

Understanding Your Positive *SMARCA4* Genetic Test Result

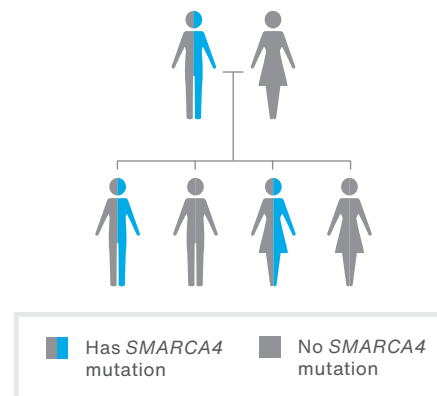
INFORMATION FOR PATIENTS WITH A PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

5 Things To Know

1	<i>SMARCA4</i> mutation	Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the <i>SMARCA4</i> gene.
2	Cancer risks	You have an increased chance to develop a rare ovarian tumor (small cell carcinoma of the ovary, hypercalcemic type or SCCOHT) or rhabdoid tumors of the brain, kidney, or other organs. The risk for SCCOHT is highest within the first 20-30 years of life, and the risk for rhabdoid tumors is highest prior to age 3 years.
3	Other Medical Concerns	Rare mutations in <i>SMARCA4</i> can cause another condition known as Coffin-Siris syndrome (CSS). CSS is usually detected early in childhood or infancy and can cause severe intellectual disability, epilepsy (seizures), and certain facial features. The mutations that cause CSS are usually different than those that cause the tumor condition. Talk to your healthcare provider to find out if this information is relevant for you and your family.
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>SMARCA4</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.

SMARCA4 Mutations in the Family

There is a 50/50 random chance to pass on a *SMARCA4* mutation to each of your children. The image to the right shows that everyone can carry and pass on these mutations, regardless of their sex at birth.



RESOURCES

- American Cancer Society [cancer.org](https://www.cancer.org)
- Genetic Information Nondiscrimination Act (GINA) [ginahelp.org](https://www.ginahelp.org)
- National Society of Genetic Counselors [nsgc.org](https://www.nsgc.org)
- Canadian Society of Genetic Counsellors [cagc-accg.ca](https://www.cagc-accg.ca)

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your *SMARCA4* 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.