

Understanding Your Positive *PRKAR1A* Genetic Test Result

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

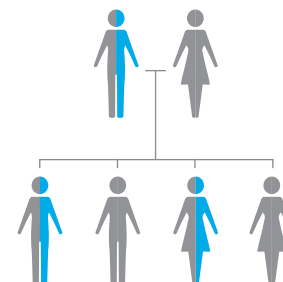
6 Things To Know

1	<i>PRKAR1A</i> mutation	Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the <i>PRKAR1A</i> gene.
2	Carney complex	People with <i>PRKAR1A</i> mutations have Carney complex or primary pigmented nodular adrenocortical disease (PPNAD).
3	Cancer and non-cancerous tumor risks	You have an increased chance to develop thyroid cancer or Sertoli cell testicular tumors (males* only). In addition, you have an increased chance to develop non-cancerous (benign) tumors in the skin, heart, breast, thyroid, and nervous system. Other non-cancerous signs of Carney complex can include variations in skin color and tumors in the endocrine system that can cause the body to produce excess hormones.
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>PRKAR1A</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.
6	Other related disorders	Very rarely, mutations in <i>PRKAR1A</i> can cause a severe skeletal disorder called acrodysostosis. Acrodysostosis is present from birth and causes short stature and abnormal bone formation. The mutations that cause acrodysostosis do not cause Carney complex or PPNAD.

*Refers to sex assigned at birth

PRKAR1A Mutations in the Family

There is a 50/50 random chance to pass on a *PRKAR1A* 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.



■ Has *PRKAR1A* mutation ■ No *PRKAR1A* mutation

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

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your *PRKAR1A* 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.