



# MASONIC MEDICAL RESEARCH LABORATORY

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## Informed Consent Forms

Attached are two Informed Consent Forms

The first form titled “Informed Consent Genetic Test for Inherited Cardiac Arrhythmias” is the consent to perform genetic screening under our CLIA approved protocols to screen for known genes associated with the syndrome.

The second form titled “Consent Form for Human Subject Research” is the consent to continue to perform genetic screening under our research protocols.

All blood samples will be initially screened under CLIA-approved protocols that are performed on genes that have been previously identified for the disease. The patient’s signature on the first form titled “Informed Consent Genetic Test for Inherited Cardiac Arrhythmias” will permit us to conduct this clinical test.

If a mutation is not detected in previously identified genes or the results are not consistent with the disease, we will need a separate consent from the patient to proceed with the genetic screening of additional candidate genes under our research protocols. The patient’s signature on the second form titled “Consent Form for Human Subject Research” will permit us to continue testing for previously unidentified genes under the research protocol.

The patient’s signature on both forms at this time will avoid any delays. Any results obtained from the screening of a candidate gene will only be available following approval by the New York State Department of Health of that gene protocol.

Should you have questions regarding these forms, please contact us at [genetics@mmrl.edu](mailto:genetics@mmrl.edu) or by phone at 315-735-2217 ext. 119.

## **Informed Consent**

### **Genetic Test for Inherited Cardiac Arrhythmias**

#### **Background**

Some diseases are inherited, or passed from parent to child. The genetic material that is transmitted from parents to children is in the form of DNA. The DNA contains some segments, called genes, which, when damaged, can cause disease. It is now possible to identify gene mutations and evaluate them for abnormal function. Identification of these altered genes can help us understand the basis of the disease and how to design new treatments and cures. In some cases, identification of a faulty gene requires study of multiple members of a family affected with the disease.

1. The purpose of this test is the identification of gene mutation associated with familial inherited cardiac diseases like Brugada syndrome, Long QT syndrome, Short QT syndrome, atrial fibrillation or idiopathic ventricular fibrillation. The results of the test may assist your physician with diagnosis of the disease, its medical management and identification of affected family members.
2. By signing this consent, you authorize your doctor's office to draw and forward your/your child's blood, DNA or tissue samples, electrocardiogram and related clinical information to the Masonic Medical Research Laboratory. A blood sample of 3 tubes (approximately 1 teaspoon each) would be required for adults and a maximum of 1-2 tubes (1-2 teaspoon total) will be required for children under 14. Clinical information may be necessary for test interpretation and is often helpful in guiding the search for the genetic defect. The clinical information and results of this test will be treated in the standard medically confidential manner, and will be released only to the physician(s) ordering the test or other persons authorized by you, in writing, unless otherwise required by federal and state laws. In rare cases in which we experience difficulty in analyzing your sample, a second sample may be requested.
3. The only physical risk present in this test involves drawing blood from a vein, usually from the arm. Rarely bleeding (usually a very small amount) or infection, just like any other small scratch may occur. If any other procedure that requires drawing blood is already being performed, no added risk will occur due to this study. There is a potential risk in genetic testing for uncovering and conveying unwanted information regarding parentage or specific risk for disease.
4. Your/your child's blood or tissue sample and any DNA will be destroyed no more than 60 days after your results are finalized, unless family member testing is requested; in which case all samples will be destroyed no more than 60 days after last family member testing is complete. No tests other than those authorized shall be performed on the sample. If you choose not to proceed with the test after Masonic Medical Research Laboratory receives your sample, it will be destroyed once we have written confirmation of your wishes. If Masonic Medical Research Laboratory is unable to confirm that you

wish to proceed with testing, your sample will be destroyed no less than 60 days after Masonic Medical Research Laboratory's last contact with you.

5. The test will involve direct sequencing of DNA to detect the presence of a genetic variation that may be responsible for the disease. The results of this test are not intended to be used as the sole means for diagnosis or patient management decisions. The performance characteristics of this test were validated by Masonic Medical Research Laboratory and it is specific only for variants in genes that are associated with cardiac ion channel mutations. The U.S. Food and Drug Administration (FDA) has not approved this test; however, FDA approval is not currently required for clinical use of this test. This test was completed in a CLIA/CLEP approved laboratory.
6. By signing this consent, you give Masonic Medical Research Laboratory permission to retain your/your child's genetic information generated by this test and to contact you if Masonic Medical Research Laboratory learns new information about the genetic variants detected by this test that affects your current test results.
7. In the interest of science, summary results from this test may be presented, for example at meetings, in publications, or on the Internet; however, no information that can identify you will ever be disclosed, unless authorized in writing by you or required by law.

