The UNIVERSITY OF CHICAGO
The Division of the Biological Sciences • The University of Chicago Medical Center

CONSENT/AUTHORIZATION BY SUBJECT FOR PARTICIPATION IN A RESEARCH PROTOCOL

COLLECTION OF A SALIVA SAMPLE FOR GENETIC STUDIES

Protocol Number: 6858 Name of Subject: _____

Date of Birth / Medical History Number: _____

STUDY TITLE: Genetic Studies of Diabetes Mellitus

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You are being asked to participate in a research study. This consent form describes the study procedures, the risks and benefits of participation, as well as how your confidentiality will be maintained. Please take your time to ask questions and feel comfortable making a decision whether or not to participate. This process is called informed consent. If you decide to participate in this study, you will be asked to sign this form. Throughout this consent form, "you" refers to you or your child, as appropriate.

WHY IS THIS STUDY BEING DONE?
Diabetes is a disease that is characterized by high levels of sugar in the blood and urine. The research study in which you are being asked to participate is aimed at identifying the genes that contribute to the development of diabetes. This study will be of no direct benefit to you. However, it could provide information about the genetic causes of diabetes that may lead to better diagnosis and treatment.

HOW MANY PEOPLE WILL TAKE PART IN THE STUDY?
Up to 5000 people are expected to take part in this study at the University of Chicago.

WHAT IS INVOLVED IN THE STUDY?
You will be asked to send two small tubes of saliva (about two teaspoons) to the University of Chicago using a special container. We may also send you a piece of filter paper (similar to the paper used in newborn screening in most states) upon which we will ask you to put four (4) drops of blood. The blood spots would be collected just like when you check your blood sugar. These may be used to do tests related to Type 1 Diabetes to help clarify the type of diabetes in your family.

You will also be asked to complete a questionnaire about your medical history and diagnosis. This will take approximately 20 minutes to complete.

DNA, the genetic material in your saliva, will be prepared for analysis of genes that might increase your risk for diabetes or other conditions that you may have, such as high blood pressure. As part of this study, your DNA sample will be stored for an indefinite length of time. Your sample will be used to search for genetic alterations associated with diabetes. This includes genetic testing for mutations in genes known to cause diabetes and other conditions. The doctors directing this study will receive the results of these tests and use this information for the purposes of research only.

Because we are a research laboratory and not a clinical laboratory with certified procedures for reporting results, we cannot directly release results from this study to you. If we obtain information that we think might be significant to you or your family (e.g., identification of a mutation that has caused the disease, disorder or condition we are studying), we may suggest to your physician that these results be confirmed by a CLIA-certified laboratory. A CLIA-certified laboratory is a laboratory that is authorized to release results from patient tests for clinical and diagnostic purposes. Most CLIA laboratories will ask for a fresh blood sample in order to ensure the accuracy of the results. Please indicate below if you wish us to inform your physician if we believe confirmatory testing should be performed.

☐ Please contact my health care provider if results become available in the future:

Physician's Name: _____

Phone: _____

Address: _____

Your DNA sample will be given a number. Information about your name and this number will be kept in a locked drawer in a locked laboratory. Only Dr. Graeme Bell and his research staff will have access to this information. Although reasonable efforts will be made to keep your information confidential, there always exists a very slight risk that your information will be disclosed. Your coded DNA sample(s) will be stored indefinitely in Dr. Bell's laboratory. If you wish to have your sample(s) destroyed at any time, you may do so by calling Dr. Bell at (773) 702-9116.

Retrieval of Dried Blood Spot Card Samples from Newborn Screening Laboratories

When you were born, drops of your blood were collected on a filter paper card and used by newborn screening programs to test for rare conditions that can go unnoticed but eventually cause illness in babies. In some instances, after newborn screening is completed, these dried blood spot samples (DBS) may still be stored in a repository in the state or country where you were born.

There are several reasons why dried blood spots are kept. First, good laboratory practices require that samples (such as dried blood spots) be kept for a period of time after testing is done, in case a test needs to be checked or repeated. Additionally, because dried blood spots contain information that may be useful for studying birth defects and diseases, some states may also allow leftover dried blood spots to be used for medical research.

If any of your blood spot samples are still available, we may request that they be sent to us for research testing, such as measuring the blood sugar level in your blood soon after you were born. Your DBS samples will help researchers learn about early-onset diabetes and potentially gather support for the inclusion of early-onset diabetes into newborn screening programs.

During the course of our study on early-onset diabetes, your DBS sample will be stored in temperature-controlled laboratory settings to which only authorized personnel will have access. Your de-identified blood spot samples will be shared with laboratories that are necessary to process the samples, including the Newborn Screening Laboratory at the Wisconsin State Laboratory of Hygiene (WSLH). However, they will not be shared with individuals or entities outside the scope of our study on neonatal diabetes. Additionally, any unused portions of your blood spot samples will not be retained for any studies outside of our study on early-onset diabetes.

