
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?](#)
- [Talking with Family Members About Genetic Testing](#)

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?](#)
- [Genetic testing to help evaluate cancer risk](#)
- [Who might benefit from genetic testing?](#)
- [What is genetic counseling?](#)
- [What does a genetic counselor do?](#)

- [Before you get tested...](#)
- [Testing cancer cells for gene changes](#)
- [Home-based genetic tests](#)

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](#)¹.)

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](#)² 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](#)³, [ovarian](#)⁴, and [pancreatic cancers](#),⁵ 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](#)⁶.

What is genetic counseling?

Genetic counseling gives you information that you and your family can use to make decisions about whether to get genetic testing (see below).

Genetic counselors have special training in the field of genetic counseling. Most are board-certified, and some might have a license depending on the rules in their state. Some doctors, advanced practice oncology nurses, social workers, and other health professionals may also provide genetic counseling, although they might have different levels of training in this field. If you are offered genetic counseling, it's fair to ask about their training in this area.

What does a genetic counselor do?

Before and after genetic testing, a genetic counselor can help you understand:

- Your risk of getting certain types of cancer based on your family's medical history
- What you can do about this risk
- Which genetic tests can give you more information about your risk level
- What your test results might mean
- The privacy of your genetic information

Your counselor will explain the testing process. They can tell you more about what the tests can and cannot do and how well they work. Together, you will also discuss how knowing the test results may affect your emotions, mental health, and family. This information can help you decide if having genetic testing is right for you. It is your decision to have testing and what steps you take after.

Before you get tested...

It's important to find out how useful genetic testing might be for you **before** you do it. Talk to your health care provider and plan on getting genetic counseling before the actual test. This will help you know [what to expect](#). Your counselor can also tell you about the risks and benefits of the test, what the results might mean, and what your options are.

Your health care provider can refer you to a genetic counselor in your area, or you can find a list of certified genetic counselors on the website of the [National Society of Genetic Counselors](#)⁷.

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](#)⁸.

Home-based genetic tests

Some tests that look for gene changes can be bought without needing a doctor's order. For this type of testing, you purchase a test kit and send a sample of your DNA (often from saliva) to a lab for testing.

If you are considering using a home-based genetic test (also known as a direct-to-consumer genetic test), you need to know what it's testing for, what it can (and can't) tell you, and how reliable the test is. Even if a test from a reputable company is able to give accurate results, there are still likely to be limitations on what it can tell you.

Home-based tests do not provide information on a person's overall risk of developing any type of cancer. Sometimes these tests can sound much more helpful and certain than they have been proven to be. It may sound like the test will provide an answer to your specific health concern, such as your risk of hereditary cancer, but the test may not be able to answer that question completely. For example, a test may look for mutations in a certain gene, but it might not test for all of the possible mutations. So a negative test result, even if accurate, may miss the bigger picture regarding your cancer risk and what you can do to manage it. And you might not be provided with the important context about the test results that a genetic counselor could provide.

Home-based genetic tests should not be used instead of [cancer screening](#)⁹ or genetic counseling that may be recommended by a medical professional based on your individual risk for cancer. Always consult with your doctor if you are considering or have questions about genetic testing. Trained genetic counselors can help you know what to

expect from your test results.

Hyperlinks

1. www.cancer.org/cancer/understanding-cancer/genes-and-cancer.html
2. www.cancer.org/cancer/types/breast-cancer.html
3. www.cancer.org/cancer/types/breast-cancer.html
4. www.cancer.org/cancer/types/ovarian-cancer.html
5. www.cancer.org/cancer/types/pancreatic-cancer.html
6. www.cancer.org/cancer/risk-prevention/genetics/family-cancer-syndromes.html
7. findageneticcounselor.nsgc.org/
8. www.cancer.org/cancer/diagnosis-staging/tests/biomarker-tests.html
9. www.cancer.org/cancer/screening.html

References

American Society of Clinical Oncology. What to Expect When Meeting With a Genetic Counselor. Accessed at cancer.net. Content is no longer available.

Bonadies DC, Farmer MB, and Matloff ET. Chapter 38: Genetic counseling. In: DeVita VT, Lawrence TS, Rosenberg SA, eds. *DeVita, Hellman, and Rosenberg's Cancer: Principles & Practice of Oncology*. 11th ed. Philadelphia, Pa: Lippincott Williams & Wilkins; 2019.

