Variant of Uncertain Significance (VUS)

Your Child's Genetic Test Result

VUS is a test result that means your child has a genetic change, but it is unclear how it affects your child's health.

What are genetic tests?

Genetic tests are a way to look for specific changes (mutations) in a person's genetic information. Genetic information tells a person's body how to grow and function. "Genes," "DNA," and "chromosomes" are all genetic information. All people have genetic changes, which makes people different. Some changes can have more of an impact on health and development.

What kind of genetic tests are there?

There are 3 kinds of genetic tests:

- **Chromosome tests** look at your chromosomes to see if there are any extra or missing pieces or copies. Chromosomes are packages of genes in our cells. If there is anything extra or missing, it is a chromosome disorder.
- **Gene tests**, also called molecular tests, look at individual or groups of genes for mutations or "spelling errors" that could lead to a specific genetic disorder.
- **Biochemical tests** measure the amount of proteins in your blood. Proteins are made using the instructions from our DNA. These tests can show if changes in the DNA are causing the body to make too much or too little of a protein. Having just the right balance is important.

What does the result of the genetic test mean?

- **Normal/Negative**: This result means that the test did not find any changes in genes that were tested. This could mean that your child is not affected by a particular disorder, does not have an increased risk of developing a certain disorder, or is not a carrier of a specific genetic mutation. However, one kind of test cannot look for every possible genetic change, so it is possible that your child may need additional testing to look at other genes.
- **Abnormal/Positive**: This result means that a change was found in your child's genes. This usually will diagnose a genetic condition, confirm that your child is a carrier, or identify an increased risk of developing a disease.
- **Variant of uncertain significance (VUS)**: This means that the test found a genetic change, but there is not enough known about the change to give a diagnosis. In some cases, testing other family members can help us better understand what the genetic change means. It is more common to find this test result when testing multiple genes at once (panel test).

To Learn More

- Medical Genetics 206-987-2056
- Ask your child's healthcare provider
- www.seattlechildrens.org

Free Interpreter Services

- In the hospital, ask your child's nurse.
- From outside the hospital, call the toll-free Family Interpreting Line 1-866-583-1527. Tell the interpreter the name or extension you need.

Does the VUS affect my child's health?

Not necessarily. There are many changes that do not negatively impact a person's health (benign). Humans have more than 50 million differences, some of which are responsible for normal differences, like how we look.

In some cases, the change is associated with a disorder (pathogenic). A VUS can be frustrating and worrisome because it is unclear how that genetic change affects your child. Even though a VUS may not provide a clear answer, there are still steps that you can take with your child's healthcare team.

How should a VUS be treated?

Because it is not clear how a VUS affects your child, treatment should be based on their personal and family history. Scientists are doing research on genetic changes in other people to help understand how they are associated with genetic disorders.

Your child's healthcare provider is the best way to learn if more information is found about your child's genetic change, so we encourage you to stay in touch over the years by calling or scheduling an appointment.

Why should parents be tested for a VUS found in their child?

Sometimes we recommend genetic testing for parents because it is a way to learn more about how a VUS affects your child's health. The testing allows us to see if the change is inherited and if the VUS might be the cause:

- If a parent has the same VUS, but does not have the same symptoms or disorder, the change is probably not the cause of a disorder (benign). However, sometimes a parent shows fewer signs of a condition than a child.
- If a parent has the same VUS, and has the same symptoms or disorder, it is likely that the change is the cause of a disorder (pathogenic).
- If neither parent has the same VUS, and it is a new change in your child (de novo), it is likely that the change is the cause of the disorder (pathogenic).

Insurance pre-authorization

Genetic tests are often expensive and might not be covered by insurance. Without pre-authorization, you may have to pay for the test yourself. To learn more, read our handout "Insurance coverage for Genetic Testing" www.seattlechildrens.org/pdf/PE2051.pdf.

Genetic Counseling

A genetic counselor is available to talk about this with you. Contact the Medical Genetics clinic to speak to a genetic counselor or schedule an appointment at 206-987-2056.

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