When genetic testing is done, it is common to hope for, and sometimes expect, a specific answer to the questions:

- “Am I at risk for developing a serious illness?”
- “Do I have a genetic cause to my illness?”
- “Are my relatives at risk for inheriting this illness?”

Genetic testing cannot always provide a specific answer. We all have some differences in our genes that make us unique. These differences are called ‘variants’.
Every cell has chromosomes, which contain your genetic information (DNA). DNA is made from 4 ‘letters’ (A, T, C and G) that code for your body’s instructions. These ‘letters’ are grouped together in different ways to form genes.

Your body has approximately 20,000 genes. Genes are sections of DNA that give your body instructions for a specific trait or characteristic, such as blood type or eye color. The ‘letters’ which make up a gene have an expected order or sequence.

FOR EXAMPLE:

Expected order of part of a gene might be: A-G-C-T-T-A-G
A variant in that part of a gene might be: A-G-C-T-A-A-G

Some genetic variants are harmless. Others may cause a specific health problem. In many cases, however, there are variants that aren’t understood yet. These are known as a variant of uncertain significance or VUS. Thus, a VUS is a variation in a genetic sequence whose association with a disease risk is unknown.

IS A VUS GOOD OR BAD?

A VUS is neither good nor bad. Its meaning is just not known yet. Your doctor or genetic counselor can help you understand what the results might mean for you and your family.

As the study of genes continues, the meaning of your genetic variant may become known. It is important to follow up yearly with the health care provider who ordered your genetic test because new information about your genetic variant may be available for you in the future.

WHAT DOES THIS MEAN FOR MY MEDICAL CARE?

Your medical care will be managed based on your personal medical and family history. Your VUS result does not have any role in making your current medical decisions.

Because science is changing fast, it is very important to follow up with your health care provider to find out if more has been learned about your genetic variant.

NOTES:

If you have questions about this information, you may talk with your health care provider.

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