McGee RB, Nichols KE. Introduction to cancer genetic susceptibility syndromes. *Hematology Am Soc Hematol Educ Program*. 2016;2016(1):293-301. doi:10.1182/asheducation-2016.1.293.

National Cancer Institute. Cancer Genetics Overview (PDQ®)—Health Professional Version. 2022. Accessed at <https://www.cancer.gov/about-cancer/causes-prevention/genetics/overview-pdq> on July 26, 2022.

National Cancer Institute. Cancer Genomics Overview. 2017. Accessed at <https://www.cancer.gov/about-nci/organization/ccg/cancer-genomics-overview> on July 26, 2022.

National Cancer Institute. Genetics of Breast and Gynecologic Cancers (PDQ®)—Health Professional Version. 2022. Accessed at <https://www.cancer.gov/types/breast/hp/breast-ovarian-genetics-pdq> on July 26, 2022.

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 on January 26, 2022.

Rahner N, Steinke V. Hereditary cancer syndromes. *Dtsch Arztebl Int*. 2008;105(41):706-714. doi:10.3238/arztebl.2008.0706.

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.

Last Revised: July 3, 2024

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

the chance to better understand your risk for a certain disease, such as cancer. Testing is not perfect, but it can often help you make decisions about your health.

For families who might be at risk, a negative result on a genetic test (that is, not finding an inherited gene mutation) may help lessen anxiety. In the same way, a positive result (finding an inherited gene mutation) might help you make important decisions about your future, including things you can do that might help lower your risk. If you test positive for an inherited gene change, your health care provider might recommend that you:

- Start cancer screening tests earlier, if such tests are available for the cancer(s) you're at risk for
- Get screened for that type of cancer more often
- Get screening tests that are used only for people known to be at increased cancer risk
- Be aware of possible signs or symptoms of that type of cancer
- Learn about options to help reduce the risk of certain types of cancer, such as medicines, surgery, or lifestyle changes

If you do develop cancer, finding it early (when the cancer is small) often means that treatment is more likely to be successful.

Finding a gene variant or mutation does not mean you will get cancer

Genetic testing can only tell you if you have a specific gene variant or mutation, not if you will get cancer. So, the test can tell what *might* happen, but it cannot tell what *will* happen. A positive test result does not always mean you will get the disease. And a negative result does not mean you have no risk of getting the cancer. Cancer risk can also change over time due to lifestyle choices, exposure to cancer-causing agents, and getting older.

This is why genetic counseling **before** testing is so important – to help ensure you understand if and how the results of genetic testing might help you.

As with many medical tests, genetic tests are not perfect, and there's also always the chance that different tests might provide different results. This is not common, as many steps are taken to prevent this, but at this time genetic testing is not tightly regulated, and different labs may have different ways of looking for certain gene changes.

Sometimes the tests themselves might not give clear answers. For example, you may

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](#)¹ 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 \(HIPAA\)](#)² 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)

GINA is a federal law that prohibits the use of genetic information in workplace employment decisions for non-governmental organizations with more than 15

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](#)³ 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](#)⁴ would not be expected to fall under this category.

Hyperlinks

1. www.cancer.org/cancer/risk-prevention/genetics/family-cancer-syndromes.html
2. www.hhs.gov/hipaa/index.html
3. www.cancer.org/cancer/financial-insurance-matters/health-insurance-laws/family-and-medical-leave-act.html
4. www.cancer.org/cancer/risk-prevention/genetics/family-cancer-syndromes.html

References

Bonadies DC, Farmer MB, and Matloff ET. Chapter 38: Genetic counseling. In: DeVita VT, Lawrence TS, Rosenberg SA, eds. *DeVita, Hellman, and Rosenberg's Cancer: Principles & Practice of Oncology*. 11th ed. Philadelphia, Pa: Lippincott Williams & Wilkins; 2019.

Department of Health and Human Services. Genetic Information. June 16, 2017. Accessed at <https://www.hhs.gov/hipaa/for-professionals/special-topics/genetic-information/index.html> on August 19, 2022.

National Cancer Institute. Cancer Genetics Overview (PDQ)—Health Professional Version. 2022. Accessed at <https://www.cancer.gov/about-cancer/causes-prevention/genetics/overview-pdq> on July 26, 2022.

