NorthShore
University HealthSystem

Research Institute

IRB APPROVAL DATE 6/25/14

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# *Genomic Health Initiative (GHI)*

## *northshore.org/ghi*

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Sponsor: NorthShore University HealthSystem

*IRB Approval date: 6/25/14*

This Consent Form gives information to help you decide whether or not to participate in this research study. If you have any questions, you can ask the research staff or call the Genomic Health Initiative at (224) 364-7470.

**Purpose.** We are asking you to enroll in the study since you are an adult NorthShore patient. If you decide to participate, we will collect, process, and store your extracted electronic health record data and samples, along with data and samples from many other volunteers. We (Genomic Health Initiative) will carry out research on these samples and data.

Our broad objective is the promotion of health at NorthShore and for society as a whole. Specifically, we plan to achieve this through genomics research. Human genomics is the study of genes and their functions at the level of cells up to the whole human organism, as well as how genomic abnormalities cause or predispose to disease. Our goals are:

1- Identification of genetic risk factors to enable reduction of risk for common adulthood diseases. These disease risks can then be minimized by approaches such as diet, lifestyle, or other environmental modifications.

2- Detection of genetic risk factors before clinical diagnosis becomes feasible. This will enable preventive or earlier treatment interventions.

3- Development of new treatment interventions. As the biology of a disease becomes better known, researchers can find potential new targets for treatment interventions.

We are planning to enroll thousands of participants over the next several years. We hope you will participate.

**Nature of the Research.** Participants donate their blood and leftover tissue samples, and allow us access to their NorthShore electronic health record to conduct genomic studies. Living organisms, such as humans, are made of cells. Genes provide a set of instructions to cells which are fundamental for cell function. Genes are also responsible for the transmission from parents to children of traits, such as the color of the eyes and the predisposition to certain disorders. DNA variants are differences in the sequence of building blocks within the DNA molecule. DNA variants can also be deletions and duplications, where parts of the DNA are missing or present in excess, respectively.

Much of our research will involve comparing DNA variation between groups of volunteers with and without various conditions to search for genetic risk variants. Our approach maximizes the research value because we will analyze your genomic data in combination with your NorthShore electronic health records. We will investigate your whole genome at once. The resulting samples and data will be useful for research on many diseases; that is not just for the study of exclusively one particular disease. This approach has many advantages. For example, a research participant might have asthma, but otherwise be healthy. His/her DNA would be very useful to studies of asthma. However, the same biological sample and data can be used as a "control" or comparison sample for studies focused on diseases unrelated to asthma, such as obesity or diabetes. This greatly increases the research usefulness of each volunteer's sample.

Besides studying DNA variants, we will also conduct functional genomic studies. Examples of the type of research include the analysis of gene methylation, which is a common DNA modification that regulates gene expression. Changes of gene methylation can influence a person's response to environmental changes, such as diet, exercise, or exposure to infections. Thus, we will also study certain other characteristics of the collected samples, such as which genes are expressed, the amounts of the corresponding proteins in plasma, and metabolic screens. This is important because many disease processes affect gene expression, protein synthesis, and metabolism. Together, this gives an integrated view, known as "systems biology".

We will perform experiments that analyze all genes at one time (known as genomic scale experiments), which is a new powerful research design. However, our laboratory research methods will change over time, as biomedical technology is rapidly progressing. We use the above general descriptions since we cannot predict every specific research question that will arise over the coming years and be studied with the Genomic Health Initiative. We may preserve your blood cells for the planned studies mentioned above (genetic, expression, protein, metabolism). However, we will not use your preserved cells for isolating stem cells, inducing cells to become stem cells, producing cell lines, or making clones under this protocol.

**Procedures.** You are asked to donate three small extra tubes of blood, less than one tablespoon in total, exclusively for research use. This blood will be collected at your visit to any of NorthShore's hospital outpatient labs or patient service centers -- please refer to www.northshore.org/lab-services/locations for a list of these designated laboratories and their locations. This can be at an already scheduled clinical blood draw, without needing an extra needle stick, or you may instead request a research blood draw at any time.

