Ambry Genetics®
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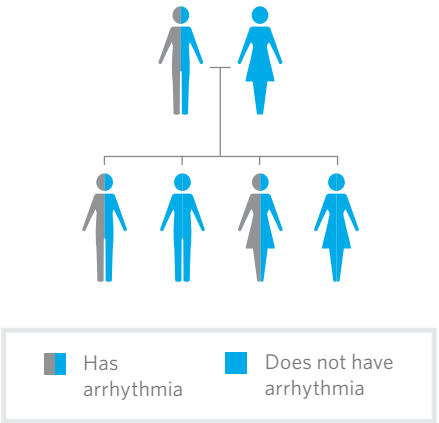
Understanding Your Positive Arrhythmia Genetic Test Result

INFORMATION FOR PATIENTS WITH A **PATHOGENIC MUTATION** OR **VARIANT THAT IS LIKELY PATHOGENIC**

Result	**POSITIVE**	Your testing shows that you have a pathogenic (disease-causing) mutation, or a variant that is likely disease-causing, in a gene that causes an inherited arrhythmia like Brugada syndrome, long QT syndrome or CPVT. Both mutations and variants that are likely disease-causing should be treated as the same type of positive result.
Gene	**DEFINITION**	Everyone has two copies of each gene. We get one copy from each of our parents. Mutations (changes in the gene, like spelling mistakes) in one copy of any of the genes in this test can cause Brugada syndrome, long QT syndrome or another inherited arrhythmia.
Diagnosis	**ARRHYTHMIA**	An arrhythmia is an abnormal heartbeat. Specific arrhythmias, like long QT syndrome, Brugada syndrome, and CPVT are inherited. This means that they run in families.
Management Options	**FOR PATIENTS WITH ARRHYTHMIA**	Treatment options may include: medications, surgery, pacemakers, implantable cardioverter defibrillators (ICDs), or avoiding certain athletic activities. Talk to your doctor about which may be right for you.
Screening Options	**FAMILY MEMBERS**	Options for screening and early detection for inherited arrhythmias include: physical exams and electrocardiograms (EKGs). Talk to your doctor about which may be right for you and/or your family.
Next Steps	**DISCUSS**	Please share this with family members so they can talk with their doctors and learn more. They can now be tested for this same mutation, if they choose to.
Reach Out	**RESOURCES**	• Ambry's Cardiology Site for Families patients.ambrygen.com/cardiology • National Society of Genetic Counselors nsgc.org • Sudden Arrhythmia Death Syndromes (SADS) Foundation sads.org • Genetic Information Nondiscrimination Act (GINA) ginahelp.org

Arrhythmias in the Family

Your close family members (like your parents, brothers, sisters, children) have a 50/50 chance of having the mutation that you carry, and other family members (like your aunts, uncles, cousins) may also have it. Your relatives can now be tested for this same mutation. Those who DO NOT have this mutation may not be at risk for arrhythmia and can avoid unneeded screening.



Please talk with your doctor or genetic counselor about this. The field of genetics is continuously changing, so updates related to your result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a healthcare provider, and should not be considered or taken as medical advice.

7 Argonaut, Aliso Viejo, CA 92656 USA Toll Free +1 866.262.7943 Fax +1 949.900.5501 ambrygen.com