Genetic Testing, Screening, and Prevention for People with a Strong Family History of Colorectal Cancer

If you have a family history of colorectal polyps or cancer, you have a higher risk of getting colorectal cancer yourself. The risk can be even higher in people with a strong family history of colorectal cancer.

Is genetic testing right for you?

Whether genetic testing is right for you will depend on your family history of colorectal cancer. Cancer in close (first-degree) relatives such as parents, brothers, and sisters is most concerning, but cancer in more distant relatives can also be important. Having 2 or more relatives with colorectal cancer is more concerning than having only 1 relative with it. It's also more concerning if your relatives were diagnosed with cancer at a younger age than usual.

If you have a family history of colorectal cancer, talk with your doctor. You might benefit from speaking with a genetic counselor or other health professional who is trained in genetic counseling. They can review your family history to see how likely it is that you have a family cancer syndrome. The counselor can also help you decide if genetic testing is right for you. If you have testing and are found to have an abnormal gene, there might be steps you can take to help lower your risk of colorectal cancer, such as starting routine screening at an earlier age or even having surgery.

But before getting genetic testing, it's important to know ahead of time what the results might or might not tell you about your risk. Genetic testing is not perfect. The tests might not provide clear answers for some people. This is why meeting with a genetic counselor or cancer genetics professional is important before deciding to be tested. To learn more about this, see Understanding Genetic Testing for Cancer and What Happens During Genetic Testing for Cancer?

Genetic tests can help show if members of certain families have inherited a high risk of colorectal cancer due to inherited cancer syndromes, such as **Lynch syndrome** (also known as **hereditary non-polyposis colorectal cancer,** or **HNPCC**) or **familial adenomatous polyposis (FAP).**

In families known to have one of these inherited syndromes, family members who decide not to get tested are still usually advised to start routine screening for colorectal cancer at an early age, and to get screened more often.

Testing for Lynch syndrome (hereditary non-polyposis colorectal cancer, or HNPCC)

Lynch syndrome can greatly increase a person's risk for colorectal cancer. The lifetime risk of colorectal cancer in people with this condition can range from about 10% to about 80%, depending on which gene mutation is causing the syndrome.

People with Lynch syndrome are also at increased risk for some other cancers, such as cancers of the uterus (endometrium), ovaries, stomach, small bowel, pancreas, kidneys, brain, ureters (tubes that carry urine from the kidneys to the bladder), and bile duct.

Who should be tested for Lynch syndrome?

There are two sets of guidelines that doctors often use to determine who might be likely to benefit from genetic counseling or testing: the Amsterdam criteria (based on family history) and the revised Bethesda guidelines (for people diagnosed with colorectal cancer).

Amsterdam criteria

Doctors have found that many families with Lynch syndrome tend to have certain characteristics, which are known as the **Amsterdam criteria:**

At least 3 relatives have a cancer linked with Lynch syndrome and:

- One is a first-degree relative (parent, brother or sister, or child) of the other 2 relatives.
- At least 2 successive generations are affected.
- At least 1 relative had their cancer when they were younger than age 50.

If all of these apply to your family, then you might want to seek genetic counseling. But even if your family history satisfies the Amsterdam criteria, it doesn't always mean you have Lynch syndrome. And many families with Lynch syndrome do not meet the Amsterdam criteria.

Revised Bethesda guidelines

A second set of criteria, called the **revised Bethesda guidelines,** can be used to help decide whether a person with colorectal cancer should be tested for genetic changes that are seen with Lynch syndrome. (These changes are called **microsatellite instability or MSI.**) These criteria include at least one of the following:

- The person is younger than age 50 when diagnosed with colorectal or uterine cancer.
- The person has or had a second colorectal cancer or another cancer (endometrial, stomach, pancreas, small intestine, ovary, kidney, brain, ureters, or bile duct) linked to Lynch syndrome.
- The person is younger than age 60, and the cancer has certain characteristics seen with Lynch syndrome when it's viewed under a microscope.
- The person has a first-degree relative (parent, sibling, or child) younger than age 50 who was diagnosed with colorectal cancer or another cancer linked to Lynch syndrome.
- The person has 2 or more first- or second-degree relatives (aunts, uncles, nieces, nephews, or grandparents) who had colorectal cancer or another Lynch syndrome-related cancer at any age.

If a person with colorectal cancer has any of the Bethesda criteria, testing for MSI may be advised. If MSI is found, the doctor typically will recommend that the patient be tested for Lynch syndrome-associated gene mutations.

It's important to know that most people who meet the Bethesda criteria do not have Lynch syndrome, and that you can have Lynch syndrome and not meet any of the criteria listed. Not all doctors use the Bethesda guidelines to decide who should have MSI testing. In fact, it is now recommended that all colorectal cancers be tested for MMR deficiency or MSI. For anyone whose cancer has a high MSI level or is missing (deficient in) one of the MMR proteins, doctors will most likely recommend genetic testing for Lynch syndrome.

Even if you don't have cancer, your doctor may suspect that Lynch syndrome runs in your family based on cases of colorectal cancer and other cancers associated with this syndrome in your relatives. In that case, your doctor might recommend genetic counseling to evaluate your risk.

those who have not been tested should start colonoscopy screening during their early 20s, or 2 to 5 years younger than the youngest person in the family with a diagnosis (whichever is earlier). Testing should be done every 1 or 2 years. This way polyps can be found and removed, and any cancers can be found at the earliest possible stage. (See American Cancer Society Guideline for Colorectal Cancer Screening.)

Testing for familial adenomatous polyposis (FAP)

Familial adenomatous polyposis (FAP) typically causes hundreds of polyps in the colon and rectum, which over time leads to colorectal cancer. For people with FAP, the lifetime risk of developing colorectal cancer is nearly 100%, and in most cases, it develops before age 50. Because FAP causes polyps and cancer earlier than the usual age colorectal cancer screening is started, it sometimes isn't diagnosed until someone already has cancer.

Genetic counseling and testing are available for people who may have FAP based on their personal or family history. If changes in the gene that causes FAP are found in one person, doctors will recommend that their close relatives (brothers, sisters, and children) be tested. FAP may also be suspected if a person is found to have many polyps during a colonoscopy that was done because of problems like rectal bleeding or anemia.

People who test positive for the gene change linked to FAP should start being screened with colonoscopy at ages 10 to 15. (See American Cancer Society Recommendations for Colorectal Cancer Early Detection.) Many doctors recommend that people with FAP have their colon removed when they're in their 20s to prevent cancer from developing.

Testing for other inherited cancer syndromes

Certain other inherited syndromes, such as MUTYH-associated polyposis and Peutz-Jeghers syndrome, can also greatly increase a person's risk of colorectal cancer. If you have certain criteria that suggest you might have one of the syndromes, your doctor might recommend genetic counseling and testing to look for the gene changes that cause them.

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