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 on January 26, 2022.

Last Revised: October 24, 2023

What Happens During Genetic Testing for Cancer Risk?

Genetic testing is typically done only if you and your health care team feel that it's the best thing for you and your family. Several steps are taken before actual testing is done. The entire process might involve 1 to 3 visits or phone calls.

- [Information gathering](#)
- [Risk evaluation](#)
- [Education and informed consent](#)
- [Specimen collection and lab testing](#)
- [What if genetic testing shows an increased cancer risk?](#)
- [Sharing results with family members](#)

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](#)¹ (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](#)². 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 be offered testing.

You will want to think about how the results might affect you and your relatives, and discuss these issues with the counselor before testing.

For example, if testing shows that you have a high cancer risk, the counselor may also talk about the best ways to manage it. These may include [lifestyle changes](#)³, [early detection](#)⁴, watching for [signs and symptoms of cancer](#)⁵, medicines to reduce cancer risk, or even preventive surgery.

Many of these issues require the skills of an expert counselor. An increased risk of cancer, especially for children, and the potential for discrimination can be frightening. It often helps to discuss your family's attitudes about cancer and the possibility of a family member being "blamed" for the cancer. You may also talk about how cultural beliefs, support systems, and finances may play a role in how your family views cancer.

The counselor will explore ways to help you cope with the results of genetic testing, as well as help to ease your specific fears and concerns. The counselor can also help you with how to discuss the test results and what they mean with other family members.

It's important to check with your health insurance company to find out if genetic counseling and genetic testing will be covered. Your genetic counselor may be able to help you find out if the recommended test(s) is covered.

Remember that genetic testing is your choice. **Even if the counselor recommends you be tested (or even if the counselor tells you genetic testing might be helpful for you), you still have the right to refuse it.**

After risk assessment and genetic counseling, you can decide if you want to proceed with testing. Some people prefer to go home and think about it first, but others are ready to complete testing that day. If you decide to be tested, you'll be asked to give your **informed consent** in writing. The process of informing you and your family about testing should cover:

- The purpose of the genetic test
- The reason for offering the test to you and/or other family members
- The type and nature of the genetic condition being tested for
- Test accuracy
- The [benefits and shortcomings of testing](#) (including the limits of what the results might tell you)
- 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](#)⁶.

Specimen collection and lab testing

Once you've signed the consent form, lab tests are done on cells taken from your body. Genetic tests for cancer are typically done on a sample of blood, saliva (spit), cheek cells (from swabbing the inside of your mouth), but they can also be done on other body tissues. Those who have an active blood cancer (such as leukemia) or a history of a bone marrow (stem cell) transplant may need to give a sample other than blood or saliva for accurate results. Typically, there are no dietary restrictions before this type of test.

Be sure to ask your genetic counselor when to expect the test results, and if you need to schedule a follow-up appointment to discuss them.

Getting the test results

Once the results are ready (often 2-3 weeks later), your genetic counselor will share the results with you. This may be by phone or at a scheduled appointment. You might want to ask for a copy of the test result for your records if one is not given to you. Some people might choose to have a family member with them when they get their results. This might be for emotional support, to help make sure they understand the information, to help with asking questions and taking notes, or to help share results with other family members.

Testing does not always give you clear answers, but genetic counselors are trained to interpret and explain the test results and what they might mean to you and your family.

The results of each test might come back as:

- Positive
- Negative
- Variant of uncertain significance (VUS)

If a result is positive

What a positive result means for cancer risk

A positive result means you have a mutated gene (or genes) that may place you at higher risk for developing certain types of cancer. If you have already been diagnosed with cancer, the result might have found a factor that played a role in causing the cancer. It might also suggest a higher risk for developing other cancers.

Many people are concerned or anxious after learning they are at increased risk for cancer. This is normal. The results may also point toward a higher risk for certain family members, which can be stressful. Concern about being treated differently may also become more real.

Even after the testing is complete, there might still be a lot of uncertainty. In most cases, there's no way to know for sure if or when cancer might develop. But it's important to remember that the results of genetic testing do not change your current state of health, meaning they do NOT tell you if you have cancer or not. Your health care provider or counselor can help you sort through your options at this point, including what you might be able to do to lower your risk of these cancers or catch them at an early stage. (See "What if genetic testing shows an increased cancer risk?" below.)

