Searching for the cause of symptoms?

Genetic testing can play an important role in identifying the underlying cause of symptoms and guiding next steps in care.

What is genetic testing?

Genetic testing analyzes a person's DNA to identify changes that may explain their symptoms or condition, which plays an important role in guiding next steps.



Approximately 80 percent of rare diseases have an underlying genetic cause.

Many genetic conditions, including epilepsy, cerebral palsy, and autism spectrum disorder, can share similar symptoms while having different underlying causes.

How genetic testing can help your family

Both positive and negative results can provide information for treatments that target the causes of disease rather than just suppressing symptoms. If a genetic diagnosis is found, families can:

- Connect with other families and advocacy groups specific to their condition
- Work with their doctor to create or adjust their treatment plan
- Search for potential clinical trials or therapies based on the genetic diagnosis
- Identify risks to other family members or prepare for future family planning
- Get insurance coverage for medical equipment or therapies

Professional medical societies recommend genetic testing as a first-line option for many neurodevelopmental symptoms, including seizures, global developmental delay, and intellectual disability.

The American Academy of Pediatrics (AAP) recommends ordering exome and genome sequencing—a comprehensive form of genetic testing—as a first-line test for children with global developmental delays and intellectual disabilities.

The **American College of Medical Genetics and Genomics (ACMG)** recommends exome or genome as a first-tier test for individuals with developmental delay, intellectual disability, and congenital anomalies.

The **National Society of Genetic Counselors (NSGC)** recommends exome or genome for all individuals with unexplained epilepsy. This guideline is endorsed by the **American Epilepsy Society (AES)**.

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Understanding your genetic testing *options*



If you are new to genetic testing, terms like exome, panel testing, and genome sequencing can feel overwhelming.

This guide outlines different types of tests to help you feel confident discussing options and next steps with your health care provider.

Screening vs diagnostic testing

When considering genetic testing, it helps to understand the difference between screening tests and diagnostic tests.

Screening tests
look for signs that a genetic condition may be present. They do not provide a diagnosis, but they can indicate whether further testing may be needed. *For example, newborn screening is a type of genetic screening offered to all babies to check for certain rare conditions early, even before symptoms appear.*

Diagnostic tests
(like exome sequencing, genome sequencing, gene panels, or chromosomal microarray) are used when a person already has medical symptoms or conditions. These tests look deeper into a person's DNA to help identify the underlying cause. A diagnostic test is designed to provide answers.

If a genetic cause is suspected, exome and genome testing offer the most comprehensive view, giving you the best chance at a genetic diagnosis.

Insurance coverage can vary based on your plan and where you live, and your health care provider can help guide the decision.

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Every family is unique, and genetic testing can support different needs at different stages. This chart compares common genetic tests to help you understand what each option may offer.

Test type	Description	Use cases	Takeaway
Single gene test	Looks at one specific gene for variants (mutations).	Used when a specific genetic condition is strongly suspected.	Best used when there's a strong clinical suspicion based on symptoms or family history.
Targeted variant test	Looks for a known, gene variant (mutation) that has already been identified in a family member.	Typically used to test a child for a variant identified in a parent or another sibling.	This testing is most valuable when a known familial variant (mutation) exists.
Multi-gene or panel test	Tests several to hundreds of genes at once. Tests the genes that are known to be related to a certain condition or set of symptoms.	Can be used for conditions like epilepsy, developmental delay, autism, neuromuscular symptoms, inherited cancers, etc.	The genes included on panels can vary by lab — some may cover just several genes, while others may cover a few hundred. Not all genes are tested.
Chromosomal Microarray (CMA)	Looks for missing or extra pieces of chromosomes. It does not read the DNA sequence itself for variants (mutations) but instead checks if parts of the DNA are duplicated or deleted.	Used for developmental delay, intellectual disability, autism, or multiple birth defects.	If you or your child gets a negative result from CMA, it doesn't necessarily mean there isn't a genetic cause.
Exome	Looks for genetic variants (mutations) in the portion of a person's DNA that tells the body how to make proteins — that's more than 20,000 genes. The majority of genetic conditions are caused by changes that can be identified with exome testing.	Most highly recommended test for: Developmental delays, intellectual disabilities, congenital anomalies, epilepsy, autism spectrum disorder, cerebral palsy, or muscle/ movement differences.	The AAP recommends starting with exome testing for many pediatric cases due to its comprehensiveness. Your doctor may also order testing on biological family members, which can help increase chances of finding a diagnosis.
Genome	Looks for genetic changes across all of a person's DNA. Including everything that exom testing looks at, as well as the regions of DNA that don't code for proteins.	Most highly recommended test for: Developmental delays, intellectual disabilities, congenital anomalies, epilepsy, autism spectrum disorder, cerebral palsy, or muscle/ movement differences.	The AAP recommends starting with genome testing for many pediatric cases due to its comprehensiveness. Your doctor may also order testing on biological family members, which can help increase chances of finding a diagnosis.
Ancestry traits	These tests provide information about ancestry or non-medical traits (e.g., hair type, lactose intolerance). They are not used to diagnose any condition.	Used for recreational purposes, like understanding ethnicity or inherited physical traits. These tests are not meant for medical decision making.	This is a different type of genetic test that is not recommended for those trying to understand medical symptoms or diagnose a condition.

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Genetic Counseling 101

What is a genetic counselor?

In some cases, your physician may order genetic testing for you, or they may choose to refer you to a specialist called a genetic counselor.

