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## Genetic Testing for Cancer Risk

Genetic testing can be useful for some people when certain types of cancer seem to run in their families. It can also be helpful for certain people with cancer who don't have a family history of cancer. But genetic testing isn't recommended for everyone. Here we offer basic information to help you understand what genetic testing is and how it is used for people and families concerned about their cancer risk.

- [Understanding Genetic Testing for Cancer Risk](#)
- [What Should I Know Before Getting Genetic Testing?](#)
- [What Happens During Genetic Testing for Cancer Risk?](#)

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## Understanding Genetic Testing for Cancer Risk

For certain types of cancer, genetic tests are available to help you learn about your risk. Before deciding to get genetic testing, it's important to understand what these tests look for and what they can and cannot tell you about your cancer risk.

- [What is genetic testing?](#)
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- [Who might benefit from genetic testing?](#)
- [What is genetic counseling?](#)
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## What is genetic testing?

Genetic testing is the use of medical tests to look for certain mutations (changes) in a person's genes. Many types of genetic tests are used today, and more are being developed.

Genetic testing can be used in many ways, but here we'll focus on how it is used to look for gene changes that are linked to cancer. (To learn about the role of genes and how mutations can lead to cancer, see [Genes and Cancer](#)<sup>1</sup>.)

## Genetic testing to help evaluate cancer risk

**Predictive genetic testing** is a type of testing used to look for inherited gene mutations that might put a person at higher risk of getting certain kinds of cancer. This type of testing might be suggested for:

- **A person with a strong family history of certain types of cancer**, to see if they carry a gene mutation that increases their risk. If they do have an inherited mutation, they might want to have screening tests to look for cancer early, or even take steps to try to lower their risk. An example is testing for changes in the *BRCA1* and *BRCA2* genes (which are known to increase the risk of [breast cancer](#)<sup>2</sup> and some other cancers) in people with several family members who have had breast cancer.
- **A person already diagnosed with cancer**, especially if there are other factors to suggest the cancer might have been caused by an inherited mutation (such as a strong family history, if the cancer was diagnosed at a young age, or if the cancer is uncommon, such as breast cancer in a man). Genetic testing might show if the person has a higher risk of developing some other cancers. It can also help other family members decide if they want to be tested for the mutation.
- **Family members of a person known to have an inherited gene mutation that increases their risk of cancer**. Testing can help them know if they need screening tests to look for cancer early, or if they should take steps to try to lower their risk.

Most people (even people with cancer) do not need this type of genetic testing. It's usually done when family history suggests that a cancer may be inherited (see below)

or if cancer is diagnosed at an uncommonly young age.

## Who might benefit from genetic testing?

Genetic counseling and testing may be recommended for people who have had certain cancers or certain patterns of cancer in their family. If you have any of the following, you might consider talking to a genetic counselor about genetic testing:

- Several first-degree relatives (mother, father, sisters, brothers, children) with cancer
- Many relatives on one side of the family who have had the same type of cancer
- A cluster of cancers in your family that are known to be linked to a single gene mutation (such as [breast](#)<sup>3</sup>, [ovarian](#)<sup>4</sup>, and [pancreatic cancers](#),<sup>5</sup> which are sometimes linked to *BRCA* gene mutations)
- A family member with more than 1 type of cancer
- Family members who had cancer at a younger age than normal for that type of cancer
- Close relatives with cancers that are linked to rare hereditary cancer syndromes
- A rare cancer (in you or a family member), such as breast cancer in a man or retinoblastoma
- A particular race or ethnicity (such as Ashkenazi Jewish ancestry, which is linked to a higher risk of *BRCA* gene mutations)
- A physical finding that's linked to an inherited cancer (such as having many colon polyps)
- A known genetic mutation in one or more family members who have already had genetic testing
- Lab tests of your cancer cells that show features that might be linked to an inherited gene mutation

If you are concerned about a pattern of cancer in your family, cancer you've had in the past, or other cancer risk factors, you may want to talk to a health care provider about whether genetic counseling and testing might be a good option for you.

