GeneDx

Exome testing for pediatric patients

Including developmental delay, intellectual disability, autism spectrum disorder, and epilepsy

What is genetic testing?

Genetic testing looks at our genes, which are the instructions that tell our bodies how to develop and function. Sometimes, changes in our genes (also called genetic variants) cause our bodies to grow or develop differently than expected. Genetic testing may be able to find these gene changes. That knowledge can help determine:

- how to best manage or treat a condition
- what to expect for the future
- which additional resources and support may help

What is an exome?

Our exome is the group of all the pieces of DNA that tell our bodies how to make proteins, which are important for our body to function properly. Exome testing looks specifically at exons to try to find the genetic change that may be responsible for differences in how your body functions or developed.

Genetics glossary

DNA is the instruction manual that tells our bodies how to develop and function.

Genes are the individual instructions in the manual that tell our bodies how to make proteins. They are made of two types of pieces:

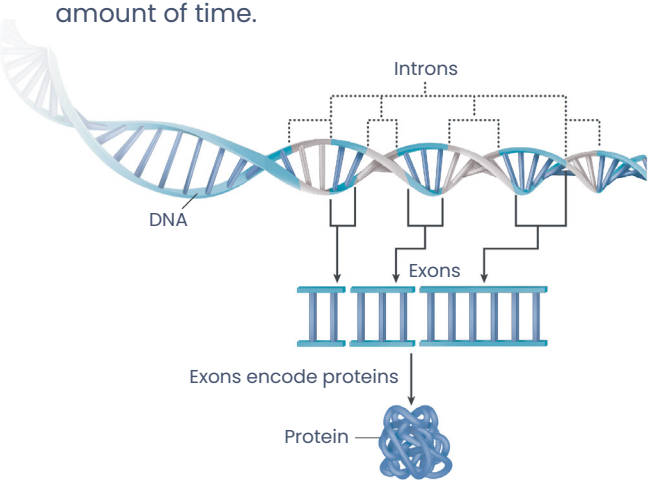
- **Exons** are the pieces that come together to tell the body how to make proteins.
- **Introns** are regions between exons that are cut out when proteins are made.

Proteins are the building blocks that make our body work. They make up our muscles, skin, hormones, and many other chemicals that allow our bodies to function.

Why is my provider recommending exome testing?

Many conditions can have similar symptoms, so your healthcare provider may have recommended a genetic test to try to identify an underlying cause for your health concerns.

Because exome testing looks at over 20,000 genes at once, it may help your healthcare provider diagnose a specific disorder or develop a more effective care plan in a shorter amount of time.



Why did my healthcare provider suggest testing family members, too?

Testing three biologically related family members and comparing the results to a person's DNA is called "trio testing." Samples from biological parents can provide the most information. However, any blood-related family member can contribute.

Trio testing is valuable because it:

- increases the chance of finding the gene change causing the symptoms
- decreases the chance of unclear or uncertain findings

What are the possible test results?

Genetic testing can deliver three types of results:

- **Positive or diagnostic** means we found a gene change that's known to cause symptoms or a specific genetic disorder.
- **Negative or non-diagnostic** means there were no gene changes identified at the time that explain a health condition, based on current knowledge. In this case, your provider may order follow-up testing.
- **Uncertain** means we found a gene change but, based on the available scientific evidence, we cannot clearly say whether this is related to a health condition. In this case, your provider might suggest additional evaluations or a future reanalysis of your genetic information.

Genetic changes may also be identified that are unrelated to the reason your provider recommended testing, known as "secondary findings." This information is optional to receive and occurs in ~3% of people. We encourage you to discuss this option with your provider.

This video explains the genetic testing process and possible results.

GeneDx.co/patient-video

What can I expect during the genetic testing process?

1. Your healthcare provider will determine the appropriate test and gather relevant medical records and family information.
2. A blood sample, cheek swab, or other specimen is collected and sent to GeneDx.
3. Our lab will receive and analyze the sample(s).
4. Your provider will receive a report from our genetic experts with an explanation of what we found.
5. Your provider will share the results with you, and together you will determine the next steps.
6. Based on the results, your provider may also suggest you talk to a genetic counselor, a healthcare professional who can help you better understand what the testing revealed.

The GeneDx difference

We have over 20 years of expertise in diagnosing rare disorders and diseases, and we are dedicated to providing clear, accurate, and meaningful genetic information to help guide healthcare decisions.

Trusted, expert support at every step

Our genetic experts have a common goal: to provide you and your healthcare provider with the information you need.

Our customer support team is also available to help answer any questions you and your family may have throughout the genetic testing process.

Call us directly at **888-729-1206, option 3,** or email Support@GeneDx.com.

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