The counselor will talk to you about whether your family members might also be affected. It will be important for those family members to know exactly which mutation was found, so they can decide if they want to be tested for it as well. To make sure family members are tested correctly, you might want to share a copy of your actual test results with family members interested in genetic testing. They can then discuss this with their health care providers and might decide to get tested, too.

How a positive result might affect cancer treatment

A positive result could also mean that you have a mutated gene (or genes) that may affect your treatment options for certain types of cancer.

For more information see [Biomarker Tests and Cancer Treatment⁷](#) and [Understanding Genetic Testing for Cancer](#).

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](#)⁸ and [breast cancer](#)⁹, 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](#)¹⁰ 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.

Sometimes insurance plans will deny coverage for increased or additional screening tests even if your doctor thinks you should have them. There are [things that you and your doctor can do](#)¹¹ to try to get your insurance plan to approve these tests after a claim is denied.

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

If you have a positive test result (a gene mutation that raises your risk), you may need to think about sharing the information with other family members who might also be at increased risk. To learn more, see [Talking with Family Members About Genetic Testing](#).

Hyperlinks

1. www.cancer.org/cancer/risk-prevention.html
2. www.cancer.org/cancer/risk-prevention/genetics/family-cancer-syndromes.html
3. www.cancer.org/cancer/risk-prevention.html
4. www.cancer.org/cancer/screening.html
5. www.cancer.org/cancer/diagnosis-staging/signs-and-symptoms-of-cancer.html
6. www.cancer.org/cancer/managing-cancer/making-treatment-decisions/informed-consent.html
7. www.cancer.org/cancer/diagnosis-staging/tests/biomarker-tests.html
8. www.cancer.org/cancer/types/colon-rectal-cancer.html
9. www.cancer.org/cancer/types/breast-cancer.html
10. www.cancer.org/cancer/types/ovarian-cancer.html
11. www.cancer.org/cancer/financial-insurance-matters/managing-health-insurance/if-your-health-insurance-claim-is-denied.html

References

Bonadies DC, Farmer MB, and Matloff ET. Chapter 38: Genetic counseling. In: DeVita VT, Lawrence TS, Rosenberg SA, eds. *DeVita, Hellman, and Rosenberg's Cancer: Principles & Practice of Oncology*. 11th ed. Philadelphia, Pa: Lippincott Williams & Wilkins; 2019.

Centers for Disease Control and Prevention. Genetic Testing. June 24, 2022. Accessed at https://www.cdc.gov/genomics/gtesting/genetic_testing.htm on August 19, 2022.

MedlinePlus [Internet]. Bethesda (MD): National Library of Medicine (US); [updated 2020 Jun 24]. Help Me Understand Genetics; [updated 2020 Jun 18; reviewed 2018 Jun 01]; Available from: <https://medlineplus.gov/genetics/understanding/>.

National Cancer Institute. The Genetics of Cancer. August 17, 2022. Accessed at <https://www.cancer.gov/about-cancer/causes-prevention/genetics> on August 19, 2022.

Last Revised: September 14, 2022

Talking with Family Members About Genetic Testing

If you've had genetic testing, you might choose to share the results with your family members. If a family member has had genetic testing, you may be interested in knowing their results.

- [Why sharing results can be important](#)
- [How do I talk to family members about genetic test results?](#)
- [Sharing results with children](#)
- [Questions to Answer Before Talking to Family Members](#)
- [Finding a genetic counselor](#)

Why sharing results can be important

When you share your genetic test results with your family, the conversation gives them a chance to understand what you are going through. It might also offer information that family members can use to learn about their own cancer risk. Here's why this information can be important:

- First-degree biological (blood) relatives (mother, father, sibling, child) of the person that tests positive have a 50% chance of having the same gene mutation.
- Second-degree biological relatives (aunt, uncle, grandparent, grandchild, niece, nephew, or half-sibling) of the person that tests positive have a 25% chance of having the same gene mutation.
- Family members can take steps to reduce their cancer risk or to find cancer early.

- Knowing the family history and genetic testing results helps people talk to their health care team and make informed decisions about their care.

How do I talk to family members about genetic test results?

