new diagnostic biomarkers, cells, proteins and/or nucleic acids, of rare diseases. You are being asked to participate because you either have a specific rare disease or are a healthy control subject.

Subject Participation: You will be asked to donate a sample of your blood, obtained during the same needle stick as your next scheduled blood draw from your primary physician. A qualified technician will use a hollow needle to enter a vein in your arm through the skin under sterile conditions. Up to five (1-5) tubes of blood (approximately 10 – 50 mL, which is equivalent to 3 – 15 teaspoons) will be removed from your vein through the needle. After the blood is collected, the needle will be withdrawn from the vein and pressure will be applied to the site of the puncture for one minute or more to prevent further bleeding. The procedure will take no more than five minutes of your time.

Use of your donated samples: Your donated samples will be used now and/or in the future in studies that compare biomarkers from patients with rare diseases to normal volunteers. The samples will be sent overnight to the Center for Biomarker Research at the Keck Graduate Institute. The samples will be treated with reagents to identify biomarkers and analyzed one cell at a time in a flow cytometer. We plan to store your sample and perform additional protein, genetic, or other testing in the future for the expressed purposes of this study.

Right to withdraw: A subject with a linked sample always has the right to withdraw from a study. You have the right to refuse to allow your samples to be studied now or saved for future study. You may withdraw from this study at any time.

Participation in this study is voluntary: You are free to take part in or withdraw from being a donor at any time. Your decision whether or not to participate will not prejudice you or your medical care. At the discretion of the protocol director, subjects may be taken out of this study due to unanticipated circumstances.

Potential benefits to medical science: Better understanding of how to diagnose and treat individuals with rare diseases may facilitate development of new more effective therapies that will improve future medical care for rare diseases.

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Subject's Initials Date

Potential Risks. The donation of blood described above may involve the following risks and/or discomforts:

1. Pain and/or bruises commonly occur at the site of puncture.
2. Less commonly, there may be formation of a small blood clot, swelling of the vein and surrounding tissue, or bleeding from the puncture site.
3. Rarely, infection may result from the puncture.
4. Lightheadedness and/or fainting may occur.

Disease testing and genetic research raises certain questions about informing you of any results. Possible risks of knowing results include: anxiety; other psychological distress; and the possibility of insurance and job discrimination. A possible risk of not knowing includes being unaware of the need for treatment. These risks can change depending on the results of the research and whether there is a treatment or cure for a particular disease. Sometimes patients have been required to furnish information from genetic testing for health insurance, life insurance, and/or a job. Donation of tissues for these research purposes is not genetic testing. (However, if you are interested in such clinical testing or genetic counseling, you should contact your physician.)

If requested, the results of these studies will be reported to your primary physician and not directly to subjects. Even with special precautions, there is no absolute protection against discrimination on the basis of disease or genetic information. For this reason, the investigator will only use the results of this study for research on diagnostic biomarkers. Disease or genetic information from tissue research can sometimes apply to family members. The investigators will not provide genetic information about you directly to you or to your family members.

Review of your medical records, confidentiality, and the potential for future contact: Your sample will be bar coded to protect your identity and we will not have access to your medical records. The bar code information will be controlled by one of the principal investigators, Jim Osborne, and will not be available to other project investigators. If additional samples or information are needed for the study, we will contact your primary physician and you will decide if you want to participate in future studies.

As a human subject you have the following rights: These rights include but are not limited to the subject's right to:

- be informed of the nature and purpose of the experiment;
- be given an explanation of the procedures to be followed in the medical experiment, and any drug or device to be utilized;
- be given a description of any attendant discomforts and risks reasonably to be expected, if applicable;
- be given a disclosure of any appropriate alternatives, drugs or devices that might be advantageous to the subject, their relative risks and benefits;
- be informed of the avenues of medical treatment, if any available to the subject after the experiment, if complications should arise;
- be given an opportunity to ask questions concerning the experiment or the procedures involved;
- be instructed that consent to participate in the medical experiment may be withdrawn at any time and the subject may discontinue participation without prejudice;
- be given a copy of the signed and dated consent form;

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Subject's Initials Date

- be given the opportunity to decide to consent or not to consent to a medical treatment experiment without intervention of any element of force, fraud, deceit, duress, coercion or undue influence on the subject's decision.

There will be no payment for participation in this study.

Your signature indicates that you have read and understand the above information, that you have discussed this study with the Principal Investigator and his or her staff, that you have decided to participate based on the information provided, and that a copy of this form has been given to you.

_____ _____
Signature of Subject Date

_____ _____
Signature of Parent or Guardian Date

_____ _____
Signature of Investigator or Witness Date

This study and its procedures have been approved by the Claremont Graduate University Institutional Review Board. This Board is responsible for ensuring the protection of research participants.

Contact Information
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Claremont Graduate University, irb@cgu.edu, 909-607-9406

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Subject's Initials Date