Information Sheet

Genetic Testing Panels

In this handout:

- What is a genetic testing panel?
- What does a testing panel look for?
- Which genes are included on a panel?
- What kind of results could I get?

What is a genetic testing panel?

A genetic testing panel is a laboratory test that looks at a select group of genes. The test lets us study many genes at one time using just one blood or saliva sample. A panel is ordered when more than one gene can cause the same symptom or condition. A genetic testing panel is carefully selected by your Genetics provider based on your medical history and your family history.

What does a testing panel look for?

A testing panel looks for differences in the DNA called genetic variants. The result shows whether or not a harmful genetic variant is found in any of the genes on the panel. Harmful variants are also called “pathogenic variants” or mutations.

Which genes are included on a panel?

Testing panels may include just a few genes to several hundred genes. The exact genes included on a panel depend on many things, such as:

- Why a test is ordered
- Which laboratory does the test
- When a sample is tested

Limits of panel testing

- **Testing panels do not include every gene that could cause problems.** There may be genes related to your condition that are not on the panel or have not been discovered yet. Testing panels are updated as information about genes changes.
- **Panels may include genes with a lower risk for medical problems.** Not all genes have the same risk for medical problems. Some genes may be less likely to cause problems than other genes on the same panel.
- **Panels may include genes with limited information.** Many genes on a testing panel have been seen in lots of people and have well-defined risks. However, some genes on a panel may not be well understood yet.
- **Testing more genes does not always mean better information.** The chance for a confusing or unclear result is often higher when a panel includes a large number of genes.

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DID YOU KNOW?

Testing sometimes finds more than one genetic variant in a person.

What kind of results could I get?

The test will show one of the following results:

Pathogenic (harmful) variant: This result means a variant was found that makes a gene work poorly or not work at all. A pathogenic variant can confirm a genetic diagnosis or identify a genetic risk for medical problems. Once a pathogenic variant is found, family members can be tested for the same variant. Sometimes a variant gets called "**likely pathogenic".** This means the variant is expected to be harmful but there is a small chance that it might not be.

Variant of uncertain significance or VUS: This result means a variant was found, but it is not clear if the variant changes the way the gene works. There are many differences in DNA that are found in healthy people. When a variant has not been seen before or has only been found in a few people, it can be hard to know if it will cause problems. A VUS result does not confirm a diagnosis or determine medical risk. Decisions about medical care will continue to be made based on the medical history and family history, rather than the test result.

Our understanding of a VUS can change as we learn more. Most will be found to be harmless, but some will be found to be pathogenic.

Negative: This result means no pathogenic variant or VUS was found in any of the genes on the panel. It is still possible to have a pathogenic variant even with a negative result. A condition could be due to a gene that is not on the panel or a variant that is not found by the test. That means the cause for some genetic conditions can be missed by panel testing.

More about GENETIC VARIANTS

Everyone has genetic variants. Having genetic variants is normal and most variants are harmless. These are called "benign variants". Benign variants do not affect your health and are not reported by the laboratory.

Not all variants are inherited. New variants happen in every generation, so each person has variants that are unique. There are also variants that are unique to your family. Most of these do not affect your health.

Some variants are not found by the test. The test is designed to find most variants, but certain DNA changes are hard to find with current testing methods.