You need to know your family history and what kinds of tests are available. For some types of cancer, no known mutations have been linked to an increased risk.

For more information on the types of cancer that may be linked to inherited genes, see [Family Cancer Syndromes](#)<sup>6</sup>.



To learn more, see [What Should I Know Before Getting Genetic Testing?](#)

## Testing cancer cells for gene changes

Sometimes after a person has been diagnosed with cancer, the doctor will order tests on a sample of cancer cells to look for certain gene or protein changes. These tests can sometimes give information on a person's outlook (prognosis), and they might also help tell if certain types of treatment may be useful.

**These types of tests look for acquired gene changes *only* in the cancer cells. These tests are not the same as the tests used to find out about inherited cancer risk.**

For more about this kind of testing and its use in cancer treatment, see [Biomarker Tests and Cancer Treatment](#)<sup>8</sup>.

expect from your test results.

## Hyperlinks

1. [www.cancer.org/cancer/understanding-cancer/genes-and-cancer.html](http://www.cancer.org/cancer/understanding-cancer/genes-and-cancer.html)
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National Cancer Institute. Genetic Testing for Inherited Cancer Susceptibility Syndromes. 2019. Accessed at [www.cancer.gov/about-cancer/causes-prevention/genetics/genetic-testing-fact-sheet](http://www.cancer.gov/about-cancer/causes-prevention/genetics/genetic-testing-fact-sheet) on January 26, 2022.

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U.S. Food & Drug Administration. Direct-to-Consumer Tests. 2019. Accessed at <https://www.fda.gov/medical-devices/in-vitro-diagnostics/direct-consumer-tests> on July 26, 2022.

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## What Should I Know Before Getting Genetic Testing?

Genetic testing can be used to learn about your risk of developing cancer and to find out if family members might be at risk. But there are many things to think about **before** you do it. If you have any factors that suggest you might benefit from genetic testing (such as certain cancers or patterns of cancer in your family), talk with your health care provider and plan to meet with a genetic counselor so you know what to expect.

- [What might I learn from genetic testing?](#)
- [How might the results affect my family?](#)
- [Will testing lead to more medical tests?](#)
- [Is genetic testing covered by insurance?](#)
- [Could the test results lead to discrimination?](#)
- [What about other privacy issues?](#)

Here are some of the things you should discuss and think about before testing.

### What might I learn from genetic testing?

Whether genetic testing would be worthwhile for you depends on what you hope to learn and what the test might be able to show. The obvious benefit of genetic testing is





be found to have a certain gene variant, but it might not be clear if this particular variant actually raises your risk (or, if it does, by how much). Researchers are always learning more about the results of genetic testing and what they might mean, but at this time there are still many unanswered questions.

Some genetic test manufacturers advertise and promote their tests to doctors and to the public. Sometimes they can make the test sound much more helpful and certain than it's been proven to be. This can be harmful because decisions about testing may then be made based on incomplete information, or even on the basis of misleading or wrong information. A lot of tests don't give the answers they seem to promise. Reputable genetics counselors can help you know what to expect from your test results.

## **How might the results affect my family?**

Many people are uneasy even before they get their test results. They may think about how the result might affect them and their families. They may worry about how to talk about and manage the information.

Learning that you or a loved one might have or develop a serious disease like cancer can be scary. It can be even more upsetting if family members have already died of the disease in question. Having a gene or passing the gene on to children can also lead to feelings of guilt or anger.

If you're tested and found to have a specific gene variant or mutation, it might help your family members decide if they want to be tested also. This might help them learn more about their own risk, and if there are things they can do to lower it.

If it turns out that the gene variant or mutation does not run in your family, testing might offer peace of mind for family members.

Sometimes, not all family members want to know if they might be at increased risk, especially if there isn't much they can do about it. Testing any family member might lead to anxiety and other concerns in other family members.

