Risk of Colorectal, Uterine, and Other Cancers

INFORMATION ABOUT YOUR GENETIC TEST RESULT

Your Result

Positive for a known pathogenic or likely pathogenic variant in the *MLH1, MSH2, MSH6, PMS2, or EPCAM* gene

What This Result Means

This result means you have a change (mutation) in one of the genes listed above. Mutations in these genes can cause Lynch syndrome. People with Lynch syndrome have an increased chance of developing certain cancers. Women have a higher risk for cancer in the uterus and ovary. Both women and men have a higher risk for cancer in the colon, rectum, stomach, small bowel, and other sites. The risk for these cancers is shown 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			
	With an *MLH1, MSH2, or EPCAM* Mutation	With an *MSH6* Mutation	With a *PMS2* Mutation	Without a Lynch Syndrome Mutation
Women				
Uterus	12 – 54%	16 – 71%	15 – 24%	3%
Ovary	3 – 24%	Increased	Increased	1%
Women and Men				
Colorectal	33 – 78%	10 – 69%	10 – 20%	4 – 5%
Stomach	2 – 18%	2 – 10%	Increased	1%
Pancreatic	1 – 6%	Increased	Increased	<1%
Other cancers	1 – 9%	Increased	Increased	<3%

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

- Colonoscopy, generally starting at age 20-25; repeat every 1-2 years
- Annual urinalysis starting at age 25-30
- Annual physical exam for central nervous system cancers starting at age 25-30

There may be other options for certain people. Women should see their doctor right away if they have abnormal vaginal bleeding or pelvic pain.

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 hysterectomy (removal of uterus)

(Turn page for more options.)

Quest Diagnostics™

• Preventive bilateral salpingo-oophorectomy (removal of ovaries and fallopian tubes)

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. 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.

If your sex partner also has a mutation in one of these genes, your child is at risk for constitutional mismatch repair deficiency (CMMRD) syndrome. Talk to your doctor and/or genetic counselor if you are thinking about having a baby.

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
- **Lynch Syndrome International**
  LynchCancers.com
- **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.