PEDIATRIC CARDIAC GENOMICS CONSORTIUM

CONGENITAL HEART DISEASE GENETIC NETWORK STUDY (CHD GENES)

(Insert site Information as required)

ADULT PARTICIPANT CONSENT in the case of a fetus with congenital heart disease

YOUR PARTICIPATION IS VOLUNTARY

You and your family are being invited to take part in a research study to discover the genetic causes of congenital heart defects (CHD). This study will include fetuses, children and adults with congenital heart defects, their parents and in some cases other family members. This form will give you information about the study. It describes the purpose of this research study and the risks and potential benefits. You can decide if you want to be in the study after reading this form and discussing it with the study doctor/staff.

This research is being performed by the Pediatric Cardiac Genetics Consortium and is sponsored by The National Heart, Lung, and Blood Institute (NHLBI) of the National Institutes of Health (NIH).

Why is this research being done?

Congenital heart defects (CHD) are the most common major birth defect. Doctors know little about the causes of these defects but would like to learn more. For this reason, a group of doctors and specialists have come together to search for genes that cause these heart defects or that influence how people with these defects do over time. Genes are the blueprint for how we look, how our bodies work, and how our bodies respond to disease. This study will use blood, saliva and in some cases tissue samples to look closely at the genes.

We hope that the results of this research will eventually help in preventing congenital heart defects, developing new tests to screen for them, or finding new treatments. We are asking many hundreds of people to be in this study at a number of medical centers in the U.S. and other countries. The research will continue through 2020 or longer if money is available. The study includes both males and females, any race or nationality, and pregnant women with a congenital heart defect.

What will happen during the study?

1. Interview and Family History Collection

Individuals with a fetus with a congenital heart defect: If you have a pregnancy with a fetus that has a congenital heart defect, you will be interviewed. Information such as the following will be collected: your age and race/ethnicity; medical history, including heart history and lifestyle; your pregnancy history; and a family history of blood relatives such as parents, grandparents, brothers, sisters and others. The family information collected will include their ages, their heart health history and other health conditions, and their cause of death if they are no longer alive.

2. Medical Records

Information will be collected from medical records at this medical center. You may also be asked if health related information can be used from other outside doctor's medical records. If outside records are to be used, we will ask you to sign a separate *Medical Release Form.*

3. Blood for use in this Research

Adults: If you are pregnant with a fetus or the biologic father of a fetus with a congenital heart defect, about 1 tablespoon will be taken. If there is left over blood available from clinical testing, the study may collect some for use in this research. If for any reason the sample taken is not enough, we will request another sample.

4. Saliva (spit)

*S*ome participants in the study may not be able to give blood. For those people, a saliva sample will be collected.

5. Tissues from a Fetus

In the case in which a pregnancy with a fetus with congenital heart disease ends, tissue from the fetus will be collected.

6. Excess or Discarded Samples

Left over DNA or excess/discarded samples from the fetus may also be collected from the laboratory if an amniocentesis, chorionic villus sampling, or percutaneous umbilical blood sampling was performed.

7. Echocardiogram and/or ECG

Based upon personal and family history, parents may be asked to have an echocardiogram and/or ECG done. An echocardiogram is a painless test using sound waves that takes a 2-dimensional picture of the heart. You will need to lie quietly on a table for about 30 minutes while the test is being done. An ECG is a painless test that is performed while you lie quietly on a bed for 5 minutes. It involves placing electrodes on the chest and arms/legs and recording the electrical activity of the heart.

8. Information from Other Family Members (grandparents, brothers and sisters, extended family)

We may ask you to tell other family members about this research study to see if they will agree to take part and to give their samples. If you agree to contact your family members, you will get some written information for them to read. You do not have to ask your family members to be in the study, and you can still take part in this study if you choose not to involve your family. (Or insert related site-specific language)

9. Follow-up Contact

   During the study, the study doctor/staff may contact you again to ask for health and medical updates. The types of information collected may include general health questions and questions about other heart related medical changes since the last interview.

What are the risks and discomforts of the research study?

There are known and possible risks and discomforts to being in this study. They are:

1. Medical History Collection

   Answering questions and sharing information about yourself and your family may be uncomfortable, cause stress, or make you nervous. You do not have to answer any question that makes you nervous.

2. Blood Collection

   If a separate blood draw (not from indwelling line) is required, there may be minor discomfort, bruising, or rarely dizziness or fainting. There is also a very small risk of infection at the site, but antiseptic solutions will be used to prevent this. Taking the research sample at the same time as a routine blood test will not cause any additional discomfort. There is no pain or discomfort if the blood sample is drawn from an indwelling line.

3. Saliva (spit) Collection

   There are no known risks to collecting spit.

