

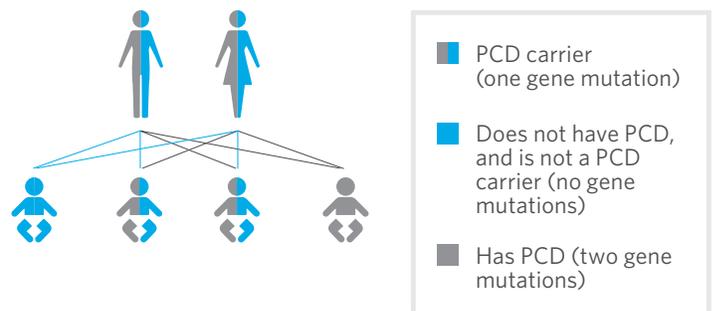
# Understanding Your Primary Ciliary Dyskinesia (PCD) Carrier Genetic Test Result

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

Result	<b>CARRIER</b>	Your testing shows that you have one pathogenic (disease-causing) mutation, or a variant that is likely pathogenic, in a gene that causes PCD. Both of these should be treated as the same type of result. This means you are what is usually called a “carrier” for PCD. It is not likely that you will have symptoms of PCD, but some carriers may have mild symptoms. Rarely, PCD can occur when only one gene mutation is present (in a male).
Gene	<b>DEFINITION</b>	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 <u>both</u> copies of a PCD gene (or genes) can cause PCD in someone. Most carriers have <u>one</u> mutation in a copy of one of their PCD genes. These carriers do not usually have common symptoms of PCD, but can have children with the condition.
Screening Options	<b>FAMILY MEMBERS</b>	If your partner also carries a mutation in the same PCD gene as you do, you could have a 1 in 4 (25%) chance to have a child with PCD in each pregnancy together (see below). Genetic testing for your partner may help you both learn more about this. In addition, your adult family members may wish to be tested to see if they carry the mutation(s) found in your family.
Next Steps	<b>DISCUSS</b>	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	<b>RESOURCES</b>	<ul style="list-style-type: none"> <li>▪ National Society of Genetic Counselors <a href="http://nsgc.org">nsgc.org</a></li> <li>▪ Canadian Association of Genetic Counsellors <a href="http://cagc-accg.ca">cagc-accg.ca</a></li> <li>▪ PCD Foundation <a href="http://pcdfoundation.org">pcdfoundation.org</a></li> <li>▪ Genetic Information Nondiscrimination Act (GINA) <a href="http://ginahelp.org">ginahelp.org</a></li> </ul>

## How PCD is Commonly Inherited

Most people who carry a mutation in only one copy 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.



*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.