



With genetic testing, there are generally three different results that we can get: a positive, a negative, or a variant of uncertain significance (VUS).

Positive

A positive result means that a harmful change (called a pathogenic variant) was found in a gene that causes that gene to not work properly. The health issues that are caused by a pathogenic variant determine what gene it is found in. For example, pathogenic variants in the [BRCA1](#) or [BRCA2](#) genes can cause an increased risk for [hereditary breast and ovarian cancer](#), while pathogenic variants in genes that help build your red blood cells can cause [inherited types of anemia](#). A positive result can also mean that the lab found a likely pathogenic variant, which is a change that probably makes the gene not work correctly, but more research is needed to say for sure.

Pathogenic variants can also travel in families in different [inheritance patterns](#), so finding out which gene is causing a particular health problem in a family may help to determine what other family members may be at risk and should also consider genetic testing.

A specialist in genetics, such as a genetic counselor, can help you understand how a positive genetic testing result may affect your health, and how it may affect the health of family members.

Negative

A negative result means that no harmful changes were found in any of the genes that were looked at. What a negative genetic testing result means depends on what the test was for, as well as the person's medical and family history. Negative test results may also sometimes list gene changes that are benign (not harmful) or likely benign (probably does not change how the gene works, but more research is needed to say for sure).

If there has previously been a pathogenic variant that was identified in the family that someone was tested for ([familial pathogenic variant](#)), then we would call the result a *true negative*. That means that we know what the specific pathogenic variant is that caused the medical condition in their family, and that they did not inherit it.

- An example of this is if your father is found to carry a pathogenic variant that is linked to an increased risk for [colon cancer](#), and you test negative for that [variant](#), then we would call that a true negative. Because we know what contributed to the [colon cancer](#) in your family and that you did not inherit it, you would likely have the same risk for



[colon cancer](#) as anyone else in the general population.

If there has not previously been a pathogenic variant identified in the family, we would call the result an *uninformative negative*. That means that there is a significantly decreased, but not zero, chance that there is not a pathogenic variant in the genes that were tested. The reason that we cannot say there is zero chance for a pathogenic variant in these genes is that our technology is not perfect, and our understanding of genetics is constantly evolving. A negative genetic testing result can mean different things depending on what the testing is looking for, your medical history, and your [family history](#).

A negative genetic testing result should always be put into context given the medical and [family history](#) to help make the most sense out of it. A healthcare professional with a background in genetics, such as a genetic counselor, can help to do that.

Variant of Uncertain Significance (VUS)

A variant of uncertain significance (VUS) means that a change was found in one of the genes that the lab was looking at, but there is not enough information on that specific change to know for sure whether it is just a normal variation (we all have many of these), or whether it is a change that damages the gene and causes a genetic condition or an increased risk for a genetic condition.

When the lab finds a VUS, they continue to do more research to try to determine whether that specific change is harmful or not. It can sometimes take months or even years for them to figure that out. Most VUS results are ultimately found by the labs to be not be harmful, so many times we will treat this as a tentative negative until we get more information about them.

Getting a VUS result can sometimes lead to more feelings of uncertainty, stress, and anxiety. A healthcare professional with a background in genetics, such as a genetic counselor, can help to put a VUS result into context given your personal medical and family history.

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