Consent Form: Example 2 (DNA Sequencing)

Important note: *This model language was developed for the NHGRI Medical Sequencing Project (MSP). It is included here only as an example of how to describe a sequencing project to potential research participants. NHGRI staff drafted the original language for this consent and have had input from numerous outside experts and advocates. The reading level of this document is above high school and may be too high for many populations*

THE NATIONAL HUMAN GENOME RESEARCH INSTITUTE
MEDICAL SEQUENCING PROGRAM

Model Consent Language

1. Purpose of the Project

We would like to invite you to participate in a research project called the Medical Sequencing Project (MSP). The purpose of the MSP is to discover genetic changes associated with diseases. This should lead to better ways to prevent, detect, and treat many human diseases. This project is being sponsored by the National Human Genome Research Institute (NHGRI). The NHGRI is part of the government agency known as the National Institutes of Health (NIH).

Body tissues are made up of cells. Cells contain DNA, which is your unique genetic material that carries the instructions for your body's development and function. Many diseases can result from changes in a person's genetic material that cause cells to not work properly. Currently, researchers and doctors know some of the genetic changes that can cause disease, but they do not know all of the genetic changes that can cause disease.

The MSP is designed to identify most of the genetic changes that can cause diseases in people. Therefore, we would like to study the genetic material from you as part of the MSP. We will compare the DNA from people with certain diseases to the DNA from people without those diseases to find the differences that exist. By combining this information with information from your medical records, it may be possible to identify the genetic changes that are associated with your particular type of disease. We will perform this same process with hundreds of other people [dozens-if rare disease] who have agreed to participate in this research project. By studying many different kinds of diseases in this way, we expect to identify most of the genetic changes associated with those diseases. Since we also will combine genetic information with information from medical records, such as different people's responses to treatments, this project could lead to more knowledge about why certain people respond differently to a treatment. With such knowledge, future treatments potentially could become customized to a patient's unique genetic make-up.

2. Description of the Research

Collection of Samples and Medical Information

- If a blood sample (or other tissue sample) was collected from you, we seek permission to receive some of this blood (or other tissue) and genetic material that already may have been extracted from this blood (or other tissue).
- If an adequate blood sample is not available for this project, we will collect a sample from you by drawing about 4 tablespoons of blood from a vein in your arm. If you object

to having blood drawn, we will collect tissue from you by swabbing cells from the inside of your cheeks.
- We also will collect information from your medical records, including your age, ethnic background, diagnosis, disease history, medical treatments, and response to treatments.

Coding of Tissue Samples and Medical Information
- Your blood (or other tissue) sample and medical information will be labeled with a code.
- Only Dr. _____ at (Institution) will have the information that matches the code to traditionally-used identifying information, such as your name, address, phone number, or social security number. Dr. ___ will keep the information that matches the code to this traditionally-used identifying information in a safeguarded database. Only very few, authorized people, who have specifically agreed to protect your identity, will have access to this database. All other researchers and personnel, including those who will be working with your samples and medical information, will not have access to any of the traditionally-used identifying information about you.

Storage and Release of Samples and Medical Information
- Your coded blood (or other tissue) samples will be sent to an NHGRI-sponsored sequencing laboratory for detailed analysis. Remaining portions of your samples will be stored for an unlimited period of time for future use in research related to diseases or, perhaps, in other research projects.
- Information from analyses of your coded samples and your coded medical information will be put into databases along with information from the other research participants. These databases will be accessible by the Internet.
    - Anonymous information from the analyses will be put in a completely public database, available to anyone on the Internet.
    - Your coded medical information and information from more detailed analyses of your coded samples will be put in a controlled-access database. The information in this database will be available only to researchers who have received approval from an NIH Data Access Committee.

Please note that traditionally-used identifying information about you, such as your name, address, telephone number, or social security number, will NOT be put into either the public or controlled-access databases for this project.

Recontact
- In the future, we may want to obtain additional samples or follow-up information about your health or medical care. Should this be needed, a person from [Insert Institution Name] will contact you to ask whether you would be interested in participating in this additional research.

3. Financial Compensation/Costs

You will not be paid to participate in this project. Your blood (or other tissue) samples and your medical information will be used only for research purposes and will not be sold. It is possible that some of the research conducted using your samples or information eventually will lead to the development of new diagnostic tests, new drugs or other commercial products. Should this occur, there is no plan to provide you with any part of the profits generated from such products.

You will not incur any expenses from participating in this project. The chance that you will be physically injured as a result of participating in this project is very small. However, if you are

physically injured as a result of participating in this project, emergency medical treatment for your research-related injury will be provided to you at no cost.

4. Potential Benefits of Participating in the Project

You should not expect to personally benefit from this research. The main reason you may want to participate is to help researchers and health professionals around the world to better understand the causes of disease so that they can find better ways to prevent, detect, treat, and cure illnesses. You may feel good knowing that you may be helping future patients.

5. Potential Risks of Participating in the Project

Physical Risks

- If a blood sample is not taken from you, there are no physical risks associated with this project.
- If a blood sample is taken from you, there are very few physical risks. Possible side effects from drawing the blood sample include mild pain, bleeding, bruising, and infection at the site of the needle insertion. Fainting or light-headedness can sometimes occur, but usually last only a few minutes.

