Genetic Insights: quick reference guide for healthcare providers

This guide is intended to facilitate a discussion between a provider and their patient.

Genetic Insights test results: ***MUTYH*-associated polyposis carrier**

Key results

A pathogenic or likely pathogenic variant associated with being a carrier of *MUTYH*-associated polyposis (MAP) was found in the *MUTYH* gene.

Carriers of MAP do not have the condition but are at higher risk of having children with the condition if the other parent is also a carrier. Carriers of MAP may have a slightly increased chance of developing colon cancer.

Next steps

Clinical recommendations	Resources
Genetic Insights is a screening test and not intended for diagnosis. A follow-up genetic test should be performed in a clinical setting before any other action is taken.	**Ready to order?** Check with your institution and/or patient's insurance about the preferred testing laboratory. Blueprint Genetics® offers MAP testing. To confirm this test result, **targeted variant testing** for the variant identified is available. You can order a confirmation test here: Blueprint Genetics/TVT **Have questions?** Call **1.866.GENE.INFO** (1.866.436.3463) to speak to a specialized Quest genetic counselor or geneticist available to healthcare providers to discuss test selection and results.
Refer your patient to a genetic counselor. *Genetic counselors can provide counseling on the implications of this test result and next steps for your patient.*	Your patient can schedule a 1-on-1 remote genetic counseling session through their online Genetic Insights Carrier Risk Report at no additional cost. To find a genetic counselor with expertise in hereditary cancer genetics practicing in your patient's area for an in-person session, please visit FindAGeneticCounselor.NSGC.org

Patient conversation starters:

Patient conversation starters summarize the preceding information in plain language to support meaningful conversations between you and your patient.

FindAGeneticCounselor.NSGC.org

Your Genetic Insights test is a screening test. The next step is to have your result confirmed with a second genetic test.

It's also important that you talk with a genetic counselor. Genetic counselors are experts in genetics and can help you understand this result and potential next steps.

You can access a genetic counselor through your online Genetic Insights dashboard at no additional cost to you, and we can discuss a referral to a local genetic counselor.

Visit QuestDiagnostics.com/Genetic-Health-Screening for more information about this test.

What is MAP?

MAP is a hereditary cancer predisposition syndrome characterized by the presence of colon polyps and a significantly increased chance of colon cancer.[1,2] Risks for duodenal, bladder, and ovarian cancer may also be increased.[1]

MAP results from 2 DNA variants associated with MAP, 1 in each copy of the *MUTYH* gene.[1] People with a DNA variant associated with MAP in only 1 copy of *MUTYH* are considered carriers of MAP. Carriers may have a slightly increased chance of developing colon cancer in their lifetime.[1,2]

Depending on their family history, people who are MAP carriers may be recommended to undergo more frequent colon cancer screening, typically starting at earlier ages than in the general population.[2]

See the Management options section for more detail.

Patient conversation starters:

MAP is a genetic condition that significantly increases the chance of developing colon cancer and certain other cancers.

People with one DNA variant in the *MUTYH* gene are considered carriers of MAP. Carriers do not have MAP, but their biological children could have MAP if their other biological parent (or sperm/egg donor) is also a MAP carrier.

People who are MAP carriers may have a slightly increased chance of developing colon cancer.

Especially if they have a family history of colon cancer, MAP carriers may be recommended to undergo colon cancer screening earlier or more often than typical.

What this result means for family members

MAP is an inherited condition. If both biological parents are carriers, each child has a 1 in 4 chance of having MAP.

If only 1 parent is a carrier, each child has a 1 in 2 chance (50%) of being a carrier and a 1 in 2 chance (50%) that they will not be a carrier.

Parents and siblings of MAP carriers may also be carriers.

In people with a confirmed *MUTYH* DNA variant, genetic testing for family members may help inform their risks and screening protocols.[1,2]

MAP carriers should discuss their carrier status with their reproductive partners, especially if planning to have children.

- Comprehensive *MUTYH* gene analysis for reproductive partners (or sperm/egg donor) or other family members may be warranted, especially for family planning purposes[2]

A genetic counselor can help determine the most appropriate testing options. Therefore, it is strongly recommended that people share their results with their biological relatives.

Patient conversation starters:

MAP runs in families.

That means the DNA variants linked to MAP can be passed down from parents to their children.

One of your biological parents is likely a MAP carrier. Your full siblings have a 1 in 2 (50%) chance of also being a MAP carrier. Your biological children's chance of being a MAP carrier or having MAP depends on if their other parent is a carrier.

Sharing these results with your family members is important they can decide if they want to have genetic testing.

Cancer risk

A person with a single confirmed variant in the *MUTYH* gene is a carrier of MAP. Individual cancer risks may be higher or lower depending on personal health history and family health history.[2]

Cancer type	Approximate lifetime risk in MAP carriers	Approximate lifetime risk in the general population
Colon	8%-12%[1]	4%[3]

Colon cancer risks may be higher for MAP carriers who have a first-degree relative with colon cancer. Associated cancers and risks may change over time as medical research advances.

Management options

There are options for cancer prevention and early detection for people who are MAP carriers. Clinical guidelines from the National Comprehensive Cancer Network® (NCCN®) for adults with this variant and no personal history of an associated cancer include[2]:

Cancer	Family history of colon cancer	Guidelines for people who are carriers of MAP
Colon	First-degree relative with colon cancer	Colonoscopy every 5 years beginning at age 40 or 10 years prior to age of first-degree relative's diagnosis of colon cancer
	Second-degree relative with colon cancer	There are no specific data available to determine screening recommendations
	No family history of colon cancer	Data are unclear as to whether specialized screening is warranted

Patient conversation starters:

People who are MAP carriers have a slightly higher risk of developing colon cancer than most people. People who are MAP carriers may need to have colon cancer screening earlier or more often than typical. This is especially true if there is a family history of a close relative with colon cancer.

If your result is confirmed, it's important to work with the right specialists, like a medical oncologist and a geneticist, to find cancer screening and risk-reducing options that are right for you.

See NCCN for complete recommendations. Recommendations may change over time.
If the test result is confirmed, local centers for excellence in hereditary cancer may be considered for further clinical management.

Additional resources

The following advocacy groups have additional information and resources about MAP:

Colorectal Cancer Alliance: CCAlliance.org
Fight Colorectal Cancer: FightColorectalCancer.org
FORCE: Facing Our Risk of Cancer Empowered: FacingOurRisk.org

References

1. Nielsen M, Infante E, and Brand R. MUTYH Polyposis. October 4, 2012. Updated May 27, 2021. In: Adam MP, Ardinger HH, Pagon RA, et al, editors. GeneReviews® [Internet]. University of Washington, Seattle; 1993-2022. www.ncbi.nlm.nih.gov/books/NBK1266
2. National Comprehensive Cancer Network®. Genetic/Familial High-Risk Assessment: Colorectal (Version 2.2022). NCCN Guidelines®. Accessed December 13, 2022. www.nccn.org
3. National Cancer Institute: Surveillance, Epidemiology, and End Results Program. Cancer Stat Facts: Colorectal Cancer. Accessed October 31, 2022. https://seer.cancer.gov/statfacts/html/colorect.html