

# Understanding Your Positive *SMARCA4* Genetic Test Result

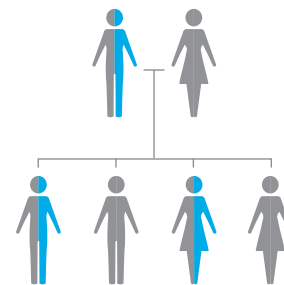
## INFORMATION FOR PATIENTS WITH A PATHOGENIC OR LIKELY PATHOGENIC VARIANT

### 5 Things To Know

1	Result	Your testing shows that you have a pathogenic or likely pathogenic variant in the <i>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 pathogenic or likely pathogenic <i>SMARCA4</i> variants 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 pathogenic and likely pathogenic variants 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 pathogenic or likely pathogenic <i>SMARCA4</i> variant 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* in the Family

There is a 50/50 random chance to pass on a pathogenic or likely pathogenic *SMARCA4* variant to each of your children.



■ Has a pathogenic or likely pathogenic *SMARCA4* variant
 ■ No pathogenic or likely pathogenic *SMARCA4* variant

### RESOURCES

- American Cancer Society [cancer.org](https://www.cancer.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.