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.

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 susceptibility 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 susceptibility genes. Like other types of genes, you have two copies of each cancer susceptibility gene - one copy from each parent.

How do cancer susceptibility genes cause cancer?
In some families, there is a mutation (genetic change) in one of the cancer susceptibility genes that affects the way the gene works. A mutation can be inherited from either parent. When a cancer susceptibility 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 it more likely to develop cancer, but it does not cause cancer. Some people who inherit a mutation never get cancer.

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 susceptibility gene. Most colon and rectal cancers are caused by other factors and not related to an inherited mutation in one of these genes.

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

Risk factors for hereditary colon cancer
You have a higher chance of carrying a mutation in a cancer susceptibility gene if you answer “YES” to one or more of the following statements:

- I have had colon or rectal cancer before age 50
- I have had tumor testing suggestive of Lynch syndrome (a hereditary cancer susceptibility syndrome)
- I have had more than 10 colon polyps
- I have 3 or more close relatives with cancer (colorectal, uterine, stomach, ovarian, urinary tract)
- I have had colorectal cancer AND one of the following cancers: uterine, stomach, ovarian, urinary tract
- I have a relative with a hereditary colon cancer syndrome

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.
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 inherited a gene mutation
- 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 susceptibility 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 susceptibility to cancer. Your genetics consultation will help determine whether or not testing is indicated for your family and which test, if any, is best based on your history.

What does it mean if the test is positive?
A positive test means a mutation has been found in one of the cancer susceptibility 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 actual cancer risk depends on which gene has a mutation. But, 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 clear plan of action is in place to deal with the results.

Negative test results:
- May give you false reassurance about your actual cancer risk.
- Can lead to 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 susceptibility 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.