You are also asked to allow the use of future leftover biological samples, such as urine or blood, for research. These are clinically unused leftover samples from your laboratory tests that would normally be discarded. This does not require any action from you, such as a separate sample collection or for you to be recontacted. We will study your donated samples now and in the future. All samples are stored in secured freezers at NorthShore,

and these samples will be labeled with a unique research code instead of your name or other identifying information.

From time to time we will update the research database with information from your electronic health record, such as demographic information, medical history, test results, medical procedures, images (such as X-rays), medicines you take, and other information in your electronic health record. Some of this clinical information could be considered sensitive, such as various diagnoses, behaviors, and conditions.

Your privacy is very important to us, and we will make every effort to protect it. Information you provide will be recorded in the research database at NorthShore. All identifiable computer records will be maintained on an internal network that is not accessible from the Internet. Access to this research database is restricted to study-associated personnel.

Your genetic research results will not become part of your electronic health record. Though we will not give you raw data from research done using your sample, you will be able to get news about general research findings from studies done and publications on the Genomic Health Initiative website (northshore.org/ghi). This study website will also have information about ongoing studies on the Genomic Health Initiative sample, which you can review in case you might have specific interests or concerns. Please also note the re-contacting section at the end of this consent form regarding suggestions for potential follow-up clinical testing.

**Other choices.** Your participation is voluntary. The alternative is to not participate. Whether or not you decide to participate, or if you later withdraw, your care at NorthShore will not be affected.

**Benefits.** If you decide to participate, there will be no direct benefit to you. This study may allow doctors to learn more about the genetic influences on various health conditions. We hope such knowledge will contribute to improvements in prevention, diagnosis, and treatment for various diseases. You may indirectly benefit by feeling that you are helping people in the future

**Risks.**

*Blood draw:* The blood draw risks include faintness, inflammation (redness and/or warmth) of the vein, pain, bruising, bleeding at the site of puncture, or (rarely) infections. Care will be taken to avoid such side effects.

*Information Privacy:* During this research, we will collect identifiable personal health information (PHI) from your electronic health record; specifically, your name, address/es, date-of-birth, phone number/s, fax number/s, email address/es, Social Security number, and medical record number. We refer to this information simply as “PHI identifiers.” The PHI identifiers in the research database are limited in this study to information contained in your electronic health record already. We have established systems to protect your PHI.

Even with the special precautions there is no absolute guarantee of confidentiality. There is a possible confidentiality risk that someone could access your stored data or trace the de-identified information in a scientific database back to you since your genetic information is unique to you. However, it should be noted that there are Federal laws against the misuse of genetic information, most notably the 2008 GINA (Genetic Information Nondiscrimination Act) law, which applies to employment and health insurance (but not to life, disability, or long-term care insurance).

Except as required by law, or as detailed in this form, we will not release identifiable information. To help ensure confidentiality, we will assign a research number to you and your name will not be used. We have obtained a Certificate of Confidentiality from the National Institutes of Health, which will be instrumental to help us protect your privacy, and allows us to legally refuse to disclose information that identifies you in any federal, state, or local civil, criminal, administrative, legislative, or other proceedings (even from a court order or subpoena). Note that this Certificate of Confidentiality does not prevent you from voluntarily releasing information about yourself, such as your involvement in this research study. Some situations legally require mandatory reporting (breaking confidentiality) such as certain communicable diseases or to protect someone from abuse or harm. When the results of the study are published in a medical book or journal or used to teach others, your PHI identifiers will not be shown.

Much of this research will occur at NorthShore. However, because of the collaborative nature of science, some research could occur elsewhere. Thus, genetic, or other biological data may be shared in scientific databases outside of NorthShore, including the National Institutes of Health, but we will not share identifying clinical information. Your biomaterials may be distributed among collaborators. Any data or biomaterial being shared outside of the Genomic Health Initiative will only have a unique code number without any identifying information. Information about the code will be kept in a secure location at NorthShore only. We will enter into collaborations only with scientists from institutions that abide by appropriate federal regulations and implement strict confidentiality protections. Researchers who want to study the information in those databases must obtain permission from the institutions managing those databases, and such researchers also must guarantee they would not attempt to identify you.

