

Sample Consent Document 4: *Genetics of Obesity, Diabetes, and Heart Disease in African Diaspora Populations*

This consent document was used for the recruitment of participants for whole exome sequencing.

Important note: This consent document was developed for the Genetics of Obesity, Diabetes, and Heart Disease in African Diaspora Populations study. It is not provided as guidance or as a template promoted by NHGRI, but as a reference to inform investigators and IRBs considering these issues. It is important to tailor consent documents for each individual study.

MEDICAL RECORD

CONSENT TO PARTICIPATE IN A CLINICAL RESEARCH STUDY

- Adult Patient or
- Parent, for Minor Patient

INSTITUTE:

STUDY NUMBER:

PRINCIPAL INVESTIGATOR:

STUDY TITLE:

Genetics of Obesity, Diabetes, and Heart Disease in African Diaspora Populations

Continuing Review Approved by the IRB on 08/19/13

Amendment Approved by the IRB on 04/08/11 (A)

Date Posted to Web: 09/07/13

Standard

INTRODUCTION

We invite you to take part in a research study at the National Institutes of Health.

First, we want you to know that:

Taking part in research is entirely voluntary.

You may choose not to take part, or you may withdraw from the study at any time. In either case, you will not lose any benefits to which you are otherwise entitled. However, to receive care at the NIH, you must be taking part in a study or be under evaluation for study participation.

You may receive no benefit from taking part. The research may give us knowledge that may help people in the future.

Second, some people have personal, religious or ethical beliefs that may limit the kinds of medical or research treatments they would want to receive (such as blood transfusions). If you have such beliefs, please discuss them with your doctors or research team before you agree to the study.

Now we will describe this research study. Before you decide to take part, please take as much time as you need to ask any questions and discuss this study with anyone you wish or with family, friends or your personal physician or other health professional.

WHY IS THIS STUDY BEING DONE?

You are being asked to participate in a research study that is looking for inherited factors (or genes) which cause obesity, adult-onset diabetes, heart disease, and other common health conditions. Researchers from the National Institutes of Health will work together to look for genes that increase the risk of getting these and other common conditions in people of African ancestry. To achieve this goal, we are seeking to study both black and white adults with and without these conditions. Your DNA and other clinical information obtained from your blood sample will be used to study the genetic basis of obesity, diabetes, and heart disease and related complications including hypertension, kidney, eye, and nerve damage. To achieve this objective, it will be necessary to study your entire genetic inheritance (i.e., your genome). Your DNA sample and information about your ethnic origin may also be used to study human population history. Also, your clinical information and biological samples, including DNA, will be stored and used for future research on the genetic basis of human diseases.

PATIENT IDENTIFICATION

CONSENT TO PARTICIPATE IN A CLINICAL RESEARCH STUDY

- Adult Patient or
- Parent, for Minor Patient

File in Section 4: Protocol Consent (1)

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WHAT IS INVOLVED IN THE STUDY?

Because your participation in this study will only involve the time to administer the informed consent and draw blood, we anticipate it will take approximately 30 minutes. Twenty ml (just over a tablespoon) of blood will be taken during your clinical visit for this study. We will not be doing a separate interview for this study; instead, we will be using your responses to the questions that you have already answered for the main study (for instance, about your medical history and lifestyle). Your sample and information will be used for genetic analysis, as described below.

Your DNA will be analyzed to look for genetic changes that might contribute to the cause of diabetes and other human diseases. We will start by looking for variants in genes that are expected to influence the levels of lipids (e.g., cholesterol and triglycerides) in your blood. For a subset of participants, the DNA samples will be used for additional testing, although we will not report to you whether or not your DNA was selected for this additional testing (unless it is necessary for us to contact you regarding the results from this testing [described in detail below]). These DNA samples will undergo "whole exome sequencing." Every cell in your body contains the genetic code of your DNA. Your genome consists of genes, short segments of DNA that code for proteins, which are the central actors in most of the biological processes in your body, and many long segments of DNA that do not contain genes. The places on the genome that contain genes are called exons. Scientists use the term "exome" to represent all of the exons, or protein-coding regions, of the genome. Whole exome sequencing looks at the entire genome, but only records the gene regions of the genome. All people have very similar genomes – about 99.6 percent identical. However, everyone is unique and between any two people there could be approximately 24 million places where the "spelling" of the code is different. Associating these differences in spelling (gene variants) with differences in lipid levels may help us to understand how these variants are related to disease and will allow us to focus on areas of your genome that are good candidates for more investigation.

Exome sequencing is a research test that can provide various types of results. For example:

- We can find gene variants that are known to cause or contribute to disease;
- We can find gene variants that are known *not* to cause or contribute to disease, meaning they are normal variations of the genome;
- We can find gene variants that are novel and of uncertain clinical importance, meaning that we do not know if they cause/contribute to disease or if they are normal variations of the genome.

