



Approved February 16, 2010

Site of Research:
Johns Hopkins Medical Institutions

Patient I.D. Plate

RESEARCH PARTICIPANT INFORMED CONSENT AND PRIVACY AUTHORIZATION FORM

Protocol Title: Genetic Modifiers of CF

Application No. : NA_00035659

Sponsor: National Institutes of Health

Principal Investigator: Garry Cutting, M.D.

1. What you should know about this study:

- You are being asked to join a research study.
- This consent form explains the research study and your part in the study.
- Please read it carefully and take as much time as you need.
- Please ask questions at any time about anything you do not understand.
- You are a volunteer. If you join the study, you can change your mind later. You can decide not to take part or you can quit at any time. There will be no penalty or loss of benefits if you decide to quit the study.
- During the study, we will tell you if we learn any new information that might affect whether you wish to continue to be in the study.
- Ask your study doctor or the study team to explain any words or information in this informed consent that you do not understand.
- If children and adults can join this study, the word “you” in this consent form will refer to both you and your child.
- During this study, you will not have access to certain medical information and test results collected for study purposes. If an emergency occurs while you are in the study, medical information needed for your treatment can be made available to your study physician and other physicians who treat you. When the study is completed, all the information in your medical record will be available to you.

2. Why is this research being done?

This research is being done to find the genes and other factors that are responsible for variation among persons with cystic fibrosis. You and a sibling (or two or more of your children) have cystic fibrosis (CF), which involves lung disease, problems with digestion, abnormal perspiration, and a shortened life span. The underlying cause of CF is changes in a gene called CFTR, but even individuals with the same change in the same family may have differences in the severity of lung, sinus, liver, or intestinal

Approved February 16, 2010

problems. This study is attempting to understand what other genes and other factors may be responsible for the variation or similarity of illness between you and your sibling (your children). Families with 2 or more siblings with CF may join. We anticipate that about 1000 families (about 4000 people) will join this study.

3. What will happen if you join this study?

If you agree to be in this study, we will ask you to do the following things:

Give investigators from Johns Hopkins permission to review the medical records of the siblings with CF as well as what was submitted to the Cystic Fibrosis Foundation. To better describe CF, we may need to review medical records and CF Registry information of the siblings in the future. Your family will be asked to complete a family medical history questionnaire and each CF patient to fill out a personal questionnaire. Researchers from Johns Hopkins may call you to get this information. If they are not able to reach you by phone after two or three attempts, they may mail you a self addressed envelope with the questionnaires enclosed. On all materials collected by this study, your name and any identifying information will be removed and replaced by a Johns Hopkins study ID number. The relevant information from your medical records and the questionnaires will be entered into a database used only for this study. Only the study ID numbers and your birthdate are present in this database. The link between the ID numbers and your identification is kept in a locked cabinet and separate electronic database that only the principal investigator and his designated staff can access.

Blood will be drawn from each of the immediate family members (parents and affected offspring). This blood will be used to extract DNA and to create white blood cell lines as a permanent source of your DNA. Some serum and plasma will be stored for later study, as additional knowledge about possible modifiers becomes available. Any tests done in the future with your banked blood or your DNA will only be to answer questions about modifier genes for CF. We will not ask your permission to do any additional tests to answer this question and the results will not be available on an individual basis since the tests are being done for research alone.

We will compare your clinical symptoms with your DNA to see if additional genes that affect the severity of CF can be identified. One to two tablespoons of blood (two teaspoons in infants) will be drawn by standard techniques from a vein in the arm. Whenever possible, blood samples for this study will be obtained at the same time blood is drawn for medically indicated tests. For each affected individual, we will determine the two mutations in the cystic fibrosis gene. If you do not already have this information, we will share it with your CF center physician in writing. A medical geneticist will be available to answer any questions you may have about your mutations.

We may ask you later to participate in additional testing to better describe the severity of your cystic fibrosis. You do not have to agree to this, even if you have already participated in this part of this study. If you agree, you will sign a different consent form.

How long will you be in the study?

