Ambry Genetics®

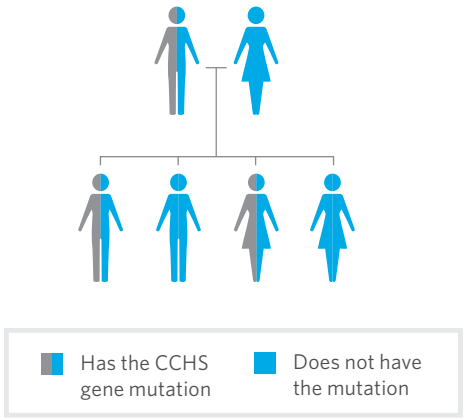
Understanding Your Positive Congenital Central Hypoventilation Syndrome (CCHS) Genetic Test Result

INFORMATION FOR PATIENTS WITH A **PATHOGENIC MUTATION** OR **VARIANT THAT IS LIKELY PATHOGENIC**

Result	**POSITIVE**	Testing shows that you/your family member have a pathogenic (disease-causing) mutation or a variant that is likely pathogenic in the gene that causes CCHS (called *PHOX2B*). Both of these results should be treated as the same type of positive result.
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	**CCHS**	People with this result have CCHS, a condition with a wide range of symptoms and severity. It typically causes troubles breathing, often while sleeping, and sometimes affects other areas of the body such as the intestinal tract. Symptoms usually begin in the newborn period, but can rarely start later (in toddlers, children, or adults).
Management Options	**FOR PEOPLE WITH CCHS**	Treatment options may include ventilator support, surgeries, or other procedures depending on the symptoms. Talk to your doctor about which options may be right to consider.
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 the same mutation, if they choose to.
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.

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