Privacy may become an issue when many family members could be affected by a single positive genetic test result. More family members may need to be tested. Sometimes family secrets, such as paternity, adoptions, or other difficult issues may be discovered because of a genetic test result.

## **Will testing lead to more medical tests?**

In some cases, more medical tests or procedures might have to be done [as a result of genetic testing](#). For example, if the test finds a gene variant or mutation that increases your risk for colorectal cancer, more tests like colonoscopy may be recommended.

This can be a good thing, if these other tests can help keep you free of cancer or if they find it early when it's to treat. But the tests can have downsides as well, such as the time and cost involved, as well as possible risks from the tests themselves.

## Is genetic testing covered by insurance?

Genetic testing is complicated, and it can cost a lot, often thousands of dollars.

Most insurance plans cover genetic testing, at least to some extent. But whether a specific person will be covered depends on a number of factors. For example, the person's personal or family cancer history may need to meet certain medical criteria for testing. Some genetic tests are also more extensive (and cost more) than others.

Federal health care laws say that genetic testing should be covered for some women who do not have cancer themselves but are found to be at higher risk for a [BRCA1 or BRCA2 gene mutation](#)<sup>1</sup> for s. The amount of coverage will depend on their health plan.

It's best to get an idea of how much testing is likely to cost before you have it done. Your genetic counselor may be able to help you understand the likelihood that your test will be covered and the policies for pre-authorization and billing.

Even if genetic testing is covered, you may decide not to ask your insurance company to help pay for testing. Some people choose to pay for it themselves in order to keep the results as private as possible.

## Could the test results lead to discrimination?

People who get genetic testing in a medical setting are protected by the [Health Insurance Portability and Accountability Act \(HIPAA\)](#)<sup>2</sup> in terms of who can receive the information and how it can be used. The federal law known as GINA provides additional protections for most people.

### **GINA (Genetic Information Nondiscrimination Act of 2008)**

employees. This law also bars health insurers from making coverage or cost decisions based on genetic information.

GINA defines genetic information as:

- A person's genetic test results
- Genetic test results of family members
- Knowledge that one or more family members have a genetic disease or disorder

### ***Employers***

GINA bars employers from discriminating on the basis of genetic information in hiring, firing or layoffs, pay, or other personnel actions such as promotions, classifications, or assignments. The law applies no matter how they got the information.

Employers are not allowed to require genetic testing and can't collect genetic information, with very limited exceptions. For instance, it may be allowed when information is needed to meet the requirements of [family and medical leave laws](#)<sup>3</sup> or to watch for harmful effects from hazardous workplace exposures.

Employers must keep genetic information confidential. They can't release or share genetic information except when they are:

- Fulfilling a request from the employee
- Fulfilling a request from a health researcher
- Complying with medical leave law
- Disclosing or reporting to a public health agency

### ***Health insurers***

GINA bars health insurers (including group health plans, individual plans, and Medicare supplemental plans) from turning down people or charging higher premiums for health insurance based on genetic information or for using genetic services. This includes genetic counseling and testing. The law also bars these insurers from asking for or requiring genetic tests. GINA applies to all health insurance plans (including federally regulated ERISA plans, state-regulated plans, and private individual plans).

### ***When GINA does not apply***

- A few states have stronger laws than GINA. GINA does not take precedence over state laws against genetic discrimination that are broader in scope. Rather, GINA establishes a national baseline protection while allowing states to impose stronger protection.
- GINA's protections do not apply to life insurance, disability insurance, or long-term care insurance. It also doesn't require health insurance to cover genetic testing.
- GINA does not apply to very small employers (with fewer than 15 employees), nor does it apply to military health plans, the Veterans Administration, the Indian Health Service, or Federal Employees Health Benefits Plans.

## What about other privacy issues?

Patients, families, and health care providers are not the only ones interested in genetic information. Here are some of the other groups who might want to use this information:

### Medical and pharmaceutical researchers

Medical researchers must get a person's informed consent before any studies of their tissue samples and DNA can be done. Some labs use a system where a person can decide to allow or not to allow their leftover DNA to be used in research once the testing has been completed.