You will be in this study for 15 years or more, but your active participation will last for one or two clinic visits.

Approved February 16, 2010

4. What are the risks or discomforts of the study?

Blood drawing may cause some discomfort and bruising. In addition, there may be a slight risk of loss of confidentiality. Every precaution will be taken to ensure that your personal information is protected as described in section 2.

You may get tired or bored when we are asking you questions or you are completing questionnaires. You do not have to answer any question you do not want to answer. There may be side effects and discomforts that are not yet known.

5. Are there benefits to being in the study?

There is no direct benefit to participants other than increasing our understanding of CF. If you take part in this study, you may help others in the future.

6. What are your options if you do not want to be in the study?

You do not have to join this study. If you do not join, your care at Johns Hopkins will not be affected.

7. Will it cost you anything to be in this study?

No. There are no costs to you for any tests done as part of this study.

8. Will you be paid if you join this study?

No. You will not be paid or reimbursed for participating in this study.

9. Can you leave the study early?

If you wish to withdraw from the study, you may do so by notifying us in writing: Garry R. Cutting, MD, 733 N. Broadway, BRB Suite 551, Baltimore, MD 21287-3914. We will destroy your DNA sample and cell line and no further testing will be performed.

10. How will your privacy be protected?

Johns Hopkins has rules to protect information about you. Federal and state laws also protect your privacy. This part of the consent form tells you what information about you may be collected in this study and who might see or use it.

Generally, only people on the research team will know that you are in the research study and will see your information. However, there are a few exceptions that are listed later in this section of the consent form.

The people working on the study will collect information about you. This includes things learned from the procedures described in this consent form. They may collect other information including your name, zip code, date of birth, and other details.

The research team will need to see your information. Sometimes other people at Johns Hopkins may see or give out your information. These include people who review the research studies, their staff, lawyers, or other Johns Hopkins staff.

People outside of Johns Hopkins may need to see your information for this study. Examples include government groups (such as the National Institutes of Health), safety monitors, other hospitals in the study and companies that sponsor the study.

Approved February 16, 2010

We cannot do this study without your permission to use and give out your information. You do not have to give us this permission. If you do not, then you may not join this study.

We will use and disclose your information only as described in this form and in our Notice of Privacy Practices; however, people outside Hopkins who receive your information may not be covered by this promise. We try to make sure that everyone who needs to see your information keeps it confidential – but we cannot guarantee this.

The use of your information has no time limit. You can cancel your permission to use and disclose your information at any time by calling the Johns Hopkins Medicine IRB at 410-955-3008 or by sending a letter to:

Office of Human Subjects Research
1620 McElderry Street
Reed Hall, Suite B130
Baltimore, MD 21205-1911

If you do cancel your permission to use and disclose your information, your part in this study will end and no further information about you will be collected. Your cancellation would not affect information already collected in this study.

11. Will the study require any of your other health care providers to share your health information with the researchers of this study?

As a part of this study, the researchers may ask to see your health care records from your other health care providers. We try to make sure that everyone who needs to see your information uses it only for the study and keeps it confidential – but we cannot guarantee this.

12. What if there is a Certificate of Confidentiality for this study?

The National Institutes of Health has given us a Certificate of Confidentiality for this study. This Certificate adds special protection for research information that identifies you and allows us, in some circumstances, to refuse to give out information that could identify you as a research subject without your consent, when such information is sought in a federal, state, or local court or public agency action. Still, we may disclose identifying information about you if, for example, you need medical help.

We may also disclose identifiable information about you as described in Section 12 of this form or in other cases. For example, the government may see your information if it audits us, and the research team will voluntarily comply with Maryland disclosure laws and will tell the local or state authorities:

- if they suspect abuse or neglect of a child or dependent adult;
- if certain diseases are present; and
- if the team learns that you plan to harm someone. In this case, the team also may warn the person who is at risk.

This Certificate does not mean the government approves or disapproves of this research project.

13. What treatment costs will be paid if you are injured in this study?

Johns Hopkins does not have a program to pay you if you are hurt or have other bad results from being in the study. However, medical care at Johns Hopkins is open to you as it is to all sick or injured people.

