

CONSENT TO PARTICIPATE IN A CLINICAL RESEARCH STUDY

STUDY TITLE: Genetic Testing of Individuals and Families with Congenital Heart Disease

PRINCIPAL INVESTIGATORS: Vidu Garg, MD and Kim McBride, MD

CONTACT TELEPHONE NUMBER: 614-355-5759 (Monday – Friday, during regular office hours).

STUDY SPONSOR: National Institutes of Health

SUBJECT'S NAME: _____ **DATE OF BIRTH:** _____

NOTE: The words “you” and “your” are used in this consent form. These words refer to the study volunteer whether a child or an adult.

1) INTRODUCTION

We invite you to be in this research study because you, your child or your relative has congenital heart disease.

Participation is voluntary. Please learn enough about this research study, its risks and benefits, to decide whether you should agree to participate. We will explain the study to you, and give you a chance to ask questions about anything you do not understand. This process is called “informed consent”. It is up to you to choose if you want to be in this study. You may refuse to be in this study or quit this study at any time, and standard medical care will still be available here or at a doctor of your choice without a penalty or loss of benefits to you.

Before agreeing to participate, it is important to read and understand the study information in this consent form. By signing the consent form, you agree to be in this study.

If this study involves a child between 9 and 18 years of age, he/she must also agree to be in the study by signing an Assent form or on the assent line of this form.

You will be given a signed and dated copy of the consent and the assent form.

2) WHY ARE WE DOING THIS RESEARCH STUDY?

Congenital heart disease is one of the most common birth defects, yet the cause is largely unknown. The study doctors, Dr. Garg and Dr. McBride, want to study the causes of congenital heart disease and how it may be passed from parent to child. They have established a repository of blood samples from patients and some families with congenital heart disease. They want to identify mutations in genes that may cause or predispose an individual to have congenital heart disease. This research is necessary because it is not known why congenital heart disease occurs.

3) WHERE WILL THE STUDY BE DONE AND HOW MANY SUBJECTS WILL TAKE PART?

This study will be done at Nationwide Children's Hospital. We hope to enroll about 2500 subjects here at this hospital.



4) WHAT WILL HAPPEN DURING THE STUDY AND HOW LONG WILL IT LAST?

- *Medical Records:* We will access your Nationwide Children's Hospital medical record to document your demographic information, any heart tests, and any genetic testing you have had. If you have had tests done at another institution, we will ask your permission to obtain these results. These records will be maintained with your other research records.
- *Blood Samples:* You will be asked to give one to two teaspoons of blood by vein for gene tests. Genetic material will be extracted from the blood sample and be used for DNA (gene) testing.
- *Results:* It is not likely that we will figure out what is causing the heart defect. If we do find a genetic cause for the heart defect because this is a research study we cannot give you your specific results. We can let you know whether or not we found a genetic change that we know or strongly think caused the heart defect. If we find this type of genetic change, we will discuss the gene, how it might be causing the heart defect and our recommendation to have this result tested in a clinical laboratory. Clinical confirmation is necessary if you wish to use this information in your medical care. This clinical test is not part of the research study and would not be paid for by the study. If you have the clinical test, you would be responsible for paying for the test and the results will become part of your medical record. It is up to you to decide whether or not to have the clinical test.
- *Other Parts of this Research Study:* You may be asked to have screening tests for cardiac disease (electrocardiogram (EKG) and/or echocardiogram (echo)), a genetic test called exome/genome sequencing, a skin biopsy, and/or give us permission to obtain a left-over tissue sample. If you are offered these procedures, they will be discussed with you and you will be given written information on them. It is up to you to decide if you want to do this and will not affect your participation in this study.
- *Relatives:* You might be asked to have family members participate in the genetic study.
- *Future Use of DNA:* Sometimes, the DNA may be stored for research at a later time, perhaps even years from now. This research includes DNA (gene) testing. This future research may be for purposes other than your medical problem. You will not be told the results of any future research. All the usual privacy protections will be applied. To further protect your confidentiality, the container, which holds your blood/DNA sample will not have any personal identifiers attached. Rather, the container will be coded with a unique study identification number. If you decide at any time that you do not want your blood or DNA stored for future research, you may contact the Study Doctor or Study Nurse.

Please initial:

I agree to allow leftover DNA to be stored for future research. I understand that they may be used for research unrelated to this study and that I will not be told the results.

I do not want leftover DNA to be stored for future research.

5) WHAT ARE THE RISKS OF BEING IN THIS STUDY?

Stress: It may be stressful knowing that researchers have personal information about you. Learning that you or a family member has an increased risk to have a heart defect might also be stressful.

