What you need to know about

Exome Sequencing

BEFORE you go to the laboratory

This handout covers:

- What ES looks for
- What you can learn from ES
- What is not reported
- Limits of ES

Exome Sequencing (ES)

Exome sequencing (ES) looks for genetic variants in DNA that can cause health problems, birth defects, or developmental disabilities. This test studies the active part of genes called **exons**. Testing is usually done on a blood sample. A single sample looks at exons from thousands of genes at the same time. Results can take two months or longer. ES covers most human genes, but some genes are hard to study with this test.

Did you know?

Everyone has variants. The average person has millions of genetic variants in their DNA. Variants are what make you unique and most variants are harmless.

What can I learn from my test result?

Your test result may include:

- Variants that could explain your medical condition
- New variants not found in either parent (when parent samples are included)

Some test results also include:

- Variants in genes that cause medical conditions we can treat (called incidental findings or secondary findings)
- Variants that reveal a genetic carrier status but do not cause health problems (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 (if this was included).

? 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 ES

- This test does not include all exons
- This test does not study all genes
- Some DNA changes cannot be found by this test
- Some genetic diseases cannot be diagnosed with ES
- Most of a person's DNA is not tested
  - Non-coding parts of a gene (introns)
  - DNA between genes

What is NOT reported in the result?

Most variants are not reported in the result. Since every person has so many variants, the laboratory focuses on genes related to the current diagnosis. The following variants are usually NOT included:

- Harmless variants (those seen before in healthy people)
- Variants in genes not known to cause a genetic condition
- Variants in genes that affect the response to certain drugs
- Variants in genes that can cause conditions with no treatment

Talk with your genetic specialist to learn more about which results to expect with your test.