Genetics in the NICU: quick-start guide

What is genetic testing?

Your baby's doctor may recommend genetic testing to better understand why your baby is experiencing certain symptoms or medical challenges.

Genetic testing looks at DNA, the instructions that help the body grow, develop, and function. Sometimes changes in DNA can help explain why a baby is sick or experiencing health concerns.

Depending on your baby's medical needs, your healthcare team may recommend:

- **Exome sequencing:** Looks for genetic changes in the portion of your DNA that tells your body how to make proteins.
- **Genome sequencing:** Looks for genetic changes across nearly all of a person's DNA, providing the most comprehensive view of genetic information available today.

Both tests can help doctors better understand your baby's health and guide medical care.

Why genetic testing may be recommended

Genetic testing may help doctors:

- Understand why your baby is experiencing certain symptoms[1,2]
- Identify conditions that may not be visible on other tests[3]
- Guide treatment and medical decisions[4]
- Avoid unnecessary tests or procedures[4]
- Connect your family with the right specialists and resources
- Help anticipate future medical needs[4]

For some babies, genetic testing can provide answers. For others, it helps rule out potential causes and narrow the path forward.

Privacy and protections

There are federal and local laws in place to protect your child's privacy and discrimination based on genetic testing results.

- **Genetic Information Nondiscrimination Act (GINA):** GINA is a federal law that helps protect individuals from discrimination based on genetic information in health insurance and employment.
- **HIPAA:** Your genetic testing results are protected health information and are safeguarded under federal privacy laws, similar to all other healthcare information.
- **Data security:** GeneDx uses secure systems and processes to protect patient information and genetic data.
- **Sample Storage:** Your consent forms explain how DNA samples and testing information may be stored and used. Your healthcare team or a GeneDx genetic counselor can answer any questions about sample storage or privacy.

What happens during testing?

Step 1: Sample collection
A DNA sample is collected from your baby, usually through a small blood sample or cheek swab.

Step 2: Parent samples may be requested
Whenever possible, biological parents may also be asked to provide a sample. This is called trio testing.

Including parent samples helps us:

- Increase the likelihood of finding an answer[5-8]
- Reduce uncertain findings[9]
- Decide whether a genetic change was inherited (passed on from mom/dad to baby) or occurred for the first time

Step 3: Laboratory analysis
Genetics experts look at your baby's DNA and compare it with information from known genetic conditions and millions of genetic variants.

Step 4: Results review
Results are reviewed by genetic experts and shared with your baby's healthcare team.

Step 5: Discussion and next steps
Your healthcare team will review the results with you, answer questions, and discuss how the information may help guide care.

When will results be available?

Turnaround times vary depending on the test ordered and your baby's medical situation. **Results may be ready as quickly as 48 hours after the lab has received the sample(s) or up to 5 days.*** Your healthcare team can provide the most accurate timeline for your family based on the test ordered.

Understanding your baby's results

Positive result

A positive result means testing identified a genetic finding that helps explain your baby's condition.

A positive result may help guide treatment, identify specialists who should be involved in care, and provide information about future health needs.

Negative result

A negative result means testing did not identify a genetic explanation based on current scientific knowledge.

A negative result does not mean your baby's symptoms are not real or that there is no underlying cause. It may still help doctors rule out many known genetic conditions and focus on other possibilities.

Variant of Uncertain Significance (VUS)

Sometimes testing identifies a genetic change that scientists do not yet fully understand.

This is called a Variant of Uncertain Significance, or VUS. A VUS is not considered a diagnosis. As scientific knowledge grows, the interpretation of a VUS may change over time.

Secondary findings

Some families may choose to receive information about certain genetic findings that are unrelated to the reason testing was ordered but could be important for future health.

Because some families want this information and others do not, you will be asked whether you would like to receive these findings before testing begins.

What happens if an answer is found?

Your healthcare team will work with you to determine the right next steps for your family. This may include treatment decisions, anticipating future medical needs, connecting your family with specialists and support resources, and providing information for future pregnancies.

