myome

Our Genome-First Approach Detects More.

All MyOme Rare Disease tests start with whole-genome sequencing to produce a comprehensive genome backbone. Exome Analysis using a genome backbone enables deeper, more uniform coverage to detect diagnostic variants that traditional platforms often miss.

Real-World Case Study

1 Meet 8-year old Sam*

Diagnosed with congenital hypotonia, autism spectrum disorder, global developmental delay, and gait abnormalities

2 Diagnostic Odyssey

Whole-exome sequencing by another lab did not detect any pathogenic variants associated with the clinical phenotype

3 MyOme test ordered

Exome Analysis identified a previously undetected single-exon deletion in the SOX5 gene, which can cause Lamb-Shaffer syndrome

4 Answers that matter

A genetic diagnosis enabled more personalized care and opened the door to specialized support networks and research opportunities

*Names are fictionalized

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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Cutting-Edge Innovation Finds Answers.

Built-in long-read sequencing** for exome and genome tests reliably detects complex variants, like tandem repeat expansions (TREs), and offers methylation insights that can aid in variant interpretation.

Real-World Case Study

1 Meet 24-year old Tom*

Began experiencing non-exercise-related muscle spasms at age 22, with a personal and family history of congenital cataracts

2 Diagnostic Odyssey

Neuromuscular genetic panels conducted by other labs (including DMPK repeat analysis) did not detect any pathogenic variants associated with the clinical phenotype

3 MyOme test ordered

Genome Analysis with confirmatory long-read sequencing detected a tandem repeat expansion in the DMPK gene (≥50 repeats) that was missed by prior testing

4 Answers that matter

A genetic diagnosis of Myotonic dystrophy 1 (DM1) explained Tom's symptoms, providing crucial answers and clarity for next steps in care

Starting With More Means Finding More

Backed by the latest genomic technology, we offer improved detection and faster results, so every family can get the answers and care they need.

Learn more at www.MyOme.com/our-tests/diagnostic

**For select genes

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