Test result FAQ: Variant of uncertain significance

Your genetic testing revealed a variant of uncertain significance in one or more of the genes tested. Here are some answers to the most commonly asked questions about this type of test result:

What is a variant of uncertain significance?

Everyone has differences in their DNA, and most of those differences are not related to disease. Genetic tests commonly identify DNA differences that have an unknown impact on disease risk. These are called variants of uncertain significance. In a small number of cases, future research may determine that a variant of uncertain significance increases the risk of a disease. In most cases, such variants are later found to be unrelated to disease risk. Genetic medicine is advancing rapidly and your doctor will be updated as new clinically relevant information becomes available.

How will this genetic test result impact my medical care?

The presence or absence of a variant of uncertain significance should not be used to guide your medical care because the effect of such a variant is unknown. Decisions about your medical care should be based on your personal and family medical history.

Does this test result rule out an inherited cancer syndrome in my family?

No, it does not. It is possible that your family has an inherited cancer predisposition syndrome, but that you did not inherit the genetic mutation in your family. It is also possible that the cancers in your family are due to a mutation in a gene or a portion of a gene not included in this testing, perhaps in a gene that is not yet known to be associated with hereditary cancer syndromes. Your family history of cancer may also be due to factors that aren't inherited.

What is the chance that I will develop cancer? If I have had a cancer diagnosis, what it the chance that I will develop cancer again?

An individual's cancer risk and medical management are not determined by genetic test results alone. Overall cancer risk assessment incorporates additional factors, including personal medical history, family medical history, and any available test results. Your physician can help devise a personalized plan for cancer surveillance and prevention.

What does this genetic test result mean for my family members?

Although your genetic testing did not identify any disease-causing mutations, this result cannot necessarily be applied to your family members. In most cases, testing your family members for this variant of uncertain significance is not indicated. If it is indicated, your physician will be notified on your test report. In addition, your relatives should seek their own personalized cancer risk assessment since in some cases comprehensive genetic testing may be indicated for them for other reasons.

What are my options to learn more about my genetic test result?

Patients may call Invitae's genetic counselors at 800-436-3037 anytime during business hours to briefly review their genetic test results (after they are released by the ordering clinician) or schedule a post-test comprehensive genetic counseling session.

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