### Employers

Employers are allowed to ask for genetic testing if it's used to monitor exposure to potentially toxic chemicals and substances in the workplace. Testing for a [hereditary cancer syndrome](#)<sup>4</sup> would not be expected to fall under this category.

## Hyperlinks

1. [www.cancer.org/cancer/risk-prevention/genetics/family-cancer-syndromes.html](http://www.cancer.org/cancer/risk-prevention/genetics/family-cancer-syndromes.html)

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# What Happens During Genetic Testing for Cancer Risk?

## Information gathering

The first step in genetic testing is to collect information about your personal and family medical history. This may be done by a genetic counselor, or a doctor or nurse trained in genetic counseling.

You will be asked about your own medical history. This may include biopsies or surgeries you have had, cancer screening tests, gynecologic history (for women), [lifestyle factors, and exposure to things that can cause cancer](#)<sup>1</sup> (carcinogens). If you have been diagnosed with cancer, information such as the type of cancer, whether it has any relevant biomarkers, and if you've had more than one type of cancer, can also be important.

A detailed review of your family medical history is also important. Often, you will be asked to complete a questionnaire before your appointment, since you might need to ask relatives for information. The questionnaire will ask about how many relatives have had cancer on your mother's and father's side, including the type of cancer and their age when they were diagnosed. This can help determine if there is a pattern of cancer that might suggest a [hereditary cancer syndrome](#)<sup>2</sup>. This is why it's important to confirm the illnesses in your family by medical records and/or death certificates whenever possible.

## Risk evaluation

Once the information about you and your family has been collected, the genetic counselor or other trained professional will go over this information to help determine:

- Your risk of developing cancer,
- If genetic testing might be helpful for you, and
- If so, what specific gene changes should be tested for

Sometimes, a test looking at only one gene might be recommended, while at other times testing for a panel or group of gene changes might be better. The genetic counselor can explain the reasons for their recommendation.

## Education and informed consent

At one of the visits, the genetic counselor will explain how families can inherit increased cancer risk and how genes are passed on. The risks, benefits, cost, and limits of genetic testing will be discussed, as well as who in the family should consider being tested. It is

often a person who has or had cancer. Sometimes, more than one family member may

- Other testing options you could use
- Screening or treatment options that might be available depending on the test results (if this applies to you)
- Further decisions that may need to be made once the results are back
- The possible consent to use the results for research purposes after the test
- Availability of counseling and support services
- Your right to refuse testing
- The timeframe you should expect to get your results
- Genetic discrimination protections and limits

For more on this process see [Informed Consent](#)<sup>6</sup>.

## **Specimen collection and lab testing**

Once you've signed the consent form, lab tests are done on cells taken from your body.





## **If a result is negative**

If the test result is negative, it means **the test did not find a mutation in the genes it tested for.**

### ***In families with a known gene mutation***

If you have a family history of a known gene mutation, knowing you don't have this mutation can be very comforting. **But it doesn't mean your risk of cancer is zero. It just means you have the same average risk as most other people.**

Family members with negative test results may feel guilty if other family members test positive. Your health care provider or genetic counselor can help you understand what the test results mean for you and your family and how to deal with them.

### ***In families without a known gene mutation***

If there is no known gene mutation in the family, a negative result can still be helpful. It means you don't have that specific mutation (or any of the other mutations that might have been tested for). But genetic testing doesn't look for every possible gene mutation, so you can also get a negative test result if:

- You have a mutation that is linked to the cancer in the family that current testing methods cannot find.
- You have a mutation, but that specific gene change was not tested for.
- You have a mutation in a gene that has not yet been discovered.

Understanding what a negative test result means for your cancer risk and what you can do next to help lower your risk is one of the more complicated elements of genetic counseling. **Depending on your situation (such as your family cancer history), you and your family might still be considered at high risk of cancer even with a negative test result.** This is why it is important to discuss your negative result in detail.

