myome® | Rare Disease

Genome-based Diagnostic Tests for Rare Disease

The diagnostic odyssey for patients with rare disease is often prolonged by multiple rounds of testing, administrative burdens, and complicated logistics. MyOme's whole-genome tests use cutting-edge technology to streamline diagnostic workflows for both patients and providers.

Providing Patients and Providers with Tools for More Informed Diagnoses

Genome First

All test options use whole-genome sequencing (WGS) to improve sensitivity and variant detection

Flexible Options

Different test options are available to meet the diagnostic needs of the patient

Future-proofed Platform

WGS enables reanalysis as clinical profiles evolve and technology and knowledge advance

MyOme's Rare Disease Offerings

Rare Disease GENOME ANALYSIS

Comprehensive analysis of single nucleotide variants (SNVs), insertions and deletions (indels), copy number variant (CNVs), mitochondrial DNA (mtDNA) variants, and select tandem repeat expansions (TREs) across the protein coding and noncoding regions of the genome. Available as proband, duo, or trio analysis*.

Rare Disease EXOME ANALYSIS

Includes SNV and indel variant analysis of the coding regions of the nuclear genome and genome-wide analysis of CNVs, along with mtDNA variant analysis and select TRE analysis. Available as proband, duo, or trio analysis*.

Rare Disease COPY NUMBER ANALYSIS

Genome-wide analysis of CNVs.

***Proband:** individual; **Duo:** + 1 immediate family member (parent or sibling); **Trio:** + 2 immediate family members (parent or sibling)

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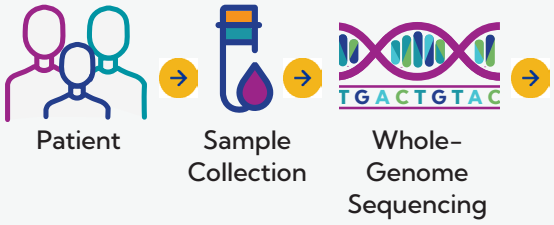
Single Platform. Multiple Insights.



Patient → Sample Collection → Whole-Genome Sequencing

Whole-Genome Platform

- SNV and indel Analysis
- CNV Analysis
- ROH and UPD Analysis*
- mtDNA variant Analysis
- TRE Analysis

*ROH: Runs of Homozygosity; UPD: Uniparental Disomy. Applicable to Duo or Trio tests with parent samples.

Key Features

Reimbursement Support

Prior authorizations, reimbursement, and affordable patient-pay options are all handled by MyOme. Financial assistance is offered through our MyOme Access program.

Provider-friendly Workflow

Cost-effective, streamlined options for reflexing to broader assays are easily accessible for providers.

Genetic Counseling

Support from a certified genetic counselor is available to help patients and families understand their results, enabling informed health decisions.

Get Started with Our Simple, Seamless Process

Order	Sample Collection	Sample Analysis	Receive Results
Submit an order via MyOme's secure portal	Use instructions provided in blood, saliva, or buccal swab collection kits	Return sample to MyOme for sequencing and data analysis	Reports are delivered through MyOme's secure portal

Interested in MyOme diagnostic testing?
Contact support@myome.com or visit our website to learn more.

This test was developed, and its 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. This test has 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.

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