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Understanding Genetic Testing for Cancer

Genetics, gene mutations, and cancer risk

Genetics is the field of science that looks at how traits (such as eye color) are passed down from parents to their children through genes.

Genes are pieces of DNA (deoxyribonucleic acid) inside our cells that tell the cell how to make the proteins the body needs to function. DNA is the genetic “blueprint” in each cell. Genes affect inherited traits passed on from a parent to a child, such as hair color, eye color, and height. They can also affect whether a person is likely to develop certain diseases, such as cancer.

Changes in genes, called **mutations**, play an important role in the development of cancer. Mutations can cause a cell to make (or not make) proteins that affect how the cell grows and divides into new cells. [Certain mutations can cause cells to grow out of control](#)¹, which can lead to cancer. Still, only about 5% to 10% of all cancers are thought to be strongly related to an **inherited** gene mutation.

Usually several gene changes or mutations are needed before a cell becomes cancer.

Most cancers start because of **acquired** gene mutations that happen during a person’s lifetime. Sometimes these gene changes have an outside cause, such as [exposure to sunlight](#)² or [tobacco](#)³. But gene mutations can also be random events that sometimes happen inside a cell, without a clear cause.

Acquired mutations only affect the cells that grow from the mutated cell. They do not affect all the cells in the person’s body. This means all the cancer cells will have the mutations, but normal cells in the body will not. Because of this, the mutations are not

passed on to a person's children. This is very different from inherited mutations, which are in every cell in the body – even the cells without cancer.

What is genetic testing?

Genetic testing is the use of medical tests to look for certain mutations 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 its use in looking for gene changes linked to cancer.

Genetic testing for cancer risk

Predictive genetic testing is the 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 advised:

- 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 tests to look for cancer early, or even take steps 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](#)⁴ and some other cancers) in a woman whose mother and sister had breast cancer.
- For 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 or if the cancer was diagnosed at a young age). Testing might show if the person has a higher risk of some other cancers. It can also help other family members decide if they want to be tested for the mutation.
- For family members of a person known to have an inherited gene mutation that increases cancer risk. Testing can help them know if they need 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 there's a cancer that may be inherited (see below).

Testing cancer cells for genetic changes

Sometimes after a person has been diagnosed with cancer, the doctor will do tests on a

sample of cancer cells to look for certain gene changes. These tests can sometimes give information on a person's outlook (prognosis) and help tell whether certain types of treatment might be useful.

These types of tests look for acquired gene changes **only** in the cancer cells that are taken from the patient. 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 our information on [specific types of cancer](#)⁵.

Who should have 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 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](#)⁶, [ovarian](#)⁷, and [pancreatic cancers](#)⁸ in your family).
- 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 family member with a rare cancer, such as breast cancer in a male or retinoblastoma
- Ethnicity (for example, Jewish ancestry is linked to ovarian and breast cancers)
- 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

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. Other

cancer types may have known mutations, but there's no way to test for them yet.

[Family Cancer Syndromes](#)⁹ gives you more information on the types of cancers that may be linked to inherited genes.

Next steps

It's important to find out how useful testing may be for you **before** you do it. Talk to your health care provider and plan to meet with a genetic counselor before the actual test. This will help you know [what to expect](#)¹⁰. The counselor can tell you about the [pros and cons of the test](#)¹¹, what the results might mean, and what your options are.

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](#)¹² or genetic counseling that may be recommended by a medical professional based on your risk for cancer.

Hyperlinks

1. www.cancer.org/cancer/cancer-causes/genetics/genes-and-cancer.html
2. www.cancer.org/cancer/cancer-causes/radiation-exposure/uv-radiation.html
3. www.cancer.org/cancer/cancer-causes/tobacco-and-cancer.html
4. www.cancer.org/cancer/breast-cancer.html
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9. www.cancer.org/cancer/cancer-causes/genetics/family-cancer-syndromes.html
10. www.cancer.org/cancer/cancer-causes/genetics/what-happens-during-genetic-testing-for-cancer.html
11. www.cancer.org/cancer/cancer-causes/genetics/should-i-get-genetic-testing-for-cancer-risk.html
12. www.cancer.org/healthy/find-cancer-early.html

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Rosenberg SA, eds. *DeVita, Hellman, and Rosenberg's Cancer: Principles & Practice of Oncology*. 10th ed. Philadelphia, Pa: Lippincott Williams & Wilkins; 2015.

National Cancer Institute. Genetic Testing for Hereditary Cancer Syndromes. April 11, 2013. Accessed at www.cancer.gov/about-cancer/causes-prevention/genetics/genetic-testing-fact-sheet on March 11, 2016.

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