**I have read this form completely and understand its contents. I have had the opportunity to ask questions about this form and have had any questions answered. I will receive a copy of this consent form.**

**NAME OF PHYSICIAN:** \_\_\_\_\_

\_\_\_\_\_  
**Signature of Patient or Legal Guardian:**

\_\_\_\_\_  
**Signature of Witness**

\_\_\_\_\_  
**Date (DD/MM/YYYY)**

\_\_\_\_\_  
**Date (DD/MM/YYYY)**

\_\_\_\_\_  
**Print Name**

\_\_\_\_\_  
**Print Name**

**If this consent is signed by a Legal Guardian, state the relationship to the Patient:** \_\_\_\_\_

**NOTE:** Genetic testing on children less than 18 years of age requires that the ordering physician obtain an informed consent from a parent or legal guardian.

Consent Form for Human Subject Research  
Conducted at the Masonic Medical Research Laboratory  
Molecular Genetics Laboratory  
Molecular Genetic Basis for Inherited Cardiac Arrhythmias  
IRB Re-approval 02/04/2010  
Amendment 1

**Background**

Some diseases are inherited, or passed from parent to child in some families. The material that is transmitted from parents to children is called DNA. The DNA contains some segments, called genes, which, when damaged, can cause disease to the patient. It is now possible to locate these genes in the DNA and analyze them for abnormal function. Identification of these altered genes in the individuals can help understand how the abnormal gene causes the disease. Identification of these genes requires the study of multiple individuals in families affected with the disease. Stem cells created from donated tissues may be used to create cellular models of the disease, which can be used to develop new therapies for the disease. Various aspects of this research study supported by the American Heart Association and National Institutes of Health.

**Purpose**

The purpose of this study is the identification of abnormal genes responsible for familial inherited cardiac diseases like Brugada syndrome, Long QT syndrome, atrial fibrillation or familial bundle branch block. The information obtained will help advance the understanding of the disease being studied. The study will be recruiting individual for the next 10 years and enroll about 500 patients each year.

**Procedures**

You will be one of approximately 500 patients each year for the next 10 years to be asked to participate in this trial. Adult individuals will be asked to donate blood (1-3 teaspoons). Children under 14 will be required to donate a maximum of 1 teaspoon (5 ml). DNA obtained will be used to determine whether the markers follow the disease gene within their particular family. While the individual will have completed his or her role in this project after giving blood, the linkage (or mapping) studies may go on for years before significant results are obtained. On occasion, if DNA is used up, that person may be asked to donate a small sample of blood once again. In some cases you will be asked to donate a small sample of tissue from any part of the body. This may be in the form a skin biopsy. In this case a biopsy refers to the removal of a very small sample of tissue from a living person for laboratory examination. This tissue may be converted to stem cells in order to create models of the disease in which to examine new therapies for your disease.

The samples will be stored in the form of DNA or cell lines in the Molecular Genetics laboratory at the Masonic Medical Research Laboratory. As part of the genetic analysis different candidate genes will be analyzed over time. The DNA will only be used to identify genetic risk factors for cardiac arrhythmias. If the patient decides to withdraw from the study, or if the study is completed, the DNA will be discarded per patient indication. The DNA or cells will not be sold or transferred for commercial purposes. They may be shared with other laboratories for the purpose of research collaboration. Because there is worldwide recruitment, my primary physician will explain the consent and the procedure and answer any questions that I may have.

## Molecular Genetic Basis for Inherited Cardiac Arrhythmias

### **Protected Health Information**

I understand that my physician will be asked to send copies of protected health information related only to this research study to the Masonic Medical Research Laboratory for proper genetic classification. This health information will be secured in compliance to all HIPAA regulations and any information used for the study will not include any personal identification. I authorize the Masonic Medical Research Laboratory to obtain said protected health information from my physician and any medical facility providing care related to my cardiac condition. I understand that I have the right to revoke this authorization at any time. I understand that should I revoke this authorization I must do so in writing to the Masonic Medical Research Laboratory or to my physician in charge of this collaboration.

