Ambry Genetics®

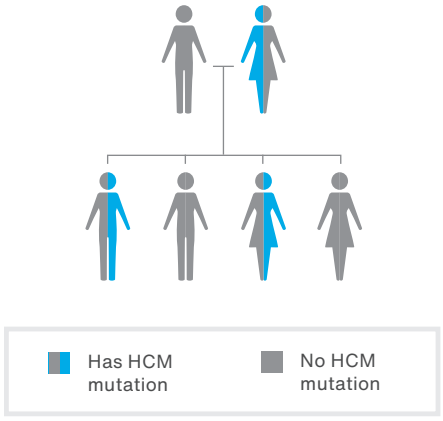
Understanding Your Positive Hypertrophic Cardiomyopathy (HCM) Genetic Test Result

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 HCM. Both of these should be treated as the same type of positive result.
Gene	DEFINITION	Everyone has two copies of each gene. We get one copy of each gene 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 HCM.
Diagnosis	HCM	People with this result have HCM, a disease that causes thickening of the heart muscle.
Management Options	FOR PATIENTS WITH HCM	Treatment options may include: medications, surgery, pacemakers, or implantable cardioverter defibrillators (ICDs). Talk to your doctor about which options may be right for you.
Screening Options	FAMILY MEMBERS	Options for screening and early detection for HCM include: physical exams, echocardiograms, electrocardiograms (EKGs) or cardiac MRI. Talk to your doctor about which options 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 • Genetic Information Nondiscrimination Act (GINA) ginahelp.org

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