Rapid genome testing for neonatal & pediatric patients

GeneDx

What is genetic testing?

Genetic testing looks at our genetic information, which tells our bodies how to develop and function. Sometimes, changes in our genetic information (also called genetic variants) cause our bodies to grow or develop differently than expected. Genetic testing may be able to find these changes.

What is a genome?

The genome refers to a person's entire set of genetic information, called DNA, which tells their body how to develop and function. Genome testing is the most comprehensive genetic test available that looks at all of a person's DNA.

What types of genetic changes can be identified by genome testing?

There are many different types of changes that can be found in our genetic information, including spelling mistakes, extra or missing pieces of DNA, or even pieces of DNA that have moved to a different location in the genome. Specific genetic changes can cause specific health conditions, so knowing if there is a change, and what that change is, can help determine:

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

Why is my provider recommending rapid genome 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 rapid genome testing delivers the most comprehensive view of an individual's entire set of genetic information, it may help your healthcare provider diagnose a specific disorder or develop a more effective care plan in a shorter amount of time. That, in turn, can improve health outcomes.

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 genetic change(s) 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 genetic change that's known to cause symptoms or a specific genetic disorder.
- **Negative or non-diagnostic** means there were no genetic 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 genetic 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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