

## **Genetic Testing** [1]

This section has been reviewed and approved by the [Cancer.Net Editorial Board](#) [2], 03/2012

### **Key Messages:**

- Genetic testing helps predict the likelihood that a person will develop a disease, including some types of cancer.
- Genetic testing is a personal decision with many factors to consider.
- Anyone who decides to undergo genetic testing should receive genetic counseling before and after the test is performed.

Genetic testing is the analysis of genes, chromosomes, or proteins to help predict the risk of disease, identify carriers (individuals who do not have the disease but have a copy of the disease gene) of disease, diagnose disease, or determine the likely course of a disease. More than 900 genetic tests are available for many different diseases, including breast, ovarian, colon, thyroid, and other cancers.

Genetic testing for cancer risk is sometimes called predictive testing, which means that it can help predict the likelihood that an individual will develop cancer in his or her lifetime. No genetic test can report with 100% certainty that a person will develop cancer, but the tests can tell a person if he or she has a higher risk of developing cancer than the general population. Not everyone with a cancer-predisposition gene will develop cancer. For example, a woman with a 75% chance of developing breast cancer may remain healthy, while a woman with a 25% chance of developing breast cancer may eventually develop cancer.

The following factors suggest that a person may be at risk for developing a hereditary cancer:

- Family history of cancer: Three or more relatives on the same side of the family with the same or related forms of cancer
- Early onset: Two or more relatives diagnosed with cancer at an early age
- Multiple sites: Two or more types of cancer occurring in the same relative

### **ASCO recommends that genetic testing be offered in the following situations:**

- The person has an individual or family history that suggests a genetic cause of cancer
- The test for the genetic condition can be adequately interpreted
- The results of the genetic test will help with the diagnosis, treatment, and/or management of

the patient and family members at risk for cancer

In addition, ASCO recommends genetic counseling both before and after the genetic test. Learn more about ASCO's latest recommendations on [genetic testing for cancer susceptibility](#) [3].

### **Reasons to consider genetic testing for cancer**

Genetic testing can help a person (and in some cases, the family) understand the cause of cancer and the risk of developing cancer in the future or passing it on to children. Genetic testing is a personal decision that should be made in collaboration with your family and doctor. Individuals choose genetic testing for the following reasons:

**To make a medical intervention.** People at a higher risk for cancer may have the option of having more frequent [cancer screenings](#) [4], avoiding specific risk factors, making lifestyle changes to lessen additional risk, or taking preventive medication ([chemoprevention](#) [5]), if available. In a few cases, people with a genetic predisposition to develop cancer may have the opportunity to reduce their risk. For example, a woman with a breast cancer susceptibility gene mutation (*BRCA1* or *BRCA2* gene mutation) may reduce her risk of breast and ovarian cancer with a risk reducing salpingo-oophorectomy (removal of the fallopian tubes and ovaries) and/or a risk reducing bilateral mastectomy (removal of both breasts).

**To relieve anxiety, especially in a family that knows that the cancer is due to a specific gene mutation.** A person who tests negative for this familial mutation may experience some relief.

**To relieve uncertainty.** Testing information may relieve anxiety and the worry of not knowing your gene status. It may reduce feelings of uncertainty. For some people, knowing is better than not knowing.

### **Additional factors to consider**

A person considering genetic testing should also be aware that it has limitations and psychological implications.

**Testing may cause depression, anxiety, or guilt.** If a person receives a positive test result (meaning a mutation exists), it may cause anxiety or depression about the possibility of developing cancer. Some people may start to think of themselves as sick, even if they never develop cancer. If a person does not have the mutation when other members of the family do, this individual may experience guilt.

**Testing may cause family tension.** In some situations, a person may feel a responsibility to tell extended family members that they have a positive test result and encourage them to be tested. This process may lead to tension in the family. Learn more about [sharing genetic test results with your family](#) [6].

**Testing may provide a false sense of security.** Just because a person's genetic test result is negative does not mean that person will never develop cancer. It only means his or her risk is similar to the risk of the general population of developing cancer, including any risk that may

come from lifestyle and environmental factors. If a person has had cancer or a pre-cancerous condition, he/she would still have some increased risk based on his/her own history.

**Testing may provide unclear results.** A person's gene may have a unique mutation that is not known to affect cancer risk. Or, the gene may have a mutation that is not detected by the available test. Many cancers cannot be tied to a specific gene, and some genes may interact in unpredictable ways with other genes or factors in the environment to cause cancer. In either case, it may be impossible to calculate the risk of the mutation as it relates to cancer, which may lead to anxiety and uncertainty.

**Testing can be costly.** Genetic testing and counseling can be expensive, especially if it is not covered by insurance or if a person pays for testing out of pocket.

**Testing may cause confidentiality concerns.** Some people fear that their test results may lead to genetic discrimination or feel concerned about maintaining the privacy of their genetic information. They may be unsure if they want to share test results with immediate relatives and are concerned about employment and health and life insurance discrimination. Consider discussing privacy concerns with a genetic counselor or doctor.

### **Genetic discrimination legislation**

In 2008, Congress passed the Genetic Information Nondiscrimination Act (GINA). The legislation protects all Americans against discrimination based on their genetic information in receiving health coverage or employment. Learn more about [GINA and genetic discrimination](#) [7].

### **Questions to ask yourself about genetic testing**

Before being tested, be sure you have a complete understanding of the risks of testing and your reasons for wanting a test. It is also helpful to think about how you will cope with the results of the test. The following are some factors to consider in your decision making:

- Do I have a family history of cancer, or have I developed cancer at an earlier-than-average age?
- How will I interpret the results of genetic testing? Who will assist me in using this information?
- Will knowing the test results affect my medical care or the medical care of my family?
- If a genetic condition is detected, are there steps I can take to lower my risk?

A genetic counselor can help address these issues. Genetic counselors are professionals specially trained to advise people about the risks and benefits of genetic testing and can help people through the process of testing and interpreting the results. Learn more about [what you can expect when meeting with a genetic counselor](#) [8].

### **More Information**

[Understanding Cancer Risk](#) [9]

[The Genetics of Cancer](#) [10]

## Understanding Statistics Used to Estimate Risk and Recommend Screening [11]

### **Additional Resources**

National Human Genome Research Institute: Issues in Genetics [12]

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#### **Links:**

[1] <http://www.cancer.net/navigating-cancer-care/cancer-basics/genetics/genetic-testing>

[2] <http://www.cancer.net/about-us>

[3] <http://www.asco.org/ascov2/Press+Center/Latest+News+Releases/ASCO+Helps+Doctors+and+Patients+Sort+Through+Genetic>

[4] <http://www.cancer.net/node/24972>

[5] <http://www.cancer.net/node/24973>

[6] <http://www.cancer.net/node/24906>

[7] <http://www.genome.gov/10002328>

[8] <http://www.cancer.net/node/24907>

[9] <http://www.cancer.net/node/25007>

[10] <http://www.cancer.net/node/24897>

[11] <http://www.cancer.net/node/24960>

[12] <http://www.genome.gov/Issues/>