As the majority of the gene variants that we find in the course of our research will fall into the third category, of uncertain clinical importance, we will not routinely return your results to you or other participants. In the unlikely event, however, that we find a gene variant that is known to cause or contribute to disease and which can be confirmed by repeating the test in a clinical laboratory, we will attempt to contact you with these results. We will *only* give you results about specific abnormal gene variants that we think are important to your health, and that have been confirmed in a clinical laboratory. In this case, we will contact you by mail or phone to find out if you are interested in learning about your results. You will be given a choice to learn or not to learn the results of your exome sequencing. You may "opt out" of learning any of your results, and still be in the study. If you choose to learn the results of your exome sequencing, you will be asked to return to the study site and will meet with a genetic counselor who will explain the findings and what they mean to your health. She or he may also make recommendations for follow-up with your physician or with a specialist. For example, if you are found to have a gene variant that causes high cholesterol, you may be recommended to discuss this with your doctor so that he/she can monitor your cholesterol closely.

It is important that you know the limits of the test we are doing:

- For some participants we will only be testing some, but not all of your genes.
- Not all gene variants that could cause disease will be detected.

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- Some gene variants that are not currently known to cause disease may be found to do so in the future. While we will attempt to contact you regarding gene variants found to have urgent clinical significance, we cannot guarantee that all such future findings will be reported. For instance, if we do not have updated contact information, we will not be able to contact you. Also, the study will not continue indefinitely, and new risks will be understood after we have stopped monitoring the results.
- Gene variants that are known not to cause disease or are of uncertain relevance (normal variation) will not be reported to you.
- It is also important to know that because you carry a variant that has been associated with a disease does not necessarily mean that you are going to develop the diseases.
- We will contact you about gene variants that are important to your health and health decisions; however, we expect to find very few of this type of gene variant.
- Even if you are not contacted by us, you could still have gene variants that could cause disease: not all gene variants that could cause disease are known and not all gene variants that could cause will be detected.

You will be able to talk with one of the researchers in the study who will answer any question you have about this study.

WHAT ARE THE RISKS OF THE STUDY?

You may feel some minor pain when the blood sample is collected. The risk involve will not be any different from what you experience during any normal blood test. The amount of blood taken is not enough to cause you any problem with anemia.

To do more powerful research, it is helpful for researchers to share information they get from studying human samples. They do this by putting it into one or more scientific databases, where it is stored along with information from other studies. Researchers can then study the combined information to learn even more about health and disease. If you agree to take part in this study, some of your genetic and health information will be placed into a scientific database called "dbGaP" that is maintained by the National Institutes of Health. A researcher who wants to study the information must apply and be approved to use the database. Researchers with an approved study may be able to see and use your information (along with that of many other people), but your name and other information that could directly identify you (such as address or social security number) will never be placed into a scientific database. As your genetic information is unique to you, however, there is a small chance that someone could trace it back to you. The risk of this happening is very small, but may grow in the future. Researchers will always have a duty to protect your privacy and to keep your information confidential.

You may be concerned that someone could get access to your genetic information and that it could be misused; for example, if your information suggested something serious about your health, it could be used to make it harder for you to get or keep a job or insurance. There are laws in place that make it illegal for an employer or health insurance company to discriminate against an individual based on their genetic information. Further, your privacy and the confidentiality of your data are very important to us and we will make every effort to protect them.

Also, by having genetic information about people from your ethnic group, researchers may make statements about your group identity that you or members of your community may disagree with.

ARE THERE BENEFITS TO TAKING PART IN THE STUDY?

If your sample is selected for whole exome sequencing, we may find gene variant(s) that are important to your health and/or the health of your relatives. In that case, you and your family may benefit from knowing that information. It is more likely, however, that you will receive no genetic information from us. As described above, a lack of contact from us does not mean that your genome does not contain gene variants that may be important to your health and/or the health of your relatives; the types of analysis that we are doing cannot determine that information.

MEDICAL RECORD**CONTINUATION SHEET for either:**

NIH 2514-1, Consent to Participate in A Clinical Research Study

NIH 2514-2, Minor Patient's Assent to Participate In A Clinical Research Study

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Although you may receive no direct benefit, your participation in this study may help scientists understand the genetic basis of obesity, adult-onset diabetes, and heart disease in individuals of African descent and may ultimately lead to better ways to prevent and treat these diseases.

WHAT ARE MY OTHER OPTIONS?

Participation in this study is voluntary. The decision to participate is up to you. You do not have to participate if you do not want to. You may withdraw from the study at any time. The care you receive now or in the future from the doctors and nurses participating in this study will not be affected, whether you participate in or withdraw from this study. You may also obtain your lipid profile and its interpretation by having a check up for heart disease with your doctor.

