



Baylor  Miraca  
Genetics Laboratories

# — ONE TEST — MORE ANSWERS

WHOLE EXOME SEQUENCING (WES)

**Y**our health care provider has advised you to have genetic testing on your DNA or your child's DNA. The specific genetic test explained by this brochure is called Whole Exome Sequencing, also known as WES. WES is a new test that has recently become available to patients who are searching for a unifying diagnosis for multiple medical issues.

Genetic testing examines a person's genetic material, also known as DNA. Our DNA is like our body's instruction manual. DNA tells our cells what type of cell to be and how to act. Scientists can study a person's DNA and identify changes that are different from standard human DNA. Sometimes these differences are significant enough that they cause medical problems.

Some genetic tests may have already been performed on your DNA or your child's DNA. These tests may not yet have identified a diagnosis. Your health care provider has recommended WES because he or she thinks that WES may identify a change that explains your or your child's medical concerns.

## WHOLE EXOME SEQUENCING (WES)

Older technology used in genetic testing could only look at one gene at a time in the body's instruction manual. Whole Exome Sequencing is a technology in which scientists look very carefully at a person's genes, or genetic information, for changes that may have caused a genetic disorder. Our genes are made up of sections of DNA called introns and exons. Exons are part of our DNA that code for proteins. Proteins give our cells instructions on what type of cells to be and how to act. All the exons in all our genes together are called the Exome.

Think of genes like sentences in the instruction manual for our bodies. Scientists know what the sentences should say. Sequencing is a technology that looks at each letter in



every sentence of DNA in the exome, allowing scientists to compare the DNA of a person who has medical issues to the DNA from a person without medical issues.

In summary, Whole Exome Sequencing can look at tens of thousands of portions of genetic material, all at the same time. But this technology still can't see all genetic changes that may lead to genetic conditions, which is why your doctor may order more than one type of test.

## RECOMMENDING WES

Your health care provider may have recommended WES because no other testing has identified a diagnosis. Or, your health care provider may have recommended WES as a first attempt to uncover a diagnosis. WES does not identify all genetic changes that can cause medical issues, but it is considered to be a powerful tool.

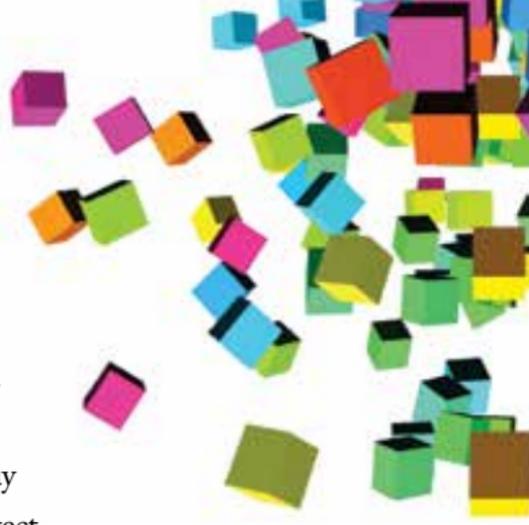
## POSSIBLE RESULTS

You and your health care provider should discuss what types of information you and your family want to receive. Your health care provider can discuss the options in more detail as some aspects of WES are customizable.

### Positive Results

Positive result or “abnormal” results mean that there is a change in genetic material. Your health care provider will discuss what this means for you or your child, and for other family members.





## Negative Results

Negative or “normal” results mean that no genetic change could be detected using WES. This does not mean that there is no genetic change, but it may mean that WES could not detect it. Your provider may decide to order additional testing.

## Results of Unclear Significance

WES can detect a change DNA that does not have a clear meaning. Every person has changes in their DNA; not all of these changes cause medical issues. Your health care provider will discuss this in more detail with you.

## Incidental Findings

This test can sometimes detect a change in a person’s DNA that is unrelated to the reason that the sample was sent for testing. If this change is of medical significance, it is called an incidental finding. Possible incidental findings include mutations that increase a person’s risk for cancer or for heart disease. Your health care provider will discuss the possibilities with you in more detail.

Information regarding adult onset dementia syndromes, such as Alzheimer’s disease, or other adult onset neurological conditions will not be reported.

## NEXT STEPS

WES takes many weeks to complete, but scientists are working on upgrades to the technology to make it quicker. Ask your health care provider for more information on when results are expected.

## OTHER QUESTIONS

The information in this brochure should be used as a supplement to a discussion with your health care provider. If you have questions about this test, or about genetic testing, contact your doctor or your genetic counselor.



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