Ambry Genetics®

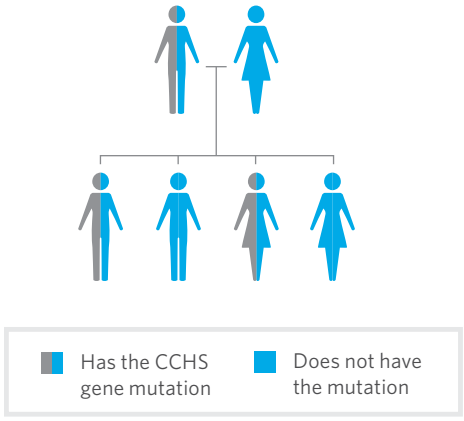
Understanding Your Negative Congenital Central Hypoventilation Syndrome (CCHS) Genetic Test Result

INFORMATION FOR PATIENTS

Result	**NEGATIVE**	Testing did not find a disease-causing change (or mutation) in you/your family member in the gene that causes CCHS (called *PHOX2B*). It is possible that you/your family member may have a mutation in a gene that was not included in this test. If you/your family member have been diagnosed with CCHS already, that will not change.
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.
Diagnosis	**NO CHANGE**	It is possible that you/your family member may have another type of condition 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.
Further Testing	**DISCUSS**	More genetic testing may be right to consider. Please talk about this with your doctor or genetic counselor.
Reach Out	**RESOURCES**	• National Society of Genetic Counselors www.nsgc.org • Canadian Association of Genetic Counsellors www.cagc-accg.org • CCHS REDcap Registry www.luriechildrens.org • Genetic Information Nondiscrimination Act (GINA) www.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.

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