Ambry Genetics®

Understanding Your *NTHL1* Carrier Genetic Test Result

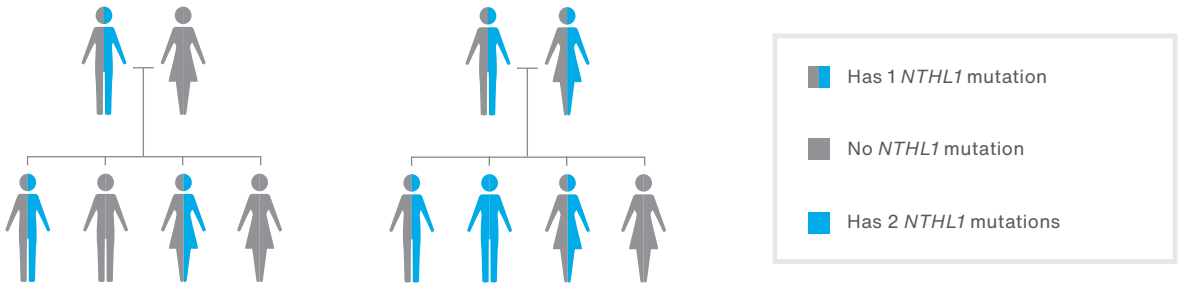
INFORMATION FOR PATIENTS WITH ONE PATHOGENIC MUTATION OR VARIANT, LIKELY PATHOGENIC

5 Things To Know

1	*NTHL1* mutation	Your testing shows that you have one pathogenic mutation or variant that is likely pathogenic in the *NTHL1* gene.
2	Carrier	People with one *NTHL1* mutation are carriers. People with two *NTHL1* mutations have an increased chance to develop colorectal polyps and colorectal cancer. Your result shows you do not have this increased risk.
3	Cancer risks	There is insufficient evidence to suggest an increased cancer risk above that of the general population for people with only one pathogenic *NTHL1* mutation.
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 also be at risk- they can be tested for the *NTHL1* mutation that was found in you, as well as other *NTHL1* mutations. It is recommended that you share this information with your family members so they can learn more and discuss with their healthcare providers.

NTHL1 Mutations in the Family

There is a 50/50 random chance to pass on the *NTHL1* mutation to your sons and daughters. If your partner happens to carry one *NTHL1* mutation, there is also a 25% chance that you will both pass on the *NTHL1* mutation to your child (who will have an increased risk for colorectal cancer), and a 25% chance that neither mutation would be passed on to your child. The images below show that both men and women can carry and pass on these mutations.



Reach Out

RESOURCES

- Ambry's Hereditary Cancer Site for Families patients.ambrygen.com/cancer
- 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 *NTHL1* 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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7 Argonaut, Aliso Viejo, CA 92656 USA Toll Free +1 866.262.7943 Fax +1 949.900.5501 ambrygen.com