You will be asked to sign a separate medical release so that study staff can obtain access to your blood spot card. All the information provided will be kept strictly confidential and available only to persons to whom it is necessary to perform the job. Specifically, your demographic information, including name, birth date, and hospital of birth, will be shared with the repository in order to locate and obtain the DBS samples.

During this study, Dr. Bell or his research team may collect information about you for the purposes of this research, including your name, age, date of birth, height, weight and the results of tests performed as part of this study. Your medical history may also be collected from your medical records.

Other Studies in Which You May be Participating or May Wish to Participate:
Please indicate whether or not you give your permission to be contacted in the future by other researchers who wish to do studies about diabetes. By agreeing below, you are under no obligation to participate in any other study, only to be contacted and given the opportunity to do so:

☐ Yes, you may contact me in the future about other studies related to diabetes

Initials: _____ Date: _____

☐ No, you may not contact me in the future about other studies related to diabetes

Initials: _____ Date: _____

HOW LONG WILL I BE IN THE STUDY?
You will be involved in a long-term study of the genetics of diabetes. The preparation of the saliva sample and collection of information about you will take about one hour. However, the identification of the genes that contribute to the development of diabetes could take many years.

WHAT ARE THE RISKS OF THE STUDY?
There is no risk associated with providing a saliva sample. There is no additional risk associated with collecting drops of blood in the same way as checking blood sugar.

Any time information is collected about you there is a potential risk for loss of confidentiality. However, the researchers will make every effort to keep your information confidential. Please refer to the 'What About Confidentiality' section for information on what measures will be taken to protect your confidentiality.

ARE THERE ANY BENEFITS TO TAKING PART IN THE STUDY?
If you agree to take part in this study, there may not be direct medical benefit to you. However, we hope the information learned from this study will benefit other individuals with diabetes in the future.

WHAT OTHER OPTIONS ARE THERE?
You may choose not to participate in this study. Your participation is entirely voluntary. The decision whether or not you wish to participate in this study will not affect your care at the University of Chicago Medical Center.

WHAT ARE THE COSTS?
Your participation in this study will not result in any cost to you or your insurance company. However, you or your insurance company will be responsible for costs related to your usual medical care.

WILL I BE PAID FOR MY PARTICIPATION?

You will not receive payment for participating in this study.

WHAT ABOUT CONFIDENTIALITY?

Study records that identify you will be kept confidential. The data collected in this study will be used solely for the purposes described herein. By signing this form, you are allowing the research team access to your medical records, which include Protected Health Information (PHI). PHI consists of any health information that is collected about you, which could include your medical history and new information collected as a result of this study. The research team includes the individuals listed on this consent form and other personnel involved in this study at the University of Chicago. This consent form document will be kept by the research team for at least six years.

On occasion, certain specialized tests might be required to clarify whether a certain genetic problem was the cause of diabetes or any related problems in your family. If this requires that any test be done in another laboratory not at the University of Chicago, we will not share any information that would identify you, and will instead give a part of your sample that will be coded only with a unique number but not your name. Such laboratories might include: a commercial laboratory; the laboratory of Deborah Mackay at the University of Southampton, UK (a specialist in transient neonatal diabetes); the laboratories of Craig Hanis or Eric Boerwinkle at the University of Texas, Houston; the lab of Jay Heinecke at University of Washington, Seattle; Jose Florez, MD, PhD and Miriam Udler, MD, PhD of the BROAD Institute; Mark Anderson, MD, PhD and Mike German, MD of University of California, San Francisco; Jorge Ferrer of Imperial College London; Joslin Diabetes Center at Harvard University; Jagiellonian University Medical College in Krakow, Poland. If you have consented to participate in the RADIANT (Rare and Atypical Diabetes Network) study, all of your research records from this study can be shared with the RADIANT researchers in an identified manner, including your name, date of birth, dates of tests and results of all tests performed as part of this research and any records collected as part of this research.

Your records may be reviewed by federal agencies whose responsibility is to protect human subjects in research, including the Food and Drug Administration (FDA) and Office of Human Research Protections (OHRP). In addition, representatives of the University of Chicago, including the Institutional Review Board, a committee that oversees the research at the University of Chicago, may also view the records of the research. If your research record is reviewed by any of these groups, they may also need to review your entire medical record.

If this study is audited by Federal Investigators, they are not required to follow the same laws regarding Protected Health Information as the University of Chicago.

The results from tests and/or procedures performed as part of this study may become part of your medical record.

During your participation in this study, you will have access to your medical record. Dr. Bell is not required to release to you research information that is not part of your medical record.

The study results will be kept in your research record and be used by the research team indefinitely. When the study is terminated, either the research information not already in your medical record will be destroyed or information identifying you will be removed from the study results. Any research information in your medical record will be kept indefinitely.

Data from this study may be used in medical publications or presentations. Your name and other identifying information will be removed before this data is used. If we wish to use identifying information in publications, we will ask for your approval at that time.

