Ambry Genetics®
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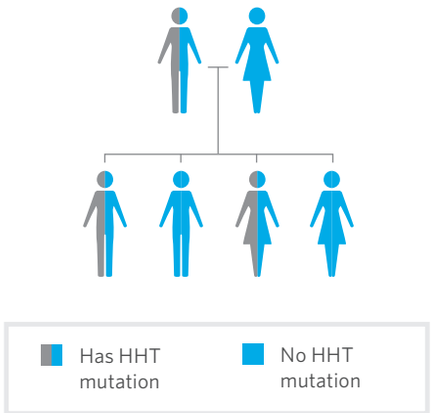
Understanding Your Positive HHTNext Genetic Test Result

INFORMATION FOR PATIENTS WITH ONE **PATHOGENIC MUTATION** OR **VARIANT, LIKELY PATHOGENIC**

Result	**POSITIVE**	The result of your genetic testing shows you either have a change that is known to be disease-causing (pathogenic mutation) or a change or gene variant that is likely to be disease-causing (likely pathogenic), in a gene that causes hereditary hemorrhagic telangiectasia (HHT). Either should be considered 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. A mutation (change in the gene, like a spelling mistake) in one copy of the *ACVRL1*, *ENG*, *SMAD4*, or *GDF2* genes can cause HHT.
Diagnosis	**HEREDITARY HEMORRHAGIC TELANGIECTASIA (HHT)**	HHT is a hereditary disease that most often causes recurrent epistaxis, mucocutaneous telangiectasias, and arteriovenous malformations in various organs.
Management Options	**FOR PATIENTS WITH HHT**	Medical management options may include: medications, surgery, or surveillance. Talk to your healthcare providers about which may be right for you.
Family Members	**50/50 CHANCE**	Your children have a 50/50 chance of inheriting the pathogenic mutation or likely pathogenic variant that you carry. Other blood-related family members (like your parents, siblings, aunts, uncles, cousins) also have a chance of carrying this mutation. Any of your blood relatives can be tested for the inherited mutation.
Next Steps	**DISCUSS**	Please share this with family members so they can talk with their healthcare providers and learn more about genetic testing with Ambry Genetics at ambrygen.com.
Reach Out	**RESOURCES**	• Cure HHT & Backpack Health curehht.org/backpack-health • National Society of Genetic Counselors nsgc.org • Canadian Association of Genetic Counsellors cagc-accg.ca • Genetic Information Nondiscrimination Act (GINA) ginahelp.org

HHT Mutations in the Family

There is a 50/50 chance to pass on a pathogenic mutation or likely pathogenic variant to your sons and daughters. The image below shows that both men and women can carry and pass on these mutations. Those who DO NOT have these mutations may not be at risk for HHT.



Please discuss this information with your healthcare providers. The field of genetics is continuously changing, so updates related to your genetic testing results and/or medical management options 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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