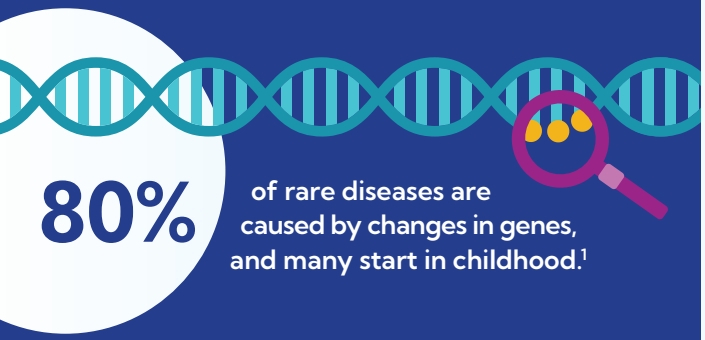
How can genetic testing lead to diagnosis?

Genetic testing looks at a person's genetic material, called DNA, to see if there are any genetic changes that could be causing a person's health problems.



80% of rare diseases are caused by changes in genes, and many start in childhood.[1]

When should testing be considered?

Medical experts* recommend genome or exome testing as a first step for patients with delays in development, learning differences, certain birth conditions, or unexplained seizures.

What are the benefits of genetic testing?

Testing can explain what is causing a person's health problem, which can help:

Guide Care

Results help doctors choose the right tests, treatments, and follow-up plans.

Help Families

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

Find Support

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

*American Academy of Pediatrics (AAP), The American College of Medical Genetics and Genomics (ACMG), The National Society of Genetic Counselors

Frequently Asked Questions

What if we already had genetic testing before?

Earlier tests may have looked at a narrow set of genes. By looking at more of the DNA, MyOme helps find answers that might have been missed before. Science also changes quickly—new research may offer new answers.

Are there things genetic testing can't find?

Yes. Exome and genome tests can find many genetic changes, but not all. Some parts of DNA are hard to read, and some conditions do not have a known genetic cause yet.

Could this testing affect other family members?

Some results may show genetic risks that run in families. Your doctor may recommend that family members consider testing or discuss what the result means for future pregnancies.

Why should I submit family samples?

You may be asked to include samples from close family members, such as parents or siblings, as part of testing. Adding family members can help make results clearer.

What happens to my sample and genetic data?

Genetic data is kept private and secure, like other medical records, and is only accessed by the lab or care team for medical care or approved research.

How can I get help understanding my results?

You should review your results with the doctor who ordered your testing to understand what they mean and how they might impact next steps. MyOme also provides access to optional genetic counseling services. Your provider can request this by emailing **support@myome.com**.

How much does testing cost?

The cost of testing depends on your payment type, insurance plan, and eligibility for our financial assistance program, MyOme Access. Learn more and apply at **myome.com/access.**

Watch our educational video to learn more

www.MyOme.com

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. The Lancet Global Health. The landscape for rare diseases in 2024. Lancet Glob Health. 2024;12(3):e341.doi:10.1016/S2214-109X(24)00056-1.

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Diagnostic Genetic Testing for Rare Diseases

Visit us at MyOme.com

MyOme's Approach to Rare Disease Diagnosis

MyOme offers three Rare Disease test options to meet the specific diagnostic needs of each patient.

- **Genome Analysis:** Looks at nearly all of your DNA, including genes and other important regions.
- **Exome Analysis:** Looks at the parts of your DNA that make up genes.
- **Copy Number Analysis:** Looks for missing or extra pieces of DNA.

Why MyOme?

Start with the Whole Story

All MyOme tests start with genome sequencing to produce a complete picture of your DNA, helping produce answers faster and with fewer steps. Your provider chooses how much of that information to review based on your symptoms and needs.

Revisit the Story Anytime

Because your DNA does not change, your provider may be able to review or expand testing later if new symptoms appear or if new genetic knowledge becomes available— often without needing a new sample.

Keep Answers Affordable

MyOme's billing team works with patients to reduce testing costs through insurance, discounts, and financial assistance.

myome® | Rare Disease

Getting Started

Collect Sample

A cheek swab or blood sample is collected in the clinic or at home using a MyOme kit. Including family samples can help improve the chance of finding answers.

Receive Results

Genetic results are shared with your provider in ~5–6 weeks through a secure portal. Your provider will review the results with you and discuss next steps.

Access Support

You'll have support at every step, including access to certified genetic counselors to explain your results and what they mean for you and your family—at no cost to you.

What Do Results Mean?

Results related to health concerns

Positive

We found a genetic change that explains the health concern. Your doctor or a genetic counselor will explain what this means for care, and whether other family members should consider testing.

Uncertain

We found a genetic change, but we do not know yet if it causes the health concern. Sometimes testing family members or checking the data again later (called re-analysis) can help us get a clearer answer.

Negative

We did not find a disease-causing genetic change related to the reported health concerns. This does not mean that there is not a genetic cause. You can ask your care team if re-analysis may be useful in the future as new information becomes available.

If you choose, you can also learn about results unrelated to the health concerns reported:

Secondary Finding

If you choose this option, we will look for additional, specific changes in the DNA linked to a small group of conditions (like certain cancers or heart problems) that are treatable or preventable if caught early. Family members submitting samples also have the option to receive their own report with these results.

Incidental Finding

This is a genetic change that we were not looking for, but it showed up during the testing process. We only tell you if there is a clear action you can take to stay healthy (like a screening test or treatment).