What you need to know about Whole Exome Sequencing (WES)

Whole exome sequencing (WES) tests the active parts of genes called exons. WES looks for genetic variants in the exons of most genes. Variants are differences in the DNA code (sequence) that might affect the way a gene works. A single blood sample studies all the exons at the same time. Results can take two months or longer. WES covers most human genes, but some genes are hard to study with this test.

**What can I learn from my WES test result?**

Your test result will let you know about:

- Variants that may explain your medical condition
- Variants in genes known to cause medical conditions we can treat
- Variants in both copies of a recessive gene
- New variants not seen in either parent (when parent samples are included)
- Variants that reveal a genetic carrier status (mainly used for pregnancy planning)

**POSITIVE**
Pathogenic variant OR likely pathogenic variant

A variant was found that is expected to change the way the gene works. This result may help explain your medical condition or tell you about your risk for a genetic disease.

**NEGATIVE**
No variant detected

No variants were found that help explain your medical condition. There were also no variants found in genes known to cause treatable conditions.

**Variant of UNKNOWN Significance (VUS)**

A variant was found in a gene related to the reason for testing, but it is not clear if the DNA change leads to disease. It may be a harmless change in the gene, but there is not enough information to decide either way.

**LIMITS OF WES**

- Testing does not include all exons or all genes.
- Some types of DNA changes cannot be found with WES
- Many parts of the DNA are not tested
  - Including the noncoding parts of a gene (introns) and most of the DNA between genes.
- Certain genetic diseases cannot be tested using WES

**What is NOT included in the test result?**

The following variants are usually not included in the test report:

- Harmless variants (those seen before in healthy people)
- Variants in genes unrelated to the reason for testing
- Variants in genes not yet known to cause a genetic condition
- Variants in genes that affect the response to certain drugs (depending on the lab, these may be included)

You may be able to ask for some of this information by signing a separate consent form.