When a person gets genetic test results from their genetic counselor, it's important that they learn as much as possible about the results before sharing them with others. In addition to other learning resources, the counselor can provide a **family letter** that includes the specific gene mutation, the name of the [family cancer syndrome](#)¹ and other helpful resources. The letter can be very helpful in planning a conversation with family members. It can even be sent to family members or used along with having a conversation.

People can talk with family about genetic testing and test results in many ways, such as a phone call, face-to-face conversation, text, email, or letter. Some people may come up with valid reasons for not sharing results with family because the time isn't quite right. But there really is no best time for everyone and every family.

Starting the conversation

Beginning the conversation can be the hardest part. If you're the one sharing genetic test results, here are some suggestions you can consider:

- *I recently had genetic testing. Would you be interested in hearing about it?*
- *I'd like to tell you about my genetic test results. I think my results could help you too.*
- *I learned some information that might be important for you to know, too. Can I tell you about it?*
- *After my cancer diagnosis, my doctor had me meet with a genetic counselor. They helped me understand that my cancer risk was higher because I have a gene mutation. They told me my close relatives could have the same gene mutation. Can I give you some information so you can talk to your doctor about it?*
- *I am going to send you an email that has my recent genetic test results. I want you to have this information so you can decide if genetic testing might be something you could benefit from having. Your doctor can help you decide.*

If you're afraid or nervous to talk to family members

It can be stressful to talk with anyone about a health problem or test result. You may feel nervous sharing personal health information with family members, even if they could benefit from hearing it. If you want to share your genetic testing results with your family, here are some tips to help you feel more comfortable and get started.

1. Make a list of who you want to share your genetic test results with.
2. Decide how you will share the information: in person, or by phone, email, text or letter.
3. Pick a date and write it on your calendar. Consider choosing an upcoming family get together or sending your family member an email or text in advance.
4. Write down what you are going to say ahead of time or practice with a friend.
5. Your doctor or genetic counselor can give you advice on how to explain your results, since they know your situation best.

Prepare for possible reactions

It's important to understand and address your own feelings before talking to your family members. It's also important to be aware that your family members might feel differently. It's hard to know how family may react to the news. It's common for people to feel a range of emotions when learning about a positive genetic test result or family cancer syndrome – whether it's a family member who was tested or if they were tested themselves. You and your family members may feel:

- Angry or scared about what it means to be at increased risk for cancer
- Guilty for possibly passing an inherited gene mutation on to a child
- Empowered or relieved to have the information they can do something about

Your doctor or genetic counselor may be able to share experiences they've had with past family conversations. These might help you anticipate how your family members may react, so you feel better prepared.

Sharing results with children

It's natural to want to protect children from bad news or information that could feel scary. But kids generally want to be informed and included. Your genetic counselor, doctor, or a pediatrician can help you find resources to guide you through these discussions, including books and online resources based on the child's age.

You might wonder if it's a good idea to have children go through genetic testing, or to

wait until they are adults to decide themselves whether to have it. It's important to know that most cancer-related gene mutations affect the risk for cancers in adulthood rather than childhood cancers. However, there are family cancer syndromes that do affect children, and it could be helpful for them to be tested in childhood. Your doctor and genetic counselor can help you and your family decide.

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

Questions to Answer Before Talking to Family Members

If you are sharing your own test results, here are some questions to think about. Addressing these first can help conversations go more smoothly. These questions can be helpful if you're waiting to hear someone else's results, too.

- *Do I have all the information I need? What specific information am I going to share?*
- *Who is at risk in my family? Are there certain family members to reach out to?*
- *What information am I comfortable sharing in person? What information would I prefer to share by letter or email or text?*
- *How do my family members like to be communicated with? Would they prefer a chat over coffee or an email?*
- *How am I feeling about sharing this information? Would it be helpful if I talked to someone else before talking to family or helpful to talk to one family member before others?*
- *Are there any family or cultural beliefs I need to consider before telling my family?*
- *What emotions might my family member(s) feel when I tell them about this?*
- *Do I have good resources handy to share with my family members?*
- *Would it be helpful to have my genetic counselor write a family letter for me that includes the details of my test results?*

Remember...

