Risk of Polyposis, Colon Cancer, and Other Cancers

INFORMATION ABOUT YOUR GENETIC TEST RESULT

Your Result

Positive for a known pathogenic or likely pathogenic variant in 1 or both copies of the *MUTYH* gene

What This Result Means

This result means you have a change (mutation) in 1 or 2 copies of the *MUTYH* gene. Everyone has 2 copies of the gene. You inherit one copy from your mother and one copy from your father. Presence of a mutation in both copies can cause *MUTYH*-associated polyposis (MAP). People with MAP have benign growths called polyps in the colon and the upper part of the small bowel. These polyps can become cancerous if not treated.

People with a mutation in only 1 copy of the *MUTYH* gene do not have MAP. But they still have a small increase in risk for cancer. The risk for cancer is listed in the table below.

This result doesn't mean you will get cancer—not everyone with a mutation develops cancer. But you are at higher risk than you would be if you didn't have the mutation.

Type of Cancer	Risk of Cancer		
	***MUTYH* Mutation in 1 Copy**	***MUTYH* Mutation in 2 Copies**	**Without an *MUTYH* Mutation**
Colon	9%	Up to 80%	4.5%
Duodenum	No increase	5 – 17%	0.2%

Options for Managing Cancer Risk in People with MAP

Your test result shows that you have an increased risk for developing cancer. There are steps you can take to lower that risk. The options depend on how many copies of the mutation you have. They also depend on your personal and family history of cancer. If you have 1 copy, your doctor might have you follow the standard guidelines for colon cancer screening. If you have 2 copies, you have the options described below. Talk with your healthcare provider to learn more and find out which is best for you.

Increased Cancer Screening

- Colonoscopy beginning at age 25-30 years; repeat frequency based on number of polyps found
- Upper endoscopy beginning at age 30-35; repeat frequency based on number of polyps found

Increased screening cannot prevent you from getting cancer. But it could detect cancer sooner. And, as you know, early detection improves your chances of survival.

Surgery

- Preventive removal of the colon when there are too many polys to remove

Surgery may be an option for lowering the risk of cancer. But surgery is not for everyone.

Medication (Chemoprevention)

Certain medicines might help prevent or reduce polyps in some people. Talk to your doctor to find out if this is an option for you.

What This Result Means for Your Family

Some of your family members may have the same mutation that you have. Your brothers and sisters each have a 25% chance of having a mutation in 2 copies of the gene. So they have a 25% chance of having MAP. Other blood relatives also have an increased risk. Your children are at risk for MAP only if your partner also has a mutation in the *MUTYH* gene.

Think about sharing your test results with your relatives. They might want to find out if they have the mutation too. If so, they should talk with their doctor and/or genetic counselor about testing. They could be tested just for the mutation that you have. Testing for a single mutation may be the best option, as it may cost less. It will still let them know if they have the same mutation you have. In some cases, testing more genes might be suggested. Either way, testing will help them know more about their risk for cancer.

Your Next Steps

- ☐ Get a copy of your test results.
- ☐ Talk with your doctor or genetic counselor about things you can do to manage your risk.
- ☐ Once you and your doctor have made a plan, set up appointments to start managing your risk.
- ☐ Think about sharing copies of your test results with your family members. That way, their doctor will know which test to order if they want to be tested.
- ☐ Talk with your doctor or genetic counselor regularly. They can keep you up to date about genetic testing and risk management options.

You can help researchers learn even more about this gene and its link to cancer risk. You can do this by participating in a research registry called PROMPT. If you would like to do this, please visit **PROMPTStudy.org** to learn more.

Additional Resources

- **Hereditary Colon Cancer Takes Guts**
  HCCTakesGuts.org
- **National Society of Genetic Counselors**
  Find a Genetic Counselor
  NSGC.org/p/cm/ld/fid=164

This information is not a substitute for medical advice, diagnosis, or treatment. The diagnosis or treatment of any disease or condition may be based on personal history, family history, symptoms, a physical examination, laboratory test results, and other information considered important by your doctor. Always talk with your doctor about the meaning of your test results and before you stop, start, or change any medication or treatment.

The classification and interpretation of the variant(s) identified reflect the current state of Quest Diagnostics' understanding at the time of the accompanying report. Variant classification and interpretation are subject to professional judgment, and may change for a variety of reasons, including but not limited to, updates in classification guidelines and availability of additional scientific and clinical information. This test result should be used in conjunction with the health care provider's clinical evaluation. Inquiry regarding potential changes to the classification of the variant is strongly recommended prior to making any clinical decision. For questions regarding variant classification updates, please call Quest Diagnostics at 866.GENE.INFO (436.3463) to speak to a genetic counselor or laboratory director.