



PATIENT EDUCATION

*Understanding Your Genetic
Test Results*

EDUCATION
learning EXCELLENCE
HEALTHY CARING
LIVING INTERACTION

BARBARA WOODWARD LIPS
PATIENT EDUCATION CENTER

About Your Results

How will you get your results?

The amount of time it takes for you to get your test results depends on the type of testing you had done. The testing lab will send your results to your health care provider. Ask your health care provider when and how you may expect to get your results.

Positive results

If your genetic test result is positive, it means that the test did find a mutation in the gene(s) tested. Figure 1 shows an example of how small a mutation may be.

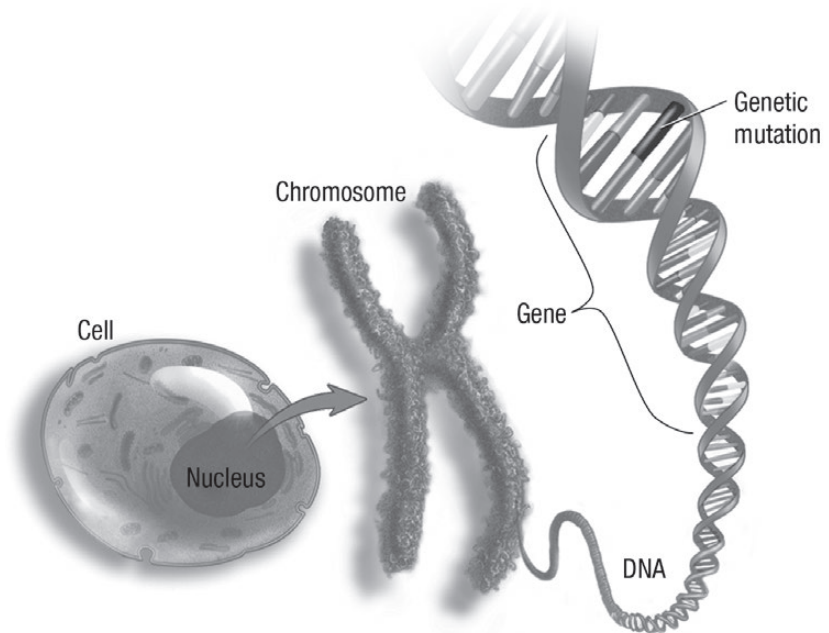


Figure 1. Each cell has chromosomes in its nucleus. Chromosomes are long chains of DNA. Your DNA is made up of many genes.

The steps you take next depend on why you were tested.

If you want to learn whether you carry an altered gene that could cause a disorder in your child: Your health care provider or a genetic counselor can help you figure out a child's risk of the disorder. A positive test result may also help you and your partner make decisions about how you want to plan for a family.

If you were tested to find out whether you might develop a certain disorder: A positive result may diagnose a disorder you have now or one that you will develop in the future. Knowing this could help you and your health care provider make plans about your treatment. Huntington's disease is an example of a disorder that usually develops when a person is an adult.

However, a positive test doesn't always mean you will get that disorder. For example, if you have a mutation in a breast cancer gene, such as the BRCA1 or BRCA2 gene, it means you are *at high risk* of having breast cancer at some point in your life. But it doesn't mean that you *will get* breast cancer.

Negative results

If your genetic test result is negative, it means that *the test did not find* a mutation in the gene(s) tested. But a negative result doesn't mean that you *don't have* a mutation.

The genetic test results may not be correct, or accurate. Result accuracy depends on what condition is being tested and whether one of your family members has the genetic mutation. Genetic testing may not be able to detect all genetic mutations.

In addition, a negative result does not mean you will never get the disorder. For example, people who don't have a mutated breast cancer gene, such as the BRCA1 or BRCA2 gene, can still get breast cancer.

Inconclusive results

"Inconclusive" means that the test did not give clear results. An inconclusive result cannot confirm or rule out a diagnosis.

Everyone has differences, or variations, in the way their genes appear. These variations are called polymorphisms. Often these variations don't affect your health. But it could be hard for your health care team to figure out the difference between a gene mutation that causes a disorder and a gene mutation that is harmless. You or other family members may need more testing.

Next Steps

If you have positive test results

- You *may* be able to make some lifestyle changes that could lower your risk of getting a disorder. This may be true even if the mutated gene makes it more likely to get the disorder. However, this is not true for every disorder.
- More testing may be helpful for you.
- Surgical treatment may be an option for some disorders.
- You may be able to make decisions now about insurance coverage.
- You may choose to help with research or join registries for your genetic disorder, if they are available. These options may help you learn news about preventing or treating a disorder.
- Family members who may be at risk of a disorder may want to be tested.

If you have negative test results

- If you test negative for a gene mutation that you know a family member has, you did not inherit the genetic mutation. Your risk of developing that disease should be the same as that of most other people.
- For some genetic disorders, a negative result may not rule out a diagnosis of the disorder. Talk to your health care provider about how to understand your test results.

If you have inconclusive test results

- Inconclusive results can be difficult to understand.
- Medical decisions have to be made while keeping in mind many different issues.
- Talk to your genetics team about how to understand your test results.

For More Information

For more information, check out the websites of the following organizations:

- U. S. National Library of Medicine
- National Society of Genetic Counselors

Your health care provider does not sponsor or endorse these websites and cannot guarantee the accuracy of the information on these sites. Websites should not be used to diagnose or treat health conditions.

If you have any questions about the information in this material, call your health care provider or genetic counselor.

Notes

Notes

BARBARA WOODWARD LIPS PATIENT EDUCATION CENTER

Mrs. Lips, a resident of San Antonio, Texas, was a loyal patient of Mayo Clinic for more than 40 years. She was a self-made business leader who significantly expanded her family's activities in oil, gas and ranching, even as she assembled a museum-quality collection of antiques and fine art. She was best known by Mayo staff for her patient advocacy and support.

Upon her death in 1995, Mrs. Lips paid the ultimate compliment by leaving her entire estate to Mayo Clinic. Mrs. Lips had a profound appreciation for the care she received at Mayo Clinic. By naming the Barbara Woodward Lips Patient Education Center, Mayo honors her generosity, her love of learning, her belief in patient empowerment and her dedication to high-quality care.

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