What happens if no answer is found?

Even when testing does not identify a genetic explanation, the information can still help guide care by ruling out many known conditions and informing future evaluations as science advances.

Glossary

We know you'll hear a lot of medical terms in your genetic testing journey. This quick guide of terms may help as you have conversations with your care team.

Genetic terms

DNA
The body's genetic instructions

Gene
A section of DNA that provides the body with instructions on how to develop or function

Genome
A person's complete set of DNA

Genetic condition
A health condition caused by changes in DNA

Congenital
Present at birth

Variant
A difference in DNA

VUS (Variant of Uncertain Significance)
A genetic variant that is not yet fully understood

Proband
The person being tested. In the NICU, this is usually your baby

Duo testing
Testing that includes the baby and one biological relative

Trio testing
Testing that includes the baby and two biological relatives

Inherited
Passed from a biological parent to a child

De novo variant
A genetic variant that occurs for the first time in a child and was not inherited from either parent

Positive result
A genetic variant that helps explain a medical condition

Negative result
No genetic explanation identified based on current knowledge

Primary finding
A result related to the reason testing was ordered

Secondary finding
A result unrelated to the reason testing was ordered but potentially important for health

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References: 1. Wenger TL, Scott A, Kruidenier L, et al. SeqFirst: Building equity access to a precise genetic diagnosis in critically ill newborns. Am J Hum Genet. 2025 Mar 6;112(3):508-522. doi: 10.1016/j.ajhg.2025.02.003. **2.** French CE, Delon I, Dolling H, et al. Whole genome sequencing reveals that genetic conditions are frequent in intensively ill children. Intensive Care Med. 2019 May;45(5):627-636. doi: 10.1007/s00134-019-05552-x **3.** Dillon OJ, Lunke S, Stark Z, et al. Exome sequencing has higher diagnostic yield compared to simulated disease-specific panels in children with suspected monogenic disorders. Eur J Hum Genet. 2018 May;26(5):644-651. doi: 10.1038/s41431-018-0099-1. Epub 2018 Feb 16. **4.** Malinowski J, Miller DT, Demmer L, et al. Systematic evidence-based review: outcomes from exome and genome sequencing for pediatric patients with congenital anomalies or intellectual disability. Genet Med. 2020 Jun;22(6):986-1004. doi: 10.1038/s41436-020-0771-z. **5.** Clark MM, Stark Z, Farnaes L, et al. Meta-analysis of the diagnostic and clinical utility of genome and exome sequencing and chromosomal microarray in children with suspected genetic diseases. NPJ Genom Med. 2018 Jul 9;3:16. doi: 10.1038/s41525-018-0053-8. eCollection 2018. **6.** Lee H, Deignan JL, Dorrani N, et al. Clinical exome sequencing for genetic identification of rare Mendelian disorders. JAMA. 2014 Nov 12;312(18):1880-7. doi: 10.1001/jama.2014.14604. **7.** Farwell KD, Shahmirzadi L, El-Khechen D, et al. Enhanced utility of family-centered diagnostic exome sequencing with inheritance model-based analysis: results from 500 unselected families with undiagnosed genetic conditions. Genet Med. 2015 Jul;17(7):578-86. doi: 10.1038/gim.2014.154. **8.** Sawyer SL, Hartley T, Dyment DA, et al. Utility of whole-exome sequencing for those near the end of the diagnostic odyssey: time to address gaps in care. Clin Genet. 2016 Mar;89(3):275-84. doi: 10.1111/cge.12654. 9. Rehm H, Alaimo JT, Aradhya S, et al. The landscape of reported VUS in multi-gene panel and genomic testing: Time for a change. Genet Med. 2023 Jul 30;100947. doi: 10.1016/j.gim.2023.100947.

*Turnaround times are estimates and begin once the sample(s) begin processing at the GeneDx lab and could be extended in situations outside GeneDx's control.