Ambry Genetics®
A Konica Minolta Company

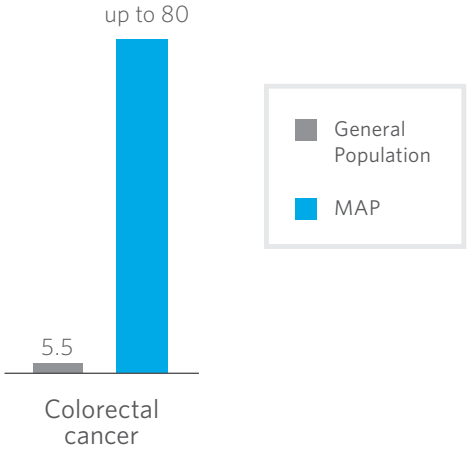
Understanding Your Positive *MUTYH* Genetic Test Result

INFORMATION FOR PATIENTS WITH **TWO PATHOGENIC MUTATIONS** OR **VARIANTS, LIKELY PATHOGENIC**

5 Things To Know

1	*MUTYH* mutation	Your testing shows that you have two pathogenic mutations or variants that are likely pathogenic in the *MUTYH* gene.
2	MUTYH-associated polyposis (MAP)	People with two *MUTYH* mutations have *MUTYH*-associated polyposis, also referred to as MAP.
3	Cancer risks and other medical concerns	You have an increased chance to develop gastrointestinal polyps, colorectal cancer, and possibly other cancers.
4	What you can do	There are risk management options to detect cancer early or lower your risk to develop cancer. It is important to discuss these options with your doctor, and decide on a plan that best manages your cancer risks.
5	Family	Family members may also be at risk – they can be tested for the *MUTYH* mutations that were identified in you.

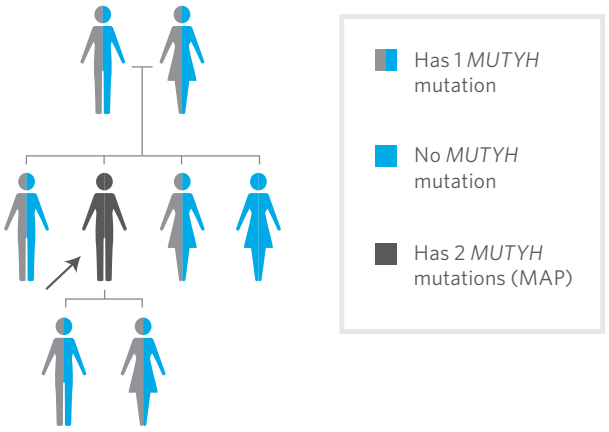
Lifetime Cancer Risks With MAP (%)*



**The above cancer risks represent the typical range for individuals with a mutation in this gene. If available, cancer risks specific to the mutation found in you will be provided in your results report.*

MUTYH Mutations in the Family

You have two *MUTYH* mutations, therefore, any children you have will inherit one of them. Your children are <u>not</u> at risk to have MAP unless your partner has at least one *MUTYH* mutation as well. Each of your parents carries at least one *MUTYH* mutation. This means your brothers and sisters have a 25% chance to have MAP, a 50% chance to inherit one *MUTYH* mutation, and a 25% chance to inherit <u>no</u> *MUTYH* mutations. The image to the right shows that both men and women can carry and pass on these mutations.



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Result	**MUTATIONS**	Your testing shows that you have two pathogenic (disease-causing) mutations (disease-causing change in the gene, like a spelling mistake) or variants that are likely disease-causing in the *MUTYH* gene, which is called a "biallelic" result. Both of these results should be considered positive.
Gene	***MUTYH***	Everyone has two copies of the *MUTYH* gene, which we randomly inherit from each of our parents. This means you inherited one of your mutations from each of your parents. Having two mutations in the *MUTYH* gene is associated with an increased lifetime chance for cancer.
Condition	**MAP**	People with two *MUTYH* mutations have *MUTYH*-associated polyposis, also referred to as MAP.
Cancer Risks and Other Medical Concerns	**INCREASED**	You have an increased chance to develop gastrointestinal polyps, colorectal cancer (up to 80% lifetime risk), and possibly cancers of the duodenum, stomach, and endometrium (uterus).
Management Options	**FOR MEN & WOMEN**	Options for early detection and prevention of cancer for women and men may include: colonoscopies, upper endoscopies, other imaging, and options for risk-reducing surgery. Talk to your doctor about what options may be right for you.
Risk Management	**VARIES**	Risk management decisions are very personal, and the best option depends on many factors. Screening typically begins earlier than in the general population and is often done more frequently. It is important to discuss these options with your doctor.
Family Members	**25-50% CHANCE**	Your parents each carry at least one *MUTYH* mutation. Since you have two *MUTYH* mutations (one from each parent), each of your children will have one of them, and are said to be carriers of MAP. If your partner happens to carry 1 *MUTYH* mutation, each of your children would have a 50% chance to have MAP (have 2 *MUTYH* mutations). Your brothers and sisters have a 25% chance to also have MAP and a 50% chance to be a carrier. Other family members (like your aunts, uncles, cousins) may also be a carriers. Talk to your healthcare provider about your family history to find out if other family members may benefit from genetic testing.
Next Steps	**DISCUSS**	It is recommended that you share this information with family members so they can learn more and discuss this with their healthcare providers.
Reach Out	**RESOURCES**	• Ambry's Hereditary Cancer Site for Families patients.ambrygen.com/cancer • Hereditary Colon Cancer Foundation hcctakesguts.org • Genetic Information Nondiscrimination Act (GINA) ginahelp.org • 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 *MUTYH* 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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