**Who may see, use, and share your PHI identifiers?:** The NorthShore research staff involved in this Genomic Health Initiative, the NorthShore Institutional Review Board (IRB) that oversees the research, and US agencies that oversee or review research, such as the National Institutes of Health. By agreeing to participate in this study you are authorizing the Genomic Health Initiative researchers to use your PHI identifiers. Your PHI identifiers will only be used for the purposes described in this Consent Form.

**Payments and costs:** There are no payments or costs to you for being in this study.

**How long will the study last?** This protocol does not have an expiration date. However, you can withdraw at any time.

**How can I withdraw from the study?** You can withdraw from this study anytime during your life by contacting the research staff at (224) 364-7470. We will send you a form so you can tell us in writing that you would like to withdraw. We would destroy your sample and remove your data from the research database. This will not have any effect on your clinical electronic health record information that resides at NorthShore.

If your genetic and health information or biosample (such as a DNA sample) were placed in scientific databases or repositories (not managed by NorthShore) or published upon before you withdrew, those may continue to be retained there. However, we would request their removal. Any information already obtained at the time you withdraw may continue to be used if necessary to ensure study integrity. We would still retain your information, biomaterials, and data derived from them for the period required by the policies of the respective scientific journal or funding agency.

**What are my rights as a research volunteer?** You may get more information about your rights as a research volunteer from the Chairperson or administrators of the IRB at (224) 364-7100. These are the people you should contact about any problems that happen during the research study.

**Will the researchers recontact me?** Qualified researchers will be able to apply for accessing samples and health information collected in this study. The Genomic Health Initiative scientific access committee will review each study data or biomaterial access request, and for any recontacting for future research. You have no obligation to volunteer for future studies. Each future study involving recontact will have its own consent process, which will provide you details to help you decide whether or not to take part. The NorthShore IRB will also review the applications.

This study may yield clinically actionable findings from the analysis of your biological sample. Clinically actionable findings are those for which therapeutic and/or preventive interventions are available. You have an option to receive clinically actionable findings from our research on your samples. We will carefully consider what results should be returned to you and your doctor, if such sharing is legally permitted at the time. A genetics counselor may recontact you to suggest you obtain additional confirmatory testing in a clinically certified laboratory informed by our research findings. This will then be followed by appropriate specific genetic counseling. Please note that the genetics field is still searching for consensus regarding medical actionability, which will evolve over time. Please visit our study website at northshore.org/ghi to learn more.

If you choose to receive clinical follow-up after this recontacting, the information generated by this process would become part of your clinical records and you (or your insurance) would be responsible for any costs including the confirmatory testing, since such activities would then be out of the scope of this research study. Some samples will be studied earlier than others, and you should not interpret lack of recontact from us as indicating you have no identifiable genetic risks. It is important for you to understand that you should not delay or omit other clinical checks or interventions because you have volunteered for this study.

**Please indicate your willingness to be recontacted by checking the appropriate "yes" or "no" box for each question, knowing that you can still take part in this study regardless of your answers:**

**Somebody may contact me with offers to take part in other studies.** ☐ **YES** ☐ **NO**

**Somebody may contact me with offers to make a clinical referral deemed appropriate for me based on my electronic health record and genetic information.** ☐ **YES** ☐ **NO**

**CONSENT TO PARTICIPATE:** I understand that Dr. Gejman and his research associates and assistants will supervise the study. I have read this consent form or have had it read to me. I understand I may obtain a copy of this consent form from the study website, or request a hardcopy at any time. I understand what will happen if I enroll in this research study. I understand the possible benefits and risks of the study. I give permission for the research study activities described in this consent form.

| | |
|---|---|
| **Volunteer's Name (Please PRINT)** | |
| **Volunteer's Signature** | |
| **Date Volunteer Signed** | |

This consent form is for informational use only. To consent for this study, please come to one of our in-person consenting/enrollment locations such as the Evanston Hospital Outpatient Laboratory, or consent/enroll electronically through NorthShore*Connect* (click on the "Enroll Now" button for GHI after logging onto NorthShore*Connect*).