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

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.

- 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].

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].

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

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.

What to Expect When Meeting With a Genetic Counselor. Cancer.net. 2021. Accessed at https://www.cancer.net/navigating-cancer-care/cancer-basics/genetics/what-expect-when-meeting-genetic-counselor.html. No longer available.

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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.

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

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

- 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.cancer.org/cancer/financial-insurance-matters/health-insurance-laws/what-is-hipaa.html
3. www.cancer.org/cancer/financial-insurance-matters/health-insurance-laws/family-and-medical-leave-act.html

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.

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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.

- 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 benefitsand 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

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.

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.

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.

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. This information could possibly be life-changing, because telling them might help them decide if they should also get genetic counseling and testing and/or adopt some of the approaches to try to lower their own risk.

Genetic test results can also cause anxiety, and some family members may not want to know their risk. This is especially true if there’s not much they can do with the results. You might want to speak with family members before you get tested to find out if they want to know your results.

Hyperlinks

1. www.cancer.org/cancer/risk-prevention.html

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.

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