

Understanding Your Positive *PHOX2B* Genetic Test Result

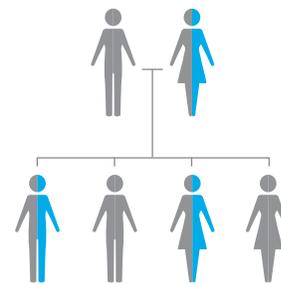
INFORMATION FOR PATIENTS WITH A PATHOGENIC OR LIKELY PATHOGENIC VARIANT

5 Things To Know

1	Result	Your testing shows that you have a pathogenic or likely pathogenic variant in the <i>PHOX2B</i> gene.
2	Non-cancerous tumor and cancer risks	You have an increased chance to develop neuroblastic tumors, which are tumors and/or cancer of the nervous system.
3	Other Medical Concerns	Some people with certain pathogenic or likely pathogenic variants in <i>PHOX2B</i> may also have Hirschsprung disease (a problem within the nerve tissue of the intestines that doesn't allow contents to move through properly) or congenital central hypoventilation syndrome (a condition that occurs in infants that may cause trouble breathing).
4	What you can do	Risk management decisions are very personal. There are options to detect cancer early or lower the risk to develop cancer. It is important to discuss these options with your healthcare provider and decide on a plan that works for you.
5	Family	Family members may also be at risk—they can be tested for the pathogenic or likely pathogenic <i>PHOX2B</i> variant that was identified in you. It is recommended that you share this information with your family members so they can learn more and discuss with their healthcare providers.

PHOX2B in the Family

There is a 50/50 random chance to pass on a pathogenic or likely pathogenic *PHOX2B* variant to each of your children.



■ Has a pathogenic or likely pathogenic *PHOX2B* variant
 ■ No pathogenic or likely pathogenic *PHOX2B* variant

RESOURCES

- American Brain Tumor Association abta.org
- American Cancer Society cancer.org
- National Society of Genetic Counselors nsgc.org
- Canadian Society of Genetic Counsellors cagc-accg.ca

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your *PHOX2B* 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 interpreted as medical advice.