Ambry Genetics®

Clinician Management Resource for individuals with only **one** (heterozygote, carrier) likely pathogenic or pathogenic mutation in *MUTYH*

This overview of clinical management guidelines is based on this patient's test result for only one *MUTYH* gene mutation. Unless otherwise stated, medical management guidelines used here are limited to those issued by the National Comprehensive Cancer Network® (NCCN®)[1] in the U.S. Please consult the referenced guideline for complete details and further information.

Clinical correlation with the patient's past medical history, treatments, surgeries and family history may lead to changes in clinical management decisions; therefore, other management recommendations may be considered. Genetic testing results and medical society guidelines help inform medical management decisions but do not constitute formal recommendations. Discussions of medical management decisions and individualized treatment plans should be made in consultation between each patient and his or her healthcare provider, and may change over time.

SCREENING/SURGICAL CONSIDERATIONS[1]	AGE TO START	FREQUENCY
Colorectal Cancer		
Colonoscopy for probands unaffected by CRC with a first-degree relative with CRC*	Beginning at age 40 y or 10 y prior to age of first-degree relative's age at CRC diagnosis	Every 5 years
For patients with colorectal cancer, please refer to the surveillance recommendations for post-colorectal cancer resection in the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Colon Cancer and Rectal Cancer.	For probands unaffected by colorectal cancer and no family history of colorectal cancer: Data are uncertain if specialized screening is warranted	N/A

* Data to support surveillance recommendations for *MUTYH* heterozygotes are evolving at this time. Caution should be used when implementing final colonoscopy surveillance regimens in context of patient preferences and new knowledge that may emerge.

1. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Colorectal. V2.2022. © National Comprehensive Cancer Network, Inc. 2022. All rights reserved. Accessed December 20, 2022. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use or application and disclaims any responsibility for their application or use in any way

Understanding Your *MUTYH* Carrier Genetic Test Result

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

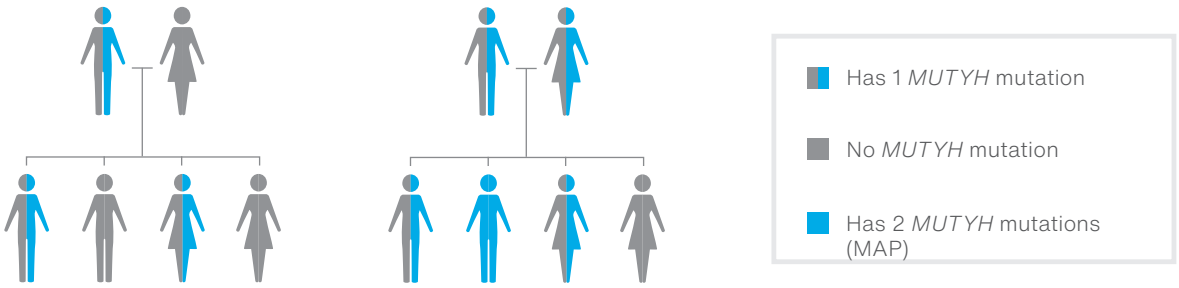
5 Things To Know

1	*MUTYH* mutation	Your testing shows that you have one pathogenic mutation or variant that is likely pathogenic in the *MUTYH* gene.
2	Carrier	People with one *MUTYH* mutation are carriers of *MUTYH*-associated polyposis, or MAP.* People with two *MUTYH* mutations have MAP. Your result shows you do not have MAP, but your family members may be at risk for it.
3	Cancer risks	You may have a slightly increased chance to develop colorectal cancer.
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 *MUTYH* mutation that was identified in you, as well as other mutations in the *MUTYH* gene. It is recommended that you share this information with family members so they can learn more and discuss this with their healthcare providers.

** MUTYH-associated polyposis (MAP) is caused by a person having two MUTYH mutations. This condition is associated with a higher risk for gastrointestinal polyps, colorectal cancer, and possibly cancers of the duodenum and stomach.*

MUTYH Mutations in the Family

There is a 50/50 random chance to pass on the *MUTYH* mutation to your sons and daughters. If your partner happens to carry one *MUTYH* mutation, there is also a 25% chance that you will both pass on the *MUTYH* mutation to your child (who will have MAP), 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
- Hereditary Colon Cancer Foundation hcctakesguts.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 *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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