The Genetic Information Nondiscrimination Act (GINA)

The Genetic Information Nondiscrimination Act (GINA) is a federal law that may help protect you from health insurance or employment discrimination based on genetic information. GINA is a federal law that will protect you in the following ways:

- Health insurance companies and group plans may not request genetic information from this research;
- Health insurance companies and group plans may not use your genetic information when making decisions regarding your eligibility or premiums;
- Employers with 15 or more employees may not use your genetic information when making a decision to hire, promote, or fire you or when setting the terms of your employment.

GINA does not protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance. GINA also does not protect you against discrimination based on an already-diagnosed genetic condition or disease.

NIH and Other Databases

If you agree to take part in this study, your genetic and health information and as applicable a portion of your specimens will be placed into one or more scientific databases. In particular, the National Institutes of Health maintains a database called "dbGaP." The NIH database is a restricted database, meaning a researcher who wants to study information from dbGaP must work with the group overseeing the database to obtain the information. Security measures are in place to protect these data.

Researchers with an approved study will be able to see and use some of your information, but your name and other information that could directly identify you (such as your name or address) will not be placed into the database. There is a risk that someone could use your unique genetic information to trace data back to you or your family, but this risk is very small. There is no direct benefit to you that is expected from any secondary research that may be conducted.

If you decide to withdraw from the study as outlined in the following section, your data will be withdrawn from these databases. However, if your data have already been submitted to an NIH database and distributed to other researchers, or your data have been de-identified and can no longer be linked back to you, your data will not be able to be withdrawn.

Certificate of Confidentiality

To help us protect your privacy, the National Institutes of Health (NIH) has issued a Certificate of Confidentiality for this research. With this Certificate, the researchers 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. The researchers will use the Certificate to resist any demands for information that would identify you, except as explained below. The Certificate cannot be used to resist a demand for information from personnel of the United States Government that is used for auditing or evaluation of federally funded projects or for information that must be disclosed in order to meet the requirements of the federal Food and Drug Administration (FDA).

A Certificate of Confidentiality does not prevent you or a member of your family from voluntarily releasing information about yourself or your involvement in this research. If an insurer, employer, or other person obtains your written consent to receive research information, then the researchers may not

use the Certificate to withhold that information. The Certificate of Confidentiality does not prevent the researchers from disclosing voluntarily, without your consent, information that would identify you as a participant in the research project, and we will do so if we have evidence of suspected child abuse, elder abuse or harm to self or others.

WHAT ARE MY RIGHTS AS A PARTICIPANT?
Taking part in this study is voluntary. If you choose not to participate in this study, your care at the University of Chicago/University of Chicago Medical Center will not be affected.

You may choose not to participate at any time during the study. Leaving the study will not affect your care at the University of Chicago/University of Chicago Medical Center.

If you choose to no longer be in the study and you do not want any of your future health information to be used, you must inform Dr. Bell in writing at the address on the first page. Dr. Bell may still use your information that was collected prior to your written notice.

This consent form document does not have an expiration date.

You will be given a signed copy of this document.

WHO DO I CALL IF I HAVE QUESTIONS OR PROBLEMS?
You have talked to _____ about this study and you have had the opportunity to ask questions concerning any and all aspects of the research. If you have further questions about the study, you may call Dr. Bell at (773) 702-9116, our main number at 773-702-0829, or email us at: monogenicdiabetes@uchicago.edu.

If you have any questions concerning your rights in this research study you may contact the Institutional Review Board, which is concerned with the protection of subjects in research projects. You may reach the Committee office between 8:30 am and 5:00 pm, Monday through Friday, by calling (773) 702-6505 or by writing: University of Chicago, Institutional Review Board, 5841 S. Maryland Ave., MC7132, I-625, Chicago, Illinois 60637.

CONSENT

SUBJECT (18 years old or older)
The research project and the procedures associated with it have been explained to me. The experimental procedures have been identified and no guarantee has been given about the possible results. I will receive a signed copy of this consent form for my records.

I agree to participate in this study. My participation is voluntary and I do not have to sign this form if I do not want to be part of this research study.

Signature of Subject: _____

Date: _____ Time: _____AM/PM
(Circle)

PERSON OBTAINING CONSENT/ASSENT

I have explained to _____ the nature and purpose of the study and the risks involved. I have answered and will answer all questions to the best of my ability. I will give a signed copy of the consent form to the subject or the family.

Signature of Person Obtaining Consent: _____

Date: _____ Time: _____AM/PM
(Circle)

PARENT/GUARDIAN/ OR LEGALLY AUTHORIZED REPRESENTATIVE:
I give my permission for my child/relative to participate in the above described research project.

Signature of Parent/Guardian/Legally Authorized Representative: _____

Date: _____ Time: _____AM/PM
(Circle)

ASSENT OF MINOR

SUBJECT (12-17 years old)

The research project and the procedures have been explained to me. I will receive a signed copy of this assent form for my records.

I agree to participate in this study. My participation is voluntary and I do not have to sign this form if I do not want to be part of this research study.

Signature of Subject: _____

Date: _____ Time: _____AM/PM
(Circle)