

Understanding Your Positive SMARCB1 Genetic Test Result

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

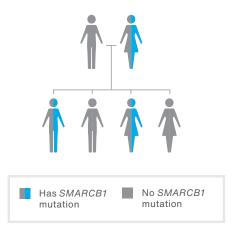
5 Things To Know

1	SMARCB1 mutation	Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the <i>SMARCB1</i> gene.
2	Rhabdoid tumor predisposition syndrome type 1 (RTPS1) or SMARCB1-related schwannomatosis (SWN)	People with <i>SMARCB1</i> mutations have rhabdoid tumor predisposition syndrome type 1 (RTPS1) or <i>SMARCB1</i> -related schwannomatosis (SWN).
3	Non-cancerous tumor and cancer risks	Some people with <i>SMARCB1</i> mutations develop aggressive tumors in the brain or kidney called rhabdoid tumors. The risk for rhabdoid tumors is highest within the first two years of life. Some individuals may also be at increased risk for benign tumors called schwannomas, or meningiomas, both of which typically develop in adulthood. These risks are variable, so it is important to discuss your specific risks with your doctor.
4	What you can do	Risk management decisions are very personal. There are options to detect tumors early. It is important to discuss these options with your doctor 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>SMARCB1</i> mutation that was found 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.

SMARCB1 Mutations in the Family

There is a 50/50 random chance to pass on an SMARCB1 mutation to your sons and daughters. The image to the right shows that both men and women can carry and pass on these mutations.

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your SMARCB1 genetic testing 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.



Reach Out RESOURCES

- 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