Risk of Colorectal, Stomach, and Other Cancers

INFORMATION ABOUT YOUR GENETIC TEST RESULT

Your Result

Positive for a known pathogenic or likely pathogenic variant in the *BMPR1A or SMAD4* gene

What This Result Means

This result means you have a change (mutation) in the *BMPR1A* or *SMAD4* gene. Mutations in these genes can cause juvenile polyposis syndrome (JPS). People with JPS have an increased chance of having benign polyps. These polyps might develop into cancer of the colon, rectum, or stomach if not treated. The risk for these cancers is shown in the table below. People with JPS might have other types of cancer too. These include cancer in the small intestine and pancreas. A small number of people with JPS may have abnormalities in the intestines, heart, brain, mouth, or fingers.

Some people with a *SMAD4* mutation may have hereditary hemorrhagic telangiectasia (HHT). People with HHT have problems with blood vessels in the skin, lungs, and some organs. HHT is a benign condition.

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		
	With a** BMPR1A **Mutation	***With a** SMAD4 **Mutation***	***Without a** BMPR1A or SMAD4 **Mutation***
Colorectal	38 – 68%	38 – 68%	4.5%
Stomach	21%	21 – 30%	<1%

Options for Managing Your Cancer Risk

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 are explained briefly below. Talk with your healthcare provider to learn more and find out which is best for you.

Increased Cancer Screening

- Regular colonoscopy and upper endoscopy starting around age 15 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.

Options for Managing Hereditary Hemorrhagic Telangiectasia Risk

Babies with a *SMAD4* mutation can be screened for HHT starting before 6 months of age.

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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. 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.

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.