A genetic counselor is a healthcare professional trained in medical genetics and counseling, and skilled at communicating the complexities of genetic testing. A genetic counselor also supports the mental health of the patients and families and can help talk through worries or concerns.

What to expect during a counseling session

During a genetic counseling session, a genetic counselor may:

- Review your personal and family health history
- Explain genetic testing, including its benefits and limitations
- Discuss your results and what they mean
- Provide education, answer questions, and offer emotional support
- Help determine whether other family members should consider testing

After testing is complete, your genetic counselor may:

- Walk through next steps and possible care plan considerations
- Connect you with advocacy resources, research opportunities, or additional support services

Some labs even offer virtual genetic counseling services after you receive your results.

How to find genetic counselor

If you're interested in finding a genetic counselor in your area:

- Use the **'Find a Genetic Counselor'** feature on the National Society of Genetic Counselors (NSGC) website
- **Find telehealth genetic counselors** through companies like Genome Medical or InformedDNA

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You got a *positive* genetic result – what now?

What does a positive genetic test result mean?

A positive genetic test result means that a change was found in your DNA that is known to cause, or increase the risk for, a specific genetic condition. This result may explain symptoms you or your child may already have, confirm a diagnosis, or indicate a higher-than-average risk of developing certain symptoms.

It is common to have a range of emotions after receiving this result, including worry, sadness, relief, or uncertainty. Some individuals may experience anticipatory grief, or benefit from anticipatory guidance to help prepare for what to expect. These reactions are normal, and many people find that, over time, this information helps them feel more prepared and in control of their health or their child's health.

What are the next steps?

After receiving a positive genetic test result, there are a few important steps you can take to better understand what it means and what to do next:

1. **Talk with your doctor or a genetic specialist**
   They can help you understand your results and what they mean for your health or your child's health. If a genetic specialist was not involved in your testing, your doctor may refer you to one.

2. **Discuss your health management options**
   Depending on the condition, your care plan may include:
   - Changes to care, including medications, equipment, or therapies
   - Referrals to specialists with expertise in the condition
   - More frequent or earlier health screenings
   - Opportunities to explore emerging treatments or join clinical trials
   - Preventive steps, such as lifestyle changes, medications, or – in some cases – surgery

3. **Consider what this may mean for your family**
   Some genetic changes can be inherited, which means your close relatives may also want to consider genetic testing or speaking with a healthcare provider. Your doctor or genetic specialist can help you understand the inheritance pattern and guide you on how to navigate these conversations with your family.

4. **Seek support if and when you need it**
   Connecting with a therapist, support groups, or others with similar experiences can be helpful. There are many organizations that support individuals and families living with genetic conditions. A list of condition-specific support groups can be found at genedx.com/advocacy. For broader support, you can also look into: Child Neurology Foundation, Courageous Parents Network, Global Genes

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Have you or your child had *genetic testing* in the past?

It might be time to take another look. Why revisit genetic testing?

Genetic science is always developing. If you or your child had testing more than a year ago, there may be new answers that were not available before. Scientists are constantly learning about new connections between genes and conditions. This means that variants that were previously considered uncertain may be reclassified.

Our understanding of genetics is constantly improving as new disease-related genes are discovered and new connections between genes and conditions are identified. As a result, existing variants may be reclassified over time as more evidence becomes available.

What is a variant?

A variant is a change in DNA. Sometimes, results are not immediately clear and may be labeled as uncertain, likely benign, or likely pathogenic. As new evidence becomes available, these classifications can change over time.

Examples of reclassification

Uncertain → ***Likely pathogenic***
May help explain a diagnosis

Likely pathogenic → ***Benign***
No longer considered the cause

What is reanalysis and when to consider it:

Reanalysis is a fresh review of your or your child's existing genetic data using the latest medical knowledge. It may identify new variants, reevaluate previous findings, and update or clarify a diagnosis.

You may want to revisit testing if:

- **It has been a year or more since testing**
- **Your child has new symptoms or clinical findings**
- **No clear genetic answer was found previously**

How to request a reanalysis

Start by talking to your doctor about whether reanalysis may be appropriate. You can then locate the original test report and ask your doctor to contact the testing lab to request a reanalysis. In some cases, a new sample may be needed.

You can ask your doctor to request a reanalysis on your behalf.

A formal request may include:

- Patient name and date of birth
- Type of testing performed
- Year and lab of original testing
- Reason for reanalysis (time elapsed, new symptoms, etc.)

Not sure what to say? Start here.

A simple message to your doctor can help start the reanalysis process.

Dear Dr. [Last Name],

I hope you are doing well. I am writing regarding the genetic testing that ***[I/my child]***, ***[Full Name]***, born ***[DOB]***, received through ***[diagnostic laboratory]*** in ***[Year]***. I have attached a copy of the original report for your reference.

Given the ongoing advances in genetic research, I would like to request a reanalysis of ***[my/my child's]*** existing genetic data. I understand that reanalysis is typically recommended at least one year after the initial test, and may be especially helpful if there have been updates to clinical symptoms, family history, or additional diagnostic findings.

As I understand it, reanalysis involves reviewing previously generated sequencing data using the most current scientific and medical knowledge. This process may help identify new variants or provide updated interpretation of previously reported findings.

Would you be able to contact ***[diagnostic laboratory]*** to initiate this request? If a new sample is needed, I am happy to help coordinate next steps.

Please let me know if you need any additional information from me. Thank you for your continued care and support.

Sincerely,
[Your Name]
[Your Contact Information]

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