



HEREDITARY COLON CANCER

Should you have genetic testing?

You or someone in your family has had colon or rectal cancer. It is normal to wonder why it happened. In most cases, there is no clear explanation. However, for some individuals with colon cancer, one critical gene is not working in the usual way. That small difference makes it more likely for cancer to develop.

Does my family history of cancer mean that I have a mutation?

Not necessarily. Only about 5-10% of colon and rectal cancers are related to inherited mutations in a cancer risk gene. Most colon and rectal cancers are caused by other factors and not related to a mutation in one of these genes.

Could I carry a mutation?

Mutations in cancer risk genes have been found in families worldwide. There are certain risk factors that help identify who might have a mutation.

Risk factors for hereditary colon cancer
You might have a mutation in a cancer risk gene if you answer "YES" to any of these statements:

- I had colon or rectal cancer before age 55
- I had tumor testing suggestive of Lynch syndrome (a hereditary colon cancer condition)
- I have had more than 10 colon polyps
- I have 3 or more close relatives with cancer (including: colorectal, uterine, stomach, ovarian, urinary tract)
- I had colorectal cancer **AND** one of the following cancers: uterine, stomach, ovarian, urinary tract
- I have a relative with a hereditary colon cancer condition

What if I do not have any risk factors?

Testing is most useful for individuals who are at high risk. If your personal or family history of cancer does not show any special risk factors, then you do not need to have genetic counseling or consider genetic testing. However, you should still follow your medical provider's advice for colon cancer screening.

What are genes?

Genes are the instructions that guide your growth and development and help control how your body works. Your genes are contained in each cell of your body. You are born with two copies of each gene - one inherited from your mother and the other inherited from your father.

What are cancer risk genes?

There are many genes that help control cell growth in your body. When one of these genes is not working normally, it raises the chance for cancer to develop. These genes are sometimes called cancer risk genes. Like other types of genes, you have two copies of each cancer risk gene - one copy from each parent.

How do cancer risk genes cause cancer?

In some families there is a genetic change (mutation) in one of the cancer risk genes that affects the way that gene works. A mutation can be inherited from either parent. When a cancer risk gene is not working in the usual way, it can allow cells to grow too much, sometimes resulting in cancer. A mutation in one of these genes makes cancer more likely to develop, but it does not **cause** cancer. Some people who inherit a mutation never get cancer.

SCREENING FOR COLON CANCER

Screening for colon cancer usually includes at least one of the following tests:

- Fecal occult blood test (FOBT) or Fecal immunochemical test (FIT)
- Flexible sigmoidoscopy
- Colonoscopy

Screening recommendations are based on your medical history, the history of cancer in your family, and your age. Talk with your medical provider about when to start screening, which test is right for you, and how often screening should be repeated.

What if I have one or more risk factors?

Talk with your provider about your concerns. After reviewing your family history and your personal health history, your provider may refer you to a genetic counselor. The genetic counselor will review your medical records, your health history and your family history of cancer.

A genetic counselor can help:

- Estimate the chance for developing cancer based on your medical and family history
- Determine if your history suggests a hereditary colon cancer condition
- Estimate the chance that you may have a mutation in a cancer risk gene
- Guide you through decisions about genetic testing and any results you receive

How is a mutation identified?

A blood test is available to look for mutations in some cancer risk genes, but testing is not for everyone. Testing is most useful when your personal and family history of cancer shows a pattern of an inherited risk to cancer. Your genetic counselor will help determine whether or not testing is indicated for your family and which test, if any, is right for you.

What does it mean if the test is positive?

A positive result means a mutation has been found in one of the cancer risk genes. A person with a mutation has a much higher chance of developing colon or rectal cancer during their lifetime. There is also a higher risk for other cancers in the body. The

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actual cancer risk depends on which gene has a mutation. Not everyone with a gene mutation will develop cancer.

What are some benefits and limits of genetic testing?

Benefits

Positive test results:

- Can help with decisions about medical care and personal choices.
- Cancer screening can start at a younger age and be done more often.
- Risk-lowering surgeries may be offered before cancer develops.
- Testing may be done for your family members.

Negative test results:

- Can let you know you did not inherit the family mutation (when the mutation is already known)

Limits

Positive test results:

- Do not tell you when or if you will develop cancer.
- Can cause stress, especially if no plan of action is in place to deal with the results.

Negative test results:

- May give you the feeling of having a lower risk for cancer than your actual risk.
- May give you a feeling of “survivor’s guilt”, when others in the family have a mutation.

Should I have genetic testing?

Not every family needs genetic testing. If genetic testing is offered, it is important to explore what testing means for you and your family before any testing is started. Testing for a cancer risk gene is very personal. Test results can have complex, and sometimes unexpected, emotional impact. Sometimes, results lead to difficult medical decisions. You will want to think carefully about how genetic testing results will help you.