Ambry Genetics

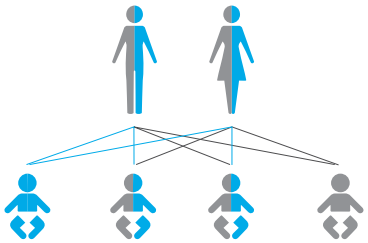
Understanding Your Positive Primary Ciliary Dyskinesia (PCD) Genetic Test Result

INFORMATION FOR PATIENTS WITH **PATHOGENIC MUTATIONS** OR **VARIANTS THAT ARE LIKELY PATHOGENIC**

Result	**POSITIVE**	Your testing shows that you have a combination of two pathogenic or likely pathogenic (disease-causing) mutations in a gene (or genes) that cause PCD. Both of these results should be treated as the same type of positive result. Rarely, PCD can occur when only one gene mutation is present (in a male).
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.
Diagnosis	**PCD**	People with this result have PCD, a medical condition that usually affects the lungs, and sometimes other areas of the body. PCD causes health concerns that can be mild or severe, including an increased chance for infections (like pneumonia).
Management Options	**FOR PATIENTS WITH PCD**	Treatment options may include: medications, inhalers, chest physical therapy, antibiotics, and other options to decrease the risk of infection. Talk to your doctor about which options may be right for you.
Screening Options	**FAMILY MEMBERS**	People who carry one mutation in a PCD gene are called "carriers." While most carriers do not have symptoms of PCD, sometimes carriers may have mild symptoms of PCD. If their partner is also a carrier, the couple has a 1 in 4 (25%) chance to have a child with PCD in each pregnancy together. Your adult family members may wish to be tested to see if they carry the mutation(s) found in your family.
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(s), if they choose to.
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 Commonly 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. There is a 2 in 4 (50%) chance for them to have a child who is a PCD carrier (usually without symptoms), and a 1 in 4 (25%) chance for them to have a child who does not have PCD, and is not a PCD carrier.



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

In rare cases, 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.

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