What Happens During Genetic Testing for Cancer Risk?

The information on this page talks about the process involved in genetic counseling and 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.

If you are considering taking a home-based genetic test, you need to know what it’s testing for. Home-based tests do not provide information on a person’s overall risk of developing any type of cancer. A home-based genetic test should not be used as a substitute for cancer screening or genetic counseling that may be recommended by a medical professional based on your risk for cancer.

Risk assessment

The first step in genetic testing is to find out how likely you are to develop a certain disease. This risk is based on things like your medical history and the pattern of disease in your family (your family history). Your risk may be looked at by a doctor, nurse, or trained genetic counselor.

You’ll be asked why you want to be tested. Your family history (as far back as possible and up to the present day) will be reviewed in depth. Your provider will look for patterns of disease in your family. It’s important to confirm the illnesses in your family by medical records and/or death certificates whenever possible.

If the genetic test is for a certain type of cancer, you may also be asked about your own medical history. This may include early detection\(^1\) practices, gynecologic history (for women), lifestyle factors\(^2\), and exposure to things that can cause cancer\(^3\) (carcinogens).
At this point, it often helps to look at your family’s attitudes about cancer and the possibility of a family member being “blamed” for the cancer. Cultural beliefs, support systems, and finances may also play a role in how your family views cancer. These topics may be discussed, too.

**Genetic counseling**

Genetic counselors have special training and often graduate degrees in their field. Some doctors, advanced practice oncology nurses, social workers, and psychologists with special training may also do genetic counseling.

The purpose of genetic counseling is to give you information in an unbiased or neutral way so that you or you and your family can make your own decisions about whether to get tested. Sometimes the need for testing is clear and a health care provider will recommend testing and follow-up care. But even if that’s the case, you have the right to refuse testing. You may feel more comfortable making a decision after you talk with a genetic counselor.

It’s important to check with your health insurer to find out if genetic counseling and genetic testing will be covered.

The counselor will explain how families inherit cancers and how genes are passed on to children. They will also talk about the types of cancer seen in the family and estimates of the person’s cancer risk. The pros and cons, cost, and limits of testing are discussed too, as well as who in the family should consider being tested. 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 the person has a high cancer risk, the counselor may also talk about the best ways to manage it. These may include lifestyle changes, early detection, watching for signs and symptoms of cancer, 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. The counselor will explore ways to cope, as well as your specific fears and concerns. How to discuss the test results and what they mean with other family members is another key topic that will be covered.

Your health care provider can probably refer you to a genetic counselor in your area. If
Informed consent

After risk assessment and genetic counseling, 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
- Pros and cons 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

For more on this process see Informed Consent.

Specimen collection and lab testing

Once you’ve signed the consent form, lab tests are done on cells taken from your body. Genetic tests may be done on a sample of blood, hair, cheek cells (from swabbing the inside of your mouth), urine, amniotic fluid (the fluid that surrounds a fetus during pregnancy), or other body tissues.

Genetic tests for cancer usually mean you will give several tubes of blood. A thin needle is put into a vein (usually in your arm) to get the blood.

Getting the test results

After the test is done, your genetic counselor will share the results with you. Some
people choose to bring a family member 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. If a mutation is found, the counselor will talk to you about which of your family members might also be affected. It will be important for those family members to know exactly which mutation was found and in which gene. They can then discuss this with their health care providers and may decide to get tested, too.

Once the testing is complete, the results are sent in writing to the provider or genetic counselor. You’ll then be given the results at another counseling session. This might not happen until several weeks or even months after the samples are taken. The accuracy of the test and the meaning of the results will be discussed with you in detail.

If the result is positive

A positive result means you have a mutated gene (or genes) that may place you at risk. Your risk of developing the cancer will be discussed in light of the fact that you have the mutation.

Many people are concerned or anxious after learning they are at increased risk for cancer. This is normal. The results may imply risk for certain blood relatives as well, and might lead to strains in family relationships as well. Concern about being treated differently may become more real.

Even after the testing is complete there’s often a great deal of uncertainty. In most cases, there’s no way to know for sure that the disease will develop. And even if you do know you’ll get the disease, you still wouldn’t know when it might develop. 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 help lower your risk.

If the result is negative

If the test result is negative, you don’t have the gene mutation you were tested for. You’ll probably feel relieved that the test did not show you are at increased risk.

But it’s important to understand that genetic test results can’t always guarantee you’re not at increased risk. For instance, there might still be a chance that you have a different mutation that you were not tested for. (Many genes can have more than one kind of mutation that can result in higher cancer risk.) And rarely, the test result may be
a “false negative.” This means the test reads negative but the mutation is actually there.

Even a result that’s truly negative does not mean your risk 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.

If the result is inconclusive

If there’s not enough information to know whether you have the genetic changes, the test result is said to be inconclusive. You (and your family) may still be at a higher risk of developing cancer. Taking steps to lower your risk or look for cancer early can be helpful for some people, but not having a sure result can still cause anxiety and frustration. Your health care provider or genetic counselor can help you understand what the results might mean for you and your family and help you cope with them. In some cases, testing blood relatives can help clarify this result.

If the result is variance of unknown or uncertain significance

When genetic tests show that a person has an unusual form of a gene, but doctors don’t know what this gene change means the result is called a variance of unknown or uncertain significance (VUS). It may be a normal variant, simply a different version of gene that isn’t seen often enough to be sure, or there may be some other explanation.

For some, a VUS result can cause anxiety, frustration, and even anger because this result gives no information to guide future decisions. Your health care provider or genetic counselor can help you understand what the results might mean for you and your family and help you cope with them.

What if genetic testing shows an increased cancer risk?

If your genetic test result is positive or inconclusive 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 include:

- **Lifestyle changes:** Making healthy choices and changing behaviors\(^2\) 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 cancer early (through screening tests and awareness of early signs and symptoms), when the cancer is likely to be small and treatment is most likely to be successful

Your health care provider may recommend one or more of these approaches, but it’s important to understand how much they could affect your risk before you decide on a course of action. You will also want to be sure you understand their risks and downsides 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](#) and [breast cancer](#), while not smoking lowers the risk for a number of cancers.

In some cases the effect 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. Medicines are being studied and used to help lower the risk of certain cancers in people known to be at high risk. For example, the drugs tamoxifen and raloxifene can be used to help reduce breast cancer 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 genetic diseases, we should find out more about using medicines to help prevent cancer.

**Prophylactic (preventive) surgery**

Prophylactic (preventive) surgery is another option in some cases. For example, some women at high risk for [ovarian cancer](#) may decide to have their ovaries removed once they’ve had their children.
Cancer detection tests and awareness

Early detection (screening) tests may be started at an earlier age or be done more often, or special tests may be needed if you have a positive genetic test result. 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.

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 gene mutation that raises your risk, you may need to think about whether to tell other family members who might also be at increased risk. Telling them might help them decide if they should get tested or adopt some of the approaches to try to lower their risk.

On the other hand, some test results may cause more anxiety than anything else, and some family members may not want to know their own 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/healthy/find-cancer-early.html
4. www.cancer.org/healthy.html
5. www.cancer.org/healthy/find-cancer-early.html

References


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Last Medical Review: April 23, 2016 Last Revised: June 9, 2020