Test type	Description	Use cases	Pros	Cons	Takeaway
Single gene test	Looks at one specific gene for variants (mutations). Looks for a known, gene variant (mutation) that has already been identified in a family member.	Used when a specific genetic condition is strongly suspected.	Highly accurate for a known of suspected condition.	Limited in scope. Won't detect possible other causes if this gene is not involved in the child's symptoms.	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.	Fast, cost-effective, and very accurate for that specific variant.	Can only detect that one change in the gene—it won't identify other variants that may exist.	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.	Broader look than a single-gene test and can be efficient for conditions with many genes associated with them.	If the variant gene isn't included on the test, a diagnosis could be missed.	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.	Can detect larger deletions or duplications not seen with other types of genetic tests.	Doesn't detect variants (mutations) within genes, so these types of genetic diagnoses can be missed.	If 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.	One of the most comprehensive options for genetic testing since it looks at more than 20,000 genes. Can diagnose conditions that are missed by other tests.	More costly out of pocket if it's not covered by insurance. Also increases the chances of finding a variant of uncertain significance (VUS), which doesn't give a clear genetic answer.	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 most comprehensive option for genetic testing. Can diagnose conditions that are missed by other tests.	More costly out of pocket if it's not covered by insurance. Also increases the chances of finding a variant of uncertain significance (VUS), which doesn't give a clear genetic answer.	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.	These can be fun tests for curiosity or family history insights.	Not clinically validated and should not be used for medical decision making. Not covered by insurance.	This is a different type of genetic test that is not recommended for those trying to understand medical symptoms or diagnose a condition.