Ambry Genetics®

Understanding Your *POLE* Carrier Genetic Test Result

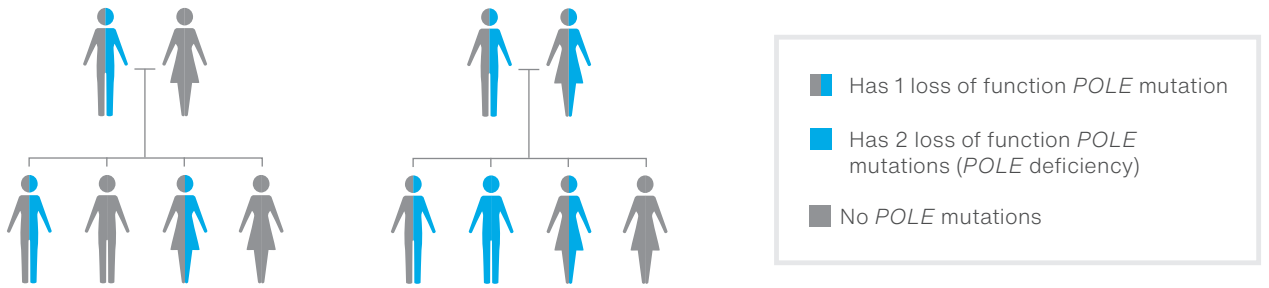
INFORMATION FOR PATIENTS WITH ONE PATHOGENIC OR LIKELY PATHOGENIC VARIANT

5 Things to know

1	Result	Your testing shows that you have a specific type of pathogenic or likely pathogenic (P/LP) variant in the *POLE* gene called a loss of function mutation.
2	Carrier	People with one loss of function mutation in the *POLE* gene are carriers of *POLE* deficiency. People with two loss of function mutations in the POLE gene have *POLE* deficiency. *POLE* deficiency is a multisystem disorder characterized by growth problems, skeletal differences, distinct facial features, a deficiency of the immune system, developmental delay, and other medical concerns. Your result shows that you do not have *POLE* deficiency, but your family members may be at risk for it.
3	Cancer risks	There is currently no evidence to suggest an increased cancer risk for carriers (people with only one loss of function *POLE* mutation) over that of the general population.
4	What you can do	Risk management decisions are very personal. It is important to discuss options with your doctor and decide on a plan that works for you.
5	Family	Family members may be at risk - they can be tested for the P/LP *POLE* variant that was identified in you, as well as other P/LP variants in the *POLE* gene. It is recommended that you share this information with family members so they can learn more and discuss with their healthcare providers.

POLE in the Family

There is a 50/50 random chance to pass on the *POLE* loss of function mutation to your children. If your partner also happens to carry one *POLE* loss of function mutation, there is a 25% chance that you will both pass on the *POLE* mutation to your child (who will have *POLE* deficiency) and a 25% chance that neither mutation would be passed on to your child.



RESOURCES

- National Society of Genetic Counselors nsgc.org
- Canadian Association 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 *POLE* 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.

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