Psychological or Social Risks Associated with Loss of Privacy

- Your privacy is very important to us and we will use many safety measures to protect your privacy. However, in spite of all of the safety measure that we will use, we cannot guarantee that your identity will never become known. Although your genetic information is unique to you, you do share some genetic information with your children, parents, brothers, sisters, and other blood relatives. Consequently, it may be possible that genetic information from them could be used to help identify you. Similarly, it may be possible that genetic information from you could be used to help identify them.
- While neither the public nor the controlled-access databases developed for this project will contain information that is traditionally used to identify you, such as your name, address, telephone number, or social security number, people may develop ways in the future that would allow someone to link your genetic or medical information in our databases back to you. For example, someone could compare information in our databases with information from you (or a blood relative) in another database and be able to identify you (or your blood relative). It also is possible that there could be violations to the security of the computer systems used to store the codes linking your genetic and medical information to you.
- Since some genetic variations can help to predict the future health problems of you and your relatives, this information might be of interest to employers, health providers, insurance companies, and others. Patterns of genetic variation also can be used by law enforcement agencies to identify a person or his/her blood relatives. Therefore, your genetic information potentially could be used in ways that could cause you or your family distress, such as by revealing that you (or a blood relative) carry a genetic disease or by leading to the denial of employment or insurance for you (or a relative).
- There also may be other privacy risks that we have not foreseen.

While we believe that the risks to you and your family are very low, we are unable to tell you exactly what all of the risks are. There are some state laws that protect against genetic discrimination by employers or insurance companies, but there is no federal law yet that prohibits such discrimination. We believe that the benefits of learning more about diseases outweigh these potential risks.

6. Confidentiality

We will make every attempt to protect your confidentiality and to make sure that your personal identity does not become known. This signed consent form will be stored in a locked file that will be accessible only to a very small number of authorized people involved in this project. We will carefully follow the coding, storage, and release plan explained in the *Description of the Research* section on pages [insert page #s] of this document.

Note to Investigators: If you have obtained a Certificate of Confidentiality, you should include the following:

To help us protect your privacy, we have obtained a Certificate of Confidentiality from the National Institutes of Health. With this Certificate, we cannot be forced to disclose information that may identify you, even by a court subpoena, in any federal, state, or local civil, criminal, administrative, legislative, or other proceedings. We will use this Certificate to resist any demands for information that would identify you, with the following exceptions:

- The Certificate cannot be used to resist a request for your information from the United States Government when the information is to be used for auditing or evaluation of federally funded projects or for information that must be disclosed to meet the requirements of the federal Food and Drug Administration (FDA).
- The Certificate does not prevent you or a member of your family from voluntarily releasing information about yourself or involvement in this research. Also, if you have given written consent to an insurer, employer, or other person to receive research information, then we may not use the Certificate to withhold the information.

7. Project Results

In general, results from this research project will not be given back to you or put into your medical records. In some situations, the results might be important to your health or medical care. If this occurs, we will contact you to see if you want to learn more. If research from this project is published in professional journals, there will be no traditionally-used identifying information, such as your name, address, telephone number, or social security number, included in the publications. Some publications from this project will be found at [insert website URL address].

8. Alternatives to Participating in the Project

The alternative option is not to participate.

9. Voluntary Participation

The choice to participate in this research by donating your tissues and medical information is completely up to you. **No matter what you decide to do, your decision will not affect your medical care. Refusal to participate will involve no other penalties or loss of benefits to which you are entitled**

10. Withdrawal from the Project

Once data are generated from the samples you provided, and those data are placed in the database as described elsewhere in this consent, you will not be able to withdraw the data, only the

samples. If you would like to withdraw from this project you can contact (insert name & contact information of Principal Investigator) at (insert name of institution) and he/she will destroy any remaining tissue samples of yours that have been obtained for the study. In addition, it may be possible for him/her to destroy the link between you and your genetic and medical information. However, the samples and data that have already been distributed to other research centers or placed in the research databases will not be able to be withdrawn.

If you chose to discontinue your participation in the study, there will be no penalty or loss of benefits to which you are entitled.

11. Contact Information

If you have any questions about the project, about your rights as a research participant, or about any research-related injury, please contact [insert specific institutional language here].

Agreeing to Participate in the Project

To participate in this research, you must agree to ALL of the following statements:

- I voluntarily agree to donate a blood (or other tissue) sample or a cheek tissue sample to be used for this and for other research projects.
- I agree to release information from my medical records for this and for other research projects.
- I agree to have my coded genetic information and coded medical information placed in databases accessible by the Internet, as described in the *Storage and Release of Samples and Medical Information* section on page 2 of this document.
- I understand that my coded genetic information and coded medical information in the Internet databases will be used in this and in other research projects.
- I understand that there is a risk that someone in the future might be able to use information in these databases to identify me or possibly my blood relative(s).
- I agree to be recontacted in the future to see if I am willing to provide additional samples or follow-up information about my health or medical care. (Only required if the study design involves re-contact.)

Please sign your name here if you agree with the above six statements.

Your signature: _____

Date: _____

Signature of Doctor/Nurse/Other Witness_____