## **If the result is a variant of uncertain significance (VUS)**

When a genetic test shows that a person has a change in a gene, but it's not known if this gene change affects cancer risk, it is reported as a variant of uncertain significance (VUS). It may be a normal variant, simply a different version of a gene that isn't seen often enough to be sure, or there may be some other explanation for it.

Your genetic counselor can help you understand what a VUS result might mean for you and your family. Typically, no changes to your medical care are recommended when a VUS is found because **most VUSs are later found to be benign (causing no medical harm) once more information on them becomes available.**

For some people, a VUS result can cause anxiety, frustration, and even anger because the result isn't clear and there is no exact timeline on when it might become clear. You usually don't need to have repeat testing if you have a VUS, but staying in contact with the healthcare provider who ordered the test is important in case updates on the VUS become available.

## What if genetic testing shows an increased cancer risk?

If your genetic test result is positive for a gene mutation that could increase your risk of cancer, managing your risk should become a priority.

Depending on which type(s) of cancer you are at increased risk for, some of the ways you might lower your risk or find cancer early include:

- **Lifestyle changes:** Making healthy choices and changing behaviors to try to help reduce cancer risk
- **Chemoprevention:** Taking medicines to help reduce risk
- **Preventive or prophylactic surgery:** Removing a healthy organ or gland to try to keep cancer from starting there
- **Early detection:** Doing what you can to find pre-cancer or cancer early through screening tests

Your health care provider may recommend one or more of these approaches, but it's important to understand how much they could lower your risk before you decide on a course of action. You will also want to be sure you understand their risks and benefits before deciding on a plan. These approaches are discussed in more detail below.

### Changes in lifestyle factors

Ask your health care provider about lifestyle changes you can make that could help lower your cancer risk. For instance, avoiding or limiting alcohol use and getting regular physical activity can help lower your risk of [colon](#)<sup>8</sup> and [breast cancer](#)<sup>9</sup>, while not smoking lowers the risk for a number of cancers.

In some cases, the effects of these changes on risk might be small compared to the

increased risk from the mutation, but you may still want to ask your provider what you can do.

## **Chemoprevention**

Chemoprevention is the use of medicines to help keep cells from developing into certain types of cancer. It is not the same as chemotherapy. These medicines are used to help lower the risk of certain cancers in people known to be at high risk. For example, tamoxifen and drugs called aromatase inhibitors are pills used mainly to treat people with breast cancer. But these drugs have also been shown to help lower breast cancer risk in people at high risk.

Each person's risk and medical situation must be considered carefully so that any harmful effects of the drugs do not outweigh the benefits. As we learn more about the genetics of cancer, we hope to learn more about using medicines to help lower cancer risk as well.

## **Prophylactic (preventive) surgery**

Prophylactic (preventive) surgery is another option in some cases. For example, some women at high risk for [ovarian cancer](#)<sup>10</sup> might decide to have their ovaries removed once they've had their children, which can greatly lower their risk. This can be important because screening tests are not considered reliable for this cancer, and many women do not have symptoms when the tumor is early stage.

## **Cancer early detection tests and awareness**

Often, those with a positive genetic test result will get different screening advice than for those at average risk for that cancer type. Early detection (screening) tests may be recommended to start at an earlier age and/or be done more often, or along with additional tests. For instance, a person known to have an increased risk of colon cancer might need to start screening earlier than usual. Likewise, a woman with a genetic mutation that raises her risk of breast cancer might need breast MRI scans along with her mammograms to look for early signs of the cancer.

But it's important to understand that many people with negative results might also get special screening recommendations based on their personal or family history of cancer. This is another reason why speaking with a genetics expert can be important for determining a care plan.

It's also important to be aware of the possible signs and symptoms of cancers you are

at higher risk for, and to see a doctor right away if anything concerns you. Finding cancer early – when it's small and has not spread – offers the best chance of treating it successfully.

## **Sharing results with family members**

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