Ambry Genetics®
A TEMPUS COMPANY

Understanding Your Positive *PRKAR1A* Genetic Test Result

INFORMATION FOR PATIENTS WITH A PATHOGENIC OR LIKELY PATHOGENIC VARIANT

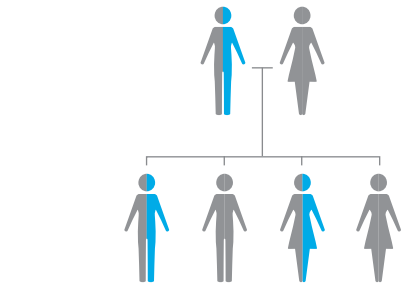
6 Things To Know

1	Result	Your testing shows that you have a pathogenic or likely pathogenic variant in the *PRKAR1A* gene.
2	Carney complex	People with a pathogenic or likely pathogenic *PRKAR1A* variant 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 pathogenic or likely pathogenic *PRKAR1A* 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.
6	Other related disorders	Very rarely, pathogenic or likely pathogenic *PRKAR1A* variants can cause a severe skeletal disorder called acrodysostosis. Acrodysostosis is present from birth and causes short stature and abnormal bone formation. The pathogenic or likely pathogenic *PRKAR1A* variants that cause acrodysostosis do not cause Carney complex or PPNAD.

*Refers to sex assigned at birth

PRKAR1A in the Family

There is a 50/50 random chance to pass on a pathogenic or likely pathogenic *PRKAR1A* variant to each of your children.



resources

- American Brain Tumor Association abta.org
- American Cancer Society cancer.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.

1 Enterprise, Aliso Viejo, CA 92656 USA Toll Free +1.866.262.7943 Fax +1.949.900.5501 **ambrygen.com**

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