Ambry Genetics®

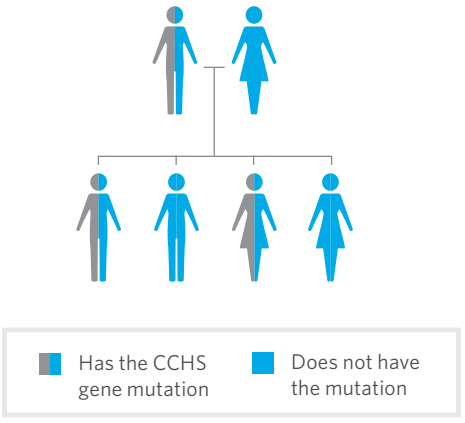
Understanding Your VUS Congenital Central Hypoventilation Syndrome (CCHS) Genetic Test Result

INFORMATION FOR PATIENTS WITH **VARIANTS OF UNKNOWN SIGNIFICANCE**

Result	**VUS**	Testing shows that you/your family member have a variant of unknown significance (VUS) in the gene that causes CCHS, which is called *PHOX2B*. A VUS is a gene change, but we do not know if it causes CCHS or not.
Gene	**DEFINITION**	Genes have instructions for traits that make us who we are, like eye color and how our bodies work. Everyone has two copies of each gene. We get one copy of each gene from each of our parents. A mutation (change in the gene, like a spelling mistake) in one copy of the *PHOX2B* gene can cause CCHS. However, some changes in a gene (like a VUS) are not well understood, so we cannot predict if they will cause CCHS or not.
Diagnosis	**NO CHANGE**	It is possible that you/your family member have CCHS or another condition, with similar symptoms, that this test cannot find. Your doctor or genetic counselor will talk to you about this further, including any medical care that might be helpful.
Screening Options	**FAMILY MEMBERS**	Testing for your family members may help explain this VUS. Talk with your doctor or genetic counselor about which family members it may be helpful to test.
	FOR YOU	Please discuss these results with your doctor or genetic counselor. They can tell you about any additional testing and/or medical care that might be right to consider.
Next Steps	**DISCUSS**	Please share this with family members so they can talk with their doctors and learn more.
Reach Out	**RESOURCES**	• National Society of Genetic Counselors nsgc.org • Canadian Association of Genetic Counsellors cagc-accg.org • CCHS REDcap Registry luriechildrens.org • Genetic Information Nondiscrimination Act (GINA) ginahelp.org

How CCHS is Inherited

People who have a mutation in one copy of their *PHOX2B* gene have CCHS. Sometimes this is passed down from a parent, and sometimes it happens for the first time in that person. When a person with CCHS has children, there is a 50/50 chance they will pass down the CCHS mutation to each of their sons or daughters.



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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