- You don't have to be an expert to have this conversation.
- Your doctor or counselor may recommend that family members be tested for the same genetic mutation that you have, but they will need to decide that themselves. A family letter that includes detailed information could be a very helpful resource.
- Encourage your family members to talk to someone about testing. Their own doctor or genetic counselor can help them decide if genetic testing is right for them.
- This may feel hard. Being brave for your family members could help them

understand their risk, take steps to stay on top of their health, and even prevent cancer or detect it early.

- Help your family member be accountable. Ask them if it would be ok to check back with them in a few weeks to see if they have questions or need help taking the next step.

Finding a genetic counselor

It's important to find out how useful genetic testing might be for you **before** you decide to do it. Talk to your doctor first and ask where you can connect with a genetic counselor or if they recommend one for you. Genetic counselors specialize in cancer risk assessment and genetic testing. Talking with a trained specialist will help you know [what to expect](#). If you decide to make an appointment with a genetic counselor, they can tell you about the risks and benefits of the best tests for you. They can also explain what the results might mean, and what your options are.

You can find a list of certified genetic counselors at the [National Society of Genetic Counselors](#)².

Hyperlinks

1. www.cancer.org/cancer/risk-prevention/genetics/family-cancer-syndromes.html
2. findageneticcounselor.nsgc.org/

References

Ballard LM, Band R, Lucassen AM. Interventions to support patients with sharing genetic test results with at-risk relatives: A synthesis without meta-analysis (SWiM). *Nature*. 2023; 31:988-1002. Available at <https://www.nature.com/articles/s41431-023-01400-1>.

Centers for Disease Control and Prevention (CDC). *Let's talk: Sharing info about your family cancer risk*. 2024. Accessed at <https://www.cdc.gov/cancer/risk-factors/family-health-history.html> on October 10, 2024.

Facing Hereditary Cancer Empowered (FORCE). *Sharing information with adult relatives*. 2020. Accessed at <https://www.facingourrisk.org/info/hereditary-cancer-and-genetic-testing/sharing-with-family/sharing-genetic-information-with-adult> on October 10, 2024.

Facing Hereditary Cancer Empowered (FORCE). *Sharing information about hereditary cancer with children*. 2020. Accessed at <https://www.facingourrisk.org/info/hereditary-cancer-and-genetic-testing/sharing-with-family/sharing-genetic-information-with-children> on October 10, 2024.

Finn KS, Pacyna J, Tsou CA, Smadder NJ, Sharp R. Patient-reported anticipated barriers and benefits to sharing cancer genetic risk information with family members. *European Journal of Human Genetics*. 2021; 30:53-61. Available at <https://www.nature.com/articles/s41431-021-00890-1>.

Hodan R et al. Family communication of cancer genetic test results in an ethnically diverse population: a qualitative exploration of more than 200 patients. *Journal of Community Genetics*. 2024; 15:363-374.

Hunter JE et al. Most people share genetic test results with relatives even if findings are normal: Family communication in a diverse population. *Genetics in Medicine*. 2023; 25:100923. Available at <https://pubmed.ncbi.nlm.nih.gov/37421176/>.

National Society of Genetic Counselors (NSGC). *How can genetic counselors help me understand results from genetic testing?* Accessed at <https://www.aboutgeneticcounselors.com/Who-Are-Genetic-Counselors/How-Can-Genetic-Counselors-Help-Me-Understand-Test-Results> on October 10, 2024.

Ofverholm A, Karlsson P, Rosen A. The experience of receiving a letter from a cancer genetics clinic about risk for hereditary cancer. *European Journal of Human Genetics*. 2024; 32:539-544. Available at <https://www.nature.com/articles/s41431-024-01551-9>.

Wynn J et al. Do research participants share genomic screening results with family members? *J Genet Couns*. 2022; 31(2):447-458. Available at <https://onlinelibrary.wiley.com/doi/10.1002/jgc4.1511>.

Last Revised: October 28, 2024

Written by

The American Cancer Society medical and editorial content team
(<https://www.cancer.org/cancer/acs-medical-content-and-news-staff.html>)

Our team is made up of doctors and oncology certified nurses with deep knowledge of cancer care as well as editors and translators with extensive experience in medical writing.

American Cancer Society medical information is copyrighted material. For reprint requests, please see our Content Usage Policy (www.cancer.org/about-us/policies/content-usage.html).

cancer.org | 1.800.227.2345