Lastly, it may cause stress if you provide information about another family member's identity and health, and he/she does not want this research to include that information. If you experience stress because you participate in this research, Dr. Garg, Dr. McBride or their colleagues can help you obtain medical care to help you manage stress.

Family Relationships: If additional family members participate in the study, the DNA tests that we perform may show that relationships in your family are different than you think they are (for example, your father is not your biologic father). This kind of information will **not** be reported to you or other family members without your permission.

Early results of DNA tests: If you obtain results of DNA tests too early, you could receive unreliable information about risks to your health and the health of other family members. Such information could unnecessarily alarm you and other members of your family.

Sample of blood: You may experience discomfort, bleeding, and/or bruising. You may feel dizzy or faint. On a rare occasion, an infection could develop at the site where the blood was collected.

Unforeseen risks: There could be risks to your participation in this research which Dr. Garg or Dr. McBride does not know about now.

6) **ARE THERE BENEFITS TO TAKING PART IN THIS STUDY?**

To you:

- Usually there are no personal benefits from participation in this kind of research.
- If any results of DNA tests indicate--with reasonable certainty--that you have a definite risk of developing congenital heart disease or any other medical problem, Dr. Garg and Dr. McBride will try to get in touch with you. The study doctors along with genetic counselors will tell you what test results mean. They could make recommendations about your future plans for having children or changing habits that could affect your health.

To others: The results of this research may help other people in the future. New information may lead to identification of the causes of heart defects and improvements in medical care for congenital heart disease. However, research tests using your sample could possibly fail to produce useful information.

7) **WILL THERE BE ANY COSTS TO ME?**

Collecting a sample of blood and testing it in a research laboratory will not cost you anything.

Expenses for routine health check-ups or care for any medical problem are your responsibility. There are no funds available to pay for parking expenses, transportation to and from the research center, lost time away from work and other activities, lost wages, or child care expenses.

8) **WHAT HAPPENS IF BEING IN THIS STUDY CAUSES INJURIES?**

If you are hurt by the blood draw that is a part of the study, you should seek medical treatment for the injuries and tell the study doctor as soon as possible at the number on the first page of this form.

In most cases, this care will be billed to your health insurance company or whoever usually pays for your health care at the usual charges, but some insurance companies will not pay for care related to a

study. If the care is provided at Nationwide Children's Hospital, we make no commitment to pay for the medical care provided to you. No funds have been set aside to compensate you in the event of injury. If no one else pays for your care, you may have to pay for the cost of this care. This does not mean that you give up any of your legal rights to seek compensation for your injuries.

9) WHAT HAPPENS IF I DO NOT FINISH THIS STUDY?

It is your choice to be in this study or to stop at any time. If you decide to stop being in this study, it is okay, but you must call the Principal Investigator(s) or the study coordinator. If you stop being in the study, there will not be a penalty or loss of benefits to which you are otherwise entitled.

If you stop participation in this research, you may ask Dr. Garg and Dr. McBride to destroy any record of your participation in this research. You will not be asked for further information or samples. Your identity will be removed from all research records. Copies of DNA and/or growing cells made from your samples will also be destroyed. In addition, we will attempt to discard any resulting data from the research if it has not yet been published. Samples sent to scientists at other medical centers cannot be identified and destroyed because your name was removed before the samples were shipped.

10) HOW WILL MY STUDY INFORMATION BE KEPT PRIVATE?

Information collected for this study will be kept confidential to the extent provided by law. Information used and/or disclosed (shared with someone outside of Nationwide Children's Hospital) may include information that can identify you. This is called "protected health information" or PHI. By agreeing to be in this study, you are giving permission or authorizing Dr. Vidu Garg, Dr. Kim McBride and their study staff to collect, use, and disclose your PHI for this research study. Information collected is the property of Dr. Vidu Garg and Dr. Kim McBride. In the event of any publication regarding this study, your identity will not be revealed.

If you have a bad outcome or adverse event from being in this study, the Principal Investigators and staff or other health care providers may need to look at your entire medical records.

The PHI collected or created under this research study will be used/disclosed as needed until the end of the study. The records of this study will be kept for an indefinite period of time.

PHI that may be used or disclosed: Names (individual); Address; Telephone/Fax Numbers; Dates (except for years); Birth Date; Date of Death; Medical Record Numbers, Email Address.

People or Companies authorized to use, disclose, and receive PHI collected or created by this research study:

PI and study staff

The Nationwide Children's Hospital Institutional Review Board (the committee that reviews all human subject research)

Nationwide Children's Hospital internal auditors

The Office for Human Research Protections (OHRP) (the federal government office that oversees human subject research)

Department of Health and Human Services, NIH

Because of the need to give information to these people, absolute confidentiality cannot be guaranteed. Information given to these people may no longer be protected by federal privacy rules.



