Yale NewHaven Health
Smilow Cancer Hospital

Yale CANCER CENTER
A Comprehensive Cancer Center Designated by the National Cancer Institute

Frequently Asked Questions about Genetic Testing for Hereditary Cancer

1. **What are genes?**
   Genes are the inherited material passed from parent to child that help to determine our body's features. Genes are like "instructions" that are "written" in a code – this code is called **DNA**. Genes have different jobs and functions in the body – some genes determine what color our eyes are or how tall we are, and other genes help to protect the body from developing cancer.

2. **What are mutations?**
   Mutations are harmful changes, or variations, in the DNA code of our genes. If a mutation is found in a gene that is important for protecting against cancer, then it causes an increased risk for that person to develop cancer in his or her lifetime.

3. **Is all cancer hereditary?**
   No, the majority of cancer is *not* hereditary. Most cancer is caused by the natural aging process, environmental factors such as pollution, lifestyle factors such as smoking, and random events. Only 5 - 10 percent of cancer is hereditary – meaning it is caused by a mutation(s) that increases a person's risk to develop cancer in his or her lifetime.

4. **Should everyone have genetic testing for hereditary cancer?**
   Genetic testing is considered for people with a personal or family history of early-onset cancer, more than one cancer in a person, the same cancer in two or more close relatives, unusual presentation of cancer (such as men with breast cancer), or certain ancestral backgrounds such as Ashkenazi Jewish ancestry. In these people, the risk for a hereditary cancer syndrome is elevated and genetic testing is recommended.

5. **I already have cancer. Why should I get tested?**
   For some people with cancer, genetic testing results can make them eligible for specific cancer treatments. For other people with cancer, the results can change their surgical decision and they may decide to have surgery to reduce their hereditary cancer risk. Additionally, people with cancer who undergo genetic testing can learn information about their risk to develop other types of cancer, and their results can provide important information to their children and close relatives.

6. **How is genetic testing for hereditary cancer done?**
   Genetic testing is conducted from a blood or saliva sample, which is used to look at the DNA code of genes related to hereditary cancer risk to determine if there is a mutation. This testing typically looks at a number of genes associated with a hereditary cancer risk. This is often called "panel genetic testing."

7. What are the potential results or outcomes of genetic testing?

There are three types of genetic test results:

- Positive: A harmful mutation (called a pathogenic variant) was detected. This means a person has an increased risk of developing cancer over his or her lifetime. This result has impact for that person, as well as his or her children and other family members.

- Negative: No harmful mutations were detected. This means a person has no known hereditary risk to develop cancer. However, this result does not completely rule out the possibility of a hereditary risk for cancer, since a mutation can be missed in one of these genes tested or there may be a mutation in a gene that has not yet been discovered.

- Variant of uncertain significance: A change or variation in the DNA was found in one of the genes tested, but the significance of this change is not yet known. This is called a variant of uncertain significance. This result means the laboratory does not know if the variation in the DNA is a *harmful* change causing an increased risk of cancer or is a *benign* change that does not cause an increased risk of cancer.

  Over time, the laboratory may be able to collect more information to better understand if the variant is harmful or benign. We typically do not change a person's cancer screening or follow-up plan based on a variant of uncertain significance. We also do not test relatives to see if they have the same variant.

8. Does a positive test result explain why I developed cancer?

If a person has cancer, a positive genetic test result may help to explain why he or she developed cancer, and clarify if there is an increased risk for other cancers in the future. It can also help to determine if the person's family members have an increased risk to develop cancer.

9. Do people with a positive test result always develop cancer?

No – a positive test result does not mean that people will always develop cancer. Instead, it means those people were born with an increased risk to develop cancer. The exact cancer risk can vary depending on the gene. For the majority of genes tested, there is *not* a 100% chance to develop cancer.

10. If my results are negative, why did I and/or my relatives develop cancer?

A negative test result does not mean a person will never develop cancer. But it does significantly reduce the likelihood he or she was born with a hereditary risk to develop cancer. Even with a negative test result, every person has a baseline risk to develop cancer in his or her lifetime. The risk to develop cancer may be modified by a person's family history of cancer, lifestyle, environmental, and other factors.

11. How long will it take to get my genetic test results?

The results of genetic testing typically take about three weeks to be reported to the ordering provider. For people with cancer whose results are considered urgent for surgical or cancer treatment planning, results can often be reported in two weeks.

12. If I have a genetic mutation, what are the risks to my children?

All of our genes are present in pairs. One copy is from our mother and the other copy is from our father. A person with a mutation has a 50 percent chance of passing the mutation on to each of their children. Both males and females are at equal risk to inherit the mutation.

13. Will this mutation skip a generation? Do my children need to be tested if I am negative?

Gene mutations do not skip generations. If you do not have a detectable mutation in one of the genes tested, you cannot pass a mutation in one of these genes to your children. If your testing is negative, we would not recommend testing your children.

Because children get half of their genes from their mother and half from their father, your children may still benefit from their own genetic counseling appointment, if there is a significant family history of cancer on the other side of their family. It is also important for your children to discuss the family history of cancer with their providers to receive the most appropriate cancer screening recommendations based on the family history and any outcomes of genetic testing.

14. What are the possible benefits of genetic testing?

Testing may provide reassurance and help reduce anxiety whether the results are positive or negative. A positive result may help to clarify cancer risks for people, their children, and other relatives. This can allow people to plan for the future and gives them the opportunity to develop a personalized cancer prevention and screening plan. In addition, some people with a mutation are interested in family planning options that can prevent the mutation from being passed to their children. A negative result may help to decrease the future cancer risks for a person and his or her children and relatives.

15. What are the possible risks of genetic testing?

There are no unusual risks involved in having blood drawn or collecting a saliva sample for genetic testing. Some people who have genetic testing may experience depression, heightened anxiety or fear. A positive test result may also bring about changes in family relationships. A negative or variant of uncertain significance test result may cause feelings of frustration, confusion, or disappointment because the results did not provide the cause of the cancers in the family or information about cancer risks for the person or his or her family members.

16. Is genetic testing covered by health insurance?

The coverage of genetic testing depends upon whether a person meets his or her insurance criteria for genetic testing. This criteria can vary with each insurance company. In most instances, people with a cancer diagnosis, especially those with pancreatic or ovarian cancer, will meet their insurance criteria for genetic testing. Even if insurance criteria are met, there still may be cost because of copays or deductibles. If you have questions about the coverage of genetic testing, please contact the Yale Medicine pre-estimation team at 203-737-7692.

17. If I test positive for a mutation, can I be discriminated against?

There are laws that provide protection from discrimination based on results of genetic testing. These laws most often provide employment and health insurance protection, but do not apply to life, long-term care, or disability insurance plans. These laws do not provide discrimination protection for people with a cancer diagnosis, but could provide protection to their relatives. Some people consider getting insurance coverage prior to genetic testing. One of these discrimination protection laws is called the Genetic Information Non-Discrimination Act (GINA). For more information on GINA, please visit www.ginahelp.org.

18. Is this genetic test a research study?

No, this genetic test is not a research study. It is performed for clinical purposes. A clinical test means the results may be used for your medical care, management, and/or treatment. The results of this testing are not used for research unless you have specifically signed a separate consent form for research purposes. Your genetic counselor will review research options with you at the time of your genetic counseling visit.

19. How can I schedule a genetic counseling visit, if an appointment has not already been requested?

You can contact the Smilow Cancer Genetics and Prevention Program at 203-200-4362 to learn more information about the process of setting up a genetic counseling appointment.