Link My Heart
BRUGADA PROJECT

For your physician

The Brugada Project is part of *Link My Heart*, an initiative with the goal of connecting patients with researchers to work towards improving research, discovery, and treatment for individuals with inherited heart conditions that may lead to sudden cardiac death.

The Brugada Project directly partners with patients with Brugada syndrome to collect health information and samples, with the goal of advancing genetic research. This condition is rare, and by directly connecting patients and researchers, we hope to rapidly expand our understanding of the genetic basis of Brugada syndrome, and ultimately other inherited heart conditions, to improve the health of patients and their family members.

The study is led by Dr. Patrick Ellinor, MD, PhD at Massachusetts General Hospital and the Broad Institute in collaboration with investigators from several academic institutions across the country.

What is the goal of the Brugada Project?

The overarching goal of the project is to create a controlled access database on inherited arrhythmia syndromes, comprising clinical elements from medical records, patient-reported data from surveys, and genomic data from patient samples.

We expect that researching the DNA of individuals with Brugada syndrome will help us learn which genetic variants cause Brugada syndrome. This may allow us to better identify individuals at risk for Brugada syndrome, and develop treatments specific for the condition.

Inherited heart conditions like Brugada syndrome are an important cause of sudden death. These conditions are individually rare, making them difficult to study. We would therefore like to enable individuals with Brugada syndrome to contribute to research to help us move the science forward. Ultimately, we hope to advance our understanding of the genetic basis of Brugada syndrome, and other inherited heart conditions, by connecting the data of patients together.

What is involved in this research?

Participants register for the study online at www.brugada.linkmyheart.org. By consenting to the study, participants allow us to request and obtain copies of their medical records and a saliva sample to perform genetic research.

What steps are involved for the physician's offices of enrolled participants?

When participants enroll, they provide information about where they have received care for their Brugada syndrome and sign a medical release form. The study team works directly with medical record departments to obtain copies of medical records.

Do participants or physicians receive individual results back from this study?

No. Because our sequencing tests are performed in a research lab and not a clinical lab, we are not currently sending study results to participants or physicians. In the future, we hope to incorporate ways to update participants with key discoveries, overall results, and progress made through this research.

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What makes this project different from existing studies?

We are building a web-based software platform to enable individuals to enroll and contribute both clinical data and DNA to advance scientific discoveries, and to develop an open science research model by providing data access to researchers. Most patients are cared for in community settings where genomic studies are not typically conducted, so this study connects patients around the country with genomics researchers, allowing them to participate regardless of where they live. We believe this approach can be an important resource for helping solve many of the unmet scientific needs related to sudden cardiac death. We hope that such an approach will lead to improved treatments, risk stratification tools, targeted therapeutics, and in general a better understanding of these conditions. Ultimately, such insights may help individuals with these conditions and family members at risk lead longer and healthier lives.

How is the genomic and clinical data shared with researchers?

Genomic assays are performed on DNA derived from the participant's cells. We intend to use both genotyping and next generation sequencing technologies such as whole exome or whole genome sequencing to identify the drivers of disease. The database of clinically annotated genomic information is shared on publicly available platforms such as those hosted by the National Institutes of Health (NIH) to allow as many researchers as possible to help make scientific advances.

What kinds of questions is this project aiming to address?

Over the past decade, genomic assays have provided insights into the causes of human traits and diseases. However, large sample sizes are necessary for making these insights. On average, a disease-causing mutation can only be identified in about 20-30% of patients with Brugada syndrome. No specific treatments have been developed for patients with Brugada syndrome. To identify the genetic causes of Brugada syndrome, much larger sample sizes will be necessary than exist today. Yet like other genetic arrhythmia syndromes, Brugada syndrome is rare, making recruitment of individuals with the condition very challenging. We seek to answer several major questions:

- What are all the genes and mutations that can lead to Brugada syndrome?
- What explains why some individuals who carry a disease-causing genetic variant have signs of Brugada syndrome whereas others do not?
- What explains why some patients with Brugada syndrome have lethal ventricular arrhythmias, whereas others do not?
- How can we develop better treatments for Brugada syndrome?

Despite the progress being made to answer these questions, much remains to be learned and we are far from the ultimate goal of developing a specific therapy for patients with Brugada syndrome. We hope that this direct to patient approach will help facilitate research and allow for larger sample sizes and critical insights into the biological mechanisms, and manifestations, of this condition.

If you have any questions about this study, please reach out to us at info@linkmyheart.org or 617-714-7560.

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Help advance our understanding of the genetic basis of

Brugada syndrome

We are enrolling patients in the Link My Heart - Brugada Project, a research study that directly connects patients and researchers, with the goal of rapidly advancing genetic research related to Brugada syndrome. The study is led by Dr. Patrick Ellinor, MD, PhD at Massachusetts General Hospital and the Broad Institute in collaboration with investigators from several academic institutions across the country.

What does the study entail?

The Link My Heart - Brugada Project empowers patients to directly connect with researchers. Participants will be asked to spend less than 30 minutes of their time to answer our brief online health questionnaires, and provide permission for their medical records to be shared with the research team. In addition, participants will be mailed a saliva collection kit and asked to provide a sample for research-based genetic testing. This study is expected to continue for many years and participants may be asked to complete follow-up questionnaires about their health history. Participants are not required to visit a study center. There are no benefits or compensation associated with this study.

Who can participate?

Individuals are eligible to participate if they have a known or suspected diagnosis of Brugada syndrome, are 21 years of age or older, and reside in the USA or Canada.

Why is it important?

Inherited heart conditions, like Brugada syndrome, are an important cause of sudden cardiac death. These conditions are individually rare, making them difficult to study. The Link My Heart - Brugada Project enables individuals with Brugada syndrome or other inherited heart conditions to contribute to research and move science forward. Ultimately, we hope to discover new genetic causes of inherited heart disease, which may allow for improved identification of at-risk individuals and the development of condition-specific treatments.

How do I participate?

For more information, or to enroll, please visit:
www.brugada.linkmyheart.org.

If you have questions and would like to speak with our study staff, please reach us at (617) 714-7560 or email info@linkmyheart.org.