Reason(s) why the use or disclosure is being made: The reason that PHI is being used to locate medical charts and to contact you in the future.

You may decide not to authorize the use and disclosure of your PHI. However, if it is necessary for this study, you will not be able to be in this study. If you agree to be in this study and later decide to withdraw your participation, you may also withdraw your authorization to use your PHI. This request must be made in writing to the Principal Investigator(s). If you withdraw your authorization, no new PHI may be collected and the PHI already collected may not be used unless it has already been used or is needed to complete the study analysis and reports.

Dr. Vidu Garg keeps a database of all subjects who participate in a research study. This database may be used to contact people about future studies. Only Dr. Vidu Garg and his staff have access to this database. The database will not be disclosed or sold to others outside Nationwide Children's Hospital.

Please initial:

I want to be contacted about future research studies.

I do not want to be contacted about future research studies.

Certificate of Confidentiality: Dr. Garg has obtained a Certificate of Confidentiality from the Federal government. This Certificate will help researchers protect your privacy. However, the Certificate will not protect your privacy if you consent in writing to the release of information about your participation in this research to anyone else.

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).

You should understand that even with a certificate of confidentiality the investigator is not prevented from taking steps, including reporting to authorities any suspected abuse of neglect of children and to prevent anyone from carrying out any threats to do serious harm to yourself or others. If the staff is given information that causes concern about your safety or the safety of others, they are required to take the necessary steps to protect the persons even if it requires reporting you to the authorities.

Only information in your records would be disclosed for the limited purpose of preventing serious harm to the person or persons endangered. The investigator may voluntarily disclose information if required by the FDA, members of Congress or law enforcement officials.

A Federal law, called the **Genetic Information Nondiscrimination Act (GINA)**, generally makes it illegal for health insurance companies, group health plans, and most employers to discriminate against you based on your genetic information. This law generally will protect you in the following ways:

- Health insurance companies and group health plans may not request your genetic information that we get from this research.
- Health insurance companies and group health 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 that we get from this research when making a decision to hire, promote, or fire you or when setting the terms of your employment.

All health insurance companies and group health plans must follow this law by May 21, 2010. All employers with 15 or more employees must follow this law as of November 21, 2009. Be aware that this new Federal law does not protect you against genetic discrimination by companies that sell life insurance, disability insurance, or long-term care insurance.

11) OTHER IMPORTANT INFORMATION

You may be told the results of this study at a later date.

Being in more than one research study at the same time may cause injury. Please tell the Principal Investigator about being in any other research study so a decision can be made about being in more than one study at the same time.

12) WHOM SHOULD I CALL IF I HAVE QUESTIONS OR PROBLEMS?

If you have questions about anything while on this study or you have been injured by the research, you may contact the Principal Investigator at 614-355-5759, Monday – Friday, between 9:00 am and 5:00 pm.

If you have questions, concerns, or complaints about the research, questions about your rights as a research volunteer, cannot reach the Principal Investigator, or want to call someone else, please call (614) 722-2708, Nationwide Children's Hospital Institutional Review Board, (IRB, the committee that reviews all research in humans at Nationwide Children's Hospital).

FOR REVIEW

Subject's Name _____ Date of Birth _____

**SUBJECT or SUBJECT'S PARENT OR PERSON AUTHORIZED TO CONSENT ON BEHALF OF
THE CHILD (SUBJECT TO THE SUBJECT'S GENERAL MEDICAL CARE)**

I have read this consent form and have had a chance to ask questions about this research study. These questions have been answered to my satisfaction. If I have more questions about participation in this study or a research-related injury, I may contact the Principal Investigator. By signing this consent form, I certify that all health information I have given is true and correct to the best of my knowledge.

I have been given a copy of the Nationwide Children's Hospital Notice of Privacy Practices. I understand that my right to my patient information that is created or collected by Nationwide Children's Hospital in the course of this research can be temporarily suspended for as long as the research is in progress. I also understand that my right to access will be reinstated upon completion of this research.

I agree to participate in this study/I give permission for my child to participate in this study. I will be given a copy of this consent form with all the signatures for my own records.

CONSENT SIGNATURES

SUBJECT or SUBJECT'S LEGAL REPRESENTATIVE

DATE & TIME AM/PM

SUBJECT or SUBJECT'S LEGAL REPRESENTATIVE

DATE & TIME AM/PM

Permission of the second parent not obtained because (select all that apply):

- Not required by the IRB (risk level 1 or 2).
- Other parent is deceased.
- Other parent is unknown.
- Other parent is not reasonably available.
- Only one parent has legal responsibility for the care and custody of subject.

PERSON OBTAINING CONSENT

DATE & TIME AM/PM

I certify that I have explained the research, it's purposes, and the procedures to the subject or subject's legal