Due to the research aspect of this study, I understand that I will not be able to receive the results of the genetic screening until the study is approved by the NYS Department of Health for clinical diagnosis. I understand that the results of the genetic study will then only be released to the physician of record. I can request, in writing, that the results be sent to additional physicians for the purpose of continuity of care or the physician on record can be changed. Results obtained will not be given to the patient directly.

### **Potential Risks and Discomforts**

The only physical risk present in this study involves drawing blood from a vein, usually from the arm. Rarely bleeding (usually a very small amount) or infection, just like any other small scratch may occur. In the case of skin biopsies, the risk is mild local pain, slight bleeding, the chance of a small scar, and slight chance of infection. If any other procedure that requires drawing blood is already being performed, no added risk will occur due to this study.

There is a potential risk in genetic testing for uncovering and conveying unwanted information regarding parentage or specific risk for disease. This risk includes: Information that may compromise your insurability and you may learn information about yourself or your family that you did not really want to know, or may be uncomfortable knowing (such as that you not biologically related).

### **Potential Benefits**

You have been told that the benefits of participating in this study may be that the information obtained during this study might result in improved diagnosis and treatment of the disease. However, you may receive no benefit from participating in this study.

### **Alternatives**

The only alternative to this study is non-participation.

### **Subject Costs and Payments**

There are no costs to you subject to your participation in this research study. The only costs that you may encounter are the cost associated with obtaining and sending the sample to us. Additional treatment or genetic counseling may be warranted as determined by the course of treatment agreed upon by you and your physician.

### **Subject's Rights**

There may be unknown risks/discomforts involved. You will receive any new information discovered during the course of this study, concerning significant treatment findings that may

## Molecular Genetic Basis for Inherited Cardiac Arrhythmias

affect your willingness to continue participating in this research study.

Every effort will be made to maintain the confidentiality of your study records. The data from the study may be published; however, you will not be identified by name. The confidentiality of the data will be maintained within legal limits. In the event of injury resulting from this research, Masonic Medical Research Laboratory is not able to offer financial compensation nor to absorb the costs of medical treatment. However, necessary facilities, emergency treatment and professional services will be available to you, just as they are to the general community. Your signature below acknowledges your voluntary participation in this research project. Such participation does not release the investigator(s), institution(s), sponsor(s) or granting agency(ies) from their professional and ethical responsibility to you.

You may refuse to participate or may discontinue your participation AT ANY TIME, without penalty, loss of benefits, or change in present or future care. The investigator has the right to withdraw you from the study at any time. Your withdrawal from the study may be for reasons related solely to you (e.g. not following study-related directions from the Investigator, a serious adverse event reaction) or because the entire study is terminated. The Sponsor has the right to terminate the study or the Investigators participation in the study at any time.

The investigator, CHARLES ANTZELEVITCH, PhD, FACC, FAHA, FHRS and/or his designee has attempted to answer all of your questions. If you have further questions or concerns, please address them with the study representative now. For questions about your rights as a research participant, you may contact the Faxton-St. Luke's Healthcare Institutional Review Board (a group of people who review the research to protect your rights) representative MARY KATE SCHUSTER, RN, OCN, Cancer Program Director at (315) 624-5722. If questions/concerns arise during the course of the study, you may speak with the principal investigator, DR. CHARLES ANTZELEVITCH at (315) 735-2217 or by e-mail at [genetics@mmrl.edu](mailto:genetics@mmrl.edu)

Signing this consent form indicates that you have read this consent form (or have had it read to you), that your questions have been answered to your satisfaction, and that you voluntarily agree to participate in this research study.

\_\_\_\_\_  
Subject

\_\_\_\_\_  
Date

\_\_\_\_\_  
Legal Representative or Next of Kin

\_\_\_\_\_  
Date

\_\_\_\_\_  
Relationship to Subject

\_\_\_\_\_  
Investigator or Designee Obtaining Consent

\_\_\_\_\_  
Date

\_\_\_\_\_  
Witness (if applicable)

\_\_\_\_\_  
Date

### CHILDREN CONSENT

Your signature on this consent form attests to the fact that your child \_\_\_\_\_ has, within limits imposed by age, maturity, and psychological state, given his/her assent (affirmative agreement) to participate in this research project.