

Understanding your genetic testing *options*



If you are new to genetic testing, terms like panel testing, exome or 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

are designed to identify whether someone is at higher or lower risk for certain conditions. They do not provide a diagnosis, but could indicate whether further testing may be needed. These tests are typically performed on someone who does not have symptoms.



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.

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 exome 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.