Ambry Genetics®

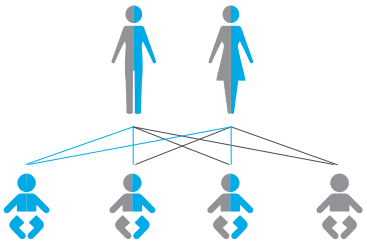
Understanding Your Negative Primary Ciliary Dyskinesia (PCD) Genetic Test Result

INFORMATION FOR PATIENTS

Result	**NEGATIVE**	Your testing did not find any disease-causing changes (mutations) in a gene that causes PCD. 60-70% of people with PCD will have a mutation in one of the genes in this test, so you may have a mutation in a gene not in this test. If you have been diagnosed with PCD, that will remain the same.
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. Usually, a mutation (change in the gene, like a spelling mistake) in both copies of a PCD gene can cause PCD in someone. Most people who carry a mutation in one of their PCD genes are called "carriers." While these carriers do not usually have symptoms, some may have mild symptoms of a PCD-related disorder. If their partner is also a carrier of a mutation in the same gene, the couple has a 1 in 4 (25%) chance to have a child with PCD in each pregnancy together (see below).
Diagnosis	**NO CHANGE**	It is possible that you have mutations in a gene that is not in this test. It is also possible that 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 for you.
Next Steps	**DISCUSS**	More genetic testing may be right for you. Please talk about this with your doctor or genetic counselor.
Reach Out	**RESOURCES**	• National Society of Genetic Counselors nsgc.org • Canadian Association of Genetic Counsellors cagc-accg.ca • PCD Foundation pcdfoundation.org • Genetic Information Nondiscrimination Act (GINA) ginahelp.org

How PCD is Inherited

Most people who carry a mutation in only one copy of one of a PCD gene are called "carriers," and do not usually have symptoms. If their partner is also a carrier of a mutation in the same gene, there is a 1 in 4 (25%) chance for them to have a son or daughter with PCD in each pregnancy together.



- PCD carrier (one gene mutation)
- Does not have PCD, and is not a PCD carrier (no gene mutations)
- Has PCD (two gene mutations)

Rarely, PCD can occur when only one gene mutation is present, usually passed down from a carrier mother to her son. Your doctor or genetic counselor can explain this type of inheritance further, if it applies to your family.

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