WHAT IF I CHANGE MY MIND?

You may stop participating in this study at any time. If you decide to withdraw from the study, your biological samples will be destroyed and your identifying information will be purged from all databases, so that your samples and data will not be included in any future analyses. However, there are limitations in our ability to exclude your information after it has already been used or shared. For example, it will be difficult or impossible to destroy your information when the data has been deposited into a scientific database or after the analysis of your genetic information has been published in a research journal.

WHO ELSE WILL KNOW THAT I AM IN THIS STUDY?

All information gathered in this study will be kept confidential. When results of this study are reported in medical journals or at medical meetings, you will not be identified. All medical records will be stored by the researchers in safe cabinets. All collected clinical information will be password protected.

Likewise, your blood samples are being collected to facilitate the study of obesity, diabetes, heart disease, and related complications. Other researchers collaborating with _____ may be interested in using your genetic material to conduct more research on diabetes and other human diseases.

Clinical information from this study will be identified with a code number instead of your name. The link connecting your personal information and your study code number will be kept in a locked file cabinet at the Center for Research on Genomics and Global Health at the National Institutes of Health. The study investigators (_____) and the person managing the clinical information (_____) will be the only people with access to that link.

Your samples and information collected about you in the main study (for instance, about your medical history and lifestyle) will be shared with co-investigators and other Institutional Review Board (IRB) approved investigators of biomedical research.

WILL I RECEIVE PAYMENT FOR BEING IN THIS STUDY?

There will be a small payment of \$75 for participating in this study.

Your DNA sample will be used only for research and will not be sold or used directly for the production of commercial products.

CONFLICTS OF INTEREST

There are no conflicts of interest with study investigators at the National Institutes of Health. The National Institutes of Health reviews NIH staff researchers at least yearly for conflicts of interest.

PATIENT IDENTIFICATION**CONTINUATION SHEET for either:**

NIH-2514-1 (10-84)

NIH-2514-2 (10-84)

P.A.: 09-25-0099

STUDY NUMBER:

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OTHER PERTINENT INFORMATION

1. Confidentiality. When results of an NIH research study are reported in medical journals or at scientific meetings, the people who take part are not named and identified. In most cases, the NIH will not release any information about your research involvement without your written permission. However, if you sign a release of information form, for example, for an insurance company, the NIH will give the insurance company information from your medical record. This information might affect (either favorably or unfavorably) the willingness of the insurance company to sell you insurance.

The Federal Privacy Act protects the confidentiality of your NIH medical records. However, you should know that the Act allows release of some information from your medical record without your permission, for example, if it is required by the Food and Drug Administration (FDA), members of Congress, law enforcement officials, or authorized hospital accreditation organizations.

2. Policy Regarding Research-Related Injuries. The _____ will provide short-term medical care for any injury resulting from your participation in research here. In general, no long-term medical care or financial compensation for research-related injuries will be provided by the _____, or the Federal Government. However, you have the right to pursue legal remedy if you believe that your injury justifies such action.

3. Payments. The amount of payment to research volunteers is guided by the National Institutes of Health policies. In general, patients are not paid for taking part in research studies at the National Institutes of Health. Reimbursement of travel and subsistence will be offered consistent with NIH guidelines.

4. Problems or Questions. If you have any problems or questions about this study, or about your rights as a research participant, or about any research-related injury, contact the Principal Investigator _____

or at _____

5. Consent Document. Please keep a copy of this document in case you want to read it again.

COMPLETE APPROPRIATE ITEM(S) BELOW:

A. Adult Patient's Consent

I have read the explanation about this study and have been given the opportunity to discuss it and to ask questions. I hereby consent to take part in this study.

Signature of Adult Patient/Legal Representative Date

Print Name

B. Parent's Permission for Minor Patient.

I have read the explanation about this study and have been given the opportunity to discuss it and to ask questions. I hereby give permission for my child to take part in this study.
(Attach NIH 2514-2, Minor's Assent, if applicable.)

Signature of Parent(s)/Guardian Date

Print Name

C. Child's Verbal Assent (If Applicable)

The information in the above consent was described to my child and my child agrees to participate in the study.

Signature of Parent(s)/Guardian Date _____
Print Name

**THIS CONSENT DOCUMENT HAS BEEN APPROVED FOR USE
FROM AUGUST 19, 2013 THROUGH AUGUST 18, 2014.**

Signature of Investigator Date _____
Signature of Witness Date

Print Name

Print Name

PATIENT IDENTIFICATION

CONSENT TO PARTICIPATE IN A CLINICAL RESEARCH STUDY (Continuation Sheet)

• Adult Patient or • Parent, for Minor Patient

NIH-2514-1 (07-09)

P.A.: 09-25-0099

File in Section 4: Protocol Consent