

Understanding Your Positive *PHOX2B* Genetic Test Result

INFORMATION FOR PATIENTS WITH A PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

5 Things To Know

1	<i>PHOX2B</i> mutation	Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic 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 mutations 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 <i>PHOX2B</i> mutation 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 Mutations in the Family

There is a 50/50 random chance to pass *PHOX2B* mutation to your sons and daughters. The image to the right shows that both men and women can carry and pass on these mutations.



Reach Out	RESOURCES	<ul style="list-style-type: none"> American Brain Tumor Association abta.org American Cancer Society cancer.org Genetic Information Nondiscrimination Act (GINA) ginahelp.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.