Risk of Colorectal Cancer, Polyposis, and Other Cancers

INFORMATION ABOUT YOUR GENETIC TEST RESULT

Your Result

Positive for a known pathogenic or likely pathogenic variant in the *POLD1* and/or *POLE* gene

What This Result Means

This result means you have a change (mutation) in the *POLD1* or *POLE* gene. Mutations in either of these genes can increase your chance of benign growths called polyps. These polyps can lead to colon cancer if not removed. There may also be an increased risk for other cancers.

Rarely, other conditions have been linked to mutations in the *POLD1* gene, too.

At this time, experts don't know a lot about the risk of cancer linked to mutations in this gene. Further research is needed to learn more. New studies will tell us how high the risk for cancer is and if there is a risk for other cancers, too. Mutations in other genes may increase the risk for cancer in you or your family.

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.

Options for Managing Your Cancer Risk

Your test result shows that you may have an increased risk for developing cancer. It's important for you to take steps to lower that risk. The options are explained briefly below. Although these options are listed in the guidelines, they may change over time. And they may not be the best options for all patients. Talk with your healthcare provider to learn more and find out which is best for you.

Increased Cancer Screening

- Colonoscopy every 1 to 3 years

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 and/or rectum if screening shows a large number of polyps

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

What This Result Means for Your Family

You share genes in common with your blood relatives, so some of them may have the same mutation that you have. Your parents, brothers, sisters, and children each have a 50% chance of having it. Other blood relatives also have an increased risk.

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. Because this type

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What This Result Means for Your Family (continued)

of testing is targeted, it is a less expensive test. It will 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.

Relative	Risk of Having the Same Mutation
Identical twin	100%
Fraternal twin	50%
Parent, brother, sister, child	50%
Grandparent, uncle, aunt, niece, nephew	25%
First cousin	12.5%

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.