GeneD

Rapid Genetic
Testing for
Neonatal
& Pediatric
Patients

Rapid genetic testing has the potential to deliver fast and accurate diagnoses to guide personalized care plans and improve health outcomes for neonatal and pediatric patients in critical care settings.

What is genetic testing?

Genes are the instructions that tell our bodies how to grow and develop. Sometimes, gene changes (also called variants) cause our bodies to grow or develop differently than expected. Genetic testing may be able to find those kinds of gene changes. Using that information, your healthcare provider may be able to diagnose a specific disorder or develop a more effective care plan.

Having a genetic diagnosis can determine how to best manage or treat a condition, what to expect for the future, and also connect you with additional support resources.



short video and learn more about genetic testing with GeneDx genedx.co/nicu-video



Why is my healthcare provider recommending rapid genetic testing?

Many conditions can have similar symptoms but different genetic causes. Utilizing comprehensive genetic testing such as rapid exome or genome testing enables your provider to cast a wider net and increase the chances of getting a genetic diagnosis in as little as 5-7 days.

Your healthcare provider will help determine which test is the best option to identify a genetic cause for your child's symptoms.

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

Testing two or more 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 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 causes symptoms or a specific genetic disorder.



Negative or non-diagnostic means there were no gene changes that explain a health condition. In this case, your provider may order follow-up testing.



Variant of uncertain significance (VUS) 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 at this time. In this case, your provider might suggest additional evaluations or a future re-analysis of your genetic information.

Your healthcare provider will discuss these results with you to help you understand the next steps, provide treatment plan recommendations, or connect you with additional resources.

Secondary Findings

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 these with your provider.

What can I expect during the genetic testing process?

- Your healthcare provider will determine the appropriate test and gather relevant medical records and family information.
- A blood sample, cheek swab, or other specimen is collected and sent to GeneDx.
- Our lab will receive and analyze the sample(s).
- Your provider will receive verbal results within 5-7 days and a full written report within 2 weeks.
- Based on the results, your provider may suggest you talk to a genetic counselor.

With a referral from your healthcare provider, GeneDx provides comprehensive post-test genetic counseling services at no additional cost. If you have questions after speaking with your provider, you can contact our expert team at (888) 729-1206, option 7.

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 Experts

Our genetic experts have a common goal to provide you and your healthcare provider with the information you need for actionable answers. And as a GeneDx customer, you will have access to remote genetic counseling services with our team or one of our trusted partners.



Support at Every Step

Our experienced customer support team is 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.