4. Tissue Collection from a Fetus After the End of a Pregnancy

   There is no risk to collecting tissue from a fetus after the pregnancy has ended.

5. Collection of Excess or Discarded Samples

   There are no risks to collecting samples that would otherwise be discarded.

6. Echocardiogram and ECG

   There are no risks to having these tests done, but the tests may discover new medical conditions that were not known.

7. Genetic Testing

The results of genetic testing will not be given to you or your family. We use a research lab, not a clinical lab with certified procedures for reporting results. We will not understand the meaning of most of the differences in this information until there is more research in the future. You should be aware that this research may detect cases where the father of a child is someone other than who it was thought to be (non-paternity). Non-paternity will be kept in the strictest confidence and will not be shared, even with you or your family members. Some people in genetic studies feel anxious if they think they might have a gene that puts them at risk or that may be passed on to children. If you have these feelings at any time during the study, you may contact us and we will arrange for you to speak with someone who can help you.

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 protects you as follows:

- 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.

However, you should 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. For this reason, we will take several steps to keep all data private and confidential. Please see the section on Confidentiality below.

There may be unknown risks to participating in this study, and they may develop during the research study.

Are there benefits to taking part in the research study?

Most families will not benefit directly from the study. The information gathered during this study may someday be of benefit to future congenital heart patients by helping the researchers understand the genetic causes of congenital heart defects. This information may lead to improvements in a doctor's ability to prevent, diagnose or treat people with congenital heart defects.

If a previously unrecognized medical finding is found by echocardiogram or ECG, you will be told about it. The study doctor/staff will talk with you about the findings and your options. You may be told to follow up with other specialists or doctors for further care.

The samples that you and/or your fetus provide may be used to develop new medical tests or treatments. It is possible that the researchers, hospitals, or companies sponsoring the research might benefit financially if the tests or treatments can be patented or commercialized (sold). There are no plans to provide you with payments or royalties if these discoveries are marketed (sold) or licensed. Although subjects and

their families will not receive any compensation now or in the future for their samples or data, income that may be derived from future research or sales of the grouped data will be used to support biomedical research.

Can I be pregnant and in the study?
Pregnant women who have a congenital heart defect or who are carrying a fetus with a congenital heart defect can be included in this study.

Will it cost you anything to be in this study?

There is no cost to you for being in the study.

Will you receive any money for joining this study?

You will receive no payment for taking part in this study.
OR
You will receive $$$ for (Insert site-specific language).

What happens if you believe you are injured during this study?

Tell the study doctor/staff immediately if you believe you have been hurt while in the study. Emergency medical treatment may be available**.** Money for pain, expenses, lost wages and other problems caused by an injury is not available. The NHLBI/NIH will not pay the bills for the care. (Insert site-specific language)

HIPAA and or site HIV policy language
(Insert site-specific language)

How will information be kept confidential and private?

We are very concerned about keeping data secure and private in this genetic study. Information collected during the study and the results of the research tests will not be placed in your medical record. All study paperwork including this signed form will be (kept in a secure location **OR** site specific location to be noted) in the hospital.

The medical information, test results, and blood, spit, tissue and/or excess/discarded samples collected for this study will be kept in special study databases and collection facilities (repositories) that will be available to the study researchers, other persons with permission for related research, and the study sponsor. It is very unlikely that the study information will get out to others within the hospital, an insurance company or employer. The genetic content of the stored DNA or cells could theoretically be used (though with enormous difficulty) to identify you. If you believe you will be bothered by this, talk with the study doctor/staff before you sign this form.

To help us protect you, your fetus, and family's privacy, there are many levels of security. First, no names will be attached to the information/data or samples collected. Samples and data will be given a unique study number, and only this study number will be put on all of the study paperwork, data files, and samples. Second, the data centers storing all of the data, as well as the laboratories and study centers all have layers of security against hacking, mis-handling and unauthorized access. Third, all study doctors

and their staff will follow hospital and study rules for keeping all study information private.

We will do everything we can to keep others from learning about your participation in this study. To further help us protect your privacy, we have obtained a Certificate of Confidentiality from the United States Department of Health and Human Services (DHHS). With this certificate, we cannot be forced (for example by court order or subpoena) to disclose information that may identify you in any federal, state, local, civil, criminal, legislative, administrative, or other proceedings. Parents or legal guardians have the right to information regarding a minor child, unless an Institutional Review Board has approved the study with a waiver of parental permission. You should understand that a Certificate of Confidentiality does not prevent you, or a member of your family, from voluntarily releasing information about yourself, your fetus, or your involvement in this study. The researchers however, will not disclose voluntarily, or without your consent, information that would identify you or your fetus as a participant in this research project.

