Exome and Genome Testing in Clinical Practice

Exome and genome testing are used to analyze a patient's genetic material to identify or rule out causes of rare or unexplained conditions. Results can provide long-awaited diagnostic answers to families and help inform care decisions. This guide helps you identify appropriate patients, know when to recommend testing, and explain results clearly to patients and families.

Guideline-based Considerations for Exome and Genome Testing

Professional Medical Societies (ACMG, AAP, NSGC) recommend either exome or genome testing as first-line tests for children with:

- Developmental Delay[1,2]
- Intellectual Disability[1,2]
- Multiple Congenital Anomalies[1]
- Idiopathic epilepsy/seizures[3]

Testing may also benefit patients with complex care needs, like those with hearing or vision issues, neuromuscular issues, autism spectrum, or growth problems.

Of note, even if prior genetic testing was negative, MyOme may analyze more variants and identify answers that were previously missed.

Benefits of Exome and Genome Testing

Choosing between exome and genome testing depends on state-wide or individual insurance coverage, previous genetic testing history, and patient preference. Since all MyOme tests start with whole-genome sequencing, upgrading from exome to genome analysis is as simple as re-analyzing existing data — no new appointments, samples, or sequencing needed.

- **Genome testing** looks at nearly all of a patient's DNA, including genes and other important regions.
- **Exome testing** looks at the parts of the DNA that make up genes, which harbor a substantial proportion of diagnostic variants associated with rare diseases.

Test results can empower providers and patients by:

Guiding care

Results inform appropriate tests, treatments, and follow-up plans.

Informing Family Risks

Testing may reveal other family members who may be at risk for the same condition.

Offering Support

Diagnosis helps empower families with support groups, services, and research opportunities.

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Explaining Results to Patients

MyOme provides patient resources and embedded genetic counseling with every test. Below are simplified definitions of each possible test result.

Results Related to Clinical Phenotype

Positive
A genetic change that explains the clinical phenotype was detected.

Uncertain
A genetic change was found, but it is unclear if it is related to the clinical phenotype.

Negative
A genetic change explaining the clinical phenotype was not found.

Patient Decision for Optional Results

Secondary finding
Reports specific, unrelated genetic risks (such as certain cancers or heart conditions) that may be preventable or treatable if identified early.

Incidental finding
A genetic change that was not looked for but was found unexpectedly during the testing process. It is only reported if there is a clear medical action to help maintain health, such as screening or treatment.

Testing Workflow

Ordering	Sample Collection	Results	Support
Submit an order via MyOme's secure provider portal.	Collect in-clinic or at-home. Including family samples* is recommended to improve diagnostic accuracy.	GINA**-protected results are delivered in ~5-6 weeks via the secure portal.	Genetic counseling sessions are included to explain results and what they mean for families.

Learn more and get started
MyOme.com

Watch our consent video
YouTube.com/@MyOmeInc

*Order duo or trio kits if family (parent or sibling) samples will be sent. **Genetic Information Nondiscrimination Act

Tests were developed, and their performance characteristics were determined, by MyOme, Inc., a clinical laboratory certified under the Clinical Laboratory Improvement Amendments of 1988 (CLIA) and College of American Pathologist (CAP) accredited to perform high complexity clinical laboratory testing. These tests have not been cleared or approved by the U.S. Food and Drug Administration (FDA). Test results should always be interpreted by a clinician in the context of clinical and familial data with the availability of genetic counseling when appropriate. MyOme is not responsible for the content or accuracy of third-party websites.

1. Manickam K, et al. Genet Med. 2021;23(11):2029-2037(ES). **2**. Lance H. Rodan, Joan Stoler, Emily Chen, Timothy Geleske, Council on Genetics; Genetic Evaluation of the Child With Intellectual Disability or Global Developmental Delay: Clinical Report. Pediatrics July 2025; 156 (1): e2025072219. 10.1542/peds.2025-072219 3. Smith L, et al. J Genet Couns. 2023 Apr;32(2):266-280.

