

A variant of uncertain significance (VUS) is when the lab found a spelling change (by sequencing) or a missing or extra piece (by deletion/duplication studies), but are not entirely sure what it means.

Most of the time, researchers get more information about the VUS and find that it is not linked with an increased risk for cancer. The majority of VUS findings are spelling errors or extra/missing pieces in the gene that don't change the instructions, and thus do not affect the protein that the gene makes.

An example would be the sentence A CAT RAN. A VUS would be similar to if the sentence said A CAT RUN instead of A CAT RAN. The spelling error that changes RAN to RUN makes the sentence not make grammatical sense, but it doesn't change the overall meaning of the sentence.

In some cases, research studies can be offered to family members to help determine whether the spelling error or missing/extra piece is harmful or not, but it can take months to years to reach more definitive answers. Due to the uncertainty of a VUS result, testing of other family members for the variant is not usually recommended outside of research studies. Until the result can be clearly interpreted as a 'positive' or 'negative', cancer risk and screening would be based on the individual and/or family history of cancer.

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Positive Result

A positive result (may also be called 'mutation detected') confirms inherited risk for cancer in the person who was tested. A mutation means that the lab found either a spelling error (by sequencing), or missing or extra letters (by deletion/duplication studies) that we know make the instructions for that gene...

Negative Result

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...