If an insurer or employer learns about your participation, and obtains your consent to receive research information, then we may not use the Certificate of Confidentiality to withhold this information. This means that you and your family must also actively protect your own privacy! You should understand that we will in all cases, take the necessary action and report to authorities, any indication of abuse, and to prevent serious harm to yourself or others as in the case of child abuse or neglect. Disclosure will be necessary, however, upon request of DHHS for the purpose of audit or evaluation, and is limited only to DHHS employees involved in Study evaluation.

At each study center, a list will be kept that links the study number with your/your fetus' name and contact information (such as address and telephone numbers). This list will be kept in a password protected computer, will be kept separately from the study data, and will not be shared with researchers outside the study center. It is important to keep this list so that the study doctor/staff can contact you to discuss any clinically relevant findings, to review your/your fetus' medical status during the course of the study, and to obtain new genetic samples, if the need arises.

For quality control purposes, the sponsor (NHLBI) or its designee (including the New England Research Institutes), your doctor, and (LOCAL CENTER NAME) Institutional Review Board (IRB) will be able to inspect your medical records and have access to confidential information, which identifies you by name. At no time will your personal information be revealed during any tabulation, presentation, or publication of the results of this research study.

What is the long term plan for my information?

The collected genetic and clinical information will be available indefinitely in a secure storage site such as the National Center for Biotechnology Information (NCBI) repository. The genetic sample from blood, tissue, and/or saliva will be stored indefinitely in a 'biobank', like the NHLBI biorepository.

As part of you/your fetus' participation in the study, a unique subject number will be assigned to you/your fetus that will allow researchers to see if your family has been involved in more than one research study or database for patients with congenital heart

disease. If your family has participated in more than one CHD study or database, this unique subject number may prevent any incorrect duplication of findings. This subject number will also allow your de-identified data to be combined with data from other research studies to increase the likelihood of meaningful analysis. Only this subject number and not your personal identifiable information will be accessible to other investigators.

Data and samples from these storage sites will be available to other, future researchers for study of cardiovascular and other diseases. Before a researcher can get any study data or samples, they must get NIH approval. The researchers will not have access to any identifying information such as your name or medical record number. With this security measure, researchers will not be able to link samples or information back to you. Knowing that samples may be used by a research team in the future to study the heart or other conditions may cause stress to some people. If you believe you will be bothered by this, talk with the study doctor/staff before you sign this form.

What other choices are there?

This study does not involve treatments. Therefore, the only alternative to participating in the study is to not participate.

Can I be withdrawn from the study or ask to be taken out of the study?

You/your fetus may be taken out of the study any time if it is in your/your fetus' best interest, if you/your fetus is unable to complete the study testing, or if the study is stopped by NIH, the Sponsor of the study.

You are free to stop participating in the study at any time. If you decide to drop out of the study, the information already collected from you/your fetus will remain in the study database, identified only with your/your fetus' study number. If you want your samples taken out of storage and destroyed, you will need to write a letter to the study doctor. Once the letter is received, the blood, spit, tissue and/or excess/discarded samples will be destroyed, but any results from testing of the sample up to that point will stay as part of the study data.

What are my rights and responsibilities as a research subject?

- Being in the study is your choice.
- Your decision about being in the study or even dropping out will not change your care or health care benefits.
- By signing this document, you are agreeing to take part in the research study which includes:
    - Completing interviews about your medical and family history.
    - Allowing collected samples (blood, saliva, tissue, and/or excess/discarded samples) to be used for genetic research testing.
    - Allowing the study team to look at your medical records to collect and store information.

- Having your coded medical and genetic data stored and shared within the Pediatric Cardiac Genomics Consortium.
- Having your coded medical and genetic data and genetic samples stored at secure storage sites and available to future researchers during and after the study has ended.
- Allowing the study team to contact you during the study to collect information about your health and changes in your medical condition.

Who do you call if you have questions about this study?

If you ever have questions about this study or in case of research-related injuries, you should contact Dr._____ at *(Telephone Number).*

If you have questions about the rights of research subjects, you can call (*Insert the name and title of the appropriate country/site-specific IRB representative)* at (*Insert the number).* If you prefer, direct your questions to the following address*: (Insert address of IRB patient representative here).*

CONSENT FOR YOU/YOUR FETUS TO TAKE PART IN THIS RESEARCH STUDY

The research study and consent form have been explained to you by:

Person Obtaining Consent	Signature of Person Obtaining Consent
	Date

By signing this form, you are indicating that you have had your questions answered, you agree to take part in this research study, or you are legally authorized to consent to your fetus' participation. You are also agreeing to let (CENTER NAME) use and share your health information as explained above. If you do not agree to our collecting, using and sharing your health information, you cannot participate in this study.

Name of Subject/Biological Mother	
Signature of Subject/Biological Mother	Date

Name of Subject/Biological Father	
Signature of Subject/Biological Father	Date