Approved February 16, 2010

- If you have health insurance: The costs for any treatment or hospital care you receive as the result of a study-related injury will be billed to your health insurer. Any costs that are not paid for by your health insurer will be billed to you.
- If you do not have health insurance: You will be billed for the costs of any treatment or hospital care you receive as the result of a study-related injury.

14. What other things should you know about this research study?

a. What is the Institutional Review Board (IRB) and how does it protect you?

The Johns Hopkins Medicine IRB is made up of:

- Doctors
- Nurses
- Ethicists
- Non-scientists
- and people from the local community.

The IRB reviews human research studies. It protects the rights and welfare of the people taking part in those studies. You may contact the IRB if you have questions about your rights as a participant or if you think you have not been treated fairly. The IRB office number is 410-955-3008. You may also call this number for other concerns or questions about the research.

b. What do you do if you have questions about the study?

Call the study doctor, Dr. Garry Cutting at 410-955-1773. If you cannot reach the principal investigator or wish to talk to someone else, call the IRB office at 410-955-3008.

c. What should you do if you are injured or ill as a result of being in this study?

Call Dr. Garry Cutting at 410-955-1773, if you have an urgent medical problem related to your taking part in this study.

Call Dr. Garry Cutting at 410-955-1773, if you think you are injured or ill because of this study.

d. What happens to Data, Tissue, Blood and Specimens that are collected in the study?

Scientists at Johns Hopkins work to find the causes and cures of disease. The data, tissue, blood and specimens collected from you during this study are important to both this study and to future research.

If you join this study:

- You will not own the data, tissue, blood and specimens given by you to the investigators for this research.
- Both Johns Hopkins and any sponsor of this research will study your data and the tissue, blood or other specimens collected from you.
- Scientists may only use data, tissue, blood and specimens that identify you for future research with your consent or IRB approval.
- You will not own any product or idea created by the investigators working on this study.
- You will not receive any financial benefit from the creation, use or sale of that product or idea.

e. What are the Organizations that are part of Johns Hopkins?



Approved February 16, 2010

Johns Hopkins includes the following:

- The Johns Hopkins University
- The Johns Hopkins Hospital
- Johns Hopkins Bayview Medical Center
- Howard County General Hospital
- Johns Hopkins Community Physicians.

15. Assent Statement

This research study has been explained to my child in my presence in language my child can understand. He/she has been encouraged to ask questions about the study now and at any time in the future.

Approved February 16, 2010

16. What does your signature on this consent form mean?

Your signature on this form means that:

- you understand the information given to you in this form
- you accept the provisions in the form
- you agree to join the study

You will not give up any legal rights by signing this consent form.

WE WILL GIVE YOU A COPY OF THIS SIGNED AND DATED CONSENT FORM

Signature of Participant Date

Signature of Person Obtaining Consent Date

Signature of Legally Authorized Representative (LAR) for **ADULTS NOT CAPABLE of GIVING CONSENT** (*Persons from the following categories in order of priority may be a Legally Authorized Representative: Health Care Agent; Legal Guardian; Spouse; Adult child; Parent; Adult sibling; Friend or other relative*) Date

Relationship of LAR to Participant (indicate why the LAR is authorized to act as a surrogate health care decision-maker under Maryland Law) Date

Signature of Parent/Guardian Date

Signature of Parent #2 (required if DHHS 45 CFR 46.406 or 46.407/FDA 21 CFR 50.53 or 50.54 study) Date

Signature of Child Participant (optional unless IRB required) Date

Signature of Witness to Consent Procedures (optional unless IRB or Sponsor required) Date

NOTE: A COPY OF THE SIGNED, DATED CONSENT FORM MUST BE KEPT BY THE PRINCIPAL INVESTIGATOR; A COPY MUST BE GIVEN TO THE PARTICIPANT; AND, IF APPROPRIATE A COPY OF THE CONSENT FORM MUST BE PLACED IN THE PARTICIPANT 'S MEDICAL RECORD.