Ambry Genetics®

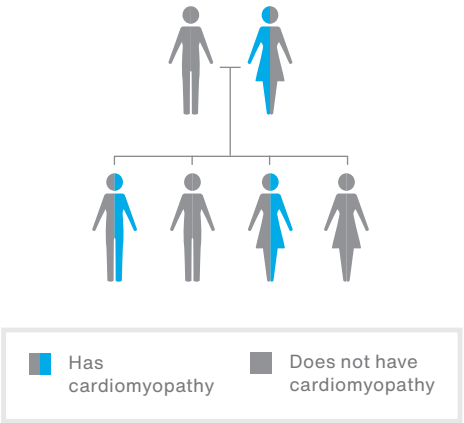
Understanding Your Positive Cardiomyopathy Genetic Test Result

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 cardiomyopathy like hypertrophic or dilated cardiomyopathy. Both mutations or 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 inherited cardiomyopathy.
Diagnosis	CARDIOMYOPATHY	Cardiomyopathy weakens, and often enlarges, the heart muscle. Some cardiomyopathies are inherited, which means they run in families.
Management Options	PATIENTS WITH CARDIOMYOPATHY	Treatment options 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 cardiomyopathy include: physical exams, echocardiograms, electrocardiograms (EKGs), and cardiac MRI. 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 • Hypertrophic Cardiomyopathy Association 4hcm.org • Children's Cardiomyopathy Foundation childrenscardiomyopathy.org • Genetic Information Nondiscrimination Act (GINA) ginahelp.org

Cardiomyopathy 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 cardiomyopathy and may 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.

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