

There are actually two different type of negative test results when it comes to hereditary cancer genetic testing: a true negative and an uninformative negative.

An uninformative negative rules out all mutations detectable by the particular test performed. The lab screens for mutations by looking for harmful spelling errors in the gene (by sequencing) or missing or extra letters (by deletion/duplication studies). An uninformative negative means that the lab looked through all of the letters in the gene(s) that were tested and did not find any mutations (harmful spelling errors or missing/extra letters). An uninformative negative genetic test result means that the chance for a strongly genetic cause for the cancer in your family is likely low but not zero.

With an uninformative negative genetic test result, it is important to interpret that result in the context of the family history. It is possible that a gene mutation is responsible for a family history of cancer, but it was just not passed down to the individual that was tested. It is also possible that there could be a mutation in the genes that were examine that cannot be found by our current technology. New genes related to an increased hereditary cancer risk are continually being discovered, so there is the potential that there could be a gene that is increasing the risk in a family that we just have not found yet.

Because of all of this, even if someone has an uninformative negative genetic test, if they have a strong family history of cancer they still may have an increased risk. If genetic testing is negative, we determine someone's risk as well as screening and management guidelines based on their family history.

If there is already a known mutation in the family and testing does not identify that mutation, this is considered a true negative result, which rules out the increased cancer risks associated with that known mutation. Unless other, unrelated risk factors are identified in the history, screening and management recommendations are based on general population guidelines. Further, any children of an individual with a true negative result are not at risk to inherit the mutation and testing for them is not necessary (unless they have a suspicious history of cancer on the other side of their family).

Click <u>here</u> to learn more about scheduling a genetic counseling appointment for questions about hereditary cancer